MENDELIOME GENE PANEL DG 3.00 (4562 genes)

Releasedate: 02-12-2020

Gene	Agilent V5 covered > 10x	Agilent V5 covered > 20x	TWIST covered > 10x	TWIST covered 20x	Associated Phenotype description and OMIM disease ID
A2M	99,9	99,6	100	100	{Alzheimer disease, susceptibility to}, 104300
A2ML1	100	99,6	100	100	{Otitis media, susceptibility to}, 166760
A4GALT	100	100	100	100	[Blood group, P1Pk system, p phenotype], 111400 NOR polyagglutination syndrome, 111400 [Blood group, P1Pk system, P(2) phenotype], 111400
AAAS	100	99,9	100	100	Achalasia-addisonianism-alacrimia syndrome, 231550
AAGAB	100	99,2	100	100	Keratoderma, palmoplantar, punctate type IA, 148600
AARS1	100	99,9	100	100	Developmental and epileptic encephalopathy 29, 616339 Charcot-Marie-Tooth disease, axonal, type 2N, 613287
AARS2	100	99,4	100	100	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889
AASS	100	99,7	100	100	Hyperlysinemia, 238700
ABAT	100	99,4	100	100	GABA-transaminase deficiency, 613163
ABCA1	99,9	99,1	100	100	HDL deficiency, familial, 1, 604091 Tangier disease, 205400
ABCA12	99,5	98,7	100	100	Ichthyosis, congenital, autosomal recessive 4A, 601277 Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500
ABCA2	99,7	98,9	100	100	Intellectual developmental disorder with poor growth and with or without seizures or ataxia, 618808
ABCA3	99,9	99,3	100	100	Surfactant metabolism dysfunction, pulmonary, 3, 610921
ABCA4	99,9	99,3	96,5	96,5	Retinal dystrophy, early-onset severe, 248200 Stargardt disease 1, 248200 Fundus flavimaculatus, 248200 {Macular degeneration, age-related, 2}, 153800 Cone-rod dystrophy 3, 604116 Retinitis pigmentosa 19, 601718

ABCA5	98,4	92,3	100	100	?Hypertrichosis, congenital generalized, with gingival hyperplasia, 135400
ABCB10	77,4	71,2	99,4	96,8	No OMIM disease ID
ABCB11	100	99,7	100	100	Cholestasis, progressive familial intrahepatic 2, 601847 Cholestasis, benign recurrent intrahepatic, 2, 605479
ABCB4	99,9	99,6	100	100	Gallbladder disease 1, 600803 Cholestasis, intrahepatic, of pregnancy, 3, 614972 Cholestasis, progressive familial intrahepatic 3, 602347
ABCB6	100	99,8	100	100	Dyschromatosis universalis hereditaria 3, 615402 Microphthalmia, isolated, with coloboma 7, 614497 [Blood group, Langereis system], 111600 Pseudohyperkalemia, familial, 2, due to red cell leak, 609153
ABCB7	99,5	98,2	99,3	98,8	Anemia, sideroblastic, with ataxia, 301310
ABCC1	98,9	97,9	100	100	?Deafness, autosomal dominant 77, 618915
ABCC2	100	99,9	100	100	Dubin-Johnson syndrome, 237500
ABCC6	93,6	92,4	100	100	Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850 Arterial calcification, generalized, of infancy, 2, 614473
ABCC8	100	99,8	100	100	Diabetes mellitus, noninsulin-dependent, 125853 Diabetes mellitus, permanent neonatal 3, with or without neurologic features, 618857 Diabetes mellitus, transient neonatal 2, 610374 Hyperinsulinemic hypoglycemia, familial, 1, 256450 Hypoglycemia of infancy, leucine-sensitive, 240800
ABCC9	100	99,9	100	100	Hypertrichotic osteochondrodysplasia, 239850 ?Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569
ABCD1	75,8	71,6	100	100	Adrenomyeloneuropathy, adult, 300100 Adrenoleukodystrophy, 300100
ABCD2	100	99,8	100	100	No OMIM disease ID
ABCD3	99,8	97,7	100	100	?Bile acid synthesis defect, congenital, 5, 616278
ABCD4	99,9	98,6	100	100	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	100	100	100	100	Sitosterolemia 2, 618666
ABCG8	99,1	97,3	100	100	{Gallbladder disease 4}, 611465 Sitosterolemia 1, 210250

ABHD12	91,2	85,2	100	99,4	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ABHD5	100	100	100	100	Chanarin-Dorfman syndrome, 275630
ABL1	100	100	100	100	Congenital heart defects and skeletal malformations syndrome, 617602 Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 608232
ACACA	98,4	98,1	100	100	Acetyl-CoA carboxylase deficiency, 613933
ACAD8	100	100	100	100	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	100	99,9	100	100	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADM	99,8	99	100	100	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	99,9	98,2	100	100	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	100	99,2	100	100	2-methylbutyrylglycinuria, 610006
ACADVL	99,4	97,3	100	100	VLCAD deficiency, 201475
ACAN	96,6	92,9	98,9	98,7	Short stature and advanced bone age, with or without early-onset osteoarthritis and/or osteochondritis dissecans, 165800 Spondyloepimetaphyseal dysplasia, aggrecan type, 612813 ?Spondyloepiphyseal dysplasia, Kimberley type, 608361
ACAT1	99,9	97,6	100	100	Alpha-methylacetoacetic aciduria, 203750
ACAT2	100	100	100	100	?ACAT2 deficiency, 614055
ACBD5	100	99,2	100	100	Retinal dystrophy with leukodystrophy, 618863
ACD	100	99,9	100	100	?Dyskeratosis congenita, autosomal dominant 6, 616553 ?Dyskeratosis congenita, autosomal recessive 7, 616553
ACE	99,9	98,4	100	100	{Stroke, hemorrhagic}, 614519 Renal tubular dysgenesis, 267430 {SARS, progression of}, 0 {Microvascular complications of diabetes 3}, 612624 {Myocardial infarction, susceptibility to}, 0 [Angiotensin I-converting enzyme, benign serum increase], 0
ACER3	99,8	98,6	100	100	?Leukodystrophy, progressive, early childhood-onset, 617762
ACO2	96,3	90,3	100	100	Infantile cerebellar-retinal degeneration, 614559 ?Optic atrophy 9, 616289
ACOX1	100	99,9	100	100	Mitchell syndrome, 618960 Peroxisomal acyl-CoA oxidase deficiency, 264470

ACOX2	100	99,2	100	100	Bile acid synthesis defect, congenital, 6, 617308
ACP4	97,2	88,8	100	100	Amelogenesis imperfecta, type IJ, 617297
ACP5	99,8	98,3	100	100	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACSF3	100	99,9	100	100	Combined malonic and methylmalonic aciduria, 614265
ACSL4	98,7	94,6	100	100	Mental retardation, X-linked 63, 300387
ACSL6	96,5	95	97,1	97,1	Myelodysplastic syndrome, 0 Myelogenous leukemia, acute, 0
ACTA1	99,6	92,3	100	100	Myopathy, actin, congenital, with cores, 161800 Nemaline myopathy 3, autosomal dominant or recessive, 161800 Myopathy, congenital, with fiber-type disproportion 1, 255310 Myopathy, actin, congenital, with excess of thin myofilaments, 161800 ?Myopathy, scapulohumeroperoneal, 616852
ACTA2	100	99	100	100	Aortic aneurysm, familial thoracic 6, 611788 Multisystemic smooth muscle dysfunction syndrome, 613834 Moyamoya disease 5, 614042
ACTB	99,7	96,1	100	100	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACTC1	100	99,7	100	100	Left ventricular noncompaction 4, 613424 Atrial septal defect 5, 612794 Cardiomyopathy, dilated, 1R, 613424 Cardiomyopathy, hypertrophic, 11, 612098
ACTG1	100	100	100	100	Baraitser-Winter syndrome 2, 614583 Deafness, autosomal dominant 20/26, 604717
ACTG2	99,9	98,2	100	100	Visceral myopathy, 155310
ACTL6A	99,8	98,7	100	100	No OMIM disease ID
ACTL6B	100	99,8	100	100	Developmental and epileptic encephalopathy 76, 618468 Intellectual developmental disorder with severe speech and ambulation defects, 618470
ACTN1	100	100	100	100	Bleeding disorder, platelet-type, 15, 615193
ACTN2	100	100	100	100	Myopathy, distal, 6, adult onset, 618655 Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158 Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158 Myopathy, congenital with structured cores and Z-line abnormalities, 618654
ACTN4	100	99,3	100	100	Glomerulosclerosis, focal segmental, 1, 603278

ACVR1	100	100	100	100	Fibrodysplasia ossificans progressiva, 135100
ACVR1B	99,7	97,4	100	100	Pancreatic cancer, somatic, 0
ACVR2B	98,3	95	100	100	Heterotaxy, visceral, 4, autosomal, 613751
ACVRL1	100	98,9	100	100	Telangiectasia, hereditary hemorrhagic, type 2, 600376
ACY1	100	98,8	100	100	Aminoacylase 1 deficiency, 609924
ADA	100	99,7	100	100	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADA2	100	99	100	100	Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688 ?Sneddon syndrome, 182410
ADAM10	94,8	93,9	100	100	{Alzheimer disease 18, susceptibility to}, 615590 Reticulate acropigmentation of Kitamura, 615537
ADAM17	99,9	99	100	100	?Inflammatory skin and bowel disease, neonatal, 1, 614328
ADAM22	99,9	99,5	100	100	?Developmental and epileptic encephalopathy 61, 617933
ADAM9	99,8	99,1	100	100	Cone-rod dystrophy 9, 612775
ADAMTS10	99,9	98,5	100	100	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS13	97,1	93,8	99,9	99,5	Thrombotic thrombocytopenic purpura, hereditary, 274150
ADAMTS17	92,8	89	97,6	95,8	Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTS18	100	99,7	100	100	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
ADAMTS19	95	91,6	100	100	No OMIM disease ID
ADAMTS2	99	96,7	98,1	97,9	Ehlers-Danlos syndrome, dermatosparaxis type, 225410
ADAMTS3	100	100	100	100	Hennekam lymphangiectasia-lymphedema syndrome 3, 618154
ADAMTS9	99,5	98,7	100	100	No OMIM disease ID
ADAMTSL2	97,1	93,3	99,8	99,4	Geleophysic dysplasia 1, 231050
ADAMTSL4	100	99,2	100	100	Ectopia lentis et pupillae, 225200 Ectopia lentis, isolated, autosomal recessive, 225100
ADAR	100	99,8	100	100	Dyschromatosis symmetrica hereditaria, 127400 Aicardi-Goutieres syndrome 6, 615010
ADARB1	97,9	95,2	95,1	95,1	Neurodevelopmental disorder with hypotonia, microcephaly, and seizures, 618862

ADAT3	100	99,7	100	100	Mental retardation, autosomal recessive 36, 615286
ADCK2	100	100	100	100	No OMIM disease ID
ADCK5	100	99,9	100	100	No OMIM disease ID
ADCY1	95,2	93,8	98,5	97,9	?Deafness, autosomal recessive 44, 610154
ADCY10	100	99,9	100	100	{Hypercalciuria, absorptive, susceptibility to}, 143870
ADCY3	100	99,1	100	100	{Obesity, susceptibility to, BMIQ19}, 617885
ADCY5	95,1	91,2	99,2	98	Dyskinesia, familial, with facial myokymia, 606703
ADCY6	100	100	100	100	?Lethal congenital contracture syndrome 8, 616287
ADD3	99,9	99,5	100	100	Cerebral palsy, spastic quadriplegic, 3, 617008
ADGRE2	96,8	96,1	99,1	98,7	Vibratory urticaria, 125630
ADGRG1	100	100	100	100	Polymicrogyria, bilateral perisylvian, 615752 Polymicrogyria, bilateral frontoparietal, 606854
ADGRG2	98,3	92,7	100	100	Congenital bilateral absence of vas deferens, X-linked, 300985
ADGRG6	99,9	99	100	100	Lethal congenital contracture syndrome 9, 616503
ADGRV1	99,6	98,6	100	100	Usher syndrome, type 2C, 605472 ?Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472
ADIPOQ	100	100	100	100	Adiponectin deficiency, 612556
ADIPOR1	99,9	97,8	100	100	No OMIM disease ID
ADK	84,1	81	84,5	84,5	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADNP	90,5	90,5	95,4	95,4	Helsmoortel-van der Aa syndrome, 615873
ADPRS	100	99,8	100	100	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
ADRB2	100	99,7	100	100	{Asthma, nocturnal, susceptibility to}, 600807 {Obesity, susceptibility to}, 601665 Beta-2-adrenoreceptor agonist, reduced response to, 0
ADSL	99,2	98,7	100	100	Adenylosuccinase deficiency, 103050
ADSS1	90,2	87,5	100	100	Myopathy, distal, 5, 617030

AEBP1	100	100	100	100	Ehlers-Danlos syndrome, classic-like, 2, 618000
AFF2	99,9	99,4	100	99,8	Mental retardation, X-linked, FRAXE type, 309548
AFF4	99,9	98,9	100	100	CHOPS syndrome, 616368
AFG3L2	95	91,1	100	99,9	Spastic ataxia 5, autosomal recessive, 614487 Optic atrophy 12, 618977 Spinocerebellar ataxia 28, 610246
AFP	96,9	89,8	100	100	Alpha-fetoprotein deficiency, 615969 [Hereditary persistence of alpha-fetoprotein], 615970
AGA	100	100	100	100	Aspartylglucosaminuria, 208400
AGBL1	98,5	98,4	100	100	Corneal dystrophy, Fuchs endothelial, 8, 615523
AGBL5	99,9	99,3	100	100	Retinitis pigmentosa 75, 617023
AGK	90,6	88,6	91,2	91,2	Sengers syndrome, 212350 Cataract 38, autosomal recessive, 614691
AGL	100	99,4	100	100	Glycogen storage disease IIIb, 232400 Glycogen storage disease IIIa, 232400
AGMO	99,2	96	100	100	No OMIM disease ID
AGO2	99,1	99,1	99,9	99,5	No OMIM disease ID
AGPAT2	99,6	96,1	100	100	Lipodystrophy, congenital generalized, type 1, 608594
AGPS	99,3	95,4	100	99,9	Rhizomelic chondrodysplasia punctata, type 3, 600121
AGRN	96,9	92,6	100	99,9	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120
AGT	100	100	100	100	Renal tubular dysgenesis, 267430 {Preeclampsia, susceptibility to}, 0 {Hypertension, essential, susceptibility to}, 145500
AGTPBP1	96	94,1	100	100	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276
AGTR1	92	91,8	100	100	Renal tubular dysgenesis, 267430 {Hypertension, essential}, 145500
AGXT	100	100	100	100	Hyperoxaluria, primary, type 1, 259900
AHCY	100	99,2	100	100	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHDC1	100	99,3	100	100	Xia-Gibbs syndrome, 615829
AHI1	99,7	97,9	100	100	Joubert syndrome 3, 608629

AHR	99,2	98,8	100	100	?Retinitis pigmentosa 85, 618345
AHSG	99,9	99,5	100	100	?Alopecia-mental retardation syndrome 1, 203650
AICDA	100	100	100	100	Immunodeficiency with hyper-IgM, type 2, 605258
AIFM1	99,9	98,8	100	100	Cowchock syndrome, 310490 Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 Combined oxidative phosphorylation deficiency 6, 300816 Deafness, X-linked 5, 300614
AIMP1	99,2	94,5	100	99,9	Leukodystrophy, hypomyelinating, 3, 260600
AIMP2	88,9	86	100	100	Leukodystrophy, hypomyelinating, 17, 618006
AIP	100	99	100	100	Pituitary adenoma 1, multiple types, 102200 Pituitary adenoma predisposition, 102200
AIPL1	100	99,8	100	100	Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393 Cone-rod dystrophy, 604393
AIRE	100	99,8	100	100	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AK1	100	100	100	100	Hemolytic anemia due to adenylate kinase deficiency, 612631
AK2	98,9	94,9	100	99,9	Reticular dysgenesis, 267500
AK7	99,7	98,1	100	100	?Spermatogenic failure 27, 617965
AKAP9	98,8	95,5	100	100	?Long QT syndrome 11, 611820
AKR1C1	95,9	89,7	100	100	No OMIM disease ID
AKR1C2	94,9	89,2	100	100	46XY sex reversal 8, 614279
AKR1D1	100	99,4	100	100	Bile acid synthesis defect, congenital, 2, 235555
AKT1	100	99,5	100	100	Breast cancer, somatic, 114480 Cowden syndrome 6, 615109 Proteus syndrome, somatic, 176920 Ovarian cancer, somatic, 167000 Colorectal cancer, somatic, 114500
AKT2	100	99,5	100	100	Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900 Diabetes mellitus, type II, 125853
AKT3	98,7	94,5	100	100	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937

ALAD	99,3	94,1	100	100	Porphyria, acute hepatic, 612740 {Lead poisoning, susceptibility to}, 612740
ALAS2	98,9	94,9	100	100	Protoporphyria, erythropoietic, X-linked, 300752 Anemia, sideroblastic, 1, 300751
ALB	100	99,4	100	100	Analbuminemia, 616000 [Dysalbuminemic hyperthyroxinemia], 615999
ALDH18A1	100	99,9	100	100	Cutis laxa, autosomal recessive, type IIIA, 219150 Cutis laxa, autosomal dominant 3, 616603 Spastic paraplegia 9B, autosomal recessive, 616586 Spastic paraplegia 9A, autosomal dominant, 601162
ALDH1A2	99,9	98,5	100	100	No OMIM disease ID
ALDH1A3	97,2	94,5	100	99,9	Microphthalmia, isolated 8, 615113
ALDH1B1	100	100	100	100	No OMIM disease ID
ALDH2	100	100	100	100	Alcohol sensitivity, acute, 610251 {Hangover, susceptibility to}, 610251 {Esophageal cancer, alcohol-related, susceptibility to}, 0 {Sublingual nitroglycerin, susceptibility to poor response to}, 0
ALDH3A2	88,8	88,1	93,2	93,2	Sjogren-Larsson syndrome, 270200
ALDH4A1	100	99,4	100	100	Hyperprolinemia, type II, 239510
ALDH5A1	91	81,5	100	100	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	100	99,9	100	100	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	94,4	88,8	100	100	Epilepsy, pyridoxine-dependent, 266100
ALDOA	98,9	96,9	100	100	Glycogen storage disease XII, 611881
ALDOB	99,4	96,6	100	100	Fructose intolerance, hereditary, 229600
ALG1	53	45,8	100	100	Congenital disorder of glycosylation, type Ik, 608540
ALG10	100	100	100	100	No OMIM disease ID
ALG11	96,8	96,8	96,8	96,8	Congenital disorder of glycosylation, type Ip, 613661
ALG12	100	100	100	100	Congenital disorder of glycosylation, type Ig, 607143
ALG13	98,4	92,6	100	99,6	Developmental and epileptic encephalopathy 36, 300884 ?Congenital disorder of glycosylation, type Is, 300884

ALG14	100	99,9	100	100	?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227 Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies, 619031 Myopathy, epilepsy, and progressive cerebral atrophy, 619036
ALG2	100	100	100	100	Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 ?Congenital disorder of glycosylation, type Ii, 607906
ALG3	100	99,7	100	100	Congenital disorder of glycosylation, type Id, 601110
ALG6	98,6	94,8	100	100	Congenital disorder of glycosylation, type Ic, 603147
ALG8	97,2	95,6	96,6	96,6	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	100	99,7	100	100	Gillessen-Kaesbach-Nishimura syndrome, 263210 Congenital disorder of glycosylation, type Il, 608776
ALK	100	99,4	100	100	{Neuroblastoma, susceptibility to, 3}, 613014
ALKBH1	100	99,9	100	100	No OMIM disease ID
ALKBH8	99,8	98,9	100	100	Intellectual developmental disorder, autosomal recessive 71, 618504
ALMS1	99,8	99,5	100	100	Alstrom syndrome, 203800
ALOX12B	100	100	100	100	Ichthyosis, congenital, autosomal recessive 2, 242100
ALOXE3	100	99,5	100	100	Ichthyosis, congenital, autosomal recessive 3, 606545
ALPI	100	99,5	100	100	No OMIM disease ID
ALPK3	97,8	94,6	100	100	Cardiomyopathy, familial hypertrophic 27, 618052
ALPL	100	100	100	100	Hypophosphatasia, adult, 146300 Odontohypophosphatasia, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500
ALS2	100	99,9	100	100	Primary lateral sclerosis, juvenile, 606353 Amyotrophic lateral sclerosis 2, juvenile, 205100 Spastic paralysis, infantile onset ascending, 607225
ALX1	99,7	97,1	100	100	Frontonasal dysplasia 3, 613456
ALX3	77,9	73,3	100	100	Frontonasal dysplasia 1, 136760
ALX4	100	99,3	100	100	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597 {Craniosynostosis 5, susceptibility to}, 615529

AMACR	100	100	100	100	Bile acid synthesis defect, congenital, 4, 214950 Alpha-methylacyl-CoA racemase deficiency, 614307
AMBN	99,8	98,5	100	100	Amelogenesis imperfecta, type IF, 616270
AMELX	99,9	96,8	100	100	Amelogenesis imperfecta, type 1E, 301200
AMER1	99,9	98,5	100	100	Osteopathia striata with cranial sclerosis, 300373
AMH	96,4	83,8	100	99,8	Persistent Mullerian duct syndrome, type I, 261550
AMHR2	100	99,5	100	100	Persistent Mullerian duct syndrome, type II, 261550
AMMECR1	100	99,1	100	100	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
AMN	89,7	80	100	100	Imerslund-Grasbeck syndrome 2, 618882
AMPD1	99,9	98,6	100	100	Myopathy due to myoadenylate deaminase deficiency, 615511
AMPD2	99,8	98,9	100	100	?Spastic paraplegia 63, 615686 Pontocerebellar hypoplasia, type 9, 615809
AMPD3	99,9	98,5	100	100	[AMP deaminase deficiency, erythrocytic], 612874
AMT	100	100	100	100	Glycine encephalopathy, 605899
AMTN	99,6	98,6	100	100	?Amelogenesis imperfecta, type IIIB, 617607
ANAPC1	59,4	57,7	100	99,9	Rothmund-Thomson syndrome, type 1, 618625
ANG	100	100	100	100	Amyotrophic lateral sclerosis 9, 611895
ANGPTL3	98,8	95,4	100	100	Hypobetalipoproteinemia, familial, 2, 605019
ANGPTL4	100	99,2	100	100	Plasma triglyceride level QTL, low, 615881
ANK1	100	99,4	100	100	Spherocytosis, type 1, 182900
ANK2	100	100	100	100	Cardiac arrhythmia, ankyrin-B-related, 600919 Long QT syndrome 4, 600919
ANK3	99,3	99	100	100	Mental retardation, autosomal recessive, 37, 615493
ANKH	100	100	100	100	Craniometaphyseal dysplasia, 123000 Chondrocalcinosis 2, 118600
ANKLE2	99,9	98,6	100	99,8	Microcephaly 16, primary, autosomal recessive, 616681
ANKRD1	100	99,4	100	100	No OMIM disease ID

ANKRD11	96,1	93,5	100	100	KBG syndrome, 148050
ANKRD26	95	89,3	97,2	97,2	Thrombocytopenia 2, 188000
ANKS1B	100	99,6	100	100	No OMIM disease ID
ANKS6	93,8	89,5	97,9	95,8	Nephronophthisis 16, 615382
ANLN	98,7	97,5	100	100	Focal segmental glomerulosclerosis 8, 616032
ANO10	99,8	97,9	100	100	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANO3	92,4	90,8	100	100	Dystonia 24, 615034
ANO5	99,5	97,3	100	100	Miyoshi muscular dystrophy 3, 613319 Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307 Gnathodiaphyseal dysplasia, 166260
ANO6	99,9	98,7	100	100	Scott syndrome, 262890
ANOS1	89,8	88,9	99,9	99,4	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
ANTXR1	99,7	97,9	100	100	GAPO syndrome, 230740 {?Hemangioma, capillary infantile, susceptibility to}, 602089
ANTXR2	100	98,2	100	100	Hyaline fibromatosis syndrome, 228600
ANXA11	100	98,5	100	100	Amytrophic lateral sclerosis 23, 617839
AP1B1	100	99,5	100	100	Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150
AP1S1	99,9	99,5	100	100	MEDNIK syndrome, 609313
AP1S2	77,9	69,9	100	100	Mental retardation, X-linked syndromic 5, 304340
AP1S3	90,4	90,1	90,5	90,5	{Psoriasis 15, pustular, susceptibility to}, 616106
AP2M1	100	100	100	100	Intellectual developmental disorder 60 with seizures, 618587
AP2S1	90,4	90,3	100	100	Hypocalciuric hypercalcemia, type III, 600740
AP3B1	99,2	95,8	100	100	Hermansky-Pudlak syndrome 2, 608233
AP3B2	93,3	89,5	99,8	98,6	Developmental and epileptic encephalopathy 48, 617276
AP3D1	99,8	98,6	100	100	?Hermansky-Pudlak syndrome 10, 617050
AP4B1	99,9	98,7	100	100	Spastic paraplegia 47, autosomal recessive, 614066

					Stuttering, familial persistent, 1, 184450
AP4E1	99,8	98,7	100	100	Spastic paraplegia 51, autosomal recessive, 613744
AP4M1	99,9	98,9	100	100	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	78,9	71,3	87,9	87,9	Spastic paraplegia 52, autosomal recessive, 614067
AP5Z1	100	99,8	100	100	Spastic paraplegia 48, autosomal recessive, 613647
					Desmoid disease, hereditary, 135290 Adenomatous polyposis coli, 175100 Gardner syndrome, 175100 Hepatoblastoma, somatic, 114550 Colorectal cancer, somatic, 114500 Brain tumor-polyposis syndrome 2, 175100 Gastric cancer, somatic, 613659
APC	100	99,7	100	100	Adenoma, periampullary, somatic, 0
					?Sotos syndrome 3, 617169
APC2	97,6	92,7	99,9	99,1	Cortical dysplasia, complex, with other brain malformations 10, 618677
APCDD1	100	99,8	100	100	Hypotrichosis 1, 605389
					Hypoalphalipoproteinemia, primary, 2, with or without corneal clouding, 618463 Amyloidosis, 3 or more types, 105200
APOA1	100	100	100	100	ApoA-I and apoC-III deficiency, combined, 618463
					{Hypercholesterolemia, familial, modifier of}, 143890
APOA2	84,2	81,5	100	100	Apolipoprotein A-II deficiency, 0
					{Hypertriglyceridemia, susceptibility to}, 145750
APOA5	100	99,9	100	99,5	Hyperchylomicronemia, late-onset, 144650
					Hypobetalipoproteinemia, 615558
APOB	99,8	99,3	100	100	Hypercholesterolemia, familial, 2, 144010
APOC2	100	100	100	100	Hyperlipoproteinemia, type Ib, 207750
APOC3	100	100	100	100	Apolipoprotein C-III deficiency, 614028
					Hyperlipoproteinemia, type III, 617347 {Coronary artery disease, severe, susceptibility to}, 617347 Lipoprotein glomerulopathy, 611771 {?Alzheimer disease, protection against, due to APOE3-Christchurch}, 607822 Sea-blue histiocyte disease, 269600 {?Macular degeneration, age-related}, 603075
APOE	98,9	90,7	100	100	Alzheimer disease 2, 104310

APOL1	100	100	100	100	{End-stage renal disease, nondiabetic, susceptibility to}, 612551 {Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551
APOO	85,7	75,3	100	100	No OMIM disease ID
APP	100	99,9	100	100	Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants, 605714 Alzheimer disease 1, familial, 104300
APRT	100	99,5	100	100	Adenine phosphoribosyltransferase deficiency, 614723
APTX	94,9	92,4	100	100	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
AQP2	100	98,6	100	100	Diabetes insipidus, nephrogenic, 125800
AQP5	100	97	100	100	Palmoplantar keratoderma, Bothnian type, 600231
AR	97,6	93,3	100	99,2	Hypospadias 1, X-linked, 300633 Androgen insensitivity, 300068 {Prostate cancer, susceptibility to}, 176807 Androgen insensitivity, partial, with or without breast cancer, 312300 Spinal and bulbar muscular atrophy of Kennedy, 313200
ARCN1	97	96,6	96,9	96,6	Short stature, rhizomelic, with microcephaly, micrognathia, and developmental delay, 617164
ARF1	100	100	100	100	Periventricular nodular heterotopia 8, 618185
ARFGEF2	99,9	99,1	100	100	Periventricular heterotopia with microcephaly, 608097
ARG1	92,9	92,9	92,9	92,9	Argininemia, 207800
ARHGAP26	90,4	90,2	100	100	Leukemia, juvenile myelomonocytic, somatic, 607785
ARHGAP29	99,5	98	100	100	No OMIM disease ID
ARHGAP31	99,9	98,8	100	100	Adams-Oliver syndrome 1, 100300
ARHGDIA	100	100	100	100	Nephrotic syndrome, type 8, 615244
ARHGEF1	99,9	98,4	100	100	?Immunodeficiency 62, 618459
ARHGEF10	99,8	98	100	100	?Slowed nerve conduction velocity, AD, 608236
ARHGEF18	95,4	92,3	100	100	Retinitis pigmentosa 78, 617433
ARHGEF2	93	93	100	100	?Neurodevelopmental disorder with midbrain and hindbrain malformations, 617523
ARHGEF28	99,2	94,4	100	100	No OMIM disease ID
ARHGEF6	99,5	96,2	100	99,9	No OMIM disease ID

ARHGEF9	76,5	74,1	97,2	97,1	Developmental and epileptic encephalopathy 8, 300607
ARID1A	98,1	96,4	100	100	Coffin-Siris syndrome 2, 614607
ARID1B	96,2	95,2	97,9	96,7	Coffin-Siris syndrome 1, 135900
ARID2	99,8	98,5	100	100	Coffin-Siris syndrome 6, 617808
ARIH1	100	99,5	100	100	No OMIM disease ID
ARL13B	100	99,2	100	100	Joubert syndrome 8, 612291
ARL2	100	100	100	100	?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 1, 619082
ARL2BP	95,9	88,3	100	100	Retinitis pigmentosa with or without situs inversus, 615434
ARL3	100	98,4	100	100	Joubert syndrome 35, 618161 Retinitis pigmentosa 83, 618173
ARL6	99,9	98,6	100	100	Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151 {Bardet-Biedl syndrome 1, modifier of}, 209900
ARL6IP1	99,4	92,6	100	100	?Spastic paraplegia 61, autosomal recessive, 615685
ARMC2	100	99,2	100	100	Spermatogenic failure 38, 618433
ARMC5	100	99,4	100	100	ACTH-independent macronodular adrenal hyperplasia 2, 615954
ARMC9	100	99,8	100	100	Joubert syndrome 30, 617622
ARNT2	100	100	100	99,6	?Webb-Dattani syndrome, 615926
ARPC1B	100	100	100	100	Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia, 617718
ARR3	100	99,8	100	100	Myopia 26, X-linked, female-limited, 301010
ARSA	100	99,8	100	100	Metachromatic leukodystrophy, 250100
ARSB	97	88,7	100	100	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARSG	100	99,5	100	100	Usher syndrome, type IV, 618144
ARSL	99	93	100	99,9	Chondrodysplasia punctata, X-linked recessive, 302950
ARV1	100	99,9	100	100	Developmental and epileptic encephalopathy 38, 617020
ARX	81	64	91,5	85,7	Lissencephaly, X-linked 2, 300215 Developmental and epileptic encephalopathy 1, 308350

					Proud syndrome, 300004 Mental retardation, X-linked 29 and others, 300419 Partington syndrome, 309510 Hydranencephaly with abnormal genitalia, 300215
ASAH1	99,7	98,6	100	100	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASB10	99,4	95,7	100	100	Glaucoma 1, open angle, F, 603383
ASCC1	93,4	90,3	87,1	87,1	Spinal muscular atrophy with congenital bone fractures 2, 616867 Barrett esophagus/esophageal adenocarcinoma, 614266
ASCL1	100	97,6	100	100	Haddad syndrome, 209880 Central hypoventilation syndrome, congenital, 209880
ASH1L	98,7	98,6	98,7	98,7	Mental retardation, autosomal dominant 52, 617796
ASIP	100	100	100	100	[Skin/hair/eye pigmentation 9, brown/nonbrown eyes], 611742 [Skin/hair/eye pigmentation 9, dark/light hair], 611742
ASL	100	99,6	100	100	Argininosuccinic aciduria, 207900
ASNS	99,4	95,2	100	100	Asparagine synthetase deficiency, 615574
ASPA	99,9	98,3	100	100	Canavan disease, 271900
ASPH	99,9	98,8	100	100	Traboulsi syndrome, 601552
ASPM	99,7	98,2	100	100	Microcephaly 5, primary, autosomal recessive, 608716
ASPSCR1	99,7	97,8	100	100	Alveolar soft-part sarcoma, 606243
ASRGL1	100	100	100	100	No OMIM disease ID
ASS1	95,4	87,9	100	100	Citrullinemia, 215700
ASXL1	99,8	99,3	99,8	99,8	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ASXL2	99,7	98,9	100	100	Shashi-Pena syndrome, 617190
ASXL3	99,9	99,7	100	100	Bainbridge-Ropers syndrome, 615485
ATAD1	99,6	95,1	100	100	Hyperekplexia 4, 618011
ATAD3A	91,9	83,2	100	100	Harel-Yoon syndrome, 617183 Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810
ATAD3B	92,1	79,2	100	100	No OMIM disease ID

