GENE LIST DG 3.6.0 (5009 GENES)

Releasedate: 05-04-2023

Gene	*TWIST X2 covered >10x*	*TWIST X2 covered >20x*	*srWGS GRCh38 covered >10x*	*srWGS GRCh38 covered >20x*	*Associated Phenotype description and OMIM disease ID*	*genepanel*
A2M	100%	100%	100%	99%	No OMIM disease ID	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME
A2ML1	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME TUMOR
A4GALT	100%	100%	100%	97%	NOR polyagglutination syndrome, 111400	MENDELIOME
AAAS	100%	100%	100%	99%	Achalasia-addisonianism-alacrimia syndrome, 231550	SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
AAGAB	100%	100%	100%	97%	Keratoderma, palmoplantar, punctate type IA, 148600	SKIN DISORDERS MENDELIOME
AARS1	100%	100%	100%	99%	Developmental and epileptic encephalopathy 29, 616339 Charcot-Marie-Tooth disease, axonal, type 2N, 613287 ?Leukoencephalopathy, hereditary diffuse, with spheroids 2, 619661 Trichothiodystrophy 8, nonphotosensitive, 619691	EPILEPSY NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
AARS2	100%	100%	100%	99%	Leukoencephalopathy, progressive, with ovarian failure, 615889 Combined oxidative phosphorylation deficiency 8, 614096	MOVEMENT DISORDERS DISORDERS OF SEX DEVELOPMENT HEART MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY
AASS	100%	100%	100%	98%	Hyperlysinemia, 238700	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

ABAT	100%	100%	100%	98%	GABA-transaminase deficiency, 613163	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
ABCA1	100%	100%	100%	99%	Tangier disease, 205400 HDL deficiency, familial, 1, 604091	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING
ABCA12	100%	100%	100%	98%	Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500 Ichthyosis, congenital, autosomal recessive 4A, 601277	SKIN DISORDERS MENDELIOME
ABCA2	100%	100%	99%	97%	Intellectual developmental disorder with poor growth and w/wo seizures or ataxia, 618808	INTELLECTUAL DISABILITY MENDELIOME
ABCA3	100%	100%	100%	99%	Surfactant metabolism dysfunction, pulmonary, 3, 610921	MENDELIOME PRE CONCEPTION SCREENING
ABCA4	100%	100%	100%	99%	Retinal dystrophy, early-onset severe, 248200 Retinitis pigmentosa 19, 601718 Cone-rod dystrophy 3, 604116 Fundus flavimaculatus, 248200 Stargardt disease 1, 248200	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ABCA5	100%	99%	100%	97%	?Hypertrichosis, congenital generalized, with gingival hyperplasia, 135400	MENDELIOME
ABCB10	100%	100%	99%	96%	No OMIM disease ID	IRON DISORDERS MENDELIOME
ABCB11	100%	99%	100%	98%	Cholestasis, benign recurrent intrahepatic, 2, 605479 Cholestasis, progressive familial intrahepatic 2, 601847	LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ABCB4	100%	100%	100%	98%	Gallbladder disease 1, 600803 Cholestasis, intrahepatic, of pregnancy, 3, 614972 Cholestasis, progressive familial intrahepatic 3, 602347	LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ABCB6	100%	100%	100%	99%	Microphthalmia, isolated, with coloboma 7, 614497 Dyschromatosis universalis hereditaria 3, 615402 Pseudohyperkalemia, familial, 2, due to red cell leak, 609153	VISION DISORDERS SKIN DISORDERS MENDELIOME
ABCB7	99%	99%	98%	75%	Anemia, sideroblastic, with ataxia, 301310	MOVEMENT DISORDERS HEREDITARY BONE MARROW FAILURE IRON DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS
ABCC1	100%	100%	100%	98%	?Deafness, autosomal dominant 77, 618915	HEARING IMPAIRMENT MENDELIOME
ABCC2	100%	100%	100%	98%	Dubin-Johnson syndrome, 237500	LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING

ABCC6	100%	100%	100%	99%	Pseudoxanthoma elasticum, 264800 Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, forme fruste, 177850	VISION DISORDERS SKIN DISORDERS HEART MENDELIOME PRE CONCEPTION SCREENING
ABCC8	100%	100%	100%	99%	Diabetes mellitus, permanent neonatal 3, w/wo neurologic features, 618857 Diabetes mellitus, transient neonatal 2, 610374 Diabetes mellitus, noninsulin-dependent, 125853 Hypoglycemia of infancy, leucine-sensitive, 240800 Hyperinsulinemic hypoglycemia, familial, 1, 256450	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ABCC9	100%	100%	100%	98%	Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850 ?Atrial fibrillation, familial, 12, 614050 Intellectual disability and myopathy syndrome, 619719	SKIN DISORDERS HEART SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
ABCD1	100%	99%	99%	76%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100	MOVEMENT DISORDERS DISORDERS OF SEX DEVELOPMENT NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME
ABCD2	100%	100%	100%	98%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME
ABCD3	100%	100%	100%	97%	?Bile acid synthesis defect, congenital, 5, 616278	LIVER DISORDERS METABOLIC DISORDERS MENDELIOME
ABCD4	100%	100%	100%	98%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857	HEREDITARY BONE MARROW FAILURE METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ABCG5	100%	100%	100%	98%	Sitosterolemia 2, 618666	HEMOSTATIC/THROMBOTIC DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ABCG8	100%	100%	100%	99%	Sitosterolemia 1, 210250	HEMOSTATIC/THROMBOTIC DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING

ABHD12	100%	100%	99%	97%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674	MOVEMENT DISORDERS VISION DISORDERS HEARING IMPAIRMENT NEUROPATHIES METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ABHD16A	100%	100%	100%	98%	Spastic paraplegia 86, autosomal recessive, 619735	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ABHD5	100%	100%	100%	99%	Chanarin-Dorfman syndrome, 275630	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ABL1	100%	100%	100%	99%	Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 608232 Congenital heart defects and skeletal malformations syndrome, 617602	ANEURYSM CONGENITAL HEART DISEASE HEART MENDELIOME
ACACA	100%	100%	100%	99%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ACAD8	100%	100%	100%	99%	Isobutyryl-CoA dehydrogenase deficiency, 611283	HEART METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ACAD9	100%	100%	100%	99%	Mitochondrial complex I deficiency, nuclear type 20, 611126	HEART METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
ACADM	100%	100%	100%	97%	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ACADS	100%	100%	100%	99%	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

ACADSB	100%	100%	100%	98%	2-methylbutyrylglycinuria, 610006	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ACADVL	100%	100%	99%	96%	VLCAD deficiency, 201475	HEART METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
ACAN	99%	99%	96%	92%	?Spondyloepiphyseal dysplasia, Kimberley type, 608361 Short stature and advanced bone age, w/wo early-onset osteoarthritis and/or osteochondritis dissecans, 165800 Spondyloepimetaphyseal dysplasia, aggrecan type, 612813	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
ACAT1	100%	100%	99%	95%	Alpha-methylacetoacetic aciduria, 203750	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ACAT2	100%	100%	100%	98%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME
ACBD5	100%	100%	100%	98%	Retinal dystrophy with leukodystrophy, 618863	VISION DISORDERS HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS METABOLIC DISORDERS MENDELIOME
ACD	100%	100%	100%	98%	?Dyskeratosis congenita, autosomal recessive 7, 616553 ?Dyskeratosis congenita, autosomal dominant 6, 616553	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS DYSKERATOSIS CONGENITA PRIMARY IMMUNODEFICIENCY MENDELIOME TUMOR
ACE	100%	100%	99%	96%	Renal tubular dysgenesis, 267430	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ACER3	100%	100%	99%	97%	?Leukodystrophy, progressive, early childhood-onset, 617762	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ACKR3	100%	100%	100%	99%	?Oculomotor-abducens synkinesis, 619215	MENDELIOME

ACO2	100%	100%	100%	99%	Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559	VISION DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
ACOX1	100%	100%	100%	99%	Mitchell syndrome, 618960 Peroxisomal acyl-CoA oxidase deficiency, 264470	NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ACOX2	100%	100%	100%	99%	Bile acid synthesis defect, congenital, 6, 617308	LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ACP4	100%	100%	100%	99%	Amelogenesis imperfecta, type IJ, 617297	CRANIOFACIAL ANOMALIES MENDELIOME
ACP5	100%	100%	100%	99%	Spondyloenchondrodysplasia with immune dysregulation, 607944	PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
ACSF3	100%	100%	100%	98%	Combined malonic and methylmalonic aciduria, 614265	HEART METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ACSL4	100%	100%	97%	73%	Intellectual developmental disorder, X-linked 63, 300387	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME
ACSL6	100%	100%	100%	99%	Myelodysplastic syndrome, Myelogenous leukemia, acute,	MENDELIOME
ACTA1	100%	100%	100%	97%	Congenital myopathy 2B, severe infantile, autosomal recessive, 620265 ?Myopathy, scapulohumeroperoneal, 616852 Congenital myopathy 2C, severe infantile, autosomal dominant, 620278 Congenital myopathy 2A, typical, autosomal dominant, 161800	FETAL AKINESIA MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS
ACTA2	99%	99%	100%	99%	Multisystemic smooth muscle dysfunction syndrome, 613834 Aortic aneurysm, familial thoracic 6, 611788 Moyamoya disease 5, 614042	ANEURYSM SKIN DISORDERS LIVER DISORDERS MENDELIOME

ACTB	100%	100%	100%	99%	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371	MOVEMENT DISORDERS SKIN DISORDERS HEARING IMPAIRMENT EPILEPSY HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS
ACTC1	100%	100%	100%	99%	Left ventricular noncompaction 4, 613424 Cardiomyopathy, hypertrophic, 11, 612098 Atrial septal defect 5, 612794 Cardiomyopathy, dilated, 1R, 613424	CONGENITAL HEART DISEASE DILATED CARDIOMYOPATHY HEART HYPERTROPHIC CARDIOMYOPATHY MENDELIOME
ACTG1	100%	100%	100%	98%	Deafness, autosomal dominant 20/26, 604717 Baraitser-Winter syndrome 2, 614583	CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS
ACTG2	100%	100%	100%	99%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 5, 619431 Visceral myopathy 1, 155310	LIVER DISORDERS MENDELIOME
ACTL6A	100%	100%	100%	98%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
ACTL6B	100%	100%	100%	99%	Developmental and epileptic encephalopathy 76, 618468 Intellectual developmental disorder with severe speech and ambulation defects, 618470	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ACTL9	100%	100%	100%	99%	Spermatogenic failure 53, 619258	MALE INFERTILITY MENDELIOME
ACTN1	100%	100%	100%	99%	Bleeding disorder, platelet-type, 15, 615193	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME
ACTN2	100%	100%	99%	97%	Myopathy, distal, 6, adult onset, 618655 Cardiomyopathy, hypertrophic, 23, w/wo LVNC, 612158 Congenital myopathy 8, 618654 Cardiomyopathy, dilated, 1AA, w/wo LVNC, 612158	DILATED CARDIOMYOPATHY HEART HYPERTROPHIC CARDIOMYOPATHY MENDELIOME
ACTN4	100%	100%	100%	98%	Glomerulosclerosis, focal segmental, 1, 603278	RENAL DISORDERS MENDELIOME

ACVR1	100%	99%	100%	98%	Fibrodysplasia ossificans progressiva, 135100	IRON DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME MUSCLE DISORDERS
ACVR1B	100%	100%	100%	99%	Pancreatic cancer, somatic,	MENDELIOME
ACVR2B	100%	100%	100%	98%	Heterotaxy, visceral, 4, autosomal, 613751	CONGENITAL HEART DISEASE CILIOPATHIES HEART MENDELIOME
ACVRL1	100%	100%	100%	99%	Telangiectasia, hereditary hemorrhagic, type 2, 600376	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME
ACY1	100%	100%	100%	99%	Aminoacylase 1 deficiency, 609924	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ADA	100%	100%	100%	99%	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700	PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)
ADA2	100%	100%	100%	99%	Sneddon syndrome, 182410 Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
ADAD2	100%	100%	100%	99%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
ADAM10	100%	100%	100%	97%	Reticulate acropigmentation of Kitamura, 615537	SKIN DISORDERS MENDELIOME
ADAM17	100%	100%	100%	98%	?Inflammatory skin and bowel disease, neonatal, 1, 614328	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
ADAM22	100%	100%	100%	98%	Developmental and epileptic encephalopathy 61, 617933	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

ADAM9	100%	100%	100%	98%	Cone-rod dystrophy 9, 612775	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ADAMTS1	100%	100%	100%	99%	No OMIM disease ID	HEARING IMPAIRMENT MENDELIOME
ADAMTS10	100%	100%	100%	99%	Weill-Marchesani syndrome 1, recessive, 277600	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
ADAMTS13	100%	100%	100%	98%	Thrombotic thrombocytopenic purpura, hereditary, 274150	HEMOSTATIC/THROMBOTIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ADAMTS17	100%	100%	100%	97%	Weill-Marchesani 4 syndrome, recessive, 613195	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
ADAMTS18	100%	100%	100%	98%	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ADAMTS19	100%	100%	100%	98%	Cardiac valvular dysplasia 2, 620067	HEART MENDELIOME
ADAMTS2	97%	97%	100%	98%	Ehlers-Danlos syndrome, dermatosparaxis type, 225410	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ADAMTS3	99%	98%	100%	99%	Hennekam lymphangiectasia-lymphedema syndrome 3, 618154	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ADAMTS9	99%	99%	100%	98%	No OMIM disease ID	CILIOPATHIES RENAL DISORDERS MENDELIOME
ADAMTSL2	100%	99%	100%	99%	Geleophysic dysplasia 1, 231050	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING

ADAMTSL4	100%	100%	100%	98%	Ectopia lentis et pupillae, 225200 Ectopia lentis, isolated, autosomal recessive, 225100	VISION DISORDERS CRANIOFACIAL ANOMALIES MENDELIOME PRE CONCEPTION SCREENING
ADAR	100%	100%	100%	98%	Dyschromatosis symmetrica hereditaria, 127400 Aicardi-Goutieres syndrome 6, 615010	MOVEMENT DISORDERS SKIN DISORDERS PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ADARB1	95%	94%	100%	99%	Neurodevelopmental disorder with hypotonia, microcephaly, and seizures, 618862	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ADAT3	100%	100%	100%	100%	Neurodevelopmental disorder with brain abnormalities, poor growth, and dysmorphic facies, 615286	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ADCK2	100%	100%	100%	98%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
ADCK5	100%	100%	100%	99%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME
ADCY1	99%	98%	99%	95%	?Deafness, autosomal recessive 44, 610154	HEARING IMPAIRMENT MENDELIOME
ADCY10	100%	100%	100%	98%	No OMIM disease ID	MALE INFERTILITY RENAL DISORDERS MENDELIOME
ADCY3	100%	100%	100%	98%	No OMIM disease ID	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME
ADCY5	100%	99%	100%	97%	Dyskinesia with orofacial involvement, autosomal dominant, 606703 Neurodevelopmental disorder with hyperkinetic movements and dyskinesia, 619651 Dyskinesia with orofacial involvement, autosomal recessive, 619647	MOVEMENT DISORDERS HEART METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME
ADCY6	100%	100%	100%	99%	Lethal congenital contracture syndrome 8, 616287	FETAL AKINESIA MENDELIOME PRE CONCEPTION SCREENING
ADD1	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
ADD3	100%	100%	100%	98%	Cerebral palsy, spastic quadriplegic, 3, 617008	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ADGRE2	99%	99%	99%	97%	Vibratory urticaria, 125630	MENDELIOME

ADGRG1	100%	100%	100%	99%	Polymicrogyria, bilateral frontoparietal, 606854 Polymicrogyria, bilateral perisylvian, 615752	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ADGRG2	100%	99%	97%	71%	Congenital bilateral absence of vas deferens, X-linked, 300985	MALE INFERTILITY MENDELIOME
ADGRG6	100%	99%	100%	98%	Lethal congenital contracture syndrome 9, 616503	FETAL AKINESIA MENDELIOME PRE CONCEPTION SCREENING
ADGRV1	100%	100%	100%	98%	Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472 ?Febrile seizures, familial, 4, 604352	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
ADH5	100%	100%	100%	98%	AMED syndrome, digenic, 619151	MENDELIOME
ADIPOQ	100%	100%	100%	99%	Adiponectin deficiency, 612556	MENDELIOME
ADIPOR1	100%	100%	100%	99%	No OMIM disease ID	VISION DISORDERS MENDELIOME
ADK	90%	90%	100%	98%	Hypermethioninemia due to adenosine kinase deficiency, 614300	LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ADNP	100%	100%	100%	98%	Helsmoortel-van der Aa syndrome, 615873	CONGENITAL HEART DISEASE HEART INTELLECTUAL DISABILITY MENDELIOME
ADPRS	100%	100%	100%	99%	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170	MOVEMENT DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
ADRB2	100%	100%	100%	99%	Beta-2-adrenoreceptor agonist, reduced response to,	MENDELIOME
ADSL	100%	100%	100%	98%	Adenylosuccinase deficiency, 103050	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ADSS1	100%	100%	100%	98%	Myopathy, distal, 5, 617030	MENDELIOME PRE CONCEPTION SCREENING
AEBP1	100%	100%	100%	98%	Ehlers-Danlos syndrome, classic-like, 2, 618000	MENDELIOME PRE CONCEPTION SCREENING

AFF2	100%	99%	98%	71%	Intellectual developmental disorder, X-linked 109, 309548	INTELLECTUAL DISABILITY MENDELIOME
AFF3	100%	100%	100%	98%	KINSSHIP syndrome, 619297	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
AFF4	100%	100%	100%	98%	CHOPS syndrome, 616368	INTELLECTUAL DISABILITY MENDELIOME
AFG3L2	100%	100%	100%	98%	Spastic ataxia 5, autosomal recessive, 614487 Optic atrophy 12, 618977 Spinocerebellar ataxia 28, 610246	MOVEMENT DISORDERS VISION DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
AFP	100%	100%	100%	98%	Alpha-fetoprotein deficiency, 615969	MENDELIOME
AGA	100%	100%	100%	98%	Aspartylglucosaminuria, 208400	MOVEMENT DISORDERS SKIN DISORDERS EPILEPSY PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
AGAP1	100%	100%	99%	91%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
AGBL1	99%	99%	100%	99%	Corneal dystrophy, Fuchs endothelial, 8, 615523	VISION DISORDERS MENDELIOME
AGBL5	100%	100%	100%	99%	Retinitis pigmentosa 75, 617023	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
AGK	91%	91%	100%	98%	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350	VISION DISORDERS HEART METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING

AGL	100%	100%	100%	98%	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400	HEART METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
AGMO	100%	100%	99%	97%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
AGO1	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
AGO2	100%	99%	99%	98%	Lessel-Kreienkamp syndrome, 619149	INTELLECTUAL DISABILITY MENDELIOME
AGPAT2	100%	100%	100%	97%	Lipodystrophy, congenital generalized, type 1, 608594	SKIN DISORDERS HEART METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
AGPS	100%	100%	100%	96%	Rhizomelic chondrodysplasia punctata, type 3, 600121	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
AGRN	100%	100%	100%	98%	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
AGT	100%	100%	100%	99%	Renal tubular dysgenesis, 267430	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
AGTPBP1	100%	100%	100%	97%	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276	MOVEMENT DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
AGTR1	100%	100%	100%	99%	Renal tubular dysgenesis, 267430	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
AGXT	100%	100%	100%	99%	Hyperoxaluria, primary, type 1, 259900	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING

AHCY	100%	100%	100%	99%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752	LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
AHDC1	100%	100%	100%	98%	Xia-Gibbs syndrome, 615829	INTELLECTUAL DISABILITY MENDELIOME
AHI1	100%	100%	100%	98%	Joubert syndrome 3, 608629	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
AHNAK2	97%	97%	95%	92%	No OMIM disease ID	NEUROPATHIES MENDELIOME
AHR	100%	100%	100%	98%	?Retinitis pigmentosa 85, 618345	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
AHSG	100%	100%	100%	98%	?Alopecia-intellectual disability syndrome 1, 203650	INTELLECTUAL DISABILITY MENDELIOME
AICDA	100%	100%	100%	98%	Immunodeficiency with hyper-IgM, type 2, 605258	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
AIFM1	100%	99%	97%	68%	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 Deafness, X-linked 5, 300614	HEARING IMPAIRMENT NEUROPATHIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS
AIMP1	100%	100%	100%	98%	Leukodystrophy, hypomyelinating, 3, 260600	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
AIMP2	100%	100%	100%	99%	Leukodystrophy, hypomyelinating, 17, 618006	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
AIP	100%	100%	100%	99%	Pituitary adenoma 1, multiple types, 102200 Pituitary adenoma predisposition, 102200	ENDOCRINE TUMOR MENDELIOME TUMOR

AIPL1	100%	100%	100%	99%	Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393 Cone-rod dystrophy, 604393	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
AIRE	100%	100%	100%	99%	Autoimmune polyendocrinopathy syndrome , type I, w/wo reversible metaphyseal dysplasia, 240300	SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
AK1	100%	100%	100%	99%	Hemolytic anemia due to adenylate kinase deficiency, 612631	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
AK2	100%	100%	100%	99%	Reticular dysgenesis, 267500	PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)
AK7	100%	100%	100%	97%	?Spermatogenic failure 27, 617965	MENDELIOME
AKAP9	100%	100%	100%	97%	?Long QT syndrome 11, 611820	HEART MENDELIOME
AKR1C1	100%	100%	100%	98%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME
AKR1C2	100%	100%	99%	98%	46XY sex reversal 8, 614279	DISORDERS OF SEX DEVELOPMENT MENDELIOME
AKR1D1	100%	100%	100%	98%	Bile acid synthesis defect, congenital, 2, 235555	LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
AKT1	100%	100%	100%	99%	Breast cancer, somatic, 114480 Cowden syndrome 6, 615109 Colorectal cancer, somatic, 114500 Proteus syndrome, somatic, 176920 Ovarian cancer, somatic, 167000	SKIN DISORDERS MENDELIOME TUMOR
AKT2	100%	100%	100%	98%	Diabetes mellitus, type II, 125853 Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900	 MENDELIOME
AKT3	100%	99%	100%	98%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME

ALAD	100%	100%	100%	99%	Porphyria, acute hepatic, 612740	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ALAS2	100%	99%	98%	73%	Anemia, sideroblastic, 1, 300751 Protoporphyria, erythropoietic, X-linked, 300752	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS IRON DISORDERS METABOLIC DISORDERS MENDELIOME
ALB	100%	100%	100%	97%	Analbuminemia, 616000	MENDELIOME PRE CONCEPTION SCREENING
ALDH18A1	100%	100%	100%	99%	Spastic paraplegia 9A, autosomal dominant, 601162 Cutis laxa, autosomal recessive, type IIIA, 219150 Spastic paraplegia 9B, autosomal recessive, 616586 Cutis laxa, autosomal dominant 3, 616603	MOVEMENT DISORDERS VISION DISORDERS SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ALDH1A2	100%	99%	100%	98%	Diaphragmatic hernia 4, with cardiovascular defects, 620025	CONGENITAL HEART DISEASE HEART MENDELIOME
ALDH1A3	100%	100%	100%	97%	Microphthalmia, isolated 8, 615113	VISION DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ALDH1B1	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
ALDH2	100%	100%	100%	98%	Alcohol sensitivity, acute, 610251	METABOLIC DISORDERS MENDELIOME
ALDH3A2	93%	93%	100%	98%	Sjogren-Larsson syndrome, 270200	MOVEMENT DISORDERS VISION DISORDERS SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ALDH4A1	100%	100%	100%	98%	Hyperprolinemia, type II, 239510	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

ALDH5A1	100%	100%	100%	97%	Succinic semialdehyde dehydrogenase deficiency, 271980	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ALDH6A1	100%	100%	99%	97%	Methylmalonate semialdehyde dehydrogenase deficiency, 614105	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ALDH7A1	100%	100%	100%	99%	Epilepsy, pyridoxine-dependent, 266100	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ALDOA	100%	100%	100%	99%	Glycogen storage disease XII, 611881	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ALDOB	100%	100%	100%	99%	Fructose intolerance, hereditary, 229600	SKIN DISORDERS LIVER DISORDERS METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ALG1	100%	100%	100%	99%	Congenital disorder of glycosylation, type Ik, 608540	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ALG10	100%	100%	100%	98%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME
ALG11	96%	96%	100%	98%	Congenital disorder of glycosylation, type Ip, 613661	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

ALG12	100%	100%	100%	99%	Congenital disorder of glycosylation, type Ig, 607143	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ALG13	99%	99%	97%	71%	Developmental and epileptic encephalopathy 36, 300884	EPILEPSY PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME
ALG14	100%	100%	100%	98%	Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies, 619031 Myopathy, epilepsy, and progressive cerebral atrophy, 619036 ?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ALG2	100%	100%	100%	99%	Congenital disorder of glycosylation, type Ii, 607906 Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
ALG3	100%	100%	100%	97%	Congenital disorder of glycosylation, type Id, 601110	FETAL AKINESIA EPILEPSY SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ALG6	100%	100%	99%	96%	Congenital disorder of glycosylation, type Ic, 603147	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ALG8	96%	96%	100%	97%	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 w/wo kidney cysts, 617874	LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING

ALG9	100%	100%	100%	98%	Gillessen-Kaesbach-Nishimura syndrome, 263210 Congenital disorder of glycosylation, type Il, 608776	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ALK	100%	99%	100%	98%	No OMIM disease ID	MENDELIOME TUMOR
ALKBH1	100%	100%	100%	97%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
ALKBH8	100%	100%	100%	99%	Intellectual developmental disorder, autosomal recessive 71, 618504	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ALMS1	100%	100%	100%	98%	Alstrom syndrome, 203800	VISION DISORDERS CILIOPATHIES HEARING IMPAIRMENT HEART SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ALOX12B	100%	100%	100%	98%	Ichthyosis, congenital, autosomal recessive 2, 242100	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ALOXE3	100%	100%	100%	98%	Ichthyosis, congenital, autosomal recessive 3, 606545	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ALPI	100%	100%	100%	99%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
ALPK1	100%	100%	100%	99%	ROSAH syndrome, 614979	PRIMARY IMMUNODEFICIENCY MENDELIOME
ALPK3	100%	100%	100%	98%	Cardiomyopathy, familial hypertrophic 27, 618052	HEART HYPERTROPHIC CARDIOMYOPATHY MENDELIOME PRE CONCEPTION SCREENING

ALPL	100%	100%	100%	99%	Odontohypophosphatasia, 146300 Hypophosphatasia, infantile, 241500 Hypophosphatasia, childhood, 241510 Hypophosphatasia, adult, 146300	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ALS2	100%	100%	100%	98%	Primary lateral sclerosis, juvenile, 606353 Spastic paralysis, infantile onset ascending, 607225 Amyotrophic lateral sclerosis 2, juvenile, 205100	ALS MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ALX1	100%	100%	100%	97%	Frontonasal dysplasia 3, 613456	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS
ALX3	100%	100%	100%	95%	Frontonasal dysplasia 1, 136760	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS
ALX4	100%	100%	100%	97%	Parietal foramina 2, 609597 Frontonasal dysplasia 2, 613451	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
AMACR	100%	100%	100%	97%	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950	VISION DISORDERS EPILEPSY NEUROPATHIES LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
AMBN	100%	99%	100%	97%	Amelogenesis imperfecta, type IF, 616270	CRANIOFACIAL ANOMALIES MENDELIOME PRE CONCEPTION SCREENING
AMELX	100%	100%	98%	69%	Amelogenesis imperfecta, type 1E, 301200	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME

AMER1	100%	100%	98%	73%	Osteopathia striata with cranial sclerosis, 300373	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS
AMH	100%	100%	100%	98%	Persistent Mullerian duct syndrome, type I, 261550	DISORDERS OF SEX DEVELOPMENT MENDELIOME TUMOR
AMHR2	100%	100%	100%	99%	Persistent Mullerian duct syndrome, type II, 261550	DISORDERS OF SEX DEVELOPMENT MENDELIOME TUMOR
AMMECR1	100%	99%	95%	65%	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990	HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS
AMN	100%	100%	100%	97%	Imerslund-Grasbeck syndrome 2, 618882	HEREDITARY BONE MARROW FAILURE METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
AMOTL1	100%	100%	100%	98%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME SCHISIS
AMPD1	100%	100%	100%	98%	Myopathy due to myoadenylate deaminase deficiency, 615511	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
AMPD2	100%	100%	99%	98%	?Spastic paraplegia 63, 615686 Pontocerebellar hypoplasia, type 9, 615809	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
AMPD3	100%	100%	100%	99%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME
AMT	100%	100%	100%	99%	Glycine encephalopathy, 605899	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

AMTN	100%	99%	100%	98%	?Amelogenesis imperfecta, type IIIB, 617607	CRANIOFACIAL ANOMALIES MENDELIOME
ANAPC1	100%	100%	100%	98%	Rothmund-Thomson syndrome, type 1, 618625	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
ANAPC7	100%	100%	100%	98%	Ferguson-Bonni neurodevelopmental syndrome, 619699	MENDELIOME
ANG	100%	100%	100%	99%	Amyotrophic lateral sclerosis 9, 611895	ALS MENDELIOME
ANGPT1	100%	100%	100%	98%	?Angioedema, hereditary, 5, 619361	PRIMARY IMMUNODEFICIENCY MENDELIOME
ANGPT2	100%	100%	100%	98%	Lymphatic malformation 10, 619369	SKIN DISORDERS MENDELIOME
ANGPTL3	100%	100%	100%	98%	Hypobetalipoproteinemia, familial, 2, 605019	MENDELIOME PRE CONCEPTION SCREENING
ANGPTL4	100%	100%	100%	99%	Plasma triglyceride level QTL, low, 615881	MENDELIOME
ANK1	100%	100%	100%	99%	Spherocytosis, type 1, 182900	MENDELIOME PRE CONCEPTION SCREENING
ANK2	100%	100%	100%	98%	Long QT syndrome 4, 600919 Cardiac arrhythmia, ankyrin-B-related, 600919	HEART INTELLECTUAL DISABILITY MENDELIOME
ANK3	100%	99%	100%	98%	Intellectual developmental disorder, autosomal recessive 37, 615493	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ANKFY1	100%	100%	100%	98%	No OMIM disease ID	RENAL DISORDERS MENDELIOME
ANKH	100%	100%	100%	99%	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ANKLE2	100%	100%	99%	94%	Microcephaly 16, primary, autosomal recessive, 616681	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ANKRD1	100%	99%	100%	96%	No OMIM disease ID	CONGENITAL HEART DISEASE HEART MENDELIOME

ANKRD11	100%	100%	100%	98%	KBG syndrome, 148050	CRANIOFACIAL ANOMALIES CONGENITAL HEART DISEASE SKIN DISORDERS EPILEPSY HEART SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS
ANKRD17	100%	100%	100%	98%	Chopra-Amiel-Gordon syndrome, 619504	INTELLECTUAL DISABILITY MENDELIOME
ANKRD26	97%	97%	100%	97%	Thrombocytopenia 2, 188000	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME TUMOR
ANKS1B	100%	100%	100%	98%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
ANKS6	99%	99%	100%	97%	Nephronophthisis 16, 615382	CILIOPATHIES LIVER DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ANLN	100%	100%	100%	98%	Focal segmental glomerulosclerosis 8, 616032	HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME
ANO10	100%	100%	100%	98%	Spinocerebellar ataxia, autosomal recessive 10, 613728	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
ANO3	100%	100%	100%	98%	Dystonia 24, 615034	MOVEMENT DISORDERS MENDELIOME
ANO5	100%	100%	100%	98%	Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307 Miyoshi muscular dystrophy 3, 613319 Gnathodiaphyseal dysplasia, 166260	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
ANO6	100%	100%	100%	98%	Scott syndrome, 262890	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING

ANOS1	100%	99%	97%	70%	Hypogonadotropic hypogonadism 1 w/wo anosmia (Kallmann syndrome 1), 308700	SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM RENAL DISORDERS MENDELIOME
ANTXR1	100%	99%	99%	94%	GAPO syndrome, 230740	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ANTXR2	100%	100%	100%	98%	Hyaline fibromatosis syndrome, 228600	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
ANXA11	100%	100%	100%	99%	Amyotrophic lateral sclerosis 23, 617839 Inclusion body myopathy and brain white matter abnormalities, 619733	ALS MENDELIOME
AOPEP	100%	100%	100%	98%	Dystonia 31, 619565	MENDELIOME
AP1B1	100%	100%	100%	99%	Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150	SKIN DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
AP1G1	100%	100%	100%	97%	Usmani-Riazuddin syndrome, autosomal recessive, 619548 Usmani-Riazuddin syndrome, autosomal dominant, 619467	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
AP1S1	100%	100%	100%	98%	MEDNIK syndrome, 609313	LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
AP1S2	100%	100%	97%	70%	Pettigrew syndrome, 304340	INTELLECTUAL DISABILITY MENDELIOME
AP1S3	90%	90%	100%	97%	No OMIM disease ID	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME
AP2M1	100%	100%	100%	98%	Intellectual developmental disorder 60 with seizures, 618587	INTELLECTUAL DISABILITY MENDELIOME
AP2S1	100%	100%	100%	94%	Hypocalciuric hypercalcemia, type III, 600740	INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME

AP3B1	100%	100%	100%	98%	Hermansky-Pudlak syndrome 2, 608233	VISION DISORDERS HEREDITARY BONE MARROW FAILURE SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
AP3B2	100%	100%	100%	98%	Developmental and epileptic encephalopathy 48, 617276	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
AP3D1	100%	100%	100%	99%	?Hermansky-Pudlak syndrome 10, 617050	VISION DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
AP4B1	100%	100%	100%	99%	Spastic paraplegia 47, autosomal recessive, 614066	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
AP4E1	100%	100%	100%	98%	Stuttering, familial persistent, 1, 184450 Spastic paraplegia 51, autosomal recessive, 613744	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
AP4M1	100%	100%	100%	98%	Spastic paraplegia 50, autosomal recessive, 612936	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
AP4S1	87%	87%	100%	99%	Spastic paraplegia 52, autosomal recessive, 614067	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
AP5Z1	100%	100%	100%	99%	Spastic paraplegia 48, autosomal recessive, 613647	MENDELIOME PRE CONCEPTION SCREENING

APC	100%	100%	100%	98%	Colorectal cancer, somatic, 114500 Brain tumor-polyposis syndrome 2, 175100 Desmoid disease, hereditary, 135290 Adenoma, periampullary, somatic, 175100 Hepatoblastoma, somatic, 114550 Gastric cancer, somatic, 613659 Gastric adenocarcinoma and proximal polyposis of the stomach, 619182 Gardner syndrome, 175100 Adenomatous polyposis coli, 175100	SKIN DISORDERS MENDELIOME TUMOR
APC2	100%	100%	100%	96%	Cortical dysplasia, complex, with other brain malformations 10, 618677 Intellectual developmental disorder, autosomal recessive 74, 617169	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
APCDD1	100%	100%	100%	99%	Hypotrichosis 1, 605389	SKIN DISORDERS MENDELIOME
APOA1	100%	100%	100%	99%	Hypoalphalipoproteinemia, primary, 2, 618463 Amyloidosis, 3 or more types, 105200 Hypoalphalipoproteinemia, primary, 2, intermediate, 619836	MENDELIOME
APOA2	100%	100%	100%	99%	Apolipoprotein A-II deficiency,	MENDELIOME
APOA5	100%	100%	100%	99%	Hyperchylomicronemia, late-onset, 144650	METABOLIC DISORDERS MENDELIOME
APOB	100%	100%	100%	98%	Hypercholesterolemia, familial, 2, 144010 Hypobetalipoproteinemia, 615558	MENDELIOME
APOC2	100%	100%	100%	97%	Hyperlipoproteinemia, type Ib, 207750	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
APOC3	100%	100%	100%	99%	Apolipoprotein C-III deficiency, 614028	MENDELIOME
APOE	100%	100%	100%	99%	Alzheimer disease 2, 104310 Sea-blue histiocyte disease, 269600 Lipoprotein glomerulopathy, 611771 Hyperlipoproteinemia, type III, 617347	 MENDELIOME PRE CONCEPTION SCREENING
APOL1	100%	100%	100%	98%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY RENAL DISORDERS MENDELIOME
APOO	100%	100%	98%	72%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
APP	100%	100%	100%	98%	Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants, 605714 Alzheimer disease 1, familial, 104300	MENDELIOME

APRT	100%	100%	100%	99%	Adenine phosphoribosyltransferase deficiency, 614723	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
APTX	100%	100%	100%	98%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920	MOVEMENT DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
AQP2	100%	100%	100%	99%	Diabetes insipidus, nephrogenic, 2, 125800	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
AQP5	100%	100%	100%	99%	Palmoplantar keratoderma, Bothnian type, 600231	SKIN DISORDERS MENDELIOME
AR	99%	99%	95%	66%	Androgen insensitivity, partial, w/wo breast cancer, 312300 Androgen insensitivity, 300068 Spinal and bulbar muscular atrophy of Kennedy, 313200 Hypospadias 1, X-linked, 300633	DISORDERS OF SEX DEVELOPMENT MALE INFERTILITY MENDELIOME
ARCN1	100%	100%	100%	99%	Short stature-micrognathia syndrome, 617164	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
ARF1	100%	100%	100%	99%	Periventricular nodular heterotopia 8, 618185	INTELLECTUAL DISABILITY MENDELIOME
ARF3	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
ARFGEF1	100%	100%	100%	97%	Developmental delay, impaired speech, and behavioral abnormalities, w/wo seizures, 619964	INTELLECTUAL DISABILITY MENDELIOME
ARFGEF2	100%	100%	100%	98%	Periventricular heterotopia with microcephaly, 608097	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ARG1	93%	93%	100%	98%	Argininemia, 207800	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ARHGAP24	100%	100%	99%	95%	No OMIM disease ID	RENAL DISORDERS MENDELIOME
ARHGAP26	99%	99%	100%	98%	Leukemia, juvenile myelomonocytic, somatic, 607785	MENDELIOME

ARHGAP29	100%	100%	100%	97%	No OMIM disease ID	CRANIOFACIAL ANOMALIES MENDELIOME SCHISIS
ARHGAP31	100%	100%	100%	98%	Adams-Oliver syndrome 1, 100300	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS
ARHGAP35	100%	100%	100%	99%	No OMIM disease ID	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM INTELLECTUAL DISABILITY MENDELIOME
ARHGDIA	100%	100%	100%	99%	Nephrotic syndrome, type 8, 615244	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ARHGEF1	100%	100%	100%	99%	?Immunodeficiency 62, 618459	PRIMARY IMMUNODEFICIENCY MENDELIOME
ARHGEF10	100%	100%	100%	99%	?Slowed nerve conduction velocity, AD, 608236	NEUROPATHIES MENDELIOME
ARHGEF18	100%	100%	100%	98%	Retinitis pigmentosa 78, 617433	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ARHGEF2	100%	100%	100%	98%	?Neurodevelopmental disorder with midbrain and hindbrain malformations, 617523	MENDELIOME PRE CONCEPTION SCREENING
ARHGEF28	100%	100%	100%	98%	No OMIM disease ID	NEUROPATHIES MENDELIOME
ARHGEF6	100%	100%	97%	69%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
ARHGEF9	96%	95%	98%	72%	Developmental and epileptic encephalopathy 8, 300607	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
ARID1A	100%	100%	99%	92%	Coffin-Siris syndrome 2, 614607	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME

ARID1B	98%	98%	98%	85%	Coffin-Siris syndrome 1, 135900	SKIN DISORDERS EPILEPSY SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
ARID2	100%	100%	100%	97%	Coffin-Siris syndrome 6, 617808	INTELLECTUAL DISABILITY MENDELIOME
ARIH1	100%	100%	100%	97%	No OMIM disease ID	ANEURYSM MENDELIOME
ARL13B	100%	100%	100%	97%	Joubert syndrome 8, 612291	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ARL2	100%	100%	100%	98%	?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 1, 619082	VISION DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS
ARL2BP	100%	100%	99%	97%	Retinitis pigmentosa w/wo situs inversus, 615434	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ARL3	100%	100%	100%	99%	Retinitis pigmentosa 83, 618173 Joubert syndrome 35, 618161	VISION DISORDERS CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING
ARL6	100%	100%	100%	95%	Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ARL6IP1	100%	100%	100%	98%	?Spastic paraplegia 61, autosomal recessive, 615685	MENDELIOME PRE CONCEPTION SCREENING
ARMC2	100%	100%	100%	96%	Spermatogenic failure 38, 618433	MALE INFERTILITY MENDELIOME
ARMC4	95%	95%	100%	98%	Ciliary dyskinesia, primary, 23, 615451	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING
ARMC5	100%	100%	100%	99%	ACTH-independent macronodular adrenal hyperplasia 2, 615954	DISORDERS OF SEX DEVELOPMENT MENDELIOME TUMOR

ARMC9	100%	100%	100%	99%	Joubert syndrome 30, 617622	CILIOPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ARNT2	100%	100%	100%	98%	?Webb-Dattani syndrome, 615926	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
ARPC1B	100%	100%	100%	99%	Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia, 617718	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
ARPC4	100%	100%	100%	98%	Developmental delay, language impairment, and ocular abnormalities, 620141	INTELLECTUAL DISABILITY MENDELIOME
ARR3	100%	100%	98%	71%	Myopia 26, X-linked, female-limited, 301010	VISION DISORDERS MENDELIOME
ARSA	100%	100%	100%	99%	Metachromatic leukodystrophy, 250100	MOVEMENT DISORDERS NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ARSB	100%	100%	100%	97%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ARSG	100%	100%	99%	98%	Usher syndrome, type IV, 618144	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
ARSK	100%	100%	100%	98%	Mucopolysaccharidosis, type X, 619698	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
ARSL	100%	100%	98%	72%	Chondrodysplasia punctata, X-linked recessive, 302950	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
ARV1	100%	100%	100%	98%	Developmental and epileptic encephalopathy 38, 617020	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

ARX	99%	96%	88%	50%	Proud syndrome, 300004 Hydranencephaly with abnormal genitalia, 300215 Partington syndrome, 309510 Developmental and epileptic encephalopathy 1, 308350 Lissencephaly, X-linked 2, 300215 Intellectual developmental disorder, X-linked 29, 300419	MOVEMENT DISORDERS DISORDERS OF SEX DEVELOPMENT EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
ASAH1	100%	100%	100%	97%	Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 Farber lipogranulomatosis, 228000	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ASB10	100%	100%	100%	99%	Glaucoma 1, open angle, F, 603383	VISION DISORDERS MENDELIOME
ASCC1	86%	86%	100%	98%	Spinal muscular atrophy with congenital bone fractures 2, 616867 Barrett esophagus/esophageal adenocarcinoma, 614266	FETAL AKINESIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
ASCL1	100%	100%	100%	96%	No OMIM disease ID	MENDELIOME
ASH1L	98%	98%	100%	98%	Intellectual developmental disorder, autosomal dominant 52, 617796	INTELLECTUAL DISABILITY MENDELIOME
ASIP	100%	100%	99%	97%	No OMIM disease ID	SKIN DISORDERS MENDELIOME
ASL	100%	100%	100%	99%	Argininosuccinic aciduria, 207900	SKIN DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ASNS	100%	100%	100%	98%	Asparagine synthetase deficiency, 615574	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ASPA	100%	100%	100%	98%	Canavan disease, 271900	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ASPH	99%	99%	100%	97%	Traboulsi syndrome, 601552	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING

ASPM	100%	99%	100%	98%	Microcephaly 5, primary, autosomal recessive, 608716	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ASPRV1	100%	100%	99%	98%	Ichthyosis, lamellar, autosomal dominant, 146750	SKIN DISORDERS MENDELIOME
ASPSCR1	100%	100%	100%	98%	Alveolar soft-part sarcoma, 606243	MENDELIOME
ASRGL1	100%	100%	100%	99%	No OMIM disease ID	VISION DISORDERS MENDELIOME
ASS1	100%	100%	100%	99%	Citrullinemia, 215700	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ASTL	100%	100%	100%	99%	?Oocyte maturation defect 11, 619643	MENDELIOME
ASXL1	100%	100%	100%	99%	Myelodysplastic syndrome, somatic, 614286 Bohring-Opitz syndrome, 605039	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS TUMOR
ASXL2	100%	100%	100%	97%	Shashi-Pena syndrome, 617190	INTELLECTUAL DISABILITY MENDELIOME
ASXL3	100%	100%	100%	97%	Bainbridge-Ropers syndrome, 615485	SKIN DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
ATAD1	100%	99%	100%	97%	Hyperekplexia 4, 618011	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
ATAD3A	100%	100%	99%	96%	Harel-Yoon syndrome, 617183 Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810	NEUROPATHIES PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
ATAD3B	100%	100%	99%	95%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS

ATCAY	100%	100%	100%	98%	Ataxia, cerebellar, Cayman type, 601238	MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ATF3	100%	100%	100%	96%	No OMIM disease ID	DISORDERS OF SEX DEVELOPMENT MENDELIOME
ATF6	100%	100%	100%	98%	Achromatopsia 7, 616517	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ATG4A	100%	100%	97%	71%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
ATG4D	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
ATG5	100%	100%	100%	97%	?Spinocerebellar ataxia, autosomal recessive 25, 617584	MENDELIOME PRE CONCEPTION SCREENING
ATG7	100%	100%	100%	99%	Spinocerebellar ataxia, autosomal recessive 31, 619422	INTELLECTUAL DISABILITY MENDELIOME
ATIC	100%	100%	100%	97%	AICA-ribosiduria due to ATIC deficiency, 608688	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ATL1	100%	100%	100%	97%	Spastic paraplegia 3A, autosomal dominant, 182600 Neuropathy, hereditary sensory, type ID, 613708	MOVEMENT DISORDERS NEUROPATHIES HNPD INTELLECTUAL DISABILITY MENDELIOME
ATL3	100%	100%	100%	97%	Neuropathy, hereditary sensory, type IF, 615632	NEUROPATHIES HNPD MENDELIOME
ATM	100%	100%	100%	98%	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic, T-cell prolymphocytic leukemia, somatic, Lymphoma, mantle cell, somatic,	MOVEMENT DISORDERS HEREDITARY BREAST AND OVARIAN CANCER PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING TUMOR
ATN1	100%	100%	99%	96%	Dentatorubral-pallidoluysian atrophy, 125370 Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME

ATOH1	100%	100%	100%	97%	?Deafness, autosomal dominant 89, 620284	HEARING IMPAIRMENT MENDELIOME
ATOH7	100%	100%	100%	97%	Persistent hyperplastic primary vitreous, autosomal recessive, 221900	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ATP11A	100%	100%	100%	99%	?Leukodystrophy, hypomyelinating, 24, 619851 Deafness, autosomal dominant 84, 619810	HEARING IMPAIRMENT MENDELIOME
ATP11C	100%	99%	97%	73%	?Hemolytic anemia, congenital, X-linked, 301015	MENDELIOME
ATP13A2	100%	100%	100%	99%	Spastic paraplegia 78, autosomal recessive, 617225 Kufor-Rakeb syndrome, 606693	MOVEMENT DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PARKINSON PRE CONCEPTION SCREENING
ATP1A1	100%	100%	100%	99%	Hypomagnesemia, seizures, and impaired intellectual development 2, 618314 Charcot-Marie-Tooth disease, axonal, type 2DD, 618036	EPILEPSY NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME
ATP1A2	100%	100%	100%	99%	Developmental and epileptic encephalopathy 98, 619605 Fetal akinesia, respiratory insufficiency, microcephaly, polymicrogyria, and dysmorphic facies, 619602 Alternating hemiplegia of childhood 1, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481	FETAL AKINESIA MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
ATP1A3	100%	100%	100%	98%	Alternating hemiplegia of childhood 2, 614820 Dystonia-12, 128235 CAPOS syndrome, 601338 Developmental and epileptic encephalopathy 99, 619606	MOVEMENT DISORDERS HEARING IMPAIRMENT EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PARKINSON
ATP2A1	100%	100%	100%	99%	Brody myopathy, 601003	MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
ATP2A2	100%	100%	100%	99%	Acrokeratosis verruciformis, 101900 Darier disease, 124200	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME
ATP2B1	100%	100%	100%	98%	Intellectual developmental disorder, autosomal dominant 66, 619910	INTELLECTUAL DISABILITY MENDELIOME

ATP2B2	100%	100%	100%	98%	Deafness, autosomal dominant 82, 619804	HEARING IMPAIRMENT MENDELIOME
ATP2B3	100%	99%	98%	74%	?Spinocerebellar ataxia, X-linked 1, 302500	MOVEMENT DISORDERS MENDELIOME
ATP2C1	100%	99%	100%	98%	Hailey-Hailey disease, 169600	SKIN DISORDERS MENDELIOME
ATP4A	100%	100%	100%	98%	No OMIM disease ID	IRON DISORDERS MENDELIOME
ATP5F1A	100%	100%	100%	99%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4, 615228 ?Combined oxidative phosphorylation deficiency 22, 616045	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
ATP5F1B	100%	100%	100%	99%	?Hypermetabolism due to uncoupled mitochondrial oxidative phosphorylation 2, 620085	MENDELIOME MITOCHONDRIAL DISORDERS
ATP5F1C	100%	100%	100%	97%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
ATP5F1D	100%	100%	100%	97%	Mitochondrial complex V (ATP synthase) deficiency, 618120	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
ATP5F1E	100%	100%	100%	96%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
ATP5IF1	100%	100%	100%	98%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
ATP5MC1	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
ATP5MC2	100%	100%	100%	98%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
ATP5MC3	100%	100%	100%	99%	Dystonia, early-onset, and/or spastic paraplegia, 619681	MENDELIOME MITOCHONDRIAL DISORDERS
ATP5MD	100%	100%	100%	97%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 6, 618683	MENDELIOME MITOCHONDRIAL DISORDERS
ATP5ME	100%	100%	100%	98%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
ATP5MF	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
ATP5MG	100%	100%	100%	98%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
ATP5MGL	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
ATP5PB	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS

ATP5PD	100%	100%	100%	98%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
ATP5PF	100%	100%	100%	97%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
ATP5PO	100%	100%	100%	98%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
ATP6AP1	100%	99%	98%	73%	Immunodeficiency 47, 300972	PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME
ATP6AP2	100%	100%	97%	71%	Intellectual developmental disorder, X-linked syndromic, Hedera type, 300423 ?Parkinsonism with spasticity, X-linked, 300911 Congenital disorder of glycosylation, type IIr, 301045	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME
ATP6V0A1	100%	100%	100%	98%	Neurodevelopmental disorder with epilepsy and brain atrophy, 619971 Developmental and epileptic encephalopathy 104, 619970	INTELLECTUAL DISABILITY MENDELIOME
ATP6V0A2	100%	100%	100%	97%	Wrinkly skin syndrome, 278250 Cutis laxa, autosomal recessive, type IIA, 219200	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ATP6V0A4	100%	100%	100%	98%	Distal renal tubular acidosis 3, w/wo sensorineural hearing loss, 602722	HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ATP6V0C	100%	100%	100%	98%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
ATP6V1A	100%	100%	100%	97%	Cutis laxa, autosomal recessive, type IID, 617403 Developmental and epileptic encephalopathy 93, 618012	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ATP6V1B1	100%	100%	100%	99%	Distal renal tubular acidosis 2 with progressive sensorineural hearing loss, 267300	HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ATP6V1B2	100%	100%	100%	98%	Zimmermann-Laband syndrome 2, 616455 Deafness, congenital, with onychodystrophy, autosomal dominant, 124480	HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME

ATP6V1E1	100%	100%	100%	98%	Cutis laxa, autosomal recessive, type IIC, 617402	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ATP7A	100%	100%	98%	72%	Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489 Menkes disease, 309400	SKIN DISORDERS EPILEPSY NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MUSCLE DISORDERS
ATP7B	100%	100%	100%	99%	Wilson disease, 277900	MOVEMENT DISORDERS LIVER DISORDERS METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ATP8A2	100%	100%	100%	98%	?Cerebellar ataxia, impaired intellectual development, and dysequilibrium syndrome 4, 615268	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ATP8B1	100%	100%	100%	97%	Cholestasis, progressive familial intrahepatic 1, 211600 Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, benign recurrent intrahepatic, 243300	LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ATP9A	100%	100%	100%	98%	Neurodevelopmental disorder with poor growth and behavioral abnormalities, 620242	INTELLECTUAL DISABILITY MENDELIOME
ATPAF1	100%	100%	99%	92%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
ATPAF2	100%	100%	100%	99%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273	HEART MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
ATR	100%	100%	100%	98%	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING TUMOR

ATRX	99%	99%	96%	66%	Alpha-thalassemia/mental retardation syndrome, 301040 Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Intellectual disability-hypotonic facies syndrome, X-linked, 309580	DISORDERS OF SEX DEVELOPMENT EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
ATXN1	100%	100%	100%	99%	Spinocerebellar ataxia 1, 164400	MENDELIOME
ATXN10	100%	100%	100%	97%	Spinocerebellar ataxia 10, 603516	MENDELIOME
ATXN2	100%	100%	99%	94%	Spinocerebellar ataxia 2, 183090	MENDELIOME
ATXN2L	100%	100%	100%	97%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
ATXN3	93%	93%	99%	98%	Machado-Joseph disease, 109150	MENDELIOME
ATXN7	100%	100%	99%	94%	Spinocerebellar ataxia 7, 164500	MENDELIOME
ATXN8OS	NC	NC	NC	NC	Spinocerebellar ataxia 8, 608768	MENDELIOME
AUH	100%	100%	100%	97%	3-methylglutaconic aciduria, type I, 250950	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
AURKC	100%	100%	100%	98%	Spermatogenic failure 5, 243060	MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING
AUTS2	100%	100%	100%	98%	Intellectual developmental disorder, autosomal dominant 26, 615834	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
AVIL	100%	100%	100%	99%	Nephrotic syndrome, type 21, 618594	RENAL DISORDERS MENDELIOME
AVP	100%	100%	100%	97%	Diabetes insipidus, neurohypophyseal, 125700	RENAL DISORDERS MENDELIOME
AVPR2	100%	100%	98%	78%	Diabetes insipidus, nephrogenic, 1, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539	INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME
AXIN1	100%	100%	100%	99%	Hepatocellular carcinoma, somatic, 114550 ?Caudal duplication anomaly, 607864	MENDELIOME
AXIN2	100%	100%	100%	99%	Colorectal cancer, somatic, 114500 Oligodontia-colorectal cancer syndrome, 608615	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME TUMOR
AXL	100%	100%	100%	98%	No OMIM disease ID	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME

B2M	100%	100%	100%	98%	?Amyloidosis, familial visceral, 105200 Immunodeficiency 43, 241600	NEUROPATHIES PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)
B3GALNT1	100%	100%	100%	97%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME
B3GALNT2	92%	92%	100%	97%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
B3GALT6	99%	98%	100%	94%	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, w/wo fractures, 271640 Al-Gazali syndrome, 609465	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS
B3GAT3	94%	93%	100%	98%	Multiple joint dislocations, short stature, craniofacial dysmorphism, w/wo congenital heart defects, 245600	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
B3GLCT	100%	100%	100%	98%	Peters-plus syndrome, 261540	VISION DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS
B4GALNT1	100%	100%	100%	99%	Spastic paraplegia 26, autosomal recessive, 609195	MOVEMENT DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
B4GALNT2	100%	100%	100%	98%	Sd(a) polyagglutination syndrome, 615018	MENDELIOME
B4GALT1	100%	100%	100%	98%	Congenital disorder of glycosylation, type IId, 607091	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

B4GALT7	100%	100%	100%	99%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS
B4GAT1	100%	100%	100%	98%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
B9D1	100%	100%	100%	99%	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120	CILIOPATHIES DISORDERS OF SEX DEVELOPMENT INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
B9D2	100%	100%	100%	99%	?Meckel syndrome 10, 614175 Joubert syndrome 34, 614175	CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS
BAAT	100%	100%	100%	99%	Bile acid conjugation defect 1, 619232	LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
BACH2	100%	100%	100%	99%	Immunodeficiency 60 and autoimmunity, 618394	PRIMARY IMMUNODEFICIENCY MENDELIOME
BAG3	100%	100%	100%	98%	Cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954	DILATED CARDIOMYOPATHY HEART NEUROPATHIES MENDELIOME MUSCLE DISORDERS
BAG5	100%	100%	100%	98%	Cardiomyopathy, dilated, 2F, 619747	MENDELIOME

BANF1	100%	100%	100%	97%	Nestor-Guillermo progeria syndrome, 614008	SKIN DISORDERS HEART SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
BAP1	100%	100%	100%	99%	Kury-Isidor syndrome, 619762 Tumor predisposition syndrome 1, 614327	SKIN DISORDERS MELA2 INTELLECTUAL DISABILITY MENDELIOME TUMOR
BARD1	100%	100%	100%	98%	No OMIM disease ID	HEREDITARY BREAST AND OVARIAN CANCER MENDELIOME TUMOR
BAX	100%	100%	100%	98%	Colorectal cancer, somatic, 114500 T-cell acute lymphoblastic leukemia, somatic, 613065	MENDELIOME
BAZ2B	100%	100%	100%	97%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
BBIP1	100%	100%	100%	98%	?Bardet-Biedl syndrome 18, 615995	VISION DISORDERS CILIOPATHIES RENAL DISORDERS MENDELIOME
BBS1	100%	100%	100%	99%	Bardet-Biedl syndrome 1, 209900	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
BBS10	100%	100%	100%	98%	Bardet-Biedl syndrome 10, 615987	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING

BBS12	100%	100%	100%	99%	Bardet-Biedl syndrome 12, 615989	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
BBS2	100%	100%	100%	98%	Retinitis pigmentosa 74, 616562 Bardet-Biedl syndrome 2, 615981	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
BBS4	100%	100%	100%	98%	Bardet-Biedl syndrome 4, 615982	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
BBS5	100%	100%	100%	98%	Bardet-Biedl syndrome 5, 615983	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
BBS7	100%	100%	100%	99%	Bardet-Biedl syndrome 7, 615984	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
BBS9	95%	95%	100%	98%	Bardet-Biedl syndrome 9, 615986	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
BCAP31	99%	92%	98%	69%	Deafness, dystonia, and cerebral hypomyelination, 300475	MOVEMENT DISORDERS HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS

BCAS3	100%	100%	100%	98%	Hengel-Maroofian-Schols syndrome, 619641	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
BCAT1	100%	100%	100%	99%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME
BCAT2	100%	100%	100%	99%	?Hypervalinemia or hyperleucine-isoleucinemia, 618850	METABOLIC DISORDERS MENDELIOME
BCHE	100%	100%	100%	98%	Butyrylcholinesterase deficiency, 617936	MENDELIOME
BCKDHA	100%	100%	100%	99%	Maple syrup urine disease, type Ia, 248600	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
BCKDHB	100%	99%	100%	97%	Maple syrup urine disease, type Ib, 248600	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
BCKDK	100%	100%	100%	99%	Branched-chain keto acid dehydrogenase kinase deficiency, 614923	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
BCL10	100%	100%	100%	99%	?Immunodeficiency 37, 616098 Lymphoma, MALT, somatic, 137245	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
BCL11A	100%	100%	100%	99%	Dias-Logan syndrome, 617101	INTELLECTUAL DISABILITY MENDELIOME
BCL11B	99%	99%	99%	97%	Immunodeficiency 49, severe combined, 617237 Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092	MOVEMENT DISORDERS PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME
BCL2	100%	100%	100%	95%	Leukemia/lymphoma, B-cell, 2,	MENDELIOME
BCL7A	100%	100%	97%	89%	B-cell non-Hodgkin lymphoma, high-grade,	MENDELIOME
BCO1	100%	100%	100%	99%	?Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300	METABOLIC DISORDERS MENDELIOME

BCOR	100%	99%	98%	74%	Microphthalmia, syndromic 2, 300166	VISION DISORDERS CRANIOFACIAL ANOMALIES SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME SCHISIS
BCORL1	100%	99%	97%	70%	Shukla-Vernon syndrome, 301029	INTELLECTUAL DISABILITY MENDELIOME
BCS1L	100%	100%	100%	99%	GRACILE syndrome, 603358 Mitochondrial complex III deficiency, nuclear type 1, 124000 Bjornstad syndrome, 262000	SKIN DISORDERS HEARING IMPAIRMENT LIVER DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
BDP1	100%	100%	100%	97%	?Deafness, autosomal recessive 112, 618257	HEARING IMPAIRMENT MENDELIOME
BEAN1	91%	91%	100%	99%	Spinocerebellar ataxia 31, 117210	MENDELIOME
BEST1	100%	100%	100%	99%	Macular dystrophy, vitelliform, 2, 153700 ?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 2, 193220 Retinitis pigmentosa-50, 613194 Retinitis pigmentosa, concentric, 613194 Vitreoretinochoroidopathy, 193220 Bestrophinopathy, autosomal recessive, 611809	VISION DISORDERS MENDELIOME
BFSP1	100%	100%	100%	98%	Cataract 33, multiple types, 611391	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
BFSP2	100%	100%	100%	99%	Cataract 12, multiple types, 611597	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
BGN	100%	99%	98%	74%	Meester-Loeys syndrome, 300989 Spondyloepimetaphyseal dysplasia, X-linked, 300106	ANEURYSM SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
BHLHA9	100%	100%	100%	96%	?Camptosynpolydactyly, complex, 607539 Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
BICC1	100%	99%	100%	99%	No OMIM disease ID	RENAL DISORDERS MENDELIOME

BICD2	100%	100%	100%	99%	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291 Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290	FETAL AKINESIA HEART NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME MUSCLE DISORDERS
BICRA	100%	100%	99%	95%	Coffin-Siris syndrome 12, 619325	INTELLECTUAL DISABILITY MENDELIOME
BIN1	100%	100%	100%	98%	Centronuclear myopathy 2, 255200	FETAL AKINESIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
BLK	100%	100%	100%	99%	Maturity-onset diabetes of the young, type 11, 613375	PRIMARY IMMUNODEFICIENCY MENDELIOME
BLM	100%	100%	100%	98%	Bloom syndrome, 210900	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING TUMOR
BLNK	100%	100%	100%	98%	?Agammaglobulinemia 4, 613502	PRIMARY IMMUNODEFICIENCY MENDELIOME
BLOC1S1	100%	100%	100%	98%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
BLOC1S3	100%	100%	100%	95%	Hermansky-Pudlak syndrome 8, 614077	VISION DISORDERS SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
BLOC1S5	100%	100%	100%	98%	Hermansky-Pudlak syndrome 11, 619172	VISION DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME
BLOC1S6	100%	100%	100%	98%	?Hermansky-Pudlak syndrome 9, 614171	VISION DISORDERS SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING

BLVRA	100%	99%	100%	98%	Hyperbiliverdinemia, 614156	LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
BMP1	100%	100%	100%	99%	Osteogenesis imperfecta, type XIII, 614856	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
BMP15	100%	100%	99%	75%	Premature ovarian failure 4, 300510 Ovarian dysgenesis 2, 300510	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRIMARY OVARIAN INSUFFICIENCY
BMP2	100%	100%	100%	98%	Short stature, facial dysmorphism, and skeletal anomalies w/wo cardiac anomalies 1, 617877 Brachydactyly, type A2, 112600	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME SCHISIS
BMP4	100%	100%	100%	99%	Orofacial cleft 11, 600625 Microphthalmia, syndromic 6, 607932	VISION DISORDERS CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT DISORDERS OF SEX DEVELOPMENT MENDELIOME
BMP6	100%	100%	99%	95%	No OMIM disease ID	IRON DISORDERS MENDELIOME
BMP7	100%	100%	100%	99%	No OMIM disease ID	DISORDERS OF SEX DEVELOPMENT MENDELIOME
BMPER	100%	100%	100%	98%	Diaphanospondylodysostosis, 608022	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS
BMPR1A	100%	100%	100%	98%	Polyposis syndrome, hereditary mixed, 2, 610069 Polyposis, juvenile intestinal, 174900	MENDELIOME TUMOR
BMPR1B	100%	100%	100%	98%	Acromesomelic dysplasia 3, 609441 Brachydactyly, type A2, 112600 Brachydactyly, type A1, D, 616849	VISION DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
BMPR2	100%	99%	100%	99%	Pulmonary hypertension, familial primary, 1, w/wo HHT, 178600 Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600 Pulmonary venoocclusive disease 1, 265450	HEART MENDELIOME

BMS1	100%	100%	100%	97%	?Aplasia cutis congenita, nonsyndromic, 107600	SKIN DISORDERS MENDELIOME
BNC1	100%	99%	100%	98%	?Premature ovarian failure 16, 618723	MENDELIOME
BNC2	100%	100%	100%	99%	Lower urinary tract obstruction, congenital, 618612	MENDELIOME
BOLA1	100%	100%	100%	100%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
BOLA2	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
BOLA3	100%	100%	100%	97%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
BPGM	100%	100%	100%	98%	Erythrocytosis, familial, 8, 222800	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
BPTF	100%	100%	99%	96%	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, 617755	INTELLECTUAL DISABILITY MENDELIOME
BPY2	50%	49%	47%	23%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
BPY2B	50%	48%	48%	24%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
BPY2C	50%	49%	47%	23%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
BRAF	100%	100%	99%	96%	Melanoma, malignant, somatic, 155600 LEOPARD syndrome 3, 613707 Cardiofaciocutaneous syndrome, 115150 Adenocarcinoma of lung, somatic, 211980 Noonan syndrome 7, 613706 Colorectal cancer, somatic, 114500 Nonsmall cell lung cancer, somatic, 211980	HEREDITARY BONE MARROW FAILURE CONGENITAL HEART DISEASE SKIN DISORDERS HEART HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME AND RASOPATHY TUMOR
BRAT1	100%	100%	100%	99%	Neurodevelopmental disorder with cerebellar atrophy and w/wo seizures, 618056 Rigidity and multifocal seizure syndrome, lethal neonatal, 614498	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

BRCA1	100%	100%	100%	98%	Fanconi anemia, complementation group S, 617883	HEREDITARY BONE MARROW FAILURE HEREDITARY BREAST AND OVARIAN CANCER MENDELIOME PRE CONCEPTION SCREENING TUMOR
BRCA2	100%	100%	100%	97%	Fanconi anemia, complementation group D1, 605724 Wilms tumor, 194070	HEREDITARY BONE MARROW FAILURE HEREDITARY BREAST AND OVARIAN CANCER MENDELIOME PRE CONCEPTION SCREENING SONIC HEDGEHOG MEDULLOBLASTOMA TUMOR
BRDT	100%	100%	100%	97%	?Spermatogenic failure 21, 617644	MENDELIOME
BRF1	100%	100%	100%	99%	Cerebellofaciodental syndrome, 616202	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
BRIP1	100%	100%	100%	97%	Fanconi anemia, complementation group J, 609054	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS HEREDITARY BREAST AND OVARIAN CANCER MENDELIOME PRE CONCEPTION SCREENING TUMOR
BRPF1	100%	99%	100%	99%	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333	INTELLECTUAL DISABILITY MENDELIOME
BRSK2	100%	100%	99%	97%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
BRWD3	100%	99%	97%	72%	Intellectual developmental disorder, X-linked 93, 300659	INTELLECTUAL DISABILITY MENDELIOME

BSCL2	100%	100%	100%	99%	Lipodystrophy, congenital generalized, type 2, 269700 Neuropathy, distal hereditary motor, type VC, 619112 Silver spastic paraplegia syndrome, 270685 Encephalopathy, progressive, w/wo lipodystrophy, 615924	MOVEMENT DISORDERS SKIN DISORDERS HEART NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
BSND	100%	100%	100%	99%	Sensorineural deafness with mild renal dysfunction, 602522 Bartter syndrome, type 4a, 602522	HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
BTD	94%	94%	100%	99%	Biotinidase deficiency, 253260	MOVEMENT DISORDERS SKIN DISORDERS HEARING IMPAIRMENT EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
BTG4	100%	100%	99%	97%	Oocyte maturation defect 8, 619009	MENDELIOME
BTK	100%	99%	98%	72%	Agammaglobulinemia, X-linked 1, 300755 Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200	PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
BTRC	100%	100%	100%	99%	No OMIM disease ID	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
BUB1	100%	100%	100%	98%	Colorectal cancer with chromosomal instability, somatic, 114500 Microcephaly 30, primary, autosomal recessive, 620183	INTELLECTUAL DISABILITY MENDELIOME TUMOR
BUB1B	100%	100%	100%	98%	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING TUMOR
BUB3	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME TUMOR

BVES	100%	100%	100%	98%	Muscular dystrophy, limb-girdle, autosomal recessive 25, 616812	HEART MENDELIOME PRE CONCEPTION SCREENING
C11orf80	92%	92%	100%	97%	Hydatidiform mole, recurrent, 4, 618432	MENDELIOME
C12orf4	100%	100%	100%	97%	Intellectual developmental disorder, autosomal recessive 66, 618221	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
C12orf57	100%	100%	100%	97%	Temtamy syndrome, 218340	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
C12orf65	100%	100%	99%	97%	Spastic paraplegia 55, autosomal recessive, 615035 Combined oxidative phosphorylation deficiency 7, 613559	MOVEMENT DISORDERS VISION DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
C14orf39	100%	100%	100%	96%	Spermatogenic failure 52, 619202 ?Premature ovarian failure 18, 619203	MALE INFERTILITY MENDELIOME
C15orf41	100%	99%	100%	99%	Dyserythropoietic anemia, congenital, type Ib, 615631	HEREDITARY BONE MARROW FAILURE IRON DISORDERS MENDELIOME PRE CONCEPTION SCREENING
C19orf12	100%	99%	99%	95%	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043	MOVEMENT DISORDERS VISION DISORDERS NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS PARKINSON PRE CONCEPTION SCREENING
C1GALT1C1	100%	100%	98%	72%	Tn polyagglutination syndrome, somatic, 300622	METABOLIC DISORDERS MENDELIOME
C1QA	100%	100%	100%	99%	C1q deficiency, 613652	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
C1QB	100%	100%	99%	94%	C1q deficiency, 613652	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING

C1QBP	100%	100%	100%	98%	Combined oxidative phosphorylation deficiency 33, 617713	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
C1QC	100%	100%	100%	97%	C1q deficiency, 613652	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
C1QTNF5	100%	100%	99%	96%	Retinal degeneration, late-onset, autosomal dominant, 605670	VISION DISORDERS MENDELIOME
C1R	99%	98%	100%	99%	Ehlers-Danlos syndrome, periodontal type, 1, 130080	PRIMARY IMMUNODEFICIENCY MENDELIOME
C1S	99%	99%	100%	98%	C1s deficiency, 613783 Ehlers-Danlos syndrome, periodontal type, 2, 617174	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
C1orf194	100%	100%	99%	98%	No OMIM disease ID	NEUROPATHIES MENDELIOME
C2	100%	100%	100%	98%	C2 deficiency, 217000	PRIMARY IMMUNODEFICIENCY MENDELIOME
C2CD3	96%	96%	100%	98%	Orofaciodigital syndrome XIV, 615948	CILIOPATHIES SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS
C2CD6	100%	100%	99%	94%	?Spermatogenic failure 68, 619805	MENDELIOME
C2orf69	100%	100%	99%	96%	Combined oxidative phosphorylation deficiency 53, 619423	PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
C3	100%	100%	100%	99%	C3 deficiency, 613779	AGE RELATED MACULAR DEGENERATION HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
C4A	99%	99%	99%	92%	C4a deficiency, 614380	MENDELIOME PRE CONCEPTION SCREENING
C4B	100%	99%	99%	92%	C4B deficiency, 614379	MENDELIOME PRE CONCEPTION SCREENING

C5	100%	100%	100%	98%	C5 deficiency, 609536	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
C6	100%	99%	100%	98%	C6 deficiency, 612446 Combined C6/C7 deficiency,	PRIMARY IMMUNODEFICIENCY MENDELIOME
C7	99%	98%	100%	98%	C7 deficiency, 610102	PRIMARY IMMUNODEFICIENCY MENDELIOME
C8A	100%	100%	100%	98%	C8 deficiency, type I, 613790	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
C8B	100%	100%	100%	98%	C8 deficiency, type II, 613789	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
C8G	100%	100%	100%	99%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
C8orf37	100%	100%	100%	98%	Retinitis pigmentosa 64, 614500 Cone-rod dystrophy 16, 614500 Bardet-Biedl syndrome 21, 617406	VISION DISORDERS CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING
C9	100%	100%	100%	97%	C9 deficiency, 613825	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
C9orf72	100%	100%	100%	98%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1, 105550	MENDELIOME
CA12	100%	100%	100%	98%	Hyperchlorhidrosis, isolated, 143860	MENDELIOME PRE CONCEPTION SCREENING
CA2	100%	100%	100%	98%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CA4	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME
CA5A	100%	100%	100%	98%	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING

CA8	100%	100%	100%	99%	Cerebellar ataxia, impaired intellectual development and dysequilibrium syndrome 3, 613227	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CABIN1	100%	100%	100%	99%	No OMIM disease ID	HNPD MENDELIOME
CABP2	100%	100%	100%	99%	Deafness, autosomal recessive 93, 614899	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
CABP4	100%	100%	100%	99%	Cone-rod synaptic disorder, congenital nonprogressive, 610427	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CACNA1A	100%	100%	100%	97%	Spinocerebellar ataxia 6, 183086 Episodic ataxia, type 2, 108500 Developmental and epileptic encephalopathy 42, 617106 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Migraine, familial hemiplegic, 1, 141500	MOVEMENT DISORDERS EPILEPSY HNPD INTELLECTUAL DISABILITY MENDELIOME
CACNA1B	100%	100%	100%	98%	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CACNA1C	100%	100%	100%	99%	Timothy syndrome, 601005 Long QT syndrome 8, 618447 Neurodevelopmental disorder with hypotonia, language delay, and skeletal defects w/wo seizures, 620029 Brugada syndrome 3, 611875	CONGENITAL HEART DISEASE HEART LONG QT SYNDROME INTELLECTUAL DISABILITY MENDELIOME ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS
CACNA1D	100%	100%	100%	98%	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896	HEARING IMPAIRMENT HEART INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CACNA1E	100%	100%	100%	99%	Developmental and epileptic encephalopathy 69, 618285	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
CACNA1F	100%	100%	97%	70%	Cone-rod dystrophy, X-linked, 3, 300476 Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071 Aland Island eye disease, 300600	VISION DISORDERS MENDELIOME
CACNA1G	100%	100%	100%	98%	Spinocerebellar ataxia 42, 616795 Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME

CACNA1H	100%	100%	99%	97%	Hyperaldosteronism, familial, type IV, 617027	HNPD RENAL DISORDERS MENDELIOME
CACNA1I	100%	100%	100%	97%	Neurodevelopmental disorder with speech impairment and w/wo seizures, 620114	INTELLECTUAL DISABILITY MENDELIOME
CACNA1S	100%	100%	100%	99%	Congenital myopathy 18 due to dihydropyridine receptor defect, 620246 Hypokalemic periodic paralysis, type 1, 170400	FETAL AKINESIA MENDELIOME MUSCLE DISORDERS
CACNA2D1	100%	100%	100%	97%	Developmental and epileptic encephalopathy 110, 620149	HEART INTELLECTUAL DISABILITY MENDELIOME
CACNA2D2	100%	100%	100%	97%	Cerebellar atrophy with seizures and variable developmental delay, 618501	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CACNA2D4	100%	100%	100%	99%	Retinal cone dystrophy 4, 610478	VISION DISORDERS MENDELIOME
CACNB2	100%	100%	100%	97%	Brugada syndrome 4, 611876	HEART MENDELIOME
CACNB4	100%	100%	100%	98%	Episodic ataxia, type 5, 613855	MOVEMENT DISORDERS EPILEPSY MENDELIOME
CACNG2	100%	100%	100%	99%	?Intellectual developmental disorder, autosomal dominant 10, 614256	MENDELIOME
CAD	100%	100%	100%	99%	Developmental and epileptic encephalopathy 50, 616457	HEREDITARY BONE MARROW FAILURE EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CADM3	100%	100%	100%	98%	Charcot-Marie-Tooth disease, axonal, type 2FF, 619519	NEUROPATHIES MENDELIOME
CALCRL	100%	100%	100%	98%	?Lymphatic malformation 8, 618773	MENDELIOME
CALM1	100%	100%	100%	99%	Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916 Long QT syndrome 14, 616247	HEART LONG QT SYNDROME MENDELIOME ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS

CALM2	73%	73%	100%	97%	Long QT syndrome 15, 616249	HEART LONG QT SYNDROME MENDELIOME ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS
CALM3	100%	100%	100%	98%	Long QT syndrome 16, 618782 ?Ventricular tachycardia, catecholaminergic polymorphic 6, 618782	HEART LONG QT SYNDROME MENDELIOME ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS
CALR	100%	100%	100%	99%	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950	HEMOSTATIC/THROMBOTIC DISORDERS IRON DISORDERS MENDELIOME
CAMK2A	100%	100%	100%	99%	Intellectual developmental disorder, autosomal dominant 53, 617798 ?Intellectual developmental disorder, autosomal recessive 63, 618095	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CAMK2B	100%	100%	100%	97%	Intellectual developmental disorder, autosomal dominant 54, 617799	INTELLECTUAL DISABILITY MENDELIOME
CAMK2G	100%	100%	100%	98%	Intellectual developmental disorder, autosomal dominant 59, 618522	INTELLECTUAL DISABILITY MENDELIOME
CAMK4	99%	99%	100%	98%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
CAMSAP1	100%	100%	99%	97%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
CAMTA1	100%	100%	99%	98%	Cerebellar dysfunction with variable cognitive and behavioral abnormalities, 614756	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME
CANT1	100%	100%	100%	99%	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CAPN1	100%	100%	100%	99%	Spastic paraplegia 76, autosomal recessive, 616907	MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CAPN10	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME PRE CONCEPTION SCREENING
CAPN12	100%	100%	99%	94%	No OMIM disease ID	SKIN DISORDERS MENDELIOME
CAPN15	100%	100%	100%	99%	Oculogastrointestinal neurodevelopmental syndrome, 619318	INTELLECTUAL DISABILITY MENDELIOME

CAPN3	100%	100%	100%	98%	Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600 Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129	MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
CAPN5	100%	100%	100%	99%	Vitreoretinopathy, neovascular inflammatory, 193235	VISION DISORDERS MENDELIOME
CARD10	100%	100%	99%	96%	?Immunodeficiency 89 and autoimmunity, 619632	PRIMARY IMMUNODEFICIENCY MENDELIOME
CARD11	100%	100%	100%	99%	B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11B with atopic dermatitis, 617638 Immunodeficiency 11A, 615206	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING TUMOR
CARD14	100%	100%	100%	99%	Psoriasis 2, 602723 Pityriasis rubra pilaris, 173200	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME
CARD8	100%	100%	100%	98%	?Inflammatory bowel disease (Crohn disease) 30, 619079	MENDELIOME
CARD9	100%	100%	100%	99%	Immunodeficiency 103, susceptibility to fungal infection, 212050	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
CARMIL2	100%	100%	100%	98%	Immunodeficiency 58, 618131	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME
CARS1	100%	100%	100%	99%	Microcephaly, developmental delay, and brittle hair syndrome, 618891	INTELLECTUAL DISABILITY MENDELIOME
CARS2	100%	100%	100%	99%	Combined oxidative phosphorylation deficiency 27, 616672	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
CASK	100%	100%	97%	72%	Intellectual developmental disorder, w/wo nystagmus, 300422 Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia, 300749 FG syndrome 4, 300422	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
CASP10	100%	100%	100%	98%	Autoimmune lymphoproliferative syndrome, type II, 603909 Gastric cancer, somatic, 613659 Lymphoma, non-Hodgkin, somatic, 605027	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCY MENDELIOME
CASP14	100%	100%	100%	97%	Ichthyosis, congenital, autosomal recessive 12, 617320	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CASP8	95%	95%	100%	98%	?Caspase 8 lymphadenopathy syndrome, 607271 Hepatocellular carcinoma, somatic, 114550	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING

CASQ1	100%	100%	100%	99%	Myopathy, vacuolar, with CASQ1 aggregates, 616231	MENDELIOME MUSCLE DISORDERS
CASQ2	100%	100%	100%	98%	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938	EPILEPSY HEART MENDELIOME PRE CONCEPTION SCREENING ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS
CASR	100%	100%	100%	98%	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 Hyperparathyroidism, neonatal, 239200 Hypocalcemia, autosomal dominant, 601198 Hypocalciuric hypercalcemia, type I, 145980	SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CAST	100%	100%	99%	98%	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CASZ1	99%	98%	99%	96%	No OMIM disease ID	HEART MENDELIOME
CAT	100%	100%	100%	98%	Acatalasemia, 614097	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CATIP	100%	100%	100%	97%	?Spermatogenic failure 54, 619379	MALE INFERTILITY MENDELIOME
CATSPER1	100%	100%	100%	97%	Spermatogenic failure 7, 612997	MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING
CATSPER2	100%	100%	100%	99%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
CAV1	100%	100%	100%	99%	Lipodystrophy, congenital generalized, type 3, 612526 Pulmonary hypertension, primary, 3, 615343 Lipodystrophy, familial partial, type 7, 606721	SKIN DISORDERS HEART METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CAV3	100%	100%	100%	99%	Myopathy, distal, Tateyama type, 614321 Creatine phosphokinase, elevated serum, 123320 Cardiomyopathy, familial hypertrophic, 192600 Rippling muscle disease 2, 606072 Long QT syndrome 9, 611818	HEART MENDELIOME MUSCLE DISORDERS

CAVIN1	100%	100%	100%	98%	Lipodystrophy, congenital generalized, type 4, 613327	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
CBFB	100%	100%	100%	94%	Cleidocranial dysplasia 2, 620099	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
CBL	100%	100%	100%	98%	Noonan syndrome-like disorder w/wo juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME AND RASOPATHY TUMOR
CBLIF	100%	100%	100%	99%	Intrinsic factor deficiency, 261000	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CBS	100%	100%	100%	99%	Thrombosis, hyperhomocysteinemic, 236200 Homocystinuria, B6-responsive and nonresponsive types, 236200	ANEURYSM VISION DISORDERS SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CBWD1	99%	97%	97%	92%	No OMIM disease ID	RENAL DISORDERS MENDELIOME
CBX2	100%	100%	100%	96%	?46XY sex reversal 5, 613080	DISORDERS OF SEX DEVELOPMENT MENDELIOME
CBY1	100%	100%	100%	99%	No OMIM disease ID	CILIOPATHIES MENDELIOME
CC2D1A	100%	100%	100%	98%	Intellectual developmental disorder, autosomal recessive 3, 608443	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

CC2D2A	98%	98%	100%	98%	COACH syndrome 2, 619111 Retinitis pigmentosa 93, 619845 Meckel syndrome 6, 612284 Joubert syndrome 9, 612285	MOVEMENT DISORDERS VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS
CCBE1	100%	100%	100%	99%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510	CRANIOFACIAL ANOMALIES SKIN DISORDERS PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CCDC103	100%	100%	100%	99%	Ciliary dyskinesia, primary, 17, 614679	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING
CCDC114	100%	100%	100%	98%	Ciliary dyskinesia, primary, 20, 615067	CONGENITAL HEART DISEASE CILIOPATHIES HEART MENDELIOME PRE CONCEPTION SCREENING
CCDC115	100%	100%	100%	96%	Congenital disorder of glycosylation, type IIo, 616828	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CCDC134	100%	100%	100%	98%	Osteogenesis imperfecta, type XXII, 619795	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
CCDC141	99%	98%	100%	98%	No OMIM disease ID	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME
CCDC151	100%	100%	100%	99%	Ciliary dyskinesia, primary, 30, 616037	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING
CCDC155	100%	100%	100%	98%	No OMIM disease ID	MALE INFERTILITY MENDELIOME