ATCAY	100	99,8	100	100	Ataxia, cerebellar, Cayman type, 601238
ATF3	99,9	97,5	100	100	No OMIM disease ID
ATF6	100	99,9	100	100	Achromatopsia 7, 616517
ATG4B	100	100	100	100	No OMIM disease ID
ATG5	99,4	97,8	100	100	?Spinocerebellar ataxia, autosomal recessive 25, 617584
ATIC	99,9	99,3	100	100	AICA-ribosiduria due to ATIC deficiency, 608688
ATL1	100	99,7	100	100	Spastic paraplegia 3A, autosomal dominant, 182600 Neuropathy, hereditary sensory, type ID, 613708
ATL3	99,8	98,3	100	100	Neuropathy, hereditary sensory, type IF, 615632
ATM	99,8	98,1	100	100	Ataxia-telangiectasia, 208900 {Breast cancer, susceptibility to}, 114480 Lymphoma, mantle cell, somatic, 0 Lymphoma, B-cell non-Hodgkin, somatic, 0 T-cell prolymphocytic leukemia, somatic, 0
ATN1	99,9	98,2	100	100	Dentatorubral-pallidoluysian atrophy, 125370 Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494
ATOH1	100	100	100	100	No OMIM disease ID
ATOH7	96	91,2	99,1	94,4	Persistent hyperplastic primary vitreous, autosomal recessive, 221900
ATP11C	98,7	93,8	100	99,6	?Hemolytic anemia, congenital, X-linked, 301015
ATP13A2	100	99,5	100	100	Kufor-Rakeb syndrome, 606693 Spastic paraplegia 78, autosomal recessive, 617225
ATP1A1	100	100	100	100	Charcot-Marie-Tooth disease, axonal, type 2DD, 618036 Hypomagnesemia, seizures, and mental retardation 2, 618314
ATP1A2	100	100	100	100	Migraine, familial hemiplegic, 2, 602481 Migraine, familial basilar, 602481 Alternating hemiplegia of childhood 1, 104290
ATP1A3	100	99,9	100	100	CAPOS syndrome, 601338 Alternating hemiplegia of childhood 2, 614820 Dystonia-12, 128235
ATP2A1	100	100	100	100	Brody myopathy, 601003
ATP2A2	100	100	100	100	Acrokeratosis verruciformis, 101900 Darier disease, 124200

ATP2B2	100	99,9	100	100	{Deafness, autosomal recessive 12, modifier of}, 601386
ATP2B3	99,5	97,5	100	100	?Spinocerebellar ataxia, X-linked 1, 302500
ATP2C1	100	99,6	100	100	Hailey-Hailey disease, 169600
ATP4A	99,9	98,9	100	100	No OMIM disease ID
ATP5F1A	95,2	87,6	100	100	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4, 615228 ?Combined oxidative phosphorylation deficiency 22, 616045
ATP5F1B	100	97,8	100	100	No OMIM disease ID
ATP5F1C	98	92,2	100	100	No OMIM disease ID
ATP5F1D	96,2	89,3	100	100	Mitochondrial complex V (ATP synthase) deficiency, 618120
ATP5F1E	100	100	100	100	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053
ATP5IF1	100	100	100	100	No OMIM disease ID
ATP5MC1	100	99,9	100	100	No OMIM disease ID
ATP5MC2	100	98,1	100	100	No OMIM disease ID
ATP5MC3	100	100	100	100	No OMIM disease ID
ATP5ME	100	100	100	100	No OMIM disease ID
ATP5MF	100	99,4	100	100	No OMIM disease ID
ATP5MG	100	100	100	100	No OMIM disease ID
ATP5MGL	100	100	100	100	No OMIM disease ID
ATP5MD	82,9	42,8	100	100	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 6, 618683
ATP5PB	98,9	90,4	100	100	No OMIM disease ID
ATP5PD	97,3	80,9	100	100	No OMIM disease ID
ATP5PF	100	97,5	100	100	No OMIM disease ID
ATP5PO	99,9	98	100	100	No OMIM disease ID
ATP6AP1	98,2	92,1	100	100	Immunodeficiency 47, 300972

ATP6AP2	94,1	76,6	100	100	Congenital disorder of glycosylation, type IIr, 301045 Mental retardation, X-linked, syndromic, Hedera type, 300423 ?Parkinsonism with spasticity, X-linked, 300911
ATP6V0A2	100	99,5	100	100	Wrinkly skin syndrome, 278250 Cutis laxa, autosomal recessive, type IIA, 219200
ATP6V0A4	100	99,9	100	100	Distal renal tubular acidosis 3, with or without sensorineural hearing loss, 602722
ATP6V1A	99,9	98,7	100	100	Epileptic encephalopathy, infantile or early childhood, 3, 618012 Cutis laxa, autosomal recessive, type IID, 617403
ATP6V1B1	100	100	100	100	Distal renal tubular acidosis 2 with progressive sensorineural hearing loss, 267300
ATP6V1B2	100	99,3	100	100	Zimmermann-Laband syndrome 2, 616455 Deafness, congenital, with onychodystrophy, autosomal dominant, 124480
ATP6V1E1	93,1	88,3	100	100	Cutis laxa, autosomal recessive, type IIC, 617402
ATP7A	99	96,9	100	100	Occipital horn syndrome, 304150 Menkes disease, 309400 Spinal muscular atrophy, distal, X-linked 3, 300489
ATP7B	99,9	99,2	100	100	Wilson disease, 277900
ATP8A2	100	99,7	100	100	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268
ATP8B1	96,5	94	100	100	Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, progressive familial intrahepatic 1, 211600 Cholestasis, benign recurrent intrahepatic, 243300
ATPAF1	78,3	70	100	100	No OMIM disease ID
ATPAF2	100	100	100	100	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
ATR	99,9	99,4	100	100	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
ATRX	99,4	96,3	100	100	Alpha-thalassemia/mental retardation syndrome, 301040 Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Mental retardation-hypotonic facies syndrome, X-linked, 309580
ATXN1	100	99,7	100	100	Spinocerebellar ataxia 1, 164400
ATXN10	99,9	99,2	100	100	Spinocerebellar ataxia 10, 603516
ATXN2	91,7	84,4	99,1	96,8	{Amyotrophic lateral sclerosis, susceptibility to, 13}, 183090 {Parkinson disease, late-onset, susceptibility to}, 168600 Spinocerebellar ataxia 2, 183090
ATXN3	94,5	89,8	95,8	95,8	Machado-Joseph disease, 109150

ATXN7	99,8	97,6	98,6	97,4	Spinocerebellar ataxia 7, 164500
ATXN8OS					Spinocerebellar ataxia 8, 608768 {Parkinson disease, susceptibility to}, 168600
AUH	100	99,8	100	100	3-methylglutaconic aciduria, type I, 250950
AURKC	100	99,2	100	100	Spermatogenic failure 5, 243060
AUTS2	98,2	95,8	100	100	Mental retardation, autosomal dominant 26, 615834
AVIL	100	99,9	100	100	Nephrotic syndrome, type 21, 618594
AVP	84,9	64,3	100	99,9	Diabetes insipidus, neurohypophyseal, 125700
AVPR2	100	99,4	100	100	Diabetes insipidus, nephrogenic, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539
AXIN1	100	99,6	100	100	Hepatocellular carcinoma, somatic, 114550 ?Caudal duplication anomaly, 607864
AXIN2	100	99,9	100	99,9	Colorectal cancer, somatic, 114500 Oligodontia-colorectal cancer syndrome, 608615
B2M	100	100	100	100	Immunodeficiency 43, 241600 ?Amyloidosis, familial visceral, 105200
B3GALNT1	100	99,8	100	100	[Blood group, globoside system], 615021 [Blood group, P1PK system, P(k) phenotype], 111400
B3GALNT2	93,8	89,4	92,5	92,5	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B3GALT6	75,7	69,7	89,8	81,6	Al-Gazali syndrome, 609465 Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B3GAT3	99,9	98,2	94,8	94,8	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B3GLCT	99,6	96,3	99,9	99,2	Peters-plus syndrome, 261540
B4GALNT1	99,3	95	100	100	Spastic paraplegia 26, autosomal recessive, 609195
B4GALT1	100	99,8	100	100	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	99,8	97,4	99,9	98,6	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
B4GAT1	100	100	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
B9D1	85,2	85,1	94,2	93,9	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120

B9D2	100	100	100	100	Joubert syndrome 34, 614175 ?Meckel syndrome 10, 614175
BAAT	99,8	98,4	100	100	Hypercholanemia, familial, 607748
BACH2	100	100	100	100	Immunodeficiency 60, 618394
BAG3	100	100	100	100	Cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954
BANF1	98,3	86,6	100	100	Nestor-Guillermo progeria syndrome, 614008
BAP1	84,4	83	100	100	Tumor predisposition syndrome, 614327
BARD1	100	99,8	100	100	{Breast cancer, susceptibility to}, 114480
BAX	98	95,4	100	100	T-cell acute lymphoblastic leukemia, somatic, 613065 Colorectal cancer, somatic, 114500
BAZ2B	99,9	99	100	100	No OMIM disease ID
BBIP1	98,6	92,4	100	100	?Bardet-Biedl syndrome 18, 615995
BBS1	100	100	100	100	Bardet-Biedl syndrome 1, 209900
BBS10	100	99,8	100	100	Bardet-Biedl syndrome 10, 615987
BBS12	100	100	100	100	Bardet-Biedl syndrome 12, 615989
BBS2	100	99,5	100	100	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	99,9	99,3	100	100	Bardet-Biedl syndrome 4, 615982
BBS5	99	93,9	100	100	Bardet-Biedl syndrome 5, 615983
BBS7	98,7	95,5	100	100	Bardet-Biedl syndrome 7, 615984
BBS9	92,3	90,4	95,8	95,8	Bardet-Biedl syndrome 9, 615986
BCAP31	92,6	83,2	100	99,9	Deafness, dystonia, and cerebral hypomyelination, 300475
BCAT1	100	100	100	100	No OMIM disease ID
BCAT2	100	100	100	100	?Hypervalinemia or hyperleucine-isoleucinemia, 618850
BCHE	100	99,9	100	100	{Apnea, postanesthetic, susceptibility to, due to BCHE deficiency}, 617936 Butyrylcholinesterase deficiency, 617936
BCKDHA	99,9	99,2	100	100	Maple syrup urine disease, type Ia, 248600

BCKDHB	99,5	94,4	100	100	Maple syrup urine disease, type Ib, 248600
BCKDK	100	100	100	100	Branched-chain ketoacid dehydrogenase kinase deficiency, 614923
BCL10	100	100	100	100	?Immunodeficiency 37, 616098 {Male germ cell tumor, somatic}, 273300 Lymphoma, MALT, somatic, 137245 {Lymphoma, follicular, somatic}, 605027 {Mesothelioma, somatic}, 156240 {Sezary syndrome, somatic}, 0
BCL11A	97,7	96	100	100	Dias-Logan syndrome, 617101
BCL11B	99,1	95,6	98,8	97,3	Immunodeficiency 49, 617237 Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092
BCL2	100	100	100	100	Leukemia/lymphoma, B-cell, 2, 0
BCL7A	100	100	100	100	B-cell non-Hodgkin lymphoma, high-grade, 0
BCO1	100	100	100	100	?Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300
BCOR	99,6	97,4	100	99,9	Microphthalmia, syndromic 2, 300166
BCORL1	99,6	97,9	100	100	Shukla-Vernon syndrome, 301029
BCS1L	100	100	100	100	GRACILE syndrome, 603358 Bjornstad syndrome, 262000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BDP1	98,8	95,3	100	100	?Deafness, autosomal recessive 112, 618257
BEAN1	98,7	96,4	92,2	92,2	Spinocerebellar ataxia 31, 117210
BECN1	100	100	100	100	No OMIM disease ID
BEST1	99,4	96,4	99,9	99,4	Retinitis pigmentosa-50, 613194 Bestrophinopathy, autosomal recessive, 611809 Retinitis pigmentosa, concentric, 613194 Vitreoretinochoroidopathy, 193220 ?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 2, 193220 Macular dystrophy, vitelliform, 2, 153700
BFSP1	99	89,9	100	99,9	Cataract 33, multiple types, 611391
BFSP2	99,8	97,6	100	100	Cataract 12, multiple types, 611597

BGN	100	100	100	100	Meester-Loeys syndrome, 300989 Spondyloepimetaphyseal dysplasia, X-linked, 300106
BHLHA9	70,9	50,4	99,8	97,3	Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432 ?Camptosynpolydactyly, complex, 607539
BICC1	100	100	100	100	{Renal dysplasia, cystic, susceptibility to}, 601331
BICD2	100	99,7	100	100	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291 Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290
BIN1	99,6	95,7	100	100	Centronuclear myopathy 2, 255200
BLK	100	100	100	100	Maturity-onset diabetes of the young, type 11, 613375
BLM	99,8	98,3	100	100	Bloom syndrome, 210900
BLNK	97,1	95,5	100	100	?Agammaglobulinemia 4, 613502
BLOC1S3	98,5	81,3	100	100	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	99,9	97,8	100	100	?Hermansky-pudlak syndrome 9, 614171
BLVRA	100	99,4	100	100	Hyperbiliverdinemia, 614156
BMP1	100	100	100	100	Osteogenesis imperfecta, type XIII, 614856
BMP15	100	99,3	100	100	Ovarian dysgenesis 2, 300510 Premature ovarian failure 4, 300510
BMP2	100	100	100	100	Brachydactyly, type A2, 112600 Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 617877 {HFE hemochromatosis, modifier of}, 235200
BMP4	100	100	100	100	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625
BMP6	95,7	93,6	99	95,8	No OMIM disease ID
BMP7	99,9	98,5	100	100	No OMIM disease ID
BMPER	100	99,8	100	100	Diaphanospondylodysostosis, 608022
BMPR1A	99,8	96,6	100	100	Polyposis syndrome, hereditary mixed, 2, 610069 Polyposis, juvenile intestinal, 174900 Juvenile polyposis syndrome, infantile form, 174900
BMPR1B	100	99,9	100	100	Brachydactyly, type A2, 112600 Brachydactyly, type A1, D, 616849 Acromesomelic dysplasia, Demirhan type, 609441

BMPR2	99,9	99,9	99,9	99,9	Pulmonary venoocclusive disease 1, 265450 Pulmonary hypertension, familial primary, 1, with or without HHT, 178600 Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600
BMS1	66,7	66,4	100	100	?Aplasia cutis congenita, nonsyndromic, 107600
BNC2	99,1	99,1	100	100	Lower urinary tract obstruction, congenital, 618612
BNIP3	78,3	67,2	100	99,2	No OMIM disease ID
BOLA1	100	100	100	100	No OMIM disease ID
BOLA2	100	100	100	100	No OMIM disease ID
BOLA3	99,4	90,2	100	100	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BPGM	100	100	100	100	Erythrocytosis, familial, 8, 222800
IMPAD1	100	100	100	100	Chondrodysplasia with joint dislocations, GPAPP type, 614078
BPTF	96,2	94,3	99,6	98,6	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, 617755
BRAF	91	81,1	100	100	Noonan syndrome 7, 613706 Cardiofaciocutaneous syndrome, 115150 Adenocarcinoma of lung, somatic, 211980 LEOPARD syndrome 3, 613707 Nonsmall cell lung cancer, somatic, 0 Melanoma, malignant, somatic, 0 Colorectal cancer, somatic, 0
BRAT1	99,7	98,2	100	100	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498 Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056
BRCA1	99,4	98,8	100	100	Fanconi anemia, complementation group S, 617883 {Pancreatic cancer, susceptibility to, 4}, 614320 {Breast-ovarian cancer, familial, 1}, 604370
BRCA2	99,8	98,5	100	100	{Pancreatic cancer 2}, 613347 {Breast cancer, male, susceptibility to}, 114480 {Glioblastoma 3}, 613029 Wilms tumor, 194070 Fanconi anemia, complementation group D1, 605724 {Medulloblastoma}, 155255 {Prostate cancer}, 176807 {Breast-ovarian cancer, familial, 2}, 612555
BRDT	95,9	91,6	100	100	?Spermatogenic failure 21, 617644

BRF1	99,9	98,4	100	100	Cerebellofaciodental syndrome, 616202
BRIP1	99,9	99	100	100	Fanconi anemia, complementation group J, 609054 {Breast cancer, early-onset, susceptibility to}, 114480
BRPF1	100	100	100	100	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333
BRSK2	99,5	97,4	100	100	No OMIM disease ID
BRWD3	99,3	97,2	100	100	Mental retardation, X-linked 93, 300659
BSCL2	100	100	100	100	Neuropathy, distal hereditary motor, type VC, 619112 Lipodystrophy, congenital generalized, type 2, 269700 Silver spastic paraplegia syndrome, 270685 Encephalopathy, progressive, with or without lipodystrophy, 615924
BSND	100	100	100	100	Sensorineural deafness with mild renal dysfunction, 602522 Bartter syndrome, type 4a, 602522
BTD	83,1	83	83,1	83,1	Biotinidase deficiency, 253260
BTK	100	99,9	100	99,9	Agammaglobulinemia, X-linked 1, 300755 Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200
BTRC	97,6	97,3	100	100	No OMIM disease ID
BUB1	99,8	98,8	100	100	Colorectal cancer with chromosomal instability, somatic, 114500
BUB1B	99,6	98,9	100	100	Colorectal cancer, somatic, 114500 [Premature chromatid separation trait], 176430 Mosaic variegated aneuploidy syndrome 1, 257300
BUB3	99,8	99,1	100	100	No OMIM disease ID
BVES	99,9	98,8	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 25, 616812
C11orf80	98,3	95	91,9	91,9	Hydatidiform mole, recurrent, 4, 618432
C12orf4	100	99,3	100	100	Mental retardation, autosomal recessive 66, 618221
C12orf57	100	98,9	100	100	Temtamy syndrome, 218340
C12orf65	99,8	98,5	100	100	Spastic paraplegia 55, autosomal recessive, 615035 Combined oxidative phosphorylation deficiency 7, 613559
C19orf12	100	99,8	100	100	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
C1GALT1C1	100	99,5	100	100	Tn polyagglutination syndrome, somatic, 300622
C1orf194	100	99,6	100	100	No OMIM disease ID

C1QA	100	100	100	100	C1q deficiency, 613652
C1QB	100	100	100	100	C1q deficiency, 613652
C1QBP	86,9	77,3	100	100	Combined oxidative phosphorylation deficiency 33, 617713
C1QC	100	99,2	100	100	C1q deficiency, 613652
C1QTNF5	90,9	78,5	100	100	Retinal degeneration, late-onset, autosomal dominant, 605670
C1R	100	100	99	96,9	Ehlers-Danlos syndrome, periodontal type, 1, 130080
C1S	99,9	99	99,5	97,7	Ehlers-Danlos syndrome, periodontal type, 2, 617174 C1s deficiency, 613783
C2	100	100	100	100	C2 deficiency, 217000 {Macular degeneration, age-related, 14, reduced risk of}, 615489
C2CD3	95,8	95,6	95,9	95,9	Orofaciodigital syndrome XIV, 615948
C3	99,9	99,2	100	100	{Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 C3 deficiency, 613779 {Macular degeneration, age-related, 9}, 611378
C4A	98,4	95,1	99,6	99,2	C4a deficiency, 614380 [Blood group, Rodgers], 614374
C4B	99,2	96,9	99,9	99,8	C4B deficiency, 614379
C5	99,9	98,5	100	100	[Eculizumab, poor response to], 615749 C5 deficiency, 609536
C6	100	99,7	100	100	C6 deficiency, 612446 Combined C6/C7 deficiency, 0
C7	100	98,9	100	100	C7 deficiency, 610102
C8A	100	99,6	100	100	C8 deficiency, type I, 613790
C8B	100	99,2	100	100	C8 deficiency, type II, 613789
C8G	100	100	100	100	No OMIM disease ID
C8orf37	100	99,4	100	100	Retinitis pigmentosa 64, 614500 Bardet-Biedl syndrome 21, 617406 Cone-rod dystrophy 16, 614500
C9	99,9	99,5	100	100	{Macular degeneration, age-related, 15, susceptibility to}, 615591 C9 deficiency, 613825
C9orf72	97,7	96,2	100	100	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1, 105550

CA12	100	100	100	100	Hyperchlorhidrosis, isolated, 143860
CA2	100	100	100	100	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA4	100	100	100	100	Retinitis pigmentosa 17, 600852
CA5A	87,4	85,2	87,7	87,7	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CA8	99,6	97,3	100	100	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CABIN1	100	99,6	100	99,9	No OMIM disease ID
CABP2	75,9	68	100	100	Deafness, autosomal recessive 93, 614899
CABP4	100	99,9	100	100	Cone-rod synaptic disorder, congenital nonprogressive, 610427
CACNA1A	93,2	90	100	99,9	Spinocerebellar ataxia 6, 183086 Episodic ataxia, type 2, 108500 Migraine, familial hemiplegic, 1, 141500 Developemental and epileptic encephalopathy 42, 617106 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500
CACNA1B	97,5	95,7	99,1	97,7	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497
CACNA1C	99,9	99,2	100	100	Timothy syndrome, 601005 Long QT syndrome 8, 618447 Brugada syndrome 3, 611875
CACNA1D	98	97,9	100	100	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896
CACNA1E	100	99,9	100	100	Developmental and epileptic encephalopathy 69, 618285
CACNA1F	99,7	97,5	100	100	Cone-rod dystrophy, X-linked, 3, 300476 Aland Island eye disease, 300600 Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071
CACNA1G	100	99,6	100	100	Spinocerebellar ataxia 42, 616795 Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087
CACNA1H	98,7	96,4	100	99,9	{Epilepsy, childhood absence, susceptibility to, 6}, 611942 {Epilepsy, idiopathic generalized, susceptibility to, 6}, 611942 Hyperaldosteronism, familial, type IV, 617027
CACNA1S	100	99,9	100	100	{Malignant hyperthermia susceptibility 5}, 601887 {Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580 {Malignant hyperthermia, susceptibility to, 5}, 601887 Hypokalemic periodic paralysis, type 1, 170400

CACNA2D1	98,6	95,3	100	100	No OMIM disease ID
CACNA2D2	94	93,2	99,2	97,6	Cerebellar atrophy with seizures and variable developmental delay, 618501
CACNA2D4	98,9	97,7	100	100	Retinal cone dystrophy 4, 610478
CACNB2	98,6	98,5	100	100	Brugada syndrome 4, 611876
CACNB4	95,5	94,3	100	100	{Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 {Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682 Episodic ataxia, type 5, 613855
CACNG2	100	100	100	100	?Mental retardation, autosomal dominant 10, 614256
CAD	100	99,2	100	100	Developmental and epileptic encephalopathy 50, 616457
CADM3	100	99,9	100	100	No OMIM disease ID
CALM1	100	99,4	100	100	Long QT syndrome 14, 616247 Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916
CALM2	67,8	65,1	72	72	Long QT syndrome 15, 616249
CALM3	100	99,1	100	100	Long QT syndrome 16, 618782 ?Ventricular tachycardia, catecholaminergic polymorphic 6, 618782
CALR	94,8	89,1	100	100	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
CAMK2A	99,9	99	99,8	98,7	?Mental retardation, autosomal recessive 63, 618095 Mental retardation, autosomal dominant 53, 617798
CAMK2B	100	99,8	100	100	Mental retardation, autosomal dominant 54, 617799
CAMK2G	99,9	98,1	100	100	Mental retardation, autosomal dominant 59, 618522
CAMTA1	100	99,5	100	100	Cerebellar ataxia, nonprogressive, with mental retardation, 614756
CANT1	100	99,9	100	100	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719
CAPN1	100	100	100	100	Spastic paraplegia 76, autosomal recessive, 616907
CAPN10	100	99,6	100	100	{Diabetes mellitus, noninsulin-dependent 1}, 601283
CAPN12	94	88,6	100	100	No OMIM disease ID
CAPN3	97,8	97,2	97,9	97,9	Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600 Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129
CAPN5	100	100	100	100	Vitreoretinopathy, neovascular inflammatory, 193235

CARD11	100	99,9	100	100	Immunodeficiency 11B with atopic dermatitis, 617638 B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11A, 615206
CARD14	100	98,9	100	100	Pityriasis rubra pilaris, 173200 Psoriasis 2, 602723
CARD9	99,9	98,4	100	100	Candidiasis, familial, 2, autosomal recessive, 212050
CARMIL2	96,3	94,5	99,7	98,2	Immunodeficiency 58, 618131
CARS2	100	100	100	99,2	Combined oxidative phosphorylation deficiency 27, 616672
CASK	97,3	94,2	100	100	Mental retardation, with or without nystagmus, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 FG syndrome 4, 300422
CASP10	99,5	97,3	100	100	Lymphoma, non-Hodgkin, somatic, 605027 Gastric cancer, somatic, 613659 Autoimmune lymphoproliferative syndrome, type II, 603909
CASP14	100	100	100	100	Ichthyosis, congenital, autosomal recessive 12, 617320
CASP8	95,6	95,4	95,6	95,6	{Lung cancer, protection against}, 211980 ?Autoimmune lymphoproliferative syndrome, type IIB, 607271 Hepatocellular carcinoma, somatic, 114550 {Breast cancer, protection against}, 114480
CASQ1	100	99,5	100	100	Myopathy, vacuolar, with CASQ1 aggregates, 616231
CASQ2	100	100	100	100	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CASR	100	99,9	100	100	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 Hypocalciuric hypercalcemia, type I, 145980 {Epilepsy idiopathic generalized, susceptibility to, 8}, 612899 Hypocalcemia, autosomal dominant, 601198 Hyperparathyroidism, neonatal, 239200
CAST	98,3	95,4	100	100	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295
CAT	100	100	100	100	Acatalasemia, 614097
CATSPER1	100	100	100	100	Spermatogenic failure 7, 612997
CAV1	100	100	100	100	Lipodystrophy, familial partial, type 7, 606721 ?Lipodystrophy, congenital generalized, type 3, 612526 Pulmonary hypertension, primary, 3, 615343

CAV3	100	100	100	100	Creatine phosphokinase, elevated serum, 123320 Long QT syndrome 9, 611818 Myopathy, distal, Tateyama type, 614321 Rippling muscle disease 2, 606072 Cardiomyopathy, familial hypertrophic, 192600
CAVIN1	100	100	100	100	Lipodystrophy, congenital generalized, type 4, 613327
CBL	97,3	97,1	100	100	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
CBLIF	100	99,7	100	100	Intrinsic factor deficiency, 261000
CBS	99,8	98,3	100	100	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CBWD1	20,8	19,4	99,6	99,3	No OMIM disease ID
CBX2	100	99,8	100	100	?46XY sex reversal 5, 613080
CC2D1A	100	99,3	100	100	Mental retardation, autosomal recessive 3, 608443
CC2D2A	98,5	96,5	97,1	97,1	Meckel syndrome 6, 612284 Joubert syndrome 9, 612285 COACH syndrome 2, 619111
CCBE1	99,8	98,8	100	100	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CCDC103	100	100	100	100	Ciliary dyskinesia, primary, 17, 614679
CCDC115	95,3	90	100	100	Congenital disorder of glycosylation, type IIo, 616828
CCDC134	100	100	100	100	No OMIM disease ID
CCDC141	100	99,5	100	100	No OMIM disease ID
CCDC174	99,5	97,1	100	100	Hypotonia, infantile, with psychomotor retardation, 616816
CCDC22	99,6	96,5	100	100	Ritscher-Schinzel syndrome 2, 300963
CCDC28B	100	99,7	100	100	{Bardet-Biedl syndrome 1, modifier of}, 209900
CCDC32	100	99,5	100	100	Cardiofacioneurodevelopmental syndrome, 619123
CCDC39	99,5	96,5	100	100	Ciliary dyskinesia, primary, 14, 613807
CCDC40	99,1	98,1	100	100	Ciliary dyskinesia, primary, 15, 613808
CCDC47	99,4	97,5	100	100	Trichohepatoneurodevelopmental syndrome, 618268

CCDC50	100	99,7	100	100	?Deafness, autosomal dominant 44, 607453
CCDC65	99,6	97,1	100	100	Ciliary dyskinesia, primary, 27, 615504
CCDC78	100	100	100	100	?Centronuclear myopathy 4, 614807
CCDC8	100	100	100	100	3-M syndrome 3, 614205
CCDC88A	96,4	93,1	97,5	97,5	?PEHO syndrome-like, 617507
CCDC88C	100	99,3	100	100	?Spinocerebellar ataxia 40, 616053 Hydrocephalus, congenital, 1, 236600
CCL2	100	100	100	100	{HIV-1, resistance to}, 609423 {Mycobacterium tuberculosis, susceptibility to}, 607948 {Spina bifida, susceptibility to}, 182940 {Coronary artery disease, modifier of}, 0
CCM2	98,7	97,8	100	100	Cerebral cavernous malformations-2, 603284
CCN6	84,7	84,6	84,6	84,6	Progressive pseudorheumatoid dysplasia, 208230
CCND2	100	100	100	100	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938
CCNK	92,6	89,8	100	99,8	?Intellectual developmental disorder with hypertelorism and distinctive facies, 618147
CCNO	100	99,2	100	100	Ciliary dyskinesia, primary, 29, 615872
CCNQ	83,1	78,5	98,9	94,6	STAR syndrome, 300707
CCR5	100	100	100	100	{Diabetes mellitus, insulin-dependent, 22}, 612522 {Hepatitis C virus, resistance to}, 609532 {West nile virus, susceptibility to}, 610379 {HIV infection, susceptibility/resistance to}, 0
CCT2	100	100	100	100	No OMIM disease ID
CCT5	100	99,7	100	100	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CD151	100	100	100	100	Nephropathy with pretibial epidermolysis bullosa and deafness, 609057 [Blood group, Raph], 179620
CD164	99,1	94,8	100	100	?Deafness, autosomal dominant 66, 616969
CD19	100	100	100	100	Immunodeficiency, common variable, 3, 613493
CD247	100	100	100	100	?Immunodeficiency 25, 610163
CD27	99,9	96,9	100	100	Lymphoproliferative syndrome 2, 615122

CD2AP	99,9	98,8	100	100	Glomerulosclerosis, focal segmental, 3, 607832
CD320	100	99,8	100	100	Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646
CD36	99,7	98,7	100	100	{Coronary heart disease, susceptibility to, 7}, 610938 {Malaria, cerebral, susceptibility to}, 611162 Platelet glycoprotein IV deficiency, 608404 {Malaria, cerebral, reduced risk of}, 611162
CD3D	100	100	100	100	Immunodeficiency 19, 615617
CD3E	100	99,5	100	100	Immunodeficiency 18, 615615 Immunodeficiency 18, SCID variant, 615615
CD3G	100	100	100	100	Immunodeficiency 17, CD3 gamma deficient, 615607
CD4	100	99,9	100	100	OKT4 epitope deficiency, 613949
CD40	100	100	100	100	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	97,3	88,1	100	100	Immunodeficiency, X-linked, with hyper-IgM, 308230
CD46	99,9	99,4	100	100	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922
CD55	92,9	85,4	94,1	92	[Blood group Cromer], 613793 Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300
CD59	80	71,6	64,5	64,5	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD70	99,8	97,7	100	100	Lymphoproliferative syndrome 3, 618261
CD79A	100	100	100	100	Agammaglobulinemia 3, 613501
CD79B	100	100	100	100	Agammaglobulinemia 6, 612692
CD81	100	99,9	100	100	Immunodeficiency, common variable, 6, 613496
CD8A	100	99,8	100	100	CD8 deficiency, familial, 608957
CD96	99,9	99,7	100	100	C syndrome, 211750
CDAN1	100	99,6	100	100	Dyserythropoietic anemia, congenital, type Ia, 224120
CDC14A	100	99	100	100	Deafness, autosomal recessive 32, with or without immotile sperm, 608653
CDC42	97,9	90,9	100	100	Takenouchi-Kosaki syndrome, 616737
CDC42BPB	100	99,3	100	100	No OMIM disease ID

CDC45	99,8	98,5	100	100	Meier-Gorlin syndrome 7, 617063
CDC6	100	100	100	100	?Meier-Gorlin syndrome 5, 613805
CDC73	100	99,4	100	100	Parathyroid carcinoma, 608266 Parathyroid adenoma with cystic changes, 145001 Hyperparathyroidism-jaw tumor syndrome, 145001 Hyperparathyroidism, familial primary, 145000
CDCA7	100	99,6	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910
CDH1	99,2	99,1	96,1	96	Endometrial carcinoma, somatic, 608089 {Prostate cancer, susceptibility to}, 176807 Blepharocheilodontic syndrome 1, 119580 {Breast cancer, lobular}, 114480 Ovarian cancer, somatic, 167000 Gastric cancer, hereditary diffuse, with or without cleft lip and/or palate, 137215
CDH11	100	100	100	100	Elsahy-Waters syndrome, 211380
CDH15	99,9	98,7	100	100	Mental retardation, autosomal dominant 3, 612580
CDH2	99,3	97,7	100	100	Arrhythmogenic right ventricular dysplasia, familial, 14, 618920 Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929
CDH23	100	100	100	100	{Pituitary adenoma 5, multiple types}, 617540 Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1D, 601067
CDH3	100	99,5	100	100	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 Hypotrichosis, congenital, with juvenile macular dystrophy, 601553
CDHR1	99,2	98,1	100	100	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660
C15orf41	85,9	85,6	100	100	Dyserythropoietic anemia, congenital, type Ib, 615631
CDK10	100	99,9	100	100	Al Kaissi syndrome, 617694
CDK13	98	92,7	100	100	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360
CDK19	100	99,9	100	100	Developmental and epileptic encephalopathy 87, 618916
CDK4	100	99,7	100	100	{Melanoma, cutaneous malignant, 3}, 609048
CDK5	100	100	100	100	?Lissencephaly 7 with cerebellar hypoplasia, 616342
CDK5RAP2	99,8	98,9	100	100	Microcephaly 3, primary, autosomal recessive, 604804

CDK6	100	99,6	100	100	?Microcephaly 12, primary, autosomal recessive, 616080
CDK8	99,7	97,9	100	100	Intellectual developmental disorder with hypotonia and behavioral abnormalities, 618748
CDKL5	91,7	90,2	92,3	91,7	Developmental and epileptic encephalopathy 2, 300672
CDKN1A	100	100	100	100	No OMIM disease ID
CDKN1B	100	99,8	100	100	Multiple endocrine neoplasia, type IV, 610755
CDKN1C	88	77,8	99,3	97,3	IMAGE syndrome, 614732 Beckwith-Wiedemann syndrome, 130650
CDKN2A	92,3	92,1	100	100	{Melanoma and neural system tumor syndrome}, 155755 {Melanoma, cutaneous malignant, 2}, 155601 {Melanoma-pancreatic cancer syndrome}, 606719
CDKN2B	100	99,9	100	100	No OMIM disease ID
CDKN2B-AS1					No OMIM disease ID
CDKN2C	100	100	100	100	No OMIM disease ID
CDON	100	99,6	100	100	Holoprosencephaly 11, 614226
CDSN	100	100	100	100	Hypotrichosis 2, 146520 Peeling skin syndrome 1, 270300
CDT1	99,7	97,5	100	99,1	Meier-Gorlin syndrome 4, 613804
CEACAM16	100	99,5	100	100	Deafness, autosomal recessive 113, 618410 Deafness, autosomal dominant 4B, 614614
CEBPA	98,6	83,9	99,3	94,7	Leukemia, acute myeloid, somatic, 601626 ?Leukemia, acute myeloid, 601626
CEBPE	100	100	100	100	Specific granule deficiency, 245480
CEL	89,7	88	100	99,8	Maturity-onset diabetes of the young, type VIII, 609812
CELA2A	98,3	95,5	100	100	Abdominal obesity-metabolic syndrome 4, 618620
CELSR1	94,7	92,8	99,5	98,7	No OMIM disease ID
CENPE	98,2	92,2	100	100	?Microcephaly 13, primary, autosomal recessive, 616051
CENPF	99,8	98,5	100	100	Stromme syndrome, 243605
CENPJ	100	99,6	100	100	Microcephaly 6, primary, autosomal recessive, 608393 ?Seckel syndrome 4, 613676

CEP104	100	99,2	100	100	Joubert syndrome 25, 616781
CEP120	100	99,5	100	100	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP135	99,1	93,6	100	100	Microcephaly 8, primary, autosomal recessive, 614673
CEP152	99,7	98,2	100	100	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823
CEP164	99,9	98,3	100	100	Nephronophthisis 15, 614845
CEP19	100	100	100	100	Morbid obesity and spermatogenic failure, 615703
CEP250	100	99,2	100	100	Cone-rod dystrophy and hearing loss 2, 618358
CEP290	96,1	90	100	100	?Bardet-Biedl syndrome 14, 615991 Leber congenital amaurosis 10, 611755 Senior-Loken syndrome 6, 610189 Meckel syndrome 4, 611134 Joubert syndrome 5, 610188
CEP41	99,8	97,4	100	100	Joubert syndrome 15, 614464
CEP55	100	99,8	100	100	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500
CEP57	99,2	93	100	100	Mosaic variegated aneuploidy syndrome 2, 614114
CEP63	99,3	96,5	100	100	?Seckel syndrome 6, 614728
CEP78	98,9	96,8	100	100	Cone-rod dystrophy and hearing loss, 617236
CEP83	99,8	97,4	100	100	Nephronophthisis 18, 615862
CEP89	96	94,5	100	100	No OMIM disease ID
CERKL	99,5	96,9	100	100	Retinitis pigmentosa 26, 608380
CERS1	75,4	63,7	94,2	86,4	?Epilepsy, progressive myoclonic, 8, 616230
CERS3	99,9	98,9	100	100	Ichthyosis, congenital, autosomal recessive 9, 615023
CERT1	90,2	87,3	100	100	Mental retardation, autosomal dominant 34, 616351
CES1	99,8	99,3	99,9	99,9	Drug metabolism, altered, CES1-related, 618057
CETP	100	99,9	100	100	[High density lipoprotein cholesterol level QTL 10], 143470 Hyperalphalipoproteinemia, 143470

WDR66	100	100	100	100	Spermatogenic failure 33, 618152
CFAP298	100	99,7	100	100	Ciliary dyskinesia, primary, 26, 615500
CFAP300	99,3	95,9	100	100	Ciliary dyskinesia, primary, 38, 618063
CFAP410	100	99,3	100	100	Spondylometaphyseal dysplasia, axial, 602271 Retinal dystrophy with macular staphyloma, 617547
CFAP43	99,9	98,9	100	100	Hydrocephalus, normal pressure, 1, 236690 Spermatogenic failure 19, 617592
CFAP44	99,8	98,9	100	100	?Spermatogenic failure 20, 617593
CFAP53	99,6	97,4	100	100	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFAP58	99,9	98,7	100	100	Spermatogenic failure 49, 619144
CFAP69	98,7	93,5	100	100	Spermatogenic failure 24, 617959
CFB	100	100	100	100	?Complement factor B deficiency, 615561 {Macular degeneration, age-related, 14, reduced risk of}, 615489 {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924
CFC1	84,2	74,1	100	100	Heterotaxy, visceral, 2, autosomal, 605376
CFD	89,3	83,7	100	100	Complement factor D deficiency, 613912
CFH	99,9	99	100	99,9	Complement factor H deficiency, 609814 {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 Basal laminar drusen, 126700 {Macular degeneration, age-related, 4}, 610698
CFHR1	96,4	94,9	95,4	93,8	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075
CFHR2	76	74,3	76,5	76,1	No OMIM disease ID
CFHR3	94	92,2	96	95,2	{Macular degeneration, age-related, reduced risk of}, 603075 {Hemolytic uremic syndrome, atypical, susceptibility to}, 235400
CFHR4	100	99,9	100	100	No OMIM disease ID
CFHR5	99,6	98,4	100	100	Nephropathy due to CFHR5 deficiency, 614809
CFI	99,2	96,8	100	100	{Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 Complement factor I deficiency, 610984 {Macular degeneration, age-related, 13, susceptibility to}, 615439
CFL2	100	99,6	100	100	Nemaline myopathy 7, autosomal recessive, 610687

CFP	100	99	100	100	Properdin deficiency, X-linked, 312060
CFTR	99,6	97,9	100	100	{Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400 Cystic fibrosis, 219700 Congenital bilateral absence of vas deferens, 277180 {Pancreatitis, hereditary}, 167800 Sweat chloride elevation without CF, 0 {Hypertrypsinemia, neonatal}, 0
CHAMP1	100	100	100	100	Mental retardation, autosomal dominant 40, 616579
CHAT	93,5	85,7	100	100	Myasthenic syndrome, congenital, 6, presynaptic, 254210
CHCHD10	59,1	43,9	100	100	Spinal muscular atrophy, Jokela type, 615048 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 ?Myopathy, isolated mitochondrial, autosomal dominant, 616209
CHCHD2	98,4	83,8	100	100	Parkinson disease 22, autosomal dominant, 616710
CHD1	99,3	94,9	100	100	Pilarowski-Bjornsson syndrome, 617682
CHD2	99,4	99,2	100	100	Epileptic encephalopathy, childhood-onset, 615369
CHD3	94,8	92,6	99,8	99,5	Snijders Blok-Campeau syndrome, 618205
CHD4	100	99,9	100	100	Sifrim-Hitz-Weiss syndrome, 617159
CHD7	100	99,5	100	100	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
CHD8	100	99,9	100	100	{Autism, susceptibility to, 18}, 615032
CHEK2	85	81,5	100	100	Li-Fraumeni syndrome, 609265 Osteosarcoma, somatic, 259500 {Prostate cancer, familial, susceptibility to}, 176807 {Breast cancer, susceptibility to}, 114480 {Breast and colorectal cancer, susceptibility to}, 0
CHIT1	99,7	98,1	100	100	[Chitotriosidase deficiency], 614122
CHKB	100	99,7	100	100	Muscular dystrophy, congenital, megaconial type, 602541
CHM	98,5	94,5	98,8	97,4	Choroideremia, 303100
CHMP1A	100	99,8	100	100	Pontocerebellar hypoplasia, type 8, 614961
CHMP2B	99,7	96,7	100	100	Amyotrophic lateral sclerosis 17, 614696 Dementia, familial, nonspecific, 600795

CHMP4B	100	99,3	100	100	Cataract 31, multiple types, 605387
CHN1	99,6	98,8	97	97	Duane retraction syndrome 2, 604356
CHP1	98,5	89,1	100	100	?Spastic ataxia 9, autosomal recessive, 618438
CHRDL1	100	99,8	100	100	Megalocornea 1, X-linked, 309300
CHRM2	100	99,9	100	100	No OMIM disease ID
CHRM3	100	100	100	100	Prune belly syndrome, 100100
CHRNA1	100	99,2	100	100	Myasthenic syndrome, congenital, 1A, slow-channel, 601462 Myasthenic syndrome, congenital, 1B, fast-channel, 608930 Multiple pterygium syndrome, lethal type, 253290
CHRNA2	100	100	100	100	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA3	100	99,4	100	100	{Lung cancer susceptibility 2}, 612052 Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT, 191800
CHRNA4	98,3	96,2	100	100	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction, susceptibility to}, 188890
CHRNB1	100	99,4	100	100	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314 Myasthenic syndrome, congenital, 2A, slow-channel, 616313
CHRNB2	99,3	96	100	100	Epilepsy, nocturnal frontal lobe, 3, 605375
CHRND	99,7	97,9	100	100	?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323 ?Myasthenic syndrome, congenital, 3A, slow-channel, 616321 Myasthenic syndrome, congenital, 3B, fast-channel, 616322 Multiple pterygium syndrome, lethal type, 253290
CHRNE	100	100	100	100	Myasthenic syndrome, congenital, 4A, slow-channel, 605809 Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931 Myasthenic syndrome, congenital, 4B, fast-channel, 616324
CHRNG	100	100	100	100	Escobar syndrome, 265000 Multiple pterygium syndrome, lethal type, 253290
CHST11	100	100	100	100	?Osteochondrodysplasia, brachydactyly, and overlapping malformed digits, 618167
CHST14	99,9	98,9	100	100	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	100	99,4	100	100	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHST6	100	100	100	100	Macular corneal dystrophy, 217800
CHST8	100	100	100	100	?Peeling skin syndrome 3, 616265

CHSY1	97,2	95,7	99,7	98	Temtamy preaxial brachydactyly syndrome, 605282
CHUK	100	99,1	100	100	Cocoon syndrome, 613630
CIB1	97,3	93,6	100	100	Epidermodysplasia verruciformis 3, 618267
CIB2	99,7	97	100	99,9	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
FAM92A	90,6	86	100	100	?Polydactyly, postaxial, type A9, 618219
CIC	63,3	63,3	100	99,9	Mental retardation, autosomal dominant 45, 617600
CIDEC	100	97,9	100	100	?Lipodystrophy, familial partial, type 5, 615238
CIITA	100	99,5	100	100	Bare lymphocyte syndrome, type II, complementation group A, 209920 {Rheumatoid arthritis, susceptibility to}, 180300
CILK1	99,9	98,7	100	100	Endocrine-cerebroosteodysplasia, 612651 {Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924
CISD2	83,4	83,4	100	100	Wolfram syndrome 2, 604928
CIT	100	99,4	100	100	Microcephaly 17, primary, autosomal recessive, 617090
CITED2	99,2	99	100	100	Atrial septal defect 8, 614433 Ventricular septal defect 2, 614431
CKAP2L	99,7	98,6	100	100	Filippi syndrome, 272440
CLCC1	99,8	98	100	100	Retinitis pigmentosa 32, 609913
CLCF1	100	99,4	100	100	Cold-induced sweating syndrome 2, 610313
CLCN1	100	99,2	100	100	Myotonia congenita, dominant, 160800 Myotonia congenita, recessive, 255700 Myotonia levior, recessive, 0
CLCN2	100	99,5	100	100	{Epilepsy, juvenile absence, susceptibility to, 2}, 607628 {Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628 {Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628 Leukoencephalopathy with ataxia, 615651 Hyperaldosteronism, familial, type II, 605635
CLCN4	99,9	98,9	100	100	Raynaud-Claes syndrome, 300114
CLCN5	99,9	98,3	100	100	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990 Dent disease, 300009 Hypophosphatemic rickets, 300554 Nephrolithiasis, type I, 310468

CLCN7	99,7	98,4	100	100	Osteopetrosis, autosomal recessive 4, 611490 Osteopetrosis, autosomal dominant 2, 166600 Hypopigmentation, organomegaly, and delayed myelination and development, 618541
CLCNKA	99,8	97,8	100	100	Bartter syndrome, type 4b, digenic, 613090
CLCNKB	99,1	95,9	100	100	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090
CLDN1	100	100	100	100	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626
CLDN10	100	100	100	100	HELIX syndrome, 617671
CLDN14	100	99,7	100	100	Deafness, autosomal recessive 29, 614035
CLDN16	100	100	100	100	Hypomagnesemia 3, renal, 248250
CLDN19	98,5	93,1	100	100	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLDN9	100	100	100	100	?Deafness, autosomal recessive 116, 619093
CLEC4D	100	99,8	100	100	No OMIM disease ID
CLEC7A	100	100	100	100	{Aspergillosis, susceptibility to}, 614079 Candidiasis, familial, 4, autosomal recessive, 613108
CLIC2	99,9	96,5	100	100	?Mental retardation, X-linked, syndromic 32, 300886
CLIC5	89,9	88	100	100	?Deafness, autosomal recessive 103, 616042
CLIP1	100	99	100	100	No OMIM disease ID
CLMP	100	99,6	100	100	Congenital short bowel syndrome, 615237
CLN3	92,5	91,8	92,5	92,5	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	69,3	66,3	72,1	71,6	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	99,9	97,1	100	100	Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300 Ceroid lipofuscinosis, neuronal, 6, 601780
CLN8	83,5	83,5	100	100	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLP1	100	100	100	100	Pontocerebellar hypoplasia, type 10, 615803
CLPB	94,9	94,9	100	100	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CLPP	100	99,1	100	100	Perrault syndrome 3, 614129

CLPX	99,9	99,4	100	100	?Protoporphyria, erythropoietic, 2, 618015
CLRN1	100	99,8	100	100	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902
CLRN2	99,7	96,6	100	100	No OMIM disease ID
CLTC	100	99,9	100	100	Mental retardation, autosomal dominant 56, 617854
CLTCL1	98,6	98,2	100	100	No OMIM disease ID
CLUAP1	100	99,8	100	100	No OMIM disease ID
CMAS	99,9	97,8	100	100	No OMIM disease ID
CNBP	100	100	100	100	Myotonic dystrophy 2, 602668
CNGA1	91,7	86,3	91	91	Retinitis pigmentosa 49, 613756
CNGA3	100	99,7	100	100	Achromatopsia 2, 216900
CNGB1	99,4	97,5	100	100	Retinitis pigmentosa 45, 613767
CNGB3	99,4	95,9	100	100	Achromatopsia 3, 262300
CNKSR2	95,5	90,8	100	100	Mental retardation, X-linked, syndromic, Houge type, 301008
CNNM2	100	100	100	100	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNNM4	99,8	98,9	99,7	98,8	Jalili syndrome, 217080
CNOT1	100	99,9	100	100	Holoprosencephaly 12, with or without pancreatic agenesis, 618500 Vissers-Bodmer syndrome, 619033
CNOT2	99,9	99,5	100	100	Intellectual developmental disorder with nasal speech, dysmorphic facies, and variable skeletal anomalies, 618608
CNOT3	100	100	100	100	Intellectual developmental disorder with speech delay, autism, and dysmorphic facies, 618672
CNPY3	100	99,3	100	100	Developmental and epileptic encephalopathy 60, 617929
CNTN1	99,9	98,9	100	100	?Myopathy, congenital, Compton-North, 612540
CNTN2	92,7	92,7	100	100	?Epilepsy, myoclonic, familial adult, 5, 615400
CNTN3	100	99,8	100	100	No OMIM disease ID
CNTNAP1	100	99,8	100	100	Lethal congenital contracture syndrome 7, 616286 Hypomyelinating neuropathy, congenital, 3, 618186

CNTNAP2	100	99,8	100	100	{Autism susceptibility 15}, 612100 Pitt-Hopkins like syndrome 1, 610042 Cortical dysplasia-focal epilepsy syndrome, 610042
COA1	100	100	100	100	No OMIM disease ID
COA3	100	100	100	100	?Mitochondrial complex IV deficiency, nuclear type 14, 619058
COA5	99,1	88,9	85,2	85,2	?Mitochondrial complex IV, deficiency, nuclear type 9, 616500
COA6	99,9	98,4	100	100	Mitochondrial complex IV deficiency, nuclear type 13, 616501
COA7	100	100	100	100	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387
COA8	81,9	80,7	93,5	93,4	Mitochondrial complex IV deficiency, nuclear type 17, 619061
COASY	100	100	100	100	Neurodegeneration with brain iron accumulation 6, 615643 Pontocerebellar hypoplasia, type 12, 618266
COCH	95,2	93,2	100	100	?Deafness, autosomal recessive 110, 618094 Deafness, autosomal dominant 9, 601369
COG1	100	100	100	100	Congenital disorder of glycosylation, type IIg, 611209
COG2	99,9	98,5	100	100	?Congenital disorder of glycosylation, type IIq, 617395
COG4	100	99,9	100	100	Saul-Wilson syndrome, 618150 Congenital disorder of glycosylation, type IIj, 613489
COG5	99,7	97,6	100	100	Congenital disorder of glycosylation, type IIi, 613612
COG6	99,1	93,9	100	100	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328
COG7	100	100	100	100	Congenital disorder of glycosylation, type IIe, 608779
COG8	99,9	98,6	100	100	Congenital disorder of glycosylation, type IIh, 611182
COL10A1	100	98,4	100	100	Metaphyseal chondrodysplasia, Schmid type, 156500
COL11A1	96,2	92,8	100	100	Stickler syndrome, type II, 604841 Marshall syndrome, 154780 ?Deafness, autosomal dominant 37, 618533 {Lumbar disc herniation, susceptibility to}, 603932 Fibrochondrogenesis 1, 228520
COL11A2	100	99,7	100	100	Deafness, autosomal dominant 13, 601868 Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150 Fibrochondrogenesis 2, 614524

					Deafness, autosomal recessive 53, 609706 Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840
COL12A1	100	99,4	100	100	Bethlem myopathy 2, 616471 ?Ullrich congenital muscular dystrophy 2, 616470
COL13A1	93,9	93,8	100	100	Myasthenic syndrome, congenital, 19, 616720
COL14A1	100	99,4	100	100	No OMIM disease ID
COL17A1	98,7	96,8	100	100	Epithelial recurrent erosion dystrophy, 122400 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa, junctional, localisata variant, 226650
COL18A1	98,1	95,6	100	100	Knobloch syndrome, type 1, 267750 Glaucoma, primary closed-angle, 618880
COL1A1	99,9	98,6	100	100	Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1, 619115 Osteogenesis imperfecta, type I, 166200 Osteogenesis imperfecta, type IV, 166220 Osteogenesis imperfecta, type II, 166210 {Bone mineral density variation QTL, osteoporosis}, 166710 Caffey disease, 114000 Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060 Osteogenesis imperfecta, type III, 259420
COL1A2	99,4	96,9	100	100	Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2, 619120 {Osteoporosis, postmenopausal}, 166710 Ehlers-Danlos syndrome, cardiac valvular type, 225320 Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type IV, 166220 Osteogenesis imperfecta, type III, 259420
COL25A1	95,8	95,3	99,9	99,9	Fibrosis of extraocular muscles, congenital, 5, 616219
COL27A1	99,9	99,7	100	100	Steel syndrome, 615155
COL2A1	100	99,7	100	100	Achondrogenesis, type II or hypochondrogenesis, 200610 Spondyloperipheral dysplasia, 271700 Kniest dysplasia, 156550 Stickler syndrome, type I, 108300 Osteoarthritis with mild chondrodysplasia, 604864 Platyspondylic skeletal dysplasia, Torrance type, 151210 Avascular necrosis of the femoral head, 608805 ?Epiphyseal dysplasia, multiple, with myopia and deafness, 132450

					SED congenita, 183900 Legg-Calve-Perthes disease, 150600 SMED Strudwick type, 184250 Czech dysplasia, 609162 Stickler sydrome, type I, nonsyndromic ocular, 609508 Spondyloepiphyseal dysplasia, Stanescu type, 616583 Vitreoretinopathy with phalangeal epiphyseal dysplasia, 0
COL3A1	99,6	97,6	100	100	Ehlers-Danlos syndrome, vascular type, 130050 Polymicrogyria with or without vascular-type EDS, 618343
COL4A1	98,7	97,4	100	100	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Brain small vessel disease with or without ocular anomalies, 175780 {Hemorrhage, intracerebral, susceptibility to}, 614519 ?Retinal arteries, tortuosity of, 180000 Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564
COL4A2	100	99,6	100	100	Brain small vessel disease 2, 614483 {Hemorrhage, intracerebral, susceptibility to}, 614519
COL4A3	98,7	98	100	100	Alport syndrome 2, autosomal recessive, 203780 Alport syndrome 3, autosomal dominant, 104200 Hematuria, benign familial, 141200
COL4A4	99,9	98,2	100	100	Alport syndrome 2, autosomal recessive, 203780 Hematuria, familial benign, 141200
COL4A5	97,8	89,1	100	99,8	Alport syndrome 1, X-linked, 301050
COL4A6	97,5	93,3	100	99,9	?Deafness, X-linked 6, 300914
COL5A1	98,8	98	100	99,9	Ehlers-Danlos syndrome, classic type, 1, 130000
COL5A2	100	99,5	100	100	Ehlers-Danlos syndrome, classic type, 2, 130010
COL6A1	100	99,4	100	100	Ullrich congenital muscular dystrophy 1, 254090 Bethlem myopathy 1, 158810
COL6A2	100	99,8	100	100	Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090 ?Myosclerosis, congenital, 255600
COL6A3	100	99,8	100	100	Bethlem myopathy 1, 158810 Dystonia 27, 616411 Ullrich congenital muscular dystrophy 1, 254090
COL6A5	99,9	99,5	100	100	No OMIM disease ID

COL7A1	99,9	99,1	100	100	EBD inversa, 226600 Epidermolysis bullosa dystrophica, AR, 226600 Toenail dystrophy, isolated, 607523 EBD, Bart type, 132000 Transient bullous of the newborn, 131705 Epidermolysis bullosa dystrophica, AD, 131750 Epidermolysis bullosa pruriginosa, 604129 Epidermolysis bullosa, pretibial, 131850 EBD, localisata variant, 0
COL8A2	99,9	97,9	100	100	Corneal dystrophy, posterior polymorphous 2, 609140 Corneal dystrophy, Fuchs endothelial, 1, 136800
COL9A1	100	99,2	100	100	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	99,9	99	100	100	?Stickler syndrome, type V, 614284 Epiphyseal dysplasia, multiple, 2, 600204
COL9A3	98,7	95,5	99,7	98,6	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 {Intervertebral disc disease, susceptibility to}, 603932
COLEC10	100	100	100	100	3MC syndrome 3, 248340
COLEC11	100	100	100	100	3MC syndrome 2, 265050
COLGALT1	93,3	89	98,6	97	Brain small vessel disease 3, 618360
COLQ	100	99,2	100	100	Myasthenic syndrome, congenital, 5, 603034
COMP	93,4	92,3	100	100	Epiphyseal dysplasia, multiple, 1, 132400 Pseudoachondroplasia, 177170
COMT	100	99,9	100	100	{Schizophrenia, susceptibility to}, 181500 {Panic disorder, susceptibility to}, 167870
COPA	100	99,2	100	100	{Autoimmune interstitial lung, joint, and kidney disease}, 616414
COPB2	99,9	99,3	100	100	?Microcephaly 19, primary, autosomal recessive, 617800
COQ2	98	95,3	97,2	97,2	{Multiple system atrophy, susceptibility to}, 146500 Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	90,9	89,3	100	100	Coenzyme Q10 deficiency, primary, 7, 616276
COQ5	100	100	100	100	?Coenzyme Q10 deficiency, primary, 9, 619028
COQ6	99,9	98,4	100	100	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	100	99,8	100	100	?Coenzyme Q10 deficiency, primary, 8, 616733

COQ8A	100	99,5	100	100	Coenzyme Q10 deficiency, primary, 4, 612016
COQ8B	100	99,3	100	100	Nephrotic syndrome, type 9, 615573
COQ9	100	97,9	100	100	Coenzyme Q10 deficiency, primary, 5, 614654
CORIN	100	99,9	100	100	Preeclampsia/eclampsia 5, 614595
CORO1A	100	98,6	100	100	Immunodeficiency 8, 615401
COX10	100	100	100	100	Mitochondrial complex IV deficiency, nuclear type 3, 619046
COX14	100	100	100	100	?Mitochondrial complex IV deficiency, nuclear type 10, 619053
COX15	99,9	98,8	100	100	Mitochondrial complex IV deficiency, nuclear type 6, 615119
COX20	97,8	88,3	100	100	Mitochondrial complex IV deficiency, nuclear type 11, 619054
COX4I1	100	100	100	100	Mitochondrial complex IV deficiency, nuclear type 16, 619060
COX4I2	100	100	100	100	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX5A	74,7	47,1	100	100	?Mitochondrial complex IV deficiency, nuclear type 20, 619064
COX5B	100	100	100	100	No OMIM disease ID
COX6A1	100	99,5	100	100	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
COX6A2	99,2	93,7	100	100	Mitochondrial complex IV deficiency, nuclear type 18, 619062
COX6B1	100	100	100	100	Mitochondrial complex IV deficiency, nuclear type 7, 619051
COX6B2	100	99,8	100	100	No OMIM disease ID
COX6C	100	97,4	100	100	No OMIM disease ID
COX7A1	100	99,9	100	100	No OMIM disease ID
COX7A2	100	99,8	100	100	No OMIM disease ID
COX7B	78,2	49,4	100	100	Linear skin defects with multiple congenital anomalies 2, 300887
COX7B2	100	100	100	100	No OMIM disease ID
COX7C	99,3	86,9	100	100	No OMIM disease ID
COX8A	100	100	100	100	?Mitochondrial complex IV deficiency, nuclear type 15, 619059
COX8C	100	99,9	100	100	No OMIM disease ID

CP	94,8	88,9	100	100	[Hypoceruloplasminemia, hereditary], 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 Cerebellar ataxia, 604290
CPA6	99,6	97,5	100	100	Febrile seizures, familial, 11, 614418 Epilepsy, familial temporal lobe, 5, 614417
CPAMD8	95,8	92,8	99,9	99,6	Anterior segment dysgenesis 8, 617319
CPLANE1	99,7	98,4	100	100	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
CPLX1	100	100	100	100	Developmental and epileptic encephalopathy 63, 617976
CPN1	99,9	99,4	100	100	Carboxypeptidase N deficiency, 212070
CPOX	99,9	95,4	100	100	Harderoporphyria, 618892 Coproporphyria, 121300
CPS1	100	99,9	100	100	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371
CPSF1	98,2	96,5	100	100	Myopia 27, 618827
CPT1A	100	98,9	100	100	CPT deficiency, hepatic, type IA, 255120
CPT1C	100	99,9	100	100	?Spastic paraplegia 73, autosomal dominant, 616282
CPT2	98,2	97,8	100	100	CPT II deficiency, myopathic, stress-induced, 255110 CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212
CR2	100	99,8	100	100	{Systemic lupus erythematosus, susceptibility to, 9}, 610927 Immunodeficiency, common variable, 7, 614699
CRADD	99,5	96,3	100	100	Mental retardation, autosomal recessive 34, with variant lissencephaly, 614499
CRAT	100	99,8	100	100	?Neurodegeneration with brain iron accumulation 8, 617917
CRB1	100	99,9	100	100	Pigmented paravenous chorioretinal atrophy, 172870 Retinitis pigmentosa-12, 600105 Leber congenital amaurosis 8, 613835
CRB2	98,5	93	100	100	Ventriculomegaly with cystic kidney disease, 219730 Focal segmental glomerulosclerosis 9, 616220
CRBN	88,2	87,7	97	92,9	Mental retardation, autosomal recessive 2, 607417
CREB1	99,7	96,7	100	100	Histiocytoma, angiomatoid fibrous, somatic, 612160

CREB3L1	100	99,9	100	100	Osteogenesis imperfecta, type XVI, 616229
CREBBP	99,7	98,5	100	100	Rubinstein-Taybi syndrome 1, 180849 Menke-Hennekam syndrome 1, 618332
CRELD1	99,9	95	100	100	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217 {Atrioventricular septal defect, susceptibility to, 2}, 606217
CRIPT	98,1	93,2	100	100	Short stature with microcephaly and distinctive facies, 615789
CRLF1	91	89,8	97,9	95,2	Cold-induced sweating syndrome 1, 272430
CRP	99,7	96,7	100	100	No OMIM disease ID
CRPPA	98,5	94,8	100	99,4	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
CRTAP	99,8	98,8	100	100	Osteogenesis imperfecta, type VII, 610682
CRTC1	99,8	99,7	100	100	Mucoepidermoid salivary gland carcinoma, 0
CRX	100	100	100	100	Cone-rod retinal dystrophy-2, 120970 Leber congenital amaurosis 7, 613829
CRYAA	99,9	97,5	100	100	Cataract 9, multiple types, 604219
CRYAB	100	99,2	100	100	Myopathy, myofibrillar, 2, 608810 Cardiomyopathy, dilated, 1II, 615184 Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869 Cataract 16, multiple types, 613763
CRYBA1	100	99,4	100	100	Cataract 10, multiple types, 600881
CRYBA2	100	100	100	100	?Cataract 42, 115900
CRYBA4	100	100	100	100	Cataract 23, 610425
CRYBB1	100	100	100	100	Cataract 17, multiple types, 611544
CRYBB2	100	100	100	100	Cataract 3, multiple types, 601547
CRYBB3	100	100	100	100	Cataract 22, 609741
CRYGB	100	99,6	100	100	Cataract 39, multiple types, autosomal dominant, 615188
CRYGC	99,8	96,9	100	100	Cataract 2, multiple types, 604307
CRYGD	100	98,9	100	100	Cataract 4, multiple types, 115700
CRYGS	94,1	86,6	100	100	Cataract 20, multiple types, 116100

CRYL1	100	99,9	100	100	No OMIM disease ID
CRYM	100	99,6	100	100	Deafness, autosomal dominant 40, 616357
CSDE1	99,9	99,5	100	100	No OMIM disease ID
CSF1R	99,9	99,3	100	100	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
CSF2RA	89,9	87,5	95,6	92,1	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF2RB	100	99	100	100	Surfactant metabolism dysfunction, pulmonary, 5, 614370
CSF3R	99,6	98,2	100	100	Neutropenia, severe congenital, 7, autosomal recessive, 617014
CSGALNACT1	100	99,8	100	100	Skeletal dysplasia, mild, with joint laxity and advanced bone age, 618870
CSNK1D	97,7	95,1	100	100	Advanced sleep-phase syndrome, familial, 2, 615224
CSNK2A1	81,5	77,7	94	94	Okur-Chung neurodevelopmental syndrome, 617062
CSNK2B	100	100	100	100	Poirier-Bienvenu neurodevelopmental syndrome, 618732
CSPP1	99,8	98,7	100	100	Joubert syndrome 21, 615636
CSRP3	100	99,1	100	100	Cardiomyopathy, hypertrophic, 12, 612124 ?Cardiomyopathy, dilated, 1M, 607482
CST3	93,4	66	100	100	Cerebral amyloid angiopathy, 105150 {Macular degeneration, age-related, 11}, 611953
CST6	98,2	92,5	100	100	?Ectodermal dysplasia 15, hypohidrotic/hair type, 618535
CSTA	100	99,8	100	100	Peeling skin syndrome 4, 607936
CSTB	99,6	89,8	100	100	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTBP1	93,2	86,9	99,5	98,6	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915
CTC1	100	99,6	100	100	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTCF	100	99,3	100	100	Mental retardation, autosomal dominant 21, 615502
CTDP1	88,4	84,3	100	99,4	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTH	100	100	100	100	Cystathioninuria, 219500
CTHRC1	93,8	87,5	100	100	Barrett esophagus/esophageal adenocarcinoma, 614266