CCDC174	100%	100%	100%	97%	Hypotonia, infantile, with psychomotor retardation, 616816	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CCDC186	100%	100%	100%	97%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
CCDC22	100%	99%	98%	72%	Ritscher-Schinzel syndrome 2, 300963	INTELLECTUAL DISABILITY MENDELIOME
CCDC28B	100%	100%	100%	98%	No OMIM disease ID	CILIOPATHIES MENDELIOME
CCDC32	100%	100%	100%	98%	Cardiofacioneurodevelopmental syndrome, 619123	INTELLECTUAL DISABILITY MENDELIOME SCHISIS
CCDC39	100%	100%	100%	96%	Ciliary dyskinesia, primary, 14, 613807	CILIOPATHIES MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING
CCDC40	100%	100%	100%	99%	Ciliary dyskinesia, primary, 15, 613808	CILIOPATHIES MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING
CCDC47	100%	100%	100%	98%	Trichohepatoneurodevelopmental syndrome, 618268	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CCDC50	100%	100%	99%	97%	?Deafness, autosomal dominant 44, 607453	HEARING IMPAIRMENT MENDELIOME
CCDC62	100%	100%	100%	97%	?Spermatogenic failure 67, 619803	MENDELIOME
CCDC65	100%	100%	100%	97%	Ciliary dyskinesia, primary, 27, 615504	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING
CCDC78	100%	100%	100%	99%	?Centronuclear myopathy 4, 614807	MENDELIOME MUSCLE DISORDERS
CCDC8	100%	100%	100%	98%	3-M syndrome 3, 614205	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
CCDC88A	97%	97%	99%	96%	?PEHO syndrome-like, 617507	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CCDC88C	100%	100%	100%	98%	?Spinocerebellar ataxia 40, 616053 Hydrocephalus, congenital, 1, 236600	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

CCL2	100%	100%	100%	95%	No OMIM disease ID	IRON DISORDERS MENDELIOME
CCM2	100%	100%	99%	98%	Cerebral cavernous malformations-2, 603284	EPILEPSY MENDELIOME
CCN6	100%	100%	100%	98%	Progressive pseudorheumatoid dysplasia, 208230	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
CCND2	100%	100%	100%	99%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938	INTELLECTUAL DISABILITY MENDELIOME
CCNF	100%	100%	100%	99%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 5, 619141	MENDELIOME
CCNK	99%	95%	96%	87%	?Intellectual developmental disorder with hypertelorism and distinctive facies, 618147	INTELLECTUAL DISABILITY MENDELIOME
CCNO	100%	100%	100%	98%	Ciliary dyskinesia, primary, 29, 615872	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING
CCNQ	100%	99%	96%	74%	STAR syndrome, 300707	DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME
CCT2	100%	100%	100%	98%	No OMIM disease ID	VISION DISORDERS MENDELIOME
CCT5	100%	100%	100%	98%	Neuropathy, hereditary sensory, with spastic paraplegia, 256840	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING
CD151	100%	100%	100%	99%	Epidermolysis bullosa simplex 7, with nephropathy and deafness, 609057	SKIN DISORDERS HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CD164	100%	100%	100%	97%	?Deafness, autosomal dominant 66, 616969	HEARING IMPAIRMENT MENDELIOME
CD19	100%	100%	100%	98%	Immunodeficiency, common variable, 3, 613493	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
CD247	100%	100%	100%	99%	?Immunodeficiency 25, 610163	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)

CD27	100%	100%	100%	99%	Lymphoproliferative syndrome 2, 615122	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING TUMOR
CD28	100%	100%	100%	98%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
CD2AP	100%	100%	100%	96%	Glomerulosclerosis, focal segmental, 3, 607832	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CD320	100%	100%	100%	99%	Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CD36	100%	99%	100%	98%	Platelet glycoprotein IV deficiency, 608404	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME
CD3D	100%	100%	100%	98%	Immunodeficiency 19, severe combined, 615617	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)
CD3E	100%	100%	100%	98%	Immunodeficiency 18, 615615 Immunodeficiency 18, SCID variant, 615615	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)
CD3G	100%	100%	100%	99%	Immunodeficiency 17, CD3 gamma deficient, 615607	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)
CD4	100%	100%	100%	98%	Immunodeficiency 79, 619238 OKT4 epitope deficiency, 613949	PRIMARY IMMUNODEFICIENCY MENDELIOME
CD40	100%	100%	100%	99%	Immunodeficiency with hyper-IgM, type 3, 606843	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
CD40LG	100%	99%	97%	69%	Immunodeficiency, X-linked, with hyper-IgM, 308230	PRIMARY IMMUNODEFICIENCY MENDELIOME
CD46	100%	100%	100%	98%	No OMIM disease ID	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY RENAL DISORDERS MENDELIOME
CD48	100%	100%	100%	99%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME

CD55	95%	92%	100%	98%	Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
CD59	100%	100%	100%	99%	Hemolytic anemia, CD59-mediated, w/wo immune-mediated polyneuropathy, 612300	NEUROPATHIES PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
CD70	100%	100%	100%	97%	Lymphoproliferative syndrome 3, 618261	PRIMARY IMMUNODEFICIENCY MENDELIOME TUMOR
CD79A	100%	99%	99%	91%	Agammaglobulinemia 3, 613501	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
CD79B	100%	100%	100%	98%	Agammaglobulinemia 6, 612692	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
CD81	100%	99%	100%	98%	Immunodeficiency, common variable, 6, 613496	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
CD8A	100%	100%	100%	97%	CD8 deficiency, familial, 608957	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)
CD96	100%	100%	100%	99%	C syndrome, 211750	MENDELIOME
CDAN1	100%	100%	99%	96%	Dyserythropoietic anemia, congenital, type Ia, 224120	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS IRON DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CDC14A	100%	100%	99%	96%	Deafness, autosomal recessive 32, w/wo immotile sperm, 608653	HEARING IMPAIRMENT MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING
CDC40	100%	100%	100%	98%	?Pontocerebellar hypoplasia, type 15, 619302	MENDELIOME

CDC42	100%	100%	100%	98%	Takenouchi-Kosaki syndrome, 616737	HEARING IMPAIRMENT HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME AND RASOPATHY
CDC42BPB	100%	100%	100%	98%	Chilton-Okur-Chung neurodevelopmental syndrome, 619841	INTELLECTUAL DISABILITY MENDELIOME
CDC45	100%	100%	100%	99%	Meier-Gorlin syndrome 7, 617063	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS
CDC6	100%	100%	100%	99%	?Meier-Gorlin syndrome 5, 613805	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CDC73	100%	100%	100%	98%	Hyperparathyroidism, familial primary, 145000 Parathyroid adenoma with cystic changes, 145001 Parathyroid carcinoma, 608266 Hyperparathyroidism-jaw tumor syndrome, 145001	SHORT STATURE AND SKELETAL DYSPLASIA ENDOCRINE TUMOR MENDELIOME TUMOR
CDCA7	100%	100%	100%	98%	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
CDH1	98%	98%	100%	98%	Ovarian cancer, somatic, 167000 Blepharocheilodontic syndrome 1, 119580 Diffuse gastric and lobular breast cancer syndrome w/wo cleft lip and/or palate, 137215 Endometrial carcinoma, somatic, 608089 Breast cancer, lobular, somatic, 114480	MENDELIOME SCHISIS TUMOR
CDH11	100%	100%	100%	99%	Teebi hypertelorism syndrome 2, 619736 Elsahy-Waters syndrome, 211380	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CDH15	100%	100%	100%	98%	Intellectual developmental disorder, autosomal dominant 3, 612580	INTELLECTUAL DISABILITY MENDELIOME

CDH2	100%	100%	100%	99%	Arrhythmogenic right ventricular dysplasia, familial, 14, 618920 ?Attention deficit-hyperactivity disorder 8, 619957 Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929	VISION DISORDERS DISORDERS OF SEX DEVELOPMENT HEART INTELLECTUAL DISABILITY MENDELIOME
CDH23	100%	100%	100%	99%	Usher syndrome, type 1D, 601067 Usher syndrome, type 1D/F digenic, 601067 Deafness, autosomal recessive 12, 601386	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING TUMOR
CDH3	100%	100%	100%	98%	Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280	VISION DISORDERS SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
CDH4	100%	100%	100%	98%	No OMIM disease ID	VISION DISORDERS MENDELIOME
CDHR1	100%	100%	100%	99%	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CDK10	100%	100%	100%	98%	Al Kaissi syndrome, 617694	VISION DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CDK13	100%	100%	100%	96%	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360	INTELLECTUAL DISABILITY MENDELIOME
CDK19	100%	100%	100%	98%	Developmental and epileptic encephalopathy 87, 618916	INTELLECTUAL DISABILITY MENDELIOME
CDK4	100%	100%	100%	99%	No OMIM disease ID	SKIN DISORDERS MELA1 MELA2 MENDELIOME TUMOR
CDK5	100%	100%	100%	99%	?Lissencephaly 7 with cerebellar hypoplasia, 616342	MENDELIOME PRE CONCEPTION SCREENING
CDK5RAP2	100%	100%	100%	98%	Microcephaly 3, primary, autosomal recessive, 604804	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

CDK6	100%	100%	100%	98%	?Microcephaly 12, primary, autosomal recessive, 616080	MENDELIOME PRE CONCEPTION SCREENING
CDK8	100%	100%	100%	98%	Intellectual developmental disorder with hypotonia and behavioral abnormalities, 618748	INTELLECTUAL DISABILITY MENDELIOME
CDKL5	95%	95%	97%	69%	Developmental and epileptic encephalopathy 2, 300672	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS
CDKN1A	100%	100%	100%	99%	No OMIM disease ID	ENDOCRINE TUMOR MENDELIOME TUMOR
CDKN1B	100%	100%	100%	97%	Multiple endocrine neoplasia, type IV, 610755	ENDOCRINE TUMOR MENDELIOME TUMOR
CDKN1C	100%	100%	100%	92%	IMAGE syndrome, 614732 Beckwith-Wiedemann syndrome, 130650	DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS TUMOR
CDKN2A	100%	100%	100%	97%	No OMIM disease ID	SKIN DISORDERS MELA1 MELA2 MENDELIOME TUMOR
CDKN2B	100%	100%	100%	99%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY ENDOCRINE TUMOR MENDELIOME TUMOR
CDKN2C	100%	100%	100%	97%	No OMIM disease ID	ENDOCRINE TUMOR MENDELIOME TUMOR
CDON	100%	100%	100%	99%	Holoprosencephaly 11, 614226	VISION DISORDERS CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME
CDSN	100%	100%	100%	99%	Hypotrichosis 2, 146520 Peeling skin syndrome 1, 270300	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING

CDT1	100%	100%	100%	98%	Meier-Gorlin syndrome 4, 613804	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
CDY1	50%	50%	48%	24%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
CDY1B	50%	49%	48%	20%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
CDY2A	50%	50%	48%	24%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
CDY2B	50%	50%	47%	20%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
CEACAM16	100%	100%	100%	99%	Deafness, autosomal dominant 4B, 614614 Deafness, autosomal recessive 113, 618410	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
CEBPA	100%	100%	98%	70%	Leukemia, acute myeloid, somatic, 601626 ?Leukemia, acute myeloid, 601626	HEREDITARY BONE MARROW FAILURE MENDELIOME TUMOR
CEBPE	100%	100%	100%	98%	?Immunodeficiency 108 with autoinflammation, 260570 Specific granule deficiency, 245480	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
CEL	100%	100%	99%	92%	Maturity-onset diabetes of the young, type VIII, 609812	METABOLIC DISORDERS MENDELIOME
CELA2A	100%	100%	100%	99%	Abdominal obesity-metabolic syndrome 4, 618620	MENDELIOME
CELF2	100%	100%	100%	98%	Developmental and epileptic encephalopathy 97, 619561	INTELLECTUAL DISABILITY MENDELIOME
CELSR1	100%	100%	100%	98%	Lymphatic malformation 9, 619319	SKIN DISORDERS MENDELIOME
CENPE	100%	100%	100%	96%	?Microcephaly 13, primary, autosomal recessive, 616051	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
CENPF	100%	100%	100%	97%	Stromme syndrome, 243605	CILIOPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CENPJ	100%	100%	100%	97%	Microcephaly 6, primary, autosomal recessive, 608393 ?Seckel syndrome 4, 613676	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CENPT	100%	100%	100%	99%	?Short stature and microcephaly with genital anomalies, 618702	MENDELIOME

CEP104	100%	100%	100%	98%	Joubert syndrome 25, 616781 Intellectual developmental disorder, autosomal recessive 77, 619988	CILIOPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CEP112	100%	100%	100%	97%	Spermatogenic failure 44, 619044	MENDELIOME
CEP120	100%	100%	100%	99%	Short-rib thoracic dysplasia 13 w/wo polydactyly, 616300 Joubert syndrome 31, 617761	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CEP135	100%	100%	100%	97%	Microcephaly 8, primary, autosomal recessive, 614673	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CEP152	100%	100%	100%	98%	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CEP164	100%	100%	100%	98%	Nephronophthisis 15, 614845	VISION DISORDERS CILIOPATHIES RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CEP19	100%	100%	100%	98%	Morbid obesity and spermatogenic failure, 615703	MENDELIOME PRE CONCEPTION SCREENING
CEP250	100%	100%	100%	98%	Cone-rod dystrophy and hearing loss 2, 618358	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME
CEP290	100%	100%	100%	96%	Leber congenital amaurosis 10, 611755 Joubert syndrome 5, 610188 Senior-Loken syndrome 6, 610189 ?Bardet-Biedl syndrome 14, 615991 Meckel syndrome 4, 611134	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING

CEP41	100%	100%	100%	98%	Joubert syndrome 15, 614464	VISION DISORDERS CILIOPATHIES DISORDERS OF SEX DEVELOPMENT INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CEP55	100%	100%	100%	98%	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500	CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CEP57	100%	100%	100%	97%	Mosaic variegated aneuploidy syndrome 2, 614114	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CEP63	100%	100%	100%	98%	?Seckel syndrome 6, 614728	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CEP78	100%	100%	100%	98%	Cone-rod dystrophy and hearing loss, 617236	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
CEP83	100%	100%	100%	95%	Nephronophthisis 18, 615862	VISION DISORDERS CILIOPATHIES LIVER DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CEP85L	100%	100%	100%	97%	Lissencephaly 10, 618873	INTELLECTUAL DISABILITY MENDELIOME
CEP89	100%	100%	100%	97%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS
CERKL	98%	98%	100%	97%	Retinitis pigmentosa 26, 608380	VISION DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING

CERS1	99%	99%	99%	96%	Epilepsy, progressive myoclonic, 8, 616230	MENDELIOME PRE CONCEPTION SCREENING
CERS3	100%	100%	100%	98%	Ichthyosis, congenital, autosomal recessive 9, 615023	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CERT1	100%	100%	100%	98%	Intellectual developmental disorder, autosomal dominant 34, 616351	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
CES1	99%	99%	99%	96%	Drug metabolism, altered, CES1-related, 618057	MENDELIOME
CETP	100%	100%	100%	99%	Hyperalphalipoproteinemia, 143470	MENDELIOME
CFAP298	100%	100%	100%	97%	Ciliary dyskinesia, primary, 26, 615500	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING
CFAP300	100%	100%	100%	96%	Ciliary dyskinesia, primary, 38, 618063	CILIOPATHIES MENDELIOME
CFAP410	100%	100%	100%	99%	Retinal dystrophy with macular staphyloma, 617547 Spondylometaphyseal dysplasia, axial, 602271	ALS VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
CFAP43	100%	100%	100%	97%	Hydrocephalus, normal pressure, 1, 236690 Spermatogenic failure 19, 617592	MALE INFERTILITY MENDELIOME
CFAP44	100%	100%	100%	97%	Spermatogenic failure 20, 617593	CILIOPATHIES MALE INFERTILITY MENDELIOME
CFAP45	100%	100%	100%	98%	Heterotaxy, visceral, 11, autosomal, with male infertility, 619608	CILIOPATHIES MENDELIOME
CFAP47	99%	99%	97%	71%	Spermatogenic failure, X-linked 3, 301059	MENDELIOME
CFAP52	100%	100%	100%	98%	Heterotaxy, visceral, 10, autosomal, with male infertility, 619607	CILIOPATHIES MENDELIOME
CFAP53	100%	100%	99%	97%	Heterotaxy, visceral, 6, autosomal recessive, 614779	CONGENITAL HEART DISEASE CILIOPATHIES HEART MENDELIOME PRE CONCEPTION SCREENING
CFAP58	100%	100%	100%	97%	Spermatogenic failure 49, 619144	MALE INFERTILITY MENDELIOME MITOCHONDRIAL DISORDERS

CFAP65	100%	100%	100%	98%	Spermatogenic failure 40, 618664	MALE INFERTILITY MENDELIOME
CFAP69	100%	100%	100%	97%	Spermatogenic failure 24, 617959	CILIOPATHIES MALE INFERTILITY MENDELIOME
CFAP70	100%	100%	100%	99%	?Spermatogenic failure 41, 618670	MENDELIOME
CFB	100%	100%	100%	99%	?Complement factor B deficiency, 615561	AGE RELATED MACULAR DEGENERATION HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY RENAL DISORDERS MENDELIOME
CFC1	100%	100%	100%	99%	Heterotaxy, visceral, 2, autosomal, 605376	CONGENITAL HEART DISEASE CILIOPATHIES HEART LIVER DISORDERS MENDELIOME
CFD	100%	100%	99%	93%	Complement factor D deficiency, 613912	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
CFH	100%	100%	100%	99%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814	AGE RELATED MACULAR DEGENERATION VISION DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CFHR1	99%	97%	93%	82%	No OMIM disease ID	RENAL DISORDERS MENDELIOME
CFHR3	99%	99%	95%	84%	No OMIM disease ID	RENAL DISORDERS MENDELIOME
CFHR5	100%	100%	100%	98%	Nephropathy due to CFHR5 deficiency, 614809	RENAL DISORDERS MENDELIOME
CFI	100%	100%	100%	98%	Complement factor I deficiency, 610984	AGE RELATED MACULAR DEGENERATION HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CFL2	100%	100%	100%	96%	Nemaline myopathy 7, autosomal recessive, 610687	MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS

CFP	100%	99%	98%	75%	Properdin deficiency, X-linked, 312060	PRIMARY IMMUNODEFICIENCY MENDELIOME
CFTR	100%	100%	100%	98%	Cystic fibrosis, 219700 Congenital bilateral absence of vas deferens, 277180 Sweat chloride elevation without CF,	PRIMARY IMMUNODEFICIENCY LIVER DISORDERS MALE INFERTILITY METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CHAMP1	100%	100%	100%	96%	Neurodevelopmental disorder with hypotonia, impaired language, and dysmorphic features, 616579	INTELLECTUAL DISABILITY MENDELIOME
CHAT	100%	100%	99%	98%	Myasthenic syndrome, congenital, 6, presynaptic, 254210	FETAL AKINESIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
CHCHD10	100%	100%	100%	96%	?Myopathy, isolated mitochondrial, autosomal dominant, 616209 Spinal muscular atrophy, Jokela type, 615048 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911	ALS NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS MUSCLE DISORDERS
CHCHD2	100%	100%	100%	99%	Parkinson disease 22, autosomal dominant, 616710	MENDELIOME MITOCHONDRIAL DISORDERS PARKINSON
CHD1	100%	100%	100%	97%	Pilarowski-Bjornsson syndrome, 617682	INTELLECTUAL DISABILITY MENDELIOME
CHD2	100%	100%	100%	98%	Developmental and epileptic encephalopathy 94, 615369	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
CHD3	100%	99%	99%	95%	Snijders Blok-Campeau syndrome, 618205	INTELLECTUAL DISABILITY MENDELIOME
CHD4	100%	100%	100%	98%	Sifrim-Hitz-Weiss syndrome, 617159	CONGENITAL HEART DISEASE HEART INTELLECTUAL DISABILITY MENDELIOME
CHD5	100%	100%	100%	98%	Parenti-Mignot neurodevelopmental syndrome, 619873	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME

CHD7	100%	100%	100%	98%	Hypogonadotropic hypogonadism 5 w/wo anosmia, 612370 CHARGE syndrome, 214800	VISION DISORDERS CRANIOFACIAL ANOMALIES CONGENITAL HEART DISEASE HEARING IMPAIRMENT DISORDERS OF SEX DEVELOPMENT HEART HYPOGONADOTROPIC HYPOGONADISM PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME SCHISIS
CHD8	100%	100%	100%	98%	Intellectual developmental disorder with autism and macrocephaly, 615032	LIVER DISORDERS INTELLECTUAL DISABILITY MENDELIOME
CHEK2	100%	100%	100%	98%	Osteosarcoma, somatic, 259500 Li-Fraumeni syndrome 2, 609265	HEREDITARY BREAST AND OVARIAN CANCER MENDELIOME TUMOR
CHIT1	100%	100%	100%	99%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME
CHKA	100%	100%	100%	91%	Neurodevelopmental disorder with microcephaly, movement abnormalities, and seizures, 620023	INTELLECTUAL DISABILITY MENDELIOME
CHKB	100%	100%	100%	98%	Muscular dystrophy, congenital, megaconial type, 602541	SKIN DISORDERS HEART METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS
CHM	99%	97%	98%	73%	Choroideremia, 303100	VISION DISORDERS MENDELIOME
CHMP1A	100%	100%	100%	99%	Pontocerebellar hypoplasia, type 8, 614961	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CHMP2B	100%	100%	99%	95%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 7, 600795	ALS MENDELIOME PARKINSON
CHMP4B	100%	100%	100%	98%	Cataract 31, multiple types, 605387	VISION DISORDERS MENDELIOME

CHN1	96%	96%	100%	98%	Duane retraction syndrome 2, 604356	VISION DISORDERS MENDELIOME
CHP1	100%	100%	100%	98%	?Spastic ataxia 9, autosomal recessive, 618438	MENDELIOME PRE CONCEPTION SCREENING
CHRDL1	100%	99%	98%	75%	Megalocornea 1, X-linked, 309300	VISION DISORDERS MENDELIOME
CHRM1	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
CHRM2	99%	98%	100%	99%	No OMIM disease ID	HEART MENDELIOME
CHRM3	100%	100%	100%	99%	Prune belly syndrome, 100100	LIVER DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CHRNA1	100%	100%	100%	98%	Myasthenic syndrome, congenital, 1B, fast-channel, 608930 Myasthenic syndrome, congenital, 1A, slow-channel, 601462 Multiple pterygium syndrome, lethal type, 253290	FETAL AKINESIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
CHRNA2	100%	100%	100%	99%	Epilepsy, nocturnal frontal lobe, type 4, 610353	EPILEPSY MENDELIOME
CHRNA3	100%	100%	100%	97%	Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT, 191800	LIVER DISORDERS RENAL DISORDERS MENDELIOME
CHRNA4	100%	100%	100%	97%	Epilepsy, nocturnal frontal lobe, 1, 600513	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
CHRNB1	100%	100%	100%	97%	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314 Myasthenic syndrome, congenital, 2A, slow-channel, 616313	FETAL AKINESIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
CHRNB2	100%	100%	100%	99%	Epilepsy, nocturnal frontal lobe, 3, 605375	EPILEPSY MENDELIOME
CHRND	100%	100%	100%	99%	?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323 Multiple pterygium syndrome, lethal type, 253290 Myasthenic syndrome, congenital, 3B, fast-channel, 616322 ?Myasthenic syndrome, congenital, 3A, slow-channel, 616321	FETAL AKINESIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
CHRNE	100%	100%	100%	97%	Myasthenic syndrome, congenital, 4A, slow-channel, 605809 Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931 Myasthenic syndrome, congenital, 4B, fast-channel, 616324	FETAL AKINESIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS

CHRNG	100%	100%	100%	99%	Multiple pterygium syndrome, lethal type, 253290 Escobar syndrome, 265000	FETAL AKINESIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS MUSCLE DISORDERS
CHST11	100%	100%	100%	97%	?Osteochondrodysplasia, brachydactyly, and overlapping malformed digits, 618167	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
CHST14	100%	100%	100%	91%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776	FETAL AKINESIA SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS
CHST3	100%	100%	100%	99%	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CHST6	100%	100%	100%	99%	Macular corneal dystrophy, 217800	VISION DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CHST8	100%	100%	100%	99%	?Peeling skin syndrome 3, 616265	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CHSY1	99%	99%	100%	97%	Temtamy preaxial brachydactyly syndrome, 605282	SKIN DISORDERS HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CHUK	100%	100%	100%	98%	?Popliteal pterygium syndrome, Bartsocas-Papas type 2, 619339 ?Cocoon syndrome, 613630	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME
CIB1	100%	100%	100%	97%	Epidermodysplasia verruciformis 3, 618267	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME

CIB2	100%	99%	99%	97%	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
CIC	100%	100%	100%	99%	Intellectual developmental disorder, autosomal dominant 45, 617600	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
CIDEC	100%	100%	100%	98%	?Lipodystrophy, familial partial, type 5, 615238	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CIITA	100%	100%	100%	99%	Bare lymphocyte syndrome, type II, complementation group A, 209920	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)
CILK1	100%	100%	100%	99%	Endocrine-cerebroosteodysplasia, 612651	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS
CISD2	100%	100%	100%	98%	Wolfram syndrome 2, 604928	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
CIT	100%	100%	100%	98%	Microcephaly 17, primary, autosomal recessive, 617090	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CITED2	100%	100%	100%	96%	Atrial septal defect 8, 614433 Ventricular septal defect 2, 614431	CONGENITAL HEART DISEASE HEART MENDELIOME
CKAP2L	100%	100%	100%	98%	Filippi syndrome, 272440	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CLCC1	100%	100%	100%	98%	Retinitis pigmentosa 32, 609913	VISION DISORDERS MENDELIOME
CLCF1	100%	100%	100%	98%	Cold-induced sweating syndrome 2, 610313	MENDELIOME PRE CONCEPTION SCREENING

CLCN1	100%	100%	100%	98%	Myotonia congenita, recessive, 255700 Myotonia congenita, dominant, 160800 Myotonia levior, recessive,	MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
CLCN2	100%	100%	100%	98%	Leukoencephalopathy with ataxia, 615651 Hyperaldosteronism, familial, type II, 605635	MOVEMENT DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CLCN3	96%	96%	100%	98%	Neurodevelopmental disorder with seizures and brain abnormalities, 619517 Neurodevelopmental disorder with hypotonia and brain abnormalities, 619512	INTELLECTUAL DISABILITY MENDELIOME
CLCN4	100%	100%	98%	71%	Raynaud-Claes syndrome, 300114	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
CLCN5	100%	99%	98%	73%	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990 Hypophosphatemic rickets, 300554 Dent disease 1, 300009 Nephrolithiasis, type I, 310468	SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME
CLCN6	100%	100%	100%	99%	Neurodegeneration, childhood-onset, hypotonia, respiratory insufficiency and brain imaging abnormalities, 619173	MOVEMENT DISORDERS MENDELIOME
CLCN7	100%	100%	100%	99%	Hypopigmentation, organomegaly, and delayed myelination and development, 618541 Osteopetrosis, autosomal recessive 4, 611490 Osteopetrosis, autosomal dominant 2, 166600	PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CLCNKA	100%	100%	100%	98%	Bartter syndrome, type 4b, digenic, 613090	MENDELIOME
CLCNKB	100%	100%	100%	98%	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CLDN1	100%	100%	100%	99%	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626	SKIN DISORDERS LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CLDN10	100%	100%	100%	99%	HELIX syndrome, 617671	SKIN DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CLDN11	100%	100%	100%	99%	Leukodystrophy, hypomyelinating, 22, 619328	INTELLECTUAL DISABILITY MENDELIOME
CLDN14	100%	100%	100%	99%	Deafness, autosomal recessive 29, 614035	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING

CLDN16	100%	100%	100%	98%	Hypomagnesemia 3, renal, 248250	EPILEPSY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CLDN19	100%	100%	100%	99%	Hypomagnesemia 5, renal, with ocular involvement, 248190	VISION DISORDERS EPILEPSY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CLDN2	100%	99%	98%	71%	?Azoospermia, obstructive, with nephrolithiasis, 301060	MENDELIOME
CLDN9	100%	100%	100%	99%	?Deafness, autosomal recessive 116, 619093	HEARING IMPAIRMENT MENDELIOME
CLEC3B	100%	100%	100%	99%	Macular dystrophy, retinal, 4, 619977	VISION DISORDERS MENDELIOME
CLEC4D	100%	100%	100%	98%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
CLEC7A	100%	100%	100%	98%	Candidiasis, familial, 4, autosomal recessive, 613108	PRIMARY IMMUNODEFICIENCY MENDELIOME
CLIC2	100%	100%	98%	74%	?Intellectual developmental disorder, X-linked syndromic 32, 300886	INTELLECTUAL DISABILITY MENDELIOME
CLIC5	100%	100%	100%	96%	?Deafness, autosomal recessive 103, 616042	HEARING IMPAIRMENT MENDELIOME
CLIP1	100%	100%	100%	96%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CLMP	100%	100%	100%	98%	Congenital short bowel syndrome, 615237	LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CLN3	93%	93%	100%	98%	Ceroid lipofuscinosis, neuronal, 3, 204200	VISION DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CLN5	83%	83%	100%	96%	Ceroid lipofuscinosis, neuronal, 5, 256731	MOVEMENT DISORDERS VISION DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

CLN6	100%	100%	100%	97%	Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300 Ceroid lipofuscinosis, neuronal, 6A, 601780	MOVEMENT DISORDERS VISION DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CLN8	100%	100%	100%	99%	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 Ceroid lipofuscinosis, neuronal, 8, 600143	VISION DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CLP1	100%	100%	100%	99%	Pontocerebellar hypoplasia, type 10, 615803	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CLPB	100%	100%	99%	98%	Neutropenia, severe congenital, 9, autosomal dominant, 619813 3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271 3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835	MOVEMENT DISORDERS HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
CLPP	100%	100%	100%	96%	Perrault syndrome 3, 614129	HEARING IMPAIRMENT DISORDERS OF SEX DEVELOPMENT MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY
CLPX	100%	100%	100%	97%	?Protoporphyria, erythropoietic, 2, 618015	MENDELIOME
CLRN1	100%	100%	100%	98%	Usher syndrome, type 3A, 276902 Retinitis pigmentosa 61, 614180	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
CLRN2	100%	100%	100%	99%	?Deafness, autosomal recessive 117, 619174	HEARING IMPAIRMENT MENDELIOME
CLTC	100%	100%	100%	98%	Intellectual developmental disorder, autosomal dominant 56, 617854	INTELLECTUAL DISABILITY MENDELIOME
CLTCL1	100%	100%	100%	99%	No OMIM disease ID	HNPD MENDELIOME

CLUAP1	100%	100%	100%	98%	No OMIM disease ID	VISION DISORDERS MENDELIOME
CMAS	100%	100%	100%	98%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME
CNBP	100%	100%	100%	99%	Myotonic dystrophy 2, 602668	MENDELIOME
CNGA1	91%	91%	100%	97%	Retinitis pigmentosa 49, 613756	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CNGA2	99%	99%	97%	68%	No OMIM disease ID	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME
CNGA3	100%	100%	100%	99%	Achromatopsia 2, 216900	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CNGB1	100%	100%	100%	98%	Retinitis pigmentosa 45, 613767	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CNGB3	100%	100%	100%	98%	Achromatopsia 3, 262300	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CNKSR2	99%	98%	98%	73%	Intellectual developmental disorder, X-linked syndromic, Houge type, 301008	INTELLECTUAL DISABILITY MENDELIOME
CNNM2	100%	100%	100%	97%	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and impaired intellectual development 1, 616418	EPILEPSY INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CNNM4	100%	100%	100%	97%	Jalili syndrome, 217080	VISION DISORDERS SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CNOT1	100%	100%	100%	98%	Vissers-Bodmer syndrome, 619033 Holoprosencephaly 12, w/wo pancreatic agenesis, 618500	INTELLECTUAL DISABILITY MENDELIOME
CNOT2	100%	100%	100%	98%	Intellectual developmental disorder with nasal speech, dysmorphic facies, and variable skeletal anomalies, 618608	INTELLECTUAL DISABILITY MENDELIOME
CNOT3	100%	100%	100%	99%	Intellectual developmental disorder with speech delay, autism, and dysmorphic facies, 618672	INTELLECTUAL DISABILITY MENDELIOME
CNP	100%	100%	100%	99%	?Leukodystrophy, hypomyelinating, 20, 619071	MENDELIOME
CNPY3	100%	100%	100%	97%	Developmental and epileptic encephalopathy 60, 617929	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

CNTN1	100%	100%	100%	98%	?Congenital myopathy 12, 612540	MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
CNTN2	100%	100%	99%	99%	?Epilepsy, myoclonic, familial adult, 5, 615400	EPILEPSY MENDELIOME PRE CONCEPTION SCREENING
CNTNAP1	100%	100%	100%	98%	Lethal congenital contracture syndrome 7, 616286 Hypomyelinating neuropathy, congenital, 3, 618186	FETAL AKINESIA NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CNTNAP2	100%	100%	100%	99%	Pitt-Hopkins like syndrome 1, 610042	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
COA1	100%	100%	100%	98%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
COA3	100%	100%	100%	99%	?Mitochondrial complex IV deficiency, nuclear type 14, 619058	NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS
COA5	82%	82%	100%	98%	?Mitochondrial complex IV, deficiency, nuclear type 9, 616500	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
COA6	100%	100%	100%	96%	Mitochondrial complex IV deficiency, nuclear type 13, 616501	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
COA7	100%	100%	100%	99%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387	NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
COA8	100%	99%	100%	97%	Mitochondrial complex IV deficiency, nuclear type 17, 619061	VISION DISORDERS HEARING IMPAIRMENT EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
COASY	100%	100%	100%	99%	Pontocerebellar hypoplasia, type 12, 618266 Neurodegeneration with brain iron accumulation 6, 615643	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING

COCH	100%	100%	100%	99%	Deafness, autosomal dominant 9, 601369 ?Deafness, autosomal recessive 110, 618094	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
COG1	100%	100%	100%	97%	Congenital disorder of glycosylation, type IIg, 611209	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
COG2	100%	100%	100%	98%	?Congenital disorder of glycosylation, type IIq, 617395	METABOLIC DISORDERS MENDELIOME
COG4	100%	100%	100%	98%	Congenital disorder of glycosylation, type IIj, 613489 Saul-Wilson syndrome, 618150	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
COG5	100%	100%	100%	97%	Congenital disorder of glycosylation, type IIi, 613612	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
COG6	100%	100%	100%	98%	Shaheen syndrome, 615328 Congenital disorder of glycosylation, type IIl, 614576	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
COG7	100%	100%	100%	98%	Congenital disorder of glycosylation, type IIe, 608779	LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
COG8	100%	100%	99%	97%	Congenital disorder of glycosylation, type IIh, 611182	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
COL10A1	100%	100%	100%	97%	Metaphyseal chondrodysplasia, Schmid type, 156500	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME

COL11A1	100%	100%	100%	97%	Fibrochondrogenesis 1, 228520 Stickler syndrome, type II, 604841 Marshall syndrome, 154780 Deafness, autosomal dominant 37, 618533	VISION DISORDERS CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS
COL11A2	100%	100%	100%	98%	Deafness, autosomal dominant 13, 601868 Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150 Fibrochondrogenesis 2, 614524 Deafness, autosomal recessive 53, 609706 Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840	CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS
COL12A1	100%	100%	100%	98%	Bethlem myopathy 2, 616471 ?Ullrich congenital muscular dystrophy 2, 616470	MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
COL13A1	100%	100%	100%	99%	Myasthenic syndrome, congenital, 19, 616720	MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
COL14A1	100%	100%	100%	98%	No OMIM disease ID	SKIN DISORDERS MENDELIOME
COL17A1	100%	100%	100%	98%	Epithelial recurrent erosion dystrophy, 122400 Epidermolysis bullosa, junctional 4, intermediate, 619787	VISION DISORDERS SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
COL18A1	100%	100%	100%	99%	Knobloch syndrome, type 1, 267750 Glaucoma, primary closed-angle, 618880	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
COL1A1	100%	100%	100%	99%	Osteogenesis imperfecta, type II, 166210 Caffey disease, 114000 Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060 Osteogenesis imperfecta, type I, 166200 Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1, 619115 Osteogenesis imperfecta, type IV, 166220 Osteogenesis imperfecta, type III, 259420	HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME

COL1A2	100%	100%	100%	99%	Osteogenesis imperfecta, type III, 259420 Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821 Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2, 619120 Ehlers-Danlos syndrome, cardiac valvular type, 225320 Osteogenesis imperfecta, type IV, 166220 Osteogenesis imperfecta, type II, 166210	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
COL25A1	99%	99%	100%	98%	Fibrosis of extraocular muscles, congenital, 5, 616219	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
COL27A1	100%	100%	100%	98%	Steel syndrome, 615155	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
COL2A1	100%	100%	100%	99%	?Vitreoretinopathy with phalangeal epiphyseal dysplasia, 619248 Czech dysplasia, 609162 Achondrogenesis, type II or hypochondrogenesis, 200610 Spondyloperipheral dysplasia, 271700 SMED Strudwick type, 184250 ?Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 SED congenita, 183900 Kniest dysplasia, 156550 Stickler syndrome, type I, nonsyndromic ocular, 609508 Osteoarthritis with mild chondrodysplasia, 604864 Stickler syndrome, type I, 108300 Platyspondylic skeletal dysplasia, Torrance type, 151210 Spondyloepiphyseal dysplasia, Stanescu type, 616583 Avascular necrosis of the femoral head, 608805 Legg-Calve-Perthes disease, 150600	VISION DISORDERS CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS
COL3A1	100%	100%	100%	98%	Ehlers-Danlos syndrome, vascular type, 130050 Polymicrogyria w/wo vascular-type EDS, 618343	ANEURYSM CONGENITAL HEART DISEASE SKIN DISORDERS HEART HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
COL4A1	100%	100%	100%	98%	?Retinal arteries, tortuosity of, 180000 Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564 Brain small vessel disease w/wo ocular anomalies, 175780	MOVEMENT DISORDERS VISION DISORDERS EPILEPSY INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME

COL4A2	100%	100%	100%	99%	Brain small vessel disease 2, 614483	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME
COL4A3	100%	100%	100%	98%	Hematuria, benign familial, 141200 Alport syndrome 3, autosomal dominant, 104200 Alport syndrome 2, autosomal recessive, 203780	HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
COL4A4	100%	100%	100%	98%	Hematuria, familial benign, 141200 Alport syndrome 2, autosomal recessive, 203780	HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
COL4A5	99%	98%	97%	69%	Alport syndrome 1, X-linked, 301050	HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME
COL4A6	99%	98%	97%	67%	?Deafness, X-linked 6, 300914	HEARING IMPAIRMENT MENDELIOME
COL5A1	100%	100%	100%	99%	Ehlers-Danlos syndrome, classic type, 1, 130000 Fibromuscular dysplasia, multifocal, 619329	ANEURYSM SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME
COL5A2	100%	100%	100%	98%	Ehlers-Danlos syndrome, classic type, 2, 130010	ANEURYSM SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME
COL6A1	100%	100%	100%	99%	Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090	FETAL AKINESIA MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
COL6A2	100%	100%	100%	99%	Bethlem myopathy 1, 158810 ?Myosclerosis, congenital, 255600 Ullrich congenital muscular dystrophy 1, 254090	FETAL AKINESIA MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
COL6A3	100%	100%	100%	99%	Ullrich congenital muscular dystrophy 1, 254090 Dystonia 27, 616411 Bethlem myopathy 1, 158810	FETAL AKINESIA MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
COL6A5	100%	99%	100%	98%	No OMIM disease ID	NEUROPATHIES HNPD MENDELIOME

COL7A1	100%	100%	100%	99%	Epidermolysis bullosa, pretibial, 131850 Transient bullous of the newborn, 131705 EBD, Bart type, 132000 Epidermolysis bullosa dystrophica, AD, 131750 Epidermolysis bullosa pruriginosa, 604129 EBD inversa, 226600 Epidermolysis bullosa dystrophica, AR, 226600 Toenail dystrophy, isolated, 607523 EBD, localisata variant,	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
COL8A2	100%	100%	100%	91%	Corneal dystrophy, posterior polymorphous 2, 609140 Corneal dystrophy, Fuchs endothelial, 1, 136800	VISION DISORDERS MENDELIOME
COL9A1	100%	100%	100%	97%	Stickler syndrome, type IV, 614134 ?Epiphyseal dysplasia, multiple, 6, 614135	VISION DISORDERS CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS
COL9A2	100%	100%	100%	97%	Epiphyseal dysplasia, multiple, 2, 600204 ?Stickler syndrome, type V, 614284	VISION DISORDERS CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
COL9A3	100%	100%	100%	98%	Epiphyseal dysplasia, multiple, 3, w/wo myopathy, 600969 Stickler syndrome, type VI, 620022	VISION DISORDERS CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
COLEC10	100%	100%	100%	97%	3MC syndrome 3, 248340	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS

COLEC11	100%	100%	100%	99%	3MC syndrome 2, 265050	CRANIOFACIAL ANOMALIES PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS
COLGALT1	100%	100%	99%	95%	Brain small vessel disease 3, 618360	EPILEPSY MENDELIOME PRE CONCEPTION SCREENING
COLQ	100%	100%	100%	99%	Myasthenic syndrome, congenital, 5, 603034	MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
COMP	100%	100%	100%	98%	Pseudoachondroplasia, 177170 Carpal tunnel syndrome 2, 619161 Epiphyseal dysplasia, multiple, 1, 132400	NEUROPATHIES HNPD SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
COMT	100%	100%	100%	99%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME
COPA	100%	100%	100%	99%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
COPB1	100%	100%	100%	97%	Baralle-Macken syndrome, 619255	INTELLECTUAL DISABILITY MENDELIOME
COPB2	100%	100%	100%	98%	Osteoporosis, childhood- or juvenile-onset, with developmental delay, 619884 ?Microcephaly 19, primary, autosomal recessive, 617800	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
COPG1	100%	100%	100%	99%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
COQ2	96%	96%	100%	98%	Coenzyme Q10 deficiency, primary, 1, 607426	MOVEMENT DISORDERS EPILEPSY HEART METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING

COQ4	100%	100%	100%	99%	Coenzyme Q10 deficiency, primary, 7, 616276	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
COQ5	100%	100%	100%	97%	?Coenzyme Q10 deficiency, primary, 9, 619028	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS
COQ6	100%	100%	99%	98%	Coenzyme Q10 deficiency, primary, 6, 614650	HNPD METABOLIC DISORDERS RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
COQ7	100%	100%	100%	98%	?Coenzyme Q10 deficiency, primary, 8, 616733	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS
COQ8A	100%	100%	100%	99%	Coenzyme Q10 deficiency, primary, 4, 612016	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
COQ8B	100%	100%	100%	99%	Nephrotic syndrome, type 9, 615573	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
COQ9	100%	100%	100%	98%	Coenzyme Q10 deficiency, primary, 5, 614654	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
CORIN	100%	99%	100%	99%	Preeclampsia/eclampsia 5, 614595	MENDELIOME

CORO1A	100%	100%	100%	98%	Immunodeficiency 8, 615401	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)
COX10	100%	100%	100%	99%	Mitochondrial complex IV deficiency, nuclear type 3, 619046	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
COX11	100%	100%	100%	94%	Mitochondrial complex IV deficiency, nuclear type 23, 620275	MENDELIOME MITOCHONDRIAL DISORDERS
COX14	100%	100%	100%	100%	?Mitochondrial complex IV deficiency, nuclear type 10, 619053	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
COX15	100%	100%	100%	98%	Mitochondrial complex IV deficiency, nuclear type 6, 615119	FETAL AKINESIA HEART INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
COX16	100%	100%	99%	98%	Mitochondrial complex IV deficiency, nuclear type 22, 619355	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS
COX20	100%	100%	100%	98%	Mitochondrial complex IV deficiency, nuclear type 11, 619054	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
COX4I1	100%	100%	100%	99%	Mitochondrial complex IV deficiency, nuclear type 16, 619060	MENDELIOME MITOCHONDRIAL DISORDERS
COX4I2	100%	100%	100%	98%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
COX5A	100%	100%	100%	98%	?Mitochondrial complex IV deficiency, nuclear type 20, 619064	MENDELIOME MITOCHONDRIAL DISORDERS
COX5B	100%	100%	100%	98%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS

COX6A1	100%	100%	100%	97%	Charcot-Marie-Tooth disease, recessive intermediate D, 616039	NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
COX6A2	100%	99%	100%	95%	Mitochondrial complex IV deficiency, nuclear type 18, 619062	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
COX6B1	100%	100%	100%	99%	Mitochondrial complex IV deficiency, nuclear type 7, 619051	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
COX6B2	100%	100%	100%	93%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
COX6C	100%	100%	100%	98%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
COX7A1	100%	100%	100%	92%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
COX7A2	100%	100%	100%	95%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
COX7B	100%	99%	98%	78%	Linear skin defects with multiple congenital anomalies 2, 300887	VISION DISORDERS SKIN DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS
COX7B2	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
COX7C	100%	100%	100%	97%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
COX8A	100%	100%	100%	99%	?Mitochondrial complex IV deficiency, nuclear type 15, 619059	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
COX8C	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
CP	100%	100%	100%	98%	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290	MOVEMENT DISORDERS IRON DISORDERS METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
CPA6	100%	100%	100%	99%	Febrile seizures, familial, 11, 614418 Epilepsy, familial temporal lobe, 5, 614417	EPILEPSY MENDELIOME PRE CONCEPTION SCREENING

CPAMD8	100%	100%	100%	98%	Anterior segment dysgenesis 8, 617319	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CPE	100%	100%	100%	98%	BDV syndrome, 619326	INTELLECTUAL DISABILITY MENDELIOME
CPLANE1	100%	100%	100%	98%	Orofaciodigital syndrome VI, 277170 Joubert syndrome 17, 614615	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS
CPLX1	100%	100%	100%	97%	Developmental and epileptic encephalopathy 63, 617976	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CPN1	100%	100%	100%	97%	Carboxypeptidase N deficiency, 212070	MENDELIOME PRE CONCEPTION SCREENING
CPOX	100%	100%	100%	97%	Coproporphyria, 121300 Harderoporphyria, 618892	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CPS1	100%	100%	100%	98%	Carbamoylphosphate synthetase I deficiency, 237300	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CPSF1	100%	100%	99%	98%	Myopia 27, 618827	VISION DISORDERS MENDELIOME
CPSF3	100%	100%	100%	98%	Neurodevelopmental disorder with microcephaly, hypotonia, nystagmus, and seizures, 619876	INTELLECTUAL DISABILITY MENDELIOME
CPT1A	100%	100%	100%	98%	CPT deficiency, hepatic, type IA, 255120	HEART METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CPT1C	100%	100%	99%	98%	?Spastic paraplegia 73, autosomal dominant, 616282	MENDELIOME

CPT2	100%	100%	100%	98%	CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 CPT II deficiency, myopathic, stress-induced, 255110	EPILEPSY HEART METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
CR2	100%	100%	100%	99%	?Immunodeficiency, common variable, 7, 614699	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
CRACR2A	100%	100%	100%	98%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
CRADD	100%	100%	100%	97%	Intellectual developmental disorder, autosomal recessive 34, with variant lissencephaly, 614499	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CRAT	100%	100%	100%	99%	?Neurodegeneration with brain iron accumulation 8, 617917	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
CRB1	100%	100%	100%	98%	Leber congenital amaurosis 8, 613835 Retinitis pigmentosa-12, 600105 Pigmented paravenous chorioretinal atrophy, 172870	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CRB2	100%	100%	100%	98%	Focal segmental glomerulosclerosis 9, 616220 Ventriculomegaly with cystic kidney disease, 219730	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CRBN	100%	99%	100%	97%	Intellectual developmental disorder, autosomal recessive 2, 607417	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CREB1	100%	100%	100%	98%	Histiocytoma, angiomatoid fibrous, somatic, 612160	MENDELIOME
CREB3L1	100%	100%	100%	98%	Osteogenesis imperfecta, type XVI, 616229	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
CREB3L3	100%	100%	100%	98%	Hypertriglyceridemia 2, 619324	MENDELIOME
CREBBP	100%	100%	100%	98%	Menke-Hennekam syndrome 1, 618332 Rubinstein-Taybi syndrome 1, 180849	DISORDERS OF SEX DEVELOPMENT PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME TUMOR

CRELD1	100%	100%	100%	98%	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217	CONGENITAL HEART DISEASE HEART MENDELIOME
CRIPT	100%	100%	100%	97%	Short stature with microcephaly and distinctive facies, 615789	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
CRLF1	99%	98%	96%	82%	Cold-induced sweating syndrome 1, 272430	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CRLS1	100%	100%	100%	95%	Combined oxidative phosphorylation deficiency 57, 620167	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS
CRPPA	100%	100%	100%	98%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643	FETAL AKINESIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
CRTAP	100%	100%	100%	98%	Osteogenesis imperfecta, type VII, 610682	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
CRTC1	100%	100%	99%	97%	Mucoepidermoid salivary gland carcinoma,	MENDELIOME
CRX	100%	100%	100%	99%	Leber congenital amaurosis 7, 613829 Cone-rod retinal dystrophy-2, 120970	VISION DISORDERS MENDELIOME
CRYAA	100%	100%	100%	99%	Cataract 9, multiple types, 604219	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CRYAB	100%	100%	100%	99%	Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869 Myopathy, myofibrillar, 2, 608810 Cataract 16, multiple types, 613763 Cardiomyopathy, dilated, 1II, 615184	VISION DISORDERS HEART MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
CRYBA1	100%	100%	100%	99%	Cataract 10, multiple types, 600881	VISION DISORDERS MENDELIOME
CRYBA2	100%	100%	100%	97%	?Cataract 42, 115900	VISION DISORDERS MENDELIOME
CRYBA4	100%	100%	100%	99%	Cataract 23, 610425	VISION DISORDERS MENDELIOME

CRYBB1	100%	100%	100%	99%	Cataract 17, multiple types, 611544	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CRYBB2	100%	100%	100%	99%	Cataract 3, multiple types, 601547	VISION DISORDERS MENDELIOME
CRYBB3	100%	100%	100%	99%	Cataract 22, 609741	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CRYGB	100%	100%	100%	98%	Cataract 39, multiple types, autosomal dominant, 615188	VISION DISORDERS MENDELIOME
CRYGC	100%	100%	100%	99%	Cataract 2, multiple types, 604307	VISION DISORDERS MENDELIOME
CRYGD	100%	100%	100%	98%	Cataract 4, multiple types, 115700	VISION DISORDERS MENDELIOME
CRYGS	100%	100%	100%	99%	Cataract 20, multiple types, 116100	VISION DISORDERS MENDELIOME
CRYL1	100%	100%	100%	98%	No OMIM disease ID	HEARING IMPAIRMENT MENDELIOME
CRYM	100%	100%	100%	97%	Deafness, autosomal dominant 40, 616357	HEARING IMPAIRMENT MENDELIOME
CSDE1	100%	100%	100%	98%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
CSF1R	100%	100%	100%	99%	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 Leukoencephalopathy, diffuse hereditary, with spheroids 1, 221820	MOVEMENT DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PARKINSON PRE CONCEPTION SCREENING
CSF2RA	97%	94%	50%	48%	Surfactant metabolism dysfunction, pulmonary, 4, 300770	PRIMARY IMMUNODEFICIENCY MENDELIOME
CSF2RB	100%	100%	100%	99%	Surfactant metabolism dysfunction, pulmonary, 5, 614370	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
CSF3R	100%	100%	100%	99%	Neutropenia, severe congenital, 7, autosomal recessive, 617014 ?Neutrophilia, hereditary, 162830	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCY MENDELIOME
CSGALNACT1	100%	100%	100%	99%	Skeletal dysplasia, mild, with joint laxity and advanced bone age, 618870	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
CSNK1D	100%	100%	100%	98%	Advanced sleep-phase syndrome, familial, 2, 615224	MENDELIOME

CSNK1G1	100%	100%	100%	98%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
CSNK2A1	94%	94%	99%	98%	Okur-Chung neurodevelopmental syndrome, 617062	INTELLECTUAL DISABILITY MENDELIOME
CSNK2B	100%	100%	100%	99%	Poirier-Bienvenu neurodevelopmental syndrome, 618732	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
CSPP1	100%	100%	100%	97%	Joubert syndrome 21, 615636	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CSRP3	100%	100%	100%	99%	?Cardiomyopathy, dilated, 1M, 607482 Cardiomyopathy, hypertrophic, 12, 612124	HEART HYPERTROPHIC CARDIOMYOPATHY MENDELIOME
CST3	100%	100%	100%	97%	Cerebral amyloid angiopathy, 105150	MENDELIOME
CST6	100%	100%	100%	98%	?Ectodermal dysplasia 15, hypohidrotic/hair type, 618535	SKIN DISORDERS MENDELIOME
CSTA	100%	100%	100%	97%	Peeling skin syndrome 4, 607936	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CSTB	100%	100%	100%	95%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CTBP1	100%	99%	99%	96%	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS
CTC1	100%	100%	100%	98%	Cerebroretinal microangiopathy with calcifications and cysts, 612199	MOVEMENT DISORDERS HEREDITARY BONE MARROW FAILURE SKIN DISORDERS DYSKERATOSIS CONGENITA PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING TUMOR

CTCF	100%	100%	100%	99%	Intellectual developmental disorder, autosomal dominant 21, 615502	INTELLECTUAL DISABILITY MENDELIOME SCHISIS
CTDP1	100%	100%	100%	99%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168	VISION DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CTH	100%	100%	100%	98%	Cystathioninuria, 219500	METABOLIC DISORDERS MENDELIOME
CTHRC1	100%	100%	100%	98%	Barrett esophagus/esophageal adenocarcinoma, 614266	MENDELIOME
CTLA4	100%	100%	100%	98%	Immune dysregulation with autoimmunity, immunodeficiency, and lymphoproliferation, 616100	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME TUMOR
CTNNA1	100%	100%	100%	98%	Macular dystrophy, patterned, 2, 608970	VISION DISORDERS MENDELIOME TUMOR
CTNNA2	99%	99%	100%	99%	Cortical dysplasia, complex, with other brain malformations 9, 618174	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CTNNA3	99%	99%	100%	98%	Arrhythmogenic right ventricular dysplasia, familial, 13, 615616	HEART MENDELIOME
CTNNB1	100%	100%	100%	99%	Exudative vitreoretinopathy 7, 617572 Pilomatricoma, somatic, 132600 Colorectal cancer, somatic, 114500 Neurodevelopmental disorder with spastic diplegia and visual defects, 615075 Medulloblastoma, somatic, 155255 Ovarian cancer, somatic, 167000 Hepatocellular carcinoma, somatic, 114550	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME
CTNNBL1	100%	100%	100%	99%	?Immunodeficiency 99 with hypogammaglobulinemia and autoimmune cytopenias, 619846	PRIMARY IMMUNODEFICIENCY MENDELIOME
CTNND1	100%	100%	100%	98%	Blepharocheilodontic syndrome 2, 617681	INTELLECTUAL DISABILITY MENDELIOME SCHISIS
CTNND2	100%	99%	99%	95%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME

CTNS	100%	100%	99%	98%	Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, atypical nephropathic, 219800	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CTPS1	100%	100%	100%	99%	Immunodeficiency 24, 615897	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
CTR9	100%	100%	100%	97%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME TUMOR
CTSA	100%	100%	100%	98%	Galactosialidosis, 256540	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CTSB	100%	100%	100%	98%	No OMIM disease ID	SKIN DISORDERS MENDELIOME
CTSC	100%	100%	100%	98%	Periodontitis 1, juvenile, 170650 Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CTSD	100%	100%	100%	99%	Ceroid lipofuscinosis, neuronal, 10, 610127	MOVEMENT DISORDERS VISION DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CTSF	100%	100%	100%	98%	Ceroid lipofuscinosis, neuronal, 13 (Kufs type), 615362	MOVEMENT DISORDERS EPILEPSY MENDELIOME PRE CONCEPTION SCREENING
CTSH	100%	100%	100%	98%	No OMIM disease ID	VISION DISORDERS MENDELIOME

CTSK	100%	100%	100%	99%	Pycnodysostosis, 265800	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CTTNBP2	100%	100%	100%	98%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
CTU2	100%	100%	100%	99%	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CUBN	100%	100%	100%	99%	Imerslund-Grasbeck syndrome 1, 261100	HEREDITARY BONE MARROW FAILURE METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CUL3	100%	100%	100%	97%	Neurodevelopmental disorder w/wo autism or seizures, 619239 Pseudohypoaldosteronism, type IIE, 614496	INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME
CUL4B	100%	99%	97%	68%	Intellectual developmental disorder, X-linked syndromic, Cabezas type, 300354	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
CUL7	100%	100%	100%	99%	3-M syndrome 1, 273750	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
CUX1	100%	100%	99%	96%	Global developmental delay w/wo impaired intellectual development, 618330	INTELLECTUAL DISABILITY MENDELIOME
CUX2	100%	100%	99%	98%	Developmental and epileptic encephalopathy 67, 618141	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
CWC27	100%	100%	100%	96%	Retinitis pigmentosa w/wo skeletal anomalies, 250410	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CWF19L1	100%	100%	100%	98%	Spinocerebellar ataxia, autosomal recessive 17, 616127	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CXCR2	100%	100%	100%	99%	?WHIM syndrome 2, 619407	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCY MENDELIOME

CXCR4	100%	100%	100%	97%	WHIM syndrome 1, 193670 Myelokathexis, isolated, 193670	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME
CXorf56	100%	99%	98%	74%	?Intellectual developmental disorder, X-linked 107, 301013	INTELLECTUAL DISABILITY MENDELIOME
CYB561	100%	100%	100%	97%	Orthostatic hypotension 2, 618182	METABOLIC DISORDERS MENDELIOME
CYB5A	100%	100%	100%	99%	Methemoglobinemia and ambiguous genitalia, 250790	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRE CONCEPTION SCREENING
CYB5R3	100%	100%	100%	99%	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CYBA	100%	100%	100%	99%	Chronic granulomatous disease 4, autosomal recessive, 233690	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
CYBB	99%	98%	97%	72%	Immunodeficiency 34, mycobacteriosis, X-linked, 300645 Chronic granulomatous disease, X-linked, 306400	PRIMARY IMMUNODEFICIENCY MENDELIOME
CYBC1	100%	100%	100%	99%	Chronic granulomatous disease 5, autosomal recessive, 618935	PRIMARY IMMUNODEFICIENCY MENDELIOME
CYBRD1	100%	100%	100%	98%	No OMIM disease ID	IRON DISORDERS MENDELIOME
CYC1	100%	100%	100%	96%	Mitochondrial complex III deficiency, nuclear type 6, 615453	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
CYCS	100%	100%	100%	98%	Thrombocytopenia 4, 612004	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS
CYFIP2	100%	100%	100%	99%	Developmental and epileptic encephalopathy 65, 618008	INTELLECTUAL DISABILITY MENDELIOME
CYLD	100%	100%	100%	98%	Brooke-Spiegler syndrome, 605041 Cylindromatosis, familial, 132700 Trichoepithelioma, multiple familial, 1, 601606 ?Frontotemporal dementia and/or amyotrophic lateral sclerosis 8, 619132	SKIN DISORDERS MENDELIOME TUMOR
CYP11A1	100%	100%	100%	99%	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743	DISORDERS OF SEX DEVELOPMENT METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING

CYP11B1	100%	100%	100%	99%	Aldosteronism, glucocorticoid-remediable, 103900 Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010	DISORDERS OF SEX DEVELOPMENT METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CYP11B2	100%	100%	100%	98%	Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 Aldosterone to renin ratio raised,	DISORDERS OF SEX DEVELOPMENT METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CYP17A1	100%	100%	100%	99%	17,20-lyase deficiency, isolated, 202110 17-alpha-hydroxylase/17,20-lyase deficiency, 202110	DISORDERS OF SEX DEVELOPMENT METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY
CYP19A1	100%	99%	100%	98%	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300	DISORDERS OF SEX DEVELOPMENT METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY
CYP1B1	100%	100%	100%	98%	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 Anterior segment dysgenesis 6, multiple subtypes, 617315	VISION DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CYP21A2	100%	99%	100%	99%	Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910 Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910	DISORDERS OF SEX DEVELOPMENT METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CYP24A1	100%	100%	100%	98%	Hypercalcemia, infantile, 1, 143880	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CYP26B1	100%	100%	100%	97%	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
CYP26C1	100%	100%	100%	99%	Focal facial dermal dysplasia 4, 614974	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING

CYP27A1	100%	100%	100%	99%	Cerebrotendinous xanthomatosis, 213700	MOVEMENT DISORDERS NEUROPATHIES LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CYP27B1	100%	100%	100%	99%	Vitamin D-dependent rickets, type I, 264700	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CYP2A6	100%	100%	99%	94%	Coumarin resistance, 122700	MENDELIOME
CYP2B6	100%	100%	100%	97%	Efavirenz, poor metabolism of, 614546	MENDELIOME
CYP2C19	100%	100%	100%	98%	Proguanil poor metabolizer, 609535 Mephenytoin poor metabolizer, 609535 Clopidogrel, impaired responsiveness to, 609535 Omeprazole poor metabolizer, 609535	MENDELIOME
CYP2C8	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME PRE CONCEPTION SCREENING
CYP2C9	100%	99%	100%	98%	Warfarin sensitivity, 122700 Tolbutamide poor metabolizer,	MENDELIOME
CYP2R1	100%	100%	100%	96%	Rickets due to defect in vitamin D 25-hydroxylation deficiency, 600081	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CYP2U1	100%	100%	100%	96%	Spastic paraplegia 56, autosomal recessive, 615030	MOVEMENT DISORDERS NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
CYP3A4	100%	99%	100%	98%	Vitamin D-dependent rickets, type 3, 619073	MENDELIOME
CYP4F22	100%	100%	100%	99%	Ichthyosis, congenital, autosomal recessive 5, 604777	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
CYP4V2	100%	100%	100%	98%	Bietti crystalline corneoretinal dystrophy, 210370	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING

CYP7B1	100%	100%	100%	98%	Spastic paraplegia 5A, autosomal recessive, 270800 Bile acid synthesis defect, congenital, 3, 613812	MOVEMENT DISORDERS NEUROPATHIES LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
D2HGDH	100%	100%	100%	99%	D-2-hydroxyglutaric aciduria, 600721	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
DAAM2	100%	100%	100%	99%	Nephrotic syndrome, type 24, 619263	RENAL DISORDERS MENDELIOME
DAB1	100%	99%	100%	99%	Spinocerebellar ataxia 37, 615945	MENDELIOME
DACT1	100%	100%	100%	98%	Townes-Brocks syndrome 2, 617466	MENDELIOME
DAG1	100%	100%	100%	99%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
DALRD3	100%	100%	100%	99%	?Developmental and epileptic encephalopathy 86, 618910	MENDELIOME
DAO	100%	100%	100%	99%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME
DARS1	100%	100%	100%	97%	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
DARS2	100%	100%	100%	96%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105	MOVEMENT DISORDERS EPILEPSY NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS
DAZ1	50%	49%	46%	20%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
DAZ2	50%	49%	44%	17%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
DAZ3	49%	49%	42%	18%	No OMIM disease ID	MALE INFERTILITY MENDELIOME

DAZ4	49%	49%	42%	15%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
DBF4	100%	100%	99%	96%	No OMIM disease ID	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCY MENDELIOME
DBH	100%	100%	100%	99%	Orthostatic hypotension 1, due to DBH deficiency, 223360	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
DBR1	100%	100%	100%	98%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
DBT	100%	100%	100%	98%	Maple syrup urine disease, type II, 248600	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
DCAF12L1	100%	100%	99%	80%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
DCAF17	100%	100%	99%	98%	Woodhouse-Sakati syndrome, 241080	MOVEMENT DISORDERS SKIN DISORDERS HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
DCAF8	100%	100%	100%	99%	?Giant axonal neuropathy 2, autosomal dominant, 610100	NEUROPATHIES MENDELIOME
DCC	100%	100%	100%	98%	Mirror movements 1 and/or agenesis of the corpus callosum, 157600 Esophageal carcinoma, somatic, 133239 Colorectal cancer, somatic, 114500 Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542	MOVEMENT DISORDERS DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
DCDC2	100%	100%	100%	97%	Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212 Sclerosing cholangitis, neonatal, 617394	CILIOPATHIES HEARING IMPAIRMENT LIVER DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING

DCHS1	100%	100%	100%	99%	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390	HEART INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
DCLRE1C	100%	100%	100%	98%	Severe combined immunodeficiency, Athabascan type, 602450 Omenn syndrome, 603554	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)
DCN	95%	95%	100%	98%	Corneal dystrophy, congenital stromal, 610048	VISION DISORDERS MENDELIOME
DCPS	100%	100%	100%	98%	Al-Raqad syndrome, 616459	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
DCT	100%	100%	100%	98%	Oculocutaneous albinism, type VIII, 619165	VISION DISORDERS MENDELIOME
DCTN1	100%	100%	100%	99%	Neuronopathy, distal hereditary motor, type VIIB, 607641 Perry syndrome, 168605	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME PARKINSON
DCTN2	100%	100%	100%	98%	No OMIM disease ID	NEUROPATHIES MENDELIOME
DCX	98%	98%	98%	73%	Subcortical laminal heterotopia, X-linked, 300067 Lissencephaly, X-linked, 300067	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
DCXR	100%	100%	100%	99%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME
DDB1	100%	100%	100%	99%	White-Kernohan syndrome, 619426	INTELLECTUAL DISABILITY MENDELIOME
DDB2	100%	100%	100%	98%	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING TUMOR
DDC	100%	100%	100%	98%	Aromatic L-amino acid decarboxylase deficiency, 608643	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

DDHD1	100%	100%	100%	97%	Spastic paraplegia 28, autosomal recessive, 609340	MOVEMENT DISORDERS VISION DISORDERS METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
DDHD2	100%	100%	100%	98%	Spastic paraplegia 54, autosomal recessive, 615033	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
DDOST	100%	100%	100%	98%	Congenital disorder of glycosylation, type Ir, 614507	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
DDR2	100%	100%	100%	98%	Warburg-Cinotti syndrome, 618175 Spondylometaepiphyseal dysplasia, short limb-hand type, 271665	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
DDRGK1	100%	100%	100%	98%	Spondyloepimetaphyseal dysplasia, Shohat type, 602557	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
DDX11	100%	100%	100%	99%	Warsaw breakage syndrome, 613398	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING TUMOR
DDX23	100%	100%	100%	98%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
DDX3X	99%	98%	98%	71%	Intellectual developmental disorder, X-linked syndromic, Snijders Blok type, 300958	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME SCHISIS
DDX3Y	50%	50%	48%	21%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
DDX41	100%	100%	100%	99%	No OMIM disease ID	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCY MENDELIOME TUMOR
DDX58	100%	100%	100%	99%	Singleton-Merten syndrome 2, 616298	VISION DISORDERS PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME

DDX59	100%	100%	100%	98%	Orofaciodigital syndrome V, 174300	CILIOPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS
DDX6	100%	100%	100%	99%	Intellectual developmental disorder with impaired language and dysmorphic facies, 618653	INTELLECTUAL DISABILITY MENDELIOME
DEAF1	100%	100%	99%	95%	Vulto-van Silfout-de Vries syndrome, 615828 Neurodevelopmental disorder with hypotonia, impaired expressive language, and w/wo seizures, 617171	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
DEF6	100%	100%	100%	98%	Immunodeficiency 87 and autoimmunity, 619573	PRIMARY IMMUNODEFICIENCY MENDELIOME
DEGS1	100%	100%	100%	99%	Leukodystrophy, hypomyelinating, 18, 618404	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
DENND5A	100%	100%	100%	97%	Developmental and epileptic encephalopathy 49, 617281	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
DEPDC5	100%	100%	100%	99%	Epilepsy, familial focal, with variable foci 1, 604364	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
DES	100%	100%	100%	98%	Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419	ARRHYTHMOGENIC CARDIOMYOPATHY (ACM/ARVC) DILATED CARDIOMYOPATHY HEART HYPERTROPHIC CARDIOMYOPATHY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS MUSCLE DISORDERS
DGAT1	100%	100%	100%	99%	Diarrhea 7, protein-losing enteropathy type, 615863	METABOLIC DISORDERS MENDELIOME
DGAT2	100%	100%	100%	99%	No OMIM disease ID	NEUROPATHIES MENDELIOME
DGCR8	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME TUMOR

DGKE	100%	100%	100%	98%	Nephrotic syndrome, type 7, 615008	HEMOSTATIC/THROMBOTIC DISORDERS METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
DGUOK	100%	100%	100%	98%	Portal hypertension, noncirrhotic, 1, 617068 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880	LIVER DISORDERS METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS
DHCR24	100%	100%	100%	99%	Desmosterolosis, 602398	FETAL AKINESIA SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
DHCR7	100%	100%	100%	99%	Smith-Lemli-Opitz syndrome, 270400	FETAL AKINESIA SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS
DHDDS	94%	94%	100%	99%	Developmental delay and seizures w/wo movement abnormalities, 617836 ?Congenital disorder of glycosylation, type 1bb, 613861 Retinitis pigmentosa 59, 613861	MOVEMENT DISORDERS VISION DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
DHFR	100%	100%	100%	98%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
DHH	100%	100%	100%	99%	46XY gonadal dysgenesis with minifascicular neuropathy, 607080 46XY sex reversal 7, 233420	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRE CONCEPTION SCREENING

DHODH	100%	100%	100%	98%	Miller syndrome, 263750	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS
DHPS	96%	93%	100%	99%	Neurodevelopmental disorder with seizures and speech and walking impairment, 618480	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
DHTKD1	100%	100%	100%	98%	?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025 Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750	NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
DHX16	100%	100%	100%	98%	Neuromuscular disease and ocular or auditory anomalies w/wo seizures, 618733	INTELLECTUAL DISABILITY MENDELIOME
DHX30	100%	100%	100%	98%	Neurodevelopmental disorder with variable motor and speech impairment, 617804	INTELLECTUAL DISABILITY MENDELIOME
DHX37	100%	100%	100%	99%	Neurodevelopmental disorder with brain anomalies and w/wo vertebral or cardiac anomalies, 618731 46, XY sex reversal 11, 273250	DISORDERS OF SEX DEVELOPMENT INTELLECTUAL DISABILITY MENDELIOME
DHX38	100%	100%	100%	99%	Retinitis pigmentosa 84, 618220	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
DIABLO	100%	100%	100%	98%	Deafness, autosomal dominant 64, 614152	HEARING IMPAIRMENT MENDELIOME
DIAPH1	100%	100%	99%	95%	Deafness, autosomal dominant 1, w/wo thrombocytopenia, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632	HEARING IMPAIRMENT EPILEPSY HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
DIAPH2	100%	99%	97%	70%	?Premature ovarian failure 2A, 300511	MENDELIOME
DIAPH3	100%	99%	100%	98%	Auditory neuropathy, autosomal dominant 1, 609129	HEARING IMPAIRMENT MENDELIOME
DICER1	100%	100%	100%	98%	Pleuropulmonary blastoma, 601200 Goiter, multinodular 1, w/wo Sertoli-Leydig cell tumors, 138800 GLOW syndrome, somatic mosaic, 618272 Rhabdomyosarcoma, embryonal, 2, 180295	HEREDITARY BONE MARROW FAILURE MENDELIOME TUMOR

DIP2B	100%	100%	100%	99%	Intellectual developmental disorder, autosomal dominant, FRA12A type, 136630	INTELLECTUAL DISABILITY MENDELIOME
DIS3L2	100%	100%	100%	98%	Perlman syndrome, 267000	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING TUMOR
DISP1	100%	100%	100%	98%	No OMIM disease ID	CRANIOFACIAL ANOMALIES MENDELIOME
DKC1	100%	100%	98%	73%	Dyskeratosis congenita, X-linked, 305000	VISION DISORDERS HEREDITARY BONE MARROW FAILURE SKIN DISORDERS DYSKERATOSIS CONGENITA PRIMARY IMMUNODEFICIENCY LIVER DISORDERS INTELLECTUAL DISABILITY MENDELIOME TUMOR
DLAT	100%	100%	100%	98%	Pyruvate dehydrogenase E2 deficiency, 245348	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
DLC1	100%	100%	100%	98%	Colorectal cancer, somatic, 114500	RENAL DISORDERS MENDELIOME
DLD	100%	100%	100%	98%	Dihydrolipoamide dehydrogenase deficiency, 246900	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
DLG3	100%	99%	97%	70%	Intellectual developmental disorder, X-linked 90, 300850	INTELLECTUAL DISABILITY MENDELIOME
DLG4	100%	100%	100%	99%	Intellectual developmental disorder, autosomal dominant 62, 618793	ANEURYSM INTELLECTUAL DISABILITY MENDELIOME
DLK1	100%	100%	100%	99%	No OMIM disease ID	DISORDERS OF SEX DEVELOPMENT MENDELIOME
DLL1	100%	100%	100%	98%	Neurodevelopmental disorder with nonspecific brain abnormalities and w/wo seizures, 618709	HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME

DLL3	100%	100%	100%	97%	Spondylocostal dysostosis 1, autosomal recessive, 277300	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
DLL4	100%	100%	100%	99%	Adams-Oliver syndrome 6, 616589	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS
DLST	100%	100%	100%	98%	Paragangliomas 7, 618475	MENDELIOME MITOCHONDRIAL DISORDERS TUMOR
DLX3	100%	100%	100%	98%	Trichodontoosseous syndrome, 190320 Amelogenesis imperfecta, type IV, 104510	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
DLX4	100%	100%	100%	99%	?Orofacial cleft 15, 616788	CRANIOFACIAL ANOMALIES MENDELIOME
DLX5	100%	100%	100%	99%	Split-hand/foot malformation 1, 183600 ?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600	SKIN DISORDERS HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
DLX6	100%	100%	100%	95%	No OMIM disease ID	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
DMAC1	100%	100%	100%	96%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
DMAC2	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
DMAC2L	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
DMC1	100%	100%	100%	97%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
DMD	99%	99%	97%	72%	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200	DILATED CARDIOMYOPATHY HEART INTELLECTUAL DISABILITY MENDELIOME MUSCLE DISORDERS

DMGDH	100%	100%	100%	98%	Dimethylglycine dehydrogenase deficiency, 605850	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
DMP1	100%	100%	100%	99%	Hypophosphatemic rickets, AR, 241520	SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
DMPK	100%	100%	100%	98%	Myotonic dystrophy 1, 160900	INTELLECTUAL DISABILITY MENDELIOME
DMRT1	100%	100%	100%	99%	No OMIM disease ID	DISORDERS OF SEX DEVELOPMENT MALE INFERTILITY MENDELIOME
DMRT2	100%	100%	100%	97%	No OMIM disease ID	DISORDERS OF SEX DEVELOPMENT MENDELIOME
DMXL2	100%	100%	100%	98%	Developmental and epileptic encephalopathy 81, 618663 ?Deafness, autosomal dominant 71, 617605 ?Polyendocrine-polyneuropathy syndrome, 616113	HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
DNA2	100%	100%	100%	97%	?Seckel syndrome 8, 615807 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS
DNAAF1	100%	100%	100%	99%	Ciliary dyskinesia, primary, 13, 613193	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING
DNAAF2	100%	100%	100%	96%	Ciliary dyskinesia, primary, 10, 612518	CILIOPATHIES MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING
DNAAF3	100%	100%	99%	97%	Ciliary dyskinesia, primary, 2, 606763	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING
DNAAF4	100%	100%	100%	96%	Ciliary dyskinesia, primary, 25, 615482	CILIOPATHIES MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING

DNAAF5	100%	99%	99%	96%	Ciliary dyskinesia, primary, 18, 614874	CILIOPATHIES MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING
DNAH1	100%	100%	100%	99%	Spermatogenic failure 18, 617576 ?Ciliary dyskinesia, primary, 37, 617577	CILIOPATHIES MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING
DNAH10	100%	100%	100%	98%	Spermatogenic failure 56, 619515	MENDELIOME
DNAH11	100%	100%	100%	98%	Ciliary dyskinesia, primary, 7, w/wo situs inversus, 611884	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING
DNAH17	100%	100%	100%	99%	Spermatogenic failure 39, 618643	CILIOPATHIES MALE INFERTILITY MENDELIOME
DNAH2	100%	99%	100%	98%	Spermatogenic failure 45, 619094	MENDELIOME
DNAH5	99%	99%	100%	98%	Ciliary dyskinesia, primary, 3, w/wo situs inversus, 608644	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING
DNAH7	100%	100%	100%	98%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
DNAH8	100%	99%	100%	97%	Spermatogenic failure 46, 619095	CILIOPATHIES MALE INFERTILITY MENDELIOME
DNAH9	100%	100%	100%	98%	Ciliary dyskinesia, primary, 40, 618300	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING
DNAI1	100%	100%	100%	99%	Ciliary dyskinesia, primary, 1, w/wo situs inversus, 244400	CILIOPATHIES MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING
DNAI2	100%	100%	100%	97%	Ciliary dyskinesia, primary, 9, w/wo situs inversus, 612444	CILIOPATHIES MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING
DNAJA3	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
DNAJB11	100%	100%	100%	97%	Polycystic kidney disease 6 w/wo polycystic liver disease, 618061	LIVER DISORDERS RENAL DISORDERS MENDELIOME

DNAJB13	100%	100%	100%	98%	Ciliary dyskinesia, primary, 34, 617091	CILIOPATHIES MALE INFERTILITY MENDELIOME
DNAJB2	100%	100%	100%	98%	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING
DNAJB5	100%	100%	100%	99%	No OMIM disease ID	NEUROPATHIES MENDELIOME
DNAJB6	100%	100%	100%	98%	Muscular dystrophy, limb-girdle, autosomal dominant 1, 603511	MENDELIOME MUSCLE DISORDERS
DNAJC12	100%	100%	100%	97%	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
DNAJC19	100%	100%	100%	98%	3-methylglutaconic aciduria, type V, 610198	HEART METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
DNAJC21	100%	100%	99%	95%	Bone marrow failure syndrome 3, 617052	HEREDITARY BONE MARROW FAILURE SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING TUMOR
DNAJC3	100%	100%	99%	97%	Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
DNAJC30	100%	100%	100%	99%	Leber hereditary optic neuropathy, autosomal recessive, 619382	VISION DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS
DNAJC5	100%	100%	100%	99%	Ceroid lipofuscinosis, neuronal, 4 (Kufs type), autosomal dominant, 162350	EPILEPSY MENDELIOME
DNAJC6	100%	100%	100%	98%	Parkinson disease 19a, juvenile-onset, 615528 Parkinson disease 19b, early-onset, 615528	MENDELIOME PARKINSON PRE CONCEPTION SCREENING

DNAL1	100%	100%	100%	98%	Ciliary dyskinesia, primary, 16, 614017	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING
DNAL4	100%	100%	100%	98%	?Mirror movements 3, 616059	MOVEMENT DISORDERS MENDELIOME
DNASE1	100%	100%	100%	99%	No OMIM disease ID	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME
DNASE1L3	100%	100%	100%	98%	Systemic lupus erythematosus 16, 614420	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
DNASE2	100%	100%	100%	98%	Autoinflammatory-pancytopenia syndrome, 619858	PRIMARY IMMUNODEFICIENCY MENDELIOME
DNHD1	100%	100%	100%	99%	Spermatogenic failure 65, 619712	MALE INFERTILITY MENDELIOME
DNM1	100%	100%	100%	98%	Developmental and epileptic encephalopathy 31, 616346	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
DNM1L	100%	100%	100%	98%	Optic atrophy 5, 610708 Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388	MOVEMENT DISORDERS VISION DISORDERS EPILEPSY NEUROPATHIES HNPD METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
DNM2	100%	100%	100%	98%	Centronuclear myopathy 1, 160150 Charcot-Marie-Tooth disease, axonal type 2M, 606482 Charcot-Marie-Tooth disease, dominant intermediate B, 606482 Lethal congenital contracture syndrome 5, 615368	FETAL AKINESIA NEUROPATHIES METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS
DNMBP	100%	100%	100%	99%	Cataract 48, 618415	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING

DNMT1	99%	99%	100%	99%	Neuropathy, hereditary sensory, type IE, 614116 Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121	MOVEMENT DISORDERS NEUROPATHIES HNPD METABOLIC DISORDERS MENDELIOME
DNMT3A	100%	100%	100%	99%	Tatton-Brown-Rahman syndrome, 615879 Acute myeloid leukemia, somatic, 601626 Heyn-Sproul-Jackson syndrome, 618724	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
DNMT3B	100%	100%	100%	99%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 Facioscapulohumeral muscular dystrophy 4, digenic, 619478	PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
DOCK2	99%	99%	100%	99%	Immunodeficiency 40, 616433	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)
DOCK3	100%	100%	100%	98%	Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
DOCK6	100%	100%	100%	98%	Adams-Oliver syndrome 2, 614219	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS
DOCK7	100%	100%	100%	98%	Developmental and epileptic encephalopathy 23, 615859	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
DOCK8	100%	100%	100%	98%	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)
DOHH	100%	100%	100%	99%	Neurodevelopmental disorder with microcephaly, cerebral atrophy, and visual impairment, 620066	INTELLECTUAL DISABILITY MENDELIOME

DOK7	100%	100%	100%	98%	Fetal akinesia deformation sequence 3, 618389 Myasthenic syndrome, congenital, 10, 254300	FETAL AKINESIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
DOLK	100%	100%	100%	98%	Congenital disorder of glycosylation, type Im, 610768	SKIN DISORDERS HEART METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
DONSON	100%	100%	100%	97%	Microcephaly, short stature, and limb abnormalities, 617604 Microcephaly-micromelia syndrome, 251230	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
DPAGT1	100%	100%	100%	99%	Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750 Congenital disorder of glycosylation, type Ij, 608093	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
DPCD	100%	100%	100%	98%	No OMIM disease ID	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
DPF2	100%	100%	100%	99%	Coffin-Siris syndrome 7, 618027	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
DPH1	100%	100%	100%	98%	Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
DPH5	100%	100%	100%	98%	Neurodevelopmental disorder with short stature, prominent forehead, and feeding difficulties, 620070	INTELLECTUAL DISABILITY MENDELIOME
DPM1	99%	96%	100%	98%	Congenital disorder of glycosylation, type Ie, 608799	EPILEPSY SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS

DPM2	100%	100%	100%	99%	Congenital disorder of glycosylation, type Iu, 615042	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
DPM3	100%	100%	100%	95%	?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937	HEART METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
DPP6	100%	99%	100%	98%	Intellectual developmental disorder, autosomal dominant 33, 616311	HEART INTELLECTUAL DISABILITY MENDELIOME
DPY19L2	100%	100%	99%	95%	Spermatogenic failure 9, 613958	MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING
DPYD	99%	99%	100%	98%	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
DPYS	100%	100%	100%	98%	Dihydropyrimidinuria, 222748	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
DPYSL2	100%	100%	100%	97%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
DPYSL5	100%	100%	100%	98%	Ritscher-Schinzel syndrome 4, 619435	INTELLECTUAL DISABILITY MENDELIOME
DRAM2	100%	100%	100%	98%	Cone-rod dystrophy 21, 616502	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
DRC1	100%	100%	100%	98%	Spermatogenic failure 80, 620222 Ciliary dyskinesia, primary, 21, 615294	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING
DRD4	100%	100%	99%	97%	Autonomic nervous system dysfunction,	MENDELIOME
DRP2	100%	99%	98%	72%	No OMIM disease ID	NEUROPATHIES MENDELIOME