CTLA4	100	100	100	100	{Systemic lupus erythematosus, susceptibility to}, 152700 {Diabetes mellitus, insulin-dependent, 12}, 601388 {Celiac disease, susceptibility to, 3}, 609755 Autoimmune lymphoproliferative syndrome, type V, 616100 {Hashimoto thyroiditis}, 140300
CTNNA1	99,3	98,1	100	100	Macular dystrophy, patterned, 2, 608970
CTNNA2	100	99,8	100	100	Cortical dysplasia, complex, with other brain malformations 9, 618174
CTNNA3	100	99,8	100	100	Arrhythmogenic right ventricular dysplasia, familial, 13, 615616
CTNNB1	100	99,9	100	100	Ovarian cancer, somatic, 167000 Colorectal cancer, somatic, 114500 Medulloblastoma, somatic, 155255 Hepatocellular carcinoma, somatic, 114550 Pilomatricoma, somatic, 132600 Neurodevelopmental disorder with spastic diplegia and visual defects, 615075 Exudative vitreoretinopathy 7, 617572
CTNND1	100	100	100	100	Blepharocheilodontic syndrome 2, 617681
CTNND2	93,5	91,1	97,7	95,5	No OMIM disease ID
CTNS	100	99,8	100	100	Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, atypical nephropathic, 219800
CTPS1	93	93	93	93	Immunodeficiency 24, 615897
CTR9	100	99,9	100	100	No OMIM disease ID
CTSA	100	100	100	100	Galactosialidosis, 256540
CTSB	100	100	100	100	Keratolytic winter erythema, 148370
CTSC	100	100	100	100	Periodontitis 1, juvenile, 170650 Papillon-Lefevre syndrome, 245000 Haim-Munk syndrome, 245010
CTSD	98,4	95	100	100	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	84	79,3	100	99,9	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362
CTSH	100	100	100	100	No OMIM disease ID

CTSK	100	99,9	100	100	Pycnodysostosis, 265800
CTTNBP2	99,5	97,3	100	100	No OMIM disease ID
CTU2	99,7	97,7	100	100	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142
CUBN	99,7	98,3	100	100	[Proteinuria, chronic benign], 618884 Imerslund-Grasbeck syndrome 1, 261100
CUL3	99,9	98,8	100	100	Pseudohypoaldosteronism, type IIE, 614496
CUL4B	98	90,8	99,9	99,2	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
CUL7	100	99,3	100	100	3-M syndrome 1, 273750
CUX1	96,4	94,8	99,3	98,4	Global developmental delay with or without impaired intellectual development, 618330
CUX2	99,9	99,1	100	100	Developmental and epileptic encephalopathy 67, 618141
CWC27	99,3	96,5	100	100	Retinitis pigmentosa with or without skeletal anomalies, 250410
CWF19L1	100	99,8	100	100	Spinocerebellar ataxia, autosomal recessive 17, 616127
CXCL10	100	100	100	100	No OMIM disease ID
CXCL13	100	100	100	100	No OMIM disease ID
CXCL2	100	100	100	100	No OMIM disease ID
CXCR4	100	100	100	100	WHIM syndrome, 193670 Myelokathexis, isolated, 0
CYB561	92,8	92,6	100	99,9	Orthostatic hypotension 2, 618182
CYB5A	100	100	100	100	Methemoglobinemia and ambiguous genitalia, 250790
CYB5R3	98,4	98	99,8	98,9	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYBA	95	82,4	100	100	Chronic granulomatous disease 4, autosomal recessive, 233690
CYBB	99,9	99,3	100	100	Immunodeficiency 34, mycobacteriosis, X-linked, 300645 Chronic granulomatous disease, X-linked, 306400
CYBC1	99,6	97	100	100	Chronic granulomatous disease 5, autosomal recessive, 618935
CYBRD1	100	99,9	100	100	No OMIM disease ID
CYC1	97,5	89,2	99,9	98,7	Mitochondrial complex III deficiency, nuclear type 6, 615453

CYCS	99,1	94,9	100	100	Thrombocytopenia 4, 612004
CYFIP2	100	99,4	100	100	Developmental and epileptic encephalopathy 65, 618008
CYLD	99,8	98	100	100	Cylindromatosis, familial, 132700 Brooke-Spiegler syndrome, 605041 Trichoepithelioma, multiple familial, 1, 601606
CYP11A1	99,3	96,1	100	100	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	100	100	100	100	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 Aldosteronism, glucocorticoid-remediable, 103900
CYP11B2	100	100	100	100	Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 Aldosterone to renin ratio raised, 0 {Low renin hypertension, susceptibility to}, 0
CYP17A1	100	99,5	100	100	17-alpha-hydroxylase/17,20-lyase deficiency, 202110 17,20-lyase deficiency, isolated, 202110
CYP19A1	98,8	96,8	100	100	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300
CYP1B1	100	100	100	100	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 Anterior segment dysgenesis 6, multiple subtypes, 617315
CYP21A2	97,8	88,4	100	100	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910
CYP24A1	100	99,9	100	100	Hypercalcemia, infantile, 1, 143880
CYP26B1	100	99,9	100	100	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
CYP26C1	99,7	97,1	100	99,8	Focal facial dermal dysplasia 4, 614974
CYP27A1	98,9	96,7	100	100	Cerebrotendinous xanthomatosis, 213700
CYP27B1	99,9	99,3	100	100	Vitamin D-dependent rickets, type I, 264700
CYP2A6	100	99,9	100	100	Coumarin resistance, 122700 {Lung cancer, resistance to}, 211980 {Nicotine addiction, protection from}, 188890
CYP2B6	99,9	98,2	100	100	{Efavirenz central nervous system toxicity, susceptibility to}, 614546 Efavirenz, poor metabolism of, 614546
CYP2C19	100	98,9	100	100	Clopidogrel, impaired responsiveness to, 609535 Mephenytoin poor metabolizer, 609535 Proguanil poor metabolizer, 609535 Omeprazole poor metabolizer, 609535

CYP2C8	99,9	98,6	100	100	{Drug metabolism, altered, CYP2C8-related}, 618018
CYP2C9	99,9	98,4	100	100	Warfarin sensitivity, 122700 Tolbutamide poor metabolizer, 0
CYP2R1	99,4	95,6	100	100	Rickets due to defect in vitamin D 25-hydroxylation deficiency, 600081
CYP2U1	94,8	91,5	100	99,9	Spastic paraplegia 56, autosomal recessive, 615030
CYP4F22	100	99,4	100	100	Ichthyosis, congenital, autosomal recessive 5, 604777
CYP4V2	99,9	98,4	100	100	Bietti crystalline corneoretinal dystrophy, 210370
CYP7B1	98	92,8	100	100	Spastic paraplegia 5A, autosomal recessive, 270800 Bile acid synthesis defect, congenital, 3, 613812
D2HGDH	99,2	97,2	100	100	D-2-hydroxyglutaric aciduria, 600721
DAB1	100	100	100	100	Spinocerebellar ataxia 37, 615945
DAB2IP	95,1	94,2	99,9	99,4	No OMIM disease ID
DACT1	93,6	91,1	100	99,9	?Townes-Brocks syndrome 2, 617466
DAG1	100	100	100	100	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538
DAO	100	100	100	100	No OMIM disease ID
DARS1	100	99,3	100	100	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2	94,9	94,3	100	100	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBF4	96,6	89,6	100	100	No OMIM disease ID
DBH	100	100	100	100	Orthostatic hypotension 1, due to DBH deficiency, 223360
DBR1	100	99,3	100	100	No OMIM disease ID
DBT	99,8	98	100	100	Maple syrup urine disease, type II, 248600
DCAF17	98,9	93,3	100	100	Woodhouse-Sakati syndrome, 241080
DCAF8	100	99,9	100	100	?Giant axonal neuropathy 2, autosomal dominant, 610100
DCC	100	100	100	100	Esophageal carcinoma, somatic, 133239 Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 Mirror movements 1 and/or agenesis of the corpus callosum, 157600 Colorectal cancer, somatic, 114500

DCDC2	100	99,9	100	100	Sclerosing cholangitis, neonatal, 617394 Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212
DCHS1	99,8	99,1	100	100	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390
DCLRE1C	100	99,4	100	100	Omenn syndrome, 603554 Severe combined immunodeficiency, Athabascan type, 602450
DCN	95,7	95,6	95,7	95,7	Corneal dystrophy, congenital stromal, 610048
DCPS	91,3	91,2	100	100	Al-Raqad syndrome, 616459
DCTN1	100	98,8	100	100	{Amyotrophic lateral sclerosis, susceptibility to}, 105400 Perry syndrome, 168605 Neuronopathy, distal hereditary motor, type VIIB, 607641
DCTN2	100	99,7	100	100	No OMIM disease ID
DCX	100	99,9	100	100	Subcortical laminal heterotopia, X-linked, 300067 Lissencephaly, X-linked, 300067
DCXR	98,6	93,6	100	100	[Pentosuria], 260800
DDB2	99,6	97,5	100	100	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DDC	99,7	96,4	100	100	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	97,9	95,8	100	100	Spastic paraplegia 28, autosomal recessive, 609340
DDHD2	100	99,6	100	100	Spastic paraplegia 54, autosomal recessive, 615033
DDOST	100	99,9	100	100	?Congenital disorder of glycosylation, type Ir, 614507
DDR2	100	99,9	100	100	Spondylometaepiphyseal dysplasia, short limb-hand type, 271665 Warburg-Cinotti syndrome, 618175
DDRGK1	100	99,9	100	100	Spondyloepimetaphyseal dysplasia, Shohat type, 602557
DDX11	85,2	80,7	100	100	Warsaw breakage syndrome, 613398
DDX3X	81,2	78,9	98	96,1	Intellectual developmental disorder, X-linked, syndrome, Snijders Blok type, 300958
DDX41	100	100	100	100	{Myeloproliferative/lymphoproliferative neoplasms, familial (multiple types), susceptibility to}, 616871
DDX58	99,9	99	100	100	Singleton-Merten syndrome 2, 616298
DDX59	100	100	100	100	Orofaciodigital syndrome V, 174300
DDX6	97,7	88,7	100	100	Intellectual developmental disorder with impaired language and dysmorphic facies, 618653

DEAF1	97,3	88,8	100	98,7	Vulto-van Silfout-de Vries syndrome, 615828 Neurodevelopmental disorder with hypotonia, impaired expressive language, and with or without seizures, 617171
DEF6	96,7	93,8	100	99,9	No OMIM disease ID
DEGS1	100	100	100	100	Leukodystrophy, hypomyelinating, 18, 618404
DENND5A	100	99,4	100	100	Developmental and epileptic encephalopathy 49, 617281
DEPDC5	100	99,8	100	100	Epilepsy, familial focal, with variable foci 1, 604364
DES	100	99,7	100	100	Cardiomyopathy, dilated, 1I, 604765 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 Myopathy, myofibrillar, 1, 601419
DGAT1	91,9	87,6	99,7	98,6	?Diarrhea 7, protein-losing enteropathy type, 615863
DGAT2	99,1	95,5	100	100	No OMIM disease ID
DGKE	99,8	98,1	100	100	{Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008 Nephrotic syndrome, type 7, 615008
DGUOK	100	99,4	100	100	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 Portal hypertension, noncirrhotic, 617068 Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHCR24	97,7	97,7	97,7	97,7	Desmosterolosis, 602398
DHCR7	100	100	100	100	Smith-Lemli-Opitz syndrome, 270400
DHDDS	99	95	95,2	95,2	Retinitis pigmentosa 59, 613861 Developmental delay and seizures with or without movement abnormalities, 617836 ?Congenital disorder of glycosylation, type 1bb, 613861
DHFR	92,1	78,9	100	100	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHH	100	100	100	100	46XY sex reversal 7, 233420 46XY gonadal dysgenesis with minifascicular neuropathy, 607080
DHODH	100	100	100	100	Miller syndrome, 263750
DHPS	100	99,7	93,3	93,2	Neurodevelopmental disorder with seizures and speech and walking impairment, 618480
DHTKD1	99,9	98,9	100	100	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DHX30	100	99,9	100	100	Neurodevelopmental disorder with severe motor impairment and absent language, 617804
DHX38	100	99,3	100	100	Retinitis pigmentosa 84, 618220

DIABLO	100	99,9	100	100	Deafness, autosomal dominant 64, 614152
DIAPH1	99,8	99	99,5	98	Seizures, cortical blindness, microcephaly syndrome, 616632 Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900
DIAPH2	95,9	87,6	99,9	99	?Premature ovarian failure 2A, 300511
DIAPH3	99,6	97	100	100	Auditory neuropathy, autosomal dominant, 1, 609129
DICER1	99,8	99	100	100	GLOW syndrome, somatic mosaic, 618272 Rhabdomyosarcoma, embryonal, 2, 180295 Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800 Pleuropulmonary blastoma, 601200
DIP2B	100	99,3	100	100	Mental retardation, FRA12A type, 136630
DIS3L2	100	99,8	100	100	Perlman syndrome, 267000
DISP1	100	99,9	100	100	No OMIM disease ID
DKC1	99,8	98,7	100	99,7	Dyskeratosis congenita, X-linked, 305000
DLAT	100	99,7	100	100	Pyruvate dehydrogenase E2 deficiency, 245348
DLC1	100	99,9	100	100	Colorectal cancer, somatic, 114500
DLD	100	99,7	100	100	Dihydrolipoamide dehydrogenase deficiency, 246900
DLG3	99,1	93,2	100	100	Mental retardation, X-linked 90, 300850
DLG4	99,1	99	98,8	98,8	Intellectual developmental disorder 62, 618793
DLL3	92,1	87	100	99,1	Spondylocostal dysostosis 1, autosomal recessive, 277300
DLL4	100	99,2	100	100	Adams-Oliver syndrome 6, 616589
DLST	96,7	90,3	100	100	Paragangliomas 7, 618475
DLX3	99,9	98,4	100	100	Trichodontoosseous syndrome, 190320 Amelogenesis imperfecta, type IV, 104510
DLX4	100	100	100	100	?Orofacial cleft 15, 616788
DLX5	100	99,9	100	100	Split-hand/foot malformation 1, 183600 ?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600
DLX6	100	100	100	100	No OMIM disease ID
DMAC1	100	100	100	100	No OMIM disease ID

DMAC2	98,3	98,3	100	100	No OMIM disease ID
DMAC2L	100	100	100	100	No OMIM disease ID
DMD	99,6	98,6	100	100	Cardiomyopathy, dilated, 3B, 302045 Becker muscular dystrophy, 300376 Duchenne muscular dystrophy, 310200
DMGDH	100	99,7	100	100	Dimethylglycine dehydrogenase deficiency, 605850
DMP1	100	99,9	100	100	Hypophosphatemic rickets, AR, 241520
DMPK	99,8	98,4	100	100	Myotonic dystrophy 1, 160900
DMRT1	100	99,8	100	100	No OMIM disease ID
DMRT2	97,7	88,4	100	100	No OMIM disease ID
DMXL2	99,9	99,1	100	100	Developmental and epileptic encephalopathy 81, 618663 ?Deafness, autosomal dominant 71, 617605 ?Polyendocrine-polyneuropathy syndrome, 616113
DNA2	99,8	98,3	100	100	?Seckel syndrome 8, 615807 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156
DNAAF1	100	99,8	100	100	Ciliary dyskinesia, primary, 13, 613193
DNAAF2	99,9	98,9	100	100	Ciliary dyskinesia, primary, 10, 612518
DNAAF3	99,5	96,1	100	100	Ciliary dyskinesia, primary, 2, 606763
DNAAF4	99,8	97	100	100	{Dyslexia, susceptibility to, 1}, 127700 Ciliary dyskinesia, primary, 25, 615482
DNAAF5	84,6	78,6	99,1	97,5	Ciliary dyskinesia, primary, 18, 614874
PIH1D3	99,1	92,5	100	100	Ciliary dyskinesia, primary, 36, X-linked, 300991
DNAH1	100	99,7	100	100	?Ciliary dyskinesia, primary, 37, 617577 Spermatogenic failure 18, 617576
DNAH10	99,9	99,4	100	100	No OMIM disease ID
DNAH11	99,9	99	100	100	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH17	100	99,6	100	99,9	Spermatogenic failure 39, 618643
DNAH5	99,9	99,3	100	100	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAH8	99,9	99	100	100	Spermatogenic failure 46, 619095

DNAH9	99,5	98,3	100	100	Ciliary dyskinesia, primary, 40, 618300
DNAI1	100	100	100	100	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	98,6	96,2	100	100	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJA3	98,6	96	100	100	No OMIM disease ID
DNAJB11	100	99,5	100	100	Polycystic kidney disease 6 with or without polycystic liver disease, 618061
DNAJB13	100	100	100	100	Ciliary dyskinesia, primary, 34, 617091
DNAJB2	100	100	100	100	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881
DNAJB5	95,7	91,3	100	100	No OMIM disease ID
DNAJB6	96,5	88,5	100	100	Muscular dystrophy, limb-girdle, autosomal dominant 1, 603511
DNAJC12	87,4	87,4	100	100	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC19	98,9	96,2	100	100	3-methylglutaconic aciduria, type V, 610198
DNAJC21	99,8	98,7	100	100	Bone marrow failure syndrome 3, 617052
DNAJC3	100	99,7	100	100	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192
DNAJC5	100	100	100	100	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350
DNAJC6	100	99,4	100	100	Parkinson disease 19b, early-onset, 615528 Parkinson disease 19a, juvenile-onset, 615528
DNAL1	99	96,8	100	100	Ciliary dyskinesia, primary, 16, 614017
DNAL4	100	98,9	100	100	?Mirror movements 3, 616059
DNASE1	100	99,9	100	100	{Systemic lupus erythematosus, susceptibility to}, 152700
DNASE1L3	100	100	100	100	Systemic lupus erythematosus 16, 614420
DNASE2	99,7	97,1	100	100	No OMIM disease ID
DNM1	92,6	89,1	97,4	97,4	Developmental and epileptic encephalopathy 31, 616346
DNM1L	99,9	98,5	100	100	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 Optic atrophy 5, 610708
DNM2	98,1	94,5	100	100	Lethal congenital contracture syndrome 5, 615368 Charcot-Marie-Tooth disease, axonal type 2M, 606482

					Centronuclear myopathy 1, 160150 Charcot-Marie-Tooth disease, dominant intermediate B, 606482
DNMBP	100	99,7	100	100	Cataract 48, 618415
DNMT1	99,2	99	99,7	99,2	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 Neuropathy, hereditary sensory, type IE, 614116
DNMT3A	99,8	98,6	100	100	Heyn-Sproul-Jackson syndrome, 618724 Acute myeloid leukemia, somatic, 601626 Tatton-Brown-Rahman syndrome, 615879
DNMT3B	100	100	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK2	100	99,6	100	100	Immunodeficiency 40, 616433
DOCK3	100	99	100	100	Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292
DOCK6	99,3	98,9	100	100	Adams-Oliver syndrome 2, 614219
DOCK7	99,8	98,2	100	99,9	Developmental and epileptic encephalopathy 23, 615859
DOCK8	100	99,6	100	100	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
DOK7	95,1	91,6	100	100	Fetal akinesia deformation sequence 3, 618389 Myasthenic syndrome, congenital, 10, 254300
DOLK	100	100	100	100	Congenital disorder of glycosylation, type Im, 610768
DONSON	91,7	85,3	100	100	Microcephaly-micromelia syndrome, 251230 Microcephaly, short stature, and limb abnormalities, 617604
DPAGT1	100	100	100	100	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPCD	100	100	100	100	No OMIM disease ID
DPF2	99,9	98,4	100	100	Coffin-Siris syndrome 7, 618027
DPH1	100	99,9	100	100	Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901
DPM1	98,2	91,3	99,7	97,1	Congenital disorder of glycosylation, type Ie, 608799
DPM2	100	98,7	100	100	Congenital disorder of glycosylation, type Iu, 615042
DPM3	100	100	100	100	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937 ?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992
DPP6	99,7	97,8	99,4	97,6	Mental retardation, autosomal dominant 33, 616311 {Ventricular fibrillation, paroxysmal familial, 2}, 612956

DPY19L2	74,5	71,2	100	100	Spermatogenic failure 9, 613958
DPYD	99,7	97,7	100	100	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270
DPYS	100	99,9	100	100	Dihydropyrimidinuria, 222748
DRAM2	100	99,9	100	100	Cone-rod dystrophy 21, 616502
DRC1	100	99,5	100	100	Ciliary dyskinesia, primary, 21, 615294
DRD4	93,8	81,8	100	99,7	{Attention deficit-hyperactivity disorder}, 143465 Autonomic nervous system dysfunction, 0
DRP2	99,1	96,5	100	99,9	No OMIM disease ID
DSC2	99,8	98,4	100	100	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 Arrhythmogenic right ventricular dysplasia 11, 610476
DSC3	99,5	96,8	100	100	?Hypotrichosis and recurrent skin vesicles, 613102
DSE	99	96,1	100	100	Ehlers-Danlos syndrome, musculocontractural type 2, 615539
DSG1	99,3	97,5	100	100	Keratosis palmoplantaris striata I, AD, 148700 Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508
DSG2	100	99,6	100	100	Arrhythmogenic right ventricular dysplasia 10, 610193 Cardiomyopathy, dilated, 1BB, 612877
DSG3	99,9	99,4	100	100	No OMIM disease ID
DSG4	100	99,2	100	100	Hypotrichosis 6, 607903
DSP	100	99,6	100	100	Keratosis palmoplantaris striata II, 612908 Arrhythmogenic right ventricular dysplasia 8, 607450 Epidermolysis bullosa, lethal acantholytic, 609638 Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 Skin fragility-woolly hair syndrome, 607655 Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821
DSPP	96,8	86,1	100	100	Dentin dysplasia, type II, 125420 Deafness, autosomal dominant 39, with dentinogenesis, 605594 Dentinogenesis imperfecta, Shields type II, 125490 Dentinogenesis imperfecta, Shields type III, 125500
DST	95,5	95	95,6	95,6	?Neuropathy, hereditary sensory and autonomic, type VI, 614653 Epidermolysis bullosa simplex, autosomal recessive 2, 615425
DSTYK	99,9	99,2	100	100	Congenital anomalies of kidney and urinary tract 1, 610805 Spastic paraplegia 23, 270750

DTNA	100	99,9	100	100	Left ventricular noncompaction 1, with or without congenital heart defects, 604169
DTNBP1	99,8	98,7	100	100	Hermansky-Pudlak syndrome 7, 614076
DTYMK	100	99,8	100	100	No OMIM disease ID
DUOX2	96,7	94,7	100	100	Thyroid dyshormonogenesis 6, 607200
DUOXA2	100	100	100	100	Thyroid dyshormonogenesis 5, 274900
DUSP6	100	100	100	100	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
DVL1	97,2	95	100	100	Robinow syndrome, autosomal dominant 2, 616331
DVL3	100	100	100	100	Robinow syndrome, autosomal dominant 3, 616894
DYM	97,4	96,5	100	100	Smith-McCort dysplasia, 607326 Dyggve-Melchior-Clausen disease, 223800
DYNC1H1	99,9	99,4	100	100	Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 Charcot-Marie-Tooth disease, axonal, type 20, 614228
DYNC1I2	84,4	68,8	100	100	Neurodevelopmental disorder with microcephaly and structural brain anomalies, 618492
DYNC2H1	98,8	95,5	100	100	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
WDR60	99,5	97	100	100	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WDR34	100	99,6	100	100	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
DYNC2LI1	99,7	97,6	100	100	Short-rib thoracic dysplasia 15 with polydactyly, 617088
TCTEX1D2	100	100	100	100	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405
DYRK1A	100	100	100	100	Mental retardation, autosomal dominant 7, 614104
DYRK1B	98,4	94	100	100	Abdominal obesity-metabolic syndrome 3, 615812
DYSF	100	99,9	100	100	Miyoshi muscular dystrophy 1, 254130 Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601 Myopathy, distal, with anterior tibial onset, 606768
DZIP1	98,4	96,6	100	100	?Mitral valve prolapse 3, 610840 Spermatogenic failure 47, 619102
DZIP1L	99,9	99	100	100	Polycystic kidney disease 5, 617610
EARS2	99,8	97,7	100	100	Combined oxidative phosphorylation deficiency 12, 614924

EBF3	100	100	100	100	Hypotonia, ataxia, and delayed development syndrome, 617330
EBP	99,7	95,8	100	100	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
ECE1	88,6	88,3	90,2	90,2	{Hypertension, essential, susceptibility to}, 145500 ?Hirschsprung disease, cardiac defects, and autonomic dysfunction, 613870
ECEL1	95,4	90	100	100	Arthrogryposis, distal, type 5D, 615065
ECHS1	99,9	99	100	100	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ECM1	100	99,6	100	100	Urbach-Wiethe disease, 247100
ECSIT	100	99,9	100	100	No OMIM disease ID
EDA	98,1	91,6	100	99,9	Tooth agenesis, selective, X-linked 1, 313500 Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100
EDAR	100	99,9	100	100	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490 Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900 [Hair morphology 1, hair thickness], 612630
EDARADD	99,9	98,8	100	100	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941 Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940
EDC3	100	99,9	100	100	?Mental retardation, autosomal recessive 50, 616460
EDN1	100	100	100	100	Auriculocondylar syndrome 3, 615706 Question mark ears, isolated, 612798 {High density lipoprotein cholesterol level QTL 7}, 0
EDN3	98,8	98,8	100	100	Waardenburg syndrome, type 4B, 613265 Central hypoventilation syndrome, congenital, 209880 {Hirschsprung disease, susceptibility to, 4}, 613712
EDNRA	100	100	100	100	Mandibulofacial dysostosis with alopecia, 616367 {Migraine, resistance to}, 157300
EDNRB	98	93,8	100	100	{Hirschsprung disease, susceptibility to, 2}, 600155 Waardenburg syndrome, type 4A, 277580 ABCD syndrome, 600501
EED	96,5	91,4	99,8	98,2	Cohen-Gibson syndrome, 617561
EEF1A2	100	100	99,9	99,1	Mental retardation, autosomal dominant 38, 616393 Developmental and epileptic encephalopathy 33, 616409
EEF1AKNMT	99,9	99,3	100	100	?{Deafness, autosomal recessive 26, modifier of}, 605429
EEF2	100	99,9	100	100	?Spinocerebellar ataxia 26, 609306

EFEMP1	100	99,9	100	100	Doyne honeycomb degeneration of retina, 126600
EFEMP2	100	100	100	100	Cutis laxa, autosomal recessive, type IB, 614437
EFHC1	93,1	91,6	98	98	{Myoclonic epilepsy, juvenile, susceptibility to, 1}, 254770 {Epilepsy, juvenile absence, susceptibility to, 1}, 607631
EFL1	99,6	98,5	100	100	Shwachman-Diamond syndrome 2, 617941
EFNA4	100	100	100	100	No OMIM disease ID
EFNB1	100	100	100	100	Craniofrontonasal dysplasia, 304110
EFNB2	100	99,8	100	100	No OMIM disease ID
EFTUD2	100	99,8	100	100	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EGF	99,9	99,7	100	100	Hypomagnesemia 4, renal, 611718
EGFR	100	100	100	99,8	?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 {Nonsmall cell lung cancer, susceptibility to}, 211980
EGLN1	89,3	82,2	100	100	[Hemoglobin, high altitude adaptation], 609070 Erythrocytosis, familial, 3, 609820
EGLN2	100	99,8	100	100	No OMIM disease ID
EGR2	100	100	100	100	Dejerine-Sottas disease, 145900 Hypomyelinating neuropathy, congenital, 1, 605253 Charcot-Marie-Tooth disease, type 1D, 607678
EHHADH	100	100	100	100	?Fanconi renotubular syndrome 3, 615605
EHMT1	94,5	93,7	99,6	99,5	Kleefstra syndrome 1, 610253
EIF2AK1	98,8	95,4	100	100	?Leukoencephalopathy, motor delay, spasticity, and dysarthria syndrome, 618878
EIF2AK2	100	99,7	100	100	Leukoencephalopathy, developmental delay, and episodic neurologic regression syndrome, 618877
EIF2AK3	97,2	94,5	100	100	Wolcott-Rallison syndrome, 226980
EIF2AK4	99,8	98,6	100	100	Pulmonary venoocclusive disease 2, 234810
EIF2B1	100	100	100	100	Leukoencephalopathy with vanishing white matter, 603896
EIF2B2	99,9	99,5	100	100	Ovarioleukodystrophy, 603896 Leukoencephalopathy with vanishing white matter, 603896

EIF2B3	100	100	100	100	Leukoencephalopathy with vanishing white matter, 603896
EIF2B4	100	99,9	100	100	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B5	100	99,1	100	100	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2S3	95,4	89,1	100	100	MEHMO syndrome, 300148
EIF3F	96,8	84,1	100	100	Mental retardation, autosomal recessive 67, 618295
EIF4A3	100	99,5	100	100	Robin sequence with cleft mandible and limb anomalies, 268305
ELAC2	100	99,7	100	100	Combined oxidative phosphorylation deficiency 17, 615440 {Prostate cancer, hereditary, 2, susceptibility to}, 614731
ELANE	99,7	97,4	100	100	Neutropenia, severe congenital 1, autosomal dominant, 202700 Neutropenia, cyclic, 162800
ELF4	100	99,7	100	100	No OMIM disease ID
ELMO2	99,9	99	100	100	Vascular malformation, primary intraosseous, 606893
ELMOD3	100	99,8	100	100	?Deafness, autosomal recessive 88, 615429
ELN	99,8	97,8	100	100	Cutis laxa, autosomal dominant, 123700 Supravalvar aortic stenosis, 185500
ELOVL1	99,8	97,6	100	100	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527
ELOVL4	100	99,5	100	100	Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
ELOVL5	100	99,8	100	100	Spinocerebellar ataxia 38, 615957
ELP1	99,8	99	100	100	Dysautonomia, familial, 223900
ELP2	99,9	98,8	100	100	Mental retardation, autosomal recessive 58, 617270
ELP4	72,8	70,2	87,1	87,1	?Aniridia 2, 617141
EMC1	100	99,3	100	100	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EMD	99,9	98,4	100	99,1	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
EMG1	100	100	100	100	Bowen-Conradi syndrome, 211180
EMILIN1	99,3	89,8	100	100	No OMIM disease ID

EML1	99,7	98,4	100	100	Band heterotopia, 600348
EMP2	99,9	96,7	100	100	Nephrotic syndrome, type 10, 615861
EMX2	100	100	100	100	Schizencephaly, 269160
ENAM	100	100	100	100	Amelogenesis imperfecta, type IC, 204650 Amelogenesis imperfecta, type IB, 104500
ENG	99,6	96	100	100	Telangiectasia, hereditary hemorrhagic, type 1, 187300
ENO3	100	99,9	100	100	?Glycogen storage disease XIII, 612932
ENPP1	96,4	91,2	98,7	97,8	Hypophosphatemic rickets, autosomal recessive, 2, 613312 Cole disease, 615522 {Obesity, susceptibility to}, 601665 Arterial calcification, generalized, of infancy, 1, 208000 {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853
ENTPD1	100	100	100	100	Spastic paraplegia 64, autosomal recessive, 615683
EOGT	79,4	78,4	91,9	89	Adams-Oliver syndrome 4, 615297
EP300	99,8	99	100	100	Rubinstein-Taybi syndrome 2, 613684 Menke-Hennekam syndrome 2, 618333 Colorectal cancer, somatic, 114500
EPAS1	99,7	98,1	100	100	Erythrocytosis, familial, 4, 611783
EPB41	85,3	83,9	100	100	Elliptocytosis-1, 611804
EPB41L1	99,8	97,7	97,8	97,8	?Mental retardation, autosomal dominant 11, 614257
EPB42	100	99,5	100	100	Spherocytosis, type 5, 612690
EPCAM	98,6	90,3	99,8	98,3	Colorectal cancer, hereditary nonpolyposis, type 8, 613244 Diarrhea 5, with tufting enteropathy, congenital, 613217
EPG5	99,5	98,5	100	100	Vici syndrome, 242840
EPHA2	100	99,5	100	100	Cataract 6, multiple types, 116600
EPHB2	98,1	98,1	99,8	98,8	{Prostate cancer/brain cancer susceptibility, somatic}, 603688 ?Bleeding disorder, platelet-type, 22, 618462
EPHB4	100	99,7	100	100	Capillary malformation-arteriovenous malformation 2, 618196 Lymphatic malformation 7, 617300
EPHX1	99,9	98,8	100	100	?Hypercholanemia, familial, 607748

EPHX2	99,5	96,2	100	100	{Hypercholesterolemia, familial, due to LDLR defect, modifier of}, 143890
EPM2A	94,2	91,5	100	97,7	Epilepsy, progressive myoclonic 2A (Lafora), 254780
EPO	99,9	97,6	100	100	Erythrocytosis, familial, 5, 617907 {Microvascular complications of diabetes 2}, 612623 ?Diamond-Blackfan anemia-like, 617911
EPRS1	100	99,6	100	100	Leukodystrophy, hypomyelinating, 15, 617951
EPS8	97	96,2	100	100	?Deafness, autosomal recessive 102, 615974
EPS8L2	84,5	82,5	88	88	Deafness autosomal recessive 106, 617637
EPS8L3	98,9	97,3	100	100	?Hypotrichosis 5, 612841
ERAL1	100	99,7	100	100	Perrault syndrome 6, 617565
ERBB2	98,4	97,1	100	100	Glioblastoma, somatic, 137800 Adenocarcinoma of lung, somatic, 211980 Gastric cancer, somatic, 613659 Ovarian cancer, somatic, 0
ERBB3	100	99,8	100	100	{?Erythroleukemia, familial, susceptibility to}, 133180 ?Lethal congenital contractural syndrome 2, 607598
ERBB4	100	99,5	100	100	Amyotrophic lateral sclerosis 19, 615515
ERCC1	100	99,3	100	100	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	100	99,7	100	100	Trichothiodystrophy 1, photosensitive, 601675 ?Cerebrooculofacioskeletal syndrome 2, 610756 Xeroderma pigmentosum, group D, 278730
ERCC3	96,9	96,3	100	100	Xeroderma pigmentosum, group B, 610651 Trichothiodystrophy 2, photosensitive, 616390
ERCC4	100	99,9	100	100	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 Fanconi anemia, complementation group Q, 615272 XFE progeroid syndrome, 610965 Xeroderma pigmentosum, group F, 278760
ERCC5	100	99,7	100	100	Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 Xeroderma pigmentosum, group G, 278780 Cerebrooculofacioskeletal syndrome 3, 616570
ERCC6	100	100	100	100	{Macular degeneration, age-related, susceptibility to, 5}, 613761 {Lung cancer, susceptibility to}, 211980 Cerebrooculofacioskeletal syndrome 1, 214150

					Cockayne syndrome, type B, 133540 Premature ovarian failure 11, 616946 UV-sensitive syndrome 1, 600630 De Sanctis-Cacchione syndrome, 278800
ERCC6L2	99,9	99,2	100	100	Bone marrow failure syndrome 2, 615715
ERCC8	99,5	95,8	100	100	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621
ERF	99,9	98,5	100	100	Craniosynostosis 4, 600775 Chitayat syndrome, 617180
ERG	99,1	98,8	100	100	No OMIM disease ID
ERGIC1	95,2	94,6	98,4	98,4	?Arthrogryposis multiplex congenita 2, neurogenic type, 208100
ERLIN1	100	100	100	100	Spastic paraplegia 62, 615681
ERLIN2	100	99,9	100	100	Spastic paraplegia 18, autosomal recessive, 611225
ERMARD	99,9	99	100	100	?Periventricular nodular heterotopia 6, 615544
ESCO2	98,7	95,2	100	100	Roberts syndrome, 268300 SC phocomelia syndrome, 269000
ESPN	44,6	35,8	100	99,8	?Usher syndrome, type 1M, 618632 Deafness, autosomal recessive 36, 609006 Deafness, neurosensory, without vestibular involvement, autosomal dominant, 609006
ESR1	100	99,8	100	100	{Myocardial infarction, susceptibility to}, 608446 Estrogen resistance, 615363 Breast cancer, somatic, 114480 {Migraine, susceptibility to}, 157300
ESR2	100	99,7	100	100	?Ovarian dysgenesis 8, 618187
ESRP1	99,9	98,9	100	100	?Deafness, autosomal recessive 109, 618013
ESRRB	96,8	95	100	100	Deafness, autosomal recessive 35, 608565
ETFA	100	100	100	100	Glutaric acidemia IIA, 231680
ETFB	100	99,8	100	100	Glutaric acidemia IIB, 231680
ETFDH	100	99,8	100	100	Glutaric acidemia IIC, 231680
ETHE1	99,9	97,4	100	100	Ethylmalonic encephalopathy, 602473

ETV6	100	99,9	100	100	Leukemia, acute myeloid, somatic, 601626 Thrombocytopenia 5, 616216
EVC	93,9	88,6	96,9	94,8	Ellis-van Creveld syndrome, 225500 ?Weyers acrofacial dysostosis, 193530
EVC2	97,7	96,1	100	100	Weyers acrofacial dysostosis, 193530 Ellis-van Creveld syndrome, 225500
EWSR1	92,3	85,3	100	100	Neuroepithelioma, 612219 Ewing sarcoma, 612219
EXOC6	99,2	96,3	100	100	No OMIM disease ID
EXOC6B	99,1	97,6	100	100	Spondyloepimetaphyseal dysplasia with joint laxity, type 3, 618395
EXOC8	100	100	100	100	?Neurodevelopmental disorder with microcephaly, seizures, and brain atrophy, 619076
EXOSC2	100	100	100	100	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EXOSC3	99,5	94,9	100	100	Pontocerebellar hypoplasia, type 1B, 614678
EXOSC5	100	100	100	100	No OMIM disease ID
EXOSC8	97,9	91,2	100	100	Pontocerebellar hypoplasia, type 1C, 616081
EXOSC9	99,7	97,2	100	100	Pontocerebellar hypoplasia, type 1D, 618065
EXPH5	100	100	100	100	Epidermolysis bullosa, nonspecific, autosomal recessive, 615028
EXT1	99,9	98,4	100	100	Exostoses, multiple, type 1, 133700 Chondrosarcoma, 215300
EXT2	100	99,3	100	100	Seizures, scoliosis, and macrocephaly syndrome, 616682 Exostoses, multiple, type 2, 133701
EXTL3	100	100	100	100	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
EYA1	99,9	99,7	100	100	?Otofaciocervical syndrome, 166780 Anterior segment anomalies with or without cataract, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650 Branchiootic syndrome 1, 602588
EYA4	100	99,7	100	100	Deafness, autosomal dominant 10, 601316 ?Cardiomyopathy, dilated, 1J, 605362
EYS	99,7	98,2	100	100	Retinitis pigmentosa 25, 602772
EZH2	100	99,5	100	100	Weaver syndrome, 277590
F10	99,8	99,1	100	100	Factor X deficiency, 227600

F11	100	100	100	100	Factor XI deficiency, autosomal dominant, 612416 Factor XI deficiency, autosomal recessive, 612416
F12	99,9	98,8	100	100	Factor XII deficiency, 234000 Angioedema, hereditary, type III, 610618
F13A1	100	100	100	100	{Myocardial infarction, protection against}, 608446 Factor XIIIA deficiency, 613225 {Venous thrombosis, protection against}, 188050
F13B	98,7	93,5	100	100	Factor XIIIB deficiency, 613235
F2	99,9	97,9	100	100	{Pregnancy loss, recurrent, susceptibility to, 2}, 614390 Hypoprothrombinemia, 613679 Dysprothrombinemia, 613679 Thrombophilia due to thrombin defect, 188050 {Stroke, ischemic, susceptibility to}, 601367
F2RL3	100	100	100	100	No OMIM disease ID
F5	99,4	98,5	100	100	{Pregnancy loss, recurrent, susceptibility to, 1}, 614389 Thrombophilia due to activated protein C resistance, 188055 {Thrombophilia, susceptibility to, due to factor V Leiden}, 188055 Factor V deficiency, 227400 {Budd-Chiari syndrome}, 600880 {Stroke, ischemic, susceptibility to}, 601367
F7	100	100	100	100	{Myocardial infarction, decreased susceptibility to}, 608446 Factor VII deficiency, 227500
F8	97,3	96,1	100	99,9	Hemophilia A, 306700
F9	99,6	97,6	99,9	98,8	Thrombophilia, X-linked, due to factor IX defect, 300807 {Deep venous thrombosis, protection against}, 300807 Hemophilia B, 306900 {Warfarin sensitivity}, 301052
FA2H	92	83,1	100	100	Spastic paraplegia 35, autosomal recessive, 612319
FAAH	93,2	90	100	99,9	{Drug addiction, susceptibility to}, 606581
FAAP24	99,3	96,7	100	100	No OMIM disease ID
FADD	100	100	100	100	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations, 613759
FAH	100	100	100	100	Tyrosinemia, type I, 276700
FAM111A	99,9	99,3	100	100	Gracile bone dysplasia, 602361 Kenny-Caffey syndrome, type 2, 127000

FAM111B	100	99,9	100	100	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704
FAM126A	100	99,4	100	100	Leukodystrophy, hypomyelinating, 5, 610532
FAM149B1	99,5	95,4	100	100	Joubert syndrome 36, 618763
FAM161A	100	99,7	100	100	Retinitis pigmentosa 28, 606068
FAM20A	99,6	94,7	100	100	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM20B	100	99,9	100	100	No OMIM disease ID
FAM20C	100	100	100	99,8	Raine syndrome, 259775
FAM83G	100	100	100	100	No OMIM disease ID
FAM83H	84,9	81,5	100	100	Amelogenesis imperfecta, type IIIA, 130900
FAN1	100	99,8	100	100	Interstitial nephritis, karyomegalic, 614817
FANCA	100	99,4	100	100	Fanconi anemia, complementation group A, 227650
FANCB	98,6	94,1	100	100	Fanconi anemia, complementation group B, 300514
FANCC	97,2	96,6	97,3	97,3	Fanconi anemia, complementation group C, 227645
FANCD2	99,5	97,5	98,8	98,8	Fanconi anemia, complementation group D2, 227646
FANCE	89,8	85,1	100	99,9	Fanconi anemia, complementation group E, 600901
FANCF	100	100	100	100	Fanconi anemia, complementation group F, 603467
FANCG	100	99,9	100	100	Fanconi anemia, complementation group G, 614082
FANCI	99,9	99,2	100	100	Fanconi anemia, complementation group I, 609053
FANCL	100	98,6	100	100	Fanconi anemia, complementation group L, 614083
FANCM	99,6	97,3	100	100	Spermatogenic failure 28, 618086 ?Premature ovarian failure 15, 618096
FAR1	97,6	92,8	100	100	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154
FARS2	100	100	100	100	Spastic paraplegia 77, autosomal recessive, 617046 Combined oxidative phosphorylation deficiency 14, 614946
FARSB	98,8	94,6	100	100	Rajab interstitial lung disease with brain calcifications 1, 613658

FAS	100	99,6	100	100	Autoimmune lymphoproliferative syndrome, type IA, 601859 {Autoimmune lymphoproliferative syndrome}, 601859 Squamous cell carcinoma, burn scar-related, somatic, 0
FASLG	100	99,6	100	100	Autoimmune lymphoproliferative syndrome, type IB, 601859 {Lung cancer, susceptibility to}, 211980
FASTKD2	99,8	98,9	100	100	Combined oxidative phosphorylation deficiency 44, 618855
FAT1	100	100	100	100	No OMIM disease ID
FAT2	100	99,8	100	100	Spinocerebellar ataxia 45, 617769
FAT4	100	100	100	100	Van Maldergem syndrome 2, 615546 Hennekam lymphangiectasia-lymphedema syndrome 2, 616006
FBLN1	99,7	97,6	100	99,7	Synpolydactyly, 3/3'4, associated with metacarpal and metatarsal synostoses, 608180
FBLN5	91,8	91,8	91,8	91,8	Macular degeneration, age-related, 3, 608895 ?Cutis laxa, autosomal dominant 2, 614434 Neuropathy, hereditary, with or without age-related macular degeneration, 608895 Cutis laxa, autosomal recessive, type IA, 219100
FBN1	100	99,9	100	100	Marfan lipodystrophy syndrome, 616914 Marfan syndrome, 154700 MASS syndrome, 604308 Ectopia lentis, familial, 129600 Acromicric dysplasia, 102370 Weill-Marchesani syndrome 2, dominant, 608328 Geleophysic dysplasia 2, 614185 Stiff skin syndrome, 184900
FBN2	100	99,9	100	100	Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118
FBP1	93,7	93,3	93,7	93,7	Fructose-1,6-bisphosphatase deficiency, 229700
FBXL3	100	100	100	100	Intellectual developmental disorder with short stature, facial anomalies, and speech defects, 606220
FBXL4	100	100	100	100	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FBXO11	96,9	92,7	100	100	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities, 618089
FBXO31	96	93,1	100	99,9	?Mental retardation, autosomal recessive 45, 615979
FBXO32	100	100	100	100	No OMIM disease ID
FBXO38	99,9	99,3	100	100	Neuronopathy, distal hereditary motor, type IID, 615575

FBXO7	99,8	97,9	100	100	Parkinson disease 15, autosomal recessive, 260300
FBXW4	81,6	79	83,5	80	No OMIM disease ID
FBXW7	99,9	99,2	100	100	No OMIM disease ID
FCGR1A	46,8	44,1	100	100	[IgG receptor I, phagocytic, familial deficiency of], 0
FCGR2A	100	100	100	100	{Pseudomonas aeruginosa, susceptibility to chronic infection by, in cystic fibrosis}, 219700 {Lupus nephritis, susceptibility to}, 152700 {Malaria, severe, susceptibility to}, 611162
FCGR2B	99,5	95,4	100	100	{Malaria, resistance to}, 611162 {Systemic lupus erythematosus, susceptibility to}, 152700
FCGR2C	98,9	98,6	98,1	97,2	No OMIM disease ID
FCGR3A	99	97,1	100	100	Immunodeficiency 20, 615707
FCGR3B	99,3	97,9	98,1	98	No OMIM disease ID
FCHO1	98,9	97,7	100	100	No OMIM disease ID
FCN3	100	100	100	100	Immunodeficiency due to ficolin 3 deficiency, 613860
FCSK	97,7	95,4	100	100	Congenital disorder of glycosylation with defective fucosylation 2, 618324
FDCSP	98,7	93,6	100	100	No OMIM disease ID
FDFT1	97,7	96	100	100	Squalene synthase deficiency, 618156
FDPS	99,1	93,5	100	100	Porokeratosis 9, multiple types, 616631
FDX2	100	100	100	100	Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900
FDXR	100	99,3	100	100	Auditory neuropathy and optic atrophy, 617717
FECH	100	100	100	100	Protoporphyria, erythropoietic, 1, 177000
FERMT1	99,9	97,9	100	100	Kindler syndrome, 173650
FERMT3	100	100	100	100	Leukocyte adhesion deficiency, type III, 612840
FEZF1	100	99,9	100	100	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FGA	99,1	97,2	100	100	Dysfibrinogenemia, congenital, 616004 Amyloidosis, familial visceral, 105200 Hypodysfibrinogenemia, congenital, 616004 Afibrinogenemia, congenital, 202400

FGB	99,8	99,1	100	100	Dysfibrinogenemia, congenital, 616004 Afibrinogenemia, congenital, 202400 Hypofibrinogenemia, congenital, 202400
FGD1	97,3	92,8	100	100	Mental retardation, X-linked syndromic 16, 305400 Aarskog-Scott syndrome, 305400
FGD4	99,9	99,4	100	100	Charcot-Marie-Tooth disease, type 4H, 609311
FGF10	100	99,8	100	100	Aplasia of lacrimal and salivary glands, 180920 LADD syndrome, 149730
FGF12	99,9	98,1	100	100	Developmental and epileptic encephalopathy 47, 617166
FGF14	100	100	100	100	Spinocerebellar ataxia 27, 609307
FGF16	100	99,8	100	100	Metacarpal 4-5 fusion, 309630
FGF17	100	100	100	100	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
FGF20	97,5	87,6	100	100	?Renal hypodysplasia/aplasia 2, 615721
FGF23	99,6	97,5	100	100	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993 Hypophosphatemic rickets, autosomal dominant, 193100
FGF3	99,8	95,1	100	100	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGF5	100	100	100	100	Trichomegaly, 190330
FGF8	98,2	88,9	100	99,6	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGF9	100	100	100	100	Multiple synostoses syndrome 3, 612961
FGFR1	100	99,9	100	100	Pfeiffer syndrome, 101600 Jackson-Weiss syndrome, 123150 Trigonocephaly 1, 190440 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Hartsfield syndrome, 615465 Osteoglophonic dysplasia, 166250 Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001
FGFR2	97,7	97,1	100	100	Apert syndrome, 101200 Jackson-Weiss syndrome, 123150 Saethre-Chotzen syndrome, 101400 Gastric cancer, somatic, 613659 Scaphocephaly, maxillary retrusion, and mental retardation, 609579 Bent bone dysplasia syndrome, 614592 LADD syndrome, 149730

					Craniofacial-skeletal-dermatologic dysplasia, 101600 Pfeiffer syndrome, 101600 Crouzon syndrome, 123500 Beare-Stevenson cutis gyrata syndrome, 123790 Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Craniosynostosis, nonspecific, 0 Scaphocephaly and Axenfeld-Rieger anomaly, 0
FGFR3	99,8	97,7	100	99,8	Muenke syndrome, 602849 Nevus, epidermal, somatic, 162900 Thanatophoric dysplasia, type II, 187601 Bladder cancer, somatic, 109800 CATSHL syndrome, 610474 Crouzon syndrome with acanthosis nigricans, 612247 Hypochondroplasia, 146000 LADD syndrome, 149730 Achondroplasia, 100800 Thanatophoric dysplasia, type I, 187600 Colorectal cancer, somatic, 114500 Spermatocytic seminoma, somatic, 273300 Cervical cancer, somatic, 603956 SADDAN, 616482
FGG	99,7	98,2	100	100	Hypofibrinogenemia, congenital, 202400 Hypodysfibrinogenemia, 616004 Dysfibrinogenemia, congenital, 616004 Afibrinogenemia, congenital, 202400
FH	92,1	88,3	100	100	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FHL1	99,7	95,8	100	100	Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717 Scapuloperoneal myopathy, X-linked dominant, 300695 Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718 ?Uruguay faciocardiomusculoskeletal syndrome, 300280 Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 Myopathy, X-linked, with postural muscle atrophy, 300696
FHL2	99,9	98,7	100	100	No OMIM disease ID
FHOD3	100	99,6	100	100	No OMIM disease ID
FIBP	100	100	100	100	Thauvin-Robinet-Faivre syndrome, 617107

FIG4	100	99,8	100	100	Yunis-Varon syndrome, 216340 ?Polymicrogyria, bilateral temporooccipital, 612691 Charcot-Marie-Tooth disease, type 4J, 611228 Amyotrophic lateral sclerosis 11, 612577
FIGLA	99,7	96	100	100	Premature ovarian failure 6, 612310
FIGN	100	100	100	100	No OMIM disease ID
FITM2	100	100	100	100	Siddiqi syndrome, 618635
FKBP10	98,8	97,2	100	100	Bruck syndrome 1, 259450 Osteogenesis imperfecta, type XI, 610968
FKBP14	100	99,9	100	100	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557
FKRP	100	100	100	99,9	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153
FKTN	99,7	97	100	100	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800
FLAD1	100	99,8	100	100	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100
FLCN	100	100	100	100	Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700 Birt-Hogg-Dube syndrome, 135150 Colorectal cancer, somatic, 114500
FLG	100	99,9	100	100	{Dermatitis, atopic, susceptibility to, 2}, 605803 Ichthyosis vulgaris, 146700
FLG2	100	100	99,9	99,9	Peeling skin syndrome 6, 618084
FLI1	99,5	98,2	100	100	Bleeding disorder, platelet-type, 21, 617443
FLNA	100	99,9	100	100	Otopalatodigital syndrome, type I, 311300 Congenital short bowel syndrome, 300048 Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Cardiac valvular dysplasia, X-linked, 314400 ?FG syndrome 2, 300321 Heterotopia, periventricular, 1, 300049

					Terminal osseous dysplasia, 300244 Frontometaphyseal dysplasia 1, 305620
FLNB	99,5	98,8	100	100	Larsen syndrome, 150250 Atelosteogenesis, type I, 108720 Boomerang dysplasia, 112310 Spondylocarpotarsal synostosis syndrome, 272460 Atelosteogenesis, type III, 108721
FLNC	100	99,6	100	100	Cardiomyopathy, familial hypertrophic, 26, 617047 Myopathy, myofibrillar, 5, 609524 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065
FLRT3	100	100	100	100	Hypogonadotropic hypogonadism 21 with anosmia, 615271
FLT1	100	99,6	100	100	No OMIM disease ID
FLT3	99,9	98,9	100	100	Leukemia, acute myeloid, somatic, 601626 Leukemia, acute lymphoblastic, somatic, 613065 Leukemia, acute myeloid, reduced survival in, somatic, 601626
FLT4	99,2	98,3	100	100	Congenital heart defects, multiple types, 7, 618780 Hemangioma, capillary infantile, somatic, 602089 Lymphatic malformation 1, 153100
FLVCR1	100	98,9	100	100	Ataxia, posterior column, with retinitis pigmentosa, 609033
FLVCR2	100	100	100	100	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790
FMN1	97,3	96,3	100	100	No OMIM disease ID
FMN2	85,5	82,5	100	100	Mental retardation, autosomal recessive 47, 616193
FMO3	99,9	99,7	100	100	Trimethylaminuria, 602079
FMR1	96,1	92,1	100	100	Premature ovarian failure 1, 311360 Fragile X tremor/ataxia syndrome, 300623 Fragile X syndrome, 300624
FN1	100	99,3	100	100	Glomerulopathy with fibronectin deposits 2, 601894 Spondylometaphyseal dysplasia, corner fracture type, 184255
FNIP1	100	99,8	100	100	No OMIM disease ID
FOLR1	100	100	100	100	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXC1	98	89,6	99,9	98,5	Axenfeld-Rieger syndrome, type 3, 602482 Anterior segment dysgenesis 3, multiple subtypes, 601631

FOXC2	100	96,7	100	99,8	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXD4	26,3	13,3	100	100	No OMIM disease ID
FOXE1	96,9	78,5	99,9	99,1	Bamforth-Lazarus syndrome, 241850 {Thyroid cancer, nonmedullary, 4}, 616534
FOXE3	82,6	72	94,4	87,8	Cataract 34, multiple types, 612968 Anterior segment dysgenesis 2, multiple subtypes, 610256 {Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349
FOXF1	99,9	98,8	100	100	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380
FOXF2	93,6	86,6	96,4	94,8	No OMIM disease ID
FOXG1	88,6	82,1	99,2	96,4	Rett syndrome, congenital variant, 613454
FOXH1	100	96,5	100	100	No OMIM disease ID
FOXI1	100	100	100	100	Enlarged vestibular aqueduct, 600791
FOXJ1	99,9	98,4	100	100	Ciliary dyskinesia, primary, 43, 618699
FOXL1	96,6	89	100	100	No OMIM disease ID
FOXL2	99,7	95,5	99,8	98	Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 Premature ovarian failure 3, 608996 Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100
FOXN1	100	99,6	100	100	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806 T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXO1	99,5	93,9	99,2	96,9	Rhabdomyosarcoma, alveolar, 268220
FOXP1	100	99,8	100	100	Mental retardation with language impairment and with or without autistic features, 613670
FOXP2	99,5	99,2	100	100	Speech-language disorder-1, 602081
FOXP3	99,2	95,5	100	100	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790
FOXRED1	100	99,9	100	100	Mitochondrial complex I deficiency, nuclear type 19, 618241
FPR1	100	100	100	100	No OMIM disease ID
FRAS1	100	99,4	100	100	Fraser syndrome 1, 219000
FREM1	99,9	99,1	100	100	Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485 Bifid nose with or without anorectal and renal anomalies, 608980

FREM2	100	99,3	100	100	Fraser syndrome 2, 617666 Cryptophthalmos, unilateral or bilateral, isolated, 123570
FRMD4A	90,7	87,3	96,6	96,6	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819
FRMD7	99,9	99,1	100	99,6	Nystagmus 1, congenital, X-linked, 310700 Nystagmus, infantile periodic alternating, X-linked, 310700
FRMPD4	97,5	96,5	98,3	98,3	Mental retardation, X-linked 104, 300983
FRRS1L	79,7	69,1	99,2	95,8	Developmental and epileptic encephalopathy 37, 616981
FSCN2	100	100	100	100	Retinitis pigmentosa 30, 607921
FSHB	100	100	100	100	Hypogonadotropic hypogonadism 24 without anosmia, 229070
FSHR	99,5	97,2	100	100	Ovarian hyperstimulation syndrome, 608115 Ovarian dysgenesis 1, 233300 Ovarian response to FSH stimulation, 276400
FSIP2	99,6	98,2	100	100	Spermatogenic failure 34, 618153
FTCD	95,7	91	100	100	Glutamate formiminotransferase deficiency, 229100
FTH1	94	76,6	100	100	?Hemochromatosis, type 5, 615517
FTL	98,5	89,4	100	100	Hyperferritinemia-cataract syndrome, 600886 Neurodegeneration with brain iron accumulation 3, 606159 L-ferritin deficiency, dominant and recessive, 615604
FTO	83,8	83,7	94,2	94,2	Growth retardation, developmental delay, facial dysmorphism, 612938 {Obesity, susceptibility to, BMIQ14}, 612460
FTSJ1	98	93,8	100	100	Mental retardation, X-linked 9/44, 309549
FUCA1	100	99,9	100	100	Fucosidosis, 230000
FUS	99,2	96,4	100	100	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 Essential tremor, hereditary, 4, 614782
FUT2	100	100	100	100	{Vitamin B12 plasma level QTL1}, 612542 [Bombay phenotype, digenic], 616754 {Norwalk virus infection, resistance to}, 0
FUT6	100	100	100	100	[Fucosyltransferase 6 deficiency], 613852
FUT8	100	99,2	100	100	Congenital disorder of glycosylation with defective fucosylation 1, 618005
FUZ	100	100	100	100	{Neural tube defects, susceptibility to}, 182940

FXN	95,5	80,1	100	100	Friedreich ataxia with retained reflexes, 229300 Friedreich ataxia, 229300
FXYD2	100	100	100	100	Hypomagnesemia 2, renal, 154020
FYB1	99,4	97	100	100	Thrombocytopenia 3, 273900
FYCO1	100	99,9	100	100	Cataract 18, autosomal recessive, 610019
FZD1	89,6	85,3	100	100	No OMIM disease ID
FZD2	99,9	98,2	100	100	Omodysplasia 2, 164745
FZD4	100	100	100	100	Exudative vitreoretinopathy 1, 133780 Retinopathy of prematurity, 133780
FZD6	100	100	100	100	Nail disorder, nonsyndromic congenital, 1, 161050
G6PC	100	100	100	100	Glycogen storage disease Ia, 232200
G6PC3	100	99,9	100	100	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
G6PD	99,3	98	100	100	Hemolytic anemia, G6PD deficient (favism), 300908 {Resistance to malaria due to G6PD deficiency}, 611162
GAA	100	99,9	100	100	Glycogen storage disease II, 232300
GAB1	100	99,4	100	100	?Deafness, autosomal recessive 26, 605428
GABBR2	96,2	92	99,1	98,4	Neurodevelopmental disorder with poor language and loss of hand skills, 617903 {Nicotine dependence, susceptibility to}, 188890 {Nicotine dependence, protection against}, 188890 Developmental and epileptic encephalopathy 59, 617904
GABRA1	100	100	100	100	{Epilepsy, childhood absence, susceptibility to, 4}, 611136 Developmental and epileptic encephalopathy 19, 615744 {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136
GABRA2	99,7	98,6	100	100	{Alcohol dependence, susceptibility to}, 103780 Developmental and epileptic encephalopathy 78, 618557
GABRA3	98,7	94,4	99,9	98,9	No OMIM disease ID
GABRA5	100	99,9	100	100	Developmental and epileptic encephalopathy 79, 618559
GABRB1	100	100	100	100	Developmental and epileptic encephalopathy 45, 617153
GABRB2	100	99,9	100	100	Epileptic encephalopathy, infantile or early childhood, 2, 617829

GABRB3	99,6	98,2	100	100	{Epilepsy, childhood absence, susceptibility to, 5}, 612269 Developmental and epileptic encephalopathy 43, 617113
GABRG2	90,8	90,2	93	93	Febrile seizures, familial, 8, 607681 Developmental and epileptic encephalopathy 74, 618396 Epilepsy, generalized, with febrile seizures plus, type 3, 607681
GAD1	100	99,9	100	100	?Cerebral palsy, spastic quadriplegic, 1, 603513 Developmental and epileptic encephalopathy 89, 619124
GAL	100	99,8	100	100	?Epilepsy, familial temporal lobe, 8, 616461
GALC	99,8	98,3	100	100	Krabbe disease, 245200
GALE	100	100	100	100	Galactose epimerase deficiency, 230350
GALK1	100	99,1	100	100	Galactokinase deficiency with cataracts, 230200
GALM	100	99,9	100	100	Galactosemia IV, 618881
GALNS	100	99,8	100	100	Mucopolysaccharidosis IVA, 253000
GALNT12	85,8	82,7	97,8	94,6	{Colorectal cancer, susceptibility to, 1}, 608812
GALNT2	99,6	97	100	100	Congenital disorder of glycosylation, type IIt, 618885
GALNT3	99,8	99	100	100	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GALT	100	99,7	100	100	Galactosemia, 230400
GAMT	93,1	82,7	100	100	Cerebral creatine deficiency syndrome 2, 612736
GAN	100	99,6	100	100	Giant axonal neuropathy-1, 256850
GANAB	99,9	99	100	100	Polycystic kidney disease 3, 600666
GAPVD1	100	99,3	100	100	No OMIM disease ID
GARS1	99,9	99,1	100	100	Charcot-Marie-Tooth disease, type 2D, 601472 Spinal muscular atrophy, infantile, James type, 619042 Neuronopathy, distal hereditary motor, type VA, 600794
GAS2	100	100	100	100	No OMIM disease ID
GAS2L2	100	100	100	100	?Ciliary dyskinesia, primary, 41, 618449
GAS8	99,9	99,3	100	100	Ciliary dyskinesia, primary, 33, 616726
GATA1	99,8	98,4	100	100	Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835

					Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367 Thrombocytopenia with beta-thalassemia, X-linked, 314050
GATA2	100	98,3	100	100	Emberger syndrome, 614038 {Myelodysplastic syndrome, susceptibility to}, 614286 Immunodeficiency 21, 614172 {Leukemia, acute myeloid, susceptibility to}, 601626
GATA3	100	100	100	100	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GATA4	84,1	74,5	100	99,9	?Testicular anomalies with or without congenital heart disease, 615542 Tetralogy of Fallot, 187500 Atrioventricular septal defect 4, 614430 Atrial septal defect 2, 607941 Ventricular septal defect 1, 614429
GATA5	99,7	93,7	100	100	Congenital heart defects, multiple types, 5, 617912
GATA6	89,8	83	99,6	98	Pancreatic agenesis and congenital heart defects, 600001 Atrial septal defect 9, 614475 Atrioventricular septal defect 5, 614474 Persistent truncus arteriosus, 217095 Tetralogy of Fallot, 187500
GATAD1	99,9	97,9	100	99,1	?Cardiomyopathy, dilated, 2B, 614672
GATAD2B	100	100	100	100	GAND syndrome, 615074
GATB	100	99,7	100	100	?Combined oxidative phosphorylation deficiency 41, 618838
GATC	100	100	100	100	Combined oxidative phosphorylation deficiency 42, 618839
GATM	100	100	100	100	Cerebral creatine deficiency syndrome 3, 612718 Fanconi renotubular syndrome 1, 134600
GBA	100	100	100	100	Gaucher disease, type III, 231000 {Parkinson disease, late-onset, susceptibility to}, 168600 Gaucher disease, type IIIC, 231005 Gaucher disease, type I, 230800 Gaucher disease, perinatal lethal, 608013 Gaucher disease, type II, 230900 {Lewy body dementia, susceptibility to}, 127750
GBA2	100	99,7	100	100	Spastic paraplegia 46, autosomal recessive, 614409
GBE1	100	99,6	100	100	Polyglucosan body disease, adult form, 263570 Glycogen storage disease IV, 232500

GBF1	98,3	98	100	100	No OMIM disease ID
GCDH	100	99,2	100	100	Glutaricaciduria, type I, 231670
GCH1	99,9	95,5	100	100	Hyperphenylalaninemia, BH4-deficient, B, 233910 Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230
GCK	95,4	95,4	92,5	92,2	Diabetes mellitus, noninsulin-dependent, late onset, 125853 MODY, type II, 125851 Diabetes mellitus, permanent neonatal 1, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485
GCLC	99,8	98	100	100	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 {Myocardial infarction, susceptibility to}, 608446
GCLM	99,6	95,8	100	100	{Myocardial infarction, susceptibility to}, 608446
GCM2	100	100	100	100	Hyperparathyroidism 4, 617343 Hypoparathyroidism, familial isolated 2, 618883
GCNT2	99,5	99,5	100	100	Adult i phenotype without cataract, 110800 Cataract 13 with adult i phenotype, 116700 [Blood group, Ii], 110800
GCSH	75,7	68,9	100	100	?Glycine encephalopathy, 605899
GDAP1	99,8	99,3	100	100	Charcot-Marie-Tooth disease, recessive intermediate, A, 608340 Charcot-Marie-Tooth disease, type 4A, 214400 Charcot-Marie-Tooth disease, axonal, type 2K, 607831 Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706
GDAP2	100	99,2	100	100	Spinocerebellar ataxia, autosomal recessive 27, 618369
GDF1	73,9	54	98,7	92	Right atrial isomerism (Ivemark), 208530 Congenital heart defects, multiple types, 6, 613854
GDF2	100	100	100	100	Telangiectasia, hereditary hemorrhagic, type 5, 615506
GDF3	100	100	100	100	Microphthalmia, isolated 7, 613704 Microphthalmia with coloboma 6, 613703 Klippel-Feil syndrome 3, autosomal dominant, 613702
GDF5	100	100	100	100	?Acromesomelic dysplasia, Hunter-Thompson type, 201250 Symphalangism, proximal, 1B, 615298 Brachydactyly, type A1, C, 615072 Chondrodysplasia, Grebe type, 200700 Brachydactyly, type A2, 112600 Du Pan syndrome, 228900 {Osteoarthritis-5}, 612400