DSC2	100%	100%	100%	98%	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 Arrhythmogenic right ventricular dysplasia 11, 610476	ARRHYTHMOGENIC CARDIOMYOPATHY (ACM/ARVC) SKIN DISORDERS HEART MENDELIOME PRE CONCEPTION SCREENING ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS
DSC3	100%	100%	100%	98%	Hypotrichosis and recurrent skin vesicles, 613102	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
DSE	100%	100%	100%	98%	Ehlers-Danlos syndrome, musculocontractural type 2, 615539	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
DSG1	100%	100%	100%	98%	Keratosis palmoplantaris striata I, AD, 148700 Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508	SKIN DISORDERS MENDELIOME
DSG2	100%	100%	100%	99%	Cardiomyopathy, dilated, 1BB, 612877 Arrhythmogenic right ventricular dysplasia 10, 610193	ARRHYTHMOGENIC CARDIOMYOPATHY (ACM/ARVC) HEART MENDELIOME ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS
DSG3	100%	100%	100%	98%	Blistering, acantholytic, of oral and laryngeal mucosa, 619226	SKIN DISORDERS MENDELIOME
DSG4	100%	100%	100%	98%	Hypotrichosis 6, 607903	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
DSP	100%	100%	100%	98%	Arrhythmogenic right ventricular dysplasia 8, 607450 Skin fragility-woolly hair syndrome, 607655 Epidermolysis bullosa, lethal acantholytic, 609638 Keratosis palmoplantaris striata II, 612908 Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821 Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676	ARRHYTHMOGENIC CARDIOMYOPATHY (ACM/ARVC) DILATED CARDIOMYOPATHY SKIN DISORDERS HEART MENDELIOME PRE CONCEPTION SCREENING ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS
DSPP	100%	100%	98%	95%	Dentinogenesis imperfecta, Shields type III, 125500 Dentinogenesis imperfecta, Shields type II, 125490 Dentin dysplasia, type II, 125420 Deafness, autosomal dominant 39, with dentinogenesis, 605594	CRANIOFACIAL ANOMALIES SKIN DISORDERS HEARING IMPAIRMENT MENDELIOME

DST	100%	100%	100%	98%	Neuropathy, hereditary sensory and autonomic, type VI, 614653 Epidermolysis bullosa simplex 3, localized or generalized intermediate, with bp230 deficiency, 615425	SKIN DISORDERS NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING
DSTYK	100%	100%	100%	98%	Congenital anomalies of kidney and urinary tract 1, 610805 Spastic paraplegia 23, 270750	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
DTNA	100%	100%	100%	98%	Left ventricular noncompaction 1, w/wo congenital heart defects, 604169	HEART MENDELIOME
DTNBP1	100%	100%	99%	97%	Hermansky-Pudlak syndrome 7, 614076	VISION DISORDERS SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
DTYMK	100%	100%	100%	99%	Neurodegeneration, childhood-onset, with progressive microcephaly, 619847	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
DUOX2	100%	100%	100%	98%	Thyroid dyshormonogenesis 6, 607200	MENDELIOME PRE CONCEPTION SCREENING
DUOXA2	100%	100%	100%	99%	Thyroid dyshormonogenesis 5, 274900	MENDELIOME PRE CONCEPTION SCREENING
DUSP6	100%	100%	100%	98%	Hypogonadotropic hypogonadism 19 w/wo anosmia, 615269	SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME
DVL1	100%	100%	99%	97%	Robinow syndrome, autosomal dominant 2, 616331	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS
DVL3	100%	100%	100%	98%	Robinow syndrome, autosomal dominant 3, 616894	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS
DYM	100%	99%	100%	98%	Smith-McCort dysplasia, 607326 Dyggve-Melchior-Clausen disease, 223800	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

DYNC1H1	100%	100%	100%	98%	Charcot-Marie-Tooth disease, axonal, type 2O, 614228 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 Intellectual developmental disorder, autosomal dominant 13, 614563	EPILEPSY NEUROPATHIES HNPD INTELLECTUAL DISABILITY MENDELIOME MUSCLE DISORDERS
DYNC1I2	100%	100%	100%	98%	Neurodevelopmental disorder with microcephaly and structural brain anomalies, 618492	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
DYNC2H1	99%	99%	100%	97%	Short-rib thoracic dysplasia 3 w/wo polydactyly, 613091	VISION DISORDERS CILIOPATHIES DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS
DYNC2LI1	100%	100%	100%	97%	Short-rib thoracic dysplasia 15 with polydactyly, 617088	CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS
DYRK1A	100%	100%	100%	98%	Intellectual developmental disorder, autosomal dominant 7, 614104	CONGENITAL HEART DISEASE EPILEPSY HEART INTELLECTUAL DISABILITY MENDELIOME
DYRK1B	100%	100%	100%	98%	Abdominal obesity-metabolic syndrome 3, 615812	MENDELIOME
DYSF	100%	100%	100%	99%	Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601 Miyoshi muscular dystrophy 1, 254130 Myopathy, distal, with anterior tibial onset, 606768	MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
DZIP1	100%	100%	100%	97%	Spermatogenic failure 47, 619102 ?Mitral valve prolapse 3, 610840	HEART MENDELIOME
DZIP1L	100%	100%	100%	99%	Polycystic kidney disease 5, 617610	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
E2F1	100%	99%	99%	92%	No OMIM disease ID	MALE INFERTILITY MENDELIOME

EARS2	100%	100%	100%	98%	Combined oxidative phosphorylation deficiency 12, 614924	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
EBF3	100%	100%	100%	96%	Hypotonia, ataxia, and delayed development syndrome, 617330	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME
EBP	100%	100%	99%	74%	MEND syndrome, 300960 Chondrodysplasia punctata, X-linked dominant, 302960	SKIN DISORDERS EPILEPSY SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME SCHISIS
ECE1	100%	100%	100%	98%	?Hirschsprung disease, cardiac defects, and autonomic dysfunction, 613870	MENDELIOME
ECEL1	100%	100%	100%	99%	Arthrogryposis, distal, type 5D, 615065	FETAL AKINESIA SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
ECHS1	100%	100%	100%	96%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
ECM1	100%	100%	100%	98%	Urbach-Wiethe disease, 247100	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ECSIT	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
EDA	100%	99%	96%	66%	Tooth agenesis, selective, X-linked 1, 313500 Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME
EDAR	100%	100%	100%	98%	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490 Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING

EDARADD	100%	100%	100%	98%	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941 Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
EDC3	100%	100%	100%	99%	?Intellectual developmental disorder, autosomal recessive 50, 616460	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
EDEM3	100%	100%	100%	98%	Congenital disorder of glycosylation, type IIv, 619493	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
EDN1	100%	100%	100%	99%	Question mark ears, isolated, 612798 Auriculocondylar syndrome 3, 615706	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS
EDN3	100%	100%	100%	99%	Waardenburg syndrome, type 4B, 613265	SKIN DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
EDNRA	100%	100%	100%	98%	Mandibulofacial dysostosis with alopecia, 616367	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS
EDNRB	100%	100%	100%	98%	?ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580	SKIN DISORDERS HEARING IMPAIRMENT LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING
EED	100%	100%	99%	94%	Cohen-Gibson syndrome, 617561	INTELLECTUAL DISABILITY MENDELIOME
EEF1A2	100%	100%	100%	98%	Developmental and epileptic encephalopathy 33, 616409 Intellectual developmental disorder, autosomal dominant 38, 616393	EPILEPSY HEART INTELLECTUAL DISABILITY MENDELIOME
EEF1AKNMT	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME
EEF1D	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
EEF2	100%	100%	100%	99%	?Spinocerebellar ataxia 26, 609306	MENDELIOME

EFEMP1	100%	100%	100%	99%	Doyne honeycomb degeneration of retina, 126600	VISION DISORDERS MENDELIOME
EFEMP2	100%	100%	100%	99%	Cutis laxa, autosomal recessive, type IB, 614437	ANEURYSM SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
EFHC1	97%	97%	100%	98%	No OMIM disease ID	EPILEPSY MENDELIOME
EFL1	100%	100%	100%	99%	Shwachman-Diamond syndrome 2, 617941	HEREDITARY BONE MARROW FAILURE SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
EFNA4	100%	100%	100%	98%	No OMIM disease ID	CRANIOFACIAL ANOMALIES MENDELIOME
EFNB1	100%	99%	98%	74%	Craniofrontonasal dysplasia, 304110	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS
EFNB2	100%	100%	100%	99%	No OMIM disease ID	HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME
EFTUD2	100%	100%	100%	99%	Mandibulofacial dysostosis, Guion-Almeida type, 610536	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS
EGF	100%	100%	100%	98%	?Hypomagnesemia 4, renal, 611718	EPILEPSY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
EGFR	100%	100%	100%	99%	?Inflammatory skin and bowel disease, neonatal, 2, 616069 Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980 Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980	MENDELIOME PRE CONCEPTION SCREENING TUMOR
EGLN1	100%	100%	99%	84%	Erythrocytosis, familial, 3, 609820	MENDELIOME TUMOR
EGLN2	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME TUMOR

EGR2	100%	100%	100%	98%	Dejerine-Sottas disease, 145900 Charcot-Marie-Tooth disease, type 1D, 607678 Hypomyelinating neuropathy, congenital, 1, 605253	FETAL AKINESIA NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING
EHD1	100%	100%	100%	99%	No OMIM disease ID	HEARING IMPAIRMENT MENDELIOME
EHHADH	100%	100%	100%	99%	?Fanconi renotubular syndrome 3, 615605	RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS
EHMT1	100%	99%	99%	98%	Kleefstra syndrome 1, 610253	CONGENITAL HEART DISEASE EPILEPSY HEART INTELLECTUAL DISABILITY MENDELIOME
EIF1AY	50%	50%	47%	19%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
EIF2AK1	100%	100%	100%	97%	?Leukoencephalopathy, motor delay, spasticity, and dysarthria syndrome, 618878	INTELLECTUAL DISABILITY MENDELIOME
EIF2AK2	100%	100%	100%	97%	Leukoencephalopathy, developmental delay, and episodic neurologic regression syndrome, 618877 Dystonia 33, 619687	INTELLECTUAL DISABILITY MENDELIOME
EIF2AK3	100%	100%	100%	98%	Wolcott-Rallison syndrome, 226980	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
EIF2AK4	100%	100%	100%	98%	Pulmonary venoocclusive disease 2, 234810	MENDELIOME PRE CONCEPTION SCREENING
EIF2B1	100%	100%	100%	99%	Leukoencephalopathy with vanishing white matter, 603896	MOVEMENT DISORDERS EPILEPSY MENDELIOME PRE CONCEPTION SCREENING
EIF2B2	100%	100%	100%	98%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896	MOVEMENT DISORDERS EPILEPSY MENDELIOME PRE CONCEPTION SCREENING
EIF2B3	100%	100%	100%	97%	Leukoencephalopathy with vanishing white matter, 603896	MOVEMENT DISORDERS EPILEPSY MENDELIOME PRE CONCEPTION SCREENING

EIF2B4	100%	100%	100%	99%	Ovarioleukodystrophy, 603896 Leukoencephalopathy with vanishing white matter, 603896	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
EIF2B5	100%	100%	100%	98%	Ovarioleukodystrophy, 603896 Leukoencephalopathy with vanishing white matter, 603896	MOVEMENT DISORDERS DISORDERS OF SEX DEVELOPMENT EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY
EIF2S3	100%	100%	97%	70%	MEHMO syndrome, 300148	INTELLECTUAL DISABILITY MENDELIOME SCHISIS
EIF3F	100%	100%	100%	99%	Intellectual developmental disorder, autosomal recessive 67, 618295	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
EIF4A2	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
EIF4A3	100%	100%	100%	98%	Robin sequence with cleft mandible and limb anomalies, 268305	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS
EIF5A	100%	100%	100%	97%	Faundes-Banka syndrome, 619376	INTELLECTUAL DISABILITY MENDELIOME
ELAC2	100%	100%	100%	99%	Combined oxidative phosphorylation deficiency 17, 615440	HEART INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
ELANE	100%	100%	100%	99%	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCY MENDELIOME TUMOR
ELF2	100%	100%	100%	98%	No OMIM disease ID	NEUROPATHIES MENDELIOME
ELF4	100%	99%	98%	73%	Autoinflammatory syndrome, familial, X-linked, Behcet-like 2, 301074	PRIMARY IMMUNODEFICIENCY MENDELIOME

ELMO2	100%	100%	100%	98%	Vascular malformation, primary intraosseous, 606893	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
ELMOD3	100%	100%	100%	99%	?Deafness, autosomal recessive 88, 615429 ?Deafness, autosomal dominant 81, 619500	HEARING IMPAIRMENT MENDELIOME
ELN	100%	100%	100%	98%	Cutis laxa, autosomal dominant, 123700 Supravalvar aortic stenosis, 185500	ANEURYSM CONGENITAL HEART DISEASE SKIN DISORDERS HEART MENDELIOME
ELOVL1	100%	100%	100%	99%	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527	VISION DISORDERS SKIN DISORDERS HEARING IMPAIRMENT METABOLIC DISORDERS MENDELIOME
ELOVL4	100%	100%	99%	97%	Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110 Ichthyosis, spastic quadriplegia, and impaired intellectual development, 614457	MOVEMENT DISORDERS VISION DISORDERS SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ELOVL5	100%	100%	100%	98%	Spinocerebellar ataxia 38, 615957	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME
ELP1	100%	100%	100%	99%	Dysautonomia, familial, 223900	NEUROPATHIES HNPD MENDELIOME PRE CONCEPTION SCREENING SONIC HEDGEHOG MEDULLOBLASTOMA TUMOR
ELP2	100%	100%	100%	98%	Intellectual developmental disorder, autosomal recessive 58, 617270	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ELP4	87%	87%	100%	97%	?Aniridia 2, 617141	MENDELIOME
EMC1	100%	100%	100%	98%	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
EMC10	100%	100%	100%	98%	Neurodevelopmental disorder with dysmorphic facies and variable seizures, 619264	INTELLECTUAL DISABILITY MENDELIOME

EMD	100%	99%	98%	70%	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300	HEART MENDELIOME MITOCHONDRIAL DISORDERS MUSCLE DISORDERS
EMG1	100%	100%	100%	98%	Bowen-Conradi syndrome, 211180	MENDELIOME PRE CONCEPTION SCREENING
EMILIN1	100%	100%	100%	99%	Neuronopathy, distal hereditary motor, type X, 620080	ANEURYSM NEUROPATHIES MENDELIOME
EML1	100%	100%	99%	97%	Band heterotopia, 600348	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
EMP2	100%	100%	100%	98%	Nephrotic syndrome, type 10, 615861	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
EMX2	100%	100%	99%	92%	Schizencephaly, 269160	INTELLECTUAL DISABILITY MENDELIOME
EN1	100%	99%	99%	78%	?ENDOVE syndrome, limb-brain type, 619218	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
ENAM	100%	100%	100%	97%	Amelogenesis imperfecta, type IC, 204650 Amelogenesis imperfecta, type IB, 104500	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ENG	100%	100%	100%	98%	Telangiectasia, hereditary hemorrhagic, type 1, 187300	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME
ENO3	100%	100%	100%	99%	Glycogen storage disease XIII, 612932	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
ENPP1	100%	99%	100%	97%	Hypophosphatemic rickets, autosomal recessive, 2, 613312 Arterial calcification, generalized, of infancy, 1, 208000 Cole disease, 615522	SKIN DISORDERS HEART SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ENTPD1	100%	100%	100%	98%	Spastic paraplegia 64, autosomal recessive, 615683	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

EOGT	98%	94%	100%	99%	Adams-Oliver syndrome 4, 615297	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS
EP300	100%	100%	100%	98%	Menke-Hennekam syndrome 2, 618333 Colorectal cancer, somatic, 114500 Rubinstein-Taybi syndrome 2, 613684	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
EPAS1	100%	100%	100%	98%	Erythrocytosis, familial, 4, 611783	MENDELIOME TUMOR
EPB41	100%	100%	100%	98%	Elliptocytosis-1, 611804	MENDELIOME PRE CONCEPTION SCREENING
EPB41L1	100%	100%	100%	99%	?Intellectual developmental disorder, autosomal dominant 11, 614257	MENDELIOME
EPB42	100%	100%	100%	99%	Spherocytosis, type 5, 612690	MENDELIOME PRE CONCEPTION SCREENING
EPCAM	100%	100%	100%	98%	Diarrhea 5, with tufting enteropathy, congenital, 613217 Lynch syndrome 8, 613244	MENDELIOME PRE CONCEPTION SCREENING TUMOR
EPG5	100%	100%	100%	98%	Vici syndrome, 242840	VISION DISORDERS SKIN DISORDERS PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING SCHISIS
EPHA2	100%	100%	100%	99%	Cataract 6, multiple types, 116600	VISION DISORDERS MENDELIOME
EPHA7	100%	100%	100%	98%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
EPHB2	100%	99%	99%	97%	?Bleeding disorder, platelet-type, 22, 618462	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME
EPHB4	100%	100%	100%	99%	Capillary malformation-arteriovenous malformation 2, 618196 Lymphatic malformation 7, 617300	SKIN DISORDERS MENDELIOME
EPHX1	100%	100%	100%	99%	No OMIM disease ID	LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING

EPHX2	100%	100%	100%	98%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME
EPM2A	100%	100%	99%	90%	Epilepsy, progressive myoclonic 2A (Lafora), 254780	EPILEPSY MENDELIOME PRE CONCEPTION SCREENING
EPO	100%	100%	100%	98%	Erythrocytosis, familial, 5, 617907 ?Diamond-Blackfan anemia-like, 617911	HEREDITARY BONE MARROW FAILURE MENDELIOME PRE CONCEPTION SCREENING
EPRS1	100%	100%	100%	98%	Leukodystrophy, hypomyelinating, 15, 617951	MENDELIOME PRE CONCEPTION SCREENING
EPS8	100%	100%	100%	98%	?Deafness, autosomal recessive 102, 615974	HEARING IMPAIRMENT MENDELIOME
EPS8L2	100%	100%	100%	95%	Deafness autosomal recessive 106, 617637	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
EPS8L3	100%	100%	100%	99%	?Hypotrichosis 5, 612841	SKIN DISORDERS MENDELIOME
ERAL1	100%	100%	100%	98%	Perrault syndrome 6, 617565	HEARING IMPAIRMENT DISORDERS OF SEX DEVELOPMENT MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY
ERBB2	100%	100%	100%	99%	Gastric cancer, somatic, 613659 Adenocarcinoma of lung, somatic, 211980 Ovarian cancer, somatic, 167000 ?Visceral neuropathy, familial, 2, autosomal recessive, 619465 Glioblastoma, somatic, 137800	NEUROPATHIES MENDELIOME
ERBB3	100%	100%	100%	99%	?Lethal congenital contractural syndrome 2, 607598 Visceral neuropathy, familial, 1, autosomal recessive, 243180	FETAL AKINESIA NEUROPATHIES LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
ERBB4	100%	99%	100%	98%	Amyotrophic lateral sclerosis 19, 615515	ALS MENDELIOME
ERBIN	100%	100%	100%	98%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
ERCC1	100%	100%	100%	98%	Cerebrooculofacioskeletal syndrome 4, 610758	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING TUMOR

ERCC2	100%	100%	100%	99%	Xeroderma pigmentosum, group D, 278730 Trichothiodystrophy 1, photosensitive, 601675 ?Cerebrooculofacioskeletal syndrome 2, 610756	MOVEMENT DISORDERS VISION DISORDERS SKIN DISORDERS PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING TUMOR
ERCC3	100%	100%	100%	98%	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651	VISION DISORDERS SKIN DISORDERS PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING TUMOR
ERCC4	100%	100%	100%	97%	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 XFE progeroid syndrome, 610965 Xeroderma pigmentosum, group F, 278760 Fanconi anemia, complementation group Q, 615272	MOVEMENT DISORDERS HEREDITARY BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING TUMOR
ERCC5	100%	100%	100%	98%	Xeroderma pigmentosum, group G, 278780 Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780	FETAL AKINESIA SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING TUMOR
ERCC6	100%	100%	100%	98%	UV-sensitive syndrome 1, 600630 Cerebrooculofacioskeletal syndrome 1, 214150 ?De Sanctis-Cacchione syndrome, 278800 Cockayne syndrome, type B, 133540 Premature ovarian failure 11, 616946	FETAL AKINESIA VISION DISORDERS SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING TUMOR
ERCC6L2	100%	99%	100%	98%	Bone marrow failure syndrome 2, 615715	HEREDITARY BONE MARROW FAILURE MENDELIOME PRE CONCEPTION SCREENING
ERCC8	100%	100%	100%	97%	UV-sensitive syndrome 2, 614621 Cockayne syndrome, type A, 216400	VISION DISORDERS SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

ERF	100%	100%	100%	99%	Craniosynostosis 4, 600775 Chitayat syndrome, 617180	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
ERGIC1	97%	97%	100%	98%	?Arthrogryposis multiplex congenita 2, neurogenic type, 208100	MENDELIOME
ERLIN1	100%	100%	100%	98%	Spastic paraplegia 62, 615681	MENDELIOME PRE CONCEPTION SCREENING
ERLIN2	100%	100%	100%	98%	Spastic paraplegia 18, autosomal recessive, 611225	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ERMARD	100%	100%	100%	98%	?Periventricular nodular heterotopia 6, 615544	MENDELIOME
ESCO2	100%	100%	100%	97%	Juberg-Hayward syndrome, 216100 Roberts-SC phocomelia syndrome, 268300	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS
ESPN	100%	100%	99%	95%	Deafness, neurosensory, without vestibular involvement, autosomal dominant, 609006 Deafness, autosomal recessive 36, 609006 ?Usher syndrome, type 1M, 618632	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
ESR1	100%	99%	100%	98%	Breast cancer, somatic, 114480 Estrogen resistance, 615363	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRE CONCEPTION SCREENING
ESR2	100%	100%	100%	99%	?Ovarian dysgenesis 8, 618187	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRIMARY OVARIAN INSUFFICIENCY TUMOR
ESRP1	100%	100%	100%	98%	?Deafness, autosomal recessive 109, 618013	HEARING IMPAIRMENT MENDELIOME
ESRRB	100%	100%	100%	99%	Deafness, autosomal recessive 35, 608565	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
ETFA	100%	100%	100%	97%	Glutaric acidemia IIA, 231680	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING

ETFB	100%	100%	100%	99%	Glutaric acidemia IIB, 231680	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ETFDH	100%	100%	100%	98%	Glutaric acidemia IIC, 231680	LIVER DISORDERS METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
ETHE1	100%	100%	100%	97%	Ethylmalonic encephalopathy, 602473	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
ETV6	100%	100%	100%	98%	Thrombocytopenia 5, 616216 Leukemia, acute myeloid, somatic, 601626	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME TUMOR
EVC	100%	99%	100%	98%	Ellis-van Creveld syndrome, 225500 ?Weyers acrofacial dysostosis, 193530	CILIOPATHIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
EVC2	100%	100%	100%	98%	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530	CILIOPATHIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
EWSR1	100%	100%	100%	99%	Neuroepithelioma, 612219 Ewing sarcoma, 612219	MENDELIOME
EXOC2	100%	100%	100%	98%	Neurodevelopmental disorder with dysmorphic facies and cerebellar hypoplasia, 619306	INTELLECTUAL DISABILITY MENDELIOME
EXOC6	100%	100%	100%	97%	No OMIM disease ID	IRON DISORDERS MENDELIOME

EXOC6B	100%	100%	100%	99%	Spondyloepimetaphyseal dysplasia with joint laxity, type 3, 618395	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
EXOC7	100%	100%	100%	98%	Neurodevelopmental disorder with seizures and brain atrophy, 619072	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
EXOC8	100%	100%	100%	97%	?Neurodevelopmental disorder with microcephaly, seizures, and brain atrophy, 619076	CILIOPATHIES MENDELIOME
EXOSC1	100%	100%	100%	98%	?Pontocerebellar hypoplasia, type 1F, 619304	MENDELIOME
EXOSC2	100%	100%	100%	97%	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763	VISION DISORDERS HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
EXOSC3	100%	100%	100%	98%	Pontocerebellar hypoplasia, type 1B, 614678	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
EXOSC5	100%	100%	100%	99%	Cerebellar ataxia, brain abnormalities, and cardiac conduction defects, 619576	MOVEMENT DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
EXOSC8	100%	100%	100%	96%	Pontocerebellar hypoplasia, type 1C, 616081	MOVEMENT DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS
EXOSC9	100%	100%	100%	98%	Pontocerebellar hypoplasia, type 1D, 618065	MOVEMENT DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
EXPH5	100%	100%	100%	98%	Epidermolysis bullosa simplex 4, localized or generalized intermediate, autosomal recessive, 615028	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING

EXT1	100%	100%	100%	98%	Exostoses, multiple, type 1, 133700 Chondrosarcoma, 215300	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME TUMOR
EXT2	100%	100%	100%	99%	Seizures, scoliosis, and macrocephaly syndrome, 616682 Exostoses, multiple, type 2, 133701	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING TUMOR
EXTL3	100%	100%	100%	99%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425	CILIOPATHIES PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
EYA1	100%	100%	100%	99%	Branchiootic syndrome 1, 602588 Branchiootorenal syndrome 1, w/wo cataracts, 113650 Anterior segment anomalies w/wo cataract, 602588 ?Otofaciocervical syndrome, 166780	VISION DISORDERS CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT METABOLIC DISORDERS RENAL DISORDERS MENDELIOME SCHISIS
EYA4	100%	100%	100%	99%	?Cardiomyopathy, dilated, 1J, 605362 Deafness, autosomal dominant 10, 601316	HEARING IMPAIRMENT MENDELIOME
EYS	100%	99%	100%	98%	Retinitis pigmentosa 25, 602772	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
EZH2	100%	100%	100%	99%	Weaver syndrome, 277590	HEREDITARY BONE MARROW FAILURE CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME TUMOR
F10	100%	100%	100%	98%	Factor X deficiency, 227600	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
F11	100%	100%	100%	98%	Factor XI deficiency, autosomal dominant, 612416 Factor XI deficiency, autosomal recessive, 612416	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING

F12	100%	100%	100%	99%	Angioedema, hereditary, 3, 610618 Factor XII deficiency, 234000	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
F13A1	100%	100%	100%	99%	Factor XIIIA deficiency, 613225	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
F13B	99%	98%	100%	98%	Factor XIIIB deficiency, 613235	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
F2	100%	100%	100%	99%	Hypoprothrombinemia, 613679 Dysprothrombinemia, 613679 Thrombophilia 1 due to thrombin defect, 188050	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
F2RL3	100%	100%	100%	99%	No OMIM disease ID	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME
F5	100%	100%	100%	98%	Thrombophilia 2 due to activated protein C resistance, 188055 Factor V deficiency, 227400	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
F7	100%	100%	100%	99%	Factor VII deficiency, 227500	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
F8	100%	99%	97%	71%	Thrombophilia 13, X-linked, due to factor VIII defect, 301071 Hemophilia A, 306700	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME
F9	100%	100%	97%	70%	Hemophilia B, 306900 Thrombophilia 8, X-linked, due to factor IX defect, 300807	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME
FA2H	100%	100%	100%	98%	Spastic paraplegia 35, autosomal recessive, 612319	MOVEMENT DISORDERS VISION DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
FAAH	100%	100%	100%	99%	No OMIM disease ID	HNPD MENDELIOME
FAAP24	100%	100%	100%	98%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
FADD	100%	100%	100%	100%	Immunodeficiency 90 with encephalopathy, functional hyposplenia, and hepatic dysfunction, 613759	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING

FAH	100%	100%	100%	98%	Tyrosinemia, type I, 276700	HEART LIVER DISORDERS METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
FAM111A	100%	100%	100%	98%	Kenny-Caffey syndrome, type 2, 127000 Gracile bone dysplasia, 602361	SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME
FAM111B	100%	100%	100%	98%	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704	SKIN DISORDERS MENDELIOME MUSCLE DISORDERS
FAM126A	100%	100%	100%	98%	Leukodystrophy, hypomyelinating, 5, 610532	MOVEMENT DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
FAM149B1	100%	100%	100%	99%	Joubert syndrome 36, 618763	CILIOPATHIES INTELLECTUAL DISABILITY MENDELIOME
FAM161A	100%	100%	100%	96%	Retinitis pigmentosa 28, 606068	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
FAM20A	100%	100%	100%	97%	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690	CRANIOFACIAL ANOMALIES SKIN DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
FAM20B	100%	100%	100%	98%	No OMIM disease ID	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
FAM20C	100%	100%	100%	97%	Raine syndrome, 259775	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS
FAM50A	100%	100%	97%	71%	Intellectual developmental disorder, X-linked syndromic, Armfield type, 300261	INTELLECTUAL DISABILITY MENDELIOME

FAM83G	100%	100%	100%	99%	No OMIM disease ID	SKIN DISORDERS MENDELIOME
FAM83H	100%	100%	100%	99%	Amelogenesis imperfecta, type IIIA, 130900	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME
FAM92A	100%	100%	100%	94%	?Polydactyly, postaxial, type A9, 618219	MENDELIOME
FAN1	100%	100%	100%	98%	Interstitial nephritis, karyomegalic, 614817	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
FANCA	100%	100%	100%	98%	Fanconi anemia, complementation group A, 227650	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING TUMOR
FANCB	100%	100%	96%	68%	Fanconi anemia, complementation group B, 300514	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING TUMOR
FANCC	100%	100%	100%	98%	Fanconi anemia, complementation group C, 227645	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING TUMOR
FANCD2	100%	100%	100%	98%	Fanconi anemia, complementation group D2, 227646	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING TUMOR
FANCE	100%	100%	100%	98%	Fanconi anemia, complementation group E, 600901	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING TUMOR
FANCF	100%	100%	100%	98%	Fanconi anemia, complementation group F, 603467	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING TUMOR

FANCG	100%	100%	100%	98%	Fanconi anemia, complementation group G, 614082	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING TUMOR
FANCI	100%	100%	100%	98%	Fanconi anemia, complementation group I, 609053	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING TUMOR
FANCL	100%	100%	100%	98%	Fanconi anemia, complementation group L, 614083	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING TUMOR
FANCM	100%	100%	100%	97%	?Premature ovarian failure 15, 618096 Spermatogenic failure 28, 618086	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT MALE INFERTILITY MENDELIOME PRIMARY OVARIAN INSUFFICIENCY TUMOR
FAR1	100%	100%	100%	98%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154 Cataracts, spastic paraparesis, and speech delay, 619338	MOVEMENT DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
FARS2	100%	100%	100%	99%	Combined oxidative phosphorylation deficiency 14, 614946 Spastic paraplegia 77, autosomal recessive, 617046	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
FARSA	100%	100%	100%	99%	?Rajab interstitial lung disease with brain calcifications 2, 619013	MENDELIOME
FARSB	100%	100%	100%	98%	Rajab interstitial lung disease with brain calcifications 1, 613658	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING

FAS	100%	100%	100%	97%	Autoimmune lymphoproliferative syndrome, type IA, 601859 Squamous cell carcinoma, burn scar-related, somatic,	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCY MENDELIOME TUMOR
FASLG	100%	100%	100%	99%	Autoimmune lymphoproliferative syndrome, type IB, 601859	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCY MENDELIOME TUMOR
FASTKD2	100%	100%	100%	97%	Combined oxidative phosphorylation deficiency 44, 618855	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
FAT1	100%	100%	100%	99%	No OMIM disease ID	RENAL DISORDERS MENDELIOME
FAT2	100%	100%	100%	99%	Spinocerebellar ataxia 45, 617769	MENDELIOME
FAT4	99%	99%	100%	98%	Van Maldergem syndrome 2, 615546 Hennekam lymphangiectasia-lymphedema syndrome 2, 616006	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
FBLN1	100%	100%	100%	99%	No OMIM disease ID	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
FBLN5	91%	91%	100%	98%	Cutis laxa, autosomal recessive, type IA, 219100 Charcot-Marie-Tooth disease, demyelinating, type 1H, 619764 Macular degeneration, age-related, 3, 608895 Neuropathy, hereditary, w/wo age-related macular degeneration, 608895 ?Cutis laxa, autosomal dominant 2, 614434	SKIN DISORDERS NEUROPATHIES HNPD MENDELIOME PRE CONCEPTION SCREENING
FBN1	100%	100%	100%	99%	Geleophysic dysplasia 2, 614185 Weill-Marchesani syndrome 2, dominant, 608328 Ectopia lentis, familial, 129600 MASS syndrome, 604308 Marfan lipodystrophy syndrome, 616914 Acromicric dysplasia, 102370 Marfan syndrome, 154700 Stiff skin syndrome, 184900	ANEURYSM VISION DISORDERS CONGENITAL HEART DISEASE HEART HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME

FBN2	100%	100%	100%	99%	Macular degeneration, early-onset, 616118 Contractural arachnodactyly, congenital, 121050	FETAL AKINESIA ANEURYSM CONGENITAL HEART DISEASE HEART NEUROPATHIES HNPD SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
FBP1	100%	100%	100%	99%	Fructose-1,6-bisphosphatase deficiency, 229700	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
FBP2	100%	100%	100%	99%	?Leukodystrophy, childhood-onset, remitting, 619864	METABOLIC DISORDERS MENDELIOME
FBRSL1	99%	99%	99%	92%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
FBXL3	100%	100%	100%	99%	Intellectual developmental disorder with short stature, facial anomalies, and speech defects, 606220	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
FBXL4	100%	100%	100%	99%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
FBXO11	100%	100%	99%	95%	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities, 618089	INTELLECTUAL DISABILITY MENDELIOME
FBXO28	100%	100%	100%	98%	Developmental and epileptic encephalopathy 100, 619777	INTELLECTUAL DISABILITY MENDELIOME
FBXO31	100%	100%	99%	96%	?Intellectual developmental disorder, autosomal recessive 45, 615979	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
FBXO32	100%	100%	100%	99%	No OMIM disease ID	HEART MENDELIOME
FBXO38	100%	100%	100%	98%	Neuronopathy, distal hereditary motor, type IID, 615575	NEUROPATHIES MENDELIOME
FBXO43	100%	100%	100%	98%	Oocyte maturation defect 12, 619697 Spermatogenic failure 64, 619696	MENDELIOME
FBXO7	100%	100%	100%	98%	Parkinson disease 15, autosomal recessive, 260300	MOVEMENT DISORDERS MENDELIOME PARKINSON PRE CONCEPTION SCREENING

FBXW11	100%	100%	100%	98%	Neurodevelopmental, jaw, eye, and digital syndrome, 618914	VISION DISORDERS PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME
FBXW4	100%	100%	100%	95%	No OMIM disease ID	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
FBXW7	99%	98%	100%	99%	Developmental delay, hypotonia, and impaired language, 620012	INTELLECTUAL DISABILITY MENDELIOME TUMOR
FCGR3A	100%	100%	100%	98%	Immunodeficiency 20, 615707	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
FCGR3B	99%	97%	92%	74%	No OMIM disease ID	MENDELIOME
FCHO1	100%	100%	100%	98%	Immunodeficiency 76, 619164	PRIMARY IMMUNODEFICIENCY MENDELIOME SEVERE COMBINED IMMUNODEFICIENCY (SCID)
FCN3	100%	100%	100%	98%	Immunodeficiency due to ficolin 3 deficiency, 613860	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
FCSK	100%	100%	100%	99%	Congenital disorder of glycosylation with defective fucosylation 2, 618324	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
FDFT1	100%	100%	100%	97%	Squalene synthase deficiency, 618156	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
FDPS	100%	100%	100%	98%	Porokeratosis 9, multiple types, 616631	SKIN DISORDERS MENDELIOME
FDX2	100%	100%	100%	98%	Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
FDXR	100%	100%	100%	99%	Auditory neuropathy and optic atrophy, 617717	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING

FECH	100%	100%	100%	99%	Protoporphyria, erythropoietic, 1, 177000	SKIN DISORDERS IRON DISORDERS LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
FERMT1	100%	100%	100%	98%	Kindler syndrome, 173650	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
FERMT3	100%	100%	100%	98%	Leukocyte adhesion deficiency, type III, 612840	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
FEZF1	100%	100%	100%	97%	Hypogonadotropic hypogonadism 22, w/wo anosmia, 616030	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME PRE CONCEPTION SCREENING
FGA	100%	100%	100%	98%	Hypodysfibrinogenemia, congenital, 616004 Dysfibrinogenemia, congenital, 616004 Amyloidosis, familial visceral, 105200 Afibrinogenemia, congenital, 202400	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
FGB	100%	100%	100%	99%	Hypofibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Afibrinogenemia, congenital, 202400	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
FGD1	99%	99%	97%	69%	Intellectual developmental disorder, X-linked syndromic 16, 305400 Aarskog-Scott syndrome, 305400	CRANIOFACIAL ANOMALIES EPILEPSY SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS
FGD4	100%	100%	100%	98%	Charcot-Marie-Tooth disease, type 4H, 609311	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING
FGF10	99%	99%	100%	97%	LADD syndrome 3, 620193 Aplasia of lacrimal and salivary glands, 180920	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME

FGF12	100%	100%	100%	99%	Developmental and epileptic encephalopathy 47, 617166	EPILEPSY HEART INTELLECTUAL DISABILITY MENDELIOME
FGF13	100%	99%	98%	69%	Developmental and epileptic encephalopathy 90, 301058 Intellectual developmental disorder, X-linked 110, 301095	INTELLECTUAL DISABILITY MENDELIOME
FGF14	100%	100%	100%	98%	Spinocerebellar ataxia 27A, 193003 Spinocerebellar ataxia 27B, late-onset, 620174	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME
FGF16	100%	99%	97%	68%	Metacarpal 4-5 fusion, 309630	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
FGF17	100%	100%	100%	99%	Hypogonadotropic hypogonadism 20 w/wo anosmia, 615270	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME
FGF20	100%	100%	100%	98%	?Renal hypodysplasia/aplasia 2, 615721	MENDELIOME PRE CONCEPTION SCREENING
FGF23	100%	100%	100%	99%	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993 Hypophosphatemic rickets, autosomal dominant, 193100	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
FGF3	100%	100%	100%	95%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706	CRANIOFACIAL ANOMALIES SKIN DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
FGF5	100%	100%	100%	98%	Trichomegaly, 190330	SKIN DISORDERS MENDELIOME
FGF8	100%	100%	99%	96%	Hypogonadotropic hypogonadism 6 w/wo anosmia, 612702	CRANIOFACIAL ANOMALIES SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS
FGF9	100%	100%	100%	99%	Multiple synostoses syndrome 3, 612961	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME

FGFR1	100%	100%	100%	99%	Pfeiffer syndrome, 101600 Hypogonadotropic hypogonadism 2 w/wo anosmia, 147950 Jackson-Weiss syndrome, 123150 Hartsfield syndrome, 615465 Trigonocephaly 1, 190440 Osteoglophonic dysplasia, 166250 Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001	CRANIOFACIAL ANOMALIES SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS
FGFR2	100%	100%	100%	99%	Bent bone dysplasia syndrome, 614592 LADD syndrome 1, 149730 Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Jackson-Weiss syndrome, 123150 Gastric cancer, somatic, 613659 Craniofacial-skeletal-dermatologic dysplasia, 101600 Apert syndrome, 101200 Pfeiffer syndrome, 101600 ?Scaphocephaly, maxillary retrusion, and impaired intellectual development, 609579 Beare-Stevenson cutis gyrata syndrome, 123790 Crouzon syndrome, 123500 Saethre-Chotzen syndrome, 101400 Scaphocephaly and Axenfeld-Rieger anomaly, Craniosynostosis, nonspecific,	CRANIOFACIAL ANOMALIES SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS
FGFR3	100%	100%	100%	99%	Muenke syndrome, 602849 SADDAN, 616482 Hypochondroplasia, 146000 Thanatophoric dysplasia, type II, 187601 Nevus, epidermal, somatic, 162900 CATSHL syndrome, 610474 Thanatophoric dysplasia, type I, 187600 Spermatocytic seminoma, somatic, 273300 Bladder cancer, somatic, 109800 LADD syndrome 2, 620192 Achondroplasia, 100800 Cervical cancer, somatic, 603956 Colorectal cancer, somatic, 114500 Crouzon syndrome with acanthosis nigricans, 612247	CRANIOFACIAL ANOMALIES SKIN DISORDERS HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
FGG	100%	100%	100%	98%	Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, 616004 Hypofibrinogenemia, congenital, 202400 Afibrinogenemia, congenital, 202400	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING

FH	100%	100%	100%	98%	Leiomyomatosis and renal cell cancer, 150800 Fumarase deficiency, 606812	SKIN DISORDERS LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING TUMOR
FHL1	100%	99%	97%	69%	Myopathy, X-linked, with postural muscle atrophy, 300696 Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 ?Uruguay faciocardiomusculoskeletal syndrome, 300280 Scapuloperoneal myopathy, X-linked dominant, 300695 Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718 Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717	HEART HYPERTROPHIC CARDIOMYOPATHY MENDELIOME MUSCLE DISORDERS
FHL2	100%	100%	100%	99%	No OMIM disease ID	HEART MENDELIOME
FHOD3	100%	100%	100%	98%	Cardiomyopathy, familial hypertrophic, 28, 619402	HEART MENDELIOME
FIBP	100%	100%	100%	98%	Thauvin-Robinet-Faivre syndrome, 617107	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
FICD	100%	100%	100%	99%	No OMIM disease ID	MOVEMENT DISORDERS MENDELIOME
FIG4	100%	100%	100%	99%	Yunis-Varon syndrome, 216340 ?Polymicrogyria, bilateral temporooccipital, 612691 Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228	ALS NEUROPATHIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
FIGLA	100%	100%	100%	99%	Premature ovarian failure 6, 612310	MENDELIOME
FIGN	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
FITM2	100%	100%	100%	99%	Siddiqi syndrome, 618635	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING

FKBP10	100%	100%	100%	98%	Osteogenesis imperfecta, type XI, 610968 Bruck syndrome 1, 259450	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
FKBP14	100%	100%	100%	97%	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
FKBP6	100%	100%	100%	97%	Spermatogenic failure 77, 620103	MALE INFERTILITY MENDELIOME
FKRP	100%	100%	100%	99%	Muscular dystrophy-dystroglycanopathy (congenital w/wo impaired intellectual development), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153	FETAL AKINESIA HEART METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
FKTN	100%	100%	99%	98%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (congenital without impaired intellectual development), type B, 4, 613152	HEART METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
FLAD1	100%	100%	100%	99%	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
FLCN	100%	100%	100%	99%	Birt-Hogg-Dube syndrome, 135150 Colorectal cancer, somatic, 114500 Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700	SKIN DISORDERS MENDELIOME TUMOR
FLG	100%	100%	100%	97%	Ichthyosis vulgaris, 146700	SKIN DISORDERS MENDELIOME
FLG2	100%	100%	100%	99%	Peeling skin syndrome 6, 618084	SKIN DISORDERS MENDELIOME
FLI1	100%	100%	100%	99%	Bleeding disorder, platelet-type, 21, 617443	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME

FLNA	100%	99%	99%	79%	Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048 Cardiac valvular dysplasia, X-linked, 314400 ?FG syndrome 2, 300321 Melnick-Needles syndrome, 309350 Terminal osseous dysplasia, 300244 Congenital short bowel syndrome, 300048 Otopalatodigital syndrome, type I, 311300 Heterotopia, periventricular, 1, 300049 Frontometaphyseal dysplasia 1, 305620	ANEURYSM CRANIOFACIAL ANOMALIES EPILEPSY HEART HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS INTELLECTUAL DISABILITY MENDELIOME SCHISIS
FLNB	100%	100%	100%	99%	Larsen syndrome, 150250 Atelosteogenesis, type I, 108720 Atelosteogenesis, type III, 108721 Spondylocarpotarsal synostosis syndrome, 272460 Boomerang dysplasia, 112310	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS
FLNC	100%	100%	100%	99%	Cardiomyopathy, familial hypertrophic, 26, 617047 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Myopathy, myofibrillar, 5, 609524	ARRHYTHMOGENIC CARDIOMYOPATHY (ACM/ARVC) FETAL AKINESIA DILATED CARDIOMYOPATHY HEART HYPERTROPHIC CARDIOMYOPATHY MENDELIOME ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS MUSCLE DISORDERS
FLRT3	100%	99%	100%	99%	Hypogonadotropic hypogonadism 21 with anosmia, 615271	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME
FLT3	100%	100%	100%	97%	Leukemia, acute lymphoblastic, somatic, 613065 Leukemia, acute myeloid, reduced survival in, somatic, 601626 Leukemia, acute myeloid, somatic, 601626	MENDELIOME
FLT4	100%	100%	100%	99%	Hemangioma, capillary infantile, somatic, 602089 Lymphatic malformation 1, 153100 Congenital heart defects, multiple types, 7, 618780	CONGENITAL HEART DISEASE SKIN DISORDERS HEART MENDELIOME

FLVCR1	100%	100%	100%	99%	Ataxia, posterior column, with retinitis pigmentosa, 609033	MOVEMENT DISORDERS VISION DISORDERS NEUROPATHIES HNPD INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
FLVCR2	100%	100%	100%	99%	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790	FETAL AKINESIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
FMN1	100%	100%	99%	96%	No OMIM disease ID	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
FMN2	100%	99%	99%	90%	Intellectual developmental disorder, autosomal recessive 47, 616193	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
FMO3	100%	100%	100%	98%	Trimethylaminuria, 602079	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
FMR1	100%	100%	97%	70%	Fragile X tremor/ataxia syndrome, 300623 Fragile X syndrome, 300624 Premature ovarian failure 1, 311360	INTELLECTUAL DISABILITY MENDELIOME
FN1	100%	100%	100%	99%	Spondylometaphyseal dysplasia, corner fracture type, 184255 Glomerulopathy with fibronectin deposits 2, 601894	SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME
FNIP1	100%	100%	100%	98%	Immunodeficiency 93 and hypertrophic cardiomyopathy, 619705	SKIN DISORDERS HEART PRIMARY IMMUNODEFICIENCY MENDELIOME
FOLR1	100%	100%	100%	99%	Neurodegeneration due to cerebral folate transport deficiency, 613068	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
FOXC1	100%	100%	99%	80%	Axenfeld-Rieger syndrome, type 3, 602482 Anterior segment dysgenesis 3, multiple subtypes, 601631	VISION DISORDERS CRANIOFACIAL ANOMALIES MENDELIOME

FOXC2	100%	100%	99%	93%	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400	SKIN DISORDERS HEART RENAL DISORDERS MENDELIOME SCHISIS
FOXD4	100%	100%	100%	99%	No OMIM disease ID	HEART MENDELIOME
FOXE1	100%	100%	99%	90%	Bamforth-Lazarus syndrome, 241850	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS
FOXE3	100%	99%	99%	89%	Anterior segment dysgenesis 2, multiple subtypes, 610256 Cataract 34, multiple types, 612968	ANEURYSM VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
FOXF1	100%	100%	100%	92%	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380	CILIOPATHIES MENDELIOME
FOXF2	99%	99%	99%	85%	No OMIM disease ID	HEARING IMPAIRMENT MENDELIOME
FOXG1	100%	99%	100%	94%	Rett syndrome, congenital variant, 613454	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
FOXH1	100%	100%	100%	99%	No OMIM disease ID	CONGENITAL HEART DISEASE HEART MENDELIOME
FOXI1	100%	100%	100%	99%	Enlarged vestibular aqueduct, 600791	HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
FOXI3	99%	99%	99%	88%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME SEVERE COMBINED IMMUNODEFICIENCY (SCID)
FOXJ1	100%	100%	100%	97%	Ciliary dyskinesia, primary, 43, 618699	CILIOPATHIES INTELLECTUAL DISABILITY MENDELIOME
FOXL1	100%	100%	99%	95%	No OMIM disease ID	CONGENITAL HEART DISEASE HEART MENDELIOME

FOXL2	100%	100%	99%	88%	Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 Premature ovarian failure 3, 608996	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRIMARY OVARIAN INSUFFICIENCY
FOXN1	100%	100%	100%	99%	T-cell lymphopenia, infantile, w/wo nail dystrophy, autosomal dominant, 618806 T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)
FOXO1	100%	100%	99%	88%	Rhabdomyosarcoma, alveolar, 268220	MENDELIOME
FOXP1	100%	100%	100%	98%	Intellectual developmental disorder with language impairment w/wo autistic features, 613670	INTELLECTUAL DISABILITY MENDELIOME
FOXP2	100%	99%	100%	98%	Speech-language disorder-1, 602081	INTELLECTUAL DISABILITY MENDELIOME
FOXP3	100%	99%	98%	75%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME
FOXRED1	100%	100%	100%	98%	Mitochondrial complex I deficiency, nuclear type 19, 618241	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
FPR1	100%	100%	100%	98%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
FRA10AC1	100%	100%	100%	96%	Neurodevelopmental disorder with growth retardation, dysmorphic facies, and corpus callosum abnormalities, 620113	INTELLECTUAL DISABILITY MENDELIOME
FRAS1	100%	99%	100%	99%	Fraser syndrome 1, 219000	DISORDERS OF SEX DEVELOPMENT INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS
FREM1	100%	100%	100%	98%	Manitoba oculotrichoanal syndrome, 248450 Bifid nose w/wo anorectal and renal anomalies, 608980 Trigonocephaly 2, 614485	VISION DISORDERS SKIN DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
FREM2	99%	99%	100%	98%	Fraser syndrome 2, 617666 Cryptophthalmos, unilateral or bilateral, isolated, 123570	DISORDERS OF SEX DEVELOPMENT RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING

FRMD4A	96%	96%	100%	98%	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819	INTELLECTUAL DISABILITY MENDELIOME
FRMD5	100%	100%	100%	98%	Neurodevelopmental disorder with eye movement abnormalities and ataxia, 620094	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME
FRMD7	99%	99%	98%	72%	Nystagmus, infantile periodic alternating, X-linked, 310700 Nystagmus 1, congenital, X-linked, 310700	MOVEMENT DISORDERS VISION DISORDERS MENDELIOME
FRMPD4	100%	99%	97%	68%	Intellectual developmental disorder, X-linked 104, 300983	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
FRRS1L	100%	100%	99%	88%	Developmental and epileptic encephalopathy 37, 616981	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
FSCN2	100%	100%	100%	98%	Retinitis pigmentosa 30, 607921	MENDELIOME
FSHB	98%	98%	100%	99%	Hypogonadotropic hypogonadism 24 without anosmia, 229070	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY
FSHR	100%	99%	100%	99%	Ovarian response to FSH stimulation, 276400 Ovarian hyperstimulation syndrome, 608115 Ovarian dysgenesis 1, 233300	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY
FSIP2	100%	100%	100%	97%	Spermatogenic failure 34, 618153	MALE INFERTILITY MENDELIOME
FTCD	100%	100%	99%	97%	Glutamate formiminotransferase deficiency, 229100	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
FTH1	100%	100%	100%	98%	?Hemochromatosis, type 5, 615517	IRON DISORDERS MENDELIOME
FTL	100%	100%	100%	96%	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159	MOVEMENT DISORDERS VISION DISORDERS IRON DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PARKINSON

FTO	94%	94%	100%	98%	Growth retardation, developmental delay, facial dysmorphism, 612938	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS
FTSJ1	100%	100%	98%	73%	Intellectual developmental disorder, X-linked 9, 309549	INTELLECTUAL DISABILITY MENDELIOME
FUCA1	100%	100%	100%	98%	Fucosidosis, 230000	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
FURIN	100%	100%	100%	99%	No OMIM disease ID	ANEURYSM MENDELIOME
FUS	100%	100%	100%	99%	Amyotrophic lateral sclerosis 6, w/wo frontotemporal dementia, 608030 Essential tremor, hereditary, 4, 614782	ALS MENDELIOME
FUT2	100%	100%	100%	99%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME
FUT6	100%	100%	100%	99%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME
FUT8	100%	99%	100%	99%	Congenital disorder of glycosylation with defective fucosylation 1, 618005	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
FUZ	100%	100%	100%	98%	No OMIM disease ID	CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
FXN	100%	100%	100%	96%	Friedreich ataxia with retained reflexes, 229300 Friedreich ataxia, 229300	NEUROPATHIES IRON DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
FXR1	100%	100%	100%	97%	Congenital myopathy 9B, proximal, with minicore lesions, 618823 ?Congenital myopathy 9A with respiratory insufficiency and bone fractures, 618822	MENDELIOME
FXYD2	100%	100%	100%	99%	Hypomagnesemia 2, renal, 154020	EPILEPSY RENAL DISORDERS MENDELIOME
FYB1	100%	100%	100%	97%	Thrombocytopenia 3, 273900	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME

FYCO1	100%	100%	100%	99%	Cataract 18, autosomal recessive, 610019	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
FZD2	100%	100%	100%	96%	Omodysplasia 2, 164745	DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
FZD4	100%	100%	100%	97%	Retinopathy of prematurity, 133780 Exudative vitreoretinopathy 1, 133780	VISION DISORDERS MENDELIOME
FZD6	100%	100%	100%	98%	Nail disorder, nonsyndromic congenital, 1, 161050	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
FZR1	100%	100%	100%	99%	Developmental and epileptic encephalopathy 109, 620145	INTELLECTUAL DISABILITY MENDELIOME
G6PC	100%	100%	100%	99%	Glycogen storage disease Ia, 232200	PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
G6PC3	100%	100%	100%	99%	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING TUMOR
G6PD	100%	99%	99%	76%	Hemolytic anemia, G6PD deficient (favism), 300908	PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS MENDELIOME
GAA	100%	100%	100%	99%	Glycogen storage disease II, 232300	HEART METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
GAB1	100%	100%	100%	98%	?Deafness, autosomal recessive 26, 605428	HEARING IMPAIRMENT MENDELIOME
GABBR1	100%	100%	100%	98%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
GABBR2	99%	99%	99%	97%	Developmental and epileptic encephalopathy 59, 617904 Neurodevelopmental disorder with poor language and loss of hand skills, 617903	INTELLECTUAL DISABILITY MENDELIOME

GABRA1	100%	100%	100%	99%	Developmental and epileptic encephalopathy 19, 615744	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
GABRA2	100%	100%	100%	97%	Developmental and epileptic encephalopathy 78, 618557	INTELLECTUAL DISABILITY MENDELIOME
GABRA3	100%	99%	98%	73%	Epilepsy, X-linked 2, w/wo impaired intellectual development and dysmorphic features, 301091	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
GABRA5	100%	100%	100%	98%	Developmental and epileptic encephalopathy 79, 618559	INTELLECTUAL DISABILITY MENDELIOME
GABRB1	100%	100%	100%	98%	Developmental and epileptic encephalopathy 45, 617153	INTELLECTUAL DISABILITY MENDELIOME
GABRB2	100%	100%	100%	99%	Developmental and epileptic encephalopathy 92, 617829	INTELLECTUAL DISABILITY MENDELIOME
GABRB3	100%	100%	99%	97%	Developmental and epileptic encephalopathy 43, 617113	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
GABRD	100%	100%	99%	96%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
GABRG2	92%	92%	100%	99%	Developmental and epileptic encephalopathy 74, 618396 Febrile seizures, familial, 8, 607681 Generalized epilepsy with febrile seizures plus, type 3, 607681	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
GAD1	100%	100%	100%	98%	Developmental and epileptic encephalopathy 89, 619124	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
GAL	100%	100%	100%	99%	?Epilepsy, familial temporal lobe, 8, 616461	MENDELIOME
GALC	100%	100%	100%	98%	Krabbe disease, 245200	MOVEMENT DISORDERS NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
GALE	100%	100%	100%	99%	Galactose epimerase deficiency, 230350	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

GALK1	100%	100%	100%	99%	Galactokinase deficiency with cataracts, 230200	VISION DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
GALM	100%	100%	100%	98%	Galactosemia IV, 618881	VISION DISORDERS METABOLIC DISORDERS MENDELIOME
GALNS	100%	100%	100%	98%	Mucopolysaccharidosis IVA, 253000	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
GALNT12	100%	100%	99%	97%	No OMIM disease ID	MENDELIOME TUMOR
GALNT2	100%	100%	100%	97%	Congenital disorder of glycosylation, type IIt, 618885	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
GALNT3	100%	100%	100%	97%	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
GALNTL5	100%	100%	100%	99%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
GALT	100%	100%	100%	99%	Galactosemia, 230400	VISION DISORDERS LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY

GAMT	100%	100%	100%	97%	Cerebral creatine deficiency syndrome 2, 612736	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
GAN	100%	100%	100%	98%	Giant axonal neuropathy-1, 256850	MOVEMENT DISORDERS SKIN DISORDERS NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING
GANAB	100%	100%	100%	99%	Polycystic kidney disease 3, 600666	LIVER DISORDERS METABOLIC DISORDERS RENAL DISORDERS MENDELIOME
GAPVD1	100%	100%	100%	98%	No OMIM disease ID	RENAL DISORDERS MENDELIOME
GARS1	100%	100%	100%	98%	Spinal muscular atrophy, infantile, James type, 619042 Neuronopathy, distal hereditary motor, type VA, 600794 Charcot-Marie-Tooth disease, type 2D, 601472	NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS
GAS2	100%	100%	100%	98%	No OMIM disease ID	HEARING IMPAIRMENT MENDELIOME
GAS2L2	100%	100%	100%	99%	?Ciliary dyskinesia, primary, 41, 618449	MENDELIOME
GAS8	100%	100%	100%	99%	Ciliary dyskinesia, primary, 33, 616726	CILIOPATHIES MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING
GATA1	100%	100%	97%	69%	Leukemia, megakaryoblastic, w/wo Down syndrome, somatic, 190685 Thrombocytopenia, X-linked, w/wo dyserythropoietic anemia, 300367 Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835 Thrombocytopenia with beta-thalassemia, X-linked, 314050 Hemolytic anemia due to elevated adenosine deaminase, 301083	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS IRON DISORDERS MENDELIOME
GATA2	100%	100%	100%	99%	Emberger syndrome, 614038 Immunodeficiency 21, 614172	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME TUMOR
GATA3	100%	100%	100%	99%	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255	HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME

GATA4	100%	100%	99%	96%	Tetralogy of Fallot, 187500 Atrial septal defect 2, 607941 Ventricular septal defect 1, 614429 Atrioventricular septal defect 4, 614430 ?Testicular anomalies w/wo congenital heart disease, 615542	CONGENITAL HEART DISEASE DISORDERS OF SEX DEVELOPMENT HEART MENDELIOME
GATA5	100%	100%	100%	97%	Congenital heart defects, multiple types, 5, 617912	CONGENITAL HEART DISEASE HEART MENDELIOME
GATA6	100%	100%	100%	92%	Atrial septal defect 9, 614475 Persistent truncus arteriosus, 217095 Pancreatic agenesis and congenital heart defects, 600001 Atrioventricular septal defect 5, 614474 Tetralogy of Fallot, 187500	CONGENITAL HEART DISEASE HEART MENDELIOME
GATAD1	100%	100%	100%	98%	?Cardiomyopathy, dilated, 2B, 614672	HEART MENDELIOME
GATAD2B	100%	100%	100%	98%	GAND syndrome, 615074	INTELLECTUAL DISABILITY MENDELIOME
GATB	100%	100%	100%	99%	?Combined oxidative phosphorylation deficiency 41, 618838	HEART MENDELIOME MITOCHONDRIAL DISORDERS
GATC	100%	100%	100%	99%	Combined oxidative phosphorylation deficiency 42, 618839	HEART MENDELIOME MITOCHONDRIAL DISORDERS
GATM	100%	100%	100%	97%	Cerebral creatine deficiency syndrome 3, 612718 Fanconi renotubular syndrome 1, 134600	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS
GBA	100%	100%	100%	99%	Gaucher disease, type II, 230900 Gaucher disease, type IIIC, 231005 Gaucher disease, type III, 231000 Gaucher disease, type I, 230800 Gaucher disease, perinatal lethal, 608013	FETAL AKINESIA MOVEMENT DISORDERS HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PARKINSON PRE CONCEPTION SCREENING

GBA2	100%	100%	100%	99%	Spastic paraplegia 46, autosomal recessive, 614409	MOVEMENT DISORDERS NEUROPATHIES METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
GBE1	100%	99%	100%	98%	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570	FETAL AKINESIA MOVEMENT DISORDERS HEART NEUROPATHIES LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
GBF1	100%	100%	100%	99%	Charcot-Marie-Tooth disease, axonal, type 2GG, 606483	NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS
GCDH	100%	100%	100%	99%	Glutaricaciduria, type I, 231670	MOVEMENT DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
GCGR	100%	100%	100%	99%	Mahvash disease, 619290	MENDELIOME
GCH1	100%	100%	99%	98%	Dystonia, DOPA-responsive, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PARKINSON PRE CONCEPTION SCREENING
GCK	100%	100%	100%	99%	MODY, type II, 125851 Diabetes mellitus, permanent neonatal 1, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485 Diabetes mellitus, noninsulin-dependent, late onset, 125853	EPILEPSY METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
GCLC	100%	100%	100%	98%	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
GCLM	100%	100%	100%	96%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME

GCM2	100%	100%	100%	99%	Hypoparathyroidism, familial isolated 2, 618883 Hyperparathyroidism 4, 617343	SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME
GCNA	100%	100%	98%	72%	Spermatogenic failure, X-linked, 4, 301077	MALE INFERTILITY MENDELIOME
GCNT2	100%	100%	100%	99%	Adult i phenotype without cataract, 110800 Cataract 13 with adult i phenotype, 116700	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
GCSH	100%	100%	100%	98%	?Glycine encephalopathy, 605899	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
GDAP1	100%	100%	100%	99%	Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706 Charcot-Marie-Tooth disease, recessive intermediate, A, 608340 Charcot-Marie-Tooth disease, axonal, type 2K, 607831 Charcot-Marie-Tooth disease, type 4A, 214400	NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
GDAP2	100%	99%	100%	98%	Spinocerebellar ataxia, autosomal recessive 27, 618369	MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING
GDF1	100%	100%	100%	99%	Congenital heart defects, multiple types, 6, 613854 Right atrial isomerism (Ivemark), 208530	CONGENITAL HEART DISEASE CILIOPATHIES HEART MENDELIOME PRE CONCEPTION SCREENING
GDF11	100%	100%	98%	84%	?Vertebral hypersegmentation and orofacial anomalies, 619122	MENDELIOME
GDF2	100%	100%	100%	99%	Telangiectasia, hereditary hemorrhagic, type 5, 615506	SKIN DISORDERS HEART HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME
GDF3	100%	100%	100%	98%	Klippel-Feil syndrome 3, autosomal dominant, 613702 Microphthalmia, isolated, with coloboma 6, 613703 Microphthalmia, isolated 7, 613704	VISION DISORDERS CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME

GDF5	100%	100%	100%	99%	Acromesomelic dysplasia 2A, 200700 Acromesomelic dysplasia 2B, 228900 Multiple synostoses syndrome 2, 610017 Symphalangism, proximal, 1B, 615298 Brachydactyly, type A2, 112600 ?Acromesomelic dysplasia 2C, Hunter-Thompson type, 201250 Brachydactyly, type C, 113100 Brachydactyly, type A1, C, 615072	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
GDF6	100%	100%	100%	95%	Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094 Leber congenital amaurosis 17, 615360 Multiple synostoses syndrome 4, 617898 Klippel-Feil syndrome 1, autosomal dominant, 118100	VISION DISORDERS CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS
GDF9	100%	100%	100%	98%	?Premature ovarian failure 14, 618014	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRIMARY OVARIAN INSUFFICIENCY
GDI1	100%	100%	98%	76%	Intellectual developmental disorder, X-linked 41, 300849	INTELLECTUAL DISABILITY MENDELIOME
GDNF	100%	100%	100%	98%	No OMIM disease ID	LIVER DISORDERS MENDELIOME TUMOR
GDPD1	100%	100%	100%	96%	No OMIM disease ID	VISION DISORDERS MENDELIOME
GEMIN4	100%	100%	100%	98%	Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities, 617913	MENDELIOME PRE CONCEPTION SCREENING
GEMIN5	100%	100%	100%	98%	Neurodevelopmental disorder with cerebellar atrophy and motor dysfunction, 619333	INTELLECTUAL DISABILITY MENDELIOME
GFAP	100%	100%	100%	98%	Alexander disease, 203450	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME
GFER	100%	100%	99%	91%	Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
GFI1	100%	100%	100%	98%	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 Neutropenia, severe congenital 2, autosomal dominant, 613107	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCY MENDELIOME TUMOR
GFI1B	100%	100%	100%	99%	Bleeding disorder, platelet-type, 17, 187900	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME

GFM1	100%	100%	100%	98%	Combined oxidative phosphorylation deficiency 1, 609060	LIVER DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
GFM2	100%	100%	100%	98%	Combined oxidative phosphorylation deficiency 39, 618397	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
GFPT1	100%	100%	100%	98%	Myasthenia, congenital, 12, with tubular aggregates, 610542	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
GFRA1	100%	100%	100%	98%	Renal hypodysplasia/aplasia 4, 619887	RENAL DISORDERS MENDELIOME
GGCX	100%	100%	100%	98%	Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450 Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
GGPS1	100%	100%	100%	98%	Muscular dystrophy, congenital hearing loss, and ovarian insufficiency syndrome, 619518	METABOLIC DISORDERS MENDELIOME
GGT1	100%	100%	100%	98%	?Glutathioninuria, 231950	MENDELIOME PRE CONCEPTION SCREENING
GH1	100%	100%	100%	99%	Kowarski syndrome, 262650 Growth hormone deficiency, isolated, type II, 173100 Growth hormone deficiency, isolated, type IB, 612781 Growth hormone deficiency, isolated, type IA, 262400	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
GHR	99%	99%	99%	97%	Laron dwarfism, 262500 Increased responsiveness to growth hormone, 604271 Growth hormone insensitivity, partial, 604271	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
GHRHR	100%	100%	100%	98%	Growth hormone deficiency, isolated, type IV, 618157	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
GHSR	100%	100%	100%	98%	Growth hormone deficiency, isolated partial, 615925	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
GIGYF1	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME

GIMAP5	100%	100%	100%	98%	Portal hypertension, noncirrhotic, 2, 619463	PRIMARY IMMUNODEFICIENCY MENDELIOME
GINS1	100%	100%	100%	99%	Immunodeficiency 55, 617827	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
GINS2	100%	100%	100%	98%	No OMIM disease ID	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
GINS4	100%	100%	100%	99%	No OMIM disease ID	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCY MENDELIOME
GIPC1	100%	100%	100%	99%	Oculopharyngodistal myopathy 2, 618940	MENDELIOME
GIPC3	100%	100%	100%	97%	Deafness, autosomal recessive 15, 601869	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
GJA1	100%	100%	100%	98%	Erythrokeratodermia variabilis et progressiva 3, 617525 Craniometaphyseal dysplasia, autosomal recessive, 218400 Oculodentodigital dysplasia, 164200 Hypoplastic left heart syndrome 1, 241550 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100 Oculodentodigital dysplasia, autosomal recessive, 257850 Atrioventricular septal defect 3, 600309	VISION DISORDERS CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS
GJA3	100%	100%	100%	99%	Cataract 14, multiple types, 601885	VISION DISORDERS MENDELIOME
GJA5	100%	100%	100%	99%	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic (GJA5/SCN5A), 108770	CONGENITAL HEART DISEASE HEART MENDELIOME
GJA8	100%	100%	100%	99%	Cataract 1, multiple types, 116200	VISION DISORDERS MENDELIOME
GJB1	100%	100%	98%	76%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800	NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME
GJB2	100%	100%	100%	99%	Keratoderma, palmoplantar, with deafness, 148350 Deafness, autosomal recessive 1A, 220290 Deafness, autosomal dominant 3A, 601544 Hystrix-like ichthyosis with deafness, 602540 Bart-Pumphrey syndrome, 149200 Keratitis-ichthyosis-deafness syndrome, 148210 Vohwinkel syndrome, 124500	SKIN DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING

GJB3	100%	100%	100%	99%	Deafness, digenic, GJB2/GJB3, 220290 Deafness, autosomal dominant 2B, 612644 Erythrokeratodermia variabilis et progressiva 1, 133200 Deafness, autosomal recessive, Deafness, autosomal dominant, with peripheral neuropathy,	SKIN DISORDERS HEARING IMPAIRMENT NEUROPATHIES MENDELIOME
GJB4	100%	100%	100%	99%	Erythrokeratodermia variabilis et progressiva 2, 617524	SKIN DISORDERS MENDELIOME
GJB6	100%	100%	99%	97%	Ectodermal dysplasia 2, Clouston type, 129500 Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290	CRANIOFACIAL ANOMALIES SKIN DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
GJC2	99%	98%	100%	96%	Lymphatic malformation 3, 613480 ?Spastic paraplegia 44, autosomal recessive, 613206 Leukodystrophy, hypomyelinating, 2, 608804	MOVEMENT DISORDERS SKIN DISORDERS PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
GK	100%	100%	97%	71%	Glycerol kinase deficiency, 307030	DISORDERS OF SEX DEVELOPMENT METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME
GLA	90%	90%	98%	74%	Fabry disease, cardiac variant, 301500 Fabry disease, 301500	SKIN DISORDERS HEARING IMPAIRMENT HEART HYPERTROPHIC CARDIOMYOPATHY NEUROPATHIES HNPD METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PAINFUL PERIPHERAL NEUROPATHIES
GLB1	100%	100%	100%	98%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010 GM1-gangliosidosis, type II, 230600	MOVEMENT DISORDERS SKIN DISORDERS HEART SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

GLDC	100%	100%	100%	98%	Glycine encephalopathy, 605899	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
GLDN	100%	100%	100%	98%	Lethal congenital contracture syndrome 11, 617194	FETAL AKINESIA MENDELIOME PRE CONCEPTION SCREENING
GLE1	100%	100%	100%	99%	Lethal congenital contracture syndrome 1, 253310 Congenital arthrogryposis with anterior horn cell disease, 611890	FETAL AKINESIA NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING
GLI1	100%	100%	100%	99%	Polydactyly, preaxial I, 174400 Polydactyly, postaxial, type A8, 618123	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
GLI2	100%	100%	100%	99%	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS
GLI3	100%	100%	100%	99%	Greig cephalopolysyndactyly syndrome, 175700 Polydactyly, postaxial, types A1 and B, 174200 Pallister-Hall syndrome, 146510 Polydactyly, preaxial, type IV, 174700	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME SCHISIS
GLIS1	100%	100%	100%	99%	No OMIM disease ID	HEART MENDELIOME
GLIS2	100%	100%	100%	99%	Nephronophthisis 7, 611498	CILIOPATHIES RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
GLIS3	100%	100%	100%	99%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199	LIVER DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
GLMN	100%	100%	100%	97%	Glomuvenous malformations, 138000	SKIN DISORDERS MENDELIOME

GLRA1	100%	100%	100%	99%	Hyperekplexia 1, 149400	EPILEPSY METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
GLRA2	99%	98%	98%	72%	Intellectual developmental disorder, X-linked syndromic, Pilorge type, 301076	INTELLECTUAL DISABILITY MENDELIOME
GLRB	100%	100%	100%	98%	Hyperekplexia 2, 614619	EPILEPSY MENDELIOME PRE CONCEPTION SCREENING
GLRX5	100%	100%	100%	97%	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859	IRON DISORDERS METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
GLS	100%	100%	100%	98%	Global developmental delay, progressive ataxia, and elevated glutamine, 618412 ?Infantile cataract, skin abnormalities, glutamate excess, and impaired intellectual development, 618339 Developmental and epileptic encephalopathy 71, 618328	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
GLUD1	100%	100%	99%	94%	Hyperinsulinism-hyperammonemia syndrome, 606762	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS
GLUL	100%	100%	100%	98%	Glutamine deficiency, congenital, 610015	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
GLYCTK	100%	100%	100%	99%	D-glyceric aciduria, 220120	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
GLYR1	100%	100%	100%	98%	No OMIM disease ID	CONGENITAL HEART DISEASE HEART MENDELIOME
GM2A	100%	100%	100%	99%	GM2-gangliosidosis, AB variant, 272750	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

GMNN	100%	100%	100%	98%	Meier-Gorlin syndrome 6, 616835	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
GMPPA	100%	100%	100%	99%	Alacrima, achalasia, and impaired intellectual development syndrome, 615510	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
GMPPB	100%	100%	100%	99%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352 Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350	FETAL AKINESIA HEART METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
GMPR	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
GMPS	100%	100%	100%	98%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME
GNA11	100%	100%	100%	97%	Hypocalciuric hypercalcemia, type II, 145981 Hypocalcemia, autosomal dominant 2, 615361	SKIN DISORDERS RENAL DISORDERS MENDELIOME
GNA14	100%	100%	100%	97%	No OMIM disease ID	SKIN DISORDERS MENDELIOME
GNAI1	100%	100%	99%	97%	Neurodevelopmental disorder with hypotonia, impaired speech, and behavioral abnormalities, 619854	INTELLECTUAL DISABILITY MENDELIOME
GNAI2	100%	100%	100%	97%	Ventricular tachycardia, idiopathic, 192605 Pituitary adenoma, ACTH-secreting, somatic,	MENDELIOME
GNAI3	100%	100%	100%	98%	Auriculocondylar syndrome 1, 602483	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS
GNAL	100%	100%	100%	96%	Dystonia 25, 615073	MOVEMENT DISORDERS MENDELIOME
GNAO1	100%	100%	100%	98%	Developmental and epileptic encephalopathy 17, 615473 Neurodevelopmental disorder with involuntary movements, 617493	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
GNAQ	100%	99%	100%	96%	Capillary malformations, congenital, 1, somatic, mosaic, 163000 Sturge-Weber syndrome, somatic, mosaic, 185300	SKIN DISORDERS MENDELIOME

GNAS	100%	99%	99%	94%	ACTH-independent macronodular adrenal hyperplasia, 219080 Pituitary adenoma 3, multiple types, somatic, 617686 Pseudohypoparathyroidism Ic, 612462 Pseudohypoparathyroidism Ia, 103580 Osseous heteroplasia, progressive, 166350 Pseudohypoparathyroidism Ib, 603233 McCune-Albright syndrome, somatic, mosaic, 174800 Pseudopseudohypoparathyroidism, 612463	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
GNAS-AS1	NC	NC	NC	NC	Pseudohypoparathyroidism, type IB, 603233	MENDELIOME
GNAT1	100%	100%	100%	99%	Night blindness, congenital stationary, autosomal dominant 3, 610444 Night blindness, congenital stationary, type 1G, 616389	VISION DISORDERS MENDELIOME
GNAT2	100%	100%	100%	98%	Achromatopsia 4, 613856	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
GNB1	100%	100%	100%	99%	Myelodysplastic syndrome, somatic, 614286 Leukemia, acute lymphoblastic, somatic, 613065 Intellectual developmental disorder, autosomal dominant 42, 616973	INTELLECTUAL DISABILITY MENDELIOME SCHISIS
GNB2	100%	100%	100%	99%	Neurodevelopmental disorder with hypotonia and dysmorphic facies, 619503 ?Sick sinus syndrome 4, 619464	HEART INTELLECTUAL DISABILITY MENDELIOME ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS
GNB3	100%	100%	100%	98%	Night blindness, congenital stationary, type 1H, 617024	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
GNB4	100%	100%	99%	98%	Charcot-Marie-Tooth disease, dominant intermediate F, 615185	NEUROPATHIES MENDELIOME
GNB5	100%	100%	100%	96%	Language delay and ADHD/cognitive impairment w/wo cardiac arrhythmia, 617182 Intellectual developmental disorder with cardiac arrhythmia, 617173	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
GNE	100%	100%	100%	99%	Sialuria, 269921 Nonaka myopathy, 605820	HEMOSTATIC/THROMBOTIC DISORDERS NEUROPATHIES METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
GNMT	100%	100%	100%	98%	Glycine N-methyltransferase deficiency, 606664	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING

GNPAT	100%	100%	100%	98%	Rhizomelic chondrodysplasia punctata, type 2, 222765	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
GNPNAT1	100%	100%	100%	98%	?Rhizomelic dysplasia, Ain-Naz type, 616510	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
GNPTAB	100%	100%	100%	98%	Mucolipidosis III alpha/beta, 252600 Mucolipidosis II alpha/beta, 252500	CRANIOFACIAL ANOMALIES HEART SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
GNPTG	100%	100%	100%	97%	Mucolipidosis III gamma, 252605	VISION DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
GNRH1	100%	100%	100%	97%	?Hypogonadotropic hypogonadism 12 w/wo anosmia, 614841	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME
GNRHR	100%	100%	100%	98%	Hypogonadotropic hypogonadism 7 without anosmia, 146110	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME PRE CONCEPTION SCREENING
GNS	100%	100%	100%	99%	Mucopolysaccharidosis type IIID, 252940	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
GOLGA2	100%	100%	100%	98%	Developmental delay with hypotonia, myopathy, and brain abnormalities, 620240	INTELLECTUAL DISABILITY MENDELIOME
GON7	100%	100%	100%	99%	Galloway-Mowat syndrome 9, 619603	MENDELIOME

GORAB	100%	100%	100%	97%	Geroderma osteodysplasticum, 231070	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
GOSR2	100%	100%	100%	99%	Epilepsy, progressive myoclonic 6, 614018 Muscular dystrophy, congenital, w/wo seizures, 620166	MOVEMENT DISORDERS EPILEPSY MENDELIOME PRE CONCEPTION SCREENING
GOT1	100%	100%	100%	99%	Aspartate aminotransferase, serum level of, QTL1, 614419	METABOLIC DISORDERS MENDELIOME
GOT2	100%	100%	100%	99%	Developmental and epileptic encephalopathy 82, 618721	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS
GP1BA	100%	100%	99%	95%	Bernard-Soulier syndrome, type A1 (recessive), 231200 Bernard-Soulier syndrome, type A2 (dominant), 153670 von Willebrand disease, platelet-type, 177820	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
GP1BB	100%	100%	100%	98%	Giant platelet disorder, isolated, 231200 Bernard-Soulier syndrome, type B, 231200	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
GP6	99%	96%	100%	99%	Bleeding disorder, platelet-type, 11, 614201	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
GP9	100%	100%	100%	99%	Bernard-Soulier syndrome, type C, 231200	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
GPAA1	100%	100%	100%	99%	Glycosylphosphatidylinositol biosynthesis defect 15, 617810	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
GPC3	99%	98%	97%	69%	Wilms tumor, somatic, 194070 Simpson-Golabi-Behmel syndrome, type 1, 312870	EPILEPSY SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS TUMOR

GPC4	100%	99%	98%	73%	Keipert syndrome, 301026	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
GPC6	99%	99%	100%	98%	Omodysplasia 1, 258315	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
GPD1	100%	100%	100%	99%	Hypertriglyceridemia, transient infantile, 614480	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
GPD1L	100%	100%	100%	97%	Brugada syndrome 2, 611777	HEART METABOLIC DISORDERS MENDELIOME
GPHN	100%	99%	100%	98%	Molybdenum cofactor deficiency C, 615501	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
GPI	100%	100%	100%	98%	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
GPIHBP1	100%	100%	100%	99%	Hyperlipoproteinemia, type 1D, 615947	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
GPNMB	95%	95%	100%	99%	Amyloidosis, primary localized cutaneous, 3, 617920	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
GPR101	100%	100%	97%	69%	Pituitary adenoma 2, GH-secreting, 300943	MENDELIOME
GPR143	100%	99%	96%	68%	Ocular albinism, type I, Nettleship-Falls type, 300500 Nystagmus 6, congenital, X-linked, 300814	MOVEMENT DISORDERS VISION DISORDERS SKIN DISORDERS MENDELIOME
GPR161	100%	100%	100%	99%	No OMIM disease ID	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SONIC HEDGEHOG MEDULLOBLASTOMA TUMOR
GPR179	100%	100%	100%	98%	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING

GPR68	100%	100%	100%	99%	Amelogenesis imperfecta, hypomaturation type, IIA6, 617217	CRANIOFACIAL ANOMALIES MENDELIOME
GPR88	100%	99%	99%	90%	?Chorea, childhood-onset, with psychomotor retardation, 616939	MENDELIOME
GPRASP2	100%	100%	98%	74%	?Deafness, X-linked 7, 301018	HEARING IMPAIRMENT MENDELIOME
GPSM2	100%	100%	100%	98%	Chudley-McCullough syndrome, 604213	HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
GPT2	100%	100%	100%	98%	Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
GPX1	100%	100%	100%	97%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME
GPX4	100%	100%	100%	98%	Spondylometaphyseal dysplasia, Sedaghatian type, 250220	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
GRAP	100%	100%	99%	96%	Deafness, autosomal recessive 114, 618456	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
GREB1L	100%	100%	100%	98%	Deafness, autosomal dominant 80, 619274 Renal hypodysplasia/aplasia 3, 617805	HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME
GREM1	100%	100%	100%	97%	No OMIM disease ID	MENDELIOME TUMOR
GREM2	100%	100%	100%	99%	Tooth agenesis, selective, 9, 617275	MENDELIOME
GRHL2	100%	100%	100%	98%	Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029 Corneal dystrophy, posterior polymorphous, 4, 618031	VISION DISORDERS HEREDITARY BONE MARROW FAILURE SKIN DISORDERS DYSKERATOSIS CONGENITA HEARING IMPAIRMENT PRIMARY IMMUNODEFICIENCY MENDELIOME TUMOR
GRHL3	100%	100%	100%	99%	van der Woude syndrome 2, 606713	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME SCHISIS

GRHPR	100%	100%	100%	98%	Hyperoxaluria, primary, type II, 260000	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
GRIA2	100%	100%	100%	98%	Neurodevelopmental disorder with language impairment and behavioral abnormalities, 618917	INTELLECTUAL DISABILITY MENDELIOME
GRIA3	99%	99%	97%	69%	Intellectual developmental disorder, X-linked syndromic, Wu type, 300699	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
GRIA4	99%	99%	100%	99%	Neurodevelopmental disorder w/wo seizures and gait abnormalities, 617864	INTELLECTUAL DISABILITY MENDELIOME
GRID2	99%	99%	100%	99%	Spinocerebellar ataxia, autosomal recessive 18, 616204	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
GRIK2	95%	95%	100%	98%	Neurodevelopmental disorder with impaired language and ataxia and w/wo seizures, 619580 Intellectual developmental disorder, autosomal recessive 6, 611092	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
GRIN1	100%	100%	100%	97%	Neurodevelopmental disorder w/wo hyperkinetic movements and seizures, autosomal recessive, 617820 Developmental and epileptic encephalopathy 101, 619814 Neurodevelopmental disorder w/wo hyperkinetic movements and seizures, autosomal dominant, 614254	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
GRIN2A	99%	99%	100%	98%	Epilepsy, focal, with speech disorder and w/wo impaired intellectual development, 245570	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
GRIN2B	99%	99%	100%	99%	Developmental and epileptic encephalopathy 27, 616139 Intellectual developmental disorder, autosomal dominant 6, w/wo seizures, 613970	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
GRIN2D	99%	98%	99%	87%	Developmental and epileptic encephalopathy 46, 617162	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
GRIP1	100%	100%	100%	99%	Fraser syndrome 3, 617667	DISORDERS OF SEX DEVELOPMENT INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
GRK1	100%	100%	100%	99%	Oguchi disease-2, 613411	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING

GRM1	100%	100%	100%	99%	Spinocerebellar ataxia, autosomal recessive 13, 614831 Spinocerebellar ataxia 44, 617691	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
GRM6	100%	100%	100%	98%	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
GRM7	100%	99%	99%	98%	Neurodevelopmental disorder with seizures, hypotonia, and brain abnormalities, 618922	INTELLECTUAL DISABILITY MENDELIOME
GRN	100%	100%	100%	99%	Aphasia, primary progressive, 607485 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706	ALS MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PARKINSON PRE CONCEPTION SCREENING
GRXCR1	99%	99%	100%	98%	Deafness, autosomal recessive 25, 613285	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
GRXCR2	100%	100%	100%	99%	?Deafness, autosomal recessive 101, 615837	HEARING IMPAIRMENT MENDELIOME
GSC	100%	100%	100%	95%	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
GSDME	100%	100%	100%	99%	Deafness, autosomal dominant 5, 600994	HEARING IMPAIRMENT MENDELIOME
GSE1	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
GSN	100%	100%	100%	98%	Amyloidosis, Finnish type, 105120	VISION DISORDERS SKIN DISORDERS NEUROPATHIES RENAL DISORDERS MENDELIOME
GSR	100%	100%	100%	96%	Hemolytic anemia due to glutathione reductase deficiency, 618660	MENDELIOME
GSS	100%	100%	100%	98%	Hemolytic anemia due to glutathione synthetase deficiency, 231900 Glutathione synthetase deficiency, 266130	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
GSX2	100%	100%	100%	98%	Diencephalic-mesencephalic junction dysplasia syndrome 2, 618646	MENDELIOME PRE CONCEPTION SCREENING

GTF2E2	100%	100%	100%	95%	Trichothiodystrophy 6, nonphotosensitive, 616943	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
GTF2H5	70%	70%	100%	99%	Trichothiodystrophy 3, photosensitive, 616395	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
GTPBP2	100%	100%	100%	98%	Jaberi-Elahi syndrome, 617988	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
GTPBP3	100%	100%	100%	98%	Combined oxidative phosphorylation deficiency 23, 616198	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
GUCA1A	100%	100%	100%	100%	Cone-rod dystrophy 14, 602093 Cone dystrophy-3, 602093	VISION DISORDERS MENDELIOME
GUCA1B	100%	100%	100%	99%	Retinitis pigmentosa 48, 613827	VISION DISORDERS MENDELIOME
GUCY1A1	100%	100%	100%	97%	Moyamoya 6 with achalasia, 615750	MENDELIOME PRE CONCEPTION SCREENING
GUCY2C	100%	100%	100%	97%	Diarrhea 6, 614616 Meconium ileus, 614665	MENDELIOME PRE CONCEPTION SCREENING
GUCY2D	100%	100%	100%	99%	Cone-rod dystrophy 6, 601777 ?Choroidal dystrophy, central areolar 1, 215500 Leber congenital amaurosis 1, 204000 Night blindness, congenital stationary, type 1I, 618555	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
GUF1	100%	100%	99%	97%	?Developmental and epileptic encephalopathy 40, 617065	MENDELIOME MITOCHONDRIAL DISORDERS
GULOP	NC	NC	NC	NC	Scurvy,	MENDELIOME
GUSB	100%	100%	100%	99%	Mucopolysaccharidosis VII, 253220	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
GYG1	100%	100%	100%	98%	?Glycogen storage disease XV, 613507 Polyglucosan body myopathy 2, 616199	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS

GYS1	100%	100%	100%	99%	Glycogen storage disease 0, muscle, 611556	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
GYS2	100%	100%	100%	98%	Glycogen storage disease 0, liver, 240600	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
GZF1	100%	100%	100%	99%	Joint laxity, short stature, and myopia, 617662	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
H1-4	100%	100%	100%	98%	Rahman syndrome, 617537	INTELLECTUAL DISABILITY MENDELIOME
H19	NC	NC	NC	NC	No OMIM disease ID	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
H3-3A	100%	100%	100%	99%	Bryant-Li-Bhoj neurodevelopmental syndrome 1, 619720	MENDELIOME
H3-3B	100%	100%	100%	97%	Bryant-Li-Bhoj neurodevelopmental syndrome 2, 619721	INTELLECTUAL DISABILITY MENDELIOME
H4C11	100%	100%	100%	99%	?Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 2, 619759	MENDELIOME
H4C3	100%	100%	100%	98%	Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 1, 619758	INTELLECTUAL DISABILITY MENDELIOME
H4C5	100%	100%	100%	96%	Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 3, 619950	INTELLECTUAL DISABILITY MENDELIOME
H4C9	100%	100%	100%	96%	Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 4, 619951	INTELLECTUAL DISABILITY MENDELIOME
H6PD	100%	100%	100%	99%	Cortisone reductase deficiency 1, 604931	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
HAAO	100%	100%	100%	98%	Vertebral, cardiac, renal, and limb defects syndrome 1, 617660	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
HABP2	100%	100%	100%	99%	No OMIM disease ID	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME

HACE1	100%	100%	100%	97%	Spastic paraplegia and psychomotor retardation w/wo seizures, 616756	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
HADH	100%	100%	100%	98%	Hyperinsulinemic hypoglycemia, familial, 4, 609975 3-hydroxyacyl-CoA dehydrogenase deficiency, 231530	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
HADHA	100%	100%	100%	98%	HELLP syndrome, maternal, of pregnancy, 609016 Mitochondrial trifunctional protein deficiency, 609015 LCHAD deficiency, 609016 Fatty liver, acute, of pregnancy, 609016	HEART NEUROPATHIES LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS
HADHB	100%	100%	100%	99%	Mitochondrial trifunctional protein deficiency, 609015	HEART NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS
HAGH	100%	100%	100%	98%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME
HAMP	100%	100%	100%	99%	Hemochromatosis, type 2B, 613313	IRON DISORDERS LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING
HAND1	100%	100%	100%	98%	No OMIM disease ID	CONGENITAL HEART DISEASE HEART MENDELIOME
HAND2	100%	100%	98%	73%	No OMIM disease ID	CONGENITAL HEART DISEASE HEART MENDELIOME

HARS1	100%	100%	100%	98%	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504	VISION DISORDERS HEARING IMPAIRMENT NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING
HARS2	100%	100%	100%	99%	Perrault syndrome 2, 614926	HEARING IMPAIRMENT DISORDERS OF SEX DEVELOPMENT MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY
HAVCR2	100%	100%	100%	98%	T-cell lymphoma, subcutaneous panniculitis-like, 618398	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING TUMOR
HAX1	100%	100%	100%	97%	Neutropenia, severe congenital 3, autosomal recessive, 610738	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING TUMOR
HBA1	100%	100%	100%	99%	Hemoglobin H disease, nondeletional, 613978 Thalassemias, alpha-, 604131 Heinz body anemias, alpha-, 140700 Methemoglobinemia, alpha type, 617973 Erythrocytosis 7, 617981	MENDELIOME
HBA2	100%	100%	99%	89%	Heinz body anemia, 140700 Erythrocytosis 7, 617981 Thalassemia, alpha-, 604131 Hemoglobin H disease, deletional and nondeletional, 613978	MENDELIOME
HBB	100%	100%	100%	99%	Methemoglobinemia, beta type, 617971 Thalassemia-beta, dominant inclusion-body, 603902 Sickle cell anemia, 603903 Thalassemia, beta, 613985 Delta-beta thalassemia, 141749 Hereditary persistence of fetal hemoglobin, 141749 Heinz body anemia, 140700 Erythrocytosis 6, 617980	MENDELIOME PRE CONCEPTION SCREENING
HBD	100%	100%	100%	99%	Thalassemia due to Hb Lepore, Thalassemia, delta-,	MENDELIOME
HBG1	98%	94%	95%	72%	Fetal hemoglobin quantitative trait locus 1, 141749	MENDELIOME

HBG2	100%	100%	100%	98%	Fetal hemoglobin quantitative trait locus 1, 141749 Cyanosis, transient neonatal, 613977	MENDELIOME
HCCS	100%	100%	98%	71%	Linear skin defects with multiple congenital anomalies 1, 309801	VISION DISORDERS SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS
HCFC1	100%	99%	98%	77%	Methylmalonic aciduria and homocysteinemia, cblX type, 309541	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME
HCK	100%	100%	100%	99%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
HCN1	99%	99%	99%	96%	Developmental and epileptic encephalopathy 24, 615871 Generalized epilepsy with febrile seizures plus, type 10, 618482	EPILEPSY HNPD INTELLECTUAL DISABILITY MENDELIOME
HCN2	94%	92%	92%	78%	Febrile seizures, familial, 2, 602477 Generalized epilepsy with febrile seizures plus, type 11, 602477	HEART HNPD MENDELIOME
HCN3	100%	100%	100%	99%	No OMIM disease ID	HEART HNPD MENDELIOME
HCN4	100%	100%	100%	96%	Sick sinus syndrome 2, 163800 Brugada syndrome 8, 613123	ANEURYSM DILATED CARDIOMYOPATHY HEART MENDELIOME ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS
HCRT	100%	100%	100%	91%	?Narcolepsy 1, 161400	MENDELIOME
HDAC4	100%	100%	100%	98%	Neurodevelopmental disorder with central hypotonia and dysmorphic facies, 619797	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
HDAC6	100%	99%	98%	75%	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863	INTELLECTUAL DISABILITY MENDELIOME
HDAC8	97%	97%	97%	72%	Cornelia de Lange syndrome 5, 300882	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS

HEATR3	100%	100%	100%	97%	Diamond-Blackfan anemia 21, 620072	INTELLECTUAL DISABILITY MENDELIOME
HEATR5B	100%	100%	100%	98%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
HECW2	100%	100%	100%	98%	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
HELLS	100%	100%	100%	97%	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
HEPACAM	100%	100%	100%	98%	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, w/wo impaired intellectual development, 613926	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
HEPH	99%	99%	98%	73%	No OMIM disease ID	IRON DISORDERS MENDELIOME
HEPHL1	100%	100%	100%	98%	?Abnormal hair, joint laxity, and developmental delay, 261990	MENDELIOME
HERC1	100%	100%	100%	99%	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
HERC2	100%	99%	100%	99%	Intellectual developmental disorder, autosomal recessive 38, 615516	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
HES7	100%	100%	100%	96%	Spondylocostal dysostosis 4, autosomal recessive, 613686	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
HESX1	100%	100%	100%	95%	Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230 Growth hormone deficiency with pituitary anomalies, 182230	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
HEXA	100%	100%	100%	99%	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

HEXB	100%	100%	100%	97%	Sandhoff disease, infantile, juvenile, and adult forms, 268800	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
HEY2	100%	100%	100%	98%	No OMIM disease ID	CONGENITAL HEART DISEASE HEART MENDELIOME
HFE	100%	100%	100%	98%	Hemochromatosis, 235200	HEART IRON DISORDERS LIVER DISORDERS METABOLIC DISORDERS MENDELIOME
HFM1	100%	100%	100%	96%	Premature ovarian failure 9, 615724	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY
HGD	100%	99%	100%	98%	Alkaptonuria, 203500	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
HGF	100%	100%	100%	98%	Deafness, autosomal recessive 39, 608265	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
HGSNAT	92%	92%	100%	98%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544	VISION DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
HHAT	100%	100%	100%	99%	Nivelon-Nivelon-Mabille syndrome, 600092	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
HIBADH	100%	100%	100%	98%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME
HIBCH	100%	100%	100%	98%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
HID1	100%	100%	100%	98%	Developmental and epileptic encephalopathy 105 with hypopituitarism, 619983	INTELLECTUAL DISABILITY MENDELIOME

HIKESHI	100%	100%	100%	98%	Leukodystrophy, hypomyelinating, 13, 616881	MENDELIOME PRE CONCEPTION SCREENING
HINT1	100%	100%	100%	96%	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING
HIVEP2	100%	100%	100%	99%	Intellectual developmental disorder, autosomal dominant 43, 616977	INTELLECTUAL DISABILITY MENDELIOME
HJV	100%	100%	100%	98%	Hemochromatosis, type 2A, 602390	HEART IRON DISORDERS MENDELIOME PRE CONCEPTION SCREENING
HK1	100%	100%	100%	99%	Retinitis pigmentosa 79, 617460 Neuropathy, hereditary motor and sensory, Russe type, 605285 Neurodevelopmental disorder with visual defects and brain anomalies, 618547 Hemolytic anemia due to hexokinase deficiency, 235700	MOVEMENT DISORDERS VISION DISORDERS NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
HKDC1	100%	100%	100%	99%	Retinitis pigmentosa 92, 619614	VISION DISORDERS MENDELIOME
HLCS	100%	100%	100%	99%	Holocarboxylase synthetase deficiency, 253270	SKIN DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
HMBS	100%	100%	100%	99%	Porphyria, acute intermittent, nonerythroid variant, 176000 Porphyria, acute intermittent, 176000	SKIN DISORDERS NEUROPATHIES METABOLIC DISORDERS MENDELIOME
HMGA2	89%	80%	100%	96%	Silver-Russell syndrome 5, 618908	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
HMGB1	100%	100%	100%	96%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
HMGB3	100%	99%	98%	69%	?Microphthalmia, syndromic 13, 300915	SKIN DISORDERS MENDELIOME

HMGCL	100%	100%	100%	98%	HMG-CoA lyase deficiency, 246450	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
HMGCS2	100%	100%	100%	98%	HMG-CoA synthase-2 deficiency, 605911	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
HMOX1	100%	100%	100%	99%	Heme oxygenase-1 deficiency, 614034	IRON DISORDERS PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
HMX1	100%	100%	99%	90%	Oculoauricular syndrome, 612109	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
HNF1A	100%	100%	100%	99%	Hepatic adenoma, somatic, 142330 Diabetes mellitus, insulin-dependent, 20, 612520 MODY, type III, 600496 Renal cell carcinoma, 144700	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME
HNF1B	100%	100%	100%	98%	Type 2 diabetes mellitus, 125853 Renal cysts and diabetes syndrome, 137920	LIVER DISORDERS RENAL DISORDERS MENDELIOME
HNF4A	100%	100%	100%	99%	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026 MODY, type I, 125850	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME
HNMT	100%	100%	99%	97%	Intellectual developmental disorder, autosomal recessive 51, 616739	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
HNRNPA1	100%	100%	100%	99%	?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3, 615424 Amyotrophic lateral sclerosis 20, 615426	MENDELIOME
HNRNPA2B1	100%	100%	100%	97%	?Inclusion body myopathy with early-onset Paget disease w/wo frontotemporal dementia 2, 615422	MENDELIOME
HNRNPD	100%	100%	100%	97%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
HNRNPDL	100%	100%	99%	90%	Muscular dystrophy, limb-girdle, autosomal dominant 3, 609115	MENDELIOME
HNRNPH1	100%	100%	99%	97%	Neurodevelopmental disorder with craniofacial dysmorphism and skeletal defects, 620083	INTELLECTUAL DISABILITY MENDELIOME
HNRNPH2	100%	100%	99%	77%	Intellectual developmental disorder, X-linked syndromic, Bain type, 300986	INTELLECTUAL DISABILITY MENDELIOME

HNRNPK	100%	100%	100%	98%	Au-Kline syndrome, 616580	INTELLECTUAL DISABILITY MENDELIOME
HNRNPU	100%	100%	100%	97%	Developmental and epileptic encephalopathy 54, 617391	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
HOGA1	100%	100%	100%	99%	Hyperoxaluria, primary, type III, 613616	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
HOMER2	100%	99%	100%	98%	?Deafness, autosomal dominant 68, 616707	HEARING IMPAIRMENT MENDELIOME
HOXA1	100%	100%	100%	97%	Bosley-Salih-Alorainy syndrome, 601536 Athabaskan brainstem dysgenesis syndrome, 601536	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
HOXA11	100%	100%	100%	96%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
HOXA13	99%	98%	93%	60%	Hand-foot-uterus syndrome, 140000 ?Guttmacher syndrome, 176305	DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
HOXA2	100%	100%	100%	97%	Microtia w/wo hearing impairment (AD), 612290 ?Microtia, hearing impairment, and cleft palate (AR), 612290	CRANIOFACIAL ANOMALIES MENDELIOME PRE CONCEPTION SCREENING
HOXB1	100%	100%	100%	99%	Facial paresis, hereditary congenital, 3, 614744	MENDELIOME PRE CONCEPTION SCREENING
HOXB13	100%	100%	100%	97%	No OMIM disease ID	MENDELIOME TUMOR
HOXC13	100%	100%	100%	94%	Ectodermal dysplasia 9, hair/nail type, 614931	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
HOXD10	100%	100%	100%	98%	Vertical talus, congenital, 192950 Charcot-Marie-Tooth disease, foot deformity of, 192950	NEUROPATHIES MENDELIOME
HOXD13	100%	100%	100%	96%	Syndactyly, type V, 186300 Synpolydactyly 1, 186000 Brachydactyly, type E, 113300 Brachydactyly, type D, 113200 ?Brachydactyly-syndactyly syndrome, 610713	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME

HPCA	100%	100%	100%	97%	Dystonia 2, torsion, autosomal recessive, 224500	MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING
HPD	100%	100%	100%	97%	Hawkinsinuria, 140350 Tyrosinemia, type III, 276710	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
HPDL	100%	100%	100%	98%	Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026 Spastic paraplegia 83, autosomal recessive, 619027	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
HPGD	100%	100%	100%	97%	?Digital clubbing, isolated congenital, 119900 Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100 Cranioosteoarthropathy, 259100	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
HPRT1	100%	100%	98%	73%	Hyperuricemia, HRPT-related, 300323 Lesch-Nyhan syndrome, 300322	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME
HPS1	100%	100%	100%	99%	Hermansky-Pudlak syndrome 1, 203300	VISION DISORDERS SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
HPS3	100%	100%	100%	97%	Hermansky-Pudlak syndrome 3, 614072	VISION DISORDERS SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
HPS4	100%	100%	100%	99%	Hermansky-Pudlak syndrome 4, 614073	VISION DISORDERS SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
HPS5	100%	100%	100%	98%	Hermansky-Pudlak syndrome 5, 614074	VISION DISORDERS SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING

HPS6	100%	100%	100%	98%	Hermansky-Pudlak syndrome 6, 614075	VISION DISORDERS SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
HPSE2	100%	100%	100%	98%	Urofacial syndrome 1, 236730	MENDELIOME PRE CONCEPTION SCREENING
HR	100%	100%	100%	99%	Atrichia with papular lesions, 209500 Alopecia universalis, 203655	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
HRAS	100%	100%	100%	99%	Bladder cancer, somatic, 109800 Thyroid carcinoma, follicular, somatic, 188470 Congenital myopathy with excess of muscle spindles, 218040 Nevus sebaceous or woolly hair nevus, somatic, 162900 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Spitz nevus or nevus spilus, somatic, 137550 Costello syndrome, 218040	VISION DISORDERS SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME AND RASOPATHY
HRG	100%	100%	100%	98%	Thrombophilia 11 due to HRG deficiency, 613116	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME
HROB	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME PRIMARY OVARIAN INSUFFICIENCY
HS2ST1	100%	100%	100%	98%	Neurofacioskeletal syndrome w/wo renal agenesis, 619194	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
HS3ST6	100%	99%	99%	94%	?Angioedema, hereditary, 8, 619367	PRIMARY IMMUNODEFICIENCY MENDELIOME
HS6ST1	100%	100%	100%	92%	No OMIM disease ID	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM METABOLIC DISORDERS MENDELIOME
HS6ST2	99%	99%	97%	69%	?Paganini-Miozzo syndrome, 301025	MENDELIOME
HSCB	100%	100%	100%	97%	?Anemia, sideroblastic, 5, 619523	IRON DISORDERS MENDELIOME
HSD11B1	100%	100%	100%	99%	Cortisone reductase deficiency 2, 614662	METABOLIC DISORDERS MENDELIOME
HSD11B2	100%	100%	99%	94%	Apparent mineralocorticoid excess, 218030	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING

HSD17B10	100%	99%	98%	72%	HSD10 mitochondrial disease, 300438	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS
HSD17B3	100%	100%	100%	98%	Pseudohermaphroditism, male, with gynecomastia, 264300	DISORDERS OF SEX DEVELOPMENT METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
HSD17B4	96%	96%	100%	98%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400	MOVEMENT DISORDERS HEARING IMPAIRMENT DISORDERS OF SEX DEVELOPMENT EPILEPSY NEUROPATHIES LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY
HSD3B2	99%	99%	100%	98%	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810	DISORDERS OF SEX DEVELOPMENT METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
HSD3B7	100%	100%	100%	99%	Bile acid synthesis defect, congenital, 1, 607765	LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
HSF2	100%	100%	100%	98%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
HSF2BP	100%	100%	100%	98%	Premature ovarian failure 19, 619245	MENDELIOME
HSF4	100%	100%	100%	99%	Cataract 5, multiple types, 116800	VISION DISORDERS MENDELIOME
HSFY1	49%	49%	47%	17%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
HSFY2	49%	49%	47%	18%	No OMIM disease ID	MALE INFERTILITY MENDELIOME

HSPA9	100%	100%	100%	98%	Even-plus syndrome, 616854 Anemia, sideroblastic, 4, 182170	IRON DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
HSPB1	100%	100%	100%	97%	Neuronopathy, distal hereditary motor, type IIB, 608634 Charcot-Marie-Tooth disease, axonal, type 2F, 606595	NEUROPATHIES HNPD MENDELIOME
HSPB3	100%	100%	100%	98%	?Neuronopathy, distal hereditary motor, type IIC, 613376	NEUROPATHIES MENDELIOME
HSPB6	100%	100%	99%	95%	No OMIM disease ID	HEART MENDELIOME
HSPB8	100%	100%	100%	98%	Neuronopathy, distal hereditary motor, type IIA, 158590 Charcot-Marie-Tooth disease, axonal, type 2L, 608673	NEUROPATHIES MENDELIOME
HSPD1	100%	100%	100%	98%	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233	MOVEMENT DISORDERS HEART INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
HSPG2	100%	100%	100%	99%	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800	VISION DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
HTR1A	100%	100%	100%	99%	Periodic fever, menstrual cycle dependent, 614674	MENDELIOME
HTRA1	100%	100%	100%	95%	CARASIL syndrome, 600142 Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
HTRA2	100%	100%	100%	98%	3-methylglutaconic aciduria, type VIII, 617248	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
HTT	100%	100%	100%	98%	Lopes-Maciel-Rodan syndrome, 617435 Huntington disease, 143100	MENDELIOME
HUWE1	100%	99%	98%	72%	Intellectual developmental disorder, X-linked syndromic, Turner type, 309590	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME

HYAL1	100%	100%	100%	98%	Mucopolysaccharidosis type IX, 601492	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
HYAL2	100%	100%	100%	99%	No OMIM disease ID	CRANIOFACIAL ANOMALIES MENDELIOME
HYDIN	100%	100%	100%	98%	Ciliary dyskinesia, primary, 5, 608647	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING
HYLS1	100%	100%	100%	99%	Hydrolethalus syndrome, 236680	CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS
HYOU1	100%	100%	100%	99%	?Immunodeficiency 59 and hypoglycemia, 233600	PRIMARY IMMUNODEFICIENCY MENDELIOME
IARS1	100%	100%	100%	98%	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093	LIVER DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
IARS2	100%	100%	100%	98%	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
IBA57	100%	100%	100%	99%	Multiple mitochondrial dysfunctions syndrome 3, 615330 ?Spastic paraplegia 74, autosomal recessive, 616451	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
ICOS	100%	100%	100%	97%	Immunodeficiency, common variable, 1, 607594	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
ICOSLG	100%	100%	100%	99%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
ID4	100%	100%	100%	91%	No OMIM disease ID	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME

IDH1	100%	100%	100%	99%	No OMIM disease ID	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME TUMOR
IDH2	100%	100%	100%	98%	D-2-hydroxyglutaric aciduria 2, 613657	EPILEPSY SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS TUMOR
IDH3A	100%	100%	100%	98%	Retinitis pigmentosa 90, 619007	VISION DISORDERS MENDELIOME
IDH3B	100%	100%	100%	99%	Retinitis pigmentosa 46, 612572	VISION DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
IDI1	100%	100%	100%	96%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME
IDS	100%	100%	97%	71%	Mucopolysaccharidosis II, 309900	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME
IDUA	100%	100%	100%	97%	Mucopolysaccharidosis Is, 607016 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Ih, 607014	SKIN DISORDERS HEART SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
IER3IP1	100%	100%	100%	98%	Microcephaly, epilepsy, and diabetes syndrome, 614231	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
IFIH1	100%	100%	100%	98%	Immunodeficiency 95, 619773 Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250	EPILEPSY PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME

IFITM5	100%	100%	100%	99%	Osteogenesis imperfecta, type V, 610967	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
IFNAR1	97%	97%	100%	97%	Immunodeficiency 106, susceptibility to viral infections, 619935	PRIMARY IMMUNODEFICIENCY MENDELIOME
IFNAR2	100%	100%	100%	98%	Immunodeficiency 45, 616669	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
IFNG	100%	100%	100%	97%	?Immunodeficiency 69, mycobacteriosis, 618963	PRIMARY IMMUNODEFICIENCY RENAL DISORDERS MENDELIOME
IFNGR1	100%	100%	100%	98%	Immunodeficiency 27A, mycobacteriosis, AR, 209950 Immunodeficiency 27B, mycobacteriosis, AD, 615978	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
IFNGR2	100%	100%	100%	97%	Immunodeficiency 28, mycobacteriosis, 614889	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
IFNLR1	100%	100%	100%	97%	No OMIM disease ID	HEARING IMPAIRMENT MENDELIOME
IFRD1	100%	100%	100%	97%	No OMIM disease ID	NEUROPATHIES MENDELIOME
IFT122	100%	100%	100%	99%	Cranioectodermal dysplasia 1, 218330	CRANIOFACIAL ANOMALIES CILIOPATHIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
IFT140	100%	100%	100%	99%	Short-rib thoracic dysplasia 9 w/wo polydactyly, 266920 Retinitis pigmentosa 80, 617781	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS

IFT172	100%	100%	100%	99%	Retinitis pigmentosa 71, 616394 Bardet-Biedl syndrome 20, 619471 Short-rib thoracic dysplasia 10 w/wo polydactyly, 615630	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS
IFT27	100%	100%	100%	99%	Bardet-Biedl syndrome 19, 615996	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
IFT43	100%	100%	100%	98%	?Cranioectodermal dysplasia 3, 614099 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866	VISION DISORDERS CRANIOFACIAL ANOMALIES CILIOPATHIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
IFT52	100%	100%	100%	98%	Short-rib thoracic dysplasia 16 w/wo polydactyly, 617102	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
IFT57	100%	100%	100%	98%	?Orofaciodigital syndrome XVIII, 617927	MENDELIOME PRE CONCEPTION SCREENING SCHISIS
IFT74	100%	100%	100%	97%	Bardet-Biedl syndrome 22, 617119 Spermatogenic failure 58, 619585 Joubert syndrome 40, 619582	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

IFT80	100%	100%	100%	98%	Short-rib thoracic dysplasia 2 w/wo polydactyly, 611263	CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS
IFT81	94%	94%	100%	98%	Short-rib thoracic dysplasia 19 w/wo polydactyly, 617895	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
IFT88	100%	100%	100%	97%	No OMIM disease ID	CRANIOFACIAL ANOMALIES MENDELIOME
IGBP1	100%	99%	97%	68%	?Corpus callosum, agenesis of, with impaired intellectual development, ocular coloboma and micrognathia, 300472	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME
IGF1	100%	100%	100%	98%	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
IGF1R	100%	100%	100%	99%	Insulin-like growth factor I, resistance to, 270450	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
IGF2	100%	100%	100%	99%	Silver-Russell syndrome 3, 616489	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
IGF2R	100%	100%	100%	99%	Hepatocellular carcinoma, somatic, 114550	MENDELIOME
IGFALS	100%	100%	100%	99%	Acid-labile subunit, deficiency of, 615961	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
IGFBP7	100%	100%	100%	95%	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis, 614224	MENDELIOME PRE CONCEPTION SCREENING
IGHG2	100%	100%	100%	97%	IgG2 deficiency, selective,	MENDELIOME
IGHM	100%	100%	100%	99%	Agammaglobulinemia 1, 601495	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING

IGHMBP2	100%	100%	100%	99%	Neuronopathy, distal hereditary motor, type VI, 604320 Charcot-Marie-Tooth disease, axonal, type 2S, 616155	FETAL AKINESIA NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
IGKC	100%	100%	100%	99%	Kappa light chain deficiency, 614102	MENDELIOME PRE CONCEPTION SCREENING
IGLL1	100%	100%	100%	99%	Agammaglobulinemia 2, 613500	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
IGSF1	100%	99%	98%	70%	Hypothyroidism, central, and testicular enlargement, 300888	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
IGSF10	100%	100%	100%	99%	No OMIM disease ID	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME
IGSF3	100%	100%	100%	99%	?Lacrimal duct defect, 149700	VISION DISORDERS MENDELIOME
IHH	100%	100%	100%	96%	Acrocapitofemoral dysplasia, 607778 Brachydactyly, type A1, 112500	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
IKBKB	100%	100%	99%	97%	Immunodeficiency 15B, 615592 Immunodeficiency 15A, 618204	PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
IKBKG	99%	98%	98%	76%	Incontinentia pigmenti, 308300 Ectodermal dysplasia and immunodeficiency 1, 300291 Immunodeficiency 33, 300636 Autoinflammatory disease, systemic, X-linked, 301081	VISION DISORDERS CRANIOFACIAL ANOMALIES SKIN DISORDERS PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
IKZF1	100%	100%	100%	99%	Immunodeficiency, common variable, 13, 616873	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCY MENDELIOME TUMOR
IKZF2	100%	100%	100%	99%	No OMIM disease ID	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCY MENDELIOME

IKZF3	100%	100%	100%	99%	?Immunodeficiency 84, 619437	PRIMARY IMMUNODEFICIENCY MENDELIOME
IKZF5	100%	100%	100%	97%	Thrombocytopenia, autosomal dominant, 7, 619130	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME
IL10	100%	100%	100%	99%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
IL10RA	100%	100%	100%	99%	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
IL10RB	100%	100%	100%	98%	Inflammatory bowel disease 25, early onset, autosomal recessive, 612567	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
IL11RA	100%	100%	100%	98%	Craniosynostosis and dental anomalies, 614188	CRANIOFACIAL ANOMALIES MENDELIOME PRE CONCEPTION SCREENING
IL12B	100%	100%	100%	99%	Immunodeficiency 29, mycobacteriosis, 614890	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
IL12RB1	94%	94%	100%	98%	Immunodeficiency 30, 614891	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
IL17F	100%	100%	100%	99%	?Candidiasis, familial, 6, autosomal dominant, 613956	PRIMARY IMMUNODEFICIENCY MENDELIOME
IL17RA	100%	100%	100%	98%	Immunodeficiency 51, 613953	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
IL17RC	100%	100%	100%	99%	Candidiasis, familial, 9, 616445	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
IL17RD	100%	100%	100%	99%	Hypogonadotropic hypogonadism 18 w/wo anosmia, 615267	SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME
IL18BP	100%	100%	100%	99%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
IL1RAPL1	100%	100%	97%	70%	Intellectual developmental disorder, X-linked 21, 300143	INTELLECTUAL DISABILITY MENDELIOME

IL1RN	100%	100%	100%	98%	Interleukin 1 receptor antagonist deficiency, 612852	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
IL2	100%	99%	100%	97%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
IL21	100%	100%	100%	96%	?Immunodeficiency, common variable, 11, 615767	PRIMARY IMMUNODEFICIENCY MENDELIOME
IL21R	100%	100%	100%	99%	Immunodeficiency 56, 615207	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
IL2RA	100%	100%	100%	99%	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
IL2RB	100%	100%	100%	98%	Immunodeficiency 63 with lymphoproliferation and autoimmunity, 618495	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
IL2RG	100%	100%	98%	71%	Combined immunodeficiency, X-linked, moderate, 312863 Severe combined immunodeficiency, X-linked, 300400	PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SEVERE COMBINED IMMUNODEFICIENCY (SCID)
IL31RA	100%	100%	100%	98%	?Amyloidosis, primary localized cutaneous, 2, 613955	SKIN DISORDERS MENDELIOME
IL36RN	100%	100%	100%	99%	Psoriasis 14, pustular, 614204	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
IL37	100%	100%	100%	96%	?Inflammatory bowel disease (infantile ulcerative colitis) 31, 619398	MENDELIOME
IL6R	92%	92%	100%	99%	Hyper-IgE recurrent infection syndrome 5, autosomal recessive, 618944	PRIMARY IMMUNODEFICIENCY MENDELIOME
IL6ST	100%	100%	100%	98%	Stuve-Wiedemann syndrome 2, 619751 Hyper-IgE recurrent infection syndrome 4A, autosomal dominant, 619752 ?Immunodeficiency 94 with autoinflammation and dysmorphic facies, 619750 Hyper-IgE recurrent infection syndrome 4B, autosomal recessive, 618523	CRANIOFACIAL ANOMALIES PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING

IL7R	100%	100%	100%	98%	Immunodeficiency 104, severe combined, 608971	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)
ILDR1	100%	100%	100%	99%	Deafness, autosomal recessive 42, 609646	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
ILK	100%	100%	100%	99%	No OMIM disease ID	HEART MENDELIOME
IMPA1	100%	100%	100%	98%	Intellectual developmental disorder, autosomal recessive 59, 617323	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
IMPAD1	100%	100%	100%	98%	Chondrodysplasia with joint dislocations, GPAPP type, 614078	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS
IMPDH1	100%	100%	100%	98%	Retinitis pigmentosa 10, 180105 Leber congenital amaurosis 11, 613837	VISION DISORDERS METABOLIC DISORDERS MENDELIOME
IMPG1	100%	99%	100%	98%	Macular dystrophy, vitelliform, 4, 616151 Retinitis pigmentosa 91, 153870	VISION DISORDERS MENDELIOME
IMPG2	100%	100%	100%	97%	Retinitis pigmentosa 56, 613581 Macular dystrophy, vitelliform, 5, 616152	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
INF2	100%	99%	99%	96%	Glomerulosclerosis, focal segmental, 5, 613237 Charcot-Marie-Tooth disease, dominant intermediate E, 614455	NEUROPATHIES RENAL DISORDERS MENDELIOME
ING1	100%	100%	100%	97%	Squamous cell carcinoma, head and neck, somatic, 275355	MENDELIOME
INO80	100%	100%	100%	99%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
INPP5E	100%	100%	100%	96%	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156	VISION DISORDERS CILIOPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING

INPP5K	100%	100%	100%	98%	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
INPPL1	100%	100%	100%	98%	Opsismodysplasia, 258480	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
INS	100%	100%	100%	99%	Diabetes mellitus, insulin-dependent, 2, 125852 Maturity-onset diabetes of the young, type 10, 613370 Hyperproinsulinemia, 616214 Diabetes mellitus, permanent neonatal 4, 618858	MENDELIOME
INSL3	78%	78%	100%	99%	Cryptorchidism, 219050	MENDELIOME
INSR	100%	100%	100%	98%	Rabson-Mendenhall syndrome, 262190 Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Donohue syndrome, 246200 Hyperinsulinemic hypoglycemia, familial, 5, 609968	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
INTS1	100%	100%	100%	99%	Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies, 618571	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
INTS8	100%	100%	100%	97%	?Neurodevelopmental disorder with cerebellar hypoplasia and spasticity, 618572	MENDELIOME PRE CONCEPTION SCREENING
INTU	100%	100%	100%	97%	?Orofaciodigital syndrome XVII, 617926 ?Short-rib thoracic dysplasia 20 with polydactyly, 617925	CRANIOFACIAL ANOMALIES CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS
INVS	100%	100%	100%	99%	Nephronophthisis 2, infantile, 602088	VISION DISORDERS CILIOPATHIES LIVER DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
IPMK	100%	100%	100%	98%	No OMIM disease ID	MENDELIOME TUMOR

IPO8	100%	100%	100%	98%	VISS syndrome, 619472	ANEURYSM PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
IQCB1	100%	100%	100%	98%	Senior-Loken syndrome 5, 609254	VISION DISORDERS CILIOPATHIES RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
IQCE	100%	100%	100%	98%	Polydactyly, postaxial, type A7, 617642	MENDELIOME
IQSEC1	100%	99%	99%	95%	Intellectual developmental disorder with short stature and behavioral abnormalities, 618687	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
IQSEC2	99%	98%	94%	62%	Intellectual developmental disorder, X-linked 1, 309530	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
IRAK1	100%	99%	95%	71%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
IRAK4	100%	100%	100%	98%	Immunodeficiency 67, 607676	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
IREB2	100%	100%	100%	98%	Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
IRF1	100%	100%	100%	98%	Nonsmall cell lung cancer, somatic, 211980 Gastric cancer, somatic, 613659 Myelodysplastic syndrome, preleukemic, Myelogenous leukemia, acute,	MENDELIOME
IRF2BP2	100%	100%	100%	89%	?Immunodeficiency, common variable, 14, 617765	PRIMARY IMMUNODEFICIENCY MENDELIOME
IRF2BPL	100%	100%	99%	91%	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
IRF3	100%	100%	100%	98%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
IRF4	100%	100%	100%	97%	No OMIM disease ID	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME

IRF6	100%	100%	100%	99%	Popliteal pterygium syndrome 1, 119500 van der Woude syndrome 1, 119300	CRANIOFACIAL ANOMALIES SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT MENDELIOME SCHISIS
IRF7	100%	100%	100%	99%	?Immunodeficiency 39, 616345	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
IRF8	100%	100%	100%	98%	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893 Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
IRF9	100%	100%	100%	99%	Immunodeficiency 65, susceptibility to viral infections, 618648	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
IRGM	100%	100%	100%	100%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
IRS4	100%	99%	93%	57%	Hypothyroidism, congenital, nongoitrous, 9, 301035	MENDELIOME
IRX1	100%	99%	99%	91%	No OMIM disease ID	VISION DISORDERS MENDELIOME
IRX5	100%	100%	99%	91%	Hamamy syndrome, 611174	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ISCA1	92%	92%	100%	98%	Multiple mitochondrial dysfunctions syndrome 5, 617613	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
ISCA2	100%	100%	100%	98%	Multiple mitochondrial dysfunctions syndrome 4, 616370	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
ISCU	100%	100%	100%	99%	Myopathy with lactic acidosis, hereditary, 255125	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS
ISG15	100%	100%	100%	100%	Immunodeficiency 38, 616126	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
ITCH	96%	96%	100%	98%	Autoimmune disease, multisystem, with facial dysmorphism, 613385	PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING

ITGA2	100%	99%	100%	98%	No OMIM disease ID	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME
ITGA2B	100%	100%	100%	99%	Glanzmann thrombasthenia 1, 273800 Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Thrombocytopenia, neonatal alloimmune, BAK antigen related,	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ITGA3	100%	100%	100%	99%	Epidermolysis bullosa, junctional 7, with interstitial lung disease and nephrotic syndrome, 614748	SKIN DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ITGA6	100%	100%	100%	98%	Epidermolysis bullosa, junctional 6, with pyloric atresia, 619817	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ITGA7	100%	100%	100%	99%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204	HEART INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
ITGA8	100%	100%	100%	98%	Renal hypodysplasia/aplasia 1, 191830	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ITGB2	100%	100%	100%	99%	Leukocyte adhesion deficiency, 116920	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
ITGB3	100%	100%	100%	98%	Bleeding disorder, platelet-type, 24, autosomal dominant, 619271 Glanzmann thrombasthenia 2, 619267 Thrombocytopenia, neonatal alloimmune, Purpura, posttransfusion,	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ITGB4	100%	100%	100%	98%	Epidermolysis bullosa, junctional 5B, with pyloric atresia, 226730 Epidermolysis bullosa, junctional 5A, intermediate, 619816	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ITGB6	100%	100%	100%	98%	Amelogenesis imperfecta, type IH, 616221	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ITK	100%	100%	100%	99%	Lymphoproliferative syndrome 1, 613011	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING TUMOR
ITM2B	100%	100%	99%	97%	?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079 Dementia, familial British, 176500 Dementia, familial Danish, 117300	VISION DISORDERS MENDELIOME

ITPA	100%	100%	100%	97%	Developmental and epileptic encephalopathy 35, 616647	EPILEPSY HEART METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ITPKB	100%	100%	100%	98%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME SEVERE COMBINED IMMUNODEFICIENCY (SCID)
ITPR1	100%	100%	100%	98%	Gillespie syndrome, 206700 Spinocerebellar ataxia 29, congenital nonprogressive, 117360 Spinocerebellar ataxia 15, 606658	MOVEMENT DISORDERS VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ITPR2	100%	99%	100%	98%	?Anhidrosis, isolated, with normal sweat glands, 106190	MENDELIOME
ITPR3	100%	100%	100%	99%	Charcot-Marie-Tooth disease, demyelinating, type 1J, 620111	NEUROPATHIES PRIMARY IMMUNODEFICIENCY MENDELIOME
ITSN1	100%	100%	100%	98%	No OMIM disease ID	RENAL DISORDERS MENDELIOME
ITSN2	100%	100%	99%	97%	No OMIM disease ID	RENAL DISORDERS MENDELIOME
IVD	100%	100%	100%	99%	Isovaleric acidemia, 243500	HEREDITARY BONE MARROW FAILURE METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
IVNS1ABP	100%	100%	100%	97%	Immunodeficiency 70, 618969	PRIMARY IMMUNODEFICIENCY MENDELIOME
IYD	100%	100%	100%	97%	Thyroid dyshormonogenesis 4, 274800	MENDELIOME PRE CONCEPTION SCREENING
JAG1	100%	100%	100%	99%	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Charcot-Marie-Tooth disease, axonal, type 2HH, 619574 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500	VISION DISORDERS CONGENITAL HEART DISEASE HEART NEUROPATHIES LIVER DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME

JAG2	100%	99%	99%	97%	Muscular dystrophy, limb-girdle, autosomal recessive 27, 619566	INTELLECTUAL DISABILITY MENDELIOME
JAGN1	100%	100%	100%	99%	Neutropenia, severe congenital, 6, autosomal recessive, 616022	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
JAK1	100%	100%	100%	98%	Autoinflammation, immune dysregulation, and eosinophilia, 618999	PRIMARY IMMUNODEFICIENCY MENDELIOME
JAK2	100%	100%	100%	98%	Myelofibrosis, somatic, 254450 Erythrocytosis, somatic, 133100 Leukemia, acute myeloid, somatic, 601626 Thrombocythemia 3, 614521 Polycythemia vera, somatic, 263300	HEMOSTATIC/THROMBOTIC DISORDERS IRON DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME
JAK3	100%	100%	100%	99%	SCID, autosomal recessive, T-negative/B-positive type, 600802	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)
JAM2	92%	92%	100%	98%	Basal ganglia calcification, idiopathic, 8, autosomal recessive, 618824	MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING
JAM3	100%	100%	100%	98%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730	MOVEMENT DISORDERS VISION DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
JARID2	100%	100%	100%	99%	Developmental delay with variable intellectual disability and dysmorphic facies, 620098	INTELLECTUAL DISABILITY MENDELIOME
JMJD1C	100%	100%	100%	98%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
JPH1	100%	100%	100%	98%	?Charcot-Marie-Tooth disease, axonal, autosomal dominant, type 2K, 607831	MENDELIOME
JPH2	100%	99%	100%	99%	Cardiomyopathy, dilated, 2E, 619492 Cardiomyopathy, hypertrophic, 17, 613873	DILATED CARDIOMYOPATHY HEART HYPERTROPHIC CARDIOMYOPATHY MENDELIOME
JPH3	100%	100%	100%	98%	Huntington disease-like 2, 606438	MENDELIOME

JUP	100%	100%	100%	99%	Naxos disease, 601214 ?Arrhythmogenic right ventricular dysplasia 12, 611528	ARRHYTHMOGENIC CARDIOMYOPATHY (ACM/ARVC) SKIN DISORDERS HEART MENDELIOME PRE CONCEPTION SCREENING ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS
KALRN	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME PRE CONCEPTION SCREENING
KANK1	100%	100%	100%	99%	Cerebral palsy, spastic quadriplegic, 2, 612900	INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME
KANK2	100%	100%	100%	99%	Nephrotic syndrome, type 16, 617783 Palmoplantar keratoderma and woolly hair, 616099	SKIN DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
KANSL1	100%	100%	100%	99%	Koolen-De Vries syndrome, 610443	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME SCHISIS
KARS1	100%	100%	100%	98%	Deafness, autosomal recessive 89, 613916 Leukoencephalopathy, progressive, infantile-onset, w/wo deafness, 619147 ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness, congenital, and adult-onset progressive leukoencephalopathy, 619196	HEARING IMPAIRMENT NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
KAT5	100%	100%	100%	98%	Neurodevelopmental disorder with dysmorphic facies, sleep disturbance, and brain abnormalities, 619103	INTELLECTUAL DISABILITY MENDELIOME
KAT6A	100%	100%	100%	98%	Arboleda-Tham syndrome, 616268	INTELLECTUAL DISABILITY MENDELIOME SCHISIS
KAT6B	100%	100%	100%	98%	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170	CRANIOFACIAL ANOMALIES CONGENITAL HEART DISEASE SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
KAT8	100%	100%	99%	95%	Li-Ghorgani-Weisz-Hubshman syndrome, 618974	INTELLECTUAL DISABILITY MENDELIOME

KATNB1	100%	100%	100%	99%	Lissencephaly 6, with microcephaly, 616212	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
KBTBD13	100%	100%	100%	99%	Nemaline myopathy 6, autosomal dominant, 609273	HEART NEUROPATHIES MENDELIOME MUSCLE DISORDERS
KCNA1	100%	100%	100%	99%	Episodic ataxia/myokymia syndrome, 160120	MOVEMENT DISORDERS EPILEPSY MENDELIOME
KCNA2	100%	100%	100%	99%	Developmental and epileptic encephalopathy 32, 616366	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME
KCNA4	100%	100%	100%	97%	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284	INTELLECTUAL DISABILITY MENDELIOME
KCNA5	100%	100%	100%	98%	Atrial fibrillation, familial, 7, 612240	HEART MENDELIOME
KCNB1	100%	100%	100%	99%	Developmental and epileptic encephalopathy 26, 616056	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
KCNC1	100%	100%	100%	99%	Epilepsy, progressive myoclonic 7, 616187	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
KCNC3	99%	98%	99%	84%	Spinocerebellar ataxia 13, 605259	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME
KCND2	99%	99%	100%	98%	No OMIM disease ID	HEART MENDELIOME
KCND3	100%	100%	100%	99%	Spinocerebellar ataxia 19, 607346 Brugada syndrome 9, 616399	MOVEMENT DISORDERS EPILEPSY HEART MENDELIOME

KCNE1	100%	100%	100%	99%	Jervell and Lange-Nielsen syndrome 2, 612347 Long QT syndrome 5, 613695	HEARING IMPAIRMENT HEART LONG QT SYNDROME MENDELIOME PRE CONCEPTION SCREENING ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS
KCNE2	100%	100%	100%	99%	Long QT syndrome 6, 613693 Atrial fibrillation, familial, 4, 611493	HEART LONG QT SYNDROME MENDELIOME ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS
KCNE3	100%	100%	100%	99%	?Brugada syndrome 6, 613119	HEART MENDELIOME
KCNE4	100%	100%	100%	99%	No OMIM disease ID	HEART MENDELIOME
KCNE5	100%	99%	98%	73%	No OMIM disease ID	HEART MENDELIOME
KCNH1	98%	98%	100%	98%	Zimmermann-Laband syndrome 1, 135500 Temple-Baraitser syndrome, 611816	SKIN DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
KCNH2	100%	100%	100%	97%	Short QT syndrome 1, 609620 Long QT syndrome 2, 613688	HEART LONG QT SYNDROME MENDELIOME ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS
KCNJ1	100%	100%	100%	98%	Bartter syndrome, type 2, 241200	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
KCNJ10	100%	100%	100%	99%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780	MOVEMENT DISORDERS HEARING IMPAIRMENT EPILEPSY INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING

KCNJ11	100%	100%	100%	99%	Diabetes, permanent neonatal 2, w/wo neurologic features, 618856 Maturity-onset diabetes of the young, type 13, 616329 Diabetes mellitus, transient neonatal 3, 610582 Hyperinsulinemic hypoglycemia, familial, 2, 601820	EPILEPSY HEART METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
KCNJ13	100%	100%	100%	99%	Snowflake vitreoretinal degeneration, 193230 Leber congenital amaurosis 16, 614186	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
KCNJ16	100%	100%	100%	99%	Hypokalemic tubulopathy and deafness, 619406	RENAL DISORDERS MENDELIOME
KCNJ2	100%	100%	100%	99%	Atrial fibrillation, familial, 9, 613980 Andersen syndrome, 170390 Short QT syndrome 3, 609622	HEART SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS SCHISIS MUSCLE DISORDERS
KCNJ5	100%	100%	100%	99%	Long QT syndrome 13, 613485 Hyperaldosteronism, familial, type III, 613677	HEART RENAL DISORDERS MENDELIOME
KCNJ6	100%	100%	100%	99%	Keppen-Lubinsky syndrome, 614098	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME
KCNJ8	100%	100%	100%	99%	No OMIM disease ID	HEART MENDELIOME
KCNK3	100%	100%	100%	96%	Pulmonary hypertension, primary, 4, 615344	HEART INTELLECTUAL DISABILITY MENDELIOME
KCNK4	100%	100%	100%	99%	Facial dysmorphism, hypertrichosis, epilepsy, intellectual/developmental delay, and gingival overgrowth syndrome, 618381	INTELLECTUAL DISABILITY MENDELIOME
KCNK9	100%	100%	99%	96%	Birk-Barel syndrome, 612292	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME SCHISIS
KCNMA1	100%	99%	100%	97%	Paroxysmal nonkinesigenic dyskinesia, 3, w/wo generalized epilepsy, 609446 Cerebellar atrophy, developmental delay, and seizures, 617643 Liang-Wang syndrome, 618729	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

KCNN2	99%	99%	100%	99%	?Dystonia 34, myoclonic, 619724 Neurodevelopmental disorder w/wo variable movement or behavioral abnormalities, 619725	INTELLECTUAL DISABILITY MENDELIOME
KCNN3	100%	100%	100%	98%	Zimmermann-Laband syndrome 3, 618658	HEART INTELLECTUAL DISABILITY MENDELIOME
KCNN4	100%	100%	100%	98%	Dehydrated hereditary stomatocytosis 2, 616689	MENDELIOME
KCNQ1	100%	100%	100%	97%	Short QT syndrome 2, 609621 Atrial fibrillation, familial, 3, 607554 Long QT syndrome 1, 192500 Jervell and Lange-Nielsen syndrome, 220400	HEARING IMPAIRMENT HEART LONG QT SYNDROME MENDELIOME PRE CONCEPTION SCREENING ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS
KCNQ1OT1	NC	NC	NC	NC	Beckwith-Wiedemann syndrome, 130650	MENDELIOME
KCNQ2	100%	100%	100%	99%	Developmental and epileptic encephalopathy 7, 613720 Seizures, benign neonatal, 1, 121200 Myokymia, 121200	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
KCNQ3	100%	100%	100%	97%	Seizures, benign neonatal, 2, 121201	EPILEPSY HNPD INTELLECTUAL DISABILITY MENDELIOME
KCNQ4	100%	99%	100%	96%	Deafness, autosomal dominant 2A, 600101	HEARING IMPAIRMENT MENDELIOME
KCNQ5	100%	100%	100%	98%	Intellectual developmental disorder, autosomal dominant 46, 617601	INTELLECTUAL DISABILITY MENDELIOME
KCNT1	100%	100%	100%	98%	Developmental and epileptic encephalopathy 14, 614959 Epilepsy nocturnal frontal lobe, 5, 615005	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
KCNT2	99%	99%	100%	98%	Developmental and epileptic encephalopathy 57, 617771	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
KCNV2	100%	100%	100%	99%	Retinal cone dystrophy 3B, 610356	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
KCTD1	100%	100%	100%	95%	Scalp-ear-nipple syndrome, 181270	MENDELIOME
KCTD17	100%	100%	100%	95%	Dystonia 26, myoclonic, 616398	MENDELIOME
KCTD3	100%	100%	100%	95%	No OMIM disease ID	CILIOPATHIES INTELLECTUAL DISABILITY MENDELIOME

KCTD7	100%	100%	100%	98%	Epilepsy, progressive myoclonic 3, w/wo intracellular inclusions, 611726	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
KDELR2	100%	100%	100%	98%	Osteogenesis imperfecta, type XXI, 619131	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
KDF1	100%	100%	100%	99%	?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/nail type, 617337	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME
KDM1A	100%	100%	100%	98%	Cleft palate, psychomotor retardation, and distinctive facial features, 616728	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME
KDM3B	100%	100%	100%	98%	Diets-Jongmans syndrome, 618846	INTELLECTUAL DISABILITY MENDELIOME
KDM4B	100%	100%	99%	98%	Intellectual developmental disorder, autosomal dominant 65, 619320	INTELLECTUAL DISABILITY MENDELIOME
KDM5B	97%	96%	100%	98%	Intellectual developmental disorder, autosomal recessive 65, 618109	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
KDM5C	100%	99%	97%	70%	Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type, 300534	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
KDM5D	48%	48%	47%	21%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
KDM6A	100%	99%	97%	70%	Kabuki syndrome 2, 300867	CRANIOFACIAL ANOMALIES PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME SCHISIS
KDM6B	100%	100%	100%	97%	Neurodevelopmental disorder with coarse facies and mild distal skeletal abnormalities, 618505	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
KDR	100%	100%	100%	98%	Hemangioma, capillary infantile, somatic, 602089	CONGENITAL HEART DISEASE HEART MENDELIOME
KDSR	100%	100%	99%	98%	Erythrokeratodermia variabilis et progressiva 4, 617526	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME

KERA	100%	100%	100%	97%	Cornea plana 2, autosomal recessive, 217300	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
KHDC3L	100%	100%	100%	99%	Hydatidiform mole, recurrent, 2, 614293	MENDELIOME PRE CONCEPTION SCREENING
KHK	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME
KIAA0556	100%	100%	100%	99%	Joubert syndrome 26, 616784	CILIOPATHIES RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
KIAA0586	95%	95%	100%	98%	Short-rib thoracic dysplasia 14 with polydactyly, 616546 Joubert syndrome 23, 616490	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS
KIAA0753	100%	100%	100%	98%	?Orofaciodigital syndrome XV, 617127 ?Joubert syndrome 38, 619476 Short-rib thoracic dysplasia 21 without polydactyly, 619479	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
KIAA0825	100%	100%	100%	98%	Polydactyly, postaxial, type A10, 618498	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
KIAA1109	100%	99%	100%	98%	Alkuraya-Kucinskas syndrome, 617822	FETAL AKINESIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
KIAA1549	99%	99%	100%	98%	Retinitis pigmentosa 86, 618613	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
KIDINS220	100%	100%	100%	98%	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296 Ventriculomegaly and arthrogryposis, 619501	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME
KIF11	100%	100%	100%	98%	Microcephaly w/wo chorioretinopathy, lymphedema, or mental retardation, 152950	VISION DISORDERS SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME

KIF12	100%	100%	100%	98%	Cholestasis, progressive familial intrahepatic, 8, 619662	LIVER DISORDERS MENDELIOME
KIF14	100%	100%	100%	98%	Microcephaly 20, primary, autosomal recessive, 617914 ?Meckel syndrome 12, 616258	CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
KIF1A	100%	100%	100%	99%	NESCAV syndrome, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal dominant, 610357 Spastic paraplegia 30, autosomal recessive, 610357	MOVEMENT DISORDERS NEUROPATHIES HNPD INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
KIF1B	100%	100%	100%	98%	Pheochromocytoma, 171300 Charcot-Marie-Tooth disease, type 2A1, 118210	NEUROPATHIES MENDELIOME TUMOR
KIF1C	100%	100%	100%	99%	Spastic ataxia 2, autosomal recessive, 611302	MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING
KIF20A	100%	100%	100%	99%	?Cardiomyopathy, familial restrictive, 6, 619433	MENDELIOME
KIF21A	100%	100%	100%	97%	Fibrosis of extraocular muscles, congenital, 3B, 135700 Fibrosis of extraocular muscles, congenital, 1, 135700	VISION DISORDERS MENDELIOME MUSCLE DISORDERS
KIF21B	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
KIF22	100%	100%	100%	98%	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
KIF23	100%	100%	100%	98%	Anemia, congenital dyserythropoietic, type IIIA, 105600	HEREDITARY BONE MARROW FAILURE IRON DISORDERS MENDELIOME
KIF24	100%	100%	100%	98%	No OMIM disease ID	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
KIF2A	100%	100%	100%	98%	Cortical dysplasia, complex, with other brain malformations 3, 615411	INTELLECTUAL DISABILITY MENDELIOME
KIF3B	100%	100%	100%	98%	Retinitis pigmentosa 89, 618955	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME
KIF4A	100%	100%	98%	72%	?Intellectual developmental disorder, X-linked 100, 300923	INTELLECTUAL DISABILITY MENDELIOME

KIF5A	100%	100%	100%	97%	Myoclonus, intractable, neonatal, 617235 Spastic paraplegia 10, autosomal dominant, 604187	ALS MOVEMENT DISORDERS EPILEPSY NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME
KIF5C	99%	99%	100%	98%	Cortical dysplasia, complex, with other brain malformations 2, 615282	FETAL AKINESIA INTELLECTUAL DISABILITY MENDELIOME
KIF7	100%	99%	100%	98%	Joubert syndrome 12, 200990 Acrocallosal syndrome, 200990 ?Hydrolethalus syndrome 2, 614120 ?Al-Gazali-Bakalinova syndrome, 607131	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS
KIFBP	95%	95%	100%	98%	Goldberg-Shprintzen megacolon syndrome, 609460	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS
KIRREL1	100%	100%	100%	99%	Nephrotic syndrome, type 23, 619201	RENAL DISORDERS MENDELIOME
KIRREL3	100%	100%	99%	97%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
KISS1	100%	100%	100%	95%	?Hypogonadotropic hypogonadism 13 w/wo anosmia, 614842	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME
KISS1R	100%	100%	100%	98%	Hypogonadotropic hypogonadism 8 w/wo anosmia, 614837 ?Precocious puberty, central, 1, 176400	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME PRE CONCEPTION SCREENING
KIT	100%	100%	100%	99%	Gastrointestinal stromal tumor, familial, 606764 Mastocytosis, cutaneous, 154800 Piebaldism, 172800 Germ cell tumors, somatic, 273300 Mastocytosis, systemic, somatic, 154800 Leukemia, acute myeloid, somatic, 601626	SKIN DISORDERS MENDELIOME TUMOR
KITLG	100%	99%	100%	98%	Hyperpigmentation w/wo hypopigmentation, 145250 Waardenburg syndrome, type 2F, 619947 Deafness, autosomal dominant 69, unilateral or asymmetric, 616697	SKIN DISORDERS HEARING IMPAIRMENT MENDELIOME

KIZ	100%	100%	100%	99%	Retinitis pigmentosa 69, 615780	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
KL	99%	99%	99%	96%	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994	SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
KLB	100%	100%	100%	99%	No OMIM disease ID	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME
KLC2	100%	100%	100%	99%	Spastic paraplegia, optic atrophy, and neuropathy, 609541	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING
KLF1	100%	100%	100%	98%	Blood group--Lutheran inhibitor, 111150 Dyserythropoietic anemia, congenital, type IV, 613673	HEREDITARY BONE MARROW FAILURE IRON DISORDERS MENDELIOME
KLF11	100%	100%	100%	98%	Maturity-onset diabetes of the young, type VII, 610508	MENDELIOME
KLF4	100%	100%	100%	97%	No OMIM disease ID	SKIN DISORDERS MENDELIOME
KLF6	100%	100%	100%	98%	Gastric cancer, somatic, 613659 Prostate cancer, somatic, 176807	MENDELIOME
KLF7	100%	100%	100%	98%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
KLHL10	100%	100%	100%	99%	Spermatogenic failure 11, 615081	MALE INFERTILITY MENDELIOME
KLHL15	100%	100%	97%	74%	Intellectual developmental disorder, X-linked 103, 300982	INTELLECTUAL DISABILITY MENDELIOME
KLHL20	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
KLHL24	100%	100%	100%	99%	Cardiomyopathy, familial hypertrophic, 29, with polyglucosan bodies, 620236 Epidermolysis bullosa simplex 6, generalized intermediate, w/wo cardiomyopathy, 617294	SKIN DISORDERS HEART MENDELIOME
KLHL3	100%	100%	100%	99%	Pseudohypoaldosteronism, type IID, 614495	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
KLHL40	100%	100%	100%	99%	Nemaline myopathy 8, autosomal recessive, 615348	FETAL AKINESIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS

KLHL41	100%	100%	100%	96%	Nemaline myopathy 9, 615731	FETAL AKINESIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
KLHL7	100%	100%	100%	98%	Retinitis pigmentosa 42, 612943 PERCHING syndrome, 617055	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
KLHL9	100%	100%	100%	98%	No OMIM disease ID	MENDELIOME MUSCLE DISORDERS
KLK4	100%	100%	100%	98%	Amelogenesis imperfecta, type IIA1, 204700	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
KLKB1	100%	100%	100%	98%	Fletcher factor (prekallikrein) deficiency, 612423	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
KLLN	100%	100%	100%	95%	Cowden syndrome 4, 615107	SKIN DISORDERS MENDELIOME
KMT2A	100%	100%	100%	97%	Wiedemann-Steiner syndrome, 605130	CONGENITAL HEART DISEASE HEART PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME
KMT2B	99%	99%	99%	95%	Intellectual developmental disorder, autosomal dominant 68, 619934 Dystonia 28, childhood-onset, 617284	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
KMT2C	100%	100%	99%	98%	Kleefstra syndrome 2, 617768	INTELLECTUAL DISABILITY MENDELIOME
KMT2D	100%	100%	100%	98%	Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186 Kabuki syndrome 1, 147920	CRANIOFACIAL ANOMALIES CONGENITAL HEART DISEASE SKIN DISORDERS HEART PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME SCHISIS

KMT2E	100%	99%	100%	98%	O'Donnell-Luria-Rodan syndrome, 618512	INTELLECTUAL DISABILITY MENDELIOME
KMT5B	100%	100%	100%	97%	Intellectual developmental disorder, autosomal dominant 51, 617788	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
KNG1	100%	100%	100%	98%	Angioedema, hereditary, 6, 619363	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME
KNL1	98%	98%	100%	98%	Microcephaly 4, primary, autosomal recessive, 604321	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
KNSTRN	100%	100%	100%	98%	?Roifman-Chitayat syndrome, digenic, 613328	MENDELIOME
KPTN	100%	100%	100%	97%	Intellectual developmental disorder, autosomal recessive 41, 615637	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
KRAS	100%	100%	100%	99%	Gastric cancer, somatic, 613659 Oculoectodermal syndrome, somatic, 600268 Breast cancer, somatic, 114480 Noonan syndrome 3, 609942 RAS-associated autoimmune leukoproliferative disorder, 614470 Arteriovenous malformation of the brain, somatic, 108010 Lung cancer, somatic, 211980 Pancreatic carcinoma, somatic, 260350 Leukemia, acute myeloid, somatic, 601626 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Cardiofaciocutaneous syndrome 2, 615278 Bladder cancer, somatic, 109800	HEREDITARY BONE MARROW FAILURE CONGENITAL HEART DISEASE SKIN DISORDERS HEART HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME AND RASOPATHY TUMOR
KREMEN1	100%	100%	100%	97%	Ectodermal dysplasia 13, hair/tooth type, 617392	CRANIOFACIAL ANOMALIES MENDELIOME
KRIT1	100%	100%	100%	98%	Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations, 116860 Cerebral cavernous malformations-1, 116860 Cavernous malformations of CNS and retina, 116860	EPILEPSY MENDELIOME
KRT1	100%	100%	100%	98%	Ichthyosis, annular epidermolytic 2, 620148 Epidermolytic hyperkeratosis, 113800 Palmoplantar keratoderma, nonepidermolytic, 600962 Keratosis palmoplantaris striata III, 607654 Palmoplantar keratoderma, epidermolytic, 144200 Ichthyosis histrix, Curth-Macklin type, 146590	SKIN DISORDERS MENDELIOME

KRT10	100%	100%	99%	91%	Ichthyosis, annular epidermolytic 1, 607602 Epidermolytic hyperkeratosis, 113800 ?Ichthyosis histrix, Lambert type, 146600 Ichthyosis with confetti, 609165	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
KRT12	100%	100%	100%	99%	Meesmann corneal dystrophy 1, 122100	VISION DISORDERS MENDELIOME
KRT13	100%	100%	100%	99%	White sponge nevus 2, 615785	SKIN DISORDERS MENDELIOME
KRT14	100%	100%	100%	99%	Epidermolysis bullosa simplex 1D, generalized, intermediate or severe, autosomal recessive, 601001 Epidermolysis bullosa simplex 1C, localized, 131800 Dermatopathia pigmentosa reticularis, 125595 Epidermolysis bullosa simplex 1A, generalized severe, 131760 Naegeli-Franceschetti-Jadassohn syndrome, 161000 Epidermolysis bullosa simplex 1B, generalized intermediate, 131900	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
KRT16	100%	100%	100%	99%	Palmoplantar keratoderma, nonepidermolytic, focal, 613000 Pachyonychia congenita 1, 167200	SKIN DISORDERS MENDELIOME
KRT17	100%	100%	100%	99%	Steatocystoma multiplex, 184500 Pachyonychia congenita 2, 167210	SKIN DISORDERS MENDELIOME
KRT18	100%	100%	100%	97%	Cirrhosis, cryptogenic, 215600	MENDELIOME PRE CONCEPTION SCREENING
KRT2	100%	100%	100%	99%	Ichthyosis bullosa of Siemens, 146800	SKIN DISORDERS MENDELIOME
KRT25	100%	100%	100%	99%	Woolly hair, autosomal recessive 3, 616760	MENDELIOME
KRT3	100%	100%	100%	98%	Meesmann corneal dystrophy 2, 618767	VISION DISORDERS MENDELIOME
KRT4	100%	100%	100%	98%	White sponge nevus 1, 193900	SKIN DISORDERS MENDELIOME
KRT5	100%	100%	100%	98%	Epidermolysis bullosa simplex 2A, generalized severe, 619555 Dowling-Degos disease 1, 179850 Epidermolysis bullosa simplex 2F, with mottled pigmentation, 131960 Epidermolysis bullosa simplex 2D, generalized, intermediate or severe, autosomal recessive, 619599 Epidermolysis bullosa simplex 2B, generalized intermediate, 619588 Epidermolysis bullosa simplex 2C, localized, 619594 Epidermolysis bullosa simplex 2E, with migratory circinate erythema, 609352	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
KRT6A	100%	100%	100%	98%	Pachyonychia congenita 3, 615726	SKIN DISORDERS MENDELIOME
KRT6B	100%	100%	100%	99%	Pachyonychia congenita 4, 615728	SKIN DISORDERS MENDELIOME
KRT6C	99%	99%	98%	92%	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735	SKIN DISORDERS MENDELIOME

KRT71	100%	100%	100%	99%	?Hypotrichosis 13, 615896	SKIN DISORDERS MENDELIOME
KRT74	100%	100%	100%	99%	Woolly hair, autosomal dominant, 194300 ?Hypotrichosis 3, 613981 ?Ectodermal dysplasia 7, hair/nail type, 614929	SKIN DISORDERS MENDELIOME
KRT75	100%	100%	100%	98%	No OMIM disease ID	SKIN DISORDERS MENDELIOME
KRT8	100%	100%	100%	98%	No OMIM disease ID	MENDELIOME PRE CONCEPTION SCREENING
KRT81	100%	100%	100%	98%	Monilethrix, 158000	SKIN DISORDERS MENDELIOME
KRT82	100%	100%	100%	99%	No OMIM disease ID	SKIN DISORDERS MENDELIOME
KRT83	100%	100%	100%	99%	Monilethrix, 158000 Erythrokeratodermia variabilis et progressiva 5, 617756	SKIN DISORDERS MENDELIOME
KRT85	100%	100%	100%	99%	Ectodermal dysplasia 4, hair/nail type, 602032	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
KRT86	100%	100%	100%	99%	Monilethrix, 158000	SKIN DISORDERS MENDELIOME
KRT9	100%	100%	100%	97%	Palmoplantar keratoderma, epidermolytic, 144200	SKIN DISORDERS MENDELIOME
KY	100%	100%	100%	99%	Myopathy, myofibrillar, 7, 617114	MENDELIOME PRE CONCEPTION SCREENING
KYNU	100%	100%	100%	98%	?Hydroxykynureninuria, 236800 Vertebral, cardiac, renal, and limb defects syndrome 2, 617661	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
L1CAM	100%	99%	98%	74%	MASA syndrome, 303350 Hydrocephalus, congenital, X-linked, 307000 ?Corpus callosum, partial agenesis of, 304100	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME
L2HGDH	100%	100%	100%	98%	L-2-hydroxyglutaric aciduria, 236792	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
LACC1	100%	100%	100%	97%	Juvenile arthritis, 618795	PRIMARY IMMUNODEFICIENCY MENDELIOME
LACTB	100%	100%	100%	97%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
LAGE3	100%	100%	96%	70%	Galloway-Mowat syndrome 2, X-linked, 301006	RENAL DISORDERS MENDELIOME

LAMA1	100%	100%	100%	99%	Poretti-Boltshauser syndrome, 615960	MOVEMENT DISORDERS VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
LAMA2	99%	99%	100%	99%	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855	HEART NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
LAMA3	100%	100%	100%	98%	Epidermolysis bullosa, junctional 2A, intermediate, 619783 Epidermolysis bullosa, junctional 2C, laryngoonychocutaneous, 245660 Epidermolysis bullosa, junctional 2B, severe, 619784	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
LAMA4	100%	100%	100%	99%	Cardiomyopathy, dilated, 1JJ, 615235	HEART MENDELIOME
LAMA5	100%	100%	100%	99%	Nephrotic syndrome, type 26, 620049 ?Bent bone dysplasia syndrome 2, 620076	SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME
LAMB1	100%	100%	100%	98%	Lissencephaly 5, 615191	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
LAMB2	100%	100%	100%	99%	Nephrotic syndrome, type 5, w/wo ocular abnormalities, 614199 Pierson syndrome, 609049	VISION DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
LAMB3	100%	100%	100%	99%	Epidermolysis bullosa, junctional 1B, severe, 226700 Epidermolysis bullosa, junctional 1A, intermediate, 226650 Amelogenesis imperfecta, type IA, 104530	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
LAMC2	100%	100%	100%	99%	Epidermolysis bullosa, junctional 3B, severe, 619786 Epidermolysis bullosa, junctional 3A, intermediate, 619785	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
LAMC3	100%	100%	100%	99%	Cortical malformations, occipital, 614115	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

LAMP2	100%	100%	98%	72%	Danon disease, 300257	VISION DISORDERS HEART HYPERTROPHIC CARDIOMYOPATHY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MUSCLE DISORDERS
LAMTOR2	100%	100%	100%	99%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
LAPTM5	100%	100%	100%	99%	No OMIM disease ID	HEREDITARY BONE MARROW FAILURE MENDELIOME
LARGE1	100%	100%	100%	99%	Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 6, 608840 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
LARP7	100%	100%	100%	97%	Alazami syndrome, 615071	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
LARS1	100%	100%	100%	98%	?Infantile liver failure syndrome 1, 615438	LIVER DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
LARS2	100%	100%	100%	99%	Perrault syndrome 4, 615300 Hydrops, lactic acidosis, and sideroblastic anemia, 617021	HEARING IMPAIRMENT DISORDERS OF SEX DEVELOPMENT IRON DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY
LAS1L	100%	99%	98%	72%	Wilson-Turner syndrome, 309585	INTELLECTUAL DISABILITY MENDELIOME
LAT	100%	100%	100%	98%	Immunodeficiency 52, 617514	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)

LBR	100%	100%	100%	97%	Pelger-Huet anomaly, 169400 ?Reynolds syndrome, 613471 Rhizomelic skeletal dysplasia w/wo Pelger-Huet anomaly, 618019 Greenberg skeletal dysplasia, 215140	CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
LBX1	100%	100%	100%	94%	?Central hypoventilation syndrome, congenital, 3, 619483	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
LCA5	100%	100%	100%	97%	Leber congenital amaurosis 5, 604537	VISION DISORDERS CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING
LCAT	100%	100%	100%	98%	Fish-eye disease, 136120 Norum disease, 245900	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
LCK	100%	100%	100%	99%	?Immunodeficiency 22, 615758	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)
LCP2	100%	100%	100%	98%	?Immunodeficiency 81, 619374	PRIMARY IMMUNODEFICIENCY MENDELIOME SEVERE COMBINED IMMUNODEFICIENCY (SCID)
LCT	100%	100%	100%	98%	Lactase deficiency, congenital, 223000	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
LDB3	100%	100%	100%	98%	Left ventricular noncompaction 3, 601493 Cardiomyopathy, hypertrophic, 24, 601493 Myopathy, myofibrillar, 4, 609452 Cardiomyopathy, dilated, 1C, w/wo LVNC, 601493	HEART MENDELIOME MUSCLE DISORDERS
LDHA	100%	100%	100%	98%	Glycogen storage disease XI, 612933	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
LDHB	100%	100%	100%	98%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME
LDHD	100%	100%	100%	99%	D-lactic aciduria with susceptibility to gout, 245450	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING

LDLR	100%	100%	100%	98%	LDL cholesterol level QTL2, 143890 Hypercholesterolemia, familial, 1, 143890	MENDELIOME
LDLRAP1	100%	100%	100%	99%	Hypercholesterolemia, familial, 4, 603813	SKIN DISORDERS MENDELIOME
LEF1	100%	100%	100%	97%	Sebaceous tumors, somatic,	MENDELIOME
LEFTY2	100%	100%	100%	99%	No OMIM disease ID	CONGENITAL HEART DISEASE HEART MENDELIOME
LEMD2	100%	100%	100%	96%	Marbach-Rustad progeroid syndrome, 619322 Cataract 46, juvenile-onset, 212500	VISION DISORDERS HEART MENDELIOME PRE CONCEPTION SCREENING
LEMD3	100%	100%	99%	94%	Buschke-Ollendorff syndrome, 166700 Osteopoikilosis w/wo melorheostosis, 166700	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
LEP	100%	100%	100%	99%	Obesity, morbid, due to leptin deficiency, 614962	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME PRE CONCEPTION SCREENING
LEPR	94%	94%	100%	98%	Obesity, morbid, due to leptin receptor deficiency, 614963	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME PRE CONCEPTION SCREENING
LETM1	100%	100%	100%	99%	Neurodegeneration, childhood-onset, with multisystem involvement due to mitochondrial dysfunction, 620089	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS
LFNG	99%	96%	99%	90%	Spondylocostal dysostosis 3, autosomal recessive, 609813	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
LGI1	100%	100%	100%	97%	Epilepsy, familial temporal lobe, 1, 600512	EPILEPSY MENDELIOME
LGI4	100%	100%	100%	99%	Arthrogryposis multiplex congenita 1, neurogenic, with myelin defect, 617468	FETAL AKINESIA MENDELIOME PRE CONCEPTION SCREENING
LGR4	100%	100%	99%	95%	Delayed puberty, self-limited, 619613	MENDELIOME
LHB	100%	100%	100%	99%	Hypogonadotropic hypogonadism 23 w/wo anosmia, 228300	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME PRE CONCEPTION SCREENING

LHCGR	100%	100%	100%	98%	Leydig cell adenoma, somatic, with precocious puberty, 176410 Leydig cell hypoplasia with pseudohermaphroditism, 238320 Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320 Luteinizing hormone resistance, female, 238320 Precocious puberty, male, 176410	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRE CONCEPTION SCREENING TUMOR
LHFPL5	100%	100%	100%	99%	Deafness, autosomal recessive 67, 610265	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
LHX1	100%	100%	100%	97%	No OMIM disease ID	DISORDERS OF SEX DEVELOPMENT MENDELIOME
LHX3	100%	100%	100%	97%	Pituitary hormone deficiency, combined, 3, 221750	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
LHX4	100%	100%	100%	98%	Pituitary hormone deficiency, combined, 4, 262700	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
LIAS	100%	100%	100%	99%	Hyperglycinemia, lactic acidosis, and seizures, 614462	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
LIFR	100%	100%	100%	97%	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559	HNPD SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
LIG1	100%	100%	100%	99%	Immunodeficiency 96, 619774	PRIMARY IMMUNODEFICIENCY MENDELIOME
LIG3	100%	100%	100%	99%	Mitochondrial DNA depletion syndrome 20 (MNGIE type), 619780	MENDELIOME MITOCHONDRIAL DISORDERS

LIG4	100%	100%	100%	98%	LIG4 syndrome, 606593	HEREDITARY BONE MARROW FAILURE DYSKERATOSIS CONGENITA PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID) TUMOR
LIM2	100%	100%	100%	99%	Cataract 19, multiple types, 615277	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
LIMS2	100%	100%	100%	99%	?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827	HEART MENDELIOME PRE CONCEPTION SCREENING
LINGO1	100%	100%	100%	99%	Intellectual developmental disorder, autosomal recessive 64, 618103	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
LINS1	100%	100%	100%	97%	Intellectual developmental disorder, autosomal recessive 27, 614340	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
LIPA	96%	95%	100%	98%	Wolman disease, 278000 Cholesteryl ester storage disease, 278000	DISORDERS OF SEX DEVELOPMENT METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
LIPC	100%	100%	100%	99%	Hepatic lipase deficiency, 614025	METABOLIC DISORDERS MENDELIOME
LIPE	100%	100%	100%	99%	Lipodystrophy, familial partial, type 6, 615980	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
LIPH	100%	100%	100%	98%	Hypotrichosis 7, 604379 Woolly hair, autosomal recessive 2 w/wo hypotrichosis, 604379	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
LIPN	100%	100%	100%	98%	Ichthyosis, congenital, autosomal recessive 8, 613943	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
LIPT1	100%	100%	100%	96%	Lipoyltransferase 1 deficiency, 616299	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING

LIPT2	100%	100%	100%	98%	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668	EPILEPSY METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
LITAF	100%	100%	100%	99%	Charcot-Marie-Tooth disease, type 1C, 601098	NEUROPATHIES MENDELIOME
LMAN1	100%	100%	100%	98%	Combined factor V and VIII deficiency, 227300	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
LMAN2L	100%	100%	100%	98%	?Intellectual developmental disorder, autosomal dominant 69, 617863 ?Intellectual developmental disorder, autosomal recessive 52, 616887	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
LMBR1	99%	99%	100%	97%	Triphalangeal thumb, type I, 174500 Syndactyly, type IV, 186200 Laurin-Sandrow syndrome, 135750 Hypoplastic or aplastic tibia with polydactyly, 188740 Polydactyly, preaxial type II, 174500 Acheiropody, 200500 Triphalangeal thumb-polysyndactyly syndrome, 190605	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
LMBRD1	100%	99%	100%	96%	Methylmalonic aciduria and homocystinuria, cblF type, 277380	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
LMBRD2	100%	100%	100%	98%	Developmental delay with variable neurologic and brain abnormalities, 619694	INTELLECTUAL DISABILITY MENDELIOME
LMCD1	100%	100%	99%	98%	No OMIM disease ID	HEART MENDELIOME
LMF1	100%	100%	100%	99%	Lipase deficiency, combined, 246650	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING

LMNA	100%	100%	100%	99%	Mandibuloacral dysplasia, 248370 Heart-hand syndrome, Slovenian type, 610140 Cardiomyopathy, dilated, 1A, 115200 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 Restrictive dermopathy 2, 619793 Charcot-Marie-Tooth disease, type 2B1, 605588 Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 Hutchinson-Gilford progeria, 176670 Lipodystrophy, familial partial, type 2, 151660 Muscular dystrophy, congenital, 613205 Malouf syndrome, 212112	FETAL AKINESIA DILATED CARDIOMYOPATHY SKIN DISORDERS HEART NEUROPATHIES SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS MUSCLE DISORDERS
LMNB1	100%	100%	100%	98%	Leukodystrophy, adult-onset, autosomal dominant, 169500 Microcephaly 26, primary, autosomal dominant, 619179	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME
LMNB2	100%	99%	100%	97%	Microcephaly 27, primary, autosomal dominant, 619180 ?Epilepsy, progressive myoclonic, 9, 616540	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
LMOD1	100%	100%	100%	97%	?Megacystis-microcolon-intestinal hypoperistalsis syndrome 3, 619362	ANEURYSM LIVER DISORDERS MENDELIOME
LMOD2	100%	100%	99%	95%	Cardiomyopathy, dilated, 2G, 619897	HEART MENDELIOME
LMOD3	100%	100%	100%	97%	Nemaline myopathy 10, 616165	FETAL AKINESIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
LMX1A	100%	100%	100%	99%	Deafness, autosomal dominant 7, 601412	HEARING IMPAIRMENT MENDELIOME
LMX1B	100%	100%	99%	94%	Focal segmental glomerulosclerosis 10, 256020 Nail-patella syndrome, 161200	VISION DISORDERS SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME
LNPK	93%	93%	100%	97%	Neurodevelopmental disorder with epilepsy and hypoplasia of the corpus callosum, 618090	MENDELIOME PRE CONCEPTION SCREENING

LONP1	100%	100%	100%	99%	CODAS syndrome, 600373	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
LORICRIN	100%	100%	99%	82%	Vohwinkel syndrome with ichthyosis, 604117	SKIN DISORDERS MENDELIOME
LOX	100%	100%	100%	97%	Aortic aneurysm, familial thoracic 10, 617168	ANEURYSM MENDELIOME
LOXHD1	100%	100%	100%	98%	Deafness, autosomal recessive 77, 613079	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
LOXL3	100%	100%	100%	99%	Myopia 28, autosomal recessive, 619781	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME
LPAR6	100%	99%	99%	94%	Hypotrichosis 8, 278150 Woolly hair, autosomal recessive 1, w/wo hypotrichosis, 278150	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
LPIN1	100%	100%	100%	98%	Myoglobinuria, acute recurrent, autosomal recessive, 268200	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
LPIN2	100%	100%	100%	98%	Majeed syndrome, 609628	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS IRON DISORDERS PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
LPL	100%	100%	100%	98%	Lipoprotein lipase deficiency, 238600 Combined hyperlipidemia, familial, 144250	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
LPP	100%	99%	100%	99%	Leukemia, acute myeloid, 601626 Lipoma,	MENDELIOME

LRAT	100%	100%	100%	98%	Leber congenital amaurosis 14, 613341 Retinal dystrophy, early-onset severe, 613341 Retinitis pigmentosa, juvenile, 613341	VISION DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
LRBA	100%	99%	100%	98%	Immunodeficiency, common variable, 8, with autoimmunity, 614700	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
LRIF1	100%	100%	100%	98%	?Facioscapulohumeral muscular dystrophy 3, digenic, 619477	MENDELIOME
LRIG2	100%	100%	100%	98%	Urofacial syndrome 2, 615112	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
LRIG3	100%	100%	100%	98%	No OMIM disease ID	NEUROPATHIES MENDELIOME
LRIT3	100%	100%	100%	97%	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
LRMDA	97%	97%	100%	99%	Albinism, oculocutaneous, type VII, 615179	VISION DISORDERS SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
LRP1	100%	100%	100%	99%	?Keratosis pilaris atrophicans, 604093	MENDELIOME PRE CONCEPTION SCREENING
LRP12	100%	100%	100%	98%	Oculopharyngodistal myopathy 1, 164310	MENDELIOME
LRP2	100%	100%	100%	99%	Donnai-Barrow syndrome, 222448	VISION DISORDERS CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
LRP4	100%	100%	100%	99%	?Myasthenic syndrome, congenital, 17, 616304 Sclerosteosis 2, 614305 Cenani-Lenz syndactyly syndrome, 212780	SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING

LRP5	100%	100%	99%	98%	Osteopetrosis, autosomal dominant 1, 607634 Hyperostosis, endosteal, 144750 Osteosclerosis, 144750 Polycystic liver disease 4 w/wo kidney cysts, 617875 Osteoporosis-pseudoglioma syndrome, 259770 Exudative vitreoretinopathy 4, 601813 van Buchem disease, type 2, 607636	VISION DISORDERS HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
LRP6	100%	100%	100%	99%	Tooth agenesis, selective, 7, 616724	CRANIOFACIAL ANOMALIES MENDELIOME
LRPAP1	100%	100%	100%	99%	Myopia 23, autosomal recessive, 615431	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
LRPPRC	100%	100%	100%	98%	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
LRRC10	100%	100%	100%	99%	No OMIM disease ID	HEART MENDELIOME
LRRC32	100%	100%	100%	99%	Cleft palate, proliferative retinopathy, and developmental delay, 619074	PRIMARY IMMUNODEFICIENCY MENDELIOME
LRRC56	100%	100%	100%	99%	Ciliary dyskinesia, primary, 39, 618254	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING
LRRC6	100%	100%	100%	98%	Ciliary dyskinesia, primary, 19, 614935	CILIOPATHIES MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING
LRRC8A	100%	100%	100%	99%	?Agammaglobulinemia 5, 613506	PRIMARY IMMUNODEFICIENCY MENDELIOME
LRRK1	100%	100%	100%	99%	Osteosclerotic metaphyseal dysplasia, 615198	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
LRRK2	100%	100%	100%	97%	No OMIM disease ID	MENDELIOME PARKINSON
LRSAM1	100%	100%	100%	99%	Charcot-Marie-Tooth disease, axonal, type 2P, 614436	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING
LRTOMT	100%	100%	100%	98%	Deafness, autosomal recessive 63, 611451	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING

LSM11	100%	100%	100%	93%	?Aicardi-Goutieres syndrome 8, 619486	PRIMARY IMMUNODEFICIENCY MENDELIOME
LSS	100%	100%	100%	99%	Hypotrichosis 14, 618275 Cataract 44, 616509 Alopecia-intellectual disability syndrome 4, 618840	VISION DISORDERS SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
LTBP1	100%	100%	100%	97%	Cutis laxa, autosomal recessive, type IIE, 619451	ANEURYSM SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
LTBP2	100%	100%	100%	99%	Glaucoma 3, primary congenital, D, 613086 Microspherophakia and/or megalocornea, with ectopia lentis and w/wo secondary glaucoma, 251750 ?Weill-Marchesani syndrome 3, recessive, 614819	VISION DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
LTBP3	100%	100%	100%	96%	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809	ANEURYSM CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
LTBP4	100%	100%	100%	98%	Cutis laxa, autosomal recessive, type IC, 613177	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
LTC4S	100%	100%	100%	96%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
LTV1	100%	100%	100%	98%	Inflammatory poikiloderma with hair abnormalities and acral keratoses, 620199	SKIN DISORDERS MENDELIOME
LYRM4	68%	68%	100%	98%	?Combined oxidative phosphorylation deficiency 19, 615595	MENDELIOME MITOCHONDRIAL DISORDERS
LYRM7	100%	100%	100%	98%	Mitochondrial complex III deficiency, nuclear type 8, 615838	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING

LYST	100%	99%	100%	98%	Chediak-Higashi syndrome, 214500	VISION DISORDERS SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
LYZ	100%	100%	100%	99%	Amyloidosis, renal, 105200	SKIN DISORDERS RENAL DISORDERS MENDELIOME
LZTFL1	100%	100%	100%	97%	Bardet-Biedl syndrome 17, 615994	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
LZTR1	100%	100%	100%	99%	Noonan syndrome 2, 605275 Noonan syndrome 10, 616564	HEART HEMOSTATIC/THROMBOTIC DISORDERS HNPD SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING NOONAN SYNDROME AND RASOPATHY TUMOR
LZTS1	100%	100%	100%	99%	Esophageal squamous cell carcinoma, somatic, 133239	MENDELIOME
M1AP	100%	100%	100%	99%	Spermatogenic failure 48, 619108	MALE INFERTILITY MENDELIOME
MAATS1	100%	100%	100%	97%	Spermatogenic failure 51, 619177	MALE INFERTILITY MENDELIOME
MAB21L1	100%	100%	100%	90%	Cerebellar, ocular, craniofacial, and genital syndrome, 618479	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MAB21L2	100%	100%	100%	100%	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877	VISION DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

MACF1	100%	100%	99%	97%	Lissencephaly 9 with complex brainstem malformation, 618325	INTELLECTUAL DISABILITY MENDELIOME
MAD1L1	100%	100%	100%	99%	Prostate cancer, somatic, 176807 Mosaic variegated aneuploidy syndrome 7 with inflammation and tumor predisposition, 620189 Lymphoma, B-cell, somatic,	MENDELIOME
MAD2L2	100%	100%	100%	99%	?Fanconi anemia, complementation group V, 617243	HEREDITARY BONE MARROW FAILURE MENDELIOME TUMOR
MADD	100%	100%	100%	99%	Neurodevelopmental disorder with dysmorphic facies, impaired speech and hypotonia, 619005 DEEAH syndrome, 619004	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MAF	93%	89%	98%	72%	Cataract 21, multiple types, 610202 Ayme-Gripp syndrome, 601088	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME
MAFA	100%	99%	94%	45%	Insulinomatosis and diabetes mellitus, 147630	MENDELIOME
MAFB	100%	100%	100%	98%	Duane retraction syndrome 3, 617041 Multicentric carpotarsal osteolysis syndrome, 166300	VISION DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME
MAG	100%	100%	100%	98%	Spastic paraplegia 75, autosomal recessive, 616680	MOVEMENT DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MAGED2	100%	99%	97%	70%	Bartter syndrome, type 5, antenatal, transient, 300971	RENAL DISORDERS MENDELIOME
MAGEL2	100%	100%	100%	99%	Schaaf-Yang syndrome, 615547	FETAL AKINESIA INTELLECTUAL DISABILITY MENDELIOME
MAGI2	98%	97%	99%	91%	Nephrotic syndrome, type 15, 617609	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
MAGT1	97%	97%	97%	69%	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853 Congenital disorder of glycosylation, type Icc, 301031	PRIMARY IMMUNODEFICIENCY MENDELIOME
MAK	100%	100%	100%	98%	Retinitis pigmentosa 62, 614181	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING

MALT1	100%	100%	100%	98%	Immunodeficiency 12, 615468	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
MAML2	100%	100%	100%	98%	Mucoepidermoid salivary gland carcinoma,	MENDELIOME
MAMLD1	100%	99%	98%	73%	Hypospadias 2, X-linked, 300758	DISORDERS OF SEX DEVELOPMENT MENDELIOME
MAN1B1	100%	100%	100%	99%	Rafiq syndrome, 614202	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MAN2B1	100%	100%	100%	99%	Mannosidosis, alpha-, types I and II, 248500	HEARING IMPAIRMENT PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MAN2B2	100%	100%	100%	99%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS MENDELIOME
MAN2C1	100%	100%	100%	99%	Congenital disorder of deglycosylation 2, 619775	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME
MANBA	100%	100%	100%	98%	Mannosidosis, beta, 248510	PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MAOA	99%	98%	98%	73%	Brunner syndrome, 300615	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME
MAP11	100%	100%	100%	97%	?Microcephaly 25, primary, autosomal recessive, 618351	MENDELIOME PRE CONCEPTION SCREENING
MAP1B	100%	100%	100%	96%	?Deafness, autosomal dominant 83, 619808 Periventricular nodular heterotopia 9, 618918	INTELLECTUAL DISABILITY MENDELIOME
MAP1LC3B2	100%	100%	100%	98%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME

MAP2K1	100%	100%	100%	98%	Cardiofaciocutaneous syndrome 3, 615279 Melorheostosis, isolated, somatic mosaic, 155950	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME AND RASOPATHY TUMOR
MAP2K2	100%	100%	100%	98%	Cardiofaciocutaneous syndrome 4, 615280	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME AND RASOPATHY TUMOR
MAP3K1	100%	100%	99%	95%	46XY sex reversal 6, 613762	DISORDERS OF SEX DEVELOPMENT MENDELIOME
MAP3K14	100%	100%	100%	98%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
MAP3K20	100%	100%	100%	98%	Centronuclear myopathy 6 with fiber-type disproportion, 617760 Split-foot malformation with mesoaxial polydactyly, 616890	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
MAP3K7	100%	100%	100%	98%	Frontometaphyseal dysplasia 2, 617137 Cardiospondylocarpofacial syndrome, 157800	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS
MAP3K8	100%	100%	100%	98%	Lung cancer, somatic, 211980	MENDELIOME
MAP4K4	100%	100%	99%	97%	No OMIM disease ID	ANEURYSM MENDELIOME
MAPK1	100%	100%	100%	97%	Noonan syndrome 13, 619087	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME AND RASOPATHY
MAPK8	100%	100%	100%	98%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
MAPK8IP3	100%	100%	100%	99%	Neurodevelopmental disorder w/wo variable brain abnormalities, 618443	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME

MAPKAPK3	100%	100%	100%	99%	?Macular dystrophy, patterned, 3, 617111	VISION DISORDERS MENDELIOME
MAPKAPK5	100%	100%	100%	98%	Neurocardiofaciodigital syndrome, 619869	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
MAPKBP1	100%	100%	100%	99%	Nephronophthisis 20, 617271	CILIOPATHIES RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
MAPRE2	100%	100%	100%	99%	Symmetric circumferential skin creases, congenital, 2, 616734	INTELLECTUAL DISABILITY MENDELIOME SCHISIS
MAPT	100%	100%	100%	99%	Supranuclear palsy, progressive, 601104 Supranuclear palsy, progressive atypical, 260540 Dementia, frontotemporal, w/wo parkinsonism, 600274 Pick disease, 172700	ALS MENDELIOME MITOCHONDRIAL DISORDERS PARKINSON PRE CONCEPTION SCREENING
MARCHF6	100%	100%	100%	98%	Epilepsy, familial adult myoclonic, 3, 613608	MENDELIOME
MARK3	100%	100%	100%	99%	?Visual impairment and progressive phthisis bulbi, 618283	MENDELIOME
MARS1	100%	100%	100%	99%	Interstitial lung and liver disease, 615486 ?Trichothiodystrophy 9, nonphotosensitive, 619692 Charcot-Marie-Tooth disease, axonal, type 2U, 616280	NEUROPATHIES LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING
MARS2	100%	100%	100%	99%	?Combined oxidative phosphorylation deficiency 25, 616430 Spastic ataxia 3, autosomal recessive, 611390	MOVEMENT DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
MARVELD2	100%	100%	100%	98%	Deafness, autosomal recessive 49, 610153	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
MASP1	100%	100%	100%	99%	3MC syndrome 1, 257920	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS
MASP2	100%	100%	100%	99%	MASP2 deficiency, 613791	PRIMARY IMMUNODEFICIENCY MENDELIOME
MAST1	100%	100%	100%	98%	Mega-corpus-callosum syndrome with cerebellar hypoplasia and cortical malformations, 618273	INTELLECTUAL DISABILITY MENDELIOME

MAST3	100%	100%	100%	99%	Developmental and epileptic encephalopathy 108, 620115	EPILEPSY MENDELIOME
MAST4	100%	100%	100%	98%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
MASTL	100%	100%	100%	98%	No OMIM disease ID	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME
MAT1A	100%	100%	100%	99%	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MAT2A	100%	100%	100%	99%	No OMIM disease ID	ANEURYSM MENDELIOME
MATN3	100%	100%	100%	98%	Spondyloepimetaphyseal dysplasia, Borochowitz-Cormier-Daire type, 608728 Epiphyseal dysplasia, multiple, 5, 607078	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
MATR3	100%	100%	100%	97%	Amyotrophic lateral sclerosis 21, 606070	ALS MENDELIOME
MAX	100%	100%	100%	98%	No OMIM disease ID	 MENDELIOME TUMOR
MBD4	100%	100%	100%	98%	Tumor predisposition syndrome 2, 619975	HEREDITARY BONE MARROW FAILURE MENDELIOME TUMOR
MBD5	100%	100%	100%	98%	Intellectual developmental disorder, autosomal dominant 1, 156200	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
MBOAT7	100%	100%	100%	98%	Intellectual developmental disorder, autosomal recessive 57, 617188	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MBTPS1	100%	100%	100%	99%	?Spondyloepiphyseal dysplasia, Kondo-Fu type, 618392	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
MBTPS2	100%	100%	98%	72%	Keratosis follicularis spinulosa decalvans, X-linked, 308800 Osteogenesis imperfecta, type XIX, 301014 IFAP syndrome w/wo BRESHECK syndrome, 308205 ?Olmsted syndrome, X-linked, 300918	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS

MC2R	100%	100%	100%	99%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200	DISORDERS OF SEX DEVELOPMENT PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
MC4R	100%	100%	100%	99%	Obesity (BMIQ20), 618406	MENDELIOME
MCAT	100%	100%	100%	99%	No OMIM disease ID	VISION DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS
MCC	100%	100%	100%	99%	Colorectal cancer, somatic, 114500	MENDELIOME
MCCC1	100%	100%	100%	99%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MCCC2	100%	100%	100%	97%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MCEE	100%	100%	100%	98%	Methylmalonyl-CoA epimerase deficiency, 251120	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
MCFD2	100%	100%	100%	96%	Factor V and factor VIII, combined deficiency of, 613625	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
MCIDAS	100%	100%	100%	98%	Ciliary dyskinesia, primary, 42, 618695	CILIOPATHIES MENDELIOME
MCM10	100%	100%	100%	99%	Immunodeficiency 80 w/wo cardiomyopathy, 619313	PRIMARY IMMUNODEFICIENCY MENDELIOME
MCM2	100%	100%	100%	99%	?Deafness, autosomal dominant 70, 616968	HEARING IMPAIRMENT MENDELIOME
MCM3AP	100%	100%	100%	99%	Peripheral neuropathy, autosomal recessive, w/wo impaired intellectual development, 618124	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING
MCM4	95%	95%	100%	98%	Immunodeficiency 54, 609981	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING

MCM5	100%	100%	100%	98%	?Meier-Gorlin syndrome 8, 617564	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
MCM6	100%	100%	100%	98%	Lactase persistence/nonpersistence, 223100	MENDELIOME
MCM8	94%	94%	100%	98%	?Premature ovarian failure 10, 612885	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRIMARY OVARIAN INSUFFICIENCY TUMOR
MCM9	100%	100%	100%	98%	Ovarian dysgenesis 4, 616185	DISORDERS OF SEX DEVELOPMENT MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY TUMOR
MCOLN1	100%	100%	100%	99%	Mucolipidosis IV, 252650	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MCPH1	100%	100%	100%	98%	Microcephaly 1, primary, autosomal recessive, 251200	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MCTP2	100%	99%	100%	98%	No OMIM disease ID	CONGENITAL HEART DISEASE MENDELIOME
MCUR1	100%	100%	100%	95%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
MDFIC	100%	99%	100%	96%	Lymphatic malformation 12, 620014	SKIN DISORDERS MENDELIOME
MDH1	100%	100%	100%	99%	?Developmental and epileptic encephalopathy 88, 618959	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS
MDH2	100%	100%	100%	98%	Developmental and epileptic encephalopathy 51, 617339	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING TUMOR
MDM2	94%	94%	100%	97%	?Lessel-Kubisch syndrome, 618681	MENDELIOME
MDM4	100%	100%	100%	98%	?Bone marrow failure syndrome 6, 618849	HEREDITARY BONE MARROW FAILURE MENDELIOME

MECOM	100%	100%	100%	98%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
MECP2	100%	99%	98%	73%	Rett syndrome, atypical, 312750 Encephalopathy, neonatal severe, 300673 Intellectual developmental disorder, X-linked syndromic, Lubs type, 300260 Intellectual developmental disorder, X-linked syndromic 13, 300055 Rett syndrome, 312750 Rett syndrome, preserved speech variant, 312750	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
MECR	100%	100%	100%	99%	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
MED11	100%	100%	100%	97%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
MED12	100%	99%	97%	70%	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Hardikar syndrome, 301068 Opitz-Kaveggia syndrome, 305450	CRANIOFACIAL ANOMALIES SKIN DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
MED12L	100%	100%	100%	98%	Nizon-Isidor syndrome, 618872	INTELLECTUAL DISABILITY MENDELIOME
MED13	100%	100%	100%	98%	Intellectual developmental disorder, autosomal dominant 61, 618009	INTELLECTUAL DISABILITY MENDELIOME
MED13L	100%	99%	100%	98%	Impaired intellectual development and distinctive facial features w/wo cardiac defects, 616789	CONGENITAL HEART DISEASE HEART INTELLECTUAL DISABILITY MENDELIOME
MED17	100%	100%	100%	98%	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MED23	100%	100%	100%	98%	Intellectual developmental disorder, autosomal recessive 18, w/wo epilepsy, 614249	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MED25	100%	100%	100%	98%	Basel-Vanagait-Smirin-Yosef syndrome, 616449	NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS

MED27	100%	100%	100%	98%	Neurodevelopmental disorder with spasticity, cataracts, and cerebellar hypoplasia, 619286	INTELLECTUAL DISABILITY MENDELIOME
MEF2C	100%	100%	100%	99%	Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language, 613443	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
MEFV	96%	96%	100%	99%	Neutrophilic dermatosis, acute febrile, 608068 Familial Mediterranean fever, AR, 249100 Familial Mediterranean fever, AD, 134610	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
MEGF10	100%	100%	100%	99%	Congenital myopathy 10A, severe variant, 614399 Congenital myopathy 10B, mild variant, 614399	FETAL AKINESIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
MEGF8	100%	100%	99%	98%	Carpenter syndrome 2, 614976	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
mei-01	100%	100%	100%	99%	Hydatidiform mole, recurrent, 3, 618431	MALE INFERTILITY MENDELIOME
MEIOB	100%	100%	100%	97%	?Spermatogenic failure 22, 617706	MALE INFERTILITY MENDELIOME
MEIS2	100%	100%	100%	99%	Cleft palate, cardiac defects, and mental retardation, 600987	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME SCHISIS
MEN1	100%	100%	100%	98%	Multiple endocrine neoplasia 1, 131100 Lipoma, somatic, Angiofibroma, somatic, Carcinoid tumor of lung, Adrenal adenoma, somatic, Parathyroid adenoma, somatic,	ENDOCRINE TUMOR MENDELIOME TUMOR
MEOX1	100%	100%	100%	98%	Klippel-Feil syndrome 2, 214300	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
MERTK	98%	98%	100%	98%	Retinitis pigmentosa 38, 613862	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING

MESD	100%	100%	100%	96%	Osteogenesis imperfecta, type XX, 618644	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
MESP2	100%	99%	100%	98%	Spondylocostal dysostosis 2, autosomal recessive, 608681	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
MET	100%	100%	100%	98%	Renal cell carcinoma, papillary, 1, familial and somatic, 605074 ?Arthrogryposis, distal, type 11, 620019 Hepatocellular carcinoma, childhood type, somatic, 114550 ?Deafness, autosomal recessive 97, 616705	HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING TUMOR
METTL23	100%	100%	100%	99%	Intellectual developmental disorder, autosomal recessive 44, 615942	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
METTL5	100%	100%	100%	97%	Intellectual developmental disorder, autosomal recessive 72, 618665	INTELLECTUAL DISABILITY MENDELIOME
MFAP5	100%	100%	100%	98%	Aortic aneurysm, familial thoracic 9, 616166	ANEURYSM MENDELIOME
MFF	100%	100%	100%	98%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
MFN2	100%	100%	100%	98%	Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260 Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 Hereditary motor and sensory neuropathy VIA, 601152	VISION DISORDERS NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
MFRP	100%	100%	100%	99%	Microphthalmia, isolated 5, 611040 Nanophthalmos 2, 609549	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
MFSD2A	100%	100%	100%	98%	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities, 616486	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

MFSD8	100%	100%	100%	99%	Macular dystrophy with central cone involvement, 616170 Ceroid lipofuscinosis, neuronal, 7, 610951	MOVEMENT DISORDERS VISION DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MGAT2	100%	100%	100%	97%	Congenital disorder of glycosylation, type IIa, 212066	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MGME1	100%	100%	100%	96%	Mitochondrial DNA depletion syndrome 11, 615084	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
MGP	100%	100%	100%	97%	Keutel syndrome, 245150	SKIN DISORDERS HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MIA3	100%	100%	99%	97%	?Ondontochondrodysplasia 2 with hearing loss and diabetes, 619269	HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME
MIB1	100%	100%	100%	99%	Left ventricular noncompaction 7, 615092	HEART MENDELIOME
MICOS13	100%	100%	100%	99%	Combined oxidative phosphorylation deficiency 37, 618329	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
MICU1	100%	99%	100%	99%	Myopathy with extrapyramidal signs, 615673	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS
MICU2	100%	100%	99%	96%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
MID1	99%	99%	98%	72%	Opitz GBBB syndrome, 300000	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME SCHISIS

MID2	100%	99%	98%	73%	?Intellectual developmental disorder, X-linked 101, 300928	INTELLECTUAL DISABILITY MENDELIOME
MIEF2	100%	100%	100%	99%	?Combined oxidative phosphorylation deficiency 49, 619024	MENDELIOME MITOCHONDRIAL DISORDERS
MINAR2	100%	100%	100%	96%	Deafness, autosomal recessive 120, 620238	HEARING IMPAIRMENT MENDELIOME
MINPP1	100%	100%	100%	98%	Pontocerebellar hypoplasia, type 16, 619527	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME
MIP	100%	100%	100%	99%	Cataract 15, multiple types, 615274	VISION DISORDERS MENDELIOME
MIPEP	100%	100%	100%	98%	Combined oxidative phosphorylation deficiency 31, 617228	HEART MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
MIR140	NC	NC	NC	NC	Spondyloepiphyseal dysplasia, Nishimura type, 618618	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
MIR17HG	NC	NC	NC	NC	No OMIM disease ID	MENDELIOME
MIR184	NC	NC	NC	NC	EDICT syndrome, 614303	VISION DISORDERS MENDELIOME
MIR204	NC	NC	NC	NC	?Retinal dystrophy and iris coloboma w/wo cataract, 616722	VISION DISORDERS MENDELIOME
MIR96	NC	NC	NC	NC	Deafness, autosomal dominant 50, 613074	HEARING IMPAIRMENT MENDELIOME
MITF	99%	99%	100%	98%	Waardenburg syndrome, type 2A, 193510 Tietz albinism-deafness syndrome, 103500 Waardenburg syndrome/ocular albinism, digenic, 103470 COMMAD syndrome, 617306	VISION DISORDERS CRANIOFACIAL ANOMALIES SKIN DISORDERS HEARING IMPAIRMENT MELA1 MELA2 MENDELIOME PRE CONCEPTION SCREENING TUMOR
MKKS	100%	100%	100%	99%	McKusick-Kaufman syndrome, 236700 Bardet-Biedl syndrome 6, 605231	VISION DISORDERS CILIOPATHIES DISORDERS OF SEX DEVELOPMENT INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING

MKRN3	100%	100%	100%	99%	Precocious puberty, central, 2, 615346	DISORDERS OF SEX DEVELOPMENT MENDELIOME
MKS1	100%	100%	100%	98%	Bardet-Biedl syndrome 13, 615990 Meckel syndrome 1, 249000 Joubert syndrome 28, 617121	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS
MLC1	100%	100%	100%	99%	Megalencephalic leukoencephalopathy with subcortical cysts 1, 604004	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MLH1	100%	100%	100%	97%	Lynch syndrome 2, 609310 Muir-Torre syndrome, 158320 Mismatch repair cancer syndrome 1, 276300	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING TUMOR
MLH3	100%	100%	100%	98%	Colorectal cancer, somatic, 114500 Colorectal cancer, hereditary nonpolyposis, type 7, 614385	MALE INFERTILITY MENDELIOME
MLIP	100%	100%	100%	98%	Myopathy with myalgia, increased serum creatine kinase, and w/wo episodic rhabdomyolysis, 620138	MENDELIOME MUSCLE DISORDERS
MLLT10	97%	97%	100%	98%	Leukemia, acute myeloid, 601626	MENDELIOME
MLPH	100%	100%	100%	99%	Griscelli syndrome, type 3, 609227	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
MLYCD	100%	100%	100%	97%	Malonyl-CoA decarboxylase deficiency, 248360	HEART METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MMAA	100%	100%	100%	99%	Methylmalonic aciduria, vitamin B12-responsive, cblA type, 251100	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

MMAB	100%	100%	99%	98%	Methylmalonic aciduria, vitamin B12-responsive, cblB type, 251110	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MMACHC	100%	100%	100%	99%	Methylmalonic aciduria and homocystinuria, cblC type, 277400	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
MMADHC	89%	89%	100%	98%	Methylmalonic aciduria, cblD type, variant 2, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Homocystinuria, cblD type, variant 1, 277410	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MME	97%	97%	100%	97%	?Spinocerebellar ataxia 43, 617018 Charcot-Marie-Tooth disease, axonal, type 2T, 617017	NEUROPATHIES HNPD MENDELIOME PRE CONCEPTION SCREENING
MMGT1	100%	100%	97%	69%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
MMP1	100%	100%	100%	97%	COPD, rate of decline of lung function in, 606963	MENDELIOME
MMP13	92%	92%	100%	97%	?Spondyloepimetaphyseal dysplasia, Missouri type, 602111 Metaphyseal anadysplasia 1, 602111 Metaphyseal dysplasia, Spahr type, 250400	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
MMP14	100%	100%	100%	99%	?Winchester syndrome, 277950	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
MMP19	100%	100%	100%	99%	Cavitary optic disc anomalies, 611543	MENDELIOME
MMP2	100%	100%	100%	98%	Multicentric osteolysis, nodulosis, and arthropathy, 259600	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING

MMP20	100%	100%	100%	98%	Amelogenesis imperfecta, type IIA2, 612529	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
MMP21	100%	100%	100%	98%	Heterotaxy, visceral, 7, autosomal, 616749	CONGENITAL HEART DISEASE CILIOPATHIES HEART MENDELIOME PRE CONCEPTION SCREENING
MMP9	100%	100%	100%	98%	Metaphyseal anadysplasia 2, 613073	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
MMUT	100%	100%	100%	98%	Methylmalonic aciduria, mut(0) type, 251000	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MN1	100%	100%	100%	99%	CEBALID syndrome, 618774 Meningioma, 607174	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME
MNS1	100%	100%	100%	97%	Heterotaxy, visceral, 9, autosomal, with male infertility, 618948	CILIOPATHIES MALE INFERTILITY MENDELIOME
MNX1	97%	93%	97%	76%	Currarino syndrome, 176450	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
MOCOS	100%	100%	100%	98%	Xanthinuria, type II, 603592	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
MOCS1	100%	100%	100%	98%	Molybdenum cofactor deficiency A, 252150	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MOCS2	100%	100%	100%	98%	Molybdenum cofactor deficiency B, 252160	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MOG	100%	100%	100%	98%	?Narcolepsy 7, 614250	MENDELIOME

MOGS	100%	100%	100%	99%	Congenital disorder of glycosylation, type IIb, 606056	PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MORC2	100%	100%	100%	98%	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688 Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy, 619090	NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME
MPC1	100%	100%	100%	98%	Mitochondrial pyruvate carrier deficiency, 614741	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
MPC2	100%	100%	100%	96%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS
MPDU1	100%	100%	100%	97%	Congenital disorder of glycosylation, type If, 609180	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MPDZ	99%	99%	100%	98%	Hydrocephalus, congenital, 2, w/wo brain or eye anomalies, 615219	HEARING IMPAIRMENT EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MPEG1	100%	100%	100%	99%	Immunodeficiency 77, 619223	PRIMARY IMMUNODEFICIENCY MENDELIOME
MPI	100%	100%	100%	99%	Congenital disorder of glycosylation, type Ib, 602579	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
MPIG6B	100%	100%	100%	98%	?Thrombocytopenia, anemia, and myelofibrosis, 617441	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME
MPL	100%	100%	100%	98%	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS IRON DISORDERS MENDELIOME PRE CONCEPTION SCREENING TUMOR
MPLKIP	100%	100%	100%	97%	Trichothiodystrophy 4, nonphotosensitive, 234050	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

MPO	100%	100%	100%	99%	Myeloperoxidase deficiency, 254600	MENDELIOME PRE CONCEPTION SCREENING
MPP5	100%	100%	100%	98%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
MPV17	100%	100%	100%	99%	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810	NEUROPATHIES LIVER DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
MPZ	100%	100%	100%	98%	Charcot-Marie-Tooth disease, type 2I, 607677 Dejerine-Sottas disease, 145900 Charcot-Marie-Tooth disease, type 1B, 118200 Roussy-Levy syndrome, 180800 Charcot-Marie-Tooth disease, dominant intermediate D, 607791 Hypomyelinating neuropathy, congenital, 2, 618184 Charcot-Marie-Tooth disease, type 2J, 607736	FETAL AKINESIA NEUROPATHIES HNPD MENDELIOME PRE CONCEPTION SCREENING
MPZL2	100%	100%	100%	99%	Deafness, autosomal recessive 111, 618145	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
MRAP	100%	100%	100%	99%	Glucocorticoid deficiency 2, 607398	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRE CONCEPTION SCREENING
MRAS	100%	100%	100%	99%	Noonan syndrome 11, 618499	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME AND RASOPATHY
MRE11	100%	100%	100%	97%	Ataxia-telangiectasia-like disorder 1, 604391	MOVEMENT DISORDERS SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING TUMOR
MRM2	97%	97%	100%	98%	?Mitochondrial DNA depletion syndrome 17, 618567	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
MRPL12	100%	100%	100%	99%	?Combined oxidative phosphorylation deficiency 45, 618951	MENDELIOME MITOCHONDRIAL DISORDERS
MRPL24	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS

MRPL3	100%	100%	100%	98%	Combined oxidative phosphorylation deficiency 9, 614582	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
MRPL40	100%	100%	100%	98%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
MRPL44	100%	100%	100%	98%	Combined oxidative phosphorylation deficiency 16, 615395	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
MRPL57	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
MRPS14	100%	100%	100%	99%	?Combined oxidative phosphorylation deficiency 38, 618378	MENDELIOME MITOCHONDRIAL DISORDERS
MRPS16	100%	100%	100%	99%	Combined oxidative phosphorylation deficiency 2, 610498	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
MRPS2	100%	100%	100%	99%	Combined oxidative phosphorylation deficiency 36, 617950	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
MRPS22	100%	100%	100%	98%	Ovarian dysgenesis 7, 618117 Combined oxidative phosphorylation deficiency 5, 611719	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
MRPS23	100%	100%	100%	99%	?Combined oxidative phosphorylation deficiency 46, 618952	MENDELIOME MITOCHONDRIAL DISORDERS
MRPS25	83%	83%	100%	98%	?Combined oxidative phosphorylation deficiency 50, 619025	MENDELIOME MITOCHONDRIAL DISORDERS
MRPS28	85%	85%	99%	96%	?Combined oxidative phosphorylation deficiency 47, 618958	MENDELIOME MITOCHONDRIAL DISORDERS
MRPS34	100%	100%	100%	99%	Combined oxidative phosphorylation deficiency 32, 617664	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
MRPS36	100%	100%	100%	96%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS
MRPS7	100%	100%	100%	99%	?Combined oxidative phosphorylation deficiency 34, 617872	MENDELIOME MITOCHONDRIAL DISORDERS
MRRF	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
MRTFA	100%	100%	100%	98%	?Immunodeficiency 66, 618847	PRIMARY IMMUNODEFICIENCY MENDELIOME

MS4A1	100%	100%	100%	97%	?Immunodeficiency, common variable, 5, 613495	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
MSH2	100%	100%	100%	98%	Lynch syndrome 1, 120435 Muir-Torre syndrome, 158320 Mismatch repair cancer syndrome 2, 619096	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING TUMOR
MSH3	100%	100%	99%	94%	Familial adenomatous polyposis 4, 617100 Endometrial carcinoma, somatic, 608089	MENDELIOME PRE CONCEPTION SCREENING TUMOR
MSH4	100%	100%	100%	98%	Premature ovarian failure 20, 619938 Spermatogenic failure 2, 108420	DISORDERS OF SEX DEVELOPMENT MALE INFERTILITY MENDELIOME PRIMARY OVARIAN INSUFFICIENCY
MSH5	100%	100%	100%	98%	?Premature ovarian failure 13, 617442 Spermatogenic failure 74, 619937	MALE INFERTILITY MENDELIOME
MSH6	100%	100%	100%	98%	Lynch syndrome 5, 614350 Mismatch repair cancer syndrome 3, 619097	HEREDITARY BONE MARROW FAILURE MENDELIOME PRE CONCEPTION SCREENING TUMOR
MSL2	100%	100%	100%	98%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
MSL3	100%	100%	97%	66%	Basilicata-Akhtar syndrome, 301032	INTELLECTUAL DISABILITY MENDELIOME
MSMO1	100%	100%	100%	99%	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MSN	100%	98%	98%	73%	Immunodeficiency 50, 300988	PRIMARY IMMUNODEFICIENCY MENDELIOME
MSR1	100%	100%	100%	97%	Barrett esophagus/esophageal adenocarcinoma, 614266	MENDELIOME
MSRB3	100%	100%	99%	96%	Deafness, autosomal recessive 74, 613718	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
MSTN	100%	100%	100%	98%	?Muscle hypertrophy, 614160	MENDELIOME MUSCLE DISORDERS
MSTO1	100%	100%	100%	98%	Myopathy, mitochondrial, and ataxia, 617675	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS

MSX1	100%	100%	99%	96%	Tooth agenesis, selective, 1, w/wo orofacial cleft, 106600 Ectodermal dysplasia 3, Witkop type, 189500 Orofacial cleft 5, 608874	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME SCHISIS
MSX2	100%	100%	100%	97%	Parietal foramina with cleidocranial dysplasia, 168550 Craniosynostosis 2, 604757 Parietal foramina 1, 168500	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
MTAP	100%	100%	100%	97%	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME TUMOR
MTFMT	100%	100%	100%	97%	Combined oxidative phosphorylation deficiency 15, 614947 Mitochondrial complex I deficiency, nuclear type 27, 618248	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
MTHFD1	100%	100%	100%	99%	Combined immunodeficiency and megaloblastic anemia w/wo hyperhomocysteinemia, 617780	PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
MTHFR	100%	100%	100%	98%	Homocystinuria due to MTHFR deficiency, 236250	MOVEMENT DISORDERS EPILEPSY HEMOSTATIC/THROMBOTIC DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MTHFS	100%	100%	100%	96%	Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MTM1	99%	99%	97%	72%	Myopathy, centronuclear, X-linked, 310400	FETAL AKINESIA LIVER DISORDERS METABOLIC DISORDERS MENDELIOME MUSCLE DISORDERS
MTMR2	100%	100%	99%	98%	Charcot-Marie-Tooth disease, type 4B1, 601382	NEUROPATHIES METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING

MTO1	93%	91%	100%	98%	Combined oxidative phosphorylation deficiency 10, 614702	HEART INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
MTOR	100%	100%	100%	99%	Focal cortical dysplasia, type II, somatic, 607341 Smith-Kingsmore syndrome, 616638	SKIN DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
MTPAP	100%	100%	100%	98%	?Spastic ataxia 4, autosomal recessive, 613672	MOVEMENT DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
MTR	100%	100%	100%	98%	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MTRR	100%	100%	100%	98%	Homocystinuria-megaloblastic anemia, cbl E type, 236270	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MTSS2	100%	100%	100%	98%	Intellectual developmental disorder with ocular anomalies and distinctive facial features, 620086	INTELLECTUAL DISABILITY MENDELIOME
MTTP	100%	100%	99%	98%	Abetalipoproteinemia, 200100	MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING
MTX2	100%	99%	100%	98%	Mandibuloacral dysplasia progeroid syndrome, 619127	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME MITOCHONDRIAL DISORDERS
MUC1	100%	100%	80%	66%	Tubulointerstitial kidney disease, autosomal dominant, 2, 174000	MENDELIOME
MUC16	100%	100%	100%	99%	No OMIM disease ID	CONGENITAL HEART DISEASE HEART MENDELIOME
MUSK	100%	100%	100%	99%	Fetal akinesia deformation sequence 1, 208150 Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325	FETAL AKINESIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS

MUTYH	100%	100%	100%	99%	Adenomas, multiple colorectal, 608456 Gastric cancer, somatic, 613659	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING TUMOR
MVD	100%	100%	100%	99%	Porokeratosis 7, multiple types, 614714	SKIN DISORDERS HEARING IMPAIRMENT MENDELIOME
MVK	90%	90%	100%	99%	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377	VISION DISORDERS HEREDITARY BONE MARROW FAILURE SKIN DISORDERS PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MXI1	100%	100%	99%	93%	Prostate cancer, somatic, 176807 Neurofibrosarcoma, somatic,	MENDELIOME
MYBPC1	100%	100%	100%	98%	Congenital myopathy 16, 618524 Lethal congenital contracture syndrome 4, 614915 Arthrogryposis, distal, type 1B, 614335	FETAL AKINESIA MENDELIOME PRE CONCEPTION SCREENING
MYBPC3	100%	100%	100%	99%	Cardiomyopathy, hypertrophic, 4, 115197 Cardiomyopathy, dilated, 1MM, 615396 Left ventricular noncompaction 10, 615396	HEART HYPERTROPHIC CARDIOMYOPATHY MENDELIOME
MYBPHL	100%	100%	100%	99%	No OMIM disease ID	HEART MENDELIOME
MYC	100%	100%	100%	97%	Burkitt lymphoma, somatic, 113970	MENDELIOME
MYCN	100%	100%	99%	94%	Feingold syndrome 1, 164280	FETAL AKINESIA SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
MYD88	100%	100%	100%	99%	Macroglobulinemia, Waldenstrom, somatic, 153600 Immunodeficiency 68, 612260	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
MYF5	100%	100%	100%	98%	Ophthalmoplegia, external, with rib and vertebral anomalies, 618155	MENDELIOME PRE CONCEPTION SCREENING
MYH11	100%	100%	100%	98%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 2, 619351 Aortic aneurysm, familial thoracic 4, 132900 Visceral myopathy 2, 619350	ANEURYSM CONGENITAL HEART DISEASE HEART LIVER DISORDERS MENDELIOME

MYH14	100%	100%	100%	98%	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369 Deafness, autosomal dominant 4A, 600652	HEARING IMPAIRMENT NEUROPATHIES MENDELIOME
MYH2	100%	100%	100%	98%	Congenital myopathy 6 with ophthalmoplegia, 605637	MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
MYH3	100%	100%	99%	97%	Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A, 178110 Contractures, pterygia, and spondylocarpotarsal fusion syndrome 1B, 618469 Arthrogryposis, distal, type 2B3 (Sheldon-Hall), 618436 Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700	FETAL AKINESIA SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
MYH6	100%	100%	100%	97%	Atrial septal defect 3, 614089 Cardiomyopathy, dilated, 1EE, 613252 Cardiomyopathy, hypertrophic, 14, 613251	CONGENITAL HEART DISEASE HEART MENDELIOME
MYH7	100%	100%	100%	99%	Laing distal myopathy, 160500 Cardiomyopathy, hypertrophic, 1, 192600 Left ventricular noncompaction 5, 613426 Cardiomyopathy, dilated, 1S, 613426 Congenital myopathy 7B, myosin storage, autosomal recessive, 255160 Congenital myopathy 7A, myosin storage, autosomal dominant, 608358	CONGENITAL HEART DISEASE DILATED CARDIOMYOPATHY HEART HYPERTROPHIC CARDIOMYOPATHY MENDELIOME MUSCLE DISORDERS
MYH7B	100%	100%	100%	99%	No OMIM disease ID	HEART MENDELIOME
MYH8	100%	100%	100%	98%	Carney complex variant, 608837 Trismus-pseudocamptodactyly syndrome, 158300	FETAL AKINESIA SKIN DISORDERS MENDELIOME
MYH9	100%	100%	100%	98%	Macrothrombocytopenia and granulocyte inclusions w/wo nephritis or sensorineural hearing loss, 155100 Deafness, autosomal dominant 17, 603622	HEREDITARY BONE MARROW FAILURE HEARING IMPAIRMENT HEMOSTATIC/THROMBOTIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME
MYL1	100%	100%	100%	98%	Congenital myopathy 14, 618414	MENDELIOME PRE CONCEPTION SCREENING
MYL2	100%	100%	100%	98%	Cardiomyopathy, hypertrophic, 10, 608758 Myopathy, myofibrillar, 12, infantile-onset, with cardiomyopathy, 619424	HEART HYPERTROPHIC CARDIOMYOPATHY MENDELIOME PRE CONCEPTION SCREENING
MYL3	100%	100%	100%	99%	Cardiomyopathy, hypertrophic, 8, 608751	HEART HYPERTROPHIC CARDIOMYOPATHY MENDELIOME PRE CONCEPTION SCREENING

MYL4	100%	100%	100%	99%	?Atrial fibrillation, familial, 18, 617280	HEART MENDELIOME
MYL7	100%	100%	100%	98%	No OMIM disease ID	HEART MENDELIOME
MYL9	100%	100%	100%	99%	?Megacystis-microcolon-intestinal hypoperistalsis syndrome 4, 619365	LIVER DISORDERS MENDELIOME
MYLK	100%	100%	100%	98%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 1, 249210 Aortic aneurysm, familial thoracic 7, 613780	ANEURYSM LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING
MYLK2	100%	100%	100%	98%	Cardiomyopathy, hypertrophic, 1, digenic, 192600	MENDELIOME
MYLK3	100%	100%	100%	99%	No OMIM disease ID	HEART MENDELIOME
MYLPF	100%	100%	100%	99%	Arthrogryposis, distal, type 1C, 619110	FETAL AKINESIA SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
MYMK	100%	100%	100%	98%	Carey-Fineman-Ziter syndrome, 254940	MENDELIOME PRE CONCEPTION SCREENING SCHISIS
MYO15A	100%	100%	100%	99%	Deafness, autosomal recessive 3, 600316	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
MYO18B	100%	100%	100%	98%	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
MYO1A	100%	100%	100%	98%	No OMIM disease ID	NEUROPATHIES MENDELIOME
MYO1E	100%	100%	100%	98%	Glomerulosclerosis, focal segmental, 6, 614131	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
MYO1H	100%	100%	100%	98%	?Central hypoventilation syndrome, congenital, 2, and autonomic dysfunction, 619482	MENDELIOME
MYO3A	100%	100%	100%	97%	Deafness, autosomal recessive 30, 607101	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
MYO5A	100%	100%	100%	98%	Griscelli syndrome, type 1, 214450	VISION DISORDERS SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

MYO5B	100%	99%	100%	98%	Diarrhea 2, with microvillus atrophy, w/wo cholestasis, 251850 Cholestasis, progressive familial intrahepatic, 10, 619868	LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING
MYO6	100%	100%	100%	98%	Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346 Deafness, autosomal dominant 22, 606346 Deafness, autosomal recessive 37, 607821	HEARING IMPAIRMENT HEART MENDELIOME PRE CONCEPTION SCREENING
MYO7A	100%	100%	100%	98%	Deafness, autosomal recessive 2, 600060 Usher syndrome, type 1B, 276900 Deafness, autosomal dominant 11, 601317	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
MYO9A	100%	100%	100%	98%	Myasthenic syndrome, congenital, 24, presynaptic, 618198	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
MYOC	100%	100%	100%	99%	Glaucoma 1A, primary open angle, 137750	VISION DISORDERS MENDELIOME
MYOCD	100%	100%	100%	98%	Megabladder, congenital, 618719	MENDELIOME
MYOD1	100%	100%	100%	99%	Congenital myopathy 17, 618975	MENDELIOME
MYOF	100%	100%	100%	98%	?Angioedema, hereditary, 7, 619366	PRIMARY IMMUNODEFICIENCY MENDELIOME
MYOM1	100%	100%	100%	98%	No OMIM disease ID	HEART MENDELIOME
MYORG	100%	100%	100%	100%	Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317	MOVEMENT DISORDERS MENDELIOME PARKINSON PRE CONCEPTION SCREENING
MYOT	100%	100%	100%	98%	Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920	HEART MENDELIOME MUSCLE DISORDERS
MYOZ2	100%	100%	100%	99%	Cardiomyopathy, hypertrophic, 16, 613838	HEART MENDELIOME
MYPN	100%	100%	100%	98%	Cardiomyopathy, hypertrophic, 22, 615248 Cardiomyopathy, familial restrictive, 4, 615248 Cardiomyopathy, dilated, 1KK, 615248 Nemaline myopathy 11, autosomal recessive, 617336	HEART MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
MYRF	100%	100%	100%	98%	Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113 Cardiac-urogenital syndrome, 618280	CONGENITAL HEART DISEASE DISORDERS OF SEX DEVELOPMENT HEART MENDELIOME

MYSM1	100%	100%	100%	98%	Bone marrow failure syndrome 4, 618116	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
MYT1L	100%	100%	100%	99%	Intellectual developmental disorder, autosomal dominant 39, 616521	INTELLECTUAL DISABILITY MENDELIOME
NAA10	100%	100%	98%	70%	Microphthalmia, syndromic 1, 309800 Ogden syndrome, 300855	VISION DISORDERS CRANIOFACIAL ANOMALIES SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME
NAA15	96%	96%	100%	98%	Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787	CONGENITAL HEART DISEASE HEART INTELLECTUAL DISABILITY MENDELIOME
NAA20	100%	100%	99%	97%	Intellectual developmental disorder, autosomal recessive 73, 619717	INTELLECTUAL DISABILITY MENDELIOME
NACC1	100%	100%	100%	99%	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
NADK2	100%	100%	100%	95%	2,4-dienoyl-CoA reductase deficiency, 616034	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
NADSYN1	100%	100%	100%	99%	Vertebral, cardiac, renal, and limb defects syndrome 3, 618845	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
NAE1	100%	100%	100%	98%	Neurodevelopmental disorder with dysmorphic facies and ischiopubic hypoplasia, 620210	INTELLECTUAL DISABILITY MENDELIOME
NAGA	100%	100%	100%	99%	Schindler disease, type I, 609241 Kanzaki disease, 609242 Schindler disease, type III, 609241	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

NAGLU	100%	100%	100%	98%	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920	NEUROPATHIES HNPD SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
NAGS	100%	100%	100%	98%	N-acetylglutamate synthase deficiency, 237310	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
NALCN	100%	100%	100%	98%	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
NANOS1	100%	99%	99%	69%	Spermatogenic failure 12, 615413	MENDELIOME
NANS	100%	100%	100%	98%	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442	MOVEMENT DISORDERS EPILEPSY SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
NAPB	100%	100%	100%	99%	Developmental and epileptic encephalopathy 107, 620033	INTELLECTUAL DISABILITY MENDELIOME
NARS1	100%	100%	100%	98%	Neurodevelopmental disorder with microcephaly, impaired language, epilepsy, and gait abnormalities, autosomal dominant, 619092 Neurodevelopmental disorder with microcephaly, impaired language, and gait abnormalities, autosomal recessive, 619091	NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME
NARS2	100%	100%	100%	98%	Combined oxidative phosphorylation deficiency 24, 616239 ?Deafness, autosomal recessive 94, 618434	MOVEMENT DISORDERS HEARING IMPAIRMENT EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
NAT8L	98%	93%	97%	75%	?N-acetylaspartate deficiency, 614063	MENDELIOME PRE CONCEPTION SCREENING

NAXD	100%	100%	100%	99%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
NAXE	100%	100%	100%	98%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
NBAS	100%	99%	100%	98%	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 Infantile liver failure syndrome 2, 616483	VISION DISORDERS SKIN DISORDERS PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
NBEA	99%	99%	100%	98%	Neurodevelopmental disorder w/wo early-onset generalized epilepsy, 619157	EPILEPSY HEMOSTATIC/THROMBOTIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME
NBEAL2	100%	100%	100%	99%	Gray platelet syndrome, 139090	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
NBN	100%	100%	100%	97%	Leukemia, acute lymphoblastic, 613065 Aplastic anemia, 609135 Nijmegen breakage syndrome, 251260	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING TUMOR
NCAPD2	100%	100%	100%	99%	?Microcephaly 21, primary, autosomal recessive, 617983	MENDELIOME PRE CONCEPTION SCREENING
NCAPD3	100%	100%	100%	99%	Microcephaly 22, primary, autosomal recessive, 617984	MENDELIOME PRE CONCEPTION SCREENING
NCAPG2	100%	100%	100%	98%	Khan-Khan-Katsanis syndrome, 618460	CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING

NCAPH	100%	100%	100%	99%	?Microcephaly 23, primary, autosomal recessive, 617985	MENDELIOME PRE CONCEPTION SCREENING
NCDN	100%	100%	100%	99%	Neurodevelopmental disorder with infantile epileptic spasms, 619373	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
NCF1	100%	99%	100%	96%	Chronic granulomatous disease 1, autosomal recessive, 233700	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
NCF2	100%	100%	100%	98%	Chronic granulomatous disease 2, autosomal recessive, 233710	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
NCF4	100%	100%	100%	98%	Chronic granulomatous disease 3, autosomal recessive, 613960	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
NCKAP1	100%	100%	100%	97%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
NCKAP1L	100%	100%	100%	98%	Immunodeficiency 72 with autoinflammation, 618982	PRIMARY IMMUNODEFICIENCY MENDELIOME
NCOA3	100%	100%	100%	99%	No OMIM disease ID	HEARING IMPAIRMENT MENDELIOME
NCOA4	100%	100%	100%	97%	No OMIM disease ID	IRON DISORDERS MENDELIOME
NCSTN	100%	100%	100%	99%	Acne inversa, familial, 1, 142690	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME
NDE1	100%	100%	100%	98%	Microhydranencephaly, 605013 Lissencephaly 4 (with microcephaly), 614019	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
NDN	100%	100%	100%	98%	No OMIM disease ID	MENDELIOME
NDNF	100%	100%	100%	98%	Hypogonadotropic hypogonadism 25 with anosmia, 618841	MENDELIOME
NDP	100%	100%	98%	74%	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600	VISION DISORDERS HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME
NDRG1	100%	100%	100%	99%	Charcot-Marie-Tooth disease, type 4D, 601455	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING
NDST1	100%	100%	100%	99%	Intellectual developmental disorder, autosomal recessive 46, 616116	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

NDUFA1	100%	100%	97%	67%	Mitochondrial complex I deficiency, nuclear type 12, 301020	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS
NDUFA10	100%	100%	100%	98%	Mitochondrial complex I deficiency, nuclear type 22, 618243	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
NDUFA11	100%	98%	100%	96%	Mitochondrial complex I deficiency, nuclear type 14, 618236	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
NDUFA12	100%	100%	100%	97%	Mitochondrial complex I deficiency, nuclear type 23, 618244	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
NDUFA13	100%	100%	100%	99%	?Mitochondrial complex I deficiency, nuclear type 28, 618249	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
NDUFA2	100%	100%	100%	99%	Mitochondrial complex I deficiency, nuclear type 13, 618235	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
NDUFA3	91%	86%	100%	98%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
NDUFA4	100%	100%	100%	95%	?Mitochondrial complex IV deficiency, nuclear type 21, 619065	MENDELIOME MITOCHONDRIAL DISORDERS
NDUFA5	100%	100%	100%	97%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
NDUFA6	100%	100%	100%	98%	Mitochondrial complex I deficiency, nuclear type 33, 618253	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
NDUFA7	100%	100%	100%	98%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
NDUFA8	100%	100%	100%	98%	Mitochondrial complex I deficiency, nuclear type 37, 619272	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS
NDUFA9	100%	100%	100%	99%	Mitochondrial complex I deficiency, nuclear type 26, 618247	NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING

NDUFAB1	100%	100%	100%	97%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
NDUFAF1	100%	100%	100%	98%	Mitochondrial complex I deficiency, nuclear type 11, 618234	EPILEPSY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
NDUFAF2	100%	100%	100%	96%	Mitochondrial complex I deficiency, nuclear type 10, 618233	EPILEPSY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
NDUFAF3	100%	100%	100%	95%	Mitochondrial complex I deficiency, nuclear type 18, 618240	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
NDUFAF4	100%	100%	100%	95%	Mitochondrial complex I deficiency, nuclear type 15, 618237	EPILEPSY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
NDUFAF5	100%	100%	99%	96%	Mitochondrial complex I deficiency, nuclear type 16, 618238	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
NDUFAF6	100%	100%	100%	96%	Mitochondrial complex I deficiency, nuclear type 17, 618239 Fanconi renotubular syndrome 5, 618913	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
NDUFAF7	100%	100%	100%	97%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
NDUFAF8	100%	100%	100%	99%	Mitochondrial complex I deficiency, nuclear type 34, 618776	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS
NDUFB1	100%	100%	99%	93%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
NDUFB10	100%	100%	100%	95%	?Mitochondrial complex I deficiency, nuclear type 35, 619003	MENDELIOME MITOCHONDRIAL DISORDERS

NDUFB11	99%	97%	88%	62%	Linear skin defects with multiple congenital anomalies 3, 300952 ?Mitochondrial complex I deficiency, nuclear type 30, 301021	VISION DISORDERS SKIN DISORDERS HEART IRON DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
NDUFB2	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
NDUFB3	100%	100%	100%	99%	Mitochondrial complex I deficiency, nuclear type 25, 618246	EPILEPSY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
NDUFB4	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
NDUFB5	100%	100%	100%	98%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
NDUFB6	100%	100%	100%	98%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
NDUFB7	100%	100%	99%	96%	?Mitochondrial complex I deficiency, nuclear type 39, 620135	MENDELIOME MITOCHONDRIAL DISORDERS
NDUFB8	100%	100%	100%	97%	Mitochondrial complex I deficiency, nuclear type 32, 618252	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
NDUFB9	100%	100%	100%	98%	?Mitochondrial complex I deficiency, nuclear type 24, 618245	EPILEPSY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
NDUFC1	100%	100%	100%	97%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
NDUFC2	100%	100%	100%	97%	Mitochondrial complex I deficiency, nuclear type 36, 619170	MENDELIOME MITOCHONDRIAL DISORDERS
NDUFS1	100%	100%	100%	98%	Mitochondrial complex I deficiency, nuclear type 5, 618226	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING

NDUFS2	100%	100%	100%	98%	Mitochondrial complex I deficiency, nuclear type 6, 618228	VISION DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
NDUFS3	96%	91%	100%	99%	Mitochondrial complex I deficiency, nuclear type 8, 618230	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
NDUFS4	100%	99%	100%	98%	Mitochondrial complex I deficiency, nuclear type 1, 252010	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
NDUFS5	100%	100%	100%	98%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
NDUFS6	100%	100%	100%	98%	Mitochondrial complex I deficiency, nuclear type 9, 618232	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
NDUFS7	100%	100%	100%	99%	Mitochondrial complex I deficiency, nuclear type 3, 618224	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
NDUFS8	100%	100%	100%	99%	Mitochondrial complex I deficiency, nuclear type 2, 618222	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
NDUFV1	100%	100%	99%	98%	Mitochondrial complex I deficiency, nuclear type 4, 618225	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
NDUFV2	100%	100%	100%	98%	Mitochondrial complex I deficiency, nuclear type 7, 618229	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING

NDUFV3	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
NEB	99%	99%	99%	97%	Nemaline myopathy 2, autosomal recessive, 256030 Arthrogryposis multiplex congenita 6, 619334	FETAL AKINESIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
NEBL	99%	99%	100%	97%	No OMIM disease ID	HEART MENDELIOME
NECAP1	100%	100%	100%	98%	Developmental and epileptic encephalopathy 21, 615833	EPILEPSY MENDELIOME PRE CONCEPTION SCREENING
NECTIN1	100%	100%	100%	99%	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS
NECTIN4	100%	100%	100%	99%	Ectodermal dysplasia-syndactyly syndrome 1, 613573	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
NEDD4L	100%	100%	100%	97%	Periventricular nodular heterotopia 7, 617201	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME SCHISIS
NEFH	100%	100%	99%	96%	Charcot-Marie-Tooth disease, axonal, type 2CC, 616924	NEUROPATHIES MENDELIOME MUSCLE DISORDERS
NEFL	100%	100%	100%	97%	Charcot-Marie-Tooth disease, type 1F, 607734 Charcot-Marie-Tooth disease, dominant intermediate G, 617882 Charcot-Marie-Tooth disease, type 2E, 607684	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME
NEK1	100%	100%	100%	98%	Short-rib thoracic dysplasia 6 w/wo polydactyly, 263520	ALS VISION DISORDERS CILIOPATHIES DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS
NEK10	100%	99%	100%	98%	Ciliary dyskinesia, primary, 44, 618781	CILIOPATHIES MENDELIOME

NEK11	100%	99%	100%	98%	No OMIM disease ID	SKIN DISORDERS MENDELIOME
NEK2	95%	95%	100%	98%	?Retinitis pigmentosa 67, 615565	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
NEK8	100%	100%	100%	99%	Renal-hepatic-pancreatic dysplasia 2, 615415 ?Nephronophthisis 9, 613824	CILIOPATHIES RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
NEK9	100%	100%	100%	98%	?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262 Nevus comedonicus, somatic, 617025 Lethal congenital contracture syndrome 10, 617022	FETAL AKINESIA SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
NEMF	100%	100%	100%	98%	Intellectual developmental disorder with speech delay and axonal peripheral neuropathy, 619099	NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME
NEPRO	100%	100%	100%	97%	Anauxetic dysplasia 3, 618853	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
NEU1	100%	100%	100%	99%	Sialidosis, type II, 256550 Sialidosis, type I, 256550	MOVEMENT DISORDERS EPILEPSY SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
NEUROD1	100%	100%	100%	97%	Maturity-onset diabetes of the young 6, 606394	VISION DISORDERS MENDELIOME
NEUROD2	100%	100%	100%	92%	Developmental and epileptic encephalopathy 72, 618374	INTELLECTUAL DISABILITY MENDELIOME
NEUROG3	100%	100%	100%	98%	Diarrhea 4, malabsorptive, congenital, 610370	MENDELIOME PRE CONCEPTION SCREENING
NEXMIF	100%	99%	97%	69%	Intellectual developmental disorder, X-linked 98, 300912	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME

NEXN	100%	100%	99%	95%	Cardiomyopathy, dilated, 1CC, 613122 Cardiomyopathy, hypertrophic, 20, 613876	DILATED CARDIOMYOPATHY HEART MENDELIOME
NF1	100%	100%	100%	98%	Watson syndrome, 193520 Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321	HEREDITARY BONE MARROW FAILURE CONGENITAL HEART DISEASE SKIN DISORDERS HEART SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME AND RASOPATHY TUMOR
NF2	100%	100%	100%	97%	Neurofibromatosis, type 2, 101000 Meningioma, NF2-related, somatic, 607174 Schwannomatosis, somatic, 162091	MOVEMENT DISORDERS MENDELIOME TUMOR
NFASC	100%	100%	100%	99%	Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING
NFAT5	100%	100%	100%	98%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
NFATC1	100%	100%	99%	96%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
NFE2	100%	100%	100%	99%	No OMIM disease ID	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME
NFE2L2	100%	100%	100%	98%	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744	PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME
NFIA	100%	100%	99%	93%	Brain malformations w/wo urinary tract defects, 613735	INTELLECTUAL DISABILITY MENDELIOME
NFIB	100%	100%	100%	98%	Macrocephaly, acquired, with impaired intellectual development, 618286	INTELLECTUAL DISABILITY MENDELIOME
NFIX	100%	99%	99%	97%	Marshall-Smith syndrome, 602535 Malan syndrome, 614753	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
NFKB1	100%	100%	100%	98%	Immunodeficiency, common variable, 12, 616576	PRIMARY IMMUNODEFICIENCY MENDELIOME

NFKB2	100%	100%	100%	98%	Immunodeficiency, common variable, 10, 615577	PRIMARY IMMUNODEFICIENCY MENDELIOME
NFKBIA	100%	100%	100%	95%	Ectodermal dysplasia and immunodeficiency 2, 612132	CRANIOFACIAL ANOMALIES SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME
NFS1	89%	89%	100%	99%	Combined oxidative phosphorylation deficiency 52, 619386	MENDELIOME MITOCHONDRIAL DISORDERS
NFU1	100%	100%	100%	98%	Multiple mitochondrial dysfunctions syndrome 1, 605711	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
NGF	100%	100%	100%	100%	Neuropathy, hereditary sensory and autonomic, type V, 608654	NEUROPATHIES HNPD MENDELIOME PRE CONCEPTION SCREENING
NGLY1	100%	100%	100%	98%	Congenital disorder of deglycosylation 1, 615273	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
NHEJ1	100%	100%	100%	99%	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)
NHLH2	100%	100%	100%	94%	?Hypogonadotropic hypogonadism 27 without anosmia, 619755	MENDELIOME
NHLRC1	100%	100%	100%	99%	Epilepsy, progressive myoclonic 2B (Lafora), 254780	EPILEPSY MENDELIOME PRE CONCEPTION SCREENING
NHLRC2	100%	99%	100%	98%	FINCA syndrome, 618278	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

NHP2	100%	100%	100%	98%	Dyskeratosis congenita, autosomal recessive 2, 613987	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS DYSKERATOSIS CONGENITA PRIMARY IMMUNODEFICIENCY LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING TUMOR
NHS	100%	100%	97%	69%	Cataract 40, X-linked, 302200 Nance-Horan syndrome, 302350	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME
NIN	100%	100%	100%	98%	?Seckel syndrome 7, 614851	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
NIPA1	100%	100%	100%	95%	Spastic paraplegia 6, autosomal dominant, 600363	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME
NIPAL4	100%	100%	100%	98%	Ichthyosis, congenital, autosomal recessive 6, 612281	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
NIPBL	100%	100%	100%	98%	Cornelia de Lange syndrome 1, 122470	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS
NKAP	100%	100%	96%	68%	Intellectual developmental disorder, X-linked syndromic, Hackman-Di Donato type, 301039	INTELLECTUAL DISABILITY MENDELIOME
NKX2-1	100%	100%	100%	96%	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME
NKX2-5	100%	100%	100%	98%	Hypoplastic left heart syndrome 2, 614435 Tetralogy of Fallot, 187500 Hypothyroidism, congenital nongoitrous, 5, 225250 Conotruncal heart malformations, variable, 217095 Ventricular septal defect 3, 614432 Atrial septal defect 7, w/wo AV conduction defects, 108900	CONGENITAL HEART DISEASE HEART MENDELIOME

NKX2-6	100%	100%	100%	99%	Persistent truncus arteriosus, 217095 Conotruncal heart malformations, 217095	CONGENITAL HEART DISEASE HEART MENDELIOME PRE CONCEPTION SCREENING
NKX3-2	100%	100%	100%	95%	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
NKX6-2	100%	100%	99%	80%	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560	MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING
NLGN2	100%	100%	100%	97%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
NLGN3	100%	100%	98%	68%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
NLGN4X	100%	99%	98%	74%	Intellectual developmental disorder, X-linked, 300495	INTELLECTUAL DISABILITY MENDELIOME
NLRC4	100%	100%	100%	98%	?Familial cold autoinflammatory syndrome 4, 616115 Autoinflammation with infantile enterocolitis, 616050	PRIMARY IMMUNODEFICIENCY MENDELIOME
NLRP1	100%	100%	100%	98%	?Respiratory papillomatosis, juvenile recurrent, congenital, 618803 Autoinflammation with arthritis and dyskeratosis, 617388 Palmoplantar carcinoma, multiple self-healing, 615225	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
NLRP12	100%	100%	100%	97%	Familial cold autoinflammatory syndrome 2, 611762	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME
NLRP3	100%	100%	100%	98%	CINCA syndrome, 607115 Familial cold inflammatory syndrome 1, 120100 Keratoendothelitis fugax hereditaria, 148200 Deafness, autosomal dominant 34, w/wo inflammation, 617772 Muckle-Wells syndrome, 191900	SKIN DISORDERS HEARING IMPAIRMENT PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
NLRP6	100%	100%	99%	97%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
NLRP7	100%	100%	100%	98%	Hydatidiform mole, recurrent, 1, 231090	MENDELIOME PRE CONCEPTION SCREENING
NME1	100%	100%	100%	99%	No OMIM disease ID	SKIN DISORDERS MENDELIOME
NME3	100%	100%	99%	95%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
NME5	100%	100%	99%	97%	Ciliary dyskinesia, primary, 48, without situs inversus, 620032	CILIOPATHIES MENDELIOME

NME8	99%	99%	100%	98%	Ciliary dyskinesia, primary, 6, 610852	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING
NMNAT1	99%	97%	100%	97%	Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis, 619260 Leber congenital amaurosis 9, 608553	VISION DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
NMNAT2	100%	100%	100%	99%	No OMIM disease ID	NEUROPATHIES HNPD MENDELIOME
NNT	96%	96%	100%	99%	Glucocorticoid deficiency 4, w/wo mineralocorticoid deficiency, 614736	DISORDERS OF SEX DEVELOPMENT METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
NOBOX	100%	100%	100%	99%	Premature ovarian failure 5, 611548	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRIMARY OVARIAN INSUFFICIENCY
NOD2	100%	100%	100%	99%	Blau syndrome, 186580	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME
NODAL	100%	100%	100%	99%	Heterotaxy, visceral, 5, 270100	CONGENITAL HEART DISEASE CILIOPATHIES HEART MENDELIOME
NOG	100%	100%	100%	96%	Symphalangism, proximal, 1A, 185800 Brachydactyly, type B2, 611377 Stapes ankylosis with broad thumbs and toes, 184460 Tarsal-carpal coalition syndrome, 186570 Multiple synostoses syndrome 1, 186500	CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
NOL3	100%	100%	100%	99%	?Myoclonus, familial, 1, 614937	MOVEMENT DISORDERS MENDELIOME
NONO	100%	99%	98%	71%	Intellectual developmental disorder, X-linked syndromic 34, 300967	INTELLECTUAL DISABILITY MENDELIOME

NOP10	100%	100%	100%	96%	Dyskeratosis congenita, autosomal recessive 1, 224230	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS DYSKERATOSIS CONGENITA PRIMARY IMMUNODEFICIENCY LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING TUMOR
NOP56	100%	100%	100%	98%	Spinocerebellar ataxia 36, 614153	MENDELIOME
NOS1	100%	100%	100%	99%	No OMIM disease ID	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME
NOS1AP	100%	100%	100%	98%	Nephrotic syndrome, type 22, 619155	HEART RENAL DISORDERS MENDELIOME
NOS2	100%	100%	100%	99%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
NOTCH1	100%	100%	100%	99%	Adams-Oliver syndrome 5, 616028 Aortic valve disease 1, 109730	ANEURYSM CONGENITAL HEART DISEASE SKIN DISORDERS HEART SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS
NOTCH2	100%	100%	100%	99%	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500	CONGENITAL HEART DISEASE HEART SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS RENAL DISORDERS MENDELIOME
NOTCH2NLC	100%	100%	99%	96%	Tremor, hereditary essential, 6, 618866 Oculopharyngodistal myopathy 3, 619473 Neuronal intranuclear inclusion disease, 603472	MENDELIOME
NOTCH3	100%	100%	100%	98%	Lateral meningocele syndrome, 130720 ?Myofibromatosis, infantile 2, 615293 Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310	MENDELIOME
NOVA2	100%	100%	100%	94%	Neurodevelopmental disorder w/wo autistic features and/or structural brain abnormalities, 618859	INTELLECTUAL DISABILITY MENDELIOME
NPAT	100%	100%	100%	98%	No OMIM disease ID	HEREDITARY BONE MARROW FAILURE MENDELIOME

NPC1	100%	100%	100%	99%	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220	MOVEMENT DISORDERS LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
NPC2	100%	100%	100%	98%	Niemann-pick disease, type C2, 607625	MOVEMENT DISORDERS LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
NPHP1	100%	100%	100%	98%	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
NPHP3	100%	100%	100%	98%	Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540 Meckel syndrome 7, 267010	VISION DISORDERS CILIOPATHIES LIVER DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
NPHP4	100%	100%	100%	99%	Senior-Loken syndrome 4, 606996 Nephronophthisis 4, 606966	VISION DISORDERS CILIOPATHIES RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
NPHS1	100%	100%	100%	98%	Nephrotic syndrome, type 1, 256300	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
NPHS2	100%	100%	100%	98%	Nephrotic syndrome, type 2, 600995	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
NPL	100%	100%	100%	98%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME
NPM1	100%	100%	100%	96%	Leukemia, acute myeloid, somatic, 601626	HEREDITARY BONE MARROW FAILURE DYSKERATOSIS CONGENITA MENDELIOME TUMOR

NPPA	100%	100%	100%	98%	Atrial standstill 2, 615745 Atrial fibrillation, familial, 6, 612201	HEART MENDELIOME PRE CONCEPTION SCREENING
NPPB	100%	100%	100%	99%	No OMIM disease ID	HEART MENDELIOME
NPPC	100%	100%	100%	98%	No OMIM disease ID	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
NPR2	100%	100%	100%	99%	Epiphyseal chondrodysplasia, Miura type, 615923 Short stature with nonspecific skeletal abnormalities, 616255 Acromesomelic dysplasia 1, Maroteaux type, 602875	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
NPR3	100%	100%	100%	98%	Boudin-Mortier syndrome, 619543	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
NPRL2	100%	100%	100%	99%	Epilepsy, familial focal, with variable foci 2, 617116	EPILEPSY MENDELIOME
NPRL3	100%	100%	100%	98%	Epilepsy, familial focal, with variable foci 3, 617118	EPILEPSY MENDELIOME
NPTX1	100%	100%	99%	88%	Spinocerebellar ataxia 50, 620158	MOVEMENT DISORDERS MENDELIOME
NR0B1	100%	99%	98%	74%	Adrenal hypoplasia, congenital, 300200 46XY sex reversal 2, dosage-sensitive, 300018	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MALE INFERTILITY MENDELIOME
NR0B2	100%	100%	100%	97%	Obesity, mild, early-onset, 601665	MENDELIOME PRE CONCEPTION SCREENING
NR1H4	100%	100%	100%	98%	Cholestasis, progressive familial intrahepatic, 5, 617049	LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING
NR2E3	100%	100%	100%	99%	Retinitis pigmentosa 37, 611131 Enhanced S-cone syndrome, 268100	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
NR2F1	100%	99%	99%	91%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722	VISION DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS
NR2F2	100%	100%	99%	96%	46,XX sex reversal 5, 618901 Congenital heart defects, multiple types, 4, 615779	CONGENITAL HEART DISEASE HEART MENDELIOME

NR3C1	100%	100%	100%	97%	Glucocorticoid resistance, 615962	DISORDERS OF SEX DEVELOPMENT MENDELIOME
NR3C2	100%	100%	100%	98%	Pseudohypoaldosteronism type I, autosomal dominant, 177735 Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115	DISORDERS OF SEX DEVELOPMENT RENAL DISORDERS MENDELIOME
NR4A2	100%	100%	100%	98%	Intellectual developmental disorder with language impairment and early-onset DOPA-responsive dystonia-parkinsonism, 619911	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
NR4A3	100%	100%	100%	97%	Chondrosarcoma, extraskeletal myxoid, 612237	MENDELIOME
NR5A1	100%	100%	100%	98%	46, XX sex reversal 4, 617480 Premature ovarian failure 7, 612964 46XY sex reversal 3, 612965 Adrenocortical insufficiency, 612964 Spermatogenic failure 8, 613957	DISORDERS OF SEX DEVELOPMENT MALE INFERTILITY MENDELIOME PRIMARY OVARIAN INSUFFICIENCY
NRAP	100%	100%	100%	99%	No OMIM disease ID	HEART MENDELIOME
NRAS	100%	100%	100%	99%	Noonan syndrome 6, 613224 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Melanocytic nevus syndrome, congenital, somatic, 137550 Epidermal nevus, somatic, 162900 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Thyroid carcinoma, follicular, somatic, 188470 Neurocutaneous melanosis, somatic, 249400 Colorectal cancer, somatic, 114500	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS HEART HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME AND RASOPATHY TUMOR
NRCAM	100%	100%	100%	99%	Neurodevelopmental disorder with neuromuscular and skeletal abnormalities, 619833	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
NRG1	99%	99%	100%	97%	No OMIM disease ID	NEUROPATHIES MENDELIOME
NRIP1	100%	100%	100%	99%	?Congenital anomalies of kidney and urinary tract 3, 618270	MENDELIOME
NRL	100%	100%	100%	97%	Retinitis pigmentosa 27, 613750 Retinal degeneration, autosomal recessive, clumped pigment type,	VISION DISORDERS MENDELIOME
NRROS	100%	100%	100%	99%	Seizures, early-onset, with neurodegeneration and brain calcification, 618875	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING

NRXN1	99%	99%	100%	99%	Pitt-Hopkins-like syndrome 2, 614325	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
NSD1	100%	100%	100%	98%	Sotos syndrome, 117550	CRANIOFACIAL ANOMALIES CONGENITAL HEART DISEASE SKIN DISORDERS HEART SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME TUMOR
NSD2	100%	100%	99%	98%	Rauch-Steindl syndrome, 619695	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
NSDHL	100%	99%	99%	75%	CK syndrome, 300831 CHILD syndrome, 308050	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME
NSF	100%	100%	99%	87%	Developmental and epileptic encephalopathy 96, 619340	INTELLECTUAL DISABILITY MENDELIOME
NSMCE2	100%	100%	100%	97%	Seckel syndrome 10, 617253	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
NSMCE3	100%	100%	100%	96%	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
NSMF	100%	100%	100%	98%	Hypogonadotropic hypogonadism 9 w/wo anosmia, 614838	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME
NSRP1	91%	91%	100%	97%	Neurodevelopmental disorder with spasticity, seizures, and brain abnormalities, 620001	INTELLECTUAL DISABILITY MENDELIOME
NSUN2	100%	100%	100%	99%	Intellectual developmental disorder, autosomal recessive 5, 611091	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

NSUN3	100%	100%	100%	98%	Combined oxidative phosphorylation deficiency 48, 619012	MENDELIOME MITOCHONDRIAL DISORDERS
NT5C2	100%	100%	100%	98%	Spastic paraplegia 45, autosomal recessive, 613162	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
NT5C3A	100%	100%	100%	98%	Anemia, hemolytic, due to UMPH1 deficiency, 266120	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
NT5E	100%	100%	100%	98%	Calcification of joints and arteries, 211800	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
NTF4	100%	100%	100%	98%	Glaucoma 1, open angle, 1O, 613100	MENDELIOME
NTHL1	100%	100%	100%	99%	Familial adenomatous polyposis 3, 616415	MENDELIOME PRE CONCEPTION SCREENING TUMOR
NTN1	100%	100%	100%	97%	Mirror movements 4, 618264	MENDELIOME
NTNG2	100%	100%	100%	98%	Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
NTRK1	100%	100%	100%	99%	Insensitivity to pain, congenital, with anhidrosis, 256800	NEUROPATHIES HNPD INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
NTRK2	100%	100%	100%	98%	Developmental and epileptic encephalopathy 58, 617830 Obesity, hyperphagia, and developmental delay, 613886	INTELLECTUAL DISABILITY MENDELIOME
NUAK2	100%	100%	100%	99%	?Anencephaly 2, 619452	MENDELIOME
NUBPL	100%	100%	100%	98%	Mitochondrial complex I deficiency, nuclear type 21, 618242	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
NUDT2	100%	100%	100%	97%	Intellectual developmental disorder w/wo peripheral neuropathy, 619844	INTELLECTUAL DISABILITY MENDELIOME
NUMA1	100%	100%	100%	98%	Leukemia, acute promyelocytic, somatic, 612376	MENDELIOME
NUP107	100%	100%	100%	98%	?Ovarian dysgenesis 6, 618078 Galloway-Mowat syndrome 7, 618348 Nephrotic syndrome, type 11, 616730	INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING

NUP133	100%	100%	100%	98%	?Galloway-Mowat syndrome 8, 618349 Nephrotic syndrome, type 18, 618177	INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
NUP155	100%	100%	100%	97%	?Atrial fibrillation 15, 615770	HEART MENDELIOME
NUP160	100%	100%	100%	98%	?Nephrotic syndrome, type 19, 618178	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
NUP188	100%	100%	100%	98%	Sandestig-Stefanova syndrome, 618804	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
NUP205	100%	100%	100%	98%	?Nephrotic syndrome, type 13, 616893	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
NUP214	100%	100%	100%	98%	Leukemia, T-cell acute lymphoblastic, somatic, 613065 Leukemia, acute myeloid, somatic, 601626	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
NUP37	100%	100%	100%	98%	?Microcephaly 24, primary, autosomal recessive, 618179	MENDELIOME PRE CONCEPTION SCREENING
NUP54	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS
NUP62	100%	100%	100%	99%	Striatonigral degeneration, infantile, 271930	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
NUP85	100%	100%	99%	97%	Nephrotic syndrome, type 17, 618176	INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
NUP88	100%	100%	100%	98%	Fetal akinesia deformation sequence 4, 618393	FETAL AKINESIA MENDELIOME PRE CONCEPTION SCREENING
NUP93	95%	95%	100%	99%	Nephrotic syndrome, type 12, 616892	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING

NUS1	100%	100%	100%	98%	Intellectual developmental disorder, autosomal dominant 55, with seizures, 617831 ?Congenital disorder of glycosylation, type 1aa, 617082	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
NUTM2B-AS1	NC	NC	NC	NC	?Oculopharyngeal myopathy with leukoencephalopathy 1, 618637	MENDELIOME
NXF5	100%	99%	96%	66%	No OMIM disease ID	RENAL DISORDERS MENDELIOME
NXN	100%	100%	100%	94%	Robinow syndrome, autosomal recessive 2, 618529	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
NYX	100%	100%	99%	83%	Night blindness, congenital stationary (complete), 1A, X-linked, 310500	VISION DISORDERS MENDELIOME
OAS1	100%	100%	100%	98%	Immunodeficiency 100 with pulmonary alveolar proteinosis and hypogammaglobulinemia, 618042	PRIMARY IMMUNODEFICIENCY MENDELIOME
OAT	100%	100%	100%	98%	Gyrate atrophy of choroid and retina w/wo ornithinemia, 258870	VISION DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
OBSL1	100%	100%	100%	99%	3-M syndrome 2, 612921	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
OCA2	100%	100%	100%	99%	Albinism, brown oculocutaneous, 203200 Albinism, oculocutaneous, type II, 203200	VISION DISORDERS SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
OCLN	100%	100%	100%	97%	Pseudo-TORCH syndrome 1, 251290	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

OCRL	100%	100%	97%	70%	Dent disease 2, 300555 Lowe syndrome, 309000	VISION DISORDERS CILIOPATHIES HEMOSTATIC/THROMBOTIC DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME
ODAM	100%	99%	100%	98%	No OMIM disease ID	SKIN DISORDERS MENDELIOME
ODAPH	100%	100%	100%	97%	Amelogenesis imperfecta, type IIA4, 614832	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ODC1	100%	100%	100%	99%	Bachmann-Bupp syndrome, 619075	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME
OFD1	100%	100%	96%	67%	Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424 Orofaciodigital syndrome I, 311200 Joubert syndrome 10, 300804	VISION DISORDERS CRANIOFACIAL ANOMALIES CILIOPATHIES SKIN DISORDERS EPILEPSY SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME SCHISIS
OGDH	100%	100%	100%	99%	Oxoglutarate dehydrogenase deficiency, 203740	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
OGDHL	100%	100%	100%	99%	Yoon-Bellen neurodevelopmental syndrome, 619701	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
OGG1	100%	100%	100%	99%	Renal cell carcinoma, clear cell, somatic, 144700	MENDELIOME
OGT	100%	99%	98%	75%	Intellectual developmental disorder, X-linked 106, 300997	INTELLECTUAL DISABILITY MENDELIOME

OPA1	100%	100%	100%	98%	Optic atrophy plus syndrome, 125250 Optic atrophy 1, 165500 Behr syndrome, 210000 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896	MOVEMENT DISORDERS VISION DISORDERS HEARING IMPAIRMENT MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS
OPA3	100%	100%	100%	98%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300	VISION DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
OPCML	100%	100%	100%	99%	Ovarian cancer, somatic, 167000	MENDELIOME
OPHN1	100%	99%	98%	72%	Intellectual developmental disorder, X-linked syndromic, Billuart type, 300486	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
OPLAH	100%	100%	100%	99%	5-oxoprolinase deficiency, 260005	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
OPN1LW	94%	94%	95%	65%	Blue cone monochromacy, 303700 Colorblindness, protan, 303900	VISION DISORDERS MENDELIOME
OPN1MW	97%	94%	80%	45%	Colorblindness, deutan, 303800 Blue cone monochromacy, 303700	VISION DISORDERS MENDELIOME
OPN1SW	100%	100%	100%	99%	Colorblindness, tritan, 190900	MENDELIOME
OPTN	100%	100%	100%	98%	Glaucoma 1, open angle, E, 137760 Amyotrophic lateral sclerosis 12 w/wo frontotemporal dementia, 613435	ALS VISION DISORDERS MENDELIOME
ORAI1	100%	100%	99%	92%	Immunodeficiency 9, 612782 Myopathy, tubular aggregate, 2, 615883	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
ORC1	100%	100%	100%	99%	Meier-Gorlin syndrome 1, 224690	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS

ORC4	99%	98%	100%	98%	Meier-Gorlin syndrome 2, 613800	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
ORC6	100%	100%	100%	99%	Meier-Gorlin syndrome 3, 613803	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
OSBPL2	100%	100%	100%	99%	Deafness, autosomal dominant 67, 616340	HEARING IMPAIRMENT MENDELIOME
OSGEP	100%	100%	100%	99%	Galloway-Mowat syndrome 3, 617729	INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
OSMR	100%	100%	100%	99%	Amyloidosis, primary localized cutaneous, 1, 105250	SKIN DISORDERS MENDELIOME
OSTM1	100%	100%	100%	98%	Osteopetrosis, autosomal recessive 5, 259720	PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
OTC	100%	99%	96%	68%	Ornithine transcarbamylase deficiency, 311250	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME
OTOA	100%	100%	100%	98%	Deafness, autosomal recessive 22, 607039	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
OTOF	100%	100%	100%	98%	Auditory neuropathy, autosomal recessive, 1, 601071 Deafness, autosomal recessive 9, 601071	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
OTOG	100%	100%	100%	99%	Deafness, autosomal recessive 18B, 614945	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
OTOGL	100%	100%	100%	98%	Deafness, autosomal recessive 84B, 614944	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
OTUD5	100%	99%	96%	67%	Multiple congenital anomalies-neurodevelopmental syndrome, X-linked, 301056	INTELLECTUAL DISABILITY MENDELIOME
OTUD6B	100%	100%	100%	97%	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