					Brachydactyly, type C, 113100 Multiple synostoses syndrome 2, 610017
GDF6	100	99,9	100	99,4	Leber congenital amaurosis 17, 615360 Klippel-Feil syndrome 1, autosomal dominant, 118100 Multiple synostoses syndrome 4, 617898 Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094
GDF9	100	100	100	100	?Premature ovarian failure 14, 618014
GDI1	99,8	98,7	100	100	Mental retardation, X-linked 41, 300849
GDNF	100	100	100	100	{Pheochromocytoma, modifier of}, 171300 {Hirschsprung disease, susceptibility to, 3}, 613711 Central hypoventilation syndrome, 209880
GEMIN4	100	99,5	100	100	Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities, 617913
GFAP	91,8	89,7	100	100	Alexander disease, 203450
GFER	99,6	93,9	100	100	Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076
GFI1	100	99,2	100	100	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 Neutropenia, severe congenital 2, autosomal dominant, 613107
GFI1B	98,5	96,7	100	100	Bleeding disorder, platelet-type, 17, 187900
GFM1	99,9	99,4	100	100	Combined oxidative phosphorylation deficiency 1, 609060
GFM2	98,9	95,2	100	100	Combined oxidative phosphorylation deficiency 39, 618397
GFPT1	100	99,4	100	100	Myasthenia, congenital, 12, with tubular aggregates, 610542
GFRA1	100	100	100	100	No OMIM disease ID
GGCX	100	99,9	100	100	Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450 Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842
GGT1	19,9	18,4	100	100	?Glutathioninuria, 231950
GH1	100	100	100	100	Kowarski syndrome, 262650 Growth hormone deficiency, isolated, type IA, 262400 Growth hormone deficiency, isolated, type IB, 612781 Growth hormone deficiency, isolated, type II, 173100
GHR	99,6	99,5	99,8	99,8	{Hypercholesterolemia, familial, modifier of}, 143890 Increased responsiveness to growth hormone, 604271

					Laron dwarfism, 262500 Growth hormone insensitivity, partial, 604271
GHRHR	96,4	96,1	100	100	Growth hormone deficiency, isolated, type IV, 618157
GHSR	98,5	95,8	100	100	Growth hormone deficiency, isolated partial, 615925
GINS1	99,3	94,9	100	100	Immunodeficiency 55, 617827
GIPC3	24,8	23	99,9	99,6	Deafness, autosomal recessive 15, 601869
GJA1	100	100	100	100	Erythrokeratodermia variabilis et progressiva 3, 617525 Craniometaphyseal dysplasia, autosomal recessive, 218400 Atrioventricular septal defect 3, 600309 Oculodentodigital dysplasia, 164200 Syndactyly, type III, 186100 Oculodentodigital dysplasia, autosomal recessive, 257850 Hypoplastic left heart syndrome 1, 241550 Palmoplantar keratoderma with congenital alopecia, 104100
GJA3	100	99,7	100	100	Cataract 14, multiple types, 601885
GJA5	100	100	100	100	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic (GJA5/SCN5A), 108770
GJA8	100	100	100	100	Cataract 1, multiple types, 116200
GJB1	100	100	100	100	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GJB2	100	100	100	100	Deafness, autosomal dominant 3A, 601544 Deafness, autosomal recessive 1A, 220290 Bart-Pumphrey syndrome, 149200 Vohwinkel syndrome, 124500 Keratoderma, palmoplantar, with deafness, 148350 Keratitis-ichthyosis-deafness syndrome, 148210 Hystrix-like ichthyosis with deafness, 602540
GJB3	100	100	100	100	Deafness, autosomal dominant 2B, 612644 Deafness, digenic, GJB2/GJB3, 220290 Erythrokeratodermia variabilis et progressiva 1, 133200 Deafness, autosomal recessive, 0 Deafness, autosomal dominant, with peripheral neuropathy, 0
GJB4	100	100	100	100	Erythrokeratodermia variabilis et progressiva 2, 617524
GJB6	100	100	100	100	Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645

					Deafness, digenic GJB2/GJB6, 220290 Ectodermal dysplasia 2, Clouston type, 129500
GJC2	78,2	58,7	96,9	91,4	Spastic paraplegia 44, autosomal recessive, 613206 Lymphatic malformation 3, 613480 Leukodystrophy, hypomyelinating, 2, 608804
GK	88,9	70,4	100	99,9	Glycerol kinase deficiency, 307030
GLA	91,1	88,2	91,3	91,3	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	99,9	97,4	100	100	GM1-gangliosidosis, type III, 230650 GM1-gangliosidosis, type I, 230500 Mucopolysaccharidosis type IVB (Morquio), 253010 GM1-gangliosidosis, type II, 230600
GLDC	89,9	82	100	99,9	Glycine encephalopathy, 605899
GLDN	94,6	91	100	100	Lethal congenital contracture syndrome 11, 617194
GLE1	100	100	100	100	Congenital arthrogryposis with anterior horn cell disease, 611890 Lethal congenital contracture syndrome 1, 253310
GLI1	100	99,8	100	100	Polydactyly, postaxial, type A8, 618123 Polydactyly, preaxial I, 174400
GLI2	99,1	97,4	100	99,8	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829
GLI3	98,5	98	100	100	Polydactyly, postaxial, types A1 and B, 174200 Greig cephalopolysyndactyly syndrome, 175700 Polydactyly, preaxial, type IV, 174700 Pallister-Hall syndrome, 146510
GLIS2	100	99,8	100	100	Nephronophthisis 7, 611498
GLIS3	98,6	98,2	100	100	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GLMN	99,3	94,7	100	100	Glomuvenous malformations, 138000
GLRA1	100	99,8	100	100	Hyperekplexia 1, 149400
GLRB	99,2	95,1	100	100	Hyperekplexia 2, 614619
GLRX5	97,3	89,1	99,6	95,4	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GLS	96,3	87,2	100	99,9	?Infantile cataract, skin abnormalities, glutamate excess, and impaired intellectual development, 618339 Global developmental delay, progressive ataxia, and elevated glutamine, 618412 Developmental and epileptic encephalopathy 71, 618328

GLUD1	94,2	82,9	100	100	Hyperinsulinism-hyperammonemia syndrome, 606762
GLUL	74,4	73,2	100	100	Glutamine deficiency, congenital, 610015
GLYCTK	98,8	97,3	100	100	D-glyceric aciduria, 220120
GM2A	100	100	100	100	GM2-gangliosidosis, AB variant, 272750
GMNN	99,8	97,4	100	100	Meier-Gorlin syndrome 6, 616835
GMPPA	100	100	100	100	Alacrima, achalasia, and mental retardation syndrome, 615510
GMPPB	100	100	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352
GMPR	100	100	100	100	No OMIM disease ID
GMPS	99,1	96,1	100	100	No OMIM disease ID
GNA11	99,9	95	100	100	Hypocalciuric hypercalcemia, type II, 145981 Hypocalcemia, autosomal dominant 2, 615361
GNA14	100	100	100	100	No OMIM disease ID
GNAI1	99	93,9	100	100	No OMIM disease ID
GNAI2	100	100	100	100	Ventricular tachycardia, idiopathic, 192605 Pituitary adenoma, ACTH-secreting, somatic, 0
GNAI3	99,3	95,2	100	100	Auriculocondylar syndrome 1, 602483
GNAL	96,8	93,4	100	100	Dystonia 25, 615073
GNAO1	93,8	93,8	100	100	Developmental and epileptic encephalopathy 17, 615473 Neurodevelopmental disorder with involuntary movements, 617493
GNAQ	84,3	74,9	100	100	Sturge-Weber syndrome, somatic, mosaic, 185300 Capillary malformations, congenital, 1, somatic, mosaic, 163000
GNAS	86,9	85,1	82	81,7	ACTH-independent macronodular adrenal hyperplasia, 219080 Pseudohypoparathyroidism Ic, 612462 Pseudohypoparathyroidism Ib, 603233 Pseudopseudohypoparathyroidism, 612463 McCune-Albright syndrome, somatic, mosaic, 174800 Osseous heteroplasia, progressive, 166350 Pituitary adenoma 3, multiple types, somatic, 617686 Pseudohypoparathyroidism Ia, 103580

GNAS-AS1					Pseudohypoparathyroidism, type IB, 603233
GNAT1	100	100	100	100	Night blindness, congenital stationary, type 1G, 616389 Night blindness, congenital stationary, autosomal dominant 3, 610444
GNAT2	99,9	99	100	100	Achromatopsia 4, 613856
GNB1	100	100	100	100	Myelodysplastic syndrome, somatic, 614286 Mental retardation, autosomal dominant 42, 616973 Leukemia, acute lymphoblastic, somatic, 613065
GNB2	100	100	100	100	No OMIM disease ID
GNB3	100	100	100	100	{Hypertension, essential, susceptibility to}, 145500 Night blindness, congenital stationary, type 1H, 617024
GNB4	100	100	100	100	Charcot-Marie-Tooth disease, dominant intermediate F, 615185
GNB5	100	98,8	100	100	Intellectual developmental disorder with cardiac arrhythmia, 617173 Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182
GNE	100	99,7	100	100	Sialuria, 269921 Nonaka myopathy, 605820
GNMT	100	100	100	100	Glycine N-methyltransferase deficiency, 606664
GNPAT	99,7	97,3	100	100	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPNAT1	68,8	48,4	100	100	No OMIM disease ID
GNPTAB	100	99,9	100	100	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GNPTG	99,1	94,3	100	99,9	Mucolipidosis III gamma, 252605
GNRH1	100	93,7	100	100	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841
GNRHR	100	100	100	100	Hypogonadotropic hypogonadism 7 without anosmia, 146110
GNS	98,4	94,8	100	100	Mucopolysaccharidosis type IIID, 252940
GORAB	100	99,1	100	100	Geroderma osteodysplasticum, 231070
GOSR2	95,9	94,6	100	100	Epilepsy, progressive myoclonic 6, 614018
GOT1	100	99,3	100	100	Aspartate aminotransferase, serum level of, QTL1, 614419
GOT2	97,5	90,9	100	100	Epileptic encephalopathy, early infantile, 82, 618721
GP1BA	98,6	95,9	100	100	Bernard-Soulier syndrome, type A1 (recessive), 231200 {Nonarteritic anterior ischemic optic neuropathy, susceptibility to}, 258660

					von Willebrand disease, platelet-type, 177820 Bernard-Soulier syndrome, type A2 (dominant), 153670
GP1BB	72,9	59,6	99,5	95	Giant platelet disorder, isolated, 231200 Bernard-Soulier syndrome, type B, 231200
GP6	100	100	94,9	91,7	Bleeding disorder, platelet-type, 11, 614201
GP9	96,5	89,3	100	100	Bernard-Soulier syndrome, type C, 231200
GPAA1	98,9	95,9	100	100	Glycosylphosphatidylinositol biosynthesis defect 15, 617810
GPC3	99,1	94,7	100	100	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPC4	100	99,6	100	100	Keipert syndrome, 301026
GPC6	100	100	100	100	Omodysplasia 1, 258315
GPD1	100	100	100	100	Hypertriglyceridemia, transient infantile, 614480
GPD1L	100	99,8	100	100	Brugada syndrome 2, 611777
GPHN	100	99,5	100	100	Molybdenum cofactor deficiency C, 615501
GPI	100	100	100	100	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470
GPIHBP1	100	100	100	100	Hyperlipoproteinemia, type 1D, 615947
GPNMB	95,5	95,5	95,5	95,5	Amyloidosis, primary localized cutaneous, 3, 617920
GPR101	100	100	100	100	Pituitary adenoma 2, GH-secreting, 300943
GPR143	85,8	76,4	99,8	97,9	Nystagmus 6, congenital, X-linked, 300814 Ocular albinism, type I, Nettleship-Falls type, 300500
GPR161	100	100	100	100	No OMIM disease ID
GPR179	100	100	100	100	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
GPR68	99,5	96,7	100	100	Amelogenesis imperfecta, hypomaturation type, IIA6, 617217
GPR88	99,4	95,1	98,8	94,9	?Chorea, childhood-onset, with psychomotor retardation, 616939
GPRASP2	100	99,7	100	100	?Deafness, X-linked 7, 301018
GPSM2	99,9	99,2	100	100	Chudley-McCullough syndrome, 604213
GPT2	99,2	93,6	100	99,8	Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281

GPX1	95,9	86,4	100	100	Hemolytic anemia due to glutathione peroxidase deficiency, 614164
GPX4	90,5	85,8	98,2	94,9	Spondylometaphyseal dysplasia, Sedaghatian type, 250220
GRAP	82,8	78,3	100	100	Deafness, autosomal recessive 114, 618456
GREB1L	100	99,9	100	100	Renal hypodysplasia/aplasia 3, 617805
GREM1	100	100	100	100	No OMIM disease ID
GREM2	100	100	100	100	Tooth agenesis, selective, 9, 617275
GRHL2	100	100	100	100	Deafness, autosomal dominant 28, 608641 Corneal dystrophy, posterior polymorphous, 4, 618031 Ectodermal dysplasia/short stature syndrome, 616029
GRHL3	100	100	100	100	Van der Woude syndrome 2, 606713
GRHPR	84,2	81,3	100	99,3	Hyperoxaluria, primary, type II, 260000
GRIA3	99,7	96,1	100	99,6	Intellectual developmental disorder, X-linked, syndromic, Wu type, 300699
GRIA4	99,8	99	100	100	Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864
GRID2	100	99,8	100	100	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRIK2	96,2	95,4	96,3	96,3	Mental retardation, autosomal recessive, 6, 611092
GRIN1	100	100	100	100	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820 Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254
GRIN2A	100	100	100	100	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570
GRIN2B	99,8	99,2	100	100	Mental retardation, autosomal dominant 6, 613970 Developmental and epileptic encephalopathy 27, 616139
GRIN2D	79,8	65,4	93,9	88,7	Developmental and epileptic encephalopathy 46, 617162
GRIP1	100	99,7	100	100	Fraser syndrome 3, 617667
GRK1	100	100	100	100	Oguchi disease-2, 613411
GRM1	100	99,7	100	100	Spinocerebellar ataxia 44, 617691 Spinocerebellar ataxia, autosomal recessive 13, 614831
GRM6	90,2	80,6	98,3	96,3	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270

GRN	100	100	100	100	Ceroid lipofuscinosis, neuronal, 11, 614706 Aphasia, primary progressive, 607485 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
GRXCR1	100	99,8	100	100	Deafness, autosomal recessive 25, 613285
GRXCR2	100	100	100	100	?Deafness, autosomal recessive 101, 615837
GSC	99,2	92,4	100	100	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471
GSDME	100	99,2	100	100	Deafness, autosomal dominant 5, 600994
GSE1	99,8	97,7	100	100	No OMIM disease ID
GSN	95,8	93,5	99,9	99,3	Amyloidosis, Finnish type, 105120
GSS	96,5	96,4	100	100	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900
GSX2	100	100	100	100	Diencephalic-mesencephalic junction dysplasia syndrome 2, 618646
GTF2E2	100	99,8	100	100	Trichothiodystrophy 6, nonphotosensitive, 616943
GTF2H5	72,5	72,2	72,5	72,5	Trichothiodystrophy 3, photosensitive, 616395
GTPBP2	100	99,3	100	99,9	Jaberi-Elahi syndrome, 617988
GTPBP3	100	99,8	100	100	Combined oxidative phosphorylation deficiency 23, 616198
GUCA1A	100	100	100	100	Cone-rod dystrophy 14, 602093 Cone dystrophy-3, 602093
GUCA1B	100	100	100	100	Retinitis pigmentosa 48, 613827
GUCY1A1	100	99,8	100	100	Moyamoya 6 with achalasia, 615750
GUCY2C	100	99,6	100	100	Diarrhea 6, 614616 Meconium ileus, 614665
GUCY2D	99,6	96,2	100	100	Cone-rod dystrophy 6, 601777 Leber congenital amaurosis 1, 204000 Night blindness, congenital stationary, type 1I, 618555 ?Choroidal dystrophy, central areolar 1, 215500
GUF1	99,7	97,8	100	100	?Developmental and epileptic encephalopathy 40, 617065
GUSB	92,9	91,7	100	100	Mucopolysaccharidosis VII, 253220
GYG1	99,9	99,2	100	100	?Glycogen storage disease XV, 613507 Polyglucosan body myopathy 2, 616199

GYS1	100	98,6	100	100	Glycogen storage disease 0, muscle, 611556
GYS2	99,8	99	100	100	Glycogen storage disease 0, liver, 240600
GZF1	100	99,6	100	100	Joint laxity, short stature, and myopia, 617662
H1-4	100	100	100	100	Rahman syndrome, 617537
H19					No OMIM disease ID
H4C3	100	100	100	100	No OMIM disease ID
H6PD	99	99	100	100	Cortisone reductase deficiency 1, 604931
HAAO	100	99,8	100	100	Vertebral, cardiac, renal, and limb defects syndrome 1, 617660
HABP2	100	99,9	100	100	{?Thyroid cancer, nonmedullary, 5}, 616535 {Venous thromboembolism, susceptibility to}, 188050
HACE1	100	99,3	100	100	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADH	99	97,5	100	100	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HADHA	97,2	91,6	100	100	LCHAD deficiency, 609016 HELLP syndrome, maternal, of pregnancy, 609016 Mitochondrial trifunctional protein deficiency, 609015 Fatty liver, acute, of pregnancy, 609016
HADHB	98,8	89,7	100	100	Trifunctional protein deficiency, 609015
HAGH	100	99,7	98,7	96,1	[Glyoxalase II deficiency], 614033
HAMP	100	100	100	100	Hemochromatosis, type 2B, 613313
HAND1	100	100	100	100	No OMIM disease ID
HAND2	99,8	92,6	100	100	No OMIM disease ID
HARS1	100	100	100	100	Usher syndrome type 3B, 614504 Charcot-Marie-Tooth disease, axonal, type 2W, 616625
HARS2	100	100	100	100	Perrault syndrome 2, 614926
HAVCR2	100	100	100	100	T-cell lymphoma, subcutaneous panniculitis-like, 618398
HAX1	100	100	100	100	Neutropenia, severe congenital 3, autosomal recessive, 610738
HBA1	100	99,8	100	100	Hemoglobin H disease, nondeletional, 613978 Thalassemias, alpha-, 604131

					Erythrocytosis 7, 617981 Methemoglobinemia, alpha type, 617973 Heinz body anemias, alpha-, 140700
HBA2	98,9	93,1	100	100	Thalassemia, alpha-, 604131 Erythrocytosis 7, 617981 Heinz body anemia, 140700 Hemoglobin H disease, deletional and nondeletional, 613978
HBB	100	100	100	100	Thalassemia, beta, 613985 Erythrocytosis 6, 617980 Heinz body anemia, 140700 Delta-beta thalassemia, 141749 Thalassemia-beta, dominant inclusion-body, 603902 Hereditary persistence of fetal hemoglobin, 141749 Methemoglobinemia, beta type, 617971 {Malaria, resistance to}, 611162 Sickle cell anemia, 603903
HBD	100	100	100	100	Thalassemia, delta-, 0 Thalassemia due to Hb Lepore, 0
HBG1	97,7	94,9	98,4	97	Fetal hemoglobin quantitative trait locus 1, 141749
HBG2	100	100	100	100	Fetal hemoglobin quantitative trait locus 1, 141749 Cyanosis, transient neonatal, 613977
HCCS	99,8	97,6	100	100	Linear skin defects with multiple congenital anomalies 1, 309801
HCFC1	98,3	93,6	100	100	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541
HCN1	98,5	98,2	98,5	98,5	Generalized epilepsy with febrile seizures plus, type 10, 618482 Developmental and epileptic encephalopathy 24, 615871
HCN2	59,2	49,5	84,1	77,3	No OMIM disease ID
HCN3	99,9	98,5	100	100	No OMIM disease ID
HCN4	100	99,3	100	99,9	Brugada syndrome 8, 613123 Sick sinus syndrome 2, 163800
HCRT	89,8	81,1	100	99,9	?Narcolepsy 1, 161400
HDAC4	100	99,8	100	100	No OMIM disease ID
HDAC6	99,5	97,4	100	100	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863
HDAC8	86,5	85,1	96,3	94,8	Cornelia de Lange syndrome 5, 300882

HECW2	100	99,1	100	100	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268
HELLS	97,8	92,1	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911
HEPACAM	86	78,9	100	100	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926
HEPH	98,8	91,9	100	100	No OMIM disease ID
HEPHL1	100	99,9	100	100	?Abnormal hair, joint laxity, and developmental delay, 261990
HERC1	100	100	100	100	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
HERC2	79,9	77,2	100	100	[Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 Mental retardation, autosomal recessive 38, 615516 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220
HES7	84,4	53,9	100	100	Spondylocostal dysostosis 4, autosomal recessive, 613686
HESX1	99,7	97,3	100	100	Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230 Growth hormone deficiency with pituitary anomalies, 182230
HEXA	93,8	93,3	100	100	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800 [Hex A pseudodeficiency], 272800
HEXB	99,6	96,9	100	99,9	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HEY2	100	99,3	100	100	No OMIM disease ID
HFE	100	99,7	100	100	{Porphyria variegata, susceptibility to}, 176200 {Microvascular complications of diabetes 7}, 612635 {Porphyria cutanea tarda, susceptibility to}, 176100 [Transferrin serum level QTL2], 614193 {Alzheimer disease, susceptibility to}, 104300 Hemochromatosis, 235200
HFM1	96,3	89,6	100	100	Premature ovarian failure 9, 615724
HGD	100	100	100	100	Alkaptonuria, 203500
HGF	100	99,4	100	100	Deafness, autosomal recessive 39, 608265
HGSNAT	86,4	86,3	91,2	89,3	Retinitis pigmentosa 73, 616544 Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930
HIBADH	94,4	91,2	100	100	No OMIM disease ID

HIBCH	98,2	88,5	100	100	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HIKESHI	98,2	90,4	100	100	Leukodystrophy, hypomyelinating, 13, 616881
HINT1	98,3	89,3	100	100	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200
HIVEP2	100	100	100	100	Mental retardation, autosomal dominant 43, 616977
HJV	100	100	100	100	Hemochromatosis, type 2A, 602390
HK1	100	100	100	100	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285 Neurodevelopmental disorder with visual defects and brain anomalies, 618547 Retinitis pigmentosa 79, 617460
HLCS	100	100	100	100	Holocarboxylase synthetase deficiency, 253270
HMBS	99,9	99,4	100	100	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000
HMGA2	81,3	76,7	75,1	73,8	Silver-Russell syndrome 5, 618908
HMGB3	78,6	67,2	100	100	?Microphthalmia, syndromic 13, 300915
HMGCL	100	99,8	100	100	HMG-CoA lyase deficiency, 246450
HMGCS2	100	99,6	100	100	HMG-CoA synthase-2 deficiency, 605911
HMOX1	98,4	89,9	100	100	{Pulmonary disease, chronic obstructive, susceptibility to}, 606963 Heme oxygenase-1 deficiency, 614034
HMX1	62,4	42,9	99,7	96,1	Oculoauricular syndrome, 612109
HNF1A	100	99,8	100	100	{Diabetes mellitus, insulin-dependent}, 222100 MODY, type III, 600496 Hepatic adenoma, somatic, 142330 Renal cell carcinoma, 144700 Diabetes mellitus, insulin-dependent, 20, 612520 {Diabetes mellitus, noninsulin-dependent, 2}, 125853
HNF1B	99,3	96,1	100	100	Diabetes mellitus, noninsulin-dependent, 125853 Renal cysts and diabetes syndrome, 137920 {Renal cell carcinoma}, 144700
HNF4A	99,9	99	100	100	{Diabetes mellitus, noninsulin-dependent}, 125853 MODY, type I, 125850 Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026

HNMT	100	99,8	100	100	{Asthma, susceptibility to}, 600807 Mental retardation, autosomal recessive 51, 616739
HNRNPA1	98,8	90	100	100	Amyotrophic lateral sclerosis 20, 615426 ?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3, 615424
HNRNPA2B1	99,9	99,4	100	100	?Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2, 615422
HNRNPDL	97,3	88,4	100	100	Muscular dystrophy, limb-girdle, autosomal dominant 3, 609115
HNRNPH1	99,4	96,2	100	100	No OMIM disease ID
HNRNPH2	100	100	100	100	Mental retardation, X-linked, syndromic, Bain type, 300986
HNRNPK	91,5	82,8	100	100	Au-Kline syndrome, 616580
HNRNPU	99,9	98,9	100	100	Developmental and epileptic encephalopathy 54, 617391
HOGA1	100	96,4	100	100	Hyperoxaluria, primary, type III, 613616
HOMER2	99,5	99,4	100	100	?Deafness, autosomal dominant 68, 616707
HOOK1	99,6	96,5	100	100	No OMIM disease ID
HOXA1	100	100	100	100	Athabaskan brainstem dysgenesis syndrome, 601536 Bosley-Salih-Alorainy syndrome, 601536
HOXA11	97,1	87,5	100	100	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
HOXA13	77,7	69	89,7	79,7	?Guttmacher syndrome, 176305 Hand-foot-uterus syndrome, 140000
HOXA2	100	99,9	100	100	Microtia with or without hearing impairment (AD), 612290 ?Microtia, hearing impairment, and cleft palate (AR), 612290
HOXA4	87,8	77	99,4	96	No OMIM disease ID
HOXB1	100	100	100	100	Facial paresis, hereditary congenital, 3, 614744
HOXB13	100	99,1	100	100	{Prostate cancer, hereditary, 9}, 610997
HOXB4	100	97,3	100	99,9	No OMIM disease ID
HOXC13	100	99,9	100	100	Ectodermal dysplasia 9, hair/nail type, 614931
HOXD10	100	100	100	100	Charcot-Marie-Tooth disease, foot deformity of, 192950 Vertical talus, congenital, 192950
HOXD13	99,9	98,6	100	100	Brachydactyly, type D, 113200 Brachydactyly, type E, 113300 ?Brachydactyly-syndactyly syndrome, 610713

					Syndactyly, type V, 186300 Synpolydactyly 1, 186000
HPCA	100	100	100	100	Dystonia 2, torsion, autosomal recessive, 224500
HPD	100	100	100	100	Tyrosinemia, type III, 276710 Hawkinsinuria, 140350
HPDL	100	100	100	100	Spastic paraplegia 83, autosomal recessive, 619027 Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026
HPGD	100	98,9	100	100	Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100 ?Digital clubbing, isolated congenital, 119900 Cranioosteoarthropathy, 259100
HPRT1	99,3	91,8	100	99,3	Hyperuricemia, HRPT-related, 300323 Lesch-Nyhan syndrome, 300322
HPS1	100	100	100	100	Hermansky-Pudlak syndrome 1, 203300
HPS3	99,7	97,5	100	100	Hermansky-Pudlak syndrome 3, 614072
HPS4	100	100	100	100	Hermansky-Pudlak syndrome 4, 614073
HPS5	100	99,7	100	100	Hermansky-Pudlak syndrome 5, 614074
HPS6	97,1	88,9	100	100	Hermansky-Pudlak syndrome 6, 614075
HPSE2	100	99,9	100	100	Urofacial syndrome 1, 236730
HR	98,5	95,6	100	100	Hypotrichosis 4, 146550 Alopecia universalis, 203655 Atrichia with papular lesions, 209500
HRAS	100	100	100	100	Nevus sebaceous or woolly hair nevus, somatic, 162900 Congenital myopathy with excess of muscle spindles, 218040 Bladder cancer, somatic, 109800 Thyroid carcinoma, follicular, somatic, 188470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Spitz nevus or nevus spilus, somatic, 137550 Costello syndrome, 218040
HRG	95	94,2	100	100	Thrombophilia due to HRG deficiency, 613116
HS6ST1	92,9	84,5	100	100	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880
HS6ST2	97,6	96,6	100	100	?Paganini-Miozzo syndrome, 301025
HSCB	100	98,7	100	100	No OMIM disease ID

HSD11B1	100	99,6	100	100	Cortisone reductase deficiency 2, 614662
HSD11B2	86	82,7	99,9	98,1	Apparent mineralocorticoid excess, 218030
HSD17B10	100	99,1	100	100	HSD10 mitochondrial disease, 300438
HSD17B3	97,8	97,8	97,8	97,8	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	95,4	93,1	96,6	96,6	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B2	100	99,7	100	100	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810
HSD3B7	99,1	95,5	100	100	Bile acid synthesis defect, congenital, 1, 607765
HSF4	99,6	97,2	100	100	Cataract 5, multiple types, 116800
HSPA9	88,5	84,5	100	100	Even-plus syndrome, 616854 Anemia, sideroblastic, 4, 182170
HSPB1	98,8	91,6	100	100	Neuronopathy, distal hereditary motor, type IIB, 608634 Charcot-Marie-Tooth disease, axonal, type 2F, 606595
HSPB3	100	100	100	100	?Neuronopathy, distal hereditary motor, type IIC, 613376
HSPB6	91,1	81	100	100	No OMIM disease ID
HSPB8	100	100	100	100	Neuronopathy, distal hereditary motor, type IIA, 158590 Charcot-Marie-Tooth disease, axonal, type 2L, 608673
HSPD1	98,8	93,7	100	100	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233
HSPG2	99,2	97,7	100	99,9	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
HTR1A	100	100	100	100	Periodic fever, menstrual cycle dependent, 614674
HTRA1	75,8	72,3	87,2	83,2	{Macular degeneration, age-related, neovascular type}, 610149 {Macular degeneration, age-related, 7}, 610149 Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779 CARASIL syndrome, 600142
HTRA2	100	99,9	100	100	{Parkinson disease 13}, 610297 3-methylglutaconic aciduria, type VIII, 617248
HTT	97,8	96,6	100	99,9	Huntington disease, 143100 Lopes-Maciel-Rodan syndrome, 617435
HUWE1	99,2	95,8	100	100	Mental retardation, X-linked syndromic, Turner type, 309590

HYAL1	100	100	100	100	?Mucopolysaccharidosis type IX, 601492
HYAL2	100	100	100	100	No OMIM disease ID
HYDIN	99,9	99,3	100	100	Ciliary dyskinesia, primary, 5, 608647
HYLS1	100	100	100	100	Hydrolethalus syndrome, 236680
HYOU1	100	99,5	100	100	?Immunodeficiency 59 and hypoglycemia, 233600
IARS1	100	99,6	100	100	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093
IARS2	100	99,9	100	100	?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
IBA57	93,7	90,1	100	100	?Spastic paraplegia 74, autosomal recessive, 616451 Multiple mitochondrial dysfunctions syndrome 3, 615330
ICOS	99,9	99,8	100	100	Immunodeficiency, common variable, 1, 607594
ICOSLG	99,5	98,8	100	100	No OMIM disease ID
ID4	87,6	82,5	98,9	93,1	No OMIM disease ID
IDH1	93,3	80,1	100	100	{Glioma, susceptibility to, somatic}, 137800
IDH2	99,7	97,4	100	99,8	D-2-hydroxyglutaric aciduria 2, 613657
IDH3A	99,4	97,3	100	100	Retinitis pigmentosa 90, 619007
IDH3B	95,4	95,4	100	100	Retinitis pigmentosa 46, 612572
IDI1	99,8	97,6	100	100	No OMIM disease ID
IDS	99,9	98	100	100	Mucopolysaccharidosis II, 309900
IDUA	93,7	86,8	100	100	Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Is, 607016
IER3IP1	91,9	82,6	100	100	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	99,7	98,4	100	100	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFITM5	99,3	95,6	100	100	Osteogenesis imperfecta, type V, 610967
IFNAR1	97,7	97,1	97,8	97,8	No OMIM disease ID

IFNAR2	100	99,7	100	100	?Immunodeficiency 45, 616669 {Hepatitis B virus, susceptibility to}, 610424
IFNG	100	100	100	100	?Immunodeficiency 69, mycobacteriosis, 618963 {AIDS, rapid progression to}, 609423 {TSC2 angiomyolipomas, renal, modifier of}, 613254 {Hepatitis C virus, response to therapy of}, 609532 {Aplastic anemia}, 609135 {Tuberculosis, protection against}, 607948
IFNGR1	98,2	97,5	100	100	Immunodeficiency 27A, mycobacteriosis, AR, 209950 {Tuberculosis infection, protection against}, 607948 {Tuberculosis, susceptibility to}, 607948 Immunodeficiency 27B, mycobacteriosis, AD, 615978 {H. pylori infection, susceptibility to}, 600263 {Hepatitis B virus infection, susceptibility to}, 610424
IFNGR2	93,3	93,2	100	99,8	Immunodeficiency 28, mycobacteriosis, 614889
IFNLR1	99,4	97,1	100	100	No OMIM disease ID
IFRD1	99,7	98,6	100	100	No OMIM disease ID
IFT122	100	99,6	100	100	Cranioectodermal dysplasia 1, 218330
IFT140	99,8	98,8	100	100	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	99,9	99,1	100	100	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	100	100	100	100	?Bardet-Biedl syndrome 19, 615996
IFT43	100	100	100	100	?Cranioectodermal dysplasia 3, 614099 Short-rib thoracic dysplasia 18 with polydactyly, 617866 ?Retinitis pigmentosa 81, 617871
IFT52	100	99,9	100	100	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102
IFT57	99,9	99,1	100	100	?Orofaciodigital syndrome XVIII, 617927
IFT74	98,4	93,9	100	100	?Bardet-Biedl syndrome 20, 617119
IFT80	97,6	88,2	100	100	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IFT81	93,5	90,1	95	94,9	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IFT88	99,6	97,3	100	100	No OMIM disease ID

IGBP1	99,5	96,2	100	100	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472
IGF1	100	99,9	100	100	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IGF1R	100	99,9	100	100	Insulin-like growth factor I, resistance to, 270450
IGF2	100	100	100	100	Silver-Russell syndrome 3, 616489
IGF2R	98,9	97,3	99,9	99,7	Hepatocellular carcinoma, somatic, 114550
IGFALS	99,9	99,6	100	100	Acid-labile subunit, deficiency of, 615961
IGFBP7	92,7	87,2	100	100	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis, 614224
IGHG2	68,8	49,6	100	100	IgG2 deficiency, selective, 0
IGHM	100	100	100	100	Agammaglobulinemia 1, 601495
IGHMBP2	98,8	95,1	100	100	Neuronopathy, distal hereditary motor, type VI, 604320 Charcot-Marie-Tooth disease, axonal, type 2S, 616155
IGKC	100	100	100	100	Kappa light chain deficiency, 614102
IGLL1	99,9	96,9	100	100	Agammaglobulinemia 2, 613500
IGSF1	99,5	96,3	100	100	Hypothyroidism, central, and testicular enlargement, 300888
IGSF10	100	100	100	100	No OMIM disease ID
IGSF3	95,4	94	100	100	?Lacrimal duct defect, 149700
IHH	100	100	100	100	Acrocapitofemoral dysplasia, 607778 Brachydactyly, type A1, 112500
IKBKB	99,8	97,4	100	100	Immunodeficiency 15A, 618204 Immunodeficiency 15B, 615592
IKBKG	84,1	77,2	100	100	Immunodeficiency 33, 300636 Incontinentia pigmenti, 308300 Ectodermal dysplasia and immunodeficiency 1, 300291
IKZF1	99,3	99,3	100	100	Immunodeficiency, common variable, 13, 616873
IKZF5	100	100	100	100	Thrombocytopenia, autosomal dominant, 7, 619130
IL10	99,8	98,2	100	100	{Graft-versus-host disease, protection against}, 614395 {HIV-1, susceptibility to}, 609423 {Rheumatoid arthritis, progression of}, 180300
IL10RA	100	100	100	100	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148

IL10RB	99,8	98	100	100	{Hepatitis B virus, susceptibility to}, 610424 Inflammatory bowel disease 25, early onset, autosomal recessive, 612567
IL11RA	100	99,9	100	100	Craniosynostosis and dental anomalies, 614188
IL12B	100	99,3	100	100	Immunodeficiency 29, mycobacteriosis, 614890
IL12RB1	98,9	96,3	94,1	94,1	Immunodeficiency 30, 614891
IL17F	99,9	97,2	100	100	?Candidiasis, familial, 6, autosomal dominant, 613956
IL17RA	100	99,4	100	100	Immunodeficiency 51, 613953
IL17RC	100	99,9	100	100	Candidiasis, familial, 9, 616445
IL17RD	99,9	99,1	100	100	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
IL18BP	100	100	100	100	{?Hepatitis, fulminant viral, susceptibility to}, 618549
IL1B	100	100	100	100	{Gastric cancer risk after H. pylori infection}, 137215
IL1RAPL1	99,8	98,6	100	100	Mental retardation, X-linked 21/34, 300143
IL1RN	100	100	100	100	{Gastric cancer risk after H. pylori infection}, 137215 {Microvascular complications of diabetes 4}, 612628 Interleukin 1 receptor antagonist deficiency, 612852
IL2	94,5	88	100	100	No OMIM disease ID
IL21	99,4	95,7	100	100	?Immunodeficiency, common variable, 11, 615767
IL21R	100	100	100	100	[IgE, elevated level of], 147050 Immunodeficiency 56, 615207
IL2RA	100	99,7	100	100	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367 {Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942
IL2RB	100	99,7	100	100	Immunodeficiency 63 with lymphoproliferation and autoimmunity, 618495
IL2RG	99,8	97,1	100	100	Severe combined immunodeficiency, X-linked, 300400 Combined immunodeficiency, X-linked, moderate, 312863
IL31RA	99,9	99,9	100	100	?Amyloidosis, primary localized cutaneous, 2, 613955
IL36RN	100	100	100	100	Psoriasis 14, pustular, 614204
IL6R	98,4	94,2	92,8	92,7	[Interleukin-6 receptor, soluble, serum level of, QTL], 614689 [Interleukin 6, serum level of, QTL], 614752 Hyper-IgE recurrent infection syndrome 5, autosomal recessive, 618944
IL6ST	96,4	90,3	100	100	Hyper-IgE recurrent infection syndrome 4, autosomal recessive, 618523

IL7R	100	99,8	100	100	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
ILDR1	99,9	98,4	100	100	Deafness, autosomal recessive 42, 609646
ILK	100	100	100	100	No OMIM disease ID
IMPA1	97	87	100	100	Mental retardation, autosomal recessive 59, 617323
IMPDH1	87,9	80,2	100	100	Leber congenital amaurosis 11, 613837 Retinitis pigmentosa 10, 180105
IMPG1	99,7	98,5	100	100	Macular dystrophy, vitelliform, 4, 616151
IMPG2	99,8	98,4	100	100	Macular dystrophy, vitelliform, 5, 616152 Retinitis pigmentosa 56, 613581
INF2	86,7	83,8	100	100	Glomerulosclerosis, focal segmental, 5, 613237 Charcot-Marie-Tooth disease, dominant intermediate E, 614455
ING1	100	99,9	100	100	Squamous cell carcinoma, head and neck, somatic, 275355
INO80	100	99,1	100	100	No OMIM disease ID
INPP5E	97,1	92,7	100	100	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300
INPP5K	100	100	100	100	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404
INPPL1	98,4	94,5	99,9	99,7	Opsismodysplasia, 258480
INS	99,9	97,9	100	100	Maturity-onset diabetes of the young, type 10, 613370 Hyperproinsulinemia, 616214 Diabetes mellitus, insulin-dependent, 2, 125852 Diabetes mellitus, permanent neonatal 4, 618858
INSL3	80,6	78,3	80,7	80,7	Cryptorchidism, 219050
INSR	97,8	94,7	99,9	99,2	Hyperinsulinemic hypoglycemia, familial, 5, 609968 Rabson-Mendenhall syndrome, 262190 Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Leprechaunism, 246200
INTS1	99,8	98,5	100	100	Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies, 618571
INTS8	99,9	98,8	100	100	?Neurodevelopmental disorder with cerebellar hypoplasia and spasticity, 618572
INTU	99,7	98,1	100	100	?Short-rib thoracic dysplasia 20 with polydactyly, 617925 ?Orofaciodigital syndrome XVII, 617926
INVS	100	100	100	100	Nephronophthisis 2, infantile, 602088

IPMK	99,2	92	100	100	No OMIM disease ID
IQCB1	93,9	85	100	100	Senior-Loken syndrome 5, 609254
IQCE	99,9	98,8	100	100	Polydactyly, postaxial, type A7, 617642
IQSEC1	89,1	86,3	97,6	95,2	Intellectual developmental disorder with short stature and behavioral abnormalities, 618687
IQSEC2	96,8	88,6	99,4	98,4	Mental retardation, X-linked 1/78, 309530
IRAK1	99,3	94,9	99,9	99,4	No OMIM disease ID
IRAK4	99,8	97,7	100	100	Immunodeficiency 67, 607676
IREB2	100	99,8	100	100	Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451
IRF1	100	100	100	100	Gastric cancer, somatic, 613659 Nonsmall cell lung cancer, somatic, 211980 Myelogenous leukemia, acute, 0 Myelodysplastic syndrome, preleukemic, 0
IRF2BP2	93,9	77,7	100	99,9	?Immunodeficiency, common variable, 14, 617765
IRF2BPL	99,5	95	99,9	99,2	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088
IRF3	100	99,8	100	100	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 7}, 616532
IRF4	100	100	100	100	[Skin/hair/eye pigmentation, variation in, 8], 611724
IRF6	99,6	95,9	100	100	Popliteal pterygium syndrome 1, 119500 {Orofacial cleft 6}, 608864 van der Woude syndrome, 119300
IRF7	100	99,9	100	100	?Immunodeficiency 39, 616345
IRF8	99	95,7	100	100	Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990 Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893
IRF9	100	100	100	100	Immunodeficiency 65, susceptibility to viral infections, 618648
IRS4	100	100	100	100	Hypothyroidism, congenital, nongoitrous, 9, 301035
IRX1	87,4	81,3	97,7	94	No OMIM disease ID
IRX2	92,9	76,5	100	99,7	No OMIM disease ID
IRX4	98	94,8	100	100	No OMIM disease ID
IRX5	99,9	98,2	100	99,8	Hamamy syndrome, 611174

ISCA1	94,2	85,9	95,1	95,1	Multiple mitochondrial dysfunctions syndrome 5, 617613
ISCA2	100	98,8	100	100	Multiple mitochondrial dysfunctions syndrome 4, 616370
ISCU	100	100	100	100	Myopathy with lactic acidosis, hereditary, 255125
ISG15	100	100	100	100	Immunodeficiency 38, 616126
ITCH	91,6	91,3	95,9	95	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITGA2	99,6	98,1	100	100	No OMIM disease ID
ITGA2B	99,7	97,8	100	100	Glanzmann thrombasthenia, 273800 Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Thrombocytopenia, neonatal alloimmune, BAK antigen related, 0
ITGA3	99,5	97,4	100	100	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748
ITGA6	99,9	98,9	100	100	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730
ITGA7	99,6	98	100	100	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
ITGA8	99,9	99,7	100	100	Renal hypodysplasia/aplasia 1, 191830
ITGB2	97,2	97,2	97,2	97,2	Leukocyte adhesion deficiency, 116920
ITGB3	100	99,4	100	100	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 {Myocardial infarction, susceptibility to}, 608446 Glanzmann thrombasthenia, 273800 Purpura, posttransfusion, 0 Thrombocytopenia, neonatal alloimmune, 0
ITGB4	98,4	96,2	100	100	Epidermolysis bullosa of hands and feet, 131800 Epidermolysis bullosa, junctional, with pyloric atresia, 226730 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
ITGB6	97,2	95,8	100	100	Amelogenesis imperfecta, type IH, 616221
ITK	100	98,9	100	100	Lymphoproliferative syndrome 1, 613011
ITM2B	100	99,8	100	100	Dementia, familial British, 176500 ?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079 Dementia, familial Danish, 117300
ITPA	100	100	100	100	Developmental and epileptic encephalopathy 35, 616647 [Inosine triphosphatase deficiency], 613850

ITPR1	100	99,9	100	100	Gillespie syndrome, 206700 Spinocerebellar ataxia 29, congenital nonprogressive, 117360 Spinocerebellar ataxia 15, 606658
ITPR2	99,9	99,3	100	100	?Anhidrosis, isolated, with normal sweat glands, 106190
ITPR3	100	99,7	100	100	{Diabetes, type 1, susceptibility to}, 222100
ITSN1	99,4	97,4	100	100	No OMIM disease ID
ITSN2	98,8	96,5	100	100	No OMIM disease ID
IVD	100	100	100	100	Isovaleric acidemia, 243500
IVNS1ABP	99,9	98,4	100	100	Immunodeficiency 70, 618969
IYD	99,5	95,7	100	100	Thyroid dyshormonogenesis 4, 274800
JAG1	97,7	96,8	100	100	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500
JAGN1	100	100	99,7	98	Neutropenia, severe congenital, 6, autosomal recessive, 616022
JAK1	100	99,8	100	99,7	Autoinflammation, immune dysregulation, and eosinophilia, 618999
JAK2	98,1	95,8	100	100	Myelofibrosis, somatic, 254450 Thrombocythemia 3, 614521 Polycythemia vera, somatic, 263300 {Budd-Chiari syndrome, somatic}, 600880 Leukemia, acute myeloid, somatic, 601626 Erythrocytosis, somatic, 133100
JAK3	99,9	98,7	100	100	SCID, autosomal recessive, T-negative/B-positive type, 600802
JAM2	100	99,9	92,3	92,3	Basal ganglia calcification, idiopathic, 8, autosomal recessive, 618824
JAM3	100	99,9	100	100	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
JMJD1C	99,9	99,2	100	100	No OMIM disease ID
JPH1	100	99,9	100	100	?Charcot-Marie-Tooth disease, axonal, autosomal dominant, type 2K, 607831
JPH2	95,5	80,3	100	100	Cardiomyopathy, hypertrophic, 17, 613873
JPH3	100	99,8	100	100	Huntington disease-like 2, 606438
JUP	100	99,5	100	100	Arrhythmogenic right ventricular dysplasia 12, 611528 Naxos disease, 601214

KALRN	99,9	99,6	100	100	No OMIM disease ID
KANK1	100	100	100	100	Cerebral palsy, spastic quadriplegic, 2, 612900
KANK2	100	100	100	100	Palmoplantar keratoderma and woolly hair, 616099 Nephrotic syndrome, type 16, 617783
KANSL1	99,9	99,2	100	100	Koolen-De Vries syndrome, 610443
KARS1	100	99,9	100	100	?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness, autosomal recessive 89, 613916
KAT6A	100	99,8	100	100	Arboleda-Tham syndrome, 616268
KAT6B	99,6	98,3	100	100	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KAT8	99,9	98,8	100	100	Li-Ghorgani-Weisz-Hubshman syndrome, 618974
KATNB1	100	99,9	100	100	Lissencephaly 6, with microcephaly, 616212
KIAA0556	100	99,9	100	100	Joubert syndrome 26, 616784
KBTBD13	99,8	95,8	100	100	Nemaline myopathy 6, autosomal dominant, 609273
KCNA1	100	99,9	100	100	Episodic ataxia/myokymia syndrome, 160120
KCNA2	100	99,6	100	100	Developmental and epileptic encephalopathy 32, 616366
KCNA4	100	100	100	100	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284
KCNA5	100	98,5	100	100	Atrial fibrillation, familial, 7, 612240
KCNB1	100	99,6	100	100	Developmental and epileptic encephalopathy 26, 616056
KCNC1	100	100	100	100	Epilepsy, progressive myoclonic 7, 616187
KCNC3	78,6	65,8	95	89,7	Spinocerebellar ataxia 13, 605259
KCND2	100	100	100	100	No OMIM disease ID
KCND3	100	99,4	100	100	Brugada syndrome 9, 616399 Spinocerebellar ataxia 19, 607346
KCNE1	100	100	100	100	Long QT syndrome 5, 613695 Jervell and Lange-Nielsen syndrome 2, 612347
KCNE2	100	97,2	100	100	Atrial fibrillation, familial, 4, 611493 Long QT syndrome 6, 613693
KCNE3	100	100	100	100	?Brugada syndrome 6, 613119

KCNE4	80,5	80,4	100	100	No OMIM disease ID
KCNE5	98,6	91,8	100	100	No OMIM disease ID
KCNH1	98,7	98,7	98,7	98,7	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500
KCNH2	95,8	91,9	100	100	{Long QT syndrome 2, acquired, susceptibility to}, 613688 Long QT syndrome 2, 613688 Short QT syndrome 1, 609620
KCNJ1	100	100	100	100	Bartter syndrome, type 2, 241200
KCNJ10	89,3	89	100	100	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ11	100	100	100	100	Maturity-onset diabetes of the young, type 13, 616329 {Diabetes mellitus, type 2, susceptibility to}, 125853 Diabetes, permanent neonatal 2, with or without neurologic features, 618856 Diabetes mellitus, transient neonatal 3, 610582 Hyperinsulinemic hypoglycemia, familial, 2, 601820
KCNJ13	100	100	100	100	Leber congenital amaurosis 16, 614186 Snowflake vitreoretinal degeneration, 193230
KCNJ2	100	100	100	100	Short QT syndrome 3, 609622 Atrial fibrillation, familial, 9, 613980 Andersen syndrome, 170390
KCNJ5	100	100	100	100	Long QT syndrome 13, 613485 Hyperaldosteronism, familial, type III, 613677
KCNJ6	100	100	100	100	Keppen-Lubinsky syndrome, 614098
KCNJ8	100	100	100	100	No OMIM disease ID
KCNK3	97,5	95	100	100	Pulmonary hypertension, primary, 4, 615344
KCNK4	99,1	97,4	100	100	Facial dysmorphism, hypertrichosis, epilepsy, intellectual/developmental delay, and gingival overgrowth syndrome, 618381
KCNK9	97,3	97,3	97,3	97,3	Birk-Barel syndrome, 612292
KCNMA1	94,4	93,6	100	100	Liang-Wang syndrome, 618729 {Epilepsy, idiopathic generalized, susceptibility to, 16}, 618596 Cerebellar atrophy, developmental delay, and seizures, 617643 Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446
KCNN3	100	99,7	100	100	Zimmermann-Laband syndrome 3, 618658

KCNN4	100	99,4	100	100	Dehydrated hereditary stomatocytosis 2, 616689
KCNQ1	93,3	90,6	100	99,8	Long QT syndrome 1, 192500 Jervell and Lange-Nielsen syndrome, 220400 Short QT syndrome 2, 609621 {Long QT syndrome 1, acquired, susceptibility to}, 192500 Atrial fibrillation, familial, 3, 607554
KCNQ1OT1					Beckwith-Wiedemann syndrome, 130650
KCNQ2	91,3	89,8	100	100	Seizures, benign neonatal, 1, 121200 Developmental and epileptic encephalopathy 7, 613720 Myokymia, 121200
KCNQ3	100	99,4	99,8	99,1	Seizures, benign neonatal, 2, 121201
KCNQ4	97	95,7	96,4	93,9	Deafness, autosomal dominant 2A, 600101
KCNQ5	97,8	95,5	100	100	Mental retardation, autosomal dominant 46, 617601
KCNT1	96	95,2	98,6	97,3	Epilepsy nocturnal frontal lobe, 5, 615005 Developmental and epileptic encephalopathy 14, 614959
KCNT2	99,4	97,1	100	100	?Developmental and epileptic encephalopathy 57, 617771
KCNV2	100	99,9	100	100	Retinal cone dystrophy 3B, 610356
KCTD1	97,3	88,8	99,9	99,4	Scalp-ear-nipple syndrome, 181270
KCTD17	100	99	100	100	Dystonia 26, myoclonic, 616398
KCTD3	100	99,7	100	100	No OMIM disease ID
KCTD7	95	95	100	100	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDELR2	100	100	100	100	Osteogenesis imperfecta 21, 619131
KDF1	100	99,8	100	100	?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/nail type, 617337
KDM1A	98,2	95,3	100	100	Cleft palate, psychomotor retardation, and distinctive facial features, 616728
KDM3B	97,5	96,3	100	100	Diets-Jongmans syndrome, 618846
KDM5B	94,6	92,3	93,9	92,9	Mental retardation, autosomal recessive 65, 618109
KDM5C	99,8	97,9	100	100	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
KDM6A	96,1	88,7	100	99,9	Kabuki syndrome 2, 300867

KDM6B	98,8	97,9	100	100	Neurodevelopmental disorder with coarse facies and mild distal skeletal abnormalities, 618505
KDR	100	99,8	100	100	{Hemangioma, capillary infantile, susceptibility to}, 602089 Hemangioma, capillary infantile, somatic, 602089
KDSR	100	99,5	100	100	Erythrokeratodermia variabilis et progressiva 4, 617526
KERA	100	100	100	100	Cornea plana 2, autosomal recessive, 217300
KHDC3L	100	99,8	100	100	Hydatidiform mole, recurrent, 2, 614293
KIAA0586	97,3	93,1	95,8	95,8	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546
KIAA0753	100	99,3	100	100	?Orofaciodigital syndrome XV, 617127
KIAA0825	99,4	97,7	100	100	Polydactyly, postaxial, type A10, 618498
KIAA1109	99,8	99,2	100	100	Alkuraya-Kucinskas syndrome, 617822
KIAA1549	97,9	96,4	98,8	98	Retinitis pigmentosa 86, 618613
KIDINS220	100	100	100	100	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296
KIF11	97,6	94,8	100	100	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF14	99,6	97,7	100	100	Microcephaly 20, primary, autosomal recessive, 617914 ?Meckel syndrome 12, 616258
KIF1A	97,4	95,2	98	98	NESCAV syndrome, 614255 Spastic paraplegia 30, autosomal dominant, 610357 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal recessive, 610357
KIF1B	100	99,6	100	100	Pheochromocytoma, 171300 ?Charcot-Marie-Tooth disease, type 2A1, 118210 {Neuroblastoma, susceptibility to, 1}, 256700
KIF1C	100	100	100	100	Spastic ataxia 2, autosomal recessive, 611302
KIF21A	99,9	99,3	100	100	Fibrosis of extraocular muscles, congenital, 1, 135700 Fibrosis of extraocular muscles, congenital, 3B, 135700
KIF22	100	100	100	100	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546
KIF23	99,5	96,3	100	100	No OMIM disease ID
KIF2A	99,6	95,6	100	100	Cortical dysplasia, complex, with other brain malformations 3, 615411
KIF3B	100	99,7	100	100	Retinitis pigmentosa 89, 618955

KIF4A	99,4	95,7	100	100	?Mental retardation, X-linked 100, 300923
KIF5A	100	99,9	100	100	Myoclonus, intractable, neonatal, 617235 Spastic paraplegia 10, autosomal dominant, 604187 {Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921
KIF5C	99,9	98,8	99,8	99,8	Cortical dysplasia, complex, with other brain malformations 2, 615282
KIF7	93,6	90,6	99,1	97,8	?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131
KIFBP	96,1	96,1	96,1	96,1	Goldberg-Shprintzen megacolon syndrome, 609460
KIRREL1	100	99,9	100	100	No OMIM disease ID
KIRREL3	99,8	98,9	100	100	No OMIM disease ID
KISS1	100	98,3	100	100	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842
KISS1R	100	99,5	100	100	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 ?Precocious puberty, central, 1, 176400
KIT	100	99,6	100	100	Piebaldism, 172800 Gastrointestinal stromal tumor, familial, 606764 Mastocytosis, cutaneous, 154800 Germ cell tumors, somatic, 273300 Leukemia, acute myeloid, somatic, 601626 Mastocytosis, systemic, somatic, 154800
KITLG	100	98,5	100	100	Deafness, autosomal dominant 69, unilateral or asymmetric, 616697 Hyperpigmentation with or without hypopigmentation, 145250 [Skin/hair/eye pigmentation 7, blond/brown hair], 611664
KIZ	100	99,2	100	100	Retinitis pigmentosa 69, 615780
KL	98,2	97,2	98,5	97,5	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994
KLB	100	99,9	100	100	No OMIM disease ID
KLC2	99,2	97,9	100	100	Spastic paraplegia, optic atrophy, and neuropathy, 609541
KLF1	100	97,8	100	100	Blood group--Lutheran inhibitor, 111150 [Hereditary persistence of fetal hemoglobin], 613566 Dyserythropoietic anemia, congenital, type IV, 613673
KLF10	100	99,9	100	100	No OMIM disease ID

KLF11	100	99,4	100	100	Maturity-onset diabetes of the young, type VII, 610508
KLF6	100	100	100	100	Gastric cancer, somatic, 613659 Prostate cancer, somatic, 176807
KLF7	100	99,7	100	100	No OMIM disease ID
KLHL10	100	100	100	100	Spermatogenic failure 11, 615081
KLHL15	100	99,7	100	100	Mental retardation, X-linked 103, 300982
KLHL24	100	100	100	100	Epidermolysis bullosa simplex, generalized, with scarring and hair loss, 617294
KLHL3	100	99,3	100	100	Pseudohypoaldosteronism, type IID, 614495
KLHL40	100	100	100	100	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	100	99,9	100	100	Nemaline myopathy 9, 615731
KLHL7	99,9	99,8	100	100	Retinitis pigmentosa 42, 612943 PERCHING syndrome, 617055
KLHL9	100	100	100	100	No OMIM disease ID
KLK4	100	100	100	100	Amelogenesis imperfecta, type IIA1, 204700
KLKB1	100	99,5	100	100	Fletcher factor (prekallikrein) deficiency, 612423
KLLN	100	100	100	100	Cowden syndrome 4, 615107
KMT2A	100	99,9	99,9	99,4	Wiedemann-Steiner syndrome, 605130
KMT2B	95,8	94	98,7	97,9	Dystonia 28, childhood-onset, 617284
KMT2C	92,2	91	100	100	Kleefstra syndrome 2, 617768
KMT2D	100	99,4	100	100	Kabuki syndrome 1, 147920
KMT2E	99,8	98,5	100	100	O'Donnell-Luria-Rodan syndrome, 618512
KMT5B	99,9	99,1	100	100	Mental retardation, autosomal dominant 51, 617788
KNG1	100	100	100	100	[Kininogen deficiency], 228960 [High molecular weight kininogen deficiency], 228960
KNL1	99,2	98,1	98,9	98,8	Microcephaly 4, primary, autosomal recessive, 604321
KPTN	100	100	100	100	Mental retardation, autosomal recessive 41, 615637

KRAS	99,5	96,9	100	100	Oculoectodermal syndrome, somatic, 600268 Leukemia, acute myeloid, somatic, 601626 Breast cancer, somatic, 114480 RAS-associated autoimmune leukoproliferative disorder, 614470 Cardiofaciocutaneous syndrome 2, 615278 Arteriovenous malformation of the brain, somatic, 108010 Bladder cancer, somatic, 109800 Pancreatic carcinoma, somatic, 260350 Lung cancer, somatic, 211980 Gastric cancer, somatic, 137215 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Noonan syndrome 3, 609942
KREMEN1	97,7	94,4	99,5	97,9	Ectodermal dysplasia 13, hair/tooth type, 617392
KRIT1	100	99,3	100	100	Cavernous malformations of CNS and retina, 116860 Cerebral cavernous malformations-1, 116860 Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations, 116860
KRT1	98,7	95,6	100	100	Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602 Keratosis palmoplantaris striata III, 607654 Palmoplantar keratoderma, epidermolytic, 144200 Palmoplantar keratoderma, nonepidermolytic, 600962 Ichthyosis histrix, Curth-Macklin type, 146590 Epidermolytic hyperkeratosis, 113800
KRT10	100	99,3	100	100	Epidermolytic hyperkeratosis, 113800 Ichthyosis with confetti, 609165 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602
KRT12	99,7	97,8	100	100	Meesmann corneal dystrophy 1, 122100
KRT13	100	100	100	100	White sponge nevus 2, 615785
KRT14	89	81,9	100	100	Naegeli-Franceschetti-Jadassohn syndrome, 161000 Epidermolysis bullosa simplex, Dowling-Meara type, 131760 Dermatopathia pigmentosa reticularis, 125595 Epidermolysis bullosa simplex, Koebner type, 131900 Epidermolysis bullosa simplex, recessive 1, 601001 Epidermolysis bullosa simplex, Weber-Cockayne type, 131800
KRT16	74,2	56,5	100	100	Palmoplantar keratoderma, nonepidermolytic, focal, 613000 Pachyonychia congenita 1, 167200

KRT17	39,8	22,8	100	100	Pachyonychia congenita 2, 167210 Steatocystoma multiplex, 184500
KRT18	86,7	70,9	100	100	Cirrhosis, cryptogenic, 215600 {Cirrhosis, noncryptogenic, susceptibility to}, 215600
KRT2	100	99,8	100	100	Ichthyosis bullosa of Siemens, 146800
KRT25	100	100	100	100	Woolly hair, autosomal recessive 3, 616760
KRT3	100	100	100	100	Meesmann corneal dystrophy 2, 618767
KRT4	100	99,7	100	100	White sponge nevus 1, 193900
KRT5	100	100	100	100	Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 Epidermolysis bullosa simplex-MCR, 609352 Epidermolysis bullosa simplex-MP, 131960 Dowling-Degos disease 1, 179850 Epidermolysis bullosa simplex, Koebner type, 131900 Epidermolysis bullosa simplex, Dowling-Meara type, 131760 Epidermolysis bullosa simplex, recessive 1, 601001
KRT6A	92,3	87,7	100	100	Pachyonychia congenita 3, 615726
KRT6B	93,6	88,6	100	100	Pachyonychia congenita 4, 615728
KRT6C	88,3	81,3	99,9	99,8	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735
KRT71	100	100	100	100	?Hypotrichosis 13, 615896
KRT74	100	100	100	100	?Ectodermal dysplasia 7, hair/nail type, 614929 Woolly hair, autosomal dominant, 194300 ?Hypotrichosis 3, 613981
KRT75	100	100	100	100	{Pseudofolliculitis barbae, susceptibility to}, 612318
KRT8	90,6	69,6	100	100	{Cirrhosis, noncryptogenic, susceptibility to}, 215600 Cirrhosis, cryptogenic, 215600
KRT81	99,2	94,2	100	100	Monilethrix, 158000
KRT83	96,6	84,4	100	100	Erythrokeratodermia variabilis et progressiva 5, 617756 Monilethrix, 158000
KRT85	99	93,6	100	100	Ectodermal dysplasia 4, hair/nail type, 602032
KRT86	99,7	96,3	100	100	Monilethrix, 158000
KRT9	99,2	95	100	100	Palmoplantar keratoderma, epidermolytic, 144200

KY	100	99,7	100	100	Myopathy, myofibrillar, 7, 617114
KYNU	99,6	97,1	100	100	?Hydroxykynureninuria, 236800 Vertebral, cardiac, renal, and limb defects syndrome 2, 617661
L1CAM	99,9	99,1	100	100	MASA syndrome, 303350 Hydrocephalus with Hirschsprung disease, 307000 Corpus callosum, partial agenesis of, 304100 CRASH syndrome, 303350 Hydrocephalus due to aqueductal stenosis, 307000 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000
L2HGDH	99	97,2	100	100	L-2-hydroxyglutaric aciduria, 236792
LACC1	100	99,4	100	100	Juvenile arthritis, 618795
LACTB	99,5	92,6	100	100	No OMIM disease ID
LAGE3	95,9	85,1	100	100	Galloway-Mowat syndrome 2, X-linked, 301006
LAMA1	100	99,7	100	100	Poretti-Boltshauser syndrome, 615960
LAMA2	100	99,6	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855
LAMA3	100	99,7	100	100	Epidermolysis bullosa, junctional, Herlitz type, 226700 Laryngoonychocutaneous syndrome, 245660 Epidermolysis bullosa, generalized atrophic benign, 226650
LAMA4	100	99,9	100	100	Cardiomyopathy, dilated, 1JJ, 615235
LAMB1	100	99,9	100	100	Lissencephaly 5, 615191
LAMB2	100	99,7	100	100	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 Pierson syndrome, 609049
LAMB3	100	99,6	100	100	Amelogenesis imperfecta, type IA, 104530 Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LAMC2	99,8	98	100	100	Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa, junctional, Herlitz type, 226700
LAMC3	98,6	97,1	100	99,6	Cortical malformations, occipital, 614115
LAMP2	99,2	95,6	100	100	Danon disease, 300257
LAMTOR2	100	99,7	100	100	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LAPTM5	97,9	92,9	100	100	No OMIM disease ID

LARGE1	100	99,6	100	100	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154
LARP7	88,5	78,4	100	100	Alazami syndrome, 615071
LARS1	99,8	98,4	100	100	?Infantile liver failure syndrome 1, 615438
LARS2	100	100	100	100	Perrault syndrome 4, 615300 ?Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LAS1L	99,7	97,3	100	100	Wilson-Turner syndrome, 309585
LAT	100	99,2	100	100	Immunodeficiency 52, 617514
LBR	99,4	94,5	100	100	Pelger-Huet anomaly, 169400 Greenberg skeletal dysplasia, 215140 ?Reynolds syndrome, 613471 Pelger-Huet anomaly with mild skeletal anomalies, 618019
LBX1	100	100	100	100	No OMIM disease ID
LCA5	99,9	99,2	100	100	Leber congenital amaurosis 5, 604537
LCAT	99	93,8	100	100	Norum disease, 245900 Fish-eye disease, 136120
LCK	98,9	96,6	100	100	?Immunodeficiency 22, 615758
LCT	99,8	98,5	100	100	Lactase deficiency, congenital, 223000
LDB3	95,4	94,7	100	100	Cardiomyopathy, hypertrophic, 24, 601493 Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 Myopathy, myofibrillar, 4, 609452 Left ventricular noncompaction 3, 601493
LDHA	95	91,7	100	100	Glycogen storage disease XI, 612933
LDHB	94,7	84,3	100	100	[Lactate dehydrogenase-B deficiency], 614128
LDHD	100	99,5	100	100	D-lactic aciduria with susceptibility to gout, 245450
LDLR	100	99,3	100	100	Hypercholesterolemia, familial, 1, 143890 LDL cholesterol level QTL2, 143890
LDLRAP1	98,8	94,2	100	100	Hypercholesterolemia, familial, 4, 603813
LEF1	100	100	100	100	Sebaceous tumors, somatic, 0
LEFTY2	88,9	81,4	100	100	No OMIM disease ID