OTUD7A	99%	98%	99%	89%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
OTULIN	100%	100%	100%	98%	Autoinflammation, panniculitis, and dermatosis syndrome, 617099	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
OTX2	100%	100%	100%	98%	Retinal dystrophy, early-onset, w/wo pituitary dysfunction, 610125 Pituitary hormone deficiency, combined, 6, 613986 Microphthalmia, syndromic 5, 610125	VISION DISORDERS CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS
OVOL2	100%	99%	100%	98%	Corneal dystrophy, posterior polymorphous, 1, 122000	VISION DISORDERS MENDELIOME
OXA1L	100%	100%	100%	98%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
OXCT1	100%	100%	100%	97%	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
OXGR1	100%	100%	100%	99%	No OMIM disease ID	RENAL DISORDERS MENDELIOME
OXR1	100%	100%	100%	98%	Cerebellar hypoplasia/atrophy, epilepsy, and global developmental delay, 213000	INTELLECTUAL DISABILITY MENDELIOME
P2RX2	100%	100%	99%	93%	Deafness, autosomal dominant 41, 608224	HEARING IMPAIRMENT MENDELIOME
P2RY12	100%	100%	100%	98%	Bleeding disorder, platelet-type, 8, 609821	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
P3H1	100%	100%	100%	99%	Osteogenesis imperfecta, type VIII, 610915	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
P3H2	100%	100%	100%	98%	Myopia, high, with cataract and vitreoretinal degeneration, 614292	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
P4HA2	100%	100%	99%	98%	Myopia 25, autosomal dominant, 617238	VISION DISORDERS MENDELIOME
P4HB	100%	100%	100%	99%	Cole-Carpenter syndrome 1, 112240	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME

P4HTM	100%	100%	100%	95%	Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
PABPN1	100%	100%	100%	95%	Oculopharyngeal muscular dystrophy, 164300	MENDELIOME MUSCLE DISORDERS
PACS1	100%	100%	99%	95%	Schuurs-Hoeijmakers syndrome, 615009	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
PACS2	100%	100%	99%	96%	Developmental and epileptic encephalopathy 66, 618067	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
PADI3	100%	100%	100%	99%	Uncombable hair syndrome, 191480	SKIN DISORDERS MENDELIOME
PADI6	100%	99%	99%	97%	Preimplantation embryonic lethality 2, 617234	MENDELIOME
PAFAH1B1	100%	100%	100%	98%	Subcortical laminar heterotopia, 607432 Lissencephaly 1, 607432	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
PAH	100%	100%	100%	99%	Phenylketonuria, 261600	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PAK1	100%	100%	100%	98%	Intellectual developmental disorder with macrocephaly, seizures, and speech delay, 618158	INTELLECTUAL DISABILITY MENDELIOME
PAK2	100%	100%	100%	97%	?Knobloch syndrome 2, 618458	VISION DISORDERS MENDELIOME
PAK3	99%	99%	97%	71%	Intellectual developmental disorder, X-linked 30, 300558	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
PALB2	100%	100%	100%	96%	Fanconi anemia, complementation group N, 610832	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS HEREDITARY BREAST AND OVARIAN CANCER MENDELIOME SONIC HEDGEHOG MEDULLOBLASTOMA TUMOR

PAM16	85%	84%	100%	99%	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PAN2	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
PANK2	100%	100%	100%	98%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200	MOVEMENT DISORDERS VISION DISORDERS IRON DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
PANK4	100%	100%	99%	98%	?Cataract 49, 619593	VISION DISORDERS MENDELIOME
PANX1	100%	100%	100%	99%	Oocyte maturation defect 7, 618550	INTELLECTUAL DISABILITY MENDELIOME
PAPPA2	100%	99%	100%	99%	Short stature, Dauber-Argente type, 619489	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
PAPSS2	100%	99%	100%	98%	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
PARK7	100%	100%	100%	98%	Parkinson disease 7, autosomal recessive early-onset, 606324	MENDELIOME PARKINSON PRE CONCEPTION SCREENING
PARN	97%	95%	100%	98%	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371	HEREDITARY BONE MARROW FAILURE DYSKERATOSIS CONGENITA PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING TUMOR
PARP4	100%	100%	100%	98%	No OMIM disease ID	HEREDITARY BONE MARROW FAILURE MENDELIOME
PARP6	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME

PARS2	100%	100%	100%	99%	Developmental and epileptic encephalopathy 75, 618437	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
PATL2	100%	100%	100%	98%	Oocyte maturation defect 4, 617743	MENDELIOME PRE CONCEPTION SCREENING
PAX1	100%	100%	99%	97%	Otofaciocervical syndrome 2, 615560	PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)
PAX2	100%	100%	100%	97%	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330	VISION DISORDERS RENAL DISORDERS MENDELIOME
PAX3	100%	99%	100%	98%	Craniofacial-deafness-hand syndrome, 122880 Waardenburg syndrome, type 3, 148820 Waardenburg syndrome, type 1, 193500 Rhabdomyosarcoma 2, alveolar, 268220	CRANIOFACIAL ANOMALIES SKIN DISORDERS HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS
PAX4	100%	100%	100%	99%	Maturity-onset diabetes of the young, type IX, 612225 Diabetes mellitus, type 2, 125853	MENDELIOME
PAX5	100%	100%	100%	99%	No OMIM disease ID	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCY MENDELIOME TUMOR
PAX6	100%	100%	100%	97%	Optic nerve hypoplasia, 165550 Cataract with late-onset corneal dystrophy, 106210 ?Coloboma, ocular, 120200 ?Coloboma of optic nerve, 120430 Aniridia, 106210 Anterior segment dysgenesis 5, multiple subtypes, 604229 ?Morning glory disc anomaly, 120430 Foveal hypoplasia 1, 136520 Keratitis, 148190	MOVEMENT DISORDERS VISION DISORDERS CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME
PAX7	100%	100%	100%	98%	Congenital myopathy 19, 618578 Rhabdomyosarcoma 2, alveolar, 268220	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS

PAX8	100%	100%	100%	98%	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700	INTELLECTUAL DISABILITY MENDELIOME
PAX9	100%	100%	100%	99%	Tooth agenesis, selective, 3, 604625	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME
PBRM1	100%	100%	100%	98%	?Renal cell carcinoma, clear cell, 144700	MENDELIOME
PBX1	100%	99%	100%	98%	Congenital anomalies of kidney and urinary tract syndrome w/wo hearing loss, abnormal ears, or developmental delay, 617641	DISORDERS OF SEX DEVELOPMENT PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME
PC	100%	100%	100%	99%	Pyruvate carboxylase deficiency, 266150	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
PCARE	100%	100%	100%	97%	Retinitis pigmentosa 54, 613428	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PCBD1	100%	100%	100%	99%	Hyperphenylalaninemia, BH4-deficient, D, 264070	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PCCA	100%	100%	100%	98%	Propionicacidemia, 606054	HEART PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PCCB	99%	98%	100%	98%	Propionicacidemia, 606054	HEART PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PCDH12	100%	100%	100%	99%	Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

PCDH15	100%	100%	100%	98%	Usher syndrome, type 1D/F digenic, 601067 Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1F, 602083	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
PCDH19	100%	99%	98%	74%	Developmental and epileptic encephalopathy 9, 300088	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
PCDHGC4	100%	100%	100%	99%	Neurodevelopmental disorder with poor growth and skeletal anomalies, 619880	INTELLECTUAL DISABILITY MENDELIOME
PCGF2	100%	100%	100%	97%	Turnpenny-Fry syndrome, 618371	INTELLECTUAL DISABILITY MENDELIOME
PCK1	100%	100%	100%	99%	?Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PCK2	100%	100%	100%	99%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PCLO	99%	99%	99%	97%	?Pontocerebellar hypoplasia, type 3, 608027	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PCNA	100%	100%	100%	99%	?Ataxia-telangiectasia-like disorder 2, 615919	SKIN DISORDERS MENDELIOME
PCNT	100%	100%	100%	99%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PCSK1	100%	100%	100%	98%	Endocrinopathy due to proprotein convertase 1/3 deficiency, 600955	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME PRE CONCEPTION SCREENING
PCSK9	100%	100%	100%	99%	Hypercholesterolemia, familial, 3, 603776	MENDELIOME
PCYT1A	100%	100%	100%	98%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940	VISION DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PCYT2	100%	100%	99%	97%	Spastic paraplegia 82, autosomal recessive, 618770	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME

PDCD1	100%	100%	100%	99%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
PDCD10	100%	100%	100%	97%	Cerebral cavernous malformations-3, 603285	EPILEPSY MENDELIOME
PDE10A	99%	98%	97%	85%	Striatal degeneration, autosomal dominant, 616922 Dyskinesia, limb and orofacial, infantile-onset, 616921	MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PDE11A	100%	99%	100%	98%	Pigmented nodular adrenocortical disease, primary, 2, 610475	MENDELIOME
PDE1C	99%	98%	100%	99%	?Deafness, autosomal dominant 74, 618140	HEARING IMPAIRMENT MENDELIOME
PDE2A	100%	100%	100%	98%	Intellectual developmental disorder with paroxysmal dyskinesia or seizures, 619150	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS
PDE3A	100%	100%	100%	98%	Hypertension and brachydactyly syndrome, 112410	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
PDE4D	100%	99%	100%	97%	Acrodysostosis 2, w/wo hormone resistance, 614613	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
PDE6A	100%	100%	100%	98%	Retinitis pigmentosa 43, 613810	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PDE6B	100%	100%	100%	99%	Retinitis pigmentosa-40, 613801 Night blindness, congenital stationary, autosomal dominant 2, 163500	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PDE6C	100%	100%	100%	97%	Cone dystrophy 4, 613093	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PDE6D	100%	100%	100%	97%	Joubert syndrome 22, 615665	VISION DISORDERS CILIOPATHIES RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PDE6G	100%	100%	100%	94%	Retinitis pigmentosa 57, 613582	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PDE6H	100%	99%	100%	98%	Retinal cone dystrophy 3, 610024 Achromatopsia 6, 610024	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PDE8B	100%	100%	100%	98%	Pigmented nodular adrenocortical disease, primary, 3, 614190 Striatal degeneration, autosomal dominant, 609161	MOVEMENT DISORDERS MENDELIOME

PDGFB	100%	100%	99%	96%	Meningioma, SIS-related, 607174 Basal ganglia calcification, idiopathic, 5, 615483 Dermatofibrosarcoma protuberans, 607907	MOVEMENT DISORDERS SKIN DISORDERS MENDELIOME PARKINSON TUMOR
PDGFRA	100%	100%	100%	99%	Gastrointestinal stromal tumor/GIST-plus syndrome, somatic or familial, 175510 Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685	VISION DISORDERS MENDELIOME TUMOR
PDGFRB	100%	100%	100%	99%	Premature aging syndrome, Penttinen type, 601812 Kosaki overgrowth syndrome, 616592 Myofibromatosis, infantile, 1, 228550 Basal ganglia calcification, idiopathic, 4, 615007	MOVEMENT DISORDERS SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PARKINSON
PDGFRL	100%	100%	100%	99%	Hepatocellular cancer, somatic, 114550 Colorectal cancer, somatic, 114500	MENDELIOME
PDHA1	99%	97%	97%	75%	Pyruvate dehydrogenase E1-alpha deficiency, 312170	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS
PDHA2	100%	100%	100%	99%	Spermatogenic failure 70, 619828	MALE INFERTILITY MENDELIOME
PDHB	100%	100%	100%	98%	Pyruvate dehydrogenase E1-beta deficiency, 614111	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
PDHX	100%	99%	99%	97%	Lacticacidemia due to PDX1 deficiency, 245349	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
PDK1	100%	100%	100%	97%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
PDK2	100%	100%	100%	98%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
PDK3	100%	100%	98%	75%	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905	NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS

PDK4	100%	100%	100%	98%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
PDLIM3	100%	100%	100%	99%	No OMIM disease ID	HEART MENDELIOME
PDLIM5	99%	97%	100%	99%	No OMIM disease ID	HEART MENDELIOME
PDP1	100%	100%	100%	99%	Pyruvate dehydrogenase phosphatase deficiency, 608782	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
PDSS1	100%	100%	100%	97%	Coenzyme Q10 deficiency, primary, 2, 614651	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
PDSS2	100%	100%	100%	98%	Coenzyme Q10 deficiency, primary, 3, 614652	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
PDX1	100%	100%	100%	97%	Pancreatic agenesis 1, 260370 MODY, type IV, 606392	EPILEPSY MENDELIOME PRE CONCEPTION SCREENING
PDXK	99%	97%	100%	98%	Neuropathy, hereditary motor and sensory, type VIC, with optic atrophy, 618511	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING
PDYN	100%	100%	100%	98%	Spinocerebellar ataxia 23, 610245	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME
PDZD7	100%	99%	100%	98%	Deafness, autosomal recessive 57, 618003 Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
PDZD8	100%	100%	99%	95%	Intellectual developmental disorder with autism and dysmorphic facies, 620021	INTELLECTUAL DISABILITY MENDELIOME

PEPD	100%	100%	100%	99%	Prolidase deficiency, 170100	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PER2	100%	100%	100%	99%	?Advanced sleep phase syndrome, familial, 1, 604348	MENDELIOME
PER3	100%	100%	100%	99%	?Advanced sleep phase syndrome, familial, 3, 616882	MENDELIOME
PERCC1	100%	100%	100%	99%	Diarrhea 11, malabsorptive, congenital, 618662	MENDELIOME
PERP	100%	100%	100%	98%	Erythrokeratodermia variabilis et progressiva 7, 619209 Olmsted syndrome 2, 619208	SKIN DISORDERS MENDELIOME
PET100	100%	100%	100%	99%	Mitochondrial complex IV deficiency, nuclear type 12, 619055	VISION DISORDERS HEARING IMPAIRMENT EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
PET117	100%	100%	100%	94%	?Mitochondrial complex IV deficiency, nuclear type 19, 619063	MENDELIOME MITOCHONDRIAL DISORDERS
PEX1	100%	100%	100%	98%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100	VISION DISORDERS HEARING IMPAIRMENT EPILEPSY NEUROPATHIES LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PEX10	100%	100%	100%	99%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871	MOVEMENT DISORDERS EPILEPSY NEUROPATHIES LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PEX11B	100%	100%	100%	96%	Peroxisome biogenesis disorder 14B, 614920	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

PEX12	100%	100%	100%	98%	Peroxisome biogenesis disorder 3B, 266510 Peroxisome biogenesis disorder 3A (Zellweger), 614859	EPILEPSY LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PEX13	100%	100%	100%	97%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885	EPILEPSY LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PEX14	100%	100%	100%	99%	Peroxisome biogenesis disorder 13A (Zellweger), 614887	EPILEPSY LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PEX16	100%	100%	100%	99%	Peroxisome biogenesis disorder 8B, 614877 Peroxisome biogenesis disorder 8A (Zellweger), 614876	EPILEPSY NEUROPATHIES PRIMARY IMMUNODEFICIENCY LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PEX19	100%	100%	100%	99%	Peroxisome biogenesis disorder 12A (Zellweger), 614886	EPILEPSY LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PEX2	100%	100%	100%	98%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867	MOVEMENT DISORDERS VISION DISORDERS LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

PEX26	100%	100%	100%	98%	Peroxisome biogenesis disorder 7B, 614873 Peroxisome biogenesis disorder 7A (Zellweger), 614872	VISION DISORDERS HEARING IMPAIRMENT EPILEPSY LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PEX3	100%	100%	100%	97%	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370	EPILEPSY LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PEX5	100%	100%	100%	98%	Peroxisome biogenesis disorder 2B, 202370 Peroxisome biogenesis disorder 2A (Zellweger), 214110 Rhizomelic chondrodysplasia punctata, type 5, 616716	EPILEPSY HEART SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PEX6	100%	100%	100%	97%	Peroxisome biogenesis disorder 4B, 614863 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Heimler syndrome 2, 616617	VISION DISORDERS HEARING IMPAIRMENT EPILEPSY SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

PEX7	91%	91%	100%	99%	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879	MOVEMENT DISORDERS VISION DISORDERS SKIN DISORDERS HEART NEUROPATHIES SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PFKM	100%	100%	100%	99%	Glycogen storage disease VII, 232800	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
PFN1	100%	100%	100%	98%	Amyotrophic lateral sclerosis 18, 614808	ALS MENDELIOME
PGAM2	100%	100%	100%	99%	Glycogen storage disease X, 261670	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
PGAP1	100%	100%	100%	97%	Neurodevelopmental disorder with dysmorphic features, spasticity, and brain abnormalities, 615802	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PGAP2	100%	100%	100%	98%	Hyperphosphatasia with impaired intellectual development syndrome 3, 614207	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PGAP3	100%	100%	100%	99%	Hyperphosphatasia with impaired intellectual development syndrome 4, 615716	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PGK1	100%	99%	98%	73%	Phosphoglycerate kinase 1 deficiency, 300653	VISION DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MUSCLE DISORDERS

PGM1	94%	94%	100%	98%	Congenital disorder of glycosylation, type It, 614921	CRANIOFACIAL ANOMALIES HEART METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS MUSCLE DISORDERS
PGM2L1	100%	100%	100%	98%	Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skin abnormalities, 620191	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME
PGM3	100%	100%	100%	99%	Immunodeficiency 23, 615816	PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PHACTR1	100%	100%	100%	98%	Developmental and epileptic encephalopathy 70, 618298	INTELLECTUAL DISABILITY MENDELIOME
PHC1	100%	100%	100%	98%	?Microcephaly 11, primary, autosomal recessive, 615414	MENDELIOME
PHEX	99%	99%	98%	71%	Hypophosphatemic rickets, X-linked dominant, 307800	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME
PHF21A	100%	100%	100%	99%	Intellectual developmental disorder with behavioral abnormalities and craniofacial dysmorphism w/wo seizures, 618725	INTELLECTUAL DISABILITY MENDELIOME
PHF6	100%	100%	98%	75%	Borjeson-Forssman-Lehmann syndrome, 301900	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
PHF8	100%	99%	97%	70%	Intellectual developmental disorder, X-linked syndromic, Siderius type, 300263	INTELLECTUAL DISABILITY MENDELIOME SCHISIS
PHGDH	100%	100%	100%	99%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815	FETAL AKINESIA SKIN DISORDERS EPILEPSY SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS

PHIP	100%	99%	99%	96%	Chung-Jansen syndrome, 617991	INTELLECTUAL DISABILITY MENDELIOME
PHKA1	100%	100%	97%	72%	Muscle glycogenosis, 300559	HEART METABOLIC DISORDERS MENDELIOME MUSCLE DISORDERS
PHKA2	100%	100%	98%	73%	Glycogen storage disease, type IXa2, 306000 Glycogen storage disease, type IXa1, 306000	METABOLIC DISORDERS MENDELIOME
PHKB	100%	100%	100%	98%	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PHKG1	100%	100%	100%	99%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME
PHKG2	100%	100%	99%	98%	Glycogen storage disease IXc, 613027	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PHOX2A	100%	100%	100%	96%	Fibrosis of extraocular muscles, congenital, 2, 602078	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
PHOX2B	100%	100%	99%	96%	Neuroblastoma with Hirschsprung disease, 613013 Central hypoventilation syndrome, congenital, 1, w/wo Hirschsprung disease, 209880	MENDELIOME TUMOR
PHYH	100%	100%	100%	98%	Refsum disease, 266500	MOVEMENT DISORDERS VISION DISORDERS SKIN DISORDERS HEART NEUROPATHIES METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PI4K2A	100%	100%	100%	97%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME
PI4KA	100%	99%	99%	98%	Spastic paraplegia 84, autosomal recessive, 619621 Gastrointestinal defects and immunodeficiency syndrome 2, 619708 Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PI4KB	100%	100%	100%	99%	Deafness, autosomal dominant 87, 620281	HEARING IMPAIRMENT MENDELIOME
PIBF1	100%	100%	100%	95%	Joubert syndrome 33, 617767	CILIOPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

PICALM	100%	100%	100%	98%	Leukemia, acute myeloid, somatic, 601626	MENDELIOME
PIDD1	100%	100%	100%	99%	Intellectual developmental disorder, autosomal recessive 75, with neuropsychiatric features and variant lissencephaly, 619827	INTELLECTUAL DISABILITY MENDELIOME
PIEZO1	100%	100%	100%	99%	Lymphatic malformation 6, 616843 Dehydrated hereditary stomatocytosis w/wo pseudohyperkalemia and/or perinatal edema, 194380	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PIEZO2	100%	100%	100%	98%	Arthrogryposis, distal, type 5, 108145 Arthrogryposis, distal, with impaired proprioception and touch, 617146 Arthrogryposis, distal, type 3, 114300 ?Marden-Walker syndrome, 248700	FETAL AKINESIA NEUROPATHIES HNPD MENDELIOME PRE CONCEPTION SCREENING SCHISIS MUSCLE DISORDERS
PIGA	100%	100%	97%	74%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818 Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072	SKIN DISORDERS EPILEPSY HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS
PIGB	100%	100%	100%	97%	Developmental and epileptic encephalopathy 80, 618580	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PIGC	100%	100%	100%	99%	Glycosylphosphatidylinositol biosynthesis defect 16, 617816	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PIGF	100%	100%	100%	99%	Onychodystrophy, osteodystrophy, impaired intellectual development, and seizures syndrome, 619356	INTELLECTUAL DISABILITY MENDELIOME
PIGG	100%	100%	100%	99%	Neurodevelopmental disorder w/wo hypotonia, seizures, and cerebellar atrophy, 616917	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PIGH	80%	75%	100%	99%	Glycosylphosphatidylinositol biosynthesis defect 17, 618010	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PIGK	100%	100%	99%	97%	Neurodevelopmental disorder with hypotonia and cerebellar atrophy, w/wo seizures, 618879	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

PIGL	100%	100%	100%	98%	CHIME syndrome, 280000	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PIGM	100%	100%	100%	98%	Glycosylphosphatidylinositol deficiency, 610293	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PIGN	100%	99%	100%	98%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080	SKIN DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS
PIGO	100%	100%	100%	99%	Hyperphosphatasia with impaired intellectual development syndrome 2, 614749	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS
PIGP	100%	100%	100%	96%	Developmental and epileptic encephalopathy 55, 617599	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PIGQ	100%	100%	100%	99%	Multiple congenital anomalies-hypotonia-seizures syndrome 4, 618548	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PIGS	100%	100%	100%	99%	Developmental and epileptic encephalopathy 95, 618143	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PIGT	100%	100%	100%	99%	?Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PIGU	100%	100%	100%	99%	Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis, 618590	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

PIGV	100%	100%	100%	99%	Hyperphosphatasia with impaired intellectual development syndrome 1, 239300	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS
PIGW	100%	100%	100%	98%	Glycosylphosphatidylinositol biosynthesis defect 11, 616025	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PIGY	100%	100%	100%	99%	Hyperphosphatasia with impaired intellectual development syndrome 6, 616809	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PIH1D3	100%	100%	97%	69%	Ciliary dyskinesia, primary, 36, X-linked, 300991	CILIOPATHIES MALE INFERTILITY MENDELIOME
PIK3C2A	100%	100%	100%	98%	Oculoskeletodental syndrome, 618440	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING
PIK3CA	100%	100%	100%	98%	CLOVE syndrome, somatic, 612918 Hepatocellular carcinoma, somatic, 114550 Breast cancer, somatic, 114480 Cerebral cavernous malformations 4, somatic, 619538 Ovarian cancer, somatic, 167000 Colorectal cancer, somatic, 114500 Macrodactyly, somatic, 155500 CLAPO syndrome, somatic, 613089 Keratosis, seborrheic, somatic, 182000 Nevus, epidermal, somatic, 162900 Gastric cancer, somatic, 613659 Nonsmall cell lung cancer, somatic, 211980 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Cowden syndrome 5, 615108	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME TUMOR
PIK3CD	100%	100%	100%	98%	Immunodeficiency 14A, autosomal dominant, 615513 Immunodeficiency 14B, autosomal recessive, 619281 ?Roifman-Chitayat syndrome, digenic, 613328	PRIMARY IMMUNODEFICIENCY MENDELIOME
PIK3CG	100%	100%	100%	99%	Immunodeficiency 97 with autoinflammation, 619802	PRIMARY IMMUNODEFICIENCY MENDELIOME

PIK3R1	100%	100%	100%	98%	Immunodeficiency 36, 616005 ?Agammaglobulinemia 7, autosomal recessive, 615214 SHORT syndrome, 269880	PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PIK3R2	100%	100%	100%	97%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME
PIK3R5	100%	100%	100%	99%	Ataxia-oculomotor apraxia 3, 615217	MOVEMENT DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PIKFYVE	100%	100%	100%	98%	Corneal fleck dystrophy, 121850	VISION DISORDERS METABOLIC DISORDERS MENDELIOME
PINK1	100%	100%	100%	98%	Parkinson disease 6, early onset, 605909	MENDELIOME PARKINSON PRE CONCEPTION SCREENING
PIP5K1C	100%	100%	100%	98%	Lethal congenital contractural syndrome 3, 611369	FETAL AKINESIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
PISD	100%	100%	100%	99%	Liberfarb syndrome, 618889	HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS
PITPNM3	100%	100%	99%	97%	Cone-rod dystrophy 5, 600977	MENDELIOME
PITRM1	100%	100%	100%	99%	Spinocerebellar ataxia, autosomal recessive 30, 619405	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS
PITX1	100%	100%	100%	96%	Clubfoot, congenital, w/wo deficiency of long bones and/or mirror-image polydactyly, 119800	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME

PITX2	100%	100%	100%	98%	Ring dermoid of cornea, 180550 Axenfeld-Rieger syndrome, type 1, 180500 Anterior segment dysgenesis 4, 137600	VISION DISORDERS CRANIOFACIAL ANOMALIES SKIN DISORDERS HEART SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
PITX3	100%	100%	100%	96%	Cataract 11, multiple types, 610623 Anterior segment dysgenesis 1, multiple subtypes, 107250 Cataract 11, syndromic, autosomal recessive, 610623	VISION DISORDERS MENDELIOME
PIWIL2	100%	100%	100%	99%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
PJA1	100%	99%	95%	60%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
PJVK	100%	100%	100%	98%	Deafness, autosomal recessive 59, 610220	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
PKD1	99%	99%	100%	98%	Polycystic kidney disease 1, 173900	CILIOPATHIES LIVER DISORDERS MALE INFERTILITY RENAL DISORDERS MENDELIOME
PKD1L1	100%	100%	100%	98%	Heterotaxy, visceral, 8, autosomal, 617205	CONGENITAL HEART DISEASE CILIOPATHIES HEART MENDELIOME PRE CONCEPTION SCREENING
PKD2	100%	100%	99%	93%	Polycystic kidney disease 2, 613095	CILIOPATHIES LIVER DISORDERS RENAL DISORDERS MENDELIOME
PKDCC	100%	100%	97%	77%	Rhizomelic limb shortening with dysmorphic features, 618821	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
PKHD1	100%	100%	100%	98%	Polycystic kidney disease 4, w/wo hepatic disease, 263200	CILIOPATHIES LIVER DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PKLR	100%	100%	100%	99%	Adenosine triphosphate, elevated, of erythrocytes, 102900 Pyruvate kinase deficiency, 266200	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING

PKP1	100%	100%	100%	99%	Ectodermal dysplasia/skin fragility syndrome, 604536	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PKP2	99%	99%	99%	98%	Arrhythmogenic right ventricular dysplasia 9, 609040	ARRHYTHMOGENIC CARDIOMYOPATHY (ACM/ARVC) HEART MENDELIOME ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS
PLA2G4A	100%	100%	100%	98%	Gastrointestinal ulceration, recurrent, with dysfunctional platelets, 618372	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME
PLA2G5	100%	100%	100%	98%	No OMIM disease ID	VISION DISORDERS METABOLIC DISORDERS MENDELIOME
PLA2G6	100%	99%	100%	99%	Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217 Infantile neuroaxonal dystrophy 1, 256600	MOVEMENT DISORDERS EPILEPSY NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PARKINSON PRE CONCEPTION SCREENING
PLA2G7	100%	100%	100%	96%	Platelet-activating factor acetylhydrolase deficiency, 614278	HEMOSTATIC/THROMBOTIC DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PLAA	100%	100%	100%	99%	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PLAG1	100%	100%	100%	99%	Adenomas, salivary gland pleomorphic, somatic, 181030 Silver-Russell syndrome 4, 618907	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
PLAT	100%	100%	100%	99%	No OMIM disease ID	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME
PLAU	100%	100%	100%	98%	Quebec platelet disorder, 601709	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME
PLCB1	100%	100%	100%	97%	Developmental and epileptic encephalopathy 12, 613722	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

PLCB3	100%	100%	100%	98%	Spondylometaphyseal dysplasia with corneal dystrophy, 618961	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
PLCB4	100%	99%	100%	98%	Auriculocondylar syndrome 2, 614669	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS
PLCD1	100%	100%	100%	99%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PLCE1	100%	99%	100%	98%	Nephrotic syndrome, type 3, 610725	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PLCG2	100%	100%	100%	99%	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS MENDELIOME
PLCZ1	100%	100%	100%	97%	Spermatogenic failure 17, 617214	MALE INFERTILITY MENDELIOME
PLD1	100%	100%	100%	98%	Cardiac valvular dysplasia 1, 212093	CONGENITAL HEART DISEASE HEART MENDELIOME PRE CONCEPTION SCREENING
PLD3	100%	100%	100%	99%	?Spinocerebellar ataxia 46, 617770	NEUROPATHIES MENDELIOME
PLEC	100%	100%	100%	99%	?Epidermolysis bullosa simplex 5D, generalized intermediate, autosomal recessive, 616487 Epidermolysis bullosa simplex 5B, with muscular dystrophy, 226670 Epidermolysis bullosa simplex 5C, with pyloric atresia, 612138 Epidermolysis bullosa simplex 5A, Ogna type, 131950 Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
PLEKHG2	100%	100%	100%	98%	Leukodystrophy and acquired microcephaly w/wo dystonia, 616763	MENDELIOME PRE CONCEPTION SCREENING
PLEKHG5	100%	100%	100%	99%	Spinal muscular atrophy, distal, autosomal recessive, 4, 611067 Charcot-Marie-Tooth disease, recessive intermediate C, 615376	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING

PLEKHM1	100%	100%	100%	99%	?Osteopetrosis, autosomal recessive 6, 611497 Osteopetrosis, autosomal dominant 3, 618107	PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
PLEKHM2	100%	100%	99%	97%	No OMIM disease ID	HEART MENDELIOME
PLG	100%	100%	100%	98%	Dysplasminogenemia, 217090 Angioedema, hereditary, 4, 619360 Plasminogen deficiency, type I, 217090	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
PLIN1	100%	100%	100%	98%	Lipodystrophy, familial partial, type 4, 613877	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME
PLK1	100%	100%	100%	97%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
PLK4	100%	100%	100%	98%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171	VISION DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MALE INFERTILITY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PLN	100%	100%	100%	98%	Cardiomyopathy, dilated, 1P, 609909 Cardiomyopathy, hypertrophic, 18, 613874	ARRHYTHMOGENIC CARDIOMYOPATHY (ACM/ARVC) DILATED CARDIOMYOPATHY HEART HYPERTROPHIC CARDIOMYOPATHY MENDELIOME ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS
PLOD1	100%	100%	100%	98%	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400	FETAL AKINESIA ANEURYSM SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING

PLOD2	100%	100%	99%	97%	Bruck syndrome 2, 609220	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PLOD3	100%	100%	100%	98%	Lysyl hydroxylase 3 deficiency, 612394	SKIN DISORDERS HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PLP1	99%	98%	98%	70%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
PLPBP	100%	100%	100%	99%	Epilepsy, early-onset, vitamin B6-dependent, 617290	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
PLPP6	100%	100%	100%	94%	No OMIM disease ID	MENDELIOME
PLS1	100%	99%	100%	98%	Deafness, autosomal dominant 76, 618787	HEARING IMPAIRMENT MENDELIOME
PLS3	96%	96%	97%	71%	Bone mineral density QTL18, osteoporosis, 300910	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
PLVAP	100%	100%	100%	99%	Diarrhea 10, protein-losing enteropathy type, 618183	MENDELIOME PRE CONCEPTION SCREENING
PLXNA1	100%	100%	100%	99%	Dworschak-Punetha neurodevelopmental syndrome, 619955	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM INTELLECTUAL DISABILITY MENDELIOME
PLXNA2	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
PLXND1	100%	100%	100%	98%	No OMIM disease ID	CONGENITAL HEART DISEASE HEART INTELLECTUAL DISABILITY MENDELIOME
PMEPA1	100%	99%	99%	93%	No OMIM disease ID	ANEURYSM MENDELIOME

PMFBP1	100%	100%	100%	98%	Spermatogenic failure 31, 618112	CILIOPATHIES MALE INFERTILITY MENDELIOME
PML	100%	100%	100%	98%	Leukemia, acute promyelocytic, PML/RARA type,	MENDELIOME
PMM2	100%	100%	100%	98%	Congenital disorder of glycosylation, type Ia, 212065	MOVEMENT DISORDERS EPILEPSY HEART NEUROPATHIES PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY
PMP2	100%	100%	100%	99%	Charcot-Marie-Tooth disease, demyelinating, type 1G, 618279	NEUROPATHIES MENDELIOME
PMP22	100%	100%	100%	99%	Charcot-Marie-Tooth disease, type 1A, 118220 Roussy-Levy syndrome, 180800 Charcot-Marie-Tooth disease, type 1E, 118300 ?Neuropathy, inflammatory demyelinating, 139393 Neuropathy, recurrent, with pressure palsies, 162500 Dejerine-Sottas disease, 145900	MOVEMENT DISORDERS NEUROPATHIES HNPD MENDELIOME
PMPCA	100%	100%	100%	99%	Spinocerebellar ataxia, autosomal recessive 2, 213200	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
PMPCB	100%	100%	100%	98%	Multiple mitochondrial dysfunctions syndrome 6, 617954	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
PMS2	100%	100%	99%	95%	Lynch syndrome 4, 614337 Mismatch repair cancer syndrome 4, 619101	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING TUMOR
PMS2CL	NC	NC	NC	NC	No OMIM disease ID	MENDELIOME TUMOR
PMVK	100%	100%	100%	98%	Porokeratosis 1, multiple types, 175800	SKIN DISORDERS MENDELIOME

PNKD	100%	100%	100%	97%	Paroxysmal nonkinesigenic dyskinesia 1, 118800	MOVEMENT DISORDERS MENDELIOME
PNKP	100%	100%	100%	98%	?Charcot-Marie-Tooth disease, type 2B2, 605589 Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402	MOVEMENT DISORDERS EPILEPSY NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PNLDC1	100%	100%	100%	98%	Spermatogenic failure 57, 619528	MALE INFERTILITY MENDELIOME
PNLIP	100%	100%	100%	98%	?Pancreatic lipase deficiency, 614338	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PNMT	100%	100%	100%	97%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME
PNP	100%	100%	100%	99%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179	PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)
PNPLA1	100%	99%	100%	97%	Ichthyosis, congenital, autosomal recessive 10, 615024	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PNPLA2	100%	100%	100%	99%	Neutral lipid storage disease with myopathy, 610717	SKIN DISORDERS HEART METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
PNPLA6	100%	100%	100%	99%	Spastic paraplegia 39, autosomal recessive, 612020 Oliver-McFarlane syndrome, 275400 ?Laurence-Moon syndrome, 245800 Boucher-Neuhauser syndrome, 215470	MOVEMENT DISORDERS VISION DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

PNPLA8	100%	100%	100%	97%	?Mitochondrial myopathy with lactic acidosis, 251950	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
PNPO	100%	100%	100%	99%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090	EPILEPSY METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PNPT1	100%	100%	100%	98%	Spinocerebellar ataxia 25, 608703 Deafness, autosomal recessive 70, w/wo adult-onset neurodegeneration, 614934 Combined oxidative phosphorylation deficiency 13, 614932	HEARING IMPAIRMENT NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
POC1A	100%	100%	100%	99%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813	CILIOPATHIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
POC1B	100%	100%	100%	98%	Cone-rod dystrophy 20, 615973	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
POC5	100%	100%	100%	98%	No OMIM disease ID	VISION DISORDERS MENDELIOME
PODXL	94%	93%	99%	95%	No OMIM disease ID	RENAL DISORDERS MENDELIOME
POF1B	100%	99%	97%	74%	?Premature ovarian failure 2B, 300604	MENDELIOME
POFUT1	100%	100%	100%	98%	Dowling-Degos disease 2, 615327	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME
POGLUT1	100%	100%	100%	98%	Dowling-Degos disease 4, 615696 Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
POGZ	100%	100%	100%	99%	White-Sutton syndrome, 616364	INTELLECTUAL DISABILITY MENDELIOME
POLA1	99%	99%	97%	71%	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 Van Esch-O'Driscoll syndrome, 301030	PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME

POLD1	100%	100%	100%	99%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381	SKIN DISORDERS HEARING IMPAIRMENT PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS MENDELIOME TUMOR
POLE	100%	100%	100%	99%	FILS syndrome, 615139 IMAGE-I syndrome, 618336	DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING TUMOR
POLE2	100%	100%	100%	98%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
POLG	100%	100%	100%	99%	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Progressive external ophthalmoplegia, autosomal recessive 1, 258450	MOVEMENT DISORDERS DISORDERS OF SEX DEVELOPMENT EPILEPSY HYPOGONADOTROPIC HYPOGONADISM NEUROPATHIES LIVER DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PARKINSON PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY MUSCLE DISORDERS
POLG2	100%	100%	100%	97%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131 ?Mitochondrial DNA depletion syndrome 16 (hepatic type), 618528 ?Mitochondrial DNA depletion syndrome 16B (neuroophthalmic type), 619425	VISION DISORDERS NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS MUSCLE DISORDERS
POLH	100%	100%	100%	99%	Xeroderma pigmentosum, variant type, 278750	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING TUMOR
POLL	100%	100%	100%	99%	No OMIM disease ID	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME

POLR1A	100%	100%	100%	99%	Acrofacial dysostosis, Cincinnati type, 616462	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS
POLR1B	100%	100%	100%	99%	Treacher-Collins syndrome 4, 618939	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
POLR1C	83%	83%	100%	99%	Leukodystrophy, hypomyelinating, 11, 616494 Treacher Collins syndrome 3, 248390	MOVEMENT DISORDERS CRANIOFACIAL ANOMALIES SKIN DISORDERS HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS
POLR1D	100%	100%	100%	98%	Treacher Collins syndrome 2, 613717	CRANIOFACIAL ANOMALIES SKIN DISORDERS HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS
POLR2A	100%	100%	100%	99%	Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS
POLR3A	100%	100%	100%	98%	Wiedemann-Rautenstrauch syndrome, 264090 Leukodystrophy, hypomyelinating, 7, w/wo oligodontia and/or hypogonadotropic hypogonadism, 607694	MOVEMENT DISORDERS SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

POLR3B	100%	99%	100%	98%	Leukodystrophy, hypomyelinating, 8, w/wo oligodontia and/or hypogonadotropic hypogonadism, 614381 Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742	MOVEMENT DISORDERS SKIN DISORDERS NEUROPATHIES SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
POLR3F	100%	100%	100%	99%	?Immunodeficiency 101 (varicella zoster virus-specific), 619872	PRIMARY IMMUNODEFICIENCY MENDELIOME
POLR3GL	100%	100%	100%	99%	Short stature, oligodontia, dysmorphic facies, and motor delay, 619234	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
POLR3K	100%	100%	100%	99%	Leukodystrophy, hypomyelinating, 21, 619310	MENDELIOME
POLRMT	100%	100%	100%	99%	Combined oxidative phosphorylation deficiency 55, 619743	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS
POMC	100%	100%	100%	99%	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734	SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING
POMGNT1	100%	100%	100%	99%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 3, 613151 Retinitis pigmentosa 76, 617123 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280	VISION DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
POMGNT2	100%	100%	100%	99%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 8, 614830 Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
POMK	100%	100%	100%	99%	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS

POMP	100%	100%	100%	96%	Proteasome-associated autoinflammatory syndrome 2, 618048 Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
POMT1	100%	100%	100%	98%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 1, 613155	HEART METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS MUSCLE DISORDERS
POMT2	100%	100%	100%	96%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 2, 613156	HEART METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
POP1	100%	100%	100%	99%	Anauxetic dysplasia 2, 617396	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
POPDC3	100%	100%	100%	99%	Muscular dystrophy, limb-girdle, autosomal recessive 26, 618848	MENDELIOME
POR	100%	100%	100%	99%	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571	CRANIOFACIAL ANOMALIES DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
PORCN	100%	99%	98%	72%	Focal dermal hypoplasia, 305600	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS
POT1	100%	100%	99%	98%	No OMIM disease ID	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS DYSKERATOSIS CONGENITA PRIMARY IMMUNODEFICIENCY MELA2 MENDELIOME TUMOR

POU1F1	100%	100%	100%	98%	Pituitary hormone deficiency, combined or isolated, 1, 613038	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
POU2AF1	100%	100%	100%	98%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
POU3F3	99%	97%	94%	56%	Snijders Blok-Fisher syndrome, 618604	INTELLECTUAL DISABILITY MENDELIOME
POU3F4	100%	100%	97%	69%	Deafness, X-linked 2, 304400	HEARING IMPAIRMENT MENDELIOME
POU4F1	94%	91%	98%	78%	Ataxia, intention tremor, and hypotonia syndrome, childhood-onset, 619352	MENDELIOME
POU4F3	100%	100%	100%	99%	Deafness, autosomal dominant 15, 602459	HEARING IMPAIRMENT MENDELIOME
POU6F2	100%	100%	100%	97%	No OMIM disease ID	MENDELIOME TUMOR
PPA2	100%	99%	100%	96%	?Sudden cardiac failure, alcohol-induced, 617223 Sudden cardiac failure, infantile, 617222	HEART MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
PPARG	99%	99%	100%	99%	Insulin resistance, severe, digenic, 604367 Lipodystrophy, familial partial, type 3, 604367 Obesity, severe, 601665 Carotid intimal medial thickness 1, 609338	METABOLIC DISORDERS MENDELIOME
PPCDC	100%	100%	100%	99%	No OMIM disease ID	HEART METABOLIC DISORDERS MENDELIOME
PPCS	100%	100%	100%	98%	Cardiomyopathy, dilated, 2C, 618189	HEART METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
PPFIBP1	100%	100%	100%	98%	Neurodevelopmental disorder with seizures, microcephaly, and brain abnormalities, 620024	INTELLECTUAL DISABILITY MENDELIOME
PPIB	100%	100%	100%	98%	Osteogenesis imperfecta, type IX, 259440	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
PPIL1	100%	100%	100%	98%	Pontocerebellar hypoplasia, type 14, 619301	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

PPIP5K2	100%	100%	100%	98%	Deafness, autosomal recessive 100, 618422	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
PPM1D	100%	100%	100%	98%	Breast cancer, somatic, 114480 Jansen de Vries syndrome, 617450	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME TUMOR
PPM1K	100%	100%	100%	99%	?Maple syrup urine disease, mild variant, 615135	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PPOX	100%	100%	100%	98%	Porphyria variegata, 176200	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME
PPP1CB	100%	100%	100%	98%	Noonan syndrome-like disorder with loose anagen hair 2, 617506	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME AND RASOPATHY
PPP1R12A	99%	99%	100%	98%	Genitourinary and/or/brain malformation syndrome, 618820	DISORDERS OF SEX DEVELOPMENT INTELLECTUAL DISABILITY MENDELIOME
PPP1R13L	100%	99%	99%	95%	No OMIM disease ID	CONGENITAL HEART DISEASE HEART MENDELIOME
PPP1R15B	100%	100%	100%	98%	Microcephaly, short stature, and impaired glucose metabolism 2, 616817	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PPP1R21	100%	100%	100%	98%	Neurodevelopmental disorder with hypotonia, facial dysmorphism, and brain abnormalities, 619383	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PPP1R3A	100%	100%	100%	98%	Insulin resistance, severe, digenic, 125853	MENDELIOME
PPP2CA	100%	100%	100%	98%	Neurodevelopmental disorder and language delay w/wo structural brain abnormalities, 618354	INTELLECTUAL DISABILITY MENDELIOME
PPP2R1A	93%	93%	100%	99%	Intellectual developmental disorder, autosomal dominant 36, 616362	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
PPP2R1B	100%	100%	100%	98%	Lung cancer, somatic, 211980	MENDELIOME
PPP2R2B	100%	100%	100%	99%	Spinocerebellar ataxia 12, 604326	MENDELIOME

PPP2R3C	100%	100%	100%	98%	Spermatogenic failure 36, 618420 Myoectodermal gonadal dysgenesis syndrome, 618419	DISORDERS OF SEX DEVELOPMENT INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PPP2R5B	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
PPP2R5C	100%	100%	99%	98%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
PPP2R5D	100%	100%	100%	98%	Intellectual developmental disorder, autosomal dominant 35, 616355	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
PPP3CA	100%	99%	100%	98%	Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265 Developmental and epileptic encephalopathy 91, 617711	INTELLECTUAL DISABILITY MENDELIOME
PPT1	90%	90%	100%	97%	Ceroid lipofuscinosis, neuronal, 1, 256730	MOVEMENT DISORDERS VISION DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PQBP1	100%	100%	97%	69%	Renpenning syndrome, 309500	SKIN DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME SCHISIS
PRCC	100%	100%	100%	98%	Renal cell carcinoma, papillary, 605074	MENDELIOME
PRCD	100%	100%	100%	94%	Retinitis pigmentosa 36, 610599	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PRDM10	100%	100%	100%	99%	No OMIM disease ID	SKIN DISORDERS MENDELIOME TUMOR
PRDM12	95%	92%	100%	93%	Neuropathy, hereditary sensory and autonomic, type VIII, 616488	NEUROPATHIES HNPD MENDELIOME PRE CONCEPTION SCREENING
PRDM13	100%	100%	100%	97%	Pontocerebellar hypoplasia, type 17, 619909 Cerebellar dysfunction, impaired intellectual development, and hypogonadotropic hypogonadism, 619761	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

PRDM15	100%	99%	99%	93%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
PRDM16	100%	100%	99%	98%	Left ventricular noncompaction 8, 615373 Cardiomyopathy, dilated, 1LL, 615373	HEART MENDELIOME
PRDM5	100%	100%	100%	98%	Brittle cornea syndrome 2, 614170	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PRDM6	100%	100%	100%	95%	Patent ductus arteriosus 3, 617039	HEART MENDELIOME
PRDM8	100%	100%	99%	86%	?Epilepsy, progressive myoclonic, 10, 616640	MENDELIOME
PRDX1	100%	100%	99%	96%	Methylmalonic aciduria and homocystinuria, cblC type, digenic, 277400	MENDELIOME PRE CONCEPTION SCREENING
PRDX2	100%	100%	100%	99%	No OMIM disease ID	HEREDITARY BONE MARROW FAILURE MENDELIOME
PRDX3	100%	100%	100%	98%	Spinocerebellar ataxia, autosomal recessive 32, 619862 Corneal dystrophy, punctiform and polychromatic pre-Descemet, 619871	VISION DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS
PREPL	100%	100%	100%	97%	Myasthenic syndrome, congenital, 22, 616224	MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
PRF1	100%	100%	100%	99%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Aplastic anemia, 609135 Lymphoma, non-Hodgkin, 605027	MOVEMENT DISORDERS HEREDITARY BONE MARROW FAILURE EPILEPSY PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING TUMOR
PRG4	100%	100%	99%	93%	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250	MENDELIOME PRE CONCEPTION SCREENING
PRICKLE1	100%	100%	100%	98%	Epilepsy, progressive myoclonic 1B, 612437	MOVEMENT DISORDERS EPILEPSY MENDELIOME PRE CONCEPTION SCREENING
PRICKLE2	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
PRIMPOL	100%	100%	100%	97%	Myopia 22, autosomal dominant, 615420	VISION DISORDERS MENDELIOME
PRKAA1	100%	100%	100%	97%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
PRKACA	100%	99%	99%	95%	Cushing syndrome, ACTH-independent adrenal, somatic, 615830 Cardioacrofacial dysplasia 1, 619142	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME

PRKACB	99%	99%	100%	97%	Cardioacrofacial dysplasia 2, 619143	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
PRKACG	100%	100%	100%	96%	?Bleeding disorder, platelet-type, 19, 616176	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME
PRKAG2	100%	100%	100%	96%	Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200 Cardiomyopathy, hypertrophic 6, 600858	HEART HYPERTROPHIC CARDIOMYOPATHY METABOLIC DISORDERS MENDELIOME
PRKAR1A	100%	100%	100%	98%	Pigmented nodular adrenocortical disease, primary, 1, 610489 Acrodysostosis 1, w/wo hormone resistance, 101800 Carney complex, type 1, 160980 Myxoma, intracardiac, 255960 Adrenocortical tumor, somatic,	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME TUMOR
PRKAR1B	100%	100%	100%	99%	Marbach-Schaaf neurodevelopmental syndrome, 619680	INTELLECTUAL DISABILITY MENDELIOME
PRKCA	100%	100%	100%	97%	Pituitary tumor, invasive,	MENDELIOME
PRKCB	100%	99%	100%	97%	No OMIM disease ID	HEARING IMPAIRMENT MENDELIOME
PRKCD	100%	100%	100%	99%	Autoimmune lymphoproliferative syndrome, type III, 615559	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
PRKCG	100%	100%	100%	97%	Spinocerebellar ataxia 14, 605361	MOVEMENT DISORDERS MENDELIOME
PRKCSH	100%	100%	100%	98%	Polycystic liver disease 1, 174050	LIVER DISORDERS METABOLIC DISORDERS MENDELIOME
PRKD1	100%	100%	99%	94%	Congenital heart defects and ectodermal dysplasia, 617364	CONGENITAL HEART DISEASE HEART MENDELIOME
PRKDC	100%	100%	100%	98%	Immunodeficiency 26, w/wo neurologic abnormalities, 615966	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)
PRKG1	95%	95%	99%	97%	Aortic aneurysm, familial thoracic 8, 615436	ANEURYSM MENDELIOME
PRKG2	100%	99%	100%	98%	Spondylometaphyseal dysplasia, Pagnamenta type, 619638 Acromesomelic dysplasia 4, 619636	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME

PRKN	91%	91%	100%	98%	Adenocarcinoma of lung, somatic, 211980 Parkinson disease, juvenile, type 2, 600116 Ovarian cancer, somatic, 167000	MOVEMENT DISORDERS MENDELIOME PARKINSON PRE CONCEPTION SCREENING TUMOR
PRKRA	100%	100%	99%	97%	Dystonia 16, 612067	MOVEMENT DISORDERS MENDELIOME PARKINSON PRE CONCEPTION SCREENING
PRLR	100%	100%	100%	98%	Multiple fibroadenomas of the breast, 615554 Hyperprolactinemia, 615555	MENDELIOME
PRMT7	100%	100%	100%	99%	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PRNP	100%	100%	100%	99%	Spongiform encephalopathy with neuropsychiatric features, 606688 Gerstmann-Straussler disease, 137440 Huntington disease-like 1, 603218 Insomnia, fatal familial, 600072 Cerebral amyloid angiopathy, PRNP-related, 137440 Creutzfeldt-Jakob disease, 123400	NEUROPATHIES MENDELIOME
PROC	100%	100%	100%	99%	Thrombophilia 3 due to protein C deficiency, autosomal dominant, 176860 Thrombophilia 3 due to protein C deficiency, autosomal recessive, 612304	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PRODH	100%	100%	100%	99%	Hyperprolinemia, type I, 239500	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PROK2	100%	100%	100%	98%	Hypogonadotropic hypogonadism 4 w/wo anosmia, 610628	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME
PROKR2	100%	100%	100%	99%	Hypogonadotropic hypogonadism 3 w/wo anosmia, 244200	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS
PROM1	100%	100%	100%	98%	Macular dystrophy, retinal, 2, 608051 Retinitis pigmentosa 41, 612095 Stargardt disease 4, 603786 Cone-rod dystrophy 12, 612657	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING

PROP1	100%	100%	100%	95%	Pituitary hormone deficiency, combined, 2, 262600	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
PRORP	100%	100%	100%	98%	Combined oxidative phosphorylation deficiency 54, 619737	HEARING IMPAIRMENT MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
PROS1	100%	100%	100%	97%	Thrombophilia 5 due to protein S deficiency, autosomal recessive, 614514 Thrombophilia 5 due to protein S deficiency, autosomal dominant, 612336	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PROZ	100%	100%	100%	99%	No OMIM disease ID	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME
PRPF3	100%	100%	100%	98%	Retinitis pigmentosa 18, 601414	VISION DISORDERS MENDELIOME
PRPF31	100%	100%	100%	98%	Retinitis pigmentosa 11, 600138	VISION DISORDERS MENDELIOME
PRPF4	100%	100%	100%	98%	Retinitis pigmentosa 70, 615922	VISION DISORDERS MENDELIOME
PRPF6	100%	100%	100%	99%	Retinitis pigmentosa 60, 613983	VISION DISORDERS MENDELIOME
PRPF8	100%	100%	100%	99%	Retinitis pigmentosa 13, 600059	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME
PRPH2	100%	100%	100%	98%	Macular dystrophy, patterned, 1, 169150 Choroidal dystrophy, central areolar 2, 613105 Retinitis punctata albescens, 136880 Leber congenital amaurosis 18, 608133 Macular dystrophy, vitelliform, 3, 608161 Retinitis pigmentosa 7 and digenic form, 608133	AGE RELATED MACULAR DEGENERATION VISION DISORDERS MENDELIOME
PRPS1	100%	100%	96%	70%	Arts syndrome, 301835 Phosphoribosylpyrophosphate synthetase superactivity, 300661 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Deafness, X-linked 1, 304500 Gout, PRPS-related, 300661	HEARING IMPAIRMENT NEUROPATHIES PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS MUSCLE DISORDERS
PRR11	100%	100%	100%	98%	No OMIM disease ID	VISION DISORDERS MENDELIOME

PRR12	100%	100%	100%	98%	Neuroocular syndrome, 619539	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME
PRRT2	100%	100%	100%	97%	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 Seizures, benign familial infantile, 2, 605751 Episodic kinesigenic dyskinesia 1, 128200	MOVEMENT DISORDERS EPILEPSY MENDELIOME
PRRX1	100%	100%	100%	98%	Agnathia-otocephaly complex, 202650	CRANIOFACIAL ANOMALIES MENDELIOME SCHISIS
PRSS1	100%	100%	100%	93%	Pancreatitis, hereditary, 167800	MENDELIOME TUMOR
PRSS12	100%	100%	100%	99%	Intellectual developmental disorder, autosomal recessive 1, 249500	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PRSS56	100%	100%	100%	99%	Microphthalmia, isolated 6, 613517	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PRUNE1	93%	93%	100%	98%	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PRX	100%	100%	100%	98%	Charcot-Marie-Tooth disease, type 4F, 614895 Dejerine-Sottas disease, 145900	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING
PRY	50%	50%	47%	18%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
PRY2	50%	50%	47%	21%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
PSAP	100%	100%	100%	99%	Combined SAP deficiency, 611721 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900 Gaucher disease, atypical, 610539	MOVEMENT DISORDERS EPILEPSY NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PSAT1	100%	100%	100%	98%	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992	FETAL AKINESIA SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

PSEN1	100%	100%	100%	99%	Pick disease, 172700 Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 Dementia, frontotemporal, 600274 ?Acne inversa, familial, 3, 613737 Cardiomyopathy, dilated, 1U, 613694 Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 Alzheimer disease, type 3, 607822	SKIN DISORDERS MENDELIOME PARKINSON
PSEN2	100%	100%	100%	99%	Alzheimer disease-4, 606889 Cardiomyopathy, dilated, 1V, 613697	MENDELIOME
PSENEN	100%	100%	100%	98%	Acne inversa, familial, 2, w/wo Dowling-Degos disease, 613736	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME
PSIP1	100%	100%	100%	95%	No OMIM disease ID	HEARING IMPAIRMENT MENDELIOME
PSMA3	100%	100%	100%	98%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
PSMB1	100%	100%	100%	98%	?Neurodevelopmental disorder with microcephaly, hypotonia, and absent language, 620038	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
PSMB10	100%	100%	100%	97%	Proteasome-associated autoinflammatory syndrome 5, 619175	MENDELIOME
PSMB4	100%	100%	100%	97%	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
PSMB8	100%	100%	99%	98%	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
PSMB9	100%	100%	100%	98%	?Proteasome-associated autoinflammatory syndrome 3, digenic, 617591	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
PSMC3	100%	100%	100%	99%	?Deafness, cataract, impaired intellectual development, and polyneuropathy, 619354	MENDELIOME
PSMC3IP	100%	100%	100%	99%	Ovarian dysgenesis 3, 614324	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY
PSMC5	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
PSMD12	100%	100%	100%	97%	Stankiewicz-Isidor syndrome, 617516	INTELLECTUAL DISABILITY MENDELIOME
PSMG2	100%	100%	100%	98%	?Proteasome-associated autoinflammatory syndrome 4, 619183	PRIMARY IMMUNODEFICIENCY MENDELIOME

PSPH	100%	100%	100%	98%	Phosphoserine phosphatase deficiency, 614023	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PSTPIP1	100%	100%	100%	99%	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME
PTCD3	100%	100%	100%	98%	?Combined oxidative phosphorylation deficiency 51, 619057	MENDELIOME MITOCHONDRIAL DISORDERS
PTCH1	100%	100%	100%	97%	Basal cell carcinoma, somatic, 605462 Holoprosencephaly 7, 610828 Basal cell nevus syndrome, 109400	CRANIOFACIAL ANOMALIES SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME SCHISIS SONIC HEDGEHOG MEDULLOBLASTOMA TUMOR
PTCH2	100%	100%	100%	99%	Medulloblastoma, somatic, 155255 Basal cell nevus syndrome, 109400 Basal cell carcinoma, somatic, 605462	SKIN DISORDERS MENDELIOME SCHISIS
PTCHD1	100%	99%	98%	69%	No OMIM disease ID	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME
PTDSS1	100%	100%	100%	98%	Lenz-Majewski hyperostotic dwarfism, 151050	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
PTEN	100%	100%	99%	97%	Cowden syndrome 1, 158350 Lhermitte-Duclos disease, 158350 Prostate cancer, somatic, 176807 Macrocephaly/autism syndrome, 605309	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME TUMOR
PTF1A	100%	100%	100%	89%	Pancreatic and cerebellar agenesis, 609069 Pancreatic agenesis 2, 615935	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PTGIS	100%	100%	100%	98%	Hypertension, essential, 145500	METABOLIC DISORDERS MENDELIOME
PTGS1	100%	100%	100%	98%	No OMIM disease ID	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME

PTH	100%	100%	100%	98%	Hypoparathyroidism, familial isolated 1, 146200	MENDELIOME
PTH1R	100%	100%	100%	99%	Metaphyseal chondrodysplasia, Murk Jansen type, 156400 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Chondrodysplasia, Blomstrand type, 215045	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PTHLH	100%	100%	100%	98%	Brachydactyly, type E2, 613382	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
PTPN11	100%	100%	100%	98%	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Leukemia, juvenile myelomonocytic, somatic, 607785	HEREDITARY BONE MARROW FAILURE CONGENITAL HEART DISEASE SKIN DISORDERS HEART HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME AND RASOPATHY TUMOR
PTPN12	100%	100%	100%	97%	Colon cancer, somatic, 114500	MENDELIOME
PTPN14	100%	100%	100%	99%	Choanal atresia and lymphedema, 613611	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PTPN22	100%	100%	100%	98%	No OMIM disease ID	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME
PTPN23	100%	100%	100%	98%	Neurodevelopmental disorder and structural brain anomalies w/wo seizures and spasticity, 618890	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PTPRC	100%	99%	100%	97%	Immunodeficiency 105, severe combined, 619924	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)
PTPRF	100%	100%	100%	99%	?Breasts and/or nipples, aplasia or hypoplasia of, 2, 616001	SKIN DISORDERS MENDELIOME
PTPRJ	100%	100%	99%	98%	Colon cancer, somatic, 114500	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME

PTPRO	99%	99%	100%	98%	Nephrotic syndrome, type 6, 614196	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PTPRQ	91%	91%	100%	98%	Deafness, autosomal dominant 73, 617663 Deafness, autosomal recessive 84A, 613391	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
PTRH2	100%	100%	100%	98%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263	FETAL AKINESIA MOVEMENT DISORDERS HEARING IMPAIRMENT EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS
PTRHD1	100%	100%	100%	97%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
PTS	100%	100%	100%	95%	Hyperphenylalaninemia, BH4-deficient, A, 261640	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PUF60	100%	100%	99%	97%	Verheij syndrome, 615583	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
PUM1	100%	100%	100%	98%	Spinocerebellar ataxia 47, 617931	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
PURA	100%	100%	100%	94%	Neurodevelopmental disorder with neonatal respiratory insufficiency, hypotonia, and feeding difficulties, 616158	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
PUS1	100%	100%	100%	98%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462	IRON DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING

PUS3	100%	100%	100%	99%	Neurodevelopmental disorder with microcephaly and gray sclerae, 617051	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PUS7	100%	100%	100%	98%	Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
PXDN	100%	100%	100%	99%	Anterior segment dysgenesis 7, with sclerocornea, 269400	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PYCR1	100%	100%	100%	99%	Cutis laxa, autosomal recessive, type IIIB, 614438 Cutis laxa, autosomal recessive, type IIB, 612940	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
PYCR2	100%	100%	100%	98%	Leukodystrophy, hypomyelinating, 10, 616420	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
PYGL	100%	100%	100%	99%	Glycogen storage disease VI, 232700	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
PYGM	100%	100%	100%	99%	McArdle disease, 232600	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
PYROXD1	100%	100%	100%	97%	Myopathy, myofibrillar, 8, 617258	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS
PYROXD2	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
QARS1	100%	100%	100%	99%	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

QDPR	100%	100%	100%	97%	Hyperphenylalaninemia, BH4-deficient, C, 261630	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
QRICH1	100%	100%	100%	99%	Ververi-Brady syndrome, 617982	INTELLECTUAL DISABILITY MENDELIOME
QRICH2	100%	100%	100%	99%	Spermatogenic failure 35, 618341	MALE INFERTILITY MENDELIOME
QRSL1	100%	100%	100%	98%	Combined oxidative phosphorylation deficiency 40, 618835	HEART MENDELIOME MITOCHONDRIAL DISORDERS
RAB11B	100%	100%	100%	99%	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807	INTELLECTUAL DISABILITY MENDELIOME
RAB14	100%	100%	99%	96%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
RAB18	100%	100%	100%	98%	Warburg micro syndrome 3, 614222	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
RAB23	100%	100%	100%	97%	Carpenter syndrome, 201000	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
RAB27A	100%	100%	100%	99%	Griscelli syndrome, type 2, 607624	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
RAB28	100%	100%	100%	97%	Cone-rod dystrophy 18, 615374	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
RAB33B	100%	100%	100%	97%	Smith-McCort dysplasia 2, 615222	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING

RAB39B	100%	100%	97%	69%	Intellectual developmental disorder, X-linked 72, 300271 Waisman syndrome, 311510	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
RAB3GAP1	99%	99%	99%	98%	Martsolf syndrome 2, 619420 Warburg micro syndrome 1, 600118	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
RAB3GAP2	100%	100%	100%	98%	Martsolf syndrome 1, 212720 Warburg micro syndrome 2, 614225	MOVEMENT DISORDERS VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
RAB7A	100%	100%	100%	99%	Charcot-Marie-Tooth disease, type 2B, 600882	NEUROPATHIES HNPD MENDELIOME
RAC1	100%	100%	100%	97%	Intellectual developmental disorder, autosomal dominant 48, 617751	INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME AND RASOPATHY
RAC2	100%	100%	100%	99%	Immunodeficiency 73A with defective neutrophil chemotaxix and leukocytosis, 608203 ?Immunodeficiency 73C with defective neutrophil chemotaxis and hypogammaglobulinemia, 618987 Immunodeficiency 73B with defective neutrophil chemotaxis and lymphopenia, 618986	PRIMARY IMMUNODEFICIENCY MENDELIOME SEVERE COMBINED IMMUNODEFICIENCY (SCID)
RAC3	100%	100%	99%	95%	Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
RACGAP1	100%	100%	100%	99%	?Anemia, congenital dyserythropoietic, type IIIb, autosomal recessive, 619789	MENDELIOME
RAD21	100%	100%	100%	98%	Cornelia de Lange syndrome 4, 614701 ?Mungan syndrome, 611376	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME
RAD21L1	100%	100%	100%	97%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
RAD50	100%	100%	100%	97%	Nijmegen breakage syndrome-like disorder, 613078	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING TUMOR

RAD51	89%	89%	100%	99%	Mirror movements 2, 614508 Fanconi anemia, complementation group R, 617244	MOVEMENT DISORDERS HEREDITARY BONE MARROW FAILURE MENDELIOME
RAD51C	100%	100%	100%	97%	Fanconi anemia, complementation group O, 613390	HEREDITARY BONE MARROW FAILURE HEREDITARY BREAST AND OVARIAN CANCER MENDELIOME PRE CONCEPTION SCREENING TUMOR
RAD51D	100%	100%	100%	98%	No OMIM disease ID	 HEREDITARY BREAST AND OVARIAN CANCER MENDELIOME TUMOR
RAD54B	100%	100%	100%	98%	Colon cancer, somatic, 114500 Lymphoma, non-Hodgkin, somatic, 605027	MENDELIOME
RAD54L	100%	100%	100%	98%	Lymphoma, non-Hodgkin, somatic, 605027 Adenocarcinoma, colonic, somatic,	MENDELIOME
RAF1	100%	100%	100%	98%	Cardiomyopathy, dilated, 1NN, 615916 Noonan syndrome 5, 611553 LEOPARD syndrome 2, 611554	CONGENITAL HEART DISEASE SKIN DISORDERS HEART HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME AND RASOPATHY TUMOR
RAG1	100%	100%	100%	99%	Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457 Combined cellular and humoral immune defects with granulomas, 233650 Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)
RAG2	100%	100%	100%	98%	Severe combined immunodeficiency, B cell-negative, 601457 Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)

RAI1	100%	100%	100%	98%	Smith-Magenis syndrome, 182290	SKIN DISORDERS HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME
RALA	100%	100%	100%	98%	Hiatt-Neu-Cooper neurodevelopmental syndrome, 619311	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
RALGAPA1	100%	99%	100%	98%	Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation, 618797	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
RANBP2	100%	100%	100%	97%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
RANGRF	100%	100%	100%	97%	No OMIM disease ID	HEART MENDELIOME
RAP1GDS1	100%	100%	100%	98%	Lymphocytic leukemia, acute T-cell,	MENDELIOME
RAPGEF2	100%	100%	99%	97%	?Epilepsy, familial adult myoclonic, 7, 618075	MENDELIOME
RAPSN	100%	100%	100%	99%	Fetal akinesia deformation sequence 2, 618388 Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326	FETAL AKINESIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
RARB	100%	100%	100%	99%	Microphthalmia, syndromic 12, 615524	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
RARS1	94%	94%	100%	97%	Leukodystrophy, hypomyelinating, 9, 616140	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
RARS2	100%	100%	100%	98%	Pontocerebellar hypoplasia, type 6, 611523	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
RASA1	99%	99%	100%	97%	Capillary malformation-arteriovenous malformation 1, 608354 Basal cell carcinoma, somatic, 605462	MENDELIOME
RASGRP1	100%	100%	100%	99%	Immunodeficiency 64, 618534	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING

RASGRP2	100%	100%	100%	98%	?Bleeding disorder, platelet-type, 18, 615888	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
RAX	100%	100%	100%	98%	Microphthalmia, syndromic 16, 611038	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
RAX2	100%	100%	100%	99%	Retinitis pigmentosa 95, 620102 Cone-rod dystrophy 11, 610381 ?Macular degeneration, age-related, 6, 613757	VISION DISORDERS MENDELIOME
RB1	100%	99%	100%	97%	Small cell cancer of the lung, somatic, 182280 Bladder cancer, somatic, 109800 Retinoblastoma, trilateral, 180200 Osteosarcoma, somatic, 259500 Retinoblastoma, 180200	MENDELIOME TUMOR
RB1CC1	100%	99%	100%	96%	Breast cancer, somatic, 114480	MENDELIOME
RBBP6	100%	100%	100%	96%	No OMIM disease ID	HEREDITARY BONE MARROW FAILURE MENDELIOME
RBBP7	100%	99%	97%	70%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
RBBP8	100%	100%	100%	97%	Seckel syndrome 2, 606744 Jawad syndrome, 251255 Pancreatic carcinoma, somatic,	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
RBCK1	100%	100%	99%	97%	Polyglucosan body myopathy 1 w/wo immunodeficiency, 615895	HEART PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
RBFOX1	100%	99%	100%	97%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
RBFOX2	100%	100%	100%	96%	No OMIM disease ID	CONGENITAL HEART DISEASE HEART MENDELIOME
RBL2	100%	100%	100%	98%	Brunet-Wagner neurodevelopmental syndrome, 619690	MENDELIOME

RBM10	100%	99%	98%	73%	TARP syndrome, 311900	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME SCHISIS
RBM20	100%	100%	100%	99%	Cardiomyopathy, dilated, 1DD, 613172	DILATED CARDIOMYOPATHY HEART MENDELIOME
RBM28	100%	100%	100%	98%	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME
RBM8A	100%	100%	99%	97%	Thrombocytopenia-absent radius syndrome, 274000	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
RBMX	100%	99%	97%	74%	?Intellectual developmental disorder, X-linked syndromic 11, Shashi type, 300238	MENDELIOME
RBMY1A1	50%	50%	49%	44%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
RBMY1B	50%	49%	48%	39%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
RBMY1D	49%	48%	47%	38%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
RBMY1E	50%	49%	48%	40%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
RBMY1F	49%	48%	47%	31%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
RBMY1J	49%	49%	48%	32%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
RBP3	100%	100%	100%	99%	?Retinitis pigmentosa 66, 615233	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
RBP4	100%	100%	100%	98%	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147	VISION DISORDERS SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
RBPJ	100%	100%	100%	98%	Adams-Oliver syndrome 3, 614814	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME

RC3H1	100%	100%	100%	99%	?Immune dysregulation and systemic hyperinflammation syndrome, 618998	PRIMARY IMMUNODEFICIENCY MENDELIOME
RCBTB1	100%	100%	100%	98%	Retinal dystrophy w/wo extraocular anomalies, 617175	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
RD3	100%	100%	100%	99%	Leber congenital amaurosis 12, 610612	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
RDH11	100%	100%	100%	99%	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
RDH12	100%	100%	100%	99%	Leber congenital amaurosis 13, 612712	VISION DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
RDH5	100%	100%	100%	99%	Fundus albipunctatus, 136880	VISION DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
RDX	100%	100%	100%	98%	Deafness, autosomal recessive 24, 611022	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
REC114	100%	100%	100%	99%	Oocyte maturation defect 10, 619176	MENDELIOME
RECQL4	100%	100%	100%	99%	Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2, 268400 RAPADILINO syndrome, 266280	CRANIOFACIAL ANOMALIES SKIN DISORDERS PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING TUMOR
REEP1	100%	100%	100%	98%	?Neuronopathy, distal hereditary motor, type VB, 614751 Spastic paraplegia 31, autosomal dominant, 610250 Spinal muscular atrophy, distal, autosomal recessive, 6, 620011	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME
REEP2	100%	100%	100%	98%	?Spastic paraplegia 72, autosomal dominant, 615625 ?Spastic paraplegia 72, autosomal recessive, 615625	MENDELIOME PRE CONCEPTION SCREENING
REEP6	100%	100%	100%	98%	Retinitis pigmentosa 77, 617304	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING

REL	100%	99%	100%	98%	Immunodeficiency 92, 619652	PRIMARY IMMUNODEFICIENCY MENDELIOME
RELA	100%	100%	100%	99%	?Mucocutaneous ulceration, chronic, 618287	PRIMARY IMMUNODEFICIENCY MENDELIOME
RELB	100%	99%	100%	98%	?Immunodeficiency 53, 617585	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
RELN	99%	99%	100%	99%	Lissencephaly 2 (Norman-Roberts type), 257320	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
RELT	100%	100%	100%	98%	Amelogenesis imperfecta, type IIIC, 618386	MENDELIOME
REN	100%	100%	100%	98%	Renal tubular dysgenesis, 267430 Tubulointerstitial kidney disease, autosomal dominant, 4, 613092	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
REPS1	100%	100%	100%	98%	?Neurodegeneration with brain iron accumulation 7, 617916	MENDELIOME PRE CONCEPTION SCREENING
RERE	100%	99%	99%	95%	Neurodevelopmental disorder w/wo anomalies of the brain, eye, or heart, 616975	INTELLECTUAL DISABILITY MENDELIOME
REST	98%	98%	100%	99%	Deafness, autosomal dominant 27, 612431 Fibromatosis, gingival, 5, 617626	HEARING IMPAIRMENT MENDELIOME TUMOR
RET	100%	100%	100%	99%	Multiple endocrine neoplasia IIA, 171400 Medullary thyroid carcinoma, 155240 Pheochromocytoma, 171300 Multiple endocrine neoplasia IIB, 162300	MENDELIOME TUMOR
RETREG1	100%	100%	100%	95%	Neuropathy, hereditary sensory and autonomic, type IIB, 613115	NEUROPATHIES HNPD MENDELIOME PRE CONCEPTION SCREENING
REV3L	97%	97%	100%	97%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
RFC1	100%	100%	100%	96%	Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome, 614575	MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING
RFT1	100%	100%	100%	98%	Congenital disorder of glycosylation, type In, 612015	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
RFWD3	100%	100%	100%	99%	?Fanconi anemia, complementation group W, 617784	HEREDITARY BONE MARROW FAILURE MENDELIOME PRE CONCEPTION SCREENING

RFX3	99%	98%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
RFX4	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
RFX5	100%	100%	100%	99%	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)
RFX6	100%	100%	100%	98%	Mitchell-Riley syndrome, 615710	LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING
RFX7	100%	100%	99%	96%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
RFXANK	100%	100%	100%	99%	Bare lymphocyte syndrome, type II, complementation group B, 209920	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)
RFXAP	100%	100%	100%	98%	Bare lymphocyte syndrome, type II, complementation group D, 209920	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)
RGR	100%	100%	100%	99%	Retinitis pigmentosa 44, 613769	MENDELIOME PRE CONCEPTION SCREENING
RGS10	100%	100%	100%	98%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
RGS9	100%	100%	100%	99%	Bradyopsia, 608415	VISION DISORDERS MENDELIOME
RGS9BP	100%	100%	100%	99%	Bradyopsia, 608415	VISION DISORDERS MENDELIOME
RHAG	100%	100%	100%	99%	Overhydrated hereditary stomatocytosis, 185000 Anemia, hemolytic, Rh-null, regulator type, 268150	MENDELIOME
RHBDF2	100%	100%	100%	99%	Tylosis with esophageal cancer, 148500	SKIN DISORDERS MENDELIOME TUMOR
RHCE	98%	98%	97%	93%	Rh-null disease, amorph type, 617970	MENDELIOME
RHEB	100%	100%	100%	97%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME

RHO	100%	100%	100%	99%	Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 Retinitis punctata albescens, 136880	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
RHOA	80%	80%	100%	97%	Ectodermal dysplasia with facial dysmorphism and acral, ocular, and brain anomalies, somatic mosaic, 618727	SKIN DISORDERS MENDELIOME
RHOBTB2	100%	100%	100%	98%	Developmental and epileptic encephalopathy 64, 618004	INTELLECTUAL DISABILITY MENDELIOME
RHOG	100%	100%	100%	100%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
RHOH	100%	100%	100%	98%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
RIC1	100%	99%	100%	98%	CATIFA syndrome, 618761	INTELLECTUAL DISABILITY MENDELIOME
RILPL1	100%	100%	100%	99%	Oculopharyngodistal myopathy 4, 619790	MENDELIOME
RIMS1	100%	100%	100%	98%	Cone-rod dystrophy 7, 603649	VISION DISORDERS MENDELIOME
RIMS2	100%	99%	100%	97%	Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
RIN2	100%	100%	100%	98%	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
RINT1	100%	100%	100%	98%	Infantile liver failure syndrome 3, 618641	LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
RIPK1	100%	100%	100%	98%	Immunodeficiency 57 with autoinflammation, 618108 Autoinflammation with episodic fever and lymphadenopathy, 618852	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
RIPK4	100%	100%	100%	99%	CHAND syndrome, 214350 Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650	FETAL AKINESIA CRANIOFACIAL ANOMALIES SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT MENDELIOME PRE CONCEPTION SCREENING SCHISIS
RIPOR2	100%	100%	100%	98%	Deafness, autosomal dominant 21, 607017 ?Deafness, autosomal recessive 104, 616515	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING

RIPPLY2	100%	100%	100%	96%	?Spondylocostal dysostosis 6, 616566	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
RIT1	100%	100%	100%	99%	Noonan syndrome 8, 615355	HEART HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME AND RASOPATHY TUMOR
RLBP1	100%	100%	100%	99%	Bothnia retinal dystrophy, 607475 Newfoundland rod-cone dystrophy, 607476 Retinitis punctata albescens, 136880 Fundus albipunctatus, 136880	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
RLIM	100%	100%	98%	72%	Tonne-Kalscheuer syndrome, 300978	INTELLECTUAL DISABILITY MENDELIOME
RMND1	100%	100%	100%	97%	Combined oxidative phosphorylation deficiency 11, 614922	HEARING IMPAIRMENT INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
RMRP	NC	NC	NC	NC	Anauxetic dysplasia 1, 607095 Metaphyseal dysplasia without hypotrichosis, 250460 Cartilage-hair hypoplasia, 250250	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID) TUMOR
RNASEH1	100%	100%	100%	98%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING

RNASEH2A	100%	100%	100%	99%	Aicardi-Goutieres syndrome 4, 610333	MOVEMENT DISORDERS SKIN DISORDERS EPILEPSY PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
RNASEH2B	91%	91%	100%	97%	Aicardi-Goutieres syndrome 2, 610181	MOVEMENT DISORDERS SKIN DISORDERS EPILEPSY PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
RNASEH2C	100%	100%	100%	97%	Aicardi-Goutieres syndrome 3, 610329	MOVEMENT DISORDERS SKIN DISORDERS EPILEPSY PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
RNASEL	100%	100%	99%	97%	Prostate cancer 1, 601518	MENDELIOME TUMOR
RNASET2	100%	100%	100%	98%	Leukoencephalopathy, cystic, without megalencephaly, 612951	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
RNF113A	100%	99%	95%	66%	Trichothiodystrophy 5, nonphotosensitive, 300953	INTELLECTUAL DISABILITY MENDELIOME
RNF125	100%	100%	100%	99%	Tenorio syndrome, 616260	INTELLECTUAL DISABILITY MENDELIOME
RNF13	100%	100%	100%	98%	Developmental and epileptic encephalopathy 73, 618379	INTELLECTUAL DISABILITY MENDELIOME
RNF139	100%	100%	100%	99%	Renal cell carcinoma, 144700	MENDELIOME
RNF168	100%	100%	100%	98%	RIDDLE syndrome, 611943	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
RNF170	100%	100%	100%	98%	Ataxia, sensory, 1, autosomal dominant, 608984 Spastic paraplegia 85, autosomal recessive, 619686	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME
RNF2	100%	100%	100%	99%	Luo-Schoch-Yamamoto syndrome, 619460	INTELLECTUAL DISABILITY MENDELIOME

RNF212	100%	100%	100%	98%	?Spermatogenic failure 62, 619673 Recombination rate QTL 1, 612042	MALE INFERTILITY MENDELIOME
RNF216	100%	100%	100%	98%	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840	MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING
RNF220	100%	100%	100%	98%	Leukodystrophy, hypomyelinating, 23, with ataxia, deafness, liver dysfunction, and dilated cardiomyopathy, 619688	INTELLECTUAL DISABILITY MENDELIOME
RNF31	100%	100%	100%	99%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
RNF43	100%	100%	100%	99%	Sessile serrated polyposis cancer syndrome, 617108	MENDELIOME TUMOR
RNF6	100%	100%	100%	98%	Esophageal carcinoma, somatic, 133239	MENDELIOME
RNPC3	100%	100%	100%	97%	Pituitary hormone deficiency, combined or isolated, 7, 618160	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
RNU12-2P	NC	NC	NC	NC	No OMIM disease ID	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME
RNU4ATAC	NC	NC	NC	NC	Roifman syndrome, 616651 Lowry-Wood syndrome, 226960 Microcephalic osteodysplastic primordial dwarfism, type I, 210710	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
RNU7-1	NC	NC	NC	NC	Aicardi-Goutieres syndrome 9, 619487	PRIMARY IMMUNODEFICIENCY MENDELIOME
ROBO1	100%	99%	100%	99%	No OMIM disease ID	HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME
ROBO2	100%	100%	100%	98%	Vesicoureteral reflux 2, 610878	RENAL DISORDERS MENDELIOME
ROBO3	100%	100%	100%	98%	Gaze palsy, familial horizontal, with progressive scoliosis, 1, 607313	MENDELIOME PRE CONCEPTION SCREENING
ROBO4	100%	100%	100%	98%	Aortic valve disease 3, 618496	ANEURYSM CONGENITAL HEART DISEASE MENDELIOME

ROGDI	100%	100%	100%	99%	Kohlschutter-Tonz syndrome, 226750	SKIN DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ROM1	100%	100%	100%	99%	Retinitis pigmentosa 7, digenic form, 608133	VISION DISORDERS MENDELIOME
ROR1	100%	100%	100%	99%	?Deafness, autosomal recessive 108, 617654	HEARING IMPAIRMENT MENDELIOME
ROR2	100%	100%	100%	99%	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310	DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS
RORA	100%	100%	99%	97%	Intellectual developmental disorder w/wo epilepsy or cerebellar ataxia, 618060	INTELLECTUAL DISABILITY MENDELIOME
RORB	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
RORC	100%	100%	100%	99%	Immunodeficiency 42, 616622	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
RP1	100%	100%	99%	96%	Retinitis pigmentosa 1, 180100	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
RP1L1	100%	100%	100%	98%	Occult macular dystrophy, 613587 Retinitis pigmentosa 88, 618826	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
RP2	100%	100%	97%	69%	Retinitis pigmentosa 2, 312600	VISION DISORDERS MENDELIOME
RP9	100%	100%	99%	94%	?Retinitis pigmentosa 9, 180104	VISION DISORDERS MENDELIOME
RPA1	100%	100%	100%	99%	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 6, 619767	HEREDITARY BONE MARROW FAILURE DYSKERATOSIS CONGENITA PRIMARY IMMUNODEFICIENCY MENDELIOME TUMOR
RPE65	100%	100%	100%	98%	Retinitis pigmentosa 20, 613794 Retinitis pigmentosa 87 with choroidal involvement, 618697 Leber congenital amaurosis 2, 204100	VISION DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING

RPGR	98%	95%	84%	54%	Retinitis pigmentosa, X-linked, and sinorespiratory infections, w/wo deafness, 300455 Cone-rod dystrophy, X-linked, 1, 304020 Retinitis pigmentosa 3, 300029 Macular degeneration, X-linked atrophic, 300834	VISION DISORDERS MENDELIOME
RPGRIP1	100%	100%	100%	98%	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
RPGRIP1L	100%	100%	100%	97%	Joubert syndrome 7, 611560 Meckel syndrome 5, 611561 ?COACH syndrome 3, 619113	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS
RPIA	100%	100%	100%	98%	Ribose 5-phosphate isomerase deficiency, 608611	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
RPL10	100%	99%	98%	71%	Intellectual developmental disorder, X-linked syndromic 35, 300998	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
RPL10L	100%	100%	100%	98%	?Spermatogenic failure 63, 619689	MENDELIOME
RPL11	100%	100%	100%	99%	Diamond-Blackfan anemia 7, 612562	HEREDITARY BONE MARROW FAILURE MENDELIOME SCHISIS TUMOR
RPL13	100%	100%	100%	99%	Spondyloepimetaphyseal dysplasia, Isidor-Toutain type, 618728	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
RPL15	99%	96%	100%	99%	?Diamond-Blackfan anemia 12, 615550	HEREDITARY BONE MARROW FAILURE MENDELIOME TUMOR
RPL18	100%	100%	100%	98%	?Diamond-Blackfan anemia 18, 618310	HEREDITARY BONE MARROW FAILURE MENDELIOME TUMOR
RPL21	100%	100%	100%	99%	Hypotrichosis 12, 615885	SKIN DISORDERS MENDELIOME