LEMD2	98,7	92	100	100	Cataract 46, juvenile-onset, 212500
LEMD3	99,9	98,7	100	100	Osteopoikilosis with or without melorheostosis, 166700 Buschke-Ollendorff syndrome, 166700
LEP	99,9	97,3	100	100	Obesity, morbid, due to leptin deficiency, 614962
LEPR	94,3	92,6	94,6	94,6	Obesity, morbid, due to leptin receptor deficiency, 614963
LFNG	87,9	86,4	92,2	87,7	Spondylocostal dysostosis 3, autosomal recessive, 609813
LGI1	98,5	97,5	100	100	Epilepsy, familial temporal lobe, 1, 600512
LGI4	99,9	97,9	100	100	Arthrogryposis multiplex congenita 1, neurogenic, with myelin defect, 617468
LHB	90,4	38,9	100	100	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300
LHCGR	94,1	92,3	100	100	Leydig cell adenoma, somatic, with precocious puberty, 176410 Precocious puberty, male, 176410 Luteinizing hormone resistance, female, 238320 Leydig cell hypoplasia with pseudohermaphroditism, 238320 Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320
LHFPL5	100	100	100	100	Deafness, autosomal recessive 67, 610265
LHX1	100	99,6	100	100	No OMIM disease ID
LHX3	96,6	96,5	100	100	Pituitary hormone deficiency, combined, 3, 221750
LHX4	100	100	100	100	Pituitary hormone deficiency, combined, 4, 262700
LIAS	100	99,1	100	100	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIFR	99,7	98	100	100	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LIG1	100	99,7	100	100	No OMIM disease ID
LIG4	100	99,9	100	100	{Multiple myeloma, resistance to}, 254500 LIG4 syndrome, 606593
LIM2	100	100	100	100	Cataract 19, multiple types, 615277
LIMS2	93	92,7	99,8	98,9	?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827
LINGO1	100	100	100	100	Mental retardation, autosomal recessive 64, 618103
LINS1	100	99,1	100	100	Mental retardation, autosomal recessive 27, 614340
LIPA	99,2	95,2	95,2	95,2	Wolman disease, 278000 Cholesteryl ester storage disease, 278000

LIPC	100	99,4	100	100	[High density lipoprotein cholesterol level QTL 12], 612797 Hepatic lipase deficiency, 614025 {Diabetes mellitus, noninsulin-dependent}, 125853
LIPE	100	99	100	100	Lipodystrophy, familial partial, type 6, 615980
LIPH	100	99,8	100	100	Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379 Hypotrichosis 7, 604379
LIPN	100	98,9	100	100	Ichthyosis, congenital, autosomal recessive 8, 613943
LIPT1	100	99,9	100	100	Lipoyltransferase 1 deficiency, 616299
LIPT2	94,9	75,2	100	100	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668
LITAF	98,2	92,7	100	100	Charcot-Marie-Tooth disease, type 1C, 601098
LMAN1	99,8	99,2	100	100	Combined factor V and VIII deficiency, 227300
LMAN2L	100	99,7	100	100	?Mental retardation, autosomal recessive, 52, 616887
LMBR1	98,1	96,2	98,7	98,7	Triphalangeal thumb-polysyndactyly syndrome, 174500 Syndactyly, type IV, 186200 Triphalangeal thumb, type I, 174500 Acheiropody, 200500 Laurin-Sandrow syndrome, 135750 Hypoplastic or aplastic tibia with polydactyly, 188740 Polydactyly, preaxial type II, 174500
LMBRD1	94,7	90,2	96,1	96,1	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LMF1	100	99,6	100	100	Lipase deficiency, combined, 246650
LMNA	97,4	91,9	100	100	Muscular dystrophy, congenital, 613205 Lipodystrophy, familial partial, type 2, 151660 Charcot-Marie-Tooth disease, type 2B1, 605588 Cardiomyopathy, dilated, 1A, 115200 Heart-hand syndrome, Slovenian type, 610140 Hutchinson-Gilford progeria, 176670 Restrictive dermopathy, lethal, 275210 Mandibuloacral dysplasia, 248370 Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 Malouf syndrome, 212112
LMNB1	99,9	98,9	100	100	Leukodystrophy, adult-onset, autosomal dominant, 169500

LMNB2	97,5	94,3	98,2	96,9	{Lipodystrophy, partial, acquired, susceptibility to}, 608709 ?Epilepsy, progressive myoclonic, 9, 616540
LMOD1	100	100	100	100	No OMIM disease ID
LMOD3	100	99,7	100	100	Nemaline myopathy 10, 616165
LMX1A	100	100	100	100	Deafness, autosomal dominant 7, 601412
LMX1B	99,6	96,3	100	100	Nail-patella syndrome, 161200 Focal segmental glomerulosclerosis 10, 256020
LNPK	98,4	92,8	93,3	93,3	Neurodevelopmental disorder with epilepsy and hypoplasia of the corpus callosum, 618090
LONP1	100	99,8	100	100	CODAS syndrome, 600373
LORICRIN	99	80,8	100	100	Vohwinkel syndrome with ichthyosis, 604117
LOX	100	99,6	100	100	Aortic aneurysm, familial thoracic 10, 617168
LOXHD1	100	99,7	100	100	Deafness, autosomal recessive 77, 613079
LOXL3	100	99,2	100	100	No OMIM disease ID
LPA	98,8	97,2	100	100	[LPA deficiency, congenital], 618807 {Coronary artery disease, susceptibility to}, 618807
LPAR6	99,6	97,8	100	100	Hypotrichosis 8, 278150 Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150
LPIN1	99,6	97,3	100	100	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	100	100	100	100	Majeed syndrome, 609628
LPL	100	100	100	100	Lipoprotein lipase deficiency, 238600 [High density lipoprotein cholesterol level QTL 11], 238600 Combined hyperlipidemia, familial, 144250
LPP	100	100	100	100	Leukemia, acute myeloid, 601626 Lipoma, 0
LRAT	100	100	100	100	Retinal dystrophy, early-onset severe, 613341 Leber congenital amaurosis 14, 613341 Retinitis pigmentosa, juvenile, 613341
LRBA	99,9	99,6	100	100	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRIG2	99,6	98,8	100	100	Urofacial syndrome 2, 615112
LRIG3	99,8	98,8	100	99,8	No OMIM disease ID

LRIT3	93,9	91,9	100	100	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRMDA	96,8	95,6	99,6	99,6	Albinism, oculocutaneous, type VII, 615179
LRP1	99,7	98,9	100	100	?Keratosis pilaris atrophicans, 604093
LRP12	100	99,8	100	100	Oculopharyngodistal myopathy 1, 164310
LRP2	100	99,9	100	100	Donnai-Barrow syndrome, 222448
LRP4	99,1	98,8	100	100	?Myasthenic syndrome, congenital, 17, 616304 Sclerosteosis 2, 614305 Cenani-Lenz syndactyly syndrome, 212780
LRP5	98,5	98,1	100	99,7	van Buchem disease, type 2, 607636 Exudative vitreoretinopathy 4, 601813 Hyperostosis, endosteal, 144750 Osteosclerosis, 144750 Polycystic liver disease 4 with or without kidney cysts, 617875 Osteoporosis-pseudoglioma syndrome, 259770 Osteopetrosis, autosomal dominant 1, 607634 {Osteoporosis}, 166710 [Bone mineral density variability 1], 601884
LRP6	100	99,9	100	100	{Coronary artery disease, autosomal dominant, 2}, 610947 Tooth agenesis, selective, 7, 616724
LRPAP1	100	100	100	100	Myopia 23, autosomal recessive, 615431
LRPPRC	99,9	99,1	100	100	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111
LRRC10	100	100	100	100	No OMIM disease ID
LRRC56	100	99	100	100	Ciliary dyskinesia, primary, 39, 618254
LRRC6	99,2	96,3	100	100	Ciliary dyskinesia, primary, 19, 614935
LRRC8A	100	99,8	100	100	?Agammaglobulinemia 5, 613506
LRRK1	98,6	97,5	100	100	No OMIM disease ID
LRRK2	99,7	97,8	100	100	{Parkinson disease 8}, 607060
LRSAM1	100	99,9	100	100	Charcot-Marie-Tooth disease, axonal, type 2P, 614436
LRTOMT	100	99,2	100	100	Deafness, autosomal recessive 63, 611451

LSS	100	99,9	100	100	Cataract 44, 616509 Hypotrichosis 14, 618275 Alopecia-mental retardation syndrome 4, 618840
LTBP1	98,9	97,2	100	99,9	No OMIM disease ID
LTBP2	99,9	99	100	100	Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750 Glaucoma 3, primary congenital, D, 613086 ?Weill-Marchesani syndrome 3, recessive, 614819
LTBP3	99,6	98,1	100	100	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
LTBP4	99,9	97,5	100	100	Cutis laxa, autosomal recessive, type IC, 613177
LTC4S	74,2	68,5	100	100	Leukotriene C4 synthase deficiency, 614037
LYRM4	68,5	66,2	66,3	66,3	?Combined oxidative phosphorylation deficiency 19, 615595
LYRM7	95,9	86,2	100	100	Mitochondrial complex III deficiency, nuclear type 8, 615838
LYST	99,6	98,3	100	100	Chediak-Higashi syndrome, 214500
LYZ	100	100	100	100	Amyloidosis, renal, 105200
LZTFL1	99,9	99,2	100	100	Bardet-Biedl syndrome 17, 615994
LZTR1	100	99,9	100	100	{Schwannomatosis-2, susceptibility to}, 615670 Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
LZTS1	100	99,8	100	100	Esophageal squamous cell carcinoma, somatic, 133239
MAB21L1	100	100	100	100	Cerebellar, ocular, craniofacial, and genital syndrome, 618479
MAB21L2	100	100	100	100	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877
MACF1	99,7	99,3	100	100	Lissencephaly 9 with complex brainstem malformation, 618325
MAD1L1	100	97,6	100	100	Prostate cancer, somatic, 176807 Lymphoma, somatic, 0
MAD2L2	100	99,9	100	100	?Fanconi anemia, complementation group V, 617243
MAF	83,5	78	88,6	82,2	Ayme-Gripp syndrome, 601088 Cataract 21, multiple types, 610202
MAFA	93,6	71,5	99,4	95,8	Insulinomatosis and diabetes mellitus, 147630

MAFB	100	99,4	100	100	Duane retraction syndrome 3, 617041 Multicentric carpotarsal osteolysis syndrome, 166300
MAG	100	100	100	100	Spastic paraplegia 75, autosomal recessive, 616680
MAGED2	99,8	97,7	100	99,9	Bartter syndrome, type 5, antenatal, transient, 300971
MAGEL2	93	87,2	100	100	Schaaf-Yang syndrome, 615547
MAGI2	94,5	92,4	94,7	93,3	Nephrotic syndrome, type 15, 617609
MAGT1	98,5	96,5	98,7	98,7	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853 Congenital disorder of glycosylation, type Icc, 301031
MAK	98,7	96,8	100	100	Retinitis pigmentosa 62, 614181
MAL2	99,9	98	100	99,9	No OMIM disease ID
MALT1	91,2	89,4	100	100	Immunodeficiency 12, 615468
MAML2	100	99,6	100	100	Mucoepidermoid salivary gland carcinoma, 0
MAMLD1	99,8	98,2	100	100	Hypospadias 2, X-linked, 300758
MAN1B1	100	99,7	100	99,9	Rafiq syndrome, 614202
MAN2B1	99,8	97,9	100	100	Mannosidosis, alpha-, types I and II, 248500
MAN2B2	99,9	99,1	100	100	No OMIM disease ID
MANBA	87,8	86,5	100	100	Mannosidosis, beta, 248510
MAOA	100	99,7	99,8	98,5	Brunner syndrome, 300615 {Antisocial behavior}, 300615
MAP1B	99,3	97,7	100	100	Periventricular nodular heterotopia 9, 618918
MAP2K1	99,8	97,1	100	100	Cardiofaciocutaneous syndrome 3, 615279 Melorheostosis, isolated, somatic mosaic, 155950
MAP2K2	98,5	95,1	100	100	Cardiofaciocutaneous syndrome 4, 615280
MAP3K1	96,1	91,6	99,7	98,3	46XY sex reversal 6, 613762
MAP3K14	100	99,9	100	100	No OMIM disease ID
MAP3K20	100	99,5	100	100	Split-foot malformation with mesoaxial polydactyly, 616890 Centronuclear myopathy 6 with fiber-type disproportion, 617760
MAP3K7	100	99,6	100	100	Cardiospondylocarpofacial syndrome, 157800 Frontometaphyseal dysplasia 2, 617137

MAP3K8	100	99,9	100	100	Lung cancer, somatic, 211980
MAP4K4	100	99,3	100	100	No OMIM disease ID
MAPK8	100	99,8	100	100	No OMIM disease ID
MAPK8IP3	99,4	99	100	100	Neurodevelopmental disorder with or without variable brain abnormalities, 618443
MAPKAPK3	100	99,6	100	100	?Macular dystrophy, patterned, 3, 617111
MAPKBP1	100	100	100	100	Nephronophthisis 20, 617271
MAPRE2	100	99,3	100	100	Symmetric circumferential skin creases, congenital, 2, 616734
MAPT	100	99,5	100	100	Pick disease, 172700 Dementia, frontotemporal, with or without parkinsonism, 600274 {Parkinson disease, susceptibility to}, 168600 Supranuclear palsy, progressive, 601104 Supranuclear palsy, progressive atypical, 260540
MARCHF6	99,9	98,4	100	100	Epilepsy, familial adult myoclonic, 3, 613608
MARK3	99,9	98,8	100	100	?Visual impairment and progressive phthisis bulbi, 618283
MARS1	99,7	97,4	100	100	Charcot-Marie-Tooth disease, axonal, type 2U, 616280 Interstitial lung and liver disease, 615486
MARS2	100	100	100	100	Spastic ataxia 3, autosomal recessive, 611390 ?Combined oxidative phosphorylation deficiency 25, 616430
MARVELD2	99,2	96,1	100	100	Deafness, autosomal recessive 49, 610153
MASP1	100	99,9	100	100	3MC syndrome 1, 257920
MASP2	100	99,6	100	100	MASP2 deficiency, 613791
MAST1	100	99,5	100	100	Mega-corpus-callosum syndrome with cerebellar hypoplasia and cortical malformations, 618273
MASTL	100	100	100	100	No OMIM disease ID
MAT1A	99,7	97,7	100	100	Methionine adenosyltransferase deficiency, autosomal recessive, 250850 Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850
MAT2A	99,6	96,4	100	100	No OMIM disease ID
MATN3	84,7	84,6	100	100	{Osteoarthritis susceptibility 2}, 140600 ?Spondyloepimetaphyseal dysplasia, Borochowitz Cormier-Daire type, 608728 Epiphyseal dysplasia, multiple, 5, 607078

MATR3	97	93,4	100	100	Amyotrophic lateral sclerosis 21, 606070
MAX	100	98,9	100	100	{Pheochromocytoma, susceptibility to}, 171300
MBD5	99,9	99,9	100	100	Mental retardation, autosomal dominant 1, 156200
MBL2	100	99,8	100	100	{Chronic infections, due to MBL deficiency}, 614372
MBOAT7	100	99,5	100	100	Mental retardation, autosomal recessive 57, 617188
MBTPS1	99,6	98,4	100	100	?Spondyloepiphyseal dysplasia, Kondo-Fu type, 618392
MBTPS2	100	99	100	100	IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800 Osteogenesis imperfecta, type XIX, 301014 ?Olmsted syndrome, X-linked, 300918
MC2R	99,9	98,3	100	100	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MC4R	100	100	100	100	Obesity (BMIQ20), 618406 {Obesity, resistance to (BMIQ20)}, 618406
MCC	100	99,7	100	100	Colorectal cancer, somatic, 114500
MCCC1	100	99,8	100	100	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	99,9	98,4	100	100	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	100	100	100	100	Methylmalonyl-CoA epimerase deficiency, 251120
MCFD2	99,5	96,9	100	100	Factor V and factor VIII, combined deficiency of, 613625
MCIDAS	99,3	96,2	100	100	Ciliary dyskinesia, primary, 42, 618695
MCM2	100	100	100	100	?Deafness, autosomal dominant 70, 616968
MCM3AP	99,9	99,1	100	100	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124
MCM4	95,5	95	95,5	95,5	Immunodeficiency 54, 609981
MCM5	100	99,7	100	100	?Meier-Gorlin syndrome 8, 617564
MCM6	100	100	100	100	Lactase persistence/nonpersistence, 223100
MCM8	100	99,6	94,4	94,4	?Premature ovarian failure 10, 612885
MCM9	99,9	99,8	100	100	Ovarian dysgenesis 4, 616185
MCOLN1	99,8	98,4	100	100	Mucolipidosis IV, 252650

MCPH1	100	99,4	100	100	Microcephaly 1, primary, autosomal recessive, 251200
MCTP2	99,7	98,2	100	100	No OMIM disease ID
MCUR1	99,5	91,5	100	99,8	No OMIM disease ID
MDH1	100	99	100	100	?Developmental and epileptic encephalopathy 88, 618959
MDH2	98	97,9	100	100	Developmental and epileptic encephalopathy 51, 617339
MDM4	99,9	99	100	100	?Bone marrow failure syndrome 6, 618849
MECOM	100	99,9	100	100	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MECP2	100	98,7	100	99,9	Encephalopathy, neonatal severe, 300673 Rett syndrome, atypical, 312750 {Autism susceptibility, X-linked 3}, 300496 Rett syndrome, 312750 Rett syndrome, preserved speech variant, 312750 Mental retardation, X-linked syndromic, Lubs type, 300260 Mental retardation, X-linked, syndromic 13, 300055
MECR	100	98,9	100	100	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MED12	99,8	96,7	100	100	Ohdo syndrome, X-linked, 300895 Lujan-Fryns syndrome, 309520 Opitz-Kaveggia syndrome, 305450
MED13	100	99,9	100	100	Intellectual developmental disorder 61, 618009
MED13L	100	99,8	100	100	Transposition of the great arteries, dextro-looped 1, 608808 Mental retardation and distinctive facial features with or without cardiac defects, 616789
MED17	96,3	93,5	100	100	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	100	99,7	100	100	Mental retardation, autosomal recessive 18, 614249
MED25	100	99,8	100	100	Basel-Vanagait-Smirin-Yosef syndrome, 616449
MEF2C	99,9	96	100	100	Chromosome 5q14.3 deletion syndrome, 613443 Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
MEFV	99,9	98,6	96,4	96,4	Neutrophilic dermatosis, acute febrile, 608068 Familial Mediterranean fever, AR, 249100 Familial Mediterranean fever, AD, 134610
MEGF10	100	100	100	100	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399

MEGF8	99,9	99	100	100	Carpenter syndrome 2, 614976
MEI1	100	99,3	100	100	Hydatidiform mole, recurrent, 3, 618431
MEIOB	100	98,6	100	100	?Spermatogenic failure 22, 617706
MEIS2	100	100	100	100	Cleft palate, cardiac defects, and mental retardation, 600987
MEN1	96,9	94,8	100	100	Multiple endocrine neoplasia 1, 131100 Angiofibroma, somatic, 0 Adrenal adenoma, somatic, 0 Parathyroid adenoma, somatic, 0 Lipoma, somatic, 0 Carcinoid tumor of lung, 0
MEOX1	100	98,9	100	100	Klippel-Feil syndrome 2, 214300
MERTK	99,5	98,8	99,1	99,1	Retinitis pigmentosa 38, 613862
MESD	100	99,9	100	100	Osteogenesis imperfecta, type XX, 618644
MESP2	93,9	86,9	97,5	97,5	Spondylocostal dysostosis 2, autosomal recessive, 608681
MET	100	99,5	100	100	{Osteofibrous dysplasia, susceptibility to}, 607278 Hepatocellular carcinoma, childhood type, somatic, 114550 ?Deafness, autosomal recessive 97, 616705 Renal cell carcinoma, papillary, 1, familial and somatic, 605074
METTL23	100	100	100	100	Mental retardation, autosomal recessive 44, 615942
METTL5	99,3	98,5	99,9	98,3	Intellectual developmental disorder, autosomal recessive 72, 618665
MFAP5	99,9	97,6	100	100	Aortic aneurysm, familial thoracic 9, 616166
MFF	94,3	89,9	100	100	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFN2	100	99,9	100	100	Hereditary motor and sensory neuropathy VIA, 601152 Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260
MFRP	100	100	100	100	Nanophthalmos 2, 609549 Microphthalmia, isolated 5, 611040
MFSD2A	99,7	98,5	100	100	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities, 616486
MFSD8	100	99,7	100	100	Macular dystrophy with central cone involvement, 616170 Ceroid lipofuscinosis, neuronal, 7, 610951
MGAT2	100	100	100	100	Congenital disorder of glycosylation, type IIa, 212066

MGME1	100	100	100	100	Mitochondrial DNA depletion syndrome 11, 615084
MGP	98,7	95,1	100	100	Keutel syndrome, 245150
MIA3	99,8	99,1	100	100	No OMIM disease ID
MIB1	100	99,9	100	100	Left ventricular noncompaction 7, 615092
MICOS13	100	99,7	100	99,7	Combined oxidative phosphorylation deficiency 37, 618329
MICU1	98,9	95,2	100	100	Myopathy with extrapyramidal signs, 615673
MICU2	97,2	91,8	100	100	No OMIM disease ID
MID1	99,8	98,7	100	100	Opitz GBBB syndrome, type I, 300000
MID2	99,8	98,7	100	100	?Mental retardation, X-linked 101, 300928
MIEF2	100	99	100	100	?Combined oxidative phosphorylation deficiency 49, 619024
MINPP1	100	99,5	100	100	{Thyroid carcinoma, follicular}, 188470
MIP	100	98,9	100	100	Cataract 15, multiple types, 615274
MIPEP	99,2	96,5	100	100	Combined oxidative phosphorylation deficiency 31, 617228
MIR140					Spondyloepiphyseal dysplasia, Nishimura type, 618618
MIR17HG					Feingold syndrome 2, 614326
MIR184					EDICT syndrome, 614303
MIR204					?Retinal dystrophy and iris coloboma with or without cataract, 616722
MIR96					Deafness, autosomal dominant 50, 613074
MITF	100	99,9	100	100	COMMAD syndrome, 617306 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 Tietz albinism-deafness syndrome, 103500
MKKS	100	100	100	100	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKRN3	96	96	96	96	Precocious puberty, central, 2, 615346

MKS1	99,8	97,9	100	100	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000
MLC1	100	99	100	100	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MLH1	100	99,9	100	100	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Muir-Torre syndrome, 158320 Mismatch repair cancer syndrome 1, 276300
MLH3	100	100	100	100	{Endometrial cancer, susceptibility to}, 608089 Colorectal cancer, somatic, 114500 Colorectal cancer, hereditary nonpolyposis, type 7, 614385
MLIP	99,9	99	100	100	No OMIM disease ID
MLLT10	96,8	95,5	97,1	97,1	Leukemia, acute myeloid, 601626
MLLT6	98,8	94,3	100	100	No OMIM disease ID
MLPH	100	98,8	100	100	Griscelli syndrome, type 3, 609227
MLYCD	96	90,4	100	98,9	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	100	100	100	100	Methylmalonic aciduria, vitamin B12-responsive, 251100
MMAB	100	99,6	100	100	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110
MMACHC	100	100	100	100	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	94,4	83,5	89,7	89,7	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410
MME	99,8	98,7	98	98	Charcot-Marie-Tooth disease, axonal, type 2T, 617017 ?Spinocerebellar ataxia 43, 617018
MMP1	100	98,9	100	100	COPD, rate of decline of lung function in, 606963 {Epidermolysis bullosa dystrophica, autosomal recessive, modifier of}, 226600
MMP10	99,8	98,1	100	100	No OMIM disease ID
MMP12	100	99,6	100	100	No OMIM disease ID
MMP13	95,2	92,2	92,4	92,4	Metaphyseal dysplasia, Spahr type, 250400 ?Spondyloepimetaphyseal dysplasia, Missouri type, 602111 Metaphyseal anadysplasia 1, 602111
MMP14	100	98,9	100	100	?Winchester syndrome, 277950

MMP19	100	99,5	100	100	Cavitary optic disc anomalies, 611543
MMP2	100	100	100	100	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP20	100	100	100	100	Amelogenesis imperfecta, type IIA2, 612529
MMP21	99,9	98,8	100	100	Heterotaxy, visceral, 7, autosomal, 616749
MMP3	99,5	96,7	100	100	{Coronary heart disease, susceptibility to, 6}, 614466
MMP7	100	99,9	100	100	No OMIM disease ID
MMP8	100	100	100	100	No OMIM disease ID
MMP9	99,1	96,1	100	100	Metaphyseal anadysplasia 2, 613073
MMUT	99,8	98,3	100	100	Methylmalonic aciduria, mut(0) type, 251000
MN1	100	99,3	100	100	Meningioma, 607174 CEBALID syndrome, 618774
MNX1	68,2	58,3	87,4	79,2	Currarino syndrome, 176450
MOCOS	99,8	97,7	100	100	Xanthinuria, type II, 603592
MOCS1	99,2	95,1	100	100	Molybdenum cofactor deficiency A, 252150
MOCS2	99,6	99,5	100	100	Molybdenum cofactor deficiency B, 252160
MOG	100	99,5	100	100	?Narcolepsy 7, 614250
MOGS	100	99,9	100	100	Congenital disorder of glycosylation, type IIb, 606056
MORC2	100	99,8	100	100	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688
MPC1	100	99,6	100	100	Mitochondrial pyruvate carrier deficiency, 614741
MPDU1	100	100	100	100	Congenital disorder of glycosylation, type If, 609180
MPDZ	99,8	98,8	100	100	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219
MPI	100	99,9	100	100	Congenital disorder of glycosylation, type Ib, 602579
MPIG6B	100	100	100	100	?Thrombocytopenia, anemia, and myelofibrosis, 617441
MPL	100	99,5	100	100	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocytopenia, congenital amegakaryocytic, 604498 Thrombocythemia 2, 601977

MPLKIP	100	99,4	100	100	Trichothiodystrophy 4, nonphotosensitive, 234050
MPO	100	99,9	100	100	Myeloperoxidase deficiency, 254600 {Alzheimer disease, susceptibility to}, 104300 {Lung cancer, protection against, in smokers}, 0
MPV17	100	97,2	100	100	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MPZ	87,9	84,1	100	100	Charcot-Marie-Tooth disease, type 2J, 607736 Charcot-Marie-Tooth disease, type 1B, 118200 Dejerine-Sottas disease, 145900 Hypomyelinating neuropathy, congenital, 2, 618184 Charcot-Marie-Tooth disease, dominant intermediate D, 607791 Roussy-Levy syndrome, 180800 Charcot-Marie-Tooth disease, type 2I, 607677
MPZL2	100	99,9	100	100	Deafness, autosomal recessive 111, 618145
MRAP	100	100	100	100	Glucocorticoid deficiency 2, 607398
MRAS	100	99,6	100	100	Noonan syndrome 11, 618499
MRE11	98,9	93,3	100	100	Ataxia-telangiectasia-like disorder 1, 604391
MRM2	100	99,4	98,9	98,9	?Mitochondrial DNA depletion syndrome 17, 618567
MRPL12	100	98,2	100	100	?Combined oxidative phosphorylation deficiency 45, 618951
MRPL24	100	99,9	100	100	No OMIM disease ID
MRPL3	93,2	87,2	100	100	Combined oxidative phosphorylation deficiency 9, 614582
MRPL40	99,9	96,1	100	100	No OMIM disease ID
MRPL44	99,9	98,7	100	100	?Combined oxidative phosphorylation deficiency 16, 615395
MRPL57	100	99,8	100	100	No OMIM disease ID
MRPS14	100	100	100	100	?Combined oxidative phosphorylation deficiency 38, 618378
MRPS16	100	99,6	100	100	Combined oxidative phosphorylation deficiency 2, 610498
MRPS2	99,6	96,9	100	100	Combined oxidative phosphorylation deficiency 36, 617950
MRPS22	99,8	99,1	100	100	Combined oxidative phosphorylation deficiency 5, 611719 Ovarian dysgenesis 7, 618117
MRPS23	100	99,6	100	100	?Combined oxidative phosphorylation defiency 46, 618952

MRPS25	100	99,9	82,7	82,7	?Combined oxidative phosphorylation deficiency 50, 619025
MRPS28	89,9	86,8	86,6	86,6	?Combined oxidative phosphorylation deficiency 47, 618958
MRPS34	97,6	92	100	100	Combined oxidative phosphorylation deficiency 32, 617664
MRPS36	95,2	77,6	100	100	No OMIM disease ID
MRPS7	100	100	100	100	?Combined oxidative phosphorylation deficiency 34, 617872
MRRF	100	100	100	100	No OMIM disease ID
MRTFA	91,4	90,2	92,8	92,8	?Immunodeficiency 66, 618847
MS4A1	99,8	98,8	100	100	?Immunodeficiency, common variable, 5, 613495
MSH2	99	96,9	100	100	Mismatch repair cancer syndrome 2, 619096 Muir-Torre syndrome, 158320 Colorectal cancer, hereditary nonpolyposis, type 1, 120435
MSH3	98	97,3	98	98	Familial adenomatous polyposis 4, 617100 Endometrial carcinoma, somatic, 608089
MSH5	100	100	100	100	?Premature ovarian failure 13, 617442
MSH6	100	99,8	100	100	{Endometrial cancer, familial}, 608089 Mismatch repair cancer syndrome 3, 619097 Colorectal cancer, hereditary nonpolyposis, type 5, 614350
MSL3	84,5	77,4	96,8	96,6	Basilicata-Akhtar syndrome, 301032
MSMO1	96,3	88,9	100	100	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MSN	99	95,7	100	100	Immunodeficiency 50, 300988
MSR1	99,9	99,5	100	100	Barrett esophagus/esophageal adenocarcinoma, 614266
MSRB3	100	99,4	100	100	Deafness, autosomal recessive 74, 613718
MSTN	100	100	100	100	Muscle hypertrophy, 614160
MSTO1	99,6	96,8	100	100	Myopathy, mitochondrial, and ataxia, 617675
MSX1	96,9	89,3	100	100	Orofacial cleft 5, 608874 Ectodermal dysplasia 3, Witkop type, 189500 Tooth agenesis, selective, 1, with or without orofacial cleft, 106600
MSX2	100	99,4	100	100	Parietal foramina 1, 168500 Craniosynostosis 2, 604757 Parietal foramina with cleidocranial dysplasia, 168550

MTA1	99,9	98,3	100	100	No OMIM disease ID
MTAP	99,1	93,5	100	100	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250
MTFMT	100	99,8	100	100	Combined oxidative phosphorylation deficiency 15, 614947 Mitochondrial complex I deficiency, nuclear type 27, 618248
MTHFD1	100	99,5	100	100	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTHFR	97,3	96	100	100	{Schizophrenia, susceptibility to}, 181500 Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}, 0
MTHFS	75	74,9	100	100	Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367
MTM1	99	93,3	100	100	Myotubular myopathy, X-linked, 310400
MTMR2	100	99	100	100	Charcot-Marie-Tooth disease, type 4B1, 601382
MTO1	91,3	90,4	91,6	91,4	Combined oxidative phosphorylation deficiency 10, 614702
MTOR	100	99,5	100	100	Smith-Kingsmore syndrome, 616638 Focal cortical dysplasia, type II, somatic, 607341
MTPAP	99,5	96,1	100	100	?Spastic ataxia 4, autosomal recessive, 613672
MTR	100	100	100	100	{Neural tube defects, folate-sensitive, susceptibility to}, 601634 Homocystinuria-megaloblastic anemia, cblG complementation type, 250940
MTRR	100	99,6	100	100	{Neural tube defects, folate-sensitive, susceptibility to}, 601634 Homocystinuria-megaloblastic anemia, cbl E type, 236270
MTTP	100	99,6	100	100	Abetalipoproteinemia, 200100 {Metabolic syndrome, protection against}, 605552
MTX2	98,1	91,5	100	100	Mandibuloacral dysplasia progeroid syndrome, 619127
MUC1	92,5	83,6	100	99,9	Medullary cystic kidney disease 1, 174000
MUSK	100	99,9	100	100	Fetal akinesia deformation sequence 1, 208150 Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325
MUTYH	100	100	100	100	Gastric cancer, somatic, 613659 Adenomas, multiple colorectal, 608456
MVD	99,9	98,3	100	100	Porokeratosis 7, multiple types, 614714

MVK	90,9	90,5	90,5	90,5	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377
MXI1	98,6	95,3	97,7	94,1	Prostate cancer, somatic, 176807 Neurofibrosarcoma, somatic, 0
MYBPC1	99,9	99,5	100	100	Arthrogryposis, distal, type 1B, 614335 Myopathy, congenital, with tremor, 618524 Lethal congenital contracture syndrome 4, 614915
MYBPC3	99,9	97,6	100	100	Cardiomyopathy, hypertrophic, 4, 115197 Cardiomyopathy, dilated, 1MM, 615396 Left ventricular noncompaction 10, 615396
MYBPHL	99,9	98,6	100	100	No OMIM disease ID
MYC	65,9	64,4	100	100	Burkitt lymphoma, somatic, 113970
MYCN	100	99,9	99,3	96,7	Feingold syndrome 1, 164280
MYD88	100	100	100	100	Immunodeficiency 68, 612260 Macroglobulinemia, Waldenstrom, somatic, 153600
MYF5	100	100	100	100	Ophthalmoplegia, external, with rib and vertebral anomalies, 618155
MYH11	100	100	100	100	