RPL26	100%	100%	100%	99%	?Diamond-Blackfan anemia 11, 614900	HEREDITARY BONE MARROW FAILURE MENDELIOME SCHISIS
RPL27	100%	100%	100%	98%	?Diamond-Blackfan anemia 16, 617408	HEREDITARY BONE MARROW FAILURE MENDELIOME TUMOR
RPL31	100%	100%	100%	99%	No OMIM disease ID	HEREDITARY BONE MARROW FAILURE MENDELIOME
RPL35	100%	100%	100%	99%	?Diamond-Blackfan anemia 19, 618312	HEREDITARY BONE MARROW FAILURE MENDELIOME
RPL35A	100%	100%	100%	99%	Diamond-Blackfan anemia 5, 612528	HEREDITARY BONE MARROW FAILURE MENDELIOME TUMOR
RPL3L	100%	100%	100%	99%	Cardiomyopathy, dilated, 2D, 619371	HEART MENDELIOME
RPL4	100%	100%	100%	98%	No OMIM disease ID	HEREDITARY BONE MARROW FAILURE MENDELIOME
RPL5	100%	100%	100%	98%	Diamond-Blackfan anemia 6, 612561	HEREDITARY BONE MARROW FAILURE MENDELIOME SCHISIS TUMOR
RPL9	100%	100%	100%	98%	No OMIM disease ID	HEREDITARY BONE MARROW FAILURE MENDELIOME
RPN2	100%	100%	100%	99%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME
RPS10	100%	100%	100%	97%	Diamond-Blackfan anemia 9, 613308	HEREDITARY BONE MARROW FAILURE MENDELIOME TUMOR
RPS14	100%	100%	100%	97%	Macrocytic anemia, refractory, due to 5q deletion, somatic, 153550	MENDELIOME
RPS15A	79%	79%	100%	95%	?Diamond-Blackfan anemia 20, 618313	HEREDITARY BONE MARROW FAILURE MENDELIOME TUMOR
RPS17	100%	100%	100%	97%	Diamond-Blackfan anemia 4, 612527	HEREDITARY BONE MARROW FAILURE MENDELIOME TUMOR
RPS19	100%	100%	100%	97%	Diamond-Blackfan anemia 1, 105650	HEREDITARY BONE MARROW FAILURE INTELLECTUAL DISABILITY MENDELIOME SCHISIS TUMOR
RPS20	100%	100%	99%	95%	No OMIM disease ID	MENDELIOME TUMOR

RPS23	100%	100%	100%	99%	Brachycephaly, trichomegaly, and developmental delay, 617412	MENDELIOME
RPS24	100%	100%	100%	98%	Diamond-blackfan anemia 3, 610629	HEREDITARY BONE MARROW FAILURE MENDELIOME TUMOR
RPS26	100%	98%	100%	98%	Diamond-Blackfan anemia 10, 613309	HEREDITARY BONE MARROW FAILURE MENDELIOME SCHISIS TUMOR
RPS27	100%	100%	100%	97%	?Diamond-Blackfan anemia 17, 617409	HEREDITARY BONE MARROW FAILURE MENDELIOME TUMOR
RPS28	100%	100%	100%	98%	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164	HEREDITARY BONE MARROW FAILURE MENDELIOME SCHISIS TUMOR
RPS29	100%	100%	100%	97%	Diamond-Blackfan anemia 13, 615909	HEREDITARY BONE MARROW FAILURE MENDELIOME TUMOR
RPS4Y2	50%	50%	48%	19%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
RPS6KA3	99%	99%	98%	71%	Intellectual developmental disorder, X-linked 19, 300844 Coffin-Lowry syndrome, 303600	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
RPS6KB1	100%	100%	100%	97%	No OMIM disease ID	HEART MENDELIOME
RPS7	100%	100%	100%	96%	Diamond-Blackfan anemia 8, 612563	HEREDITARY BONE MARROW FAILURE MENDELIOME TUMOR
RPSA	100%	100%	100%	98%	Asplenia, isolated congenital, 271400	PRIMARY IMMUNODEFICIENCY MENDELIOME
RRAD	100%	100%	100%	96%	No OMIM disease ID	HEART MENDELIOME
RRAGC	100%	100%	100%	98%	No OMIM disease ID	HEART MENDELIOME
RRAS	100%	99%	100%	95%	No OMIM disease ID	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME NOONAN SYNDROME AND RASOPATHY
RRAS2	100%	100%	100%	95%	Noonan syndrome 12, 618624 Ovarian carcinoma,	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME NOONAN SYNDROME AND RASOPATHY

RREB1	100%	100%	100%	99%	No OMIM disease ID	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME NOONAN SYNDROME AND RASOPATHY
RRM1	100%	100%	100%	98%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
RRM2B	100%	100%	100%	97%	Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Rod-cone dystrophy, sensorineural deafness, and Fanconi-type renal dysfunction, 268315 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077	HEARING IMPAIRMENT EPILEPSY INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS
RRP7A	100%	99%	99%	96%	?Microcephaly 28, primary, autosomal recessive, 619453	INTELLECTUAL DISABILITY MENDELIOME
RS1	100%	100%	98%	76%	Retinoschisis, 312700	VISION DISORDERS MENDELIOME
RSPH1	100%	100%	100%	98%	Ciliary dyskinesia, primary, 24, 615481	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING
RSPH3	100%	100%	100%	98%	Ciliary dyskinesia, primary, 32, 616481	CILIOPATHIES MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING
RSPH4A	100%	100%	100%	96%	Ciliary dyskinesia, primary, 11, 612649	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING
RSPH9	100%	100%	100%	98%	Ciliary dyskinesia, primary, 12, 612650	CILIOPATHIES PRIMARY IMMUNODEFICIENCY MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING
RSPO1	100%	100%	100%	99%	Palmoplantar hyperkeratosis and true hermaphroditism, 610644 Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644	SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT MENDELIOME PRE CONCEPTION SCREENING
RSPO2	100%	99%	100%	99%	?Humerofemoral hypoplasia with radiotibial ray deficiency, 618022 Tetraamelia syndrome 2, 618021	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING

RSPO4	100%	100%	100%	98%	Anonychia congenita, 206800	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
RSPRY1	100%	100%	100%	98%	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
RSRC1	100%	99%	100%	98%	Intellectual developmental disorder, autosomal recessive 70, 618402	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
RTEL1	100%	100%	100%	99%	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS DYSKERATOSIS CONGENITA PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING TUMOR
RTN2	100%	100%	100%	99%	Spastic paraplegia 12, autosomal dominant, 604805	MOVEMENT DISORDERS MENDELIOME
RTN4IP1	100%	100%	100%	97%	Optic atrophy 10 w/wo ataxia, impaired intellectual development and seizures, 616732	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
RTTN	100%	99%	100%	98%	Microcephaly, short stature, and polymicrogyria with seizures, 614833	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
RUBCN	100%	100%	100%	98%	Spinocerebellar ataxia, autosomal recessive 15, 615705	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
RUNX1	100%	100%	100%	97%	Platelet disorder, familial, with associated myeloid malignancy, 601399 Leukemia, acute myeloid, 601626	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME TUMOR

RUNX2	100%	100%	99%	95%	Metaphyseal dysplasia with maxillary hypoplasia w/wo brachydactyly, 156510 Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600 Cleidocranial dysplasia, 119600	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS
RUSC2	100%	100%	100%	99%	Intellectual developmental disorder, autosomal recessive 61, 617773	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
RXYLT1	100%	100%	100%	96%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
RYR1	100%	99%	100%	98%	Congenital myopathy 1B, autosomal recessive, 255320 Congenital myopathy 1A, autosomal dominant, with susceptibility to malignant hyperthermia, 117000 King-Denborough syndrome, 619542	FETAL AKINESIA MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS
RYR2	100%	100%	100%	98%	Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772 Ventricular arrhythmias due to cardiac ryanodine receptor calcium release deficiency syndrome, 115000	HEART MENDELIOME ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS
S1PR2	100%	100%	100%	99%	Deafness, autosomal recessive 68, 610419	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
SACS	100%	100%	100%	98%	Spastic ataxia, Charlevoix-Saguenay type, 270550	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
SAG	100%	100%	100%	98%	Retinitis pigmentosa 47, autosomal recessive, 613758 Retinitis pigmentosa 96, autosomal dominant, 620228 Oguchi disease-1, 258100	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SALL1	100%	100%	100%	98%	Townes-Brocks syndrome 1, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME
SALL2	100%	100%	100%	99%	?Coloboma, ocular, autosomal recessive, 216820	VISION DISORDERS MENDELIOME

SALL4	100%	100%	100%	99%	?IVIC syndrome, 147750 Duane-radial ray syndrome, 607323	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME SCHISIS
SAMD11	100%	100%	100%	98%	No OMIM disease ID	VISION DISORDERS MENDELIOME
SAMD12	100%	100%	100%	98%	Epilepsy, familial adult myoclonic, 1, 601068	MENDELIOME
SAMD9	100%	100%	100%	97%	Tumoral calcinosis, familial, normophosphatemic, 610455 Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041 MIRAGE syndrome, 617053	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING TUMOR
SAMD9L	100%	100%	100%	98%	Ataxia-pancytopenia syndrome, 159550 Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270 Spinocerebellar ataxia 49, 619806	MOVEMENT DISORDERS HEREDITARY BONE MARROW FAILURE NEUROPATHIES PRIMARY IMMUNODEFICIENCY MENDELIOME TUMOR
SAMHD1	100%	100%	100%	98%	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952	MOVEMENT DISORDERS SKIN DISORDERS EPILEPSY PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
SAR1B	100%	100%	99%	96%	Chylomicron retention disease, 246700	MENDELIOME PRE CONCEPTION SCREENING
SARDH	91%	91%	100%	98%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME
SARS1	100%	100%	100%	99%	Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SARS2	100%	100%	100%	98%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845	RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING

SART3	100%	100%	100%	99%	No OMIM disease ID	SKIN DISORDERS MENDELIOME
SASH1	100%	100%	100%	98%	Dyschromatosis universalis hereditaria 1, 127500 ?Cancer, alopecia, pigment dyscrasia, onychodystrophy, and keratoderma, 618373	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SASH3	100%	99%	98%	72%	Immunodeficiency 102, 301082	PRIMARY IMMUNODEFICIENCY MENDELIOME
SASS6	100%	100%	100%	96%	Microcephaly 14, primary, autosomal recessive, 616402	MENDELIOME PRE CONCEPTION SCREENING
SAT1	100%	100%	97%	67%	No OMIM disease ID	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS MENDELIOME
SATB1	100%	100%	100%	98%	Kohlschutter-Tonz syndrome-like, 619229 Developmental delay with dysmorphic facies and dental anomalies, 619228	INTELLECTUAL DISABILITY MENDELIOME
SATB2	100%	99%	100%	98%	Glass syndrome, 612313	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS SCHISIS
SBDS	100%	100%	100%	97%	Shwachman-Diamond syndrome 1, 260400	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING TUMOR
SBF1	100%	100%	100%	99%	Charcot-Marie-Tooth disease, type 4B3, 615284	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING
SBF2	100%	100%	100%	98%	Charcot-Marie-Tooth disease, type 4B2, 604563	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING

SC5D	100%	100%	100%	98%	Lathosterolosis, 607330	VISION DISORDERS LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SCAF4	100%	100%	100%	98%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
SCAMP5	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
SCAPER	100%	100%	100%	98%	Intellectual developmental disorder and retinitis pigmentosa, 618195	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SCARB2	100%	100%	100%	99%	Epilepsy, progressive myoclonic 4, w/wo renal failure, 254900	EPILEPSY NEUROPATHIES METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SCARF2	100%	100%	99%	90%	Van den Ende-Gupta syndrome, 600920	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS
SCD5	100%	100%	100%	97%	?Deafness, autosomal dominant 79, 619086	HEARING IMPAIRMENT MENDELIOME
SCLT1	95%	95%	100%	97%	No OMIM disease ID	CILIOPATHIES MENDELIOME
SCN10A	100%	100%	100%	98%	Episodic pain syndrome, familial, 2, 615551	SKIN DISORDERS HEART NEUROPATHIES HNPD MENDELIOME PAINFUL PERIPHERAL NEUROPATHIES

SCN11A	100%	99%	99%	97%	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548	MOVEMENT DISORDERS SKIN DISORDERS NEUROPATHIES HNPD MENDELIOME PAINLESS PERIPHERAL NEUROPATHIES PAINFUL PERIPHERAL NEUROPATHIES
SCN1A	100%	100%	100%	98%	Developmental and epileptic encephalopathy 6B, non-Dravet, 619317 Migraine, familial hemiplegic, 3, 609634 Dravet syndrome, 607208 Febrile seizures, familial, 3A, 604403 Generalized epilepsy with febrile seizures plus, type 2, 604403	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
SCN1B	100%	100%	100%	97%	Generalized epilepsy with febrile seizures plus, type 1, 604233 Developmental and epileptic encephalopathy 52, 617350 Cardiac conduction defect, nonspecific, 612838 Atrial fibrillation, familial, 13, 615377 Brugada syndrome 5, 612838	EPILEPSY HEART HNPD INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING PAINFUL PERIPHERAL NEUROPATHIES
SCN2A	100%	100%	100%	98%	Seizures, benign familial infantile, 3, 607745 Developmental and epileptic encephalopathy 11, 613721 Episodic ataxia, type 9, 618924	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
SCN2B	100%	100%	100%	98%	Atrial fibrillation, familial, 14, 615378	HEART HNPD MENDELIOME PAINFUL PERIPHERAL NEUROPATHIES
SCN3A	100%	100%	100%	98%	Epilepsy, familial focal, with variable foci 4, 617935 Developmental and epileptic encephalopathy 62, 617938	EPILEPSY HNPD INTELLECTUAL DISABILITY MENDELIOME PAINFUL PERIPHERAL NEUROPATHIES
SCN3B	100%	100%	100%	99%	Atrial fibrillation, familial, 16, 613120 Brugada syndrome 7, 613120	HEART HNPD MENDELIOME PAINFUL PERIPHERAL NEUROPATHIES
SCN4A	100%	100%	100%	98%	Paramyotonia congenita, 168300 Hypokalemic periodic paralysis, type 2, 613345 Myotonia congenita, atypical, acetazolamide-responsive, 608390 Myasthenic syndrome, congenital, 16, 614198 Hyperkalemic periodic paralysis, type 2, 170500	FETAL AKINESIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS

SCN4B	100%	100%	100%	98%	Atrial fibrillation, familial, 17, 611819 Long QT syndrome 10, 611819	HEART HNPD MENDELIOME PAINFUL PERIPHERAL NEUROPATHIES
SCN5A	100%	100%	100%	98%	Ventricular fibrillation, familial, 1, 603829 Heart block, progressive, type IA, 113900 Cardiomyopathy, dilated, 1E, 601154 Heart block, nonprogressive, 113900 Long QT syndrome 3, 603830 Sick sinus syndrome 1, 608567 Brugada syndrome 1, 601144 Atrial fibrillation, familial, 10, 614022	DILATED CARDIOMYOPATHY HEART LONG QT SYNDROME MENDELIOME ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS
SCN7A	100%	100%	100%	98%	No OMIM disease ID	HNPD MENDELIOME PAINFUL PERIPHERAL NEUROPATHIES
SCN8A	100%	100%	100%	98%	?Myoclonus, familial, 2, 618364 Seizures, benign familial infantile, 5, 617080 Cognitive impairment w/wo cerebellar ataxia, 614306 Developmental and epileptic encephalopathy 13, 614558	MOVEMENT DISORDERS EPILEPSY HNPD INTELLECTUAL DISABILITY MENDELIOME PAINFUL PERIPHERAL NEUROPATHIES
SCN9A	100%	99%	100%	97%	Erythermalgia, primary, 133020 Insensitivity to pain, congenital, 243000 Small fiber neuropathy, 133020 Paroxysmal extreme pain disorder, 167400 Neuropathy, hereditary sensory and autonomic, type IID, 243000	SKIN DISORDERS NEUROPATHIES HNPD MENDELIOME PRE CONCEPTION SCREENING PAINLESS PERIPHERAL NEUROPATHIES PAINFUL PERIPHERAL NEUROPATHIES
SCNN1A	100%	100%	100%	98%	Pseudohypoaldosteronism, type IB1, autosomal recessive, 264350 ?Liddle syndrome 3, 618126 Bronchiectasis w/wo elevated sweat chloride 2, 613021	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SCNN1B	100%	100%	100%	99%	Bronchiectasis w/wo elevated sweat chloride 1, 211400 Pseudohypoaldosteronism, type IB2, autosomal recessive, 620125 Liddle syndrome 1, 177200	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SCNN1G	100%	100%	100%	99%	Bronchiectasis w/wo elevated sweat chloride 3, 613071 Pseudohypoaldosteronism, type IB3, autosomal recessive, 620126 Liddle syndrome 2, 618114	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SCO1	100%	100%	100%	98%	Mitochondrial complex IV deficiency, nuclear type 4, 619048	LIVER DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING

SCO2	100%	100%	100%	99%	Myopia 6, 608908 Mitochondrial complex IV deficiency, nuclear type 2, 604377	VISION DISORDERS HEART NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
SCP2	100%	100%	100%	98%	?Leukoencephalopathy with dystonia and motor neuropathy, 613724	NEUROPATHIES METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
SCUBE3	100%	100%	100%	99%	Short stature, facial dysmorphism, and skeletal anomalies w/wo cardiac anomalies, 619184	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
SCYL1	100%	100%	100%	98%	Spinocerebellar ataxia, autosomal recessive 21, 616719	NEUROPATHIES LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SCYL2	100%	100%	100%	97%	Arthrogryposis multiplex congenita 4, neurogenic, with agenesis of the corpus callosum, 618766	FETAL AKINESIA MENDELIOME
SDCCAG8	100%	100%	100%	97%	Senior-Loken syndrome 7, 613615 Bardet-Biedl syndrome 16, 615993	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SDHA	100%	100%	100%	99%	Cardiomyopathy, dilated, 1GG, 613642 Mitochondrial complex II deficiency, nuclear type 1, 252011 Neurodegeneration with ataxia and late-onset optic atrophy, 619259 Paragangliomas 5, 614165	HEART INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING TUMOR
SDHAF1	100%	100%	100%	98%	Mitochondrial complex II deficiency, nuclear type 2, 619166	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING

SDHAF2	100%	98%	100%	98%	Paragangliomas 2, 601650	MENDELIOME TUMOR
SDHB	100%	100%	100%	98%	Paragangliomas 4, 115310 Mitochondrial complex II deficiency, nuclear type 4, 619224 Gastrointestinal stromal tumor, 606764 Pheochromocytoma, 171300 Paraganglioma and gastric stromal sarcoma, 606864	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING TUMOR
SDHC	100%	100%	100%	98%	Paragangliomas 3, 605373 Paraganglioma and gastric stromal sarcoma, 606864 Gastrointestinal stromal tumor, 606764	MENDELIOME TUMOR
SDHD	78%	78%	100%	98%	Paragangliomas 1, w/wo deafness, 168000 Paraganglioma and gastric stromal sarcoma, 606864 Mitochondrial complex II deficiency, nuclear type 3, 619167 Pheochromocytoma, 171300	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING TUMOR
SDR9C7	100%	100%	100%	99%	Ichthyosis, congenital, autosomal recessive 13, 617574	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SEC23A	100%	100%	100%	98%	Craniolenticulosutural dysplasia, 607812	MENDELIOME PRE CONCEPTION SCREENING SCHISIS
SEC23B	100%	100%	100%	98%	?Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS IRON DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SEC24D	100%	99%	100%	98%	Cole-Carpenter syndrome 2, 616294	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
SEC31A	100%	100%	100%	98%	?Halperin-Birk syndrome, 618651	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

SEC61A1	100%	100%	100%	98%	Tubulointerstitial kidney disease, autosomal dominant, 5, 617056	PRIMARY IMMUNODEFICIENCY RENAL DISORDERS MENDELIOME
SEC61B	100%	100%	100%	96%	No OMIM disease ID	LIVER DISORDERS MENDELIOME
SEC63	100%	100%	100%	98%	Polycystic liver disease 2, 617004	LIVER DISORDERS MENDELIOME
SECISBP2	100%	100%	100%	98%	Thyroid hormone metabolism, abnormal, 1, 609698	MENDELIOME PRE CONCEPTION SCREENING
SELENBP1	100%	100%	100%	99%	Extraoral halitosis due to MTO deficiency, 618148	METABOLIC DISORDERS MENDELIOME
SELENOI	100%	100%	100%	99%	Spastic paraplegia 81, autosomal recessive, 618768	MENDELIOME
SELENON	93%	91%	99%	95%	Congenital myopathy 3 with rigid spine, 602771	FETAL AKINESIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
SEMA3A	100%	100%	100%	99%	No OMIM disease ID	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
SEMA3E	100%	100%	100%	98%	No OMIM disease ID	CRANIOFACIAL ANOMALIES HYPOGONADOTROPIC HYPOGONADISM PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME SCHISIS
SEMA4A	100%	100%	99%	97%	Retinitis pigmentosa 35, 610282 Cone-rod dystrophy 10, 610283	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING TUMOR
SEMA6B	100%	100%	100%	98%	Epilepsy, progressive myoclonic, 11, 618876	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
SEMA7A	100%	100%	100%	98%	?Cholestasis, progressive familial intrahepatic, 11, 619874	LIVER DISORDERS MENDELIOME
SEPSECS	100%	100%	100%	98%	Pontocerebellar hypoplasia type 2D, 613811	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

SEPTIN12	100%	100%	100%	99%	Spermatogenic failure 10, 614822	MALE INFERTILITY MENDELIOME
SEPTIN9	100%	100%	100%	97%	Amyotrophy, hereditary neuralgic, 162100	NEUROPATHIES HNPD MENDELIOME SCHISIS
SERAC1	100%	100%	100%	98%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739	MOVEMENT DISORDERS HEARING IMPAIRMENT PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
SERPINA1	100%	100%	100%	99%	Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490 Emphysema due to AAT deficiency, 613490 Emphysema-cirrhosis, due to AAT deficiency, 613490	LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SERPINA12	100%	100%	100%	98%	No OMIM disease ID	SKIN DISORDERS MENDELIOME
SERPINA3	100%	100%	100%	98%	Alpha-1-antichymotrypsin deficiency, Cerebrovascular disease, occlusive,	SKIN DISORDERS MENDELIOME
SERPINA6	100%	100%	100%	99%	Corticosteroid-binding globulin deficiency, 611489	MENDELIOME PRE CONCEPTION SCREENING
SERPINB6	100%	100%	100%	98%	?Deafness, autosomal recessive 91, 613453	HEARING IMPAIRMENT MENDELIOME
SERPINB7	100%	100%	100%	98%	Palmoplantar keratoderma, Nagashima type, 615598	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SERPINB8	100%	100%	100%	98%	Peeling skin syndrome 5, 617115	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SERPINC1	100%	100%	100%	98%	Thrombophilia 7 due to antithrombin III deficiency, 613118	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SERPIND1	100%	100%	100%	99%	Thrombophilia 10 due to heparin cofactor II deficiency, 612356	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME
SERPINE1	100%	100%	100%	98%	Plasminogen activator inhibitor-1 deficiency, 613329	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING

SERPINF1	100%	100%	100%	98%	Osteogenesis imperfecta, type VI, 613982	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
SERPINF2	100%	100%	99%	97%	Alpha-2-plasmin inhibitor deficiency, 262850	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SERPING1	100%	100%	100%	99%	Angioedema, hereditary, 1 and 2, 106100 Complement component 4, partial deficiency of, 120790	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
SERPINH1	100%	100%	100%	99%	Osteogenesis imperfecta, type X, 613848	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
SERPINI1	100%	100%	100%	99%	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218	EPILEPSY MENDELIOME
SET	100%	99%	98%	86%	Intellectual developmental disorder, autosomal dominant 58, 618106	INTELLECTUAL DISABILITY MENDELIOME
SETBP1	100%	100%	100%	97%	Schinzel-Giedion midface retraction syndrome, 269150 Intellectual developmental disorder, autosomal dominant 29, 616078	INTELLECTUAL DISABILITY MENDELIOME
SETD1A	100%	100%	100%	98%	Epilepsy, early-onset, w/wo developmental delay, 618832 Neurodevelopmental disorder with speech impairment and dysmorphic facies, 619056	INTELLECTUAL DISABILITY MENDELIOME
SETD1B	100%	99%	99%	95%	Intellectual developmental disorder with seizures and language delay, 619000	INTELLECTUAL DISABILITY MENDELIOME
SETD2	100%	100%	100%	97%	Luscan-Lumish syndrome, 616831 Intellectual developmental disorder, autosomal dominant 70, 620157 Rabin-Pappas syndrome, 620155	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
SETD5	100%	100%	100%	99%	Intellectual developmental disorder, autosomal dominant 23, 615761	INTELLECTUAL DISABILITY MENDELIOME
SETX	100%	100%	100%	98%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 Amyotrophic lateral sclerosis 4, juvenile, 602433	ALS MOVEMENT DISORDERS NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING
SEZ6	100%	100%	100%	98%	No OMIM disease ID	HEARING IMPAIRMENT MENDELIOME
SF3B1	100%	100%	100%	98%	Myelodysplastic syndrome, somatic, 614286	IRON DISORDERS MENDELIOME

SF3B2	100%	100%	100%	98%	Craniofacial microsomia, 164210	CRANIOFACIAL ANOMALIES MENDELIOME
SF3B4	100%	100%	100%	99%	Acrofacial dysostosis 1, Nager type, 154400	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS
SFRP4	100%	100%	100%	98%	Pyle disease, 265900	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
SFTPA1	100%	100%	100%	99%	Interstitial lung disease 1, 619611	MENDELIOME TUMOR
SFTPA2	100%	100%	100%	99%	Interstitial lung disease 2, 178500	MENDELIOME TUMOR
SFTPB	100%	100%	100%	99%	Surfactant metabolism dysfunction, pulmonary, 1, 265120	MENDELIOME PRE CONCEPTION SCREENING
SFTPC	100%	100%	100%	98%	Surfactant metabolism dysfunction, pulmonary, 2, 610913	MENDELIOME
SFXN4	100%	100%	100%	97%	Combined oxidative phosphorylation deficiency 18, 615578	IRON DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
SGCA	100%	100%	100%	99%	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099	HEART MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
SGCB	100%	100%	100%	97%	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286	HEART MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
SGCD	100%	99%	100%	99%	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287	HEART MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
SGCE	90%	90%	100%	97%	Dystonia-11, myoclonic, 159900	MOVEMENT DISORDERS MENDELIOME
SGCG	100%	100%	100%	99%	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700	HEART MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS

SGMS2	100%	100%	100%	99%	Calvarial doughnut lesions with bone fragility w/wo spondylometaphyseal dysplasia, 126550	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
SGO1	100%	100%	100%	97%	Chronic atrial and intestinal dysrhythmia, 616201	LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SGPL1	100%	100%	100%	99%	Nephrotic syndrome, type 14, 617575	SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT NEUROPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SGSH	100%	100%	100%	99%	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900	VISION DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SH2B3	100%	100%	100%	97%	Thrombocythemia, somatic, 187950 Myelofibrosis, somatic, 254450 Erythrocytosis, somatic, 133100	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME TUMOR
SH2D1A	100%	100%	99%	83%	Lymphoproliferative syndrome, X-linked, 1, 308240	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCY MENDELIOME TUMOR
SH3BP2	99%	99%	100%	97%	Cherubism, 118400	VISION DISORDERS CRANIOFACIAL ANOMALIES PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
SH3KBP1	99%	99%	97%	71%	?Immunodeficiency 61, 300310	PRIMARY IMMUNODEFICIENCY MENDELIOME
SH3PXD2B	100%	100%	100%	98%	Frank-ter Haar syndrome, 249420	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING

SH3TC2	100%	100%	100%	99%	Charcot-Marie-Tooth disease, type 4C, 601596 Mononeuropathy of the median nerve, mild, 613353	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING
SHANK1	100%	100%	99%	91%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
SHANK2	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
SHANK3	99%	99%	99%	95%	Phelan-McDermid syndrome, 606232	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
SHH	100%	100%	100%	95%	Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250 Holoprosencephaly 3, 142945	VISION DISORDERS CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS
SHMT2	100%	100%	100%	99%	Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121	HEART METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME
SHOC1	100%	100%	100%	97%	Spermatogenic failure 75, 619949	MALE INFERTILITY MENDELIOME
SHOC2	100%	100%	100%	97%	Noonan syndrome-like with loose anagen hair 1, 607721	SKIN DISORDERS HEART SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME AND RASOPATHY TUMOR
SHOX	94%	94%	50%	49%	Short stature, idiopathic familial, 300582 Leri-Weill dyschondrosteosis, 127300 Langer mesomelic dysplasia, 249700 Short stature, idiopathic familial, 300582 Langer mesomelic dysplasia, 249700 Leri-Weill dyschondrosteosis, 127300	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
SHQ1	100%	100%	100%	98%	Neurodevelopmental disorder with dystonia and seizures, 619922 ?Dystonia 35, childhood-onset, 619921	INTELLECTUAL DISABILITY MENDELIOME
SHROOM3	100%	100%	100%	99%	No OMIM disease ID	CONGENITAL HEART DISEASE HEART MENDELIOME

SHROOM4	100%	99%	98%	71%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
SI	99%	98%	100%	98%	Sucrase-isomaltase deficiency, congenital, 222900	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SIAH1	100%	100%	100%	99%	Buratti-Harel syndrome, 619314	INTELLECTUAL DISABILITY MENDELIOME
SIGLEC7	100%	100%	100%	99%	No OMIM disease ID	ANEURYSM MENDELIOME
SIGMAR1	100%	100%	100%	99%	?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726 ?Amyotrophic lateral sclerosis 16, juvenile, 614373	ALS NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING
SIK1	100%	100%	100%	99%	Developmental and epileptic encephalopathy 30, 616341	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
SIK3	100%	100%	100%	96%	?Spondyloepimetaphyseal dysplasia, Krakow type, 618162	MENDELIOME PRE CONCEPTION SCREENING
SIL1	100%	100%	100%	99%	Marinesco-Sjogren syndrome, 248800	MOVEMENT DISORDERS VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
SIN3A	100%	100%	100%	99%	Witteveen-Kolk syndrome, 613406	INTELLECTUAL DISABILITY MENDELIOME
SIN3B	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
SIPA1L3	100%	100%	100%	99%	?Cataract 45, 616851	VISION DISORDERS MENDELIOME
SIX1	100%	100%	100%	97%	Deafness, autosomal dominant 23, 605192 Branchiootic syndrome 3, 608389	CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT MENDELIOME SCHISIS
SIX3	100%	100%	100%	95%	Schizencephaly, 269160 Holoprosencephaly 2, 157170	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME SCHISIS

SIX5	100%	100%	99%	94%	Branchiootorenal syndrome 2, 610896	CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME SCHISIS
SIX6	100%	100%	100%	98%	Optic disc anomalies with retinal and/or macular dystrophy, 212550	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SKI	100%	99%	99%	92%	Shprintzen-Goldberg syndrome, 182212	ANEURYSM CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS
SKIV2L	100%	100%	100%	99%	Trichohepatoenteric syndrome 2, 614602	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
SLC10A1	100%	100%	100%	99%	Hypercholanemia, familial 2, 619256	MENDELIOME
SLC10A2	100%	100%	100%	97%	?Bile acid malabsorption, primary, 1, 613291	MENDELIOME
SLC10A7	100%	100%	100%	99%	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS
SLC11A2	100%	100%	100%	98%	Anemia, hypochromic microcytic, with iron overload 1, 206100	IRON DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC12A1	96%	96%	100%	98%	Bartter syndrome, type 1, 601678	HEARING IMPAIRMENT METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC12A2	100%	100%	100%	97%	Kilquist syndrome, 619080 Delpire-McNeill syndrome, 619083 Deafness, autosomal dominant 78, 619081	HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

SLC12A3	100%	100%	100%	99%	Gitelman syndrome, 263800	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC12A5	100%	100%	100%	98%	Developmental and epileptic encephalopathy 34, 616645	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SLC12A6	100%	100%	100%	98%	Agenesis of the corpus callosum with peripheral neuropathy, 218000 Charcot-Marie-Tooth disease, axonal, type 2II, 620068	MOVEMENT DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SLC13A3	100%	100%	100%	99%	Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate, 618384	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC13A5	100%	100%	100%	98%	Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SLC16A1	100%	100%	100%	99%	Hyperinsulinemic hypoglycemia, familial, 7, 610021 Erythrocyte lactate transporter defect, 245340 Monocarboxylate transporter 1 deficiency, 616095	EPILEPSY METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC16A12	100%	100%	100%	99%	Cataract 47, juvenile, with microcornea, 612018	VISION DISORDERS RENAL DISORDERS MENDELIOME
SLC16A2	100%	99%	98%	67%	Allan-Herndon-Dudley syndrome, 300523	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME
SLC17A5	100%	100%	100%	97%	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SLC17A8	100%	100%	100%	97%	Deafness, autosomal dominant 25, 605583	HEARING IMPAIRMENT MENDELIOME
SLC17A9	100%	100%	100%	98%	Porokeratosis 8, disseminated superficial actinic type, 616063	SKIN DISORDERS MENDELIOME
SLC18A2	100%	100%	100%	98%	?Parkinsonism-dystonia, infantile, 2, 618049	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING

SLC18A3	100%	100%	100%	99%	Myasthenic syndrome, congenital, 21, presynaptic, 617239	MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
SLC19A1	100%	100%	100%	99%	?Megaloblastic anemia, folate-responsive, 601775	MENDELIOME
SLC19A2	100%	100%	100%	99%	Thiamine-responsive megaloblastic anemia syndrome, 249270	HEREDITARY BONE MARROW FAILURE HEARING IMPAIRMENT IRON DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
SLC19A3	99%	98%	100%	98%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
SLC1A1	100%	100%	100%	98%	Dicarboxylic aminoaciduria, 222730	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SLC1A2	100%	99%	100%	99%	Developmental and epileptic encephalopathy 41, 617105	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
SLC1A3	100%	100%	100%	99%	Episodic ataxia, type 6, 612656	MOVEMENT DISORDERS MENDELIOME
SLC1A4	100%	100%	100%	98%	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SLC20A2	100%	100%	100%	99%	Basal ganglia calcification, idiopathic, 1, 213600	MOVEMENT DISORDERS MENDELIOME PARKINSON
SLC22A12	100%	99%	99%	97%	Hypouricemia, renal, 220150	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC22A18	100%	100%	100%	99%	Breast cancer, somatic, 114480 Lung cancer, somatic, 211980 Rhabdomyosarcoma, somatic, 268210	MENDELIOME
SLC22A4	100%	100%	100%	98%	No OMIM disease ID	HEARING IMPAIRMENT MENDELIOME

SLC22A5	100%	100%	100%	98%	Carnitine deficiency, systemic primary, 212140	HEART METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC24A1	100%	100%	100%	98%	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC24A4	100%	100%	100%	98%	Amelogenesis imperfecta, type IIA5, 615887	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC24A5	100%	99%	100%	98%	Albinism, oculocutaneous, type VI, 113750	VISION DISORDERS SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC25A1	100%	100%	100%	93%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182 Myasthenic syndrome, congenital, 23, presynaptic, 618197	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
SLC25A10	100%	100%	100%	99%	?Mitochondrial DNA depletion syndrome 19, 618972	MENDELIOME MITOCHONDRIAL DISORDERS
SLC25A11	100%	100%	100%	98%	Paragangliomas 6, 618464	 MENDELIOME TUMOR
SLC25A12	100%	100%	100%	98%	Developmental and epileptic encephalopathy 39, 612949	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
SLC25A13	100%	100%	100%	98%	Citrullinemia, type II, neonatal-onset, 605814 Citrullinemia, adult-onset type II, 603471	LIVER DISORDERS METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING

SLC25A15	100%	100%	100%	99%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SLC25A19	100%	100%	100%	98%	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710	NEUROPATHIES METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
SLC25A20	100%	100%	100%	99%	Carnitine-acylcarnitine translocase deficiency, 212138	HEART METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC25A21	100%	100%	100%	98%	?Mitochondrial DNA depletion syndrome 18, 618811	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS
SLC25A22	100%	100%	100%	99%	Developmental and epileptic encephalopathy 3, 609304	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
SLC25A24	99%	99%	99%	97%	Fontaine progeroid syndrome, 612289	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS
SLC25A26	100%	100%	100%	98%	Combined oxidative phosphorylation deficiency 28, 616794	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
SLC25A3	100%	100%	100%	99%	Mitochondrial phosphate carrier deficiency, 610773	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
SLC25A32	100%	100%	100%	98%	?Exercise intolerance, riboflavin-responsive, 616839	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS
SLC25A36	100%	100%	100%	97%	Hyperinsulinemic hypoglycemia, familial, 8, 620211	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS

SLC25A37	100%	100%	100%	98%	No OMIM disease ID	IRON DISORDERS MENDELIOME
SLC25A38	100%	100%	100%	99%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950	HEREDITARY BONE MARROW FAILURE IRON DISORDERS METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
SLC25A4	100%	100%	100%	98%	Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283 Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184	HEART MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS
SLC25A42	100%	100%	100%	99%	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
SLC25A46	100%	100%	99%	98%	Neuropathy, hereditary motor and sensory, type VIB, 616505 Pontocerebellar hypoplasia, type 1E, 619303	VISION DISORDERS NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
SLC26A1	100%	100%	100%	99%	?Nephrolithiasis, calcium oxalate, 167030	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC26A2	100%	100%	100%	98%	Epiphyseal dysplasia, multiple, 4, 226900 De la Chapelle dysplasia, 256050 Diastrophic dysplasia, 222600 Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 Achondrogenesis Ib, 600972 Atelosteogenesis, type II, 256050	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS
SLC26A3	100%	100%	100%	98%	Diarrhea 1, secretory chloride, congenital, 214700	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC26A4	100%	100%	100%	97%	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791 Pendred syndrome, 274600	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
SLC26A5	100%	100%	100%	98%	?Deafness, autosomal recessive 61, 613865	HEARING IMPAIRMENT MENDELIOME
SLC26A8	100%	100%	100%	98%	Spermatogenic failure 3, 606766	MENDELIOME

SLC27A4	100%	100%	100%	99%	Ichthyosis prematurity syndrome, 608649	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC28A1	100%	100%	100%	99%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME
SLC29A3	100%	100%	100%	99%	Histiocytosis-lymphadenopathy plus syndrome, 602782	SKIN DISORDERS HEARING IMPAIRMENT PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
SLC2A1	100%	100%	100%	99%	Dystonia 9, 601042 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 GLUT1 deficiency syndrome 2, childhood onset, 612126	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SLC2A10	100%	100%	100%	99%	Arterial tortuosity syndrome, 208050	ANEURYSM SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC2A2	100%	100%	100%	99%	Fanconi-Bickel syndrome, 227810	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC2A9	100%	100%	100%	98%	Hypouricemia, renal, 2, 612076	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC30A10	100%	100%	100%	98%	Hypermanganesemia with dystonia 1, 613280	MOVEMENT DISORDERS METABOLIC DISORDERS MENDELIOME PARKINSON PRE CONCEPTION SCREENING
SLC30A2	100%	100%	100%	99%	Zinc deficiency, transient neonatal, 608118	MENDELIOME
SLC30A5	100%	100%	100%	97%	No OMIM disease ID	HEART MENDELIOME
SLC30A9	100%	100%	100%	98%	Birk-Landau-Perez syndrome, 617595	MENDELIOME PRE CONCEPTION SCREENING

SLC33A1	100%	100%	100%	97%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482	MOVEMENT DISORDERS VISION DISORDERS HEARING IMPAIRMENT METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SLC34A1	100%	100%	100%	98%	?Fanconi renotubular syndrome 2, 613388 Hypercalcemia, infantile, 2, 616963 Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC34A2	100%	100%	100%	98%	Pulmonary alveolar microlithiasis, 265100	MENDELIOME PRE CONCEPTION SCREENING
SLC34A3	100%	100%	100%	97%	Hypophosphatemic rickets with hypercalciuria, 241530	SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC35A1	100%	100%	100%	99%	Congenital disorder of glycosylation, type IIf, 603585	PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SLC35A2	100%	100%	98%	75%	Congenital disorder of glycosylation, type IIm, 300896	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME
SLC35A3	97%	93%	99%	96%	Arthrogryposis, impaired intellectual development, and seizures, 615553	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SLC35B2	100%	100%	100%	99%	Leukodystrophy, hypomyelinating, 26, with chondrodysplasia, 620269	INTELLECTUAL DISABILITY MENDELIOME
SLC35C1	100%	100%	100%	99%	Congenital disorder of glycosylation, type IIc, 266265	PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

SLC35D1	100%	100%	100%	97%	Schneckenbecken dysplasia, 269250	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC36A2	100%	100%	100%	98%	Iminoglycinuria, digenic, 242600 Hyperglycinuria, 138500	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME
SLC37A3	100%	100%	100%	99%	No OMIM disease ID	VISION DISORDERS MENDELIOME
SLC37A4	100%	100%	100%	99%	Glycogen storage disease Ib, 232220 Congenital disorder of glycosylation, type IIw, 619525 Glycogen storage disease Ic, 232240	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC38A3	100%	100%	100%	98%	Developmental and epileptic encephalopathy 102, 619881	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME
SLC38A8	100%	100%	100%	99%	Foveal hypoplasia 2, w/wo optic nerve misrouting and/or anterior segment dysgenesis, 609218	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC39A12	100%	100%	100%	98%	No OMIM disease ID	VISION DISORDERS MENDELIOME
SLC39A13	100%	100%	100%	99%	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
SLC39A14	93%	93%	100%	99%	?Hyperostosis cranalis interna, 144755 Hypermanganesemia with dystonia 2, 617013	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PARKINSON PRE CONCEPTION SCREENING
SLC39A4	100%	100%	100%	99%	Acrodermatitis enteropathica, 201100	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING

SLC39A5	100%	100%	100%	99%	Myopia 24, autosomal dominant, 615946	VISION DISORDERS MENDELIOME
SLC39A7	100%	100%	100%	98%	Agammaglobulinemia 9, autosomal recessive, 619693	PRIMARY IMMUNODEFICIENCY MENDELIOME
SLC39A8	100%	100%	100%	97%	Congenital disorder of glycosylation, type IIn, 616721	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
SLC3A1	96%	96%	100%	99%	Cystinuria, 220100	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC40A1	100%	100%	100%	99%	Hemochromatosis, type 4, 606069	IRON DISORDERS LIVER DISORDERS MENDELIOME
SLC41A1	100%	100%	100%	99%	?Nephronophthisis-like nephropathy 2, 619468	RENAL DISORDERS MENDELIOME
SLC44A1	100%	100%	100%	97%	Neurodegeneration, childhood-onset, with ataxia, tremor, optic atrophy, and cognitive decline, 618868	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC44A4	100%	100%	100%	98%	?Deafness, autosomal dominant 72, 617606	HEARING IMPAIRMENT MENDELIOME
SLC45A1	100%	100%	100%	97%	Intellectual developmental disorder with neuropsychiatric features, 617532	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SLC45A2	100%	100%	100%	99%	Albinism, oculocutaneous, type IV, 606574	VISION DISORDERS SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC46A1	100%	100%	100%	98%	Folate malabsorption, hereditary, 229050	HEREDITARY BONE MARROW FAILURE IRON DISORDERS PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

SLC4A1	100%	100%	100%	99%	Distal renal tubular acidosis 1, 179800 Spherocytosis, type 4, 612653 Distal renal tubular acidosis 4 with hemolytic anemia, 611590 Cryohydrocytosis, 185020 Ovalocytosis, SA type, 166900	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC4A11	100%	100%	100%	99%	Corneal endothelial dystrophy, autosomal recessive, 217700 Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy and perceptive deafness, 217400	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
SLC4A2	100%	100%	100%	98%	No OMIM disease ID	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
SLC4A3	100%	100%	100%	98%	Short QT syndrome 7, 620231	HEART MENDELIOME
SLC4A4	100%	99%	100%	98%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278	SKIN DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC4A7	100%	100%	100%	98%	No OMIM disease ID	VISION DISORDERS MENDELIOME
SLC51A	100%	100%	100%	99%	?Cholestasis, progressive familial intrahepatic, 6, 619484	LIVER DISORDERS MENDELIOME
SLC51B	100%	100%	100%	98%	?Bile acid malabsorption, primary, 2, 619481	MENDELIOME
SLC52A1	100%	100%	100%	99%	Riboflavin deficiency, 615026	METABOLIC DISORDERS MENDELIOME
SLC52A2	100%	100%	100%	99%	Brown-Vialetto-Van Laere syndrome 2, 614707	MOVEMENT DISORDERS VISION DISORDERS HEARING IMPAIRMENT NEUROPATHIES METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS

SLC52A3	100%	100%	100%	99%	?Fazio-Londe disease, 211500 Brown-Vialetto-Van Laere syndrome 1, 211530	MOVEMENT DISORDERS HEARING IMPAIRMENT NEUROPATHIES METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS
SLC5A1	100%	100%	100%	98%	Glucose/galactose malabsorption, 606824	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC5A2	100%	100%	100%	99%	Renal glucosuria, 233100	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC5A5	100%	100%	99%	97%	Thyroid dyshormonogenesis 1, 274400	MENDELIOME PRE CONCEPTION SCREENING
SLC5A6	100%	100%	100%	99%	Sodium-dependent multivitamin transporter deficiency, 618973 Peripheral motor neuropathy, childhood-onset, biotin-responsive, 619903	NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME
SLC5A7	100%	100%	100%	99%	Neuronopathy, distal hereditary motor, type VIIA, 158580 Myasthenic syndrome, congenital, 20, presynaptic, 617143	FETAL AKINESIA NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
SLC66A1	100%	100%	100%	99%	No OMIM disease ID	VISION DISORDERS MENDELIOME
SLC6A1	100%	100%	100%	99%	Myoclonic-atonic epilepsy, 616421	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
SLC6A17	100%	100%	100%	97%	Intellectual developmental disorder, autosomal recessive 48, 616269	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SLC6A19	100%	100%	100%	99%	Iminoglycinuria, digenic, 242600 Hartnup disorder, 234500 Hyperglycinuria, 138500	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING

SLC6A2	100%	100%	100%	98%	?Orthostatic intolerance, 604715	MENDELIOME
SLC6A20	100%	100%	100%	99%	Iminoglycinuria, digenic, 242600 Hyperglycinuria, 138500	RENAL DISORDERS MENDELIOME
SLC6A3	100%	100%	100%	99%	Parkinsonism-dystonia, infantile, 1, 613135	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PARKINSON PRE CONCEPTION SCREENING
SLC6A5	100%	100%	100%	98%	Hyperekplexia 3, 614618	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC6A6	100%	100%	100%	98%	Hypotaurinemic retinal degeneration and cardiomyopathy, 145350	HEART METABOLIC DISORDERS MENDELIOME
SLC6A8	100%	99%	95%	68%	Cerebral creatine deficiency syndrome 1, 300352	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME
SLC6A9	100%	100%	100%	99%	Glycine encephalopathy with normal serum glycine, 617301	FETAL AKINESIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SLC7A14	100%	100%	100%	99%	Retinitis pigmentosa 68, 615725	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
SLC7A6OS	100%	100%	100%	98%	Epilepsy, progressive myoclonic, 12, 619191	MENDELIOME
SLC7A7	100%	100%	100%	98%	Lysinuric protein intolerance, 222700	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC7A9	100%	100%	100%	99%	Cystinuria, 220100	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC8B1	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS

SLC9A1	100%	100%	100%	99%	Lichtenstein-Knorr syndrome, 616291	MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC9A3	100%	99%	99%	94%	Diarrhea 8, secretory sodium, congenital, 616868	RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLC9A3R1	100%	100%	100%	97%	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287	HEARING IMPAIRMENT NEUROPATHIES RENAL DISORDERS MENDELIOME
SLC9A6	100%	99%	97%	71%	Intellectual developmental disorder, X-linked syndromic, Christianson type, 300243	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
SLC9A7	100%	99%	98%	71%	Intellectual developmental disorder, X-linked 108, 301024	INTELLECTUAL DISABILITY MENDELIOME
SLCO1B1	100%	100%	100%	97%	Hyperbilirubinemia, Rotor type, digenic, 237450	METABOLIC DISORDERS MENDELIOME
SLCO1B3	100%	100%	100%	97%	Hyperbilirubinemia, Rotor type, digenic, 237450	METABOLIC DISORDERS MENDELIOME
SLCO2A1	100%	100%	100%	99%	Hypertrophic osteoarthropathy, primary, autosomal dominant, 167100 Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
SLCO5A1	100%	100%	100%	98%	No OMIM disease ID	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
SLFN14	100%	100%	100%	98%	Bleeding disorder, platelet-type, 20, 616913	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME
SLIRP	100%	100%	99%	95%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
SLIT3	100%	100%	100%	99%	No OMIM disease ID	RENAL DISORDERS MENDELIOME
SLITRK1	100%	100%	100%	97%	Tourette syndrome, 137580 ?Trichotillomania, 613229	MENDELIOME
SLITRK6	100%	100%	100%	98%	Deafness and myopia, 221200	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
SLMAP	100%	100%	100%	98%	No OMIM disease ID	HEART MENDELIOME

SLURP1	100%	100%	100%	99%	Meleda disease, 248300	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SLX4	100%	100%	100%	99%	Fanconi anemia, complementation group P, 613951	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING TUMOR
SMAD1	100%	100%	100%	98%	No OMIM disease ID	HEART MENDELIOME
SMAD2	100%	100%	100%	99%	Loeys-Dietz syndrome 6, 619656 Congenital heart defects, multiple types, 8, w/wo heterotaxy, 619657	ANEURYSM SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
SMAD3	100%	100%	99%	96%	Loeys-Dietz syndrome 3, 613795	ANEURYSM SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS
SMAD4	100%	100%	100%	99%	Pancreatic cancer, somatic, 260350 Myhre syndrome, 139210 Polyposis, juvenile intestinal, 174900 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050	ANEURYSM SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS TUMOR
SMAD6	100%	100%	99%	91%	Aortic valve disease 2, 614823	ANEURYSM CRANIOFACIAL ANOMALIES CONGENITAL HEART DISEASE HEART SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
SMAD9	100%	100%	100%	98%	Pulmonary hypertension, primary, 2, 615342	HEART MENDELIOME TUMOR
SMARCA1	100%	99%	97%	69%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME

SMARCA2	100%	99%	100%	98%	Nicolaides-Baraitser syndrome, 601358 Blepharophimosis-impaired intellectual development syndrome, 619293	SKIN DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
SMARCA4	100%	100%	100%	99%	Coffin-Siris syndrome 4, 614609	CONGENITAL HEART DISEASE SKIN DISORDERS HEART SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME TUMOR
SMARCA5	100%	100%	100%	97%	No OMIM disease ID	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
SMARCAD1	100%	100%	100%	97%	Basan syndrome, 129200 Huriez syndrome, 181600 Adermatoglyphia, 136000	SKIN DISORDERS MENDELIOME
SMARCAL1	100%	100%	100%	98%	Schimke immunoosseous dysplasia, 242900	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SMARCB1	100%	100%	100%	98%	Rhabdoid tumors, somatic, 609322 Coffin-Siris syndrome 3, 614608	SKIN DISORDERS HNPD SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SONIC HEDGEHOG MEDULLOBLASTOMA TUMOR
SMARCC2	100%	100%	100%	98%	Coffin-Siris syndrome 8, 618362	INTELLECTUAL DISABILITY MENDELIOME
SMARCD1	100%	100%	99%	95%	Coffin-Siris syndrome 11, 618779	INTELLECTUAL DISABILITY MENDELIOME
SMARCD2	100%	100%	100%	97%	Specific granule deficiency 2, 617475	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING

SMARCE1	100%	100%	100%	98%	Coffin-Siris syndrome 5, 616938	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME TUMOR
SMC1A	100%	99%	97%	68%	Cornelia de Lange syndrome 2, 300590 Developmental and epileptic encephalopathy 85, w/wo midline brain defects, 301044	CRANIOFACIAL ANOMALIES EPILEPSY SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS
SMC3	100%	100%	100%	98%	Cornelia de Lange syndrome 3, 610759	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS
SMCHD1	100%	100%	100%	98%	Bosma arhinia microphthalmia syndrome, 603457 Fascioscapulohumeral muscular dystrophy 2, digenic, 158901	MENDELIOME SCHISIS MUSCLE DISORDERS
SMDT1	100%	100%	100%	99%	No OMIM disease ID	MOVEMENT DISORDERS MENDELIOME MUSCLE DISORDERS
SMG8	100%	100%	100%	98%	Alzahrani-Kuwahara syndrome, 619268	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME
SMG9	100%	100%	100%	99%	Heart and brain malformation syndrome, 616920 Neurodevelopmental disorder with intention tremor, pyramidal signs, dyspraxia, and ocular anomalies, 619995	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SMN1	93%	93%	99%	92%	Spinal muscular atrophy-2, 253550 Spinal muscular atrophy-4, 271150 Spinal muscular atrophy-3, 253400 Spinal muscular atrophy-1, 253300	FETAL AKINESIA MENDELIOME PRE CONCEPTION SCREENING
SMO	100%	100%	100%	98%	Pallister-Hall-like syndrome, 241800 Basal cell carcinoma, somatic, 605462 Curry-Jones syndrome, somatic mosaic, 601707	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING

SMOC1	100%	100%	100%	99%	Microphthalmia with limb anomalies, 206920	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SMOC2	100%	100%	100%	98%	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
SMPD1	100%	100%	100%	98%	Niemann-Pick disease, type B, 607616 Niemann-Pick disease, type A, 257200	MOVEMENT DISORDERS LIVER DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SMPD4	100%	100%	100%	99%	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622	FETAL AKINESIA EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SMPX	100%	99%	98%	70%	Myopathy, distal, 7, adult-onset, X-linked, 301075 Deafness, X-linked 4, 300066	HEARING IMPAIRMENT MENDELIOME
SMS	100%	99%	97%	73%	Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type, 309583	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME SCHISIS
SNAI2	100%	100%	100%	99%	Waardenburg syndrome, type 2D, 608890 Piebaldism, 172800	CRANIOFACIAL ANOMALIES SKIN DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
SNAP25	100%	100%	100%	98%	?Myasthenic syndrome, congenital, 18, 616330	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
SNAP29	100%	100%	100%	96%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

SNCA	100%	100%	100%	98%	Dementia, Lewy body, 127750 Parkinson disease 1, 168601 Parkinson disease 4, 605543	MENDELIOME PARKINSON
SNCB	100%	100%	100%	98%	Dementia, Lewy body, 127750	MENDELIOME
SNIP1	100%	100%	100%	98%	Neurodevelopmental disorder with hypotonia, craniofacial abnormalities, and seizures, 614501	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SNORA31	NC	NC	NC	NC	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
SNORD118	NC	NC	NC	NC	Leukoencephalopathy, brain calcifications, and cysts, 614561	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SNRNP200	100%	100%	100%	99%	Retinitis pigmentosa 33, 610359	VISION DISORDERS MENDELIOME
SNRPB	100%	100%	99%	96%	Cerebrocostomandibular syndrome, 117650	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS
SNRPE	100%	100%	100%	98%	Hypotrichosis 11, 615059	SKIN DISORDERS MENDELIOME
SNRPN	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
SNTA1	100%	100%	99%	95%	Long QT syndrome 12, 612955	HEART MENDELIOME
SNX10	100%	100%	100%	99%	Osteopetrosis, autosomal recessive 8, 615085	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
SNX14	100%	100%	100%	97%	Spinocerebellar ataxia, autosomal recessive 20, 616354	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SNX27	100%	100%	100%	97%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
SOBP	100%	99%	99%	92%	?Impaired intellectual development, anterior maxillary protrusion, and strabismus, 613671	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

SOCS1	100%	100%	100%	94%	Autoinflammatory syndrome, familial, w/wo immunodeficiency, 619375	PRIMARY IMMUNODEFICIENCY MENDELIOME
SOCS4	100%	100%	100%	98%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
SOD1	100%	100%	100%	99%	Spastic tetraplegia and axial hypotonia, progressive, 618598 Amyotrophic lateral sclerosis 1, 105400	ALS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SOD2	100%	100%	100%	99%	No OMIM disease ID	HEART METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS
SOHLH1	100%	100%	100%	99%	Ovarian dysgenesis 5, 617690 Spermatogenic failure 32, 618115	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRIMARY OVARIAN INSUFFICIENCY
SON	100%	100%	100%	99%	ZTTK syndrome, 617140	INTELLECTUAL DISABILITY MENDELIOME SCHISIS
SORD	92%	89%	97%	89%	Sorbitol dehydrogenase deficiency with peripheral neuropathy, 618912	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING
SOS1	100%	100%	100%	96%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300	HEREDITARY BONE MARROW FAILURE CONGENITAL HEART DISEASE SKIN DISORDERS HEART HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME AND RASOPATHY TUMOR
SOS2	100%	100%	100%	98%	Noonan syndrome 9, 616559	HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME AND RASOPATHY
SOST	100%	100%	100%	99%	Sclerosteosis 1, 269500 Craniodiaphyseal dysplasia, autosomal dominant, 122860	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING

SOX10	100%	100%	100%	98%	Waardenburg syndrome, type 4C, 613266 PCWH syndrome, 609136 Waardenburg syndrome, type 2E, w/wo neurologic involvement, 611584	MOVEMENT DISORDERS CRANIOFACIAL ANOMALIES SKIN DISORDERS HEARING IMPAIRMENT DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM NEUROPATHIES LIVER DISORDERS INTELLECTUAL DISABILITY MENDELIOME
SOX11	100%	100%	100%	91%	Intellectual developmental disorder with microcephaly and w/wo ocular malformations or hypogonadotropic hypogonadism, 615866	INTELLECTUAL DISABILITY MENDELIOME
SOX17	100%	100%	100%	99%	Vesicoureteral reflux 3, 613674	RENAL DISORDERS MENDELIOME
SOX18	99%	98%	100%	92%	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823 Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SOX2	100%	100%	99%	95%	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 Microphthalmia, syndromic 3, 206900	VISION DISORDERS SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
SOX3	100%	100%	94%	62%	Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000	DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
SOX4	100%	100%	99%	81%	Coffin-Siris syndrome 10, 618506	INTELLECTUAL DISABILITY MENDELIOME
SOX5	100%	99%	100%	98%	Lamb-Shaffer syndrome, 616803	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME
SOX6	99%	99%	100%	98%	Tolchin-Le Caignec syndrome, 618971	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME

SOX9	100%	100%	100%	98%	Campomelic dysplasia with autosomal sex reversal, 114290 Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290	CRANIOFACIAL ANOMALIES DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS
SP110	100%	99%	100%	98%	Hepatic venoocclusive disease with immunodeficiency, 235550	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
SP7	100%	100%	100%	99%	Osteogenesis imperfecta, type XII, 613849	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
SPAG1	100%	100%	100%	96%	Ciliary dyskinesia, primary, 28, 615505	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING
SPAG17	100%	99%	100%	98%	?Spermatogenic failure 55, 619380	MENDELIOME
SPAG6	100%	100%	100%	99%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
SPARC	100%	100%	100%	99%	Osteogenesis imperfecta, type XVII, 616507	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
SPART	100%	100%	100%	98%	Troyer syndrome, 275900	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
SPAST	100%	100%	99%	93%	Spastic paraplegia 4, autosomal dominant, 182601	MOVEMENT DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME
SPATA16	100%	100%	100%	98%	?Spermatogenic failure 6, 102530	MALE INFERTILITY MENDELIOME
SPATA22	100%	100%	100%	98%	No OMIM disease ID	DISORDERS OF SEX DEVELOPMENT MALE INFERTILITY MENDELIOME PRIMARY OVARIAN INSUFFICIENCY

SPATA5	100%	100%	100%	98%	Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577	HEARING IMPAIRMENT EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
SPATA5L1	100%	100%	100%	97%	Deafness, autosomal recessive 119, 619615 Neurodevelopmental disorder with hearing loss and spasticity, 619616	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SPATA7	100%	100%	100%	97%	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa 94, variable age at onset, autosomal recessive, 604232	VISION DISORDERS CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING
SPECC1L	100%	100%	100%	98%	Teebi hypertelorism syndrome 1, 145420 ?Facial clefting, oblique, 1, 600251	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS
SPEF2	100%	100%	100%	98%	Spermatogenic failure 43, 618751	MALE INFERTILITY MENDELIOME
SPEG	100%	100%	100%	98%	Centronuclear myopathy 5, 615959	MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
SPEN	100%	100%	100%	97%	Radio-Tartaglia syndrome, 619312	INTELLECTUAL DISABILITY MENDELIOME
SPG11	100%	100%	100%	98%	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360	ALS MOVEMENT DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SPG21	100%	100%	100%	98%	Mast syndrome, 248900	MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SPG7	100%	100%	100%	98%	Spastic paraplegia 7, autosomal recessive, 607259	MOVEMENT DISORDERS VISION DISORDERS NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING

SPI1	100%	100%	100%	99%	Agammaglobulinemia 10, autosomal dominant, 619707	PRIMARY IMMUNODEFICIENCY MENDELIOME
SPIDR	100%	100%	100%	98%	Ovarian dysgenesis 9, 619665	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRIMARY OVARIAN INSUFFICIENCY
SPINK1	99%	99%	100%	98%	Tropical calcific pancreatitis, 608189 Pancreatitis, hereditary, 167800	MENDELIOME TUMOR
SPINK2	96%	96%	100%	97%	?Spermatogenic failure 29, 618091	MALE INFERTILITY MENDELIOME
SPINK5	100%	100%	100%	97%	Netherton syndrome, 256500	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
SPINT2	100%	100%	100%	98%	Diarrhea 3, secretory sodium, congenital, syndromic, 270420	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SPNS2	100%	99%	99%	95%	?Deafness, autosomal recessive 115, 618457	HEARING IMPAIRMENT MENDELIOME
SPO11	100%	100%	100%	97%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
SPOCK1	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
SPOP	100%	100%	100%	97%	Nabais Sa-de Vries syndrome, type 1, 618828 Nabais Sa-de Vries syndrome, type 2, 618829	INTELLECTUAL DISABILITY MENDELIOME
SPP2	100%	100%	100%	98%	No OMIM disease ID	VISION DISORDERS MENDELIOME
SPPL2A	100%	100%	100%	98%	Immunodeficiency 86, mycobacteriosis, 619549	PRIMARY IMMUNODEFICIENCY MENDELIOME
SPR	100%	100%	100%	99%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716	MOVEMENT DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

SPRED1	100%	100%	100%	98%	Legius syndrome, 611431	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME NOONAN SYNDROME AND RASOPATHY TUMOR
SPRED2	100%	100%	100%	99%	Noonan syndrome 14, 619745	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING NOONAN SYNDROME AND RASOPATHY
SPRTN	100%	100%	100%	98%	Ruijs-Aalfs syndrome, 616200	MENDELIOME PRE CONCEPTION SCREENING
SPRY4	100%	100%	100%	99%	Hypogonadotropic hypogonadism 17 w/wo anosmia, 615266	SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME
SPTA1	100%	99%	100%	98%	Spherocytosis, type 3, 270970 Elliptocytosis-2, 130600 Pyropoikilocytosis, 266140	MENDELIOME PRE CONCEPTION SCREENING
SPTAN1	100%	100%	100%	98%	Developmental and epileptic encephalopathy 5, 613477	MOVEMENT DISORDERS EPILEPSY NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME
SPTB	100%	100%	100%	99%	Anemia, neonatal hemolytic, fatal or near-fatal, 617948 Elliptocytosis-3, 617948 Spherocytosis, type 2, 616649	MENDELIOME PRE CONCEPTION SCREENING
SPTBN1	100%	100%	100%	98%	Developmental delay, impaired speech, and behavioral abnormalities, 619475	INTELLECTUAL DISABILITY MENDELIOME
SPTBN2	100%	99%	100%	99%	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia, autosomal recessive 14, 615386	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SPTBN4	100%	100%	100%	98%	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519	NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING

SPTLC1	100%	100%	100%	98%	Amyotrophic lateral sclerosis 27, juvenile, 620285 Neuropathy, hereditary sensory and autonomic, type IA, 162400	NEUROPATHIES HNPD METABOLIC DISORDERS MENDELIOME
SPTLC2	100%	100%	100%	98%	Neuropathy, hereditary sensory and autonomic, type IC, 613640	NEUROPATHIES HNPD METABOLIC DISORDERS MENDELIOME
SPTLC3	99%	98%	100%	99%	No OMIM disease ID	NEUROPATHIES MENDELIOME
SPTSSA	100%	100%	100%	86%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME
SQOR	100%	100%	100%	98%	Sulfide:quinone oxidoreductase deficiency, 619221	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS
SQSTM1	100%	100%	100%	99%	Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 Myopathy, distal, with rimmed vacuoles, 617158 Paget disease of bone 3, 167250	ALS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING TUMOR
SRC	100%	100%	100%	99%	?Thrombocytopenia 6, 616937 Colon cancer, advanced, somatic, 114500	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME
SRCAP	100%	100%	100%	98%	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities, 619595 Floating-Harbor syndrome, 136140	DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
SRD5A2	100%	100%	100%	99%	Pseudovaginal perineoscrotal hypospadias, 264600	DISORDERS OF SEX DEVELOPMENT METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SRD5A3	100%	100%	100%	97%	Kahrizi syndrome, 612713 Congenital disorder of glycosylation, type Iq, 612379	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SREBF1	100%	100%	99%	98%	Ichthyosis, follicular, with atrichia and photophobia syndrome 2, 619016 Mucoepithelial dysplasia, hereditary, 158310	SKIN DISORDERS MENDELIOME
SRF	100%	100%	100%	95%	No OMIM disease ID	CONGENITAL HEART DISEASE HEART MENDELIOME

SRI	100%	100%	100%	98%	No OMIM disease ID	HEART MENDELIOME
SRP54	100%	100%	100%	99%	Neutropenia, severe congenital, 8, autosomal dominant, 618752	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
SRP72	100%	100%	100%	98%	Bone marrow failure syndrome 1, 614675	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCY MENDELIOME
SRPK3	100%	99%	99%	78%	No OMIM disease ID	MENDELIOME MUSCLE DISORDERS
SRPX2	100%	99%	97%	73%	?Rolandic epilepsy, impaired intellectual development, and speech dyspraxia, 300643	INTELLECTUAL DISABILITY MENDELIOME
SRRM2	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
SRY	50%	50%	46%	19%	46XY sex reversal 1, 400044	DISORDERS OF SEX DEVELOPMENT MALE INFERTILITY MENDELIOME
SSBP1	100%	100%	100%	98%	Optic atrophy 13 with retinal and foveal abnormalities, 165510	VISION DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS
SSR4	100%	99%	97%	72%	Congenital disorder of glycosylation, type Iy, 300934	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME
SSTR5	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME
SSX1	100%	99%	96%	66%	Spermatogenic failure, X-linked, 5, 301099	MENDELIOME
SSX2	100%	100%	98%	73%	?Sarcoma, synovial, 300813	MENDELIOME
ST14	100%	100%	100%	99%	Ichthyosis, congenital, autosomal recessive 11, 602400	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ST3GAL3	97%	95%	100%	99%	Developmental and epileptic encephalopathy 15, 615006 Intellectual developmental disorder, autosomal recessive 12, 611090	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

ST3GAL5	98%	98%	100%	97%	Salt and pepper developmental regression syndrome, 609056	SKIN DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
STAB2	100%	100%	100%	98%	No OMIM disease ID	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME
STAC3	100%	100%	100%	98%	Congenital myopathy 13, 255995	FETAL AKINESIA MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING SCHISIS
STAG1	100%	100%	100%	97%	Intellectual developmental disorder, autosomal dominant 47, 617635	INTELLECTUAL DISABILITY MENDELIOME
STAG2	100%	100%	97%	71%	Holoprosencephaly 13, X-linked, 301043 Mullegama-Klein-Martinez syndrome, 301022	INTELLECTUAL DISABILITY MENDELIOME
STAG3	100%	100%	100%	98%	Spermatogenic failure 61, 619672 Premature ovarian failure 8, 615723	DISORDERS OF SEX DEVELOPMENT MALE INFERTILITY MENDELIOME PRIMARY OVARIAN INSUFFICIENCY
STAMBP	100%	100%	100%	99%	Microcephaly-capillary malformation syndrome, 614261	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS
STAR	100%	100%	100%	99%	Lipoid adrenal hyperplasia, 201710	DISORDERS OF SEX DEVELOPMENT METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
STARD7	100%	100%	100%	98%	Epilepsy, familial adult myoclonic, 2, 607876	MENDELIOME
STAT1	96%	95%	100%	99%	Immunodeficiency 31C, chronic mucocutaneous candidiasis, autosomal dominant, 614162 Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
STAT2	100%	100%	100%	99%	Pseudo-TORCH syndrome 3, 618886 Immunodeficiency 44, 616636	PRIMARY IMMUNODEFICIENCY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING

STAT3	100%	100%	100%	98%	Hyper-IgE recurrent infection syndrome, 147060 Autoimmune disease, multisystem, infantile-onset, 1, 615952	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
STAT4	100%	100%	100%	97%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
STAT5B	100%	100%	100%	98%	Growth hormone insensitivity with immune dysregulation 1, autosomal recessive, 245590 Growth hormone insensitivity with immune dysregulation 2, autosomal dominant, 618985 Leukemia, acute promyelocytic, somatic, 102578	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
STAT6	100%	100%	100%	99%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
STEAP3	100%	100%	100%	99%	?Anemia, hypochromic microcytic, with iron overload 2, 615234	IRON DISORDERS MENDELIOME
STIL	100%	100%	100%	98%	Microcephaly 7, primary, autosomal recessive, 612703	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
STIM1	100%	100%	100%	99%	Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070 Immunodeficiency 10, 612783	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
STING1	100%	100%	100%	97%	STING-associated vasculopathy, infantile-onset, 615934	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME
STK11	100%	100%	100%	98%	Melanoma, malignant, somatic, 155600 Pancreatic cancer, somatic, 260350 Peutz-Jeghers syndrome, 175200 Testicular tumor, somatic, 273300	SKIN DISORDERS MENDELIOME TUMOR
STK36	100%	100%	100%	98%	?Ciliary dyskinesia, primary, 46, 619436	CILIOPATHIES MENDELIOME

STK4	100%	100%	100%	99%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)
STN1	100%	100%	100%	98%	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341	HEREDITARY BONE MARROW FAILURE LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING
STOX1	98%	97%	91%	82%	Preeclampsia/eclampsia 4, 609404	MENDELIOME
STRA6	100%	100%	100%	98%	Microphthalmia, syndromic 9, 601186 Microphthalmia, isolated, with coloboma 8, 601186	VISION DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
STRADA	100%	100%	100%	98%	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
STRC	100%	100%	100%	98%	Deafness, autosomal recessive 16, 603720	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
STS	96%	96%	98%	72%	Ichthyosis, X-linked, 308100	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME
STT3A	100%	100%	100%	99%	Congenital disorder of glycosylation, type Iw, autosomal dominant, 619714 Congenital disorder of glycosylation, type Iw, autosomal recessive, 615596	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
STT3B	100%	100%	99%	95%	Congenital disorder of glycosylation, type Ix, 615597	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
STUB1	100%	100%	100%	97%	Spinocerebellar ataxia 48, 618093 Spinocerebellar ataxia, autosomal recessive 16, 615768	MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING
STX11	100%	100%	100%	99%	Hemophagocytic lymphohistiocytosis, familial, 4, 603552	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
STX16	100%	100%	100%	98%	Pseudohypoparathyroidism, type IB, 603233	RENAL DISORDERS MENDELIOME

STX1A	100%	100%	100%	97%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
STX1B	100%	100%	100%	97%	Generalized epilepsy with febrile seizures plus, type 9, 616172	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
STX3	100%	100%	100%	98%	Retinal dystrophy and microvillus inclusion disease, 619446 Diarrhea 12, with microvillus atrophy, 619445	VISION DISORDERS MENDELIOME
STX5	100%	100%	100%	98%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME
STXBP1	100%	100%	100%	98%	Developmental and epileptic encephalopathy 4, 612164	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS
STXBP2	100%	99%	100%	99%	Hemophagocytic lymphohistiocytosis, familial, 5, w/wo microvillus inclusion disease, 613101	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
SUCLA2	100%	99%	100%	98%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic w/wo methylmalonic aciduria), 612073	HEARING IMPAIRMENT METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
SUCLG1	100%	100%	100%	96%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
SUCLG2	100%	99%	100%	97%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS TUMOR
SUFU	100%	100%	99%	98%	Joubert syndrome 32, 617757 Basal cell nevus syndrome, 109400	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SONIC HEDGEHOG MEDULLOBLASTOMA TUMOR
SUGCT	100%	99%	100%	98%	Glutaric aciduria III, 231690	METABOLIC DISORDERS MENDELIOME

SULF1	100%	100%	100%	99%	No OMIM disease ID	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
SULT2B1	100%	100%	99%	98%	Ichthyosis, congenital, autosomal recessive 14, 617571	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SUMF1	100%	100%	100%	99%	Multiple sulfatase deficiency, 272200	MOVEMENT DISORDERS SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SUMO1	71%	71%	100%	97%	?Orofacial cleft 10, 613705	CRANIOFACIAL ANOMALIES MENDELIOME
SUN5	100%	100%	100%	99%	Spermatogenic failure 16, 617187	MALE INFERTILITY MENDELIOME
SUOX	100%	100%	100%	99%	Sulfite oxidase deficiency, 272300	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SUPT16H	100%	100%	100%	98%	Neurodevelopmental disorder with dysmorphic facies and thin corpus callosum, 619480	INTELLECTUAL DISABILITY MENDELIOME
SUPV3L1	100%	100%	100%	98%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
SURF1	100%	100%	100%	98%	Charcot-Marie-Tooth disease, type 4K, 616684 Mitochondrial complex IV deficiency, nuclear type 1, 220110	HEART NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
SUZ12	100%	100%	100%	95%	Imagawa-Matsumoto syndrome, 618786	INTELLECTUAL DISABILITY MENDELIOME
SVBP	100%	100%	100%	96%	Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SVIL	100%	100%	100%	98%	Myofibrillar myopathy 10, 619040	MENDELIOME

SYCE1	100%	100%	100%	99%	?Spermatogenic failure 15, 616950 ?Premature ovarian failure 12, 616947	DISORDERS OF SEX DEVELOPMENT MALE INFERTILITY MENDELIOME
SYCP2	100%	100%	100%	96%	Spermatogenic failure 1, 258150	MALE INFERTILITY MENDELIOME
SYCP3	100%	100%	100%	97%	Pregnancy loss, recurrent, 4, 270960 Spermatogenic failure 4, 270960	MALE INFERTILITY MENDELIOME
SYK	100%	100%	100%	99%	Immunodeficiency 82 with systemic inflammation, 619381	PRIMARY IMMUNODEFICIENCY MENDELIOME
SYN1	100%	100%	96%	66%	Epilepsy, X-linked 1, with variable learning disabilities and behavior disorders, 300491 Intellectual developmental disorder, X-linked 50, 300115	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
SYNCRIP	100%	100%	100%	98%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
SYNE1	99%	99%	100%	98%	Arthrogryposis multiplex congenita 3, myogenic type, 618484 Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 Spinocerebellar ataxia, autosomal recessive 8, 610743	FETAL AKINESIA MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING
SYNE2	100%	100%	100%	98%	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999	MENDELIOME
SYNE4	100%	100%	100%	98%	Deafness, autosomal recessive 76, 615540	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
SYNGAP1	100%	100%	100%	96%	Intellectual developmental disorder, autosomal dominant 5, 612621	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
SYNJ1	100%	100%	100%	98%	Parkinson disease 20, early-onset, 615530 Developmental and epileptic encephalopathy 53, 617389	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
SYP	100%	99%	98%	71%	Intellectual developmental disorder, X-linked 96, 300802	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
SYT1	100%	99%	100%	98%	Baker-Gordon syndrome, 618218	INTELLECTUAL DISABILITY MENDELIOME
SYT14	100%	100%	100%	98%	?Spinocerebellar ataxia, autosomal recessive 11, 614229	MENDELIOME PRE CONCEPTION SCREENING
SYT2	100%	100%	100%	99%	Myasthenic syndrome, congenital, 7A, presynaptic, and distal motor neuropathy, autosomal dominant, 616040 Myasthenic syndrome, congenital, 7B, presynaptic, autosomal recessive, 619461	NEUROPATHIES MENDELIOME MUSCLE DISORDERS

SZT2	100%	100%	100%	99%	Developmental and epileptic encephalopathy 18, 615476	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
TAB2	100%	100%	100%	98%	Congenital heart defects, nonsyndromic, 2, 614980	CONGENITAL HEART DISEASE HEART SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
TAC3	100%	100%	100%	99%	Hypogonadotropic hypogonadism 10 w/wo anosmia, 614839	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME PRE CONCEPTION SCREENING
TACO1	100%	100%	100%	98%	Mitochondrial complex IV deficiency, nuclear type 8, 619052	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
TACR3	100%	99%	100%	98%	Hypogonadotropic hypogonadism 11 w/wo anosmia, 614840	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME PRE CONCEPTION SCREENING
TACSTD2	100%	100%	100%	99%	Corneal dystrophy, gelatinous drop-like, 204870	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
TAF1	100%	99%	97%	69%	Intellectual developmental disorder, X-linked syndromic 33, 300966 Dystonia-Parkinsonism, X-linked, 314250	MOVEMENT DISORDERS CONGENITAL HEART DISEASE HEART INTELLECTUAL DISABILITY MENDELIOME PARKINSON
TAF13	100%	100%	100%	96%	Intellectual developmental disorder, autosomal recessive 60, 617432	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TAF1A	100%	100%	100%	97%	No OMIM disease ID	HEART MENDELIOME
TAF1C	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TAF2	100%	100%	100%	98%	Intellectual developmental disorder, autosomal recessive 40, 615599	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

TAF4	89%	84%	93%	73%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
TAF4B	100%	100%	100%	97%	?Spermatogenic failure 13, 615841	MALE INFERTILITY MENDELIOME
TAF6	100%	100%	100%	99%	Alazami-Yuan syndrome, 617126	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TAF8	89%	89%	100%	98%	Neurodevelopmental disorder with severe motor impairment, absent language, cerebral hypomyelination, and brain atrophy, 619972	INTELLECTUAL DISABILITY MENDELIOME
TAL1	100%	100%	100%	94%	Leukemia, T-cell acute lymphocytic, somatic, 613065	MENDELIOME
TAL2	100%	100%	100%	99%	Leukemia, T-cell acute lymphocytic, somatic, 613065	MENDELIOME
TALDO1	100%	100%	100%	98%	Transaldolase deficiency, 606003	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
TAMM41	100%	100%	100%	98%	Combined oxidative phosphorylation deficiency 56, 620139	MENDELIOME MITOCHONDRIAL DISORDERS
TANC2	100%	100%	100%	99%	Intellectual developmental disorder with autistic features and language delay, w/wo seizures, 618906	INTELLECTUAL DISABILITY MENDELIOME
TANGO2	100%	100%	100%	99%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS
TAOK1	100%	100%	100%	98%	Developmental delay w/wo intellectual impairment or behavioral abnormalities, 619575	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS
TAP1	100%	100%	100%	98%	Bare lymphocyte syndrome, type I, 604571	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)

TAP2	100%	100%	100%	98%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)
TAPBP	95%	95%	99%	97%	Bare lymphocyte syndrome, type I, 604571	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)
TAPT1	100%	100%	99%	94%	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS
TARDBP	100%	100%	100%	99%	Frontotemporal lobar degeneration, TARDBP-related, 612069 Amyotrophic lateral sclerosis 10, w/wo FTD, 612069	ALS MENDELIOME
TARS1	100%	100%	100%	98%	Trichothiodystrophy 7, nonphotosensitive, 618546	MENDELIOME
TARS2	100%	100%	100%	98%	Combined oxidative phosphorylation deficiency 21, 615918	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
TASP1	99%	99%	100%	99%	Suleiman-El-Hattab syndrome, 618950	INTELLECTUAL DISABILITY MENDELIOME
TAT	100%	100%	100%	98%	Tyrosinemia, type II, 276600	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TAX1BP3	100%	100%	100%	98%	No OMIM disease ID	ANEURYSM MENDELIOME
TAZ	100%	100%	96%	67%	Barth syndrome, 302060	HEREDITARY BONE MARROW FAILURE HEART PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS

TBC1D20	100%	100%	100%	95%	Warburg micro syndrome 4, 615663	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TBC1D23	100%	100%	100%	98%	Pontocerebellar hypoplasia, type 11, 617695	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TBC1D24	100%	100%	100%	99%	Deafness, autosomal recessive 86, 614617 Epilepsy, rolandic, with paroxysmal exercise-induce dystonia and writer's cramp, 608105 Myoclonic epilepsy, infantile, familial, 605021 Deafness, autosomal dominant 65, 616044 Developmental and epileptic encephalopathy 16, 615338 DOORS syndrome, 220500	SKIN DISORDERS HEARING IMPAIRMENT EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TBC1D2B	99%	99%	100%	98%	Neurodevelopmental disorder with seizures and gingival overgrowth, 619323	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME
TBC1D32	100%	100%	100%	98%	No OMIM disease ID	CILIOPATHIES MENDELIOME
TBC1D7	100%	100%	100%	97%	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TBC1D8B	100%	99%	97%	72%	Nephrotic syndrome, type 20, 301028	RENAL DISORDERS MENDELIOME
TBCD	100%	100%	100%	99%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193	FETAL AKINESIA MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TBCE	100%	100%	100%	98%	Kenny-Caffey syndrome, type 1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207	EPILEPSY NEUROPATHIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TBCK	100%	100%	100%	98%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING

TBK1	100%	100%	100%	98%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439	ALS MENDELIOME
TBL1X	100%	99%	98%	73%	Hypothyroidism, congenital, nongoitrous, 8, 301033	HEARING IMPAIRMENT MENDELIOME
TBL1XR1	100%	100%	100%	98%	Intellectual developmental disorder, autosomal dominant 41, 616944 Pierpont syndrome, 602342	INTELLECTUAL DISABILITY MENDELIOME
TBL1Y	50%	49%	47%	21%	?Deafness, Y-linked 2, 400047	HEARING IMPAIRMENT MENDELIOME
TBP	100%	100%	100%	97%	Spinocerebellar ataxia 17, 607136	INTELLECTUAL DISABILITY MENDELIOME
TBR1	100%	100%	100%	96%	Intellectual developmental disorder with autism and speech delay, 606053	INTELLECTUAL DISABILITY MENDELIOME
TBX1	97%	95%	99%	83%	Tetralogy of Fallot, 187500 DiGeorge syndrome, 188400 Conotruncal anomaly face syndrome, 217095 Velocardiofacial syndrome, 192430	CRANIOFACIAL ANOMALIES CONGENITAL HEART DISEASE HEART HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME SCHISIS
TBX15	100%	99%	100%	98%	Cousin syndrome, 260660	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS
TBX18	100%	100%	100%	98%	Congenital anomalies of kidney and urinary tract 2, 143400	RENAL DISORDERS MENDELIOME
TBX19	100%	100%	100%	98%	Adrenocorticotropic hormone deficiency, 201400	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRE CONCEPTION SCREENING
TBX2	100%	99%	99%	92%	Vertebral anomalies and variable endocrine and T-cell dysfunction, 618223	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS
TBX20	100%	100%	100%	98%	Atrial septal defect 4, 611363	CONGENITAL HEART DISEASE HEART MENDELIOME
TBX21	100%	100%	100%	97%	Asthma and nasal polyps, 208550 ?Immunodeficiency 88, 619630	PRIMARY IMMUNODEFICIENCY MENDELIOME
TBX22	99%	98%	98%	72%	Cleft palate with ankyloglossia, 303400 ?Abruzzo-Erickson syndrome, 302905	CRANIOFACIAL ANOMALIES MENDELIOME SCHISIS

TBX3	100%	100%	100%	98%	Ulnar-mammary syndrome, 181450	SKIN DISORDERS DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
TBX4	100%	100%	100%	98%	Ischiocoxopodopatellar syndrome w/wo pulmonary arterial hypertension, 147891 Amelia, posterior, with pelvic and pulmonary hypoplasia syndrome, 601360	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS
TBX5	100%	100%	100%	99%	Holt-Oram syndrome, 142900	CONGENITAL HEART DISEASE HEART SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
TBX6	100%	100%	100%	99%	Spondylocostal dysostosis 5, 122600	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
TBXA2R	99%	99%	100%	99%	No OMIM disease ID	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME
TBXAS1	100%	100%	100%	98%	Ghosal hematodiaphyseal syndrome, 231095	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
TBXT	100%	100%	100%	98%	Sacral agenesis with vertebral anomalies, 615709	MENDELIOME PRE CONCEPTION SCREENING
TCAP	100%	100%	100%	99%	Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954	HEART MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
TCF12	100%	100%	100%	98%	Craniosynostosis 3, 615314 Hypogonadotropic hypogonadism 26 w/wo anosmia, 619718	CRANIOFACIAL ANOMALIES DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
TCF20	100%	100%	100%	99%	Developmental delay with variable intellectual impairment and behavioral abnormalities, 618430	INTELLECTUAL DISABILITY MENDELIOME

TCF3	100%	100%	100%	98%	Agammaglobulinemia 8B, autosomal recessive, 619824 Agammaglobulinemia 8A, autosomal dominant, 616941	PRIMARY IMMUNODEFICIENCY MENDELIOME
TCF4	100%	100%	100%	98%	Pitt-Hopkins syndrome, 610954 Corneal dystrophy, Fuchs endothelial, 3, 613267	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
TCF7L2	100%	100%	99%	94%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
TCHH	100%	100%	99%	88%	?Uncombable hair syndrome 3, 617252	SKIN DISORDERS MENDELIOME
TCIRG1	100%	100%	100%	99%	Osteopetrosis, autosomal recessive 1, 259700	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
TCN2	100%	100%	100%	98%	Transcobalamin II deficiency, 275350	PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TCOF1	100%	100%	100%	99%	Treacher Collins syndrome 1, 154500	CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS
TCTEX1D2	100%	100%	100%	94%	Short-rib thoracic dysplasia 17 w/wo polydactyly, 617405	CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
TCTN1	95%	94%	100%	97%	Joubert syndrome 13, 614173	VISION DISORDERS CILIOPATHIES RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING

TCTN2	100%	100%	100%	99%	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
TCTN3	100%	100%	100%	98%	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860	VISION DISORDERS CILIOPATHIES DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS
TDGF1	100%	100%	100%	98%	No OMIM disease ID	CONGENITAL HEART DISEASE HEART MENDELIOME
TDP1	100%	100%	100%	99%	?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING
TDP2	100%	100%	100%	97%	Spinocerebellar ataxia, autosomal recessive 23, 616949	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
TDRD7	100%	100%	100%	99%	Cataract 36, 613887	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
TDRD9	100%	100%	100%	98%	?Spermatogenic failure 30, 618110	MALE INFERTILITY MENDELIOME
TDRKH	100%	100%	100%	99%	No OMIM disease ID	NEUROPATHIES MENDELIOME
TEAD1	100%	100%	100%	99%	Sveinsson chorioretinal atrophy, 108985	VISION DISORDERS MENDELIOME

TECPR2	100%	100%	100%	98%	Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031	MOVEMENT DISORDERS NEUROPATHIES HNPD INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TECR	100%	100%	100%	99%	Intellectual developmental disorder, autosomal recessive 14, 614020	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TECRL	100%	100%	100%	97%	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021	HEART MENDELIOME PRE CONCEPTION SCREENING
TECTA	100%	100%	100%	99%	Deafness, autosomal dominant 8/12, 601543 Deafness, autosomal recessive 21, 603629	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
TEFM	100%	100%	100%	98%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
TEK	100%	99%	100%	98%	Venous malformations, multiple cutaneous and mucosal, 600195 Glaucoma 3, primary congenital, E, 617272	VISION DISORDERS SKIN DISORDERS MENDELIOME
TELO2	100%	100%	100%	99%	You-Hoover-Fong syndrome, 616954	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TENM1	99%	99%	98%	74%	No OMIM disease ID	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM MENDELIOME
TENM3	100%	100%	100%	99%	Microphthalmia, syndromic 15, 615145 ?Microphthalmia, isolated, with coloboma 9, 615145	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TENM4	100%	100%	100%	99%	Essential tremor, hereditary, 5, 616736	MOVEMENT DISORDERS MENDELIOME
TENT5A	100%	100%	100%	96%	Osteogenesis imperfecta, type XVIII, 617952	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
TERB1	100%	100%	100%	97%	Spermatogenic failure 60, 619646	MALE INFERTILITY MENDELIOME
TERB2	100%	100%	100%	96%	?Spermatogenic failure 59, 619645	MALE INFERTILITY MENDELIOME

TERC	NC	NC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS DYSKERATOSIS CONGENITA PRIMARY IMMUNODEFICIENCY LIVER DISORDERS MENDELIOME TUMOR
TERF2IP	99%	96%	100%	98%	No OMIM disease ID	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS MENDELIOME TUMOR
TERT	100%	100%	100%	99%	Dyskeratosis congenita, autosomal dominant 2, 613989 Dyskeratosis congenita, autosomal recessive 4, 613989 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1, 614742	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS DYSKERATOSIS CONGENITA PRIMARY IMMUNODEFICIENCY LIVER DISORDERS MELA2 MENDELIOME TUMOR
TES	100%	100%	100%	99%	No OMIM disease ID	ANEURYSM MENDELIOME
TET2	100%	99%	100%	98%	Myelodysplastic syndrome, somatic, 614286 Immunodeficiency 75, 619126	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCY MENDELIOME
TET3	100%	100%	100%	98%	Beck-Fahrner syndrome, 618798	INTELLECTUAL DISABILITY MENDELIOME
TEX11	97%	96%	97%	70%	Spermatogenic failure, X-linked 2, 309120	MALE INFERTILITY MENDELIOME
TEX14	100%	100%	100%	98%	Spermatogenic failure 23, 617707	MALE INFERTILITY MENDELIOME
TEX15	100%	100%	100%	97%	Spermatogenic failure 25, 617960	MALE INFERTILITY MENDELIOME
TF	100%	100%	100%	99%	Atransferrinemia, 209300	IRON DISORDERS MENDELIOME PRE CONCEPTION SCREENING
TFAM	100%	100%	100%	98%	?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING

TFAP2A	100%	100%	99%	93%	Branchiooculofacial syndrome, 113620	VISION DISORDERS CRANIOFACIAL ANOMALIES SKIN DISORDERS HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME SCHISIS
TFAP2B	100%	100%	100%	98%	Patent ductus arteriosus 2, 617035 Char syndrome, 169100	CONGENITAL HEART DISEASE HEART MENDELIOME
TFB2M	100%	100%	100%	97%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
TFE3	100%	99%	97%	71%	Intellectual developmental disorder, X-linked syndromic, with pigmentary mosaicism and coarse facies, 301066 Renal cell carcinoma, papillary, 1, 300854	INTELLECTUAL DISABILITY MENDELIOME
TFG	100%	100%	100%	98%	?Spastic paraplegia 57, autosomal recessive, 615658 Hereditary motor and sensory neuropathy, Okinawa type, 604484	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING
TFPT	100%	100%	100%	97%	No OMIM disease ID	VISION DISORDERS MENDELIOME
TFR2	100%	100%	100%	97%	Hemochromatosis, type 3, 604250	IRON DISORDERS LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING
TFRC	100%	100%	100%	98%	Immunodeficiency 46, 616740	IRON DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
TG	100%	100%	100%	99%	Thyroid dyshormonogenesis 3, 274700	MENDELIOME PRE CONCEPTION SCREENING TUMOR
TGDS	100%	100%	100%	97%	Catel-Manzke syndrome, 616145	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS
TGFB1	100%	100%	100%	97%	Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213 Camurati-Engelmann disease, 131300	PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING

TGFB2	100%	100%	100%	98%	Loeys-Dietz syndrome 4, 614816	ANEURYSM SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
TGFB3	100%	100%	100%	99%	Arrhythmogenic right ventricular dysplasia 1, 107970 Loeys-Dietz syndrome 5, 615582	ANEURYSM HEART SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS
TGFBI	100%	100%	100%	99%	Corneal dystrophy, Avellino type, 607541 Corneal dystrophy, Reis-Bucklers type, 608470 Corneal dystrophy, Thiel-Behnke type, 602082 Corneal dystrophy, Groenouw type I, 121900 Corneal dystrophy, epithelial basement membrane, 121820 Corneal dystrophy, lattice type I, 122200 Corneal dystrophy, lattice type IIIA, 608471	VISION DISORDERS MENDELIOME
TGFBR1	100%	100%	100%	96%	Loeys-Dietz syndrome 1, 609192	ANEURYSM CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS
TGFBR2	100%	100%	100%	98%	Loeys-Dietz syndrome 2, 610168 Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239	ANEURYSM CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS
TGIF1	100%	100%	100%	98%	Holoprosencephaly 4, 142946	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME SCHISIS
TGM1	100%	100%	100%	99%	Ichthyosis, congenital, autosomal recessive 1, 242300	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
TGM3	100%	100%	100%	99%	?Uncombable hair syndrome 2, 617251	SKIN DISORDERS MENDELIOME

TGM5	100%	100%	100%	98%	Peeling skin syndrome 2, 609796	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
TGM6	100%	100%	100%	99%	Spinocerebellar ataxia 35, 613908	MOVEMENT DISORDERS MENDELIOME
TH	100%	100%	100%	98%	Segawa syndrome, recessive, 605407	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PARKINSON PRE CONCEPTION SCREENING
THAP1	100%	100%	100%	99%	Dystonia 6, torsion, 602629	MOVEMENT DISORDERS MENDELIOME
THBD	100%	100%	100%	97%	Thrombophilia 12 due to thrombomodulin defect, 614486	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY RENAL DISORDERS MENDELIOME
THBS4	100%	100%	100%	99%	No OMIM disease ID	HEART MENDELIOME
THG1L	100%	100%	100%	98%	Spinocerebellar ataxia, autosomal recessive 28, 618800	MENDELIOME MITOCHONDRIAL DISORDERS
THOC1	100%	100%	100%	96%	?Deafness, autosomal dominant 86, 620280	HEARING IMPAIRMENT MENDELIOME
THOC2	100%	100%	97%	70%	Intellectual developmental disorder, X-linked 12, 300957	INTELLECTUAL DISABILITY MENDELIOME
THOC6	100%	100%	100%	99%	Beaulieu-Boycott-Innes syndrome, 613680	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
THPO	100%	100%	100%	98%	Thrombocythemia 1, 187950	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME TUMOR
THRA	100%	100%	100%	99%	Hypothyroidism, congenital, nongoitrous, 6, 614450	MENDELIOME
THRB	100%	100%	100%	98%	Thyroid hormone resistance, autosomal recessive, 274300 Thyroid hormone resistance, 188570 Thyroid hormone resistance, selective pituitary, 145650	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
THSD1	100%	100%	100%	98%	?Aneurysm, intracranial berry, 12, 618734 Lymphatic malformation 13, 620244	ANEURYSM MENDELIOME

THSD4	100%	100%	100%	99%	Aortic aneurysm, familial thoracic 12, 619825	ANEURYSM MENDELIOME
THUMPD1	100%	99%	100%	96%	Neurodevelopmental disorder with speech delay and variable ocular anomalies, 619989	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TIA1	100%	100%	99%	95%	Welander distal myopathy, 604454 Amyotrophic lateral sclerosis 26 w/wo frontotemporal dementia, 619133	MENDELIOME
TIAM1	100%	100%	100%	98%	Neurodevelopmental disorder with language delay and seizures, 619908	INTELLECTUAL DISABILITY MENDELIOME
TICAM1	100%	100%	100%	99%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
TIE1	100%	100%	100%	99%	Lymphatic malformation 11, 619401	MENDELIOME
TIMM22	100%	100%	100%	99%	?Combined oxidative phosphorylation deficiency 43, 618851	MENDELIOME MITOCHONDRIAL DISORDERS
TIMM44	100%	100%	100%	98%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
TIMM50	100%	100%	100%	99%	3-methylglutaconic aciduria, type IX, 617698	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
TIMM8A	100%	99%	97%	67%	Mohr-Tranebjaerg syndrome, 304700	MOVEMENT DISORDERS VISION DISORDERS HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS
TIMMDC1	100%	100%	100%	97%	Mitochondrial complex I deficiency, nuclear type 31, 618251	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
TIMP3	100%	100%	100%	98%	Sorsby fundus dystrophy, 136900	VISION DISORDERS MENDELIOME
TINF2	100%	100%	100%	98%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS DYSKERATOSIS CONGENITA PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME TUMOR
TIRAP	100%	100%	100%	99%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME

TJP1	100%	100%	100%	98%	No OMIM disease ID	HEART MENDELIOME
TJP2	100%	100%	99%	98%	Hypercholanemia, familial 1, 607748 Cholestasis, progressive familial intrahepatic 4, 615878	HEARING IMPAIRMENT LIVER DISORDERS MENDELIOME PRE CONCEPTION SCREENING
TK2	100%	100%	100%	98%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560 ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS
TKFC	100%	100%	100%	99%	Triokinase and FMN cyclase deficiency syndrome, 618805	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME
TKT	98%	98%	100%	99%	Short stature, developmental delay, and congenital heart defects, 617044	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TLCD3B	100%	100%	100%	97%	Cone-rod dystrophy 22, 619531	VISION DISORDERS MENDELIOME
TLE6	100%	100%	100%	98%	Preimplantation embryonic lethality, 616814	MENDELIOME PRE CONCEPTION SCREENING
TLK2	100%	100%	100%	98%	Intellectual developmental disorder, autosomal dominant 57, 618050	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME
TLL1	99%	98%	100%	98%	Atrial septal defect 6, 613087	CONGENITAL HEART DISEASE HEART MENDELIOME
TLR3	100%	100%	100%	97%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
TLR4	100%	99%	100%	98%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
TLR5	100%	100%	100%	98%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
TLR7	100%	100%	97%	67%	Immunodeficiency 74, COVID19-related, X-linked, 301051 Systemic lupus erythematosus 17, 301080	PRIMARY IMMUNODEFICIENCY MENDELIOME
TLR8	100%	100%	97%	69%	Immunodeficiency 98 with autoinflammation, X-linked, 301078	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCY MENDELIOME
TMC1	100%	100%	100%	96%	Deafness, autosomal dominant 36, 606705 Deafness, autosomal recessive 7, 600974	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING

TMC6	100%	100%	100%	99%	Epidermodysplasia verruciformis, 226400	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
TMC8	100%	100%	100%	99%	Epidermodysplasia verruciformis 2, 618231	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
TMCO1	88%	87%	100%	97%	Craniofacial dysmorphism, skeletal anomalies, and impaired intellectual development 1, 213980	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS
TMCO3	100%	100%	100%	99%	No OMIM disease ID	VISION DISORDERS MENDELIOME
TMEM106B	100%	100%	100%	98%	Leukodystrophy, hypomyelinating, 16, 617964	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME
TMEM107	100%	100%	100%	98%	Orofaciodigital syndrome XVI, 617563 Meckel syndrome 13, 617562 ?Joubert syndrome 29, 617562	CILIOPATHIES RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
TMEM126A	100%	100%	100%	97%	Optic atrophy 7, 612989	VISION DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
TMEM126B	100%	100%	100%	99%	Mitochondrial complex I deficiency, nuclear type 29, 618250	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
TMEM127	100%	100%	100%	98%	No OMIM disease ID	MENDELIOME TUMOR
TMEM132E	100%	100%	100%	99%	Deafness, autosomal recessive 99, 618481	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
TMEM138	100%	100%	100%	99%	Joubert syndrome 16, 614465	VISION DISORDERS CILIOPATHIES RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING

TMEM147	100%	100%	100%	99%	Neurodevelopmental disorder with facial dysmorphism, absent language, and pseudo-Pelger-Huet anomaly, 620075	INTELLECTUAL DISABILITY MENDELIOME
TMEM14C	100%	100%	100%	98%	No OMIM disease ID	IRON DISORDERS MENDELIOME
TMEM163	100%	100%	100%	97%	Leukodystrophy, hypomyelinating, 25, 620243	INTELLECTUAL DISABILITY MENDELIOME
TMEM165	100%	100%	100%	97%	Congenital disorder of glycosylation, type IIk, 614727	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TMEM186	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
TMEM199	100%	100%	100%	98%	Congenital disorder of glycosylation, type IIp, 616829	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
TMEM216	100%	100%	100%	98%	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS
TMEM218	100%	100%	100%	98%	Joubert syndrome 39, 619562	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY MENDELIOME
TMEM222	100%	100%	99%	97%	Neurodevelopmental disorder with motor and speech delay and behavioral abnormalities, 619470	INTELLECTUAL DISABILITY MENDELIOME
TMEM231	100%	100%	100%	99%	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING

TMEM237	100%	100%	99%	97%	Joubert syndrome 14, 614424	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
TMEM240	100%	100%	99%	90%	Spinocerebellar ataxia 21, 607454	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME
TMEM251	100%	100%	100%	99%	Dysostosis multiplex, Ain-Naz type, 619345	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
TMEM260	100%	100%	100%	98%	Structural heart defects and renal anomalies syndrome, 617478	CONGENITAL HEART DISEASE CILIOPATHIES HEART RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
TMEM38B	100%	100%	100%	98%	Osteogenesis imperfecta, type XIV, 615066	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
TMEM43	100%	100%	100%	98%	Arrhythmogenic right ventricular dysplasia 5, 604400 Auditory neuropathy, autosomal dominant 3, 619832 Emery-Dreifuss muscular dystrophy 7, AD, 614302	ARRHYTHMOGENIC CARDIOMYOPATHY (ACM/ARVC) HEARING IMPAIRMENT HEART MENDELIOME ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS
TMEM53	100%	100%	100%	99%	Craniotubular dysplasia, Ikegawa type, 619727	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
TMEM63A	100%	100%	100%	98%	Leukodystrophy, hypomyelinating, 19, transient infantile, 618688	INTELLECTUAL DISABILITY MENDELIOME
TMEM63C	100%	100%	100%	98%	Spastic paraplegia 87, autosomal recessive, 619966	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS
TMEM65	100%	98%	99%	91%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS

TMEM67	99%	97%	100%	95%	Nephronophthisis 11, 613550 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 ?RHYNS syndrome, 602152 COACH syndrome 1, 216360	MOVEMENT DISORDERS VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
TMEM70	100%	100%	100%	97%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
TMEM94	100%	100%	100%	99%	Intellectual developmental disorder with cardiac defects and dysmorphic facies, 618316	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TMEM98	100%	100%	100%	99%	Nanophthalmos 4, 615972	VISION DISORDERS MENDELIOME
TMIE	100%	100%	100%	99%	Deafness, autosomal recessive 6, 600971	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
TMLHE	100%	99%	98%	80%	No OMIM disease ID	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME
TMPO	100%	100%	100%	98%	No OMIM disease ID	HEART MENDELIOME
TMPRSS15	100%	100%	100%	98%	Enterokinase deficiency, 226200	MENDELIOME PRE CONCEPTION SCREENING
TMPRSS3	100%	100%	100%	99%	Deafness, autosomal recessive 8/10, 601072	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
TMPRSS6	100%	100%	100%	99%	Iron-refractory iron deficiency anemia, 206200	IRON DISORDERS MENDELIOME PRE CONCEPTION SCREENING
TMTC2	97%	97%	100%	99%	No OMIM disease ID	HEARING IMPAIRMENT MENDELIOME
TMTC3	100%	99%	99%	97%	Lissencephaly 8, 617255	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

TMX2	100%	100%	100%	99%	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS
TNC	100%	100%	100%	99%	Deafness, autosomal dominant 56, 615629	HEARING IMPAIRMENT MENDELIOME
TNFAIP3	100%	100%	100%	99%	Autoinflammatory syndrome, familial, Behcet-like 1, 616744	PRIMARY IMMUNODEFICIENCY MENDELIOME
TNFRSF10B	100%	100%	100%	98%	Squamous cell carcinoma, head and neck, 275355	MENDELIOME
TNFRSF11A	100%	99%	99%	98%	Osteopetrosis, autosomal recessive 7, 612301 Osteolysis, familial expansile, 174810	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING TUMOR
TNFRSF11B	100%	100%	100%	98%	Paget disease of bone 5, juvenile-onset, 239000	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
TNFRSF13B	100%	100%	100%	99%	Immunodeficiency, common variable, 2, 240500 Immunoglobulin A deficiency 2, 609529	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
TNFRSF13C	100%	100%	100%	94%	Immunodeficiency, common variable, 4, 613494	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
TNFRSF1A	92%	92%	100%	99%	Periodic fever, familial, 142680	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME
TNFRSF4	100%	100%	100%	98%	?Immunodeficiency 16, 615593	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
TNFRSF9	100%	100%	100%	98%	Immunodeficiency 109 with lymphoproliferation, 620282	PRIMARY IMMUNODEFICIENCY MENDELIOME
TNFSF11	100%	100%	100%	98%	Osteopetrosis, autosomal recessive 2, 259710	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
TNFSF12	100%	100%	100%	97%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME

TNFSF13	100%	100%	100%	98%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
TNIK	100%	100%	100%	98%	Intellectual developmental disorder, autosomal recessive 54, 617028	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TNNC1	100%	100%	100%	98%	Cardiomyopathy, dilated, 1Z, 611879 Cardiomyopathy, hypertrophic, 13, 613243	DILATED CARDIOMYOPATHY HEART HYPERTROPHIC CARDIOMYOPATHY MENDELIOME
TNNI2	100%	100%	100%	99%	Arthrogryposis, distal, type 2B1, 601680	FETAL AKINESIA MENDELIOME MUSCLE DISORDERS
TNNI3	100%	100%	100%	97%	?Cardiomyopathy, dilated, 2A, 611880 Cardiomyopathy, hypertrophic, 7, 613690 Cardiomyopathy, familial restrictive, 1, 115210 Cardiomyopathy, dilated, 1FF, 613286	DILATED CARDIOMYOPATHY HEART HYPERTROPHIC CARDIOMYOPATHY MENDELIOME PRE CONCEPTION SCREENING
TNNI3K	100%	100%	100%	98%	Cardiac conduction disease w/wo dilated cardiomyopathy, 616117	CONGENITAL HEART DISEASE HEART MENDELIOME
TNNT1	100%	100%	100%	97%	Nemaline myopathy 5, Amish type, 605355	MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
TNNT2	100%	100%	100%	98%	Cardiomyopathy, dilated, 1D, 601494 Cardiomyopathy, hypertrophic, 2, 115195 Cardiomyopathy, familial restrictive, 3, 612422 Left ventricular noncompaction 6, 601494	DILATED CARDIOMYOPATHY HEART HYPERTROPHIC CARDIOMYOPATHY MENDELIOME ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS
TNNT3	100%	100%	100%	99%	Arthrogryposis, distal, type 2B2, 618435	FETAL AKINESIA MENDELIOME
TNPO2	100%	100%	100%	98%	Intellectual developmental disorder with hypotonia, impaired speech, and dysmorphic facies, 619556	INTELLECTUAL DISABILITY MENDELIOME
TNPO3	100%	100%	100%	99%	Muscular dystrophy, limb-girdle, autosomal dominant 2, 608423	MENDELIOME MUSCLE DISORDERS
TNR	100%	100%	100%	99%	Neurodevelopmental disorder, nonprogressive, with spasticity and transient opisthotonus, 619653	INTELLECTUAL DISABILITY MENDELIOME
TNRC6A	100%	100%	100%	98%	?Epilepsy, familial adult myoclonic, 6, 618074	MENDELIOME
TNRC6B	100%	100%	100%	98%	Global developmental delay with speech and behavioral abnormalities, 619243	INTELLECTUAL DISABILITY MENDELIOME MUSCLE DISORDERS

TNS1	100%	100%	100%	99%	No OMIM disease ID	HEART MENDELIOME
TNS2	100%	100%	100%	99%	No OMIM disease ID	RENAL DISORDERS MENDELIOME
TNXB	100%	100%	100%	98%	Ehlers-Danlos syndrome, classic-like, 1, 606408 Vesicoureteral reflux 8, 615963	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
TOE1	100%	100%	100%	99%	Pontocerebellar hypoplasia, type 7, 614969	MOVEMENT DISORDERS DISORDERS OF SEX DEVELOPMENT EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TOGARAM1	100%	100%	100%	97%	Joubert syndrome 37, 619185	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY MENDELIOME
TOM1	100%	100%	100%	98%	?Immunodeficiency 85 and autoimmunity, 619510	PRIMARY IMMUNODEFICIENCY MENDELIOME
TOMM70	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS
TONSL	100%	100%	100%	99%	Spondyloepimetaphyseal dysplasia, sponastrime type, 271510	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
TOP1	100%	100%	100%	98%	DNA topoisomerase I, camptothecin-resistant,	MENDELIOME
TOP2A	100%	100%	100%	97%	DNA topoisomerase II, resistance to inhibition of, by amsacrine,	MENDELIOME
TOP2B	100%	100%	100%	97%	B-cell immunodeficiency, distal limb anomalies, and urogenital malformations, 609296	PRIMARY IMMUNODEFICIENCY MENDELIOME
TOP3A	100%	100%	100%	98%	?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098 Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
TOPORS	100%	100%	100%	98%	Retinitis pigmentosa 31, 609923	VISION DISORDERS CILIOPATHIES MENDELIOME

TOR1A	91%	90%	100%	96%	Arthrogryposis multiplex congenita 5, 618947 Dystonia-1, torsion, 128100	FETAL AKINESIA MOVEMENT DISORDERS HNPD INTELLECTUAL DISABILITY MENDELIOME
TOR1AIP1	100%	100%	100%	96%	?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072	HEART MENDELIOME PRE CONCEPTION SCREENING
TP53	94%	94%	100%	97%	Hepatocellular carcinoma, somatic, 114550 Breast cancer, somatic, 114480 Li-Fraumeni syndrome, 151623 Pancreatic cancer, somatic, 260350 Nasopharyngeal carcinoma, somatic, 607107 Bone marrow failure syndrome 5, 618165	HEREDITARY BONE MARROW FAILURE MENDELIOME SONIC HEDGEHOG MEDULLOBLASTOMA TUMOR
TP53RK	100%	100%	100%	98%	Galloway-Mowat syndrome 4, 617730	INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
TP63	100%	99%	100%	99%	Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 Hay-Wells syndrome, 106260 Split-hand/foot malformation 4, 605289 Orofacial cleft 8, 618149 Rapp-Hodgkin syndrome, 129400 ADULT syndrome, 103285 Limb-mammary syndrome, 603543	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS
TP73	100%	100%	100%	99%	Ciliary dyskinesia, primary, 47, and lissencephaly, 619466	CILIOPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TPCN2	100%	100%	100%	99%	No OMIM disease ID	SKIN DISORDERS MENDELIOME
TPI1	100%	100%	100%	98%	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TPK1	100%	100%	100%	98%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING

TPM1	100%	100%	100%	98%	Left ventricular noncompaction 9, 611878 Cardiomyopathy, hypertrophic, 3, 115196 Cardiomyopathy, dilated, 1Y, 611878	DILATED CARDIOMYOPATHY HEART HYPERTROPHIC CARDIOMYOPATHY MENDELIOME
TPM2	100%	100%	100%	99%	Arthrogryposis, distal, type 2B4, 108120 Arthrogryposis, distal, type 1A, 108120 Nemaline myopathy 4, autosomal dominant, 609285 CAP myopathy 2, 609285	FETAL AKINESIA MENDELIOME MUSCLE DISORDERS
TPM3	100%	100%	100%	98%	Congenital myopathy 4A, autosomal dominant, 255310 Congenital myopathy 4B, autosomal recessive, 609284	FETAL AKINESIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
TPM4	100%	100%	99%	97%	No OMIM disease ID	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME
TPMT	100%	100%	100%	98%	No OMIM disease ID	METABOLIC DISORDERS MENDELIOME
TPO	100%	100%	100%	99%	Thyroid dyshormonogenesis 2A, 274500	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TPP1	100%	100%	100%	99%	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270	MOVEMENT DISORDERS VISION DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TPP2	100%	100%	100%	98%	Immunodeficiency 78 with autoimmunity and developmental delay, 619220	PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME
TPRKB	82%	81%	100%	98%	Galloway-Mowat syndrome 5, 617731	INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
TPRN	97%	95%	97%	80%	Deafness, autosomal recessive 79, 613307	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
TRA2B	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
TRAC	100%	100%	100%	99%	Immunodeficiency 7, TCR-alpha/beta deficient, 615387	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING

TRAF3	100%	100%	100%	98%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
TRAF3IP1	100%	100%	100%	96%	Senior-Loken syndrome 9, 616629	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA LIVER DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
TRAF3IP2	100%	100%	100%	98%	?Candidiasis, familial, 8, 615527	PRIMARY IMMUNODEFICIENCY MENDELIOME
TRAF6	100%	100%	100%	99%	No OMIM disease ID	CRANIOFACIAL ANOMALIES MENDELIOME
TRAF7	100%	100%	100%	99%	Cardiac, facial, and digital anomalies with developmental delay, 618164	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
TRAIP	100%	100%	100%	99%	Seckel syndrome 9, 616777	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TRAK1	100%	100%	100%	99%	Developmental and epileptic encephalopathy 68, 618201	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TRAPPC11	100%	100%	100%	98%	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
TRAPPC12	100%	100%	100%	99%	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TRAPPC2	100%	100%	98%	72%	Spondyloepiphyseal dysplasia tarda, 313400	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME

TRAPPC2L	100%	100%	100%	99%	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
TRAPPC4	100%	100%	100%	97%	Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy, 618741	INTELLECTUAL DISABILITY MENDELIOME
TRAPPC6B	100%	100%	100%	98%	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TRAPPC9	100%	100%	100%	98%	Intellectual developmental disorder, autosomal recessive 13, 613192	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TRDN	99%	99%	100%	96%	Cardiac arrhythmia syndrome, w/wo skeletal muscle weakness, 615441	HEART LONG QT SYNDROME MENDELIOME PRE CONCEPTION SCREENING ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS
TREH	100%	100%	100%	99%	Trehalase deficiency, 612119	METABOLIC DISORDERS MENDELIOME
TREM2	100%	100%	100%	98%	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193	MOVEMENT DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
TREX1	100%	100%	100%	99%	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448	MOVEMENT DISORDERS VISION DISORDERS SKIN DISORDERS EPILEPSY HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TRH	100%	100%	100%	98%	No OMIM disease ID	MENDELIOME PRE CONCEPTION SCREENING
TRHR	100%	100%	100%	97%	Hypothyroidism, congenital, nongoitrous, 7, 618573	MENDELIOME
TRIM2	93%	93%	100%	98%	Charcot-Marie-Tooth disease, type 2R, 615490	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING

TRIM22	100%	100%	100%	99%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
TRIM28	100%	100%	100%	97%	No OMIM disease ID	MENDELIOME TUMOR
TRIM32	100%	100%	100%	99%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110	VISION DISORDERS CILIOPATHIES SKIN DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
TRIM36	100%	100%	100%	98%	?Anencephaly 1, 206500	MENDELIOME
TRIM37	98%	98%	100%	98%	Mulibrey nanism, 253250	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING SCHISIS TUMOR
TRIM44	100%	100%	100%	97%	?Aniridia 3, 617142	MENDELIOME
TRIM63	100%	100%	100%	98%	No OMIM disease ID	HEART MENDELIOME
TRIM71	100%	100%	99%	97%	Hydrocephalus, congenital, 4, 618667	MALE INFERTILITY MENDELIOME
TRIM8	100%	100%	100%	98%	Focal segmental glomerulosclerosis and neurodevelopmental syndrome, 619428	INTELLECTUAL DISABILITY MENDELIOME
TRIO	99%	99%	99%	97%	Intellectual developmental disorder, autosomal dominant 44, with microcephaly, 617061 Intellectual developmental disorder, autosomal dominant 63, with macrocephaly, 618825	INTELLECTUAL DISABILITY MENDELIOME
TRIOBP	100%	100%	100%	98%	Deafness, autosomal recessive 28, 609823	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
TRIP11	100%	100%	100%	97%	Odontochondrodysplasia 1, 184260 Achondrogenesis, type IA, 200600	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
TRIP12	100%	100%	100%	98%	Intellectual developmental disorder, autosomal dominant 49, 617752	INTELLECTUAL DISABILITY MENDELIOME

TRIP13	100%	100%	100%	98%	Oocyte maturation defect 9, 619011 Mosaic variegated aneuploidy syndrome 3, 617598	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING TUMOR
TRIP4	100%	100%	100%	98%	?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066 Spinal muscular atrophy with congenital bone fractures 1, 616866	FETAL AKINESIA MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
TRIT1	100%	100%	100%	98%	Combined oxidative phosphorylation deficiency 35, 617873	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
TRMT1	100%	100%	100%	98%	Intellectual developmental disorder, autosomal recessive 68, 618302	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TRMT10A	100%	100%	100%	98%	Microcephaly, short stature, and impaired glucose metabolism 1, 616033	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TRMT10C	100%	100%	100%	98%	Combined oxidative phosphorylation deficiency 30, 616974	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
TRMT5	100%	100%	100%	98%	Peripheral neuropathy with variable spasticity, exercise intolerance, and developmental delay, 616539	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
TRMU	100%	100%	100%	98%	Liver failure, transient infantile, 613070	LIVER DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
TRNT1	100%	100%	100%	98%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 Retinitis pigmentosa and erythrocytic microcytosis, 616959	VISION DISORDERS IRON DISORDERS PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
TRPA1	100%	100%	100%	98%	?Episodic pain syndrome, familial, 1, 615040	HNPD MENDELIOME
TRPC3	100%	100%	100%	98%	?Spinocerebellar ataxia 41, 616410	MENDELIOME

TRPC6	100%	100%	100%	98%	Glomerulosclerosis, focal segmental, 2, 603965	RENAL DISORDERS MENDELIOME
TRPM1	100%	100%	100%	99%	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
TRPM3	100%	100%	100%	98%	?Cataract 50 w/wo glaucoma, 620253 Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skeletal anomalies, w/wo seizures, 620224	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
TRPM4	100%	100%	100%	99%	Progressive familial heart block, type IB, 604559 Erythrokeratodermia variabilis et progressiva 6, 618531	SKIN DISORDERS HEART MENDELIOME
TRPM6	100%	100%	100%	98%	Hypomagnesemia 1, intestinal, 602014	EPILEPSY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
TRPM7	100%	100%	100%	98%	No OMIM disease ID	HNPD MENDELIOME
TRPM8	100%	100%	100%	99%	No OMIM disease ID	HNPD MENDELIOME
TRPS1	100%	99%	100%	98%	Trichorhinophalangeal syndrome, type III, 190351 Trichorhinophalangeal syndrome, type I, 190350	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
TRPV1	100%	100%	100%	98%	No OMIM disease ID	HNPD MENDELIOME
TRPV3	100%	100%	100%	99%	?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400 Olmsted syndrome 1, 614594	SKIN DISORDERS HNPD MENDELIOME
TRPV4	100%	100%	100%	99%	Spondylometaphyseal dysplasia, Kozlowski type, 184252 Digital arthropathy-brachydactyly, familial, 606835 SED, Maroteaux type, 184095 Metatropic dysplasia, 156530 Scapuloperoneal spinal muscular atrophy, 181405 Hereditary motor and sensory neuropathy, type IIc, 606071 ?Avascular necrosis of femoral head, primary, 2, 617383 Neuronopathy, distal hereditary motor, type VIII, 600175 Parastremmatic dwarfism, 168400 Brachyolmia type 3, 113500	FETAL AKINESIA NEUROPATHIES HNPD SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME MUSCLE DISORDERS
TRPV6	100%	100%	100%	99%	Hyperparathyroidism, transient neonatal, 618188	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME

TRRAP	100%	100%	100%	98%	?Deafness, autosomal dominant 75, 618778 Developmental delay w/wo dysmorphic facies and autism, 618454	HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME
TSC1	100%	100%	100%	98%	Focal cortical dysplasia, type II, somatic, 607341 Tuberous sclerosis-1, 191100 Lymphangioleiomyomatosis, 606690	CONGENITAL HEART DISEASE SKIN DISORDERS EPILEPSY HEART INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME TUMOR
TSC2	100%	100%	100%	99%	Lymphangioleiomyomatosis, somatic, 606690 ?Focal cortical dysplasia, type II, somatic, 607341 Tuberous sclerosis-2, 613254	SKIN DISORDERS EPILEPSY INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME TUMOR
TSEN15	100%	100%	99%	98%	Pontocerebellar hypoplasia, type 2F, 617026	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TSEN2	100%	100%	100%	98%	Pontocerebellar hypoplasia type 2B, 612389	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TSEN34	100%	100%	100%	98%	?Pontocerebellar hypoplasia type 2C, 612390	MENDELIOME PRE CONCEPTION SCREENING
TSEN54	100%	100%	100%	98%	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753 ?Pontocerebellar hypoplasia type 5, 610204	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TSFM	94%	94%	100%	98%	Combined oxidative phosphorylation deficiency 3, 610505	HEART INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
TSGA10	100%	100%	100%	97%	?Spermatogenic failure 26, 617961	MALE INFERTILITY MENDELIOME

TSHB	100%	100%	100%	99%	Hypothyroidism, congenital, nongoitrous 4, 275100	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TSHR	100%	100%	100%	98%	Hyperthyroidism, familial gestational, 603373 Hyperthyroidism, nonautoimmune, 609152 Hypothyroidism, congenital, nongoitrous, 1, 275200 Thyroid adenoma, hyperfunctioning, somatic, Thyroid carcinoma with thyrotoxicosis, somatic,	MENDELIOME PRE CONCEPTION SCREENING
TSHZ1	100%	100%	99%	98%	Aural atresia, congenital, 607842	CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT MENDELIOME
TSPAN12	100%	100%	100%	98%	Exudative vitreoretinopathy 5, 613310	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
TSPAN7	99%	98%	97%	74%	Intellectual developmental disorder, X-linked 58, 300210	INTELLECTUAL DISABILITY MENDELIOME
TSPEAR	100%	100%	100%	98%	Tooth agenesis, selective, 10, 620173 ?Deafness, autosomal recessive 98, 614861 Ectodermal dysplasia 14, hair/tooth type w/wo hypohidrosis, 618180	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
TSPYL1	100%	100%	100%	97%	Sudden infant death with dysgenesis of the testes syndrome, 608800	DISORDERS OF SEX DEVELOPMENT MENDELIOME PRE CONCEPTION SCREENING
TSR2	100%	100%	97%	73%	?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946	HEREDITARY BONE MARROW FAILURE MENDELIOME
TTBK2	100%	100%	100%	98%	Spinocerebellar ataxia 11, 604432	MOVEMENT DISORDERS CILIOPATHIES MENDELIOME
TTC12	100%	100%	100%	99%	Ciliary dyskinesia, primary, 45, 618801	CILIOPATHIES MENDELIOME
TTC19	100%	100%	100%	97%	Mitochondrial complex III deficiency, nuclear type 2, 615157	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS
TTC21A	100%	100%	100%	99%	Spermatogenic failure 37, 618429	MENDELIOME

TTC21B	100%	99%	100%	98%	Short-rib thoracic dysplasia 4 w/wo polydactyly, 613819 Nephronophthisis 12, 613820	CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
TTC25	100%	100%	100%	98%	Ciliary dyskinesia, primary, 35, 617092	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING
TTC26	100%	100%	100%	98%	Biliary, renal, neurologic, and skeletal syndrome, 619534	CILIOPATHIES MENDELIOME
TTC29	99%	99%	100%	98%	Spermatogenic failure 42, 618745	MALE INFERTILITY MENDELIOME
TTC37	100%	100%	100%	98%	Trichohepatoenteric syndrome 1, 222470	SKIN DISORDERS PRIMARY IMMUNODEFICIENCY LIVER DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TTC5	100%	100%	100%	99%	Neurodevelopmental disorder with cerebral atrophy and variable facial dysmorphism, 619244	INTELLECTUAL DISABILITY MENDELIOME
TTC7A	100%	100%	100%	98%	Gastrointestinal defects and immunodeficiency syndrome, 243150	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)
TTC8	100%	99%	100%	98%	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
TTI1	100%	100%	100%	98%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
TTI2	100%	100%	100%	98%	Intellectual developmental disorder, autosomal recessive 39, 615541	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

TTLL5	100%	100%	100%	98%	Cone-rod dystrophy 19, 615860	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
TTN	99%	99%	100%	98%	Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807 Cardiomyopathy, familial hypertrophic, 9, 613765 Congenital myopathy 5 with cardiomyopathy, 611705 Tibial muscular dystrophy, tardive, 600334 Cardiomyopathy, dilated, 1G, 604145 Myopathy, myofibrillar, 9, with early respiratory failure, 603689	FETAL AKINESIA DILATED CARDIOMYOPATHY HEART MENDELIOME MUSCLE DISORDERS
TTPA	100%	100%	100%	98%	Ataxia with isolated vitamin E deficiency, 277460	MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING
TTR	90%	90%	100%	99%	Amyloidosis, hereditary, transthyretin-related, 105210 Carpal tunnel syndrome, familial, 115430	HEART HYPERTROPHIC CARDIOMYOPATHY NEUROPATHIES HNPD MENDELIOME PAINFUL PERIPHERAL NEUROPATHIES
TUB	100%	100%	100%	98%	?Retinal dystrophy and obesity, 616188	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
TUBA1A	100%	100%	100%	99%	Lissencephaly 3, 611603	FETAL AKINESIA MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
TUBA3D	100%	100%	100%	99%	Keratoconus 9, 617928	VISION DISORDERS MENDELIOME
TUBA4A	100%	100%	100%	99%	Amyotrophic lateral sclerosis 22 w/wo frontotemporal dementia, 616208	ALS MENDELIOME
TUBA8	100%	100%	100%	99%	Macrothrombocytopenia, isolated, 2, autosomal dominant, 619840	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TUBB	99%	98%	100%	99%	Symmetric circumferential skin creases, congenital, 1, 156610 Cortical dysplasia, complex, with other brain malformations 6, 615771	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME SCHISIS
TUBB1	100%	100%	100%	99%	Macrothrombocytopenia, isolated, 1, autosomal dominant, 613112	HEREDITARY BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME

TUBB2A	100%	100%	100%	99%	Cortical dysplasia, complex, with other brain malformations 5, 615763	EPILEPSY NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME
TUBB2B	100%	100%	100%	99%	Cortical dysplasia, complex, with other brain malformations 7, 610031	FETAL AKINESIA EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
TUBB3	100%	100%	100%	99%	Fibrosis of extraocular muscles, congenital, 3A, 600638 Cortical dysplasia, complex, with other brain malformations 1, 614039	VISION DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME MUSCLE DISORDERS
TUBB4A	98%	95%	100%	98%	Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
TUBB4B	100%	100%	100%	98%	Leber congenital amaurosis with early-onset deafness, 617879	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME
TUBB6	100%	100%	100%	99%	?Facial palsy, congenital, with ptosis and velopharyngeal dysfunction, 617732	MENDELIOME
TUBB8	100%	100%	100%	99%	Oocyte maturation defect 2, 616780	MENDELIOME
TUBG1	100%	100%	100%	98%	Cortical dysplasia, complex, with other brain malformations 4, 615412	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
TUBGCP2	96%	96%	100%	99%	Pachygyria, microcephaly, developmental delay, and dysmorphic facies, w/wo seizures, 618737	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TUBGCP4	100%	100%	100%	97%	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TUBGCP6	100%	100%	100%	99%	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TUFM	100%	100%	100%	98%	Combined oxidative phosphorylation deficiency 4, 610678	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING

TULP1	100%	100%	100%	98%	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132	VISION DISORDERS CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING
TULP3	100%	100%	100%	99%	Hepatorenocardiac degenerative fibrosis, 619902	HEART MENDELIOME
TUSC3	100%	100%	100%	98%	Intellectual developmental disorder, autosomal recessive 7, 611093	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
TWIST1	100%	100%	99%	91%	Craniosynostosis 1, 123100 Robinow-Sorauf syndrome, 180750 Sweeney-Cox syndrome, 617746 Saethre-Chotzen syndrome w/wo eyelid anomalies, 101400	CRANIOFACIAL ANOMALIES SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS
TWIST2	100%	100%	100%	94%	Ablepharon-macrostomia syndrome, 200110 Barber-Say syndrome, 209885 Focal facial dermal dysplasia 3, Setleis type, 227260	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
TWNK	100%	100%	100%	99%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 Perrault syndrome 5, 616138	MOVEMENT DISORDERS VISION DISORDERS HEARING IMPAIRMENT DISORDERS OF SEX DEVELOPMENT NEUROPATHIES LIVER DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING PRIMARY OVARIAN INSUFFICIENCY MUSCLE DISORDERS
TXN2	100%	100%	100%	99%	?Combined oxidative phosphorylation deficiency 29, 616811	MENDELIOME MITOCHONDRIAL DISORDERS
TXNDC15	100%	100%	100%	99%	Meckel syndrome 14, 619879	CILIOPATHIES MENDELIOME
TXNL4A	100%	100%	100%	98%	Burn-McKeown syndrome, 608572	MENDELIOME PRE CONCEPTION SCREENING SCHISIS
TXNRD2	100%	100%	100%	99%	?Glucocorticoid deficiency 5, 617825	DISORDERS OF SEX DEVELOPMENT HEART MENDELIOME

TYK2	100%	100%	100%	99%	Immunodeficiency 35, 611521	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
TYMP	100%	100%	100%	98%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041	NEUROPATHIES LIVER DISORDERS METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
TYMS	100%	100%	100%	96%	Dyskeratosis congenita, digenic, 620040	METABOLIC DISORDERS MENDELIOME
TYR	100%	99%	100%	98%	Albinism, oculocutaneous, type IB, 606952 Waardenburg syndrome/albinism, digenic, 103470 Albinism, oculocutaneous, type IA, 203100	VISION DISORDERS SKIN DISORDERS HEARING IMPAIRMENT METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
TYROBP	100%	100%	100%	98%	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770	MOVEMENT DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
TYRP1	100%	100%	100%	99%	Albinism, oculocutaneous, type III, 203290	VISION DISORDERS SKIN DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
U2AF2	100%	100%	99%	95%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
UBA1	100%	99%	99%	74%	Spinal muscular atrophy, X-linked 2, infantile, 301830 VEXAS syndrome, somatic, 301054	FETAL AKINESIA HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCY MENDELIOME MUSCLE DISORDERS
UBA2	100%	100%	100%	97%	ACCES syndrome, 619959	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME

UBA5	100%	100%	100%	97%	?Spinocerebellar ataxia, autosomal recessive 24, 617133 Developmental and epileptic encephalopathy 44, 617132	EPILEPSY NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
UBAP1	100%	100%	100%	96%	Spastic paraplegia 80, autosomal dominant, 618418	MOVEMENT DISORDERS MENDELIOME
UBAP2L	100%	100%	100%	98%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
UBB	100%	100%	100%	96%	No OMIM disease ID	CRANIOFACIAL ANOMALIES MENDELIOME
UBE2A	100%	100%	96%	71%	Intellectual developmental disorder, X-linked syndromic, Nascimento type, 300860	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME
UBE2T	100%	100%	100%	98%	Fanconi anemia, complementation group T, 616435	HEREDITARY BONE MARROW FAILURE MENDELIOME PRE CONCEPTION SCREENING
UBE3A	100%	100%	100%	98%	Angelman syndrome, 105830	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
UBE3B	100%	100%	100%	99%	Kaufman oculocerebrofacial syndrome, 244450	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
UBE4A	100%	100%	100%	98%	Neurodevelopmental disorder with hypotonia and gross motor and seech delay, 619639	INTELLECTUAL DISABILITY MENDELIOME
UBIAD1	100%	100%	100%	98%	Corneal dystrophy, Schnyder type, 121800	VISION DISORDERS MENDELIOME
UBQLN2	100%	100%	96%	66%	Amyotrophic lateral sclerosis 15, w/wo frontotemporal dementia, 300857	ALS MENDELIOME
UBR1	98%	98%	100%	98%	Johanson-Blizzard syndrome, 243800	SKIN DISORDERS LIVER DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
UBR2	100%	99%	100%	98%	No OMIM disease ID	MALE INFERTILITY MENDELIOME
UBR7	100%	100%	100%	98%	Li-Campeau syndrome, 619189	INTELLECTUAL DISABILITY MENDELIOME
UBTF	100%	100%	100%	97%	Neurodegeneration, childhood-onset, with brain atrophy, 617672	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME

UCHL1	100%	100%	100%	97%	Spastic paraplegia 79A, autosomal dominant, 620221 Spastic paraplegia 79B, autosomal recessive, 615491	MOVEMENT DISORDERS VISION DISORDERS NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING
UFC1	100%	100%	100%	99%	Neurodevelopmental disorder with spasticity and poor growth, 618076	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
UFM1	100%	100%	100%	99%	Leukodystrophy, hypomyelinating, 14, 617899	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
UFSP2	100%	100%	100%	98%	?Hip dysplasia, Beukes type, 142669 Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974 Developmental and epileptic encephalopathy 106, 620028	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
UGDH	100%	100%	100%	98%	Developmental and epileptic encephalopathy 84, 618792	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
UGP2	95%	94%	100%	98%	Developmental and epileptic encephalopathy 83, 618744	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
UGT1A1	100%	100%	100%	98%	Crigler-Najjar syndrome, type I, 218800 Hyperbilirubinemia, familial transient neonatal, 237900 Crigler-Najjar syndrome, type II, 606785	LIVER DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
UMOD	100%	100%	100%	99%	Tubulointerstitial kidney disease, autosomal dominant, 1, 162000	RENAL DISORDERS MENDELIOME
UMPS	100%	100%	100%	99%	Orotic aciduria, 258900	METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
UNC119	100%	100%	100%	96%	?Immunodeficiency 13, 615518 ?Cone-rod dystrophy,	MENDELIOME
UNC13A	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
UNC13D	100%	100%	100%	99%	Hemophagocytic lymphohistiocytosis, familial, 3, 608898	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
UNC45A	100%	100%	100%	98%	Osteootohepatoenteric syndrome, 619377	INTELLECTUAL DISABILITY MENDELIOME

UNC45B	100%	100%	100%	99%	?Cataract 43, 616279 Myofibrillar myopathy 11, 619178	VISION DISORDERS MENDELIOME MUSCLE DISORDERS
UNC80	100%	100%	100%	98%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
UNC93B1	100%	99%	99%	94%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
UNG	100%	100%	100%	99%	Immunodeficiency with hyper IgM, type 5, 608106	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
UPB1	100%	100%	100%	98%	Beta-ureidopropionase deficiency, 613161	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
UPF1	99%	98%	100%	98%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
UPF3B	100%	99%	96%	66%	Intellectual developmental disorder, X-linked syndromic 14, 300676	INTELLECTUAL DISABILITY MENDELIOME
UPK3A	100%	100%	100%	99%	No OMIM disease ID	RENAL DISORDERS MENDELIOME
UQCC1	100%	100%	100%	96%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
UQCC2	100%	100%	100%	99%	Mitochondrial complex III deficiency, nuclear type 7, 615824	RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
UQCC3	100%	100%	100%	97%	?Mitochondrial complex III deficiency, nuclear type 9, 616111	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
UQCR10	100%	100%	100%	97%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
UQCR11	100%	100%	100%	99%	No OMIM disease ID	MENDELIOME MITOCHONDRIAL DISORDERS
UQCRB	100%	100%	100%	98%	Mitochondrial complex III deficiency, nuclear type 3, 615158	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
UQCRC1	100%	100%	100%	99%	Parkinsonism with polyneuropathy, 619279	NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS

UQCRC2	100%	100%	100%	98%	Mitochondrial complex III deficiency, nuclear type 5, 615160	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
UQCRFS1	100%	100%	100%	99%	Mitochondrial complex III deficiency, nuclear type 10, 618775	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
UQCRH	100%	100%	100%	99%	?Mitochondrial complex III deficiency, nuclear type 11, 620137	MENDELIOME MITOCHONDRIAL DISORDERS
UQCRQ	100%	100%	100%	98%	Mitochondrial complex III deficiency, nuclear type 4, 615159	MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
UROC1	100%	100%	100%	99%	?Urocanase deficiency, 276880	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
UROD	100%	100%	100%	99%	Porphyria, hepatoerythropoietic, 176100 Porphyria cutanea tarda, 176100	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
UROS	100%	100%	100%	98%	Porphyria, congenital erythropoietic, 263700	SKIN DISORDERS IRON DISORDERS METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
USB1	100%	100%	100%	98%	Poikiloderma with neutropenia, 604173	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS DYSKERATOSIS CONGENITA PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING TUMOR
USH1C	100%	100%	100%	97%	Usher syndrome, type 1C, 276904 Deafness, autosomal recessive 18A, 602092	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
USH1G	100%	100%	100%	99%	Usher syndrome, type 1G, 606943	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING

USH2A	99%	99%	100%	99%	Usher syndrome, type 2A, 276901 Retinitis pigmentosa 39, 613809	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
USP18	100%	100%	100%	98%	Pseudo-TORCH syndrome 2, 617397	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
USP26	100%	100%	96%	63%	Spermatogenic failure, X-linked, 6, 301101	MALE INFERTILITY MENDELIOME
USP27X	100%	100%	98%	74%	Intellectual developmental disorder, X-linked 105, 300984	INTELLECTUAL DISABILITY MENDELIOME
USP45	100%	100%	100%	98%	?Leber congenital amaurosis 19, 618513	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
USP48	100%	100%	100%	97%	Deafness, autosomal dominant 85, 620227	HEARING IMPAIRMENT MENDELIOME
USP53	100%	100%	100%	97%	Cholestasis, progressive familial intrahepatic, 7, w/wo hearing loss, 619658	LIVER DISORDERS MENDELIOME
USP7	100%	99%	99%	97%	Hao-Fountain syndrome, 616863	INTELLECTUAL DISABILITY MENDELIOME
USP8	100%	100%	100%	97%	Pituitary adenoma 4, ACTH-secreting, somatic, 219090	MENDELIOME
USP9X	100%	99%	98%	73%	Intellectual developmental disorder, X-linked 99, 300919 Intellectual developmental disorder, X-linked 99, syndromic, female-restricted, 300968	INTELLECTUAL DISABILITY MENDELIOME SCHISIS
USP9Y	49%	49%	47%	22%	Spermatogenic failure, Y-linked, 2, 415000	MALE INFERTILITY MENDELIOME
UST	100%	100%	100%	98%	No OMIM disease ID	ANEURYSM MENDELIOME
UVSSA	100%	100%	100%	99%	UV-sensitive syndrome 3, 614640	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
VAC14	100%	100%	100%	99%	Striatonigral degeneration, childhood-onset, 617054	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
VAMP1	100%	100%	100%	99%	Myasthenic syndrome, congenital, 25, 618323 Spastic ataxia 1, autosomal dominant, 108600	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
VAMP2	100%	100%	100%	99%	Neurodevelopmental disorder with hypotonia and autistic features w/wo hyperkinetic movements, 618760	INTELLECTUAL DISABILITY MENDELIOME

VANGL1	100%	100%	100%	98%	Caudal regression syndrome, 600145	MENDELIOME
VANGL2	100%	100%	100%	99%	Neural tube defects, 182940	MENDELIOME
VAPB	100%	100%	100%	97%	Spinal muscular atrophy, late-onset, Finkel type, 182980 Amyotrophic lateral sclerosis 8, 608627	ALS MENDELIOME
VARS1	100%	100%	100%	98%	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
VARS2	100%	100%	100%	99%	Combined oxidative phosphorylation deficiency 20, 615917	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
VAV1	98%	98%	100%	99%	No OMIM disease ID	PRIMARY IMMUNODEFICIENCY MENDELIOME
VAX1	99%	99%	99%	85%	?Microphthalmia, syndromic 11, 614402	VISION DISORDERS CRANIOFACIAL ANOMALIES MENDELIOME PRE CONCEPTION SCREENING
VCAN	100%	100%	100%	98%	Wagner syndrome 1, 143200	VISION DISORDERS MENDELIOME
VCL	100%	100%	100%	98%	Cardiomyopathy, dilated, 1W, 611407 Cardiomyopathy, hypertrophic, 15, 613255	DILATED CARDIOMYOPATHY HEART MENDELIOME
VCP	100%	100%	100%	98%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 6, 613954 Charcot-Marie-Tooth disease, type 2Y, 616687 Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320	ALS MOVEMENT DISORDERS NEUROPATHIES MENDELIOME MUSCLE DISORDERS
VDR	100%	100%	100%	98%	Rickets, vitamin D-resistant, type IIA, 277440	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
VEGFC	100%	100%	100%	98%	Lymphatic malformation 4, 615907	SKIN DISORDERS MENDELIOME
VEZF1	100%	100%	99%	96%	?Cardiomyopathy, dilated, 1OO, 620247	HEART MENDELIOME

VHL	100%	100%	100%	99%	Erythrocytosis, familial, 2, 263400 von Hippel-Lindau syndrome, 193300 Renal cell carcinoma, somatic, 144700 Pheochromocytoma, 171300 Hemangioblastoma, cerebellar, somatic,	VISION DISORDERS CILIOPATHIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING TUMOR
VIM	100%	100%	100%	97%	Cataract 30, pulverulent, 116300	VISION DISORDERS MENDELIOME
VIPAS39	100%	100%	100%	99%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404	FETAL AKINESIA HEMOSTATIC/THROMBOTIC DISORDERS LIVER DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
VKORC1	97%	92%	100%	98%	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473 Warfarin resistance, 122700	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
VLDLR	100%	100%	100%	99%	Cerebellar hypoplasia, impaired intellectual development, and dysequilibrium syndrome 1, 224050	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
VMA21	100%	100%	98%	72%	Myopathy, X-linked, with excessive autophagy, 310440	METABOLIC DISORDERS MENDELIOME MUSCLE DISORDERS
VPS11	100%	100%	100%	99%	?Dystonia 32, 619637 Leukodystrophy, hypomyelinating, 12, 616683	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
VPS13A	100%	100%	100%	97%	Choreoacanthocytosis, 200150	MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING
VPS13B	99%	99%	100%	98%	Cohen syndrome, 216550	VISION DISORDERS HEREDITARY BONE MARROW FAILURE SKIN DISORDERS PRIMARY IMMUNODEFICIENCY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

VPS13C	100%	100%	100%	98%	Parkinson disease 23, autosomal recessive, early onset, 616840	MENDELIOME PARKINSON PRE CONCEPTION SCREENING
VPS13D	100%	100%	100%	98%	Spinocerebellar ataxia, autosomal recessive 4, 607317	MOVEMENT DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
VPS16	100%	100%	100%	99%	Dystonia 30, 619291	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME
VPS33A	89%	89%	100%	96%	Mucopolysaccharidosis-plus syndrome, 617303	SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
VPS33B	100%	100%	100%	98%	Keratoderma-ichthyosis-deafness syndrome, autosomal recessive, 620009 Cholestasis, progressive familial intrahepatic, 12, 620010 Arthrogryposis, renal dysfunction, and cholestasis 1, 208085	FETAL AKINESIA SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS LIVER DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
VPS35	100%	100%	100%	98%	No OMIM disease ID	MENDELIOME PARKINSON
VPS35L	100%	100%	100%	98%	Ritscher-Schinzel syndrome 3, 619135	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME
VPS37A	100%	100%	100%	94%	Spastic paraplegia 53, autosomal recessive, 614898	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
VPS41	100%	99%	100%	98%	Spinocerebellar ataxia, autosomal recessive 29, 619389	INTELLECTUAL DISABILITY MENDELIOME
VPS45	95%	95%	100%	98%	Neutropenia, severe congenital, 5, autosomal recessive, 615285	HEREDITARY BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
VPS4A	100%	100%	100%	98%	CIMDAG syndrome, 619273	HEREDITARY BONE MARROW FAILURE INTELLECTUAL DISABILITY MENDELIOME

VPS50	100%	100%	100%	98%	Neurodevelopmental disorder with microcephaly, seizures, and neonatal cholestasis, 619685	INTELLECTUAL DISABILITY MENDELIOME
VPS51	100%	100%	100%	98%	Pontocerebellar hypoplasia, type 13, 618606	MENDELIOME PRE CONCEPTION SCREENING
VPS53	100%	100%	100%	98%	Pontocerebellar hypoplasia, type 2E, 615851	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
VRK1	100%	99%	100%	98%	Pontocerebellar hypoplasia type 1A, 607596	MOVEMENT DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
VSX1	100%	100%	100%	99%	?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195 Keratoconus 1, 148300	VISION DISORDERS MENDELIOME
VSX2	100%	100%	100%	99%	Microphthalmia, isolated 2, 610093 Microphthalmia with coloboma 3, 610092	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
VWA1	100%	100%	100%	98%	Neuropathy, hereditary motor, with myopathic features, 619216	NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING MUSCLE DISORDERS
VWA3B	100%	100%	100%	97%	?Spinocerebellar ataxia, autosomal recessive 22, 616948	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
VWF	100%	100%	100%	99%	von Willebrand disease, type 1, 193400 von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 von Willebrand disease, type 3, 277480	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
WAC	100%	100%	99%	97%	Desanto-Shinawi syndrome, 616708	INTELLECTUAL DISABILITY MENDELIOME
WARS1	100%	100%	100%	99%	Neuronopathy, distal hereditary motor, type IX, 617721	NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME
WARS2	100%	100%	100%	99%	Parkinsonism-dystonia 3, childhood-onset, 619738 Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, w/wo seizures, 617710	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING

WAS	100%	98%	97%	68%	Wiskott-Aldrich syndrome, 301000 Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, intermittent, 313900 Thrombocytopenia, X-linked, 313900	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME TUMOR
WASF1	100%	99%	100%	99%	Neurodevelopmental disorder with absent language and variable seizures, 618707	INTELLECTUAL DISABILITY MENDELIOME
WASHC4	100%	100%	100%	97%	Intellectual developmental disorder, autosomal recessive 43, 615817	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
WASHC5	100%	100%	100%	98%	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, autosomal dominant, 603563	MOVEMENT DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS
WBP11	100%	100%	100%	99%	Vertebral, cardiac, tracheoesophageal, renal, and limb defects, 619227	MENDELIOME
WBP2	100%	100%	100%	97%	Deafness, autosomal recessive 107, 617639	HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
WDFY3	100%	100%	100%	98%	?Microcephaly 18, primary, autosomal dominant, 617520	INTELLECTUAL DISABILITY MENDELIOME
WDPCP	97%	97%	100%	98%	?Bardet-Biedl syndrome 15, 615992 Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085	VISION DISORDERS CILIOPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
WDR1	100%	100%	100%	98%	Periodic fever, immunodeficiency, and thrombocytopenia syndrome, 150550	PRIMARY IMMUNODEFICIENCY MENDELIOME
WDR11	100%	100%	100%	98%	Intellectual developmental disorder, autosomal recessive 78, 620237 Hypogonadotropic hypogonadism 14 w/wo anosmia, 614858	DISORDERS OF SEX DEVELOPMENT HYPOGONADOTROPIC HYPOGONADISM INTELLECTUAL DISABILITY MENDELIOME
WDR13	100%	99%	99%	79%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME

WDR19	100%	100%	100%	97%	Nephronophthisis 13, 614377 Cranioectodermal dysplasia 4, 614378 Senior-Loken syndrome 8, 616307 Short-rib thoracic dysplasia 5 w/wo polydactyly, 614376 ?Spermatogenic failure 72, 619867	VISION DISORDERS CRANIOFACIAL ANOMALIES CILIOPATHIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
WDR26	100%	100%	99%	93%	Skraban-Deardorff syndrome, 617616	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
WDR34	100%	100%	100%	99%	Short-rib thoracic dysplasia 11 w/wo polydactyly, 615633	VISION DISORDERS CILIOPATHIES SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
WDR35	100%	100%	100%	98%	Short-rib thoracic dysplasia 7 w/wo polydactyly, 614091 Cranioectodermal dysplasia 2, 613610	CRANIOFACIAL ANOMALIES CILIOPATHIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS
WDR36	100%	100%	100%	97%	Glaucoma 1, open angle, G, 609887	VISION DISORDERS MENDELIOME
WDR37	100%	100%	100%	99%	Neurooculocardiogenitourinary syndrome, 618652	INTELLECTUAL DISABILITY MENDELIOME
WDR4	100%	100%	100%	98%	Galloway-Mowat syndrome 6, 618347 Microcephaly, growth deficiency, seizures, and brain malformations, 618346	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
WDR45	100%	100%	99%	77%	Neurodegeneration with brain iron accumulation 5, 300894	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PARKINSON

WDR45B	100%	100%	100%	97%	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities w/wo seizures, 617977	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
WDR5	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
WDR60	100%	100%	100%	98%	Short-rib thoracic dysplasia 8 w/wo polydactyly, 615503	CILIOPATHIES DISORDERS OF SEX DEVELOPMENT SHORT STATURE AND SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
WDR62	100%	100%	100%	99%	Microcephaly 2, primary, autosomal recessive, w/wo cortical malformations, 604317	FETAL AKINESIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
WDR66	100%	100%	100%	98%	Spermatogenic failure 33, 618152	CILIOPATHIES MALE INFERTILITY MENDELIOME
WDR72	96%	96%	100%	98%	Amelogenesis imperfecta, type IIA3, 613211	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
WDR73	100%	100%	100%	98%	Galloway-Mowat syndrome 1, 251300	MOVEMENT DISORDERS VISION DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
WDR81	100%	100%	100%	99%	Cerebellar ataxia, impaired intellectual development, and dysquilibrium syndrome 2, 610185 Hydrocephalus, congenital, 3, with brain anomalies, 617967	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
WEE2	100%	100%	100%	98%	Oocyte maturation defect 5, 617996	MENDELIOME PRE CONCEPTION SCREENING
WFS1	100%	100%	100%	99%	Deafness, autosomal dominant 6/14/38, 600965 ?Cataract 41, 116400 Wolfram-like syndrome, autosomal dominant, 614296 Wolfram syndrome 1, 222300	MOVEMENT DISORDERS VISION DISORDERS HEARING IMPAIRMENT EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

WHRN	100%	100%	100%	99%	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRE CONCEPTION SCREENING
WIPF1	100%	100%	100%	98%	Wiskott-Aldrich syndrome 2, 614493	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
WIPI2	100%	100%	100%	98%	?Intellectual developmental disorder with short stature and variable skeletal anomalies, 618453	MENDELIOME PRE CONCEPTION SCREENING
WLS	100%	100%	100%	98%	Zaki syndrome, 619648	MENDELIOME
WNK1	100%	100%	100%	98%	Neuropathy, hereditary sensory and autonomic, type II, 201300 Pseudohypoaldosteronism, type IIC, 614492	NEUROPATHIES HNPD RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
WNK3	100%	100%	97%	71%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
WNK4	100%	100%	100%	97%	Pseudohypoaldosteronism, type IIB, 614491	RENAL DISORDERS MENDELIOME
WNT1	100%	100%	100%	98%	Osteogenesis imperfecta, type XV, 615220	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
WNT10A	100%	100%	100%	99%	Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400 Ectodermal dysplasia 16 (odontoonychodermal dysplasia), 257980	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
WNT10B	100%	100%	100%	99%	Tooth agenesis, selective, 8, 617073 Split-hand/foot malformation 6, 225300	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
WNT2B	100%	100%	100%	99%	Diarrhea 9, 618168	MENDELIOME
WNT3	100%	100%	99%	96%	?Tetra-amelia syndrome 1, 273395	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING

WNT4	100%	99%	99%	95%	?SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330	DISORDERS OF SEX DEVELOPMENT RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS
WNT5A	100%	100%	100%	97%	Robinow syndrome, autosomal dominant 1, 180700	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME SCHISIS
WNT6	100%	100%	100%	97%	No OMIM disease ID	SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME
WNT7A	100%	100%	100%	99%	Fuhrmann syndrome, 228930 Ulna and fibula, absence of, with severe limb deficiency, 276820	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA MENDELIOME PRE CONCEPTION SCREENING
WRAP53	100%	100%	100%	98%	Dyskeratosis congenita, autosomal recessive 3, 613988	HEREDITARY BONE MARROW FAILURE SKIN DISORDERS DYSKERATOSIS CONGENITA PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING TUMOR
WRN	100%	100%	100%	97%	Werner syndrome, 277700	VISION DISORDERS SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING TUMOR
WT1	100%	100%	99%	96%	Mesothelioma, somatic, 156240 Meacham syndrome, 608978 Frasier syndrome, 136680 Nephrotic syndrome, type 4, 256370 Denys-Drash syndrome, 194080 Wilms tumor, type 1, 194070	DISORDERS OF SEX DEVELOPMENT RENAL DISORDERS MENDELIOME TUMOR
WWOX	100%	100%	100%	99%	Esophageal squamous cell carcinoma, somatic, 133239 Developmental and epileptic encephalopathy 28, 616211 Spinocerebellar ataxia, autosomal recessive 12, 614322	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

XDH	100%	100%	100%	99%	Xanthinuria, type I, 278300	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
XIAP	100%	100%	98%	73%	Lymphoproliferative syndrome, X-linked, 2, 300635	PRIMARY IMMUNODEFICIENCY MENDELIOME
XIRP2	100%	100%	100%	97%	No OMIM disease ID	HEART MENDELIOME
XIST	NC	NC	NC	NC	X-inactivation, familial skewed, 300087	MENDELIOME
XK	100%	99%	98%	72%	McLeod syndrome w/wo chronic granulomatous disease, 300842	MOVEMENT DISORDERS EPILEPSY HEART MENDELIOME MUSCLE DISORDERS
XKRY	NC	NC	NC	NC	No OMIM disease ID	MALE INFERTILITY MENDELIOME
XKRY2	NC	NC	NC	NC	No OMIM disease ID	MALE INFERTILITY MENDELIOME
XPA	100%	100%	100%	97%	Xeroderma pigmentosum, group A, 278700	MOVEMENT DISORDERS SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING TUMOR
XPC	100%	100%	99%	95%	Xeroderma pigmentosum, group C, 278720	SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING TUMOR
XPNPEP3	100%	100%	100%	99%	Nephronophthisis-like nephropathy 1, 613159	CILIOPATHIES RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
XPO5	100%	100%	100%	99%	No OMIM disease ID	RENAL DISORDERS MENDELIOME
XPR1	100%	100%	100%	98%	Basal ganglia calcification, idiopathic, 6, 616413	MOVEMENT DISORDERS MENDELIOME PARKINSON
XRCC1	100%	100%	100%	98%	?Spinocerebellar ataxia, autosomal recessive 26, 617633	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME PRE CONCEPTION SCREENING

XRCC2	100%	100%	100%	99%	Spermatogenic failure 50, 619145 ?Premature ovarian failure 17, 619146 ?Fanconi anemia, complementation group U, 617247	HEREDITARY BONE MARROW FAILURE MALE INFERTILITY MENDELIOME PRE CONCEPTION SCREENING
XRCC4	100%	100%	100%	98%	Short stature, microcephaly, and endocrine dysfunction, 616541	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
XYLT1	100%	99%	99%	93%	Desbuquois dysplasia 2, 615777	SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING SCHISIS
XYLT2	99%	99%	100%	98%	Spondyloocular syndrome, 605822	SKIN DISORDERS HEARING IMPAIRMENT SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRE CONCEPTION SCREENING
YAP1	100%	100%	99%	97%	Coloboma, ocular, w/wo hearing impairment, cleft lip/palate, and/or impaired intellectual development, 120433	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME
YARS1	100%	100%	100%	97%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease 2, 619418 Charcot-Marie-Tooth disease, dominant intermediate C, 608323	VISION DISORDERS NEUROPATHIES MENDELIOME
YARS2	100%	100%	100%	97%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561	HEREDITARY BONE MARROW FAILURE IRON DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING MUSCLE DISORDERS
YEATS2	100%	100%	100%	98%	?Epilepsy, myoclonic, familial adult, 4, 615127	MENDELIOME
YIF1B	90%	90%	100%	98%	Kaya-Barakat-Masson syndrome, 619125	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING

YIPF5	100%	100%	100%	96%	Microcephaly, epilepsy, and diabetes syndrome 2, 619278	INTELLECTUAL DISABILITY MENDELIOME
YME1L1	100%	100%	100%	97%	?Optic atrophy 11, 617302	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRE CONCEPTION SCREENING
YPEL2	100%	100%	100%	99%	No OMIM disease ID	VISION DISORDERS MENDELIOME
YRDC	100%	100%	99%	94%	Galloway-Mowat syndrome 10, 619609	MENDELIOME
YWHAE	100%	100%	100%	98%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
YWHAG	100%	100%	100%	98%	Developmental and epileptic encephalopathy 56, 617665	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME
YWHAZ	100%	100%	100%	98%	No OMIM disease ID	SKIN DISORDERS MENDELIOME
YY1	100%	99%	98%	77%	Gabriele-de Vries syndrome, 617557	INTELLECTUAL DISABILITY MENDELIOME
YY1AP1	100%	100%	100%	98%	Grange syndrome, 602531	MENDELIOME PRE CONCEPTION SCREENING
ZAP70	100%	100%	100%	99%	Immunodeficiency 48, 269840 Autoimmune disease, multisystem, infantile-onset, 2, 617006	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING SEVERE COMBINED IMMUNODEFICIENCY (SCID)
ZBTB11	100%	100%	100%	99%	Intellectual developmental disorder, autosomal recessive 69, 618383	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ZBTB16	100%	100%	100%	99%	Leukemia, acute promyelocytic, PL2F/RARA type,	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ZBTB17	100%	100%	100%	99%	No OMIM disease ID	HEART MENDELIOME
ZBTB18	100%	100%	100%	98%	Intellectual developmental disorder, autosomal dominant 22, 612337	INTELLECTUAL DISABILITY MENDELIOME
ZBTB20	100%	100%	100%	99%	Primrose syndrome, 259050	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME

ZBTB24	100%	100%	100%	99%	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069	PRIMARY IMMUNODEFICIENCY INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ZBTB42	100%	100%	100%	99%	?Lethal congenital contracture syndrome 6, 616248	MENDELIOME PRE CONCEPTION SCREENING
ZBTB7A	100%	100%	100%	99%	Macrocephaly, neurodevelopmental delay, lymphoid hyperplasia, and persistent fetal hemoglobin, 619769	INTELLECTUAL DISABILITY MENDELIOME
ZC3H14	100%	100%	100%	98%	Intellectual developmental disorder, autosomal recessive 56, 617125	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ZC4H2	100%	99%	96%	64%	Wieacker-Wolff syndrome, 314580 Wieacker-Wolff syndrome, female-restricted, 301041	FETAL AKINESIA MOVEMENT DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME MUSCLE DISORDERS
ZCCHC8	100%	100%	100%	96%	?Pulmonary fibrosis and/or bone marrow failure, telomere-related, 5, 618674	HEREDITARY BONE MARROW FAILURE MENDELIOME
ZDHHC9	100%	99%	98%	75%	Intellectual developmental disorder, X-linked syndromic, Raymond type, 300799	INTELLECTUAL DISABILITY MENDELIOME
ZEB1	99%	99%	100%	98%	Corneal dystrophy, posterior polymorphous, 3, 609141 Corneal dystrophy, Fuchs endothelial, 6, 613270	VISION DISORDERS MENDELIOME
ZEB2	96%	96%	100%	96%	Mowat-Wilson syndrome, 235730	CRANIOFACIAL ANOMALIES EPILEPSY INTELLECTUAL DISABILITY MENDELIOME SCHISIS
ZFHX2	100%	100%	100%	98%	?Marsili syndrome, 147430	HNPD MENDELIOME
ZFHX3	100%	100%	100%	96%	Prostate cancer, somatic, 176807	MENDELIOME
ZFHX4	99%	98%	100%	97%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
ZFP57	100%	100%	100%	98%	Diabetes mellitus, transient neonatal 1, 601410	MENDELIOME
ZFPM2	100%	100%	100%	98%	Diaphragmatic hernia 3, 610187 46XY sex reversal 9, 616067 Tetralogy of Fallot, 187500	CONGENITAL HEART DISEASE DISORDERS OF SEX DEVELOPMENT HEART MENDELIOME
ZFX	100%	100%	98%	74%	No OMIM disease ID	MALE INFERTILITY MENDELIOME

ZFYVE19	100%	100%	100%	99%	Cholestasis, progressive familial intrahepatic, 9, 619849	LIVER DISORDERS MENDELIOME
ZFYVE26	100%	100%	100%	99%	Spastic paraplegia 15, autosomal recessive, 270700	MOVEMENT DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ZFYVE27	100%	100%	100%	99%	Spastic paraplegia 33, autosomal dominant, 610244	MOVEMENT DISORDERS MENDELIOME
ZIC1	100%	100%	100%	98%	?Craniosynostosis 6, 616602 Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME
ZIC2	100%	99%	99%	86%	Holoprosencephaly 5, 609637	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME SCHISIS
ZIC3	100%	100%	97%	69%	Congenital heart defects, nonsyndromic, 1, X-linked, 306955 Heterotaxy, visceral, 1, X-linked, 306955 VACTERL association, X-linked, 314390	CONGENITAL HEART DISEASE CILIOPATHIES HEART MENDELIOME SCHISIS
ZMIZ1	100%	99%	100%	99%	Neurodevelopmental disorder with dysmorphic facies and distal skeletal anomalies, 618659	INTELLECTUAL DISABILITY MENDELIOME
ZMPSTE24	100%	100%	100%	98%	Mandibuloacral dysplasia with type B lipodystrophy, 608612 Restrictive dermopathy 1, 275210	FETAL AKINESIA SKIN DISORDERS SHORT STATURE AND SKELETAL DYSPLASIA METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING SCHISIS
ZMYM2	100%	100%	100%	98%	Neurodevelopmental-craniofacial syndrome with variable renal and cardiac abnormalities, 619522	INTELLECTUAL DISABILITY MENDELIOME
ZMYM3	100%	99%	98%	72%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
ZMYND10	100%	100%	100%	99%	Ciliary dyskinesia, primary, 22, 615444	CILIOPATHIES MENDELIOME PRE CONCEPTION SCREENING
ZMYND11	100%	100%	100%	99%	Intellectual developmental disorder, autosomal dominant 30, 616083	INTELLECTUAL DISABILITY MENDELIOME

ZMYND15	100%	100%	100%	99%	?Spermatogenic failure 14, 615842	MALE INFERTILITY MENDELIOME
ZMYND8	100%	100%	100%	99%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
ZNF141	100%	100%	100%	99%	?Polydactyly, postaxial, type A6, 615226	MENDELIOME
ZNF142	100%	100%	100%	99%	Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ZNF148	100%	100%	100%	98%	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260	INTELLECTUAL DISABILITY MENDELIOME
ZNF292	99%	99%	100%	98%	Intellectual developmental disorder, autosomal dominant 64, 619188	INTELLECTUAL DISABILITY MENDELIOME
ZNF335	100%	100%	100%	99%	Microcephaly 10, primary, autosomal recessive, 615095	INTELLECTUAL DISABILITY MENDELIOME PRE CONCEPTION SCREENING
ZNF341	100%	100%	100%	98%	Hyper-IgE recurrent infection syndrome 3, autosomal recessive, 618282	PRIMARY IMMUNODEFICIENCY MENDELIOME PRE CONCEPTION SCREENING
ZNF407	100%	100%	100%	98%	SIMHA syndrome, 619557	INTELLECTUAL DISABILITY MENDELIOME
ZNF408	100%	100%	100%	99%	Retinitis pigmentosa 72, 616469 ?Exudative vitreoretinopathy 6, 616468	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ZNF41	100%	100%	98%	72%	No OMIM disease ID	INTELLECTUAL DISABILITY MENDELIOME
ZNF423	100%	100%	100%	99%	Nephronophthisis 14, 614844 Joubert syndrome 19, 614844	VISION DISORDERS CILIOPATHIES RENAL DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ZNF462	100%	100%	100%	99%	Weiss-Kruszka syndrome, 618619	INTELLECTUAL DISABILITY MENDELIOME
ZNF469	100%	100%	100%	98%	Brittle cornea syndrome 1, 229200	VISION DISORDERS SKIN DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ZNF513	100%	100%	100%	98%	?Retinitis pigmentosa 58, 613617	VISION DISORDERS MENDELIOME PRE CONCEPTION SCREENING
ZNF526	100%	100%	100%	99%	Dentici-Novelli neurodevelopmental syndrome, 619877	INTELLECTUAL DISABILITY MENDELIOME

ZNF592	100%	100%	100%	99%	No OMIM disease ID	SKIN DISORDERS MENDELIOME
ZNF644	100%	100%	100%	98%	Myopia 21, autosomal dominant, 614167	VISION DISORDERS MENDELIOME
ZNF687	100%	100%	100%	99%	Paget disease of bone 6, 616833	MENDELIOME
ZNF699	100%	100%	100%	98%	DEGCAGS syndrome, 619488	INTELLECTUAL DISABILITY MENDELIOME
ZNF711	100%	100%	98%	73%	Intellectual developmental disorder, X-linked 97, 300803	INTELLECTUAL DISABILITY MENDELIOME
ZNF750	100%	100%	100%	99%	?Seborrhea-like dermatitis with psoriasiform elements, 610227	SKIN DISORDERS MENDELIOME
ZNFX1	100%	100%	100%	99%	Immunodeficiency 91 and hyperinflammation, 619644	PRIMARY IMMUNODEFICIENCY MENDELIOME
ZNHIT3	78%	76%	100%	96%	PEHO syndrome, 260565	MENDELIOME PRE CONCEPTION SCREENING
ZP1	100%	100%	100%	99%	Oocyte maturation defect 1, 615774	MENDELIOME PRE CONCEPTION SCREENING
ZP2	100%	100%	100%	98%	Oocyte maturation defect 6, 618353	MENDELIOME
ZP3	100%	100%	100%	98%	Oocyte maturation defect 3, 617712	MENDELIOME
ZPBP	100%	100%	100%	97%	?Spermatogenic failure 66, 619799	MENDELIOME
ZPR1	100%	100%	100%	98%	?Growth restriction, hypoplastic kidneys, alopecia, and distinctive facies, 619321	MENDELIOME
ZSWIM6	97%	95%	96%	89%	Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865 Acromelic frontonasal dysostosis, 603671	SHORT STATURE AND SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME SCHISIS

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.
Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.
TWIST X2 is the chemistry used for WES analysis.
Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with coverage denoting NC are non-protein coding genes.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : March 17th, 2023
This list is accurate for panel version DG 3.6.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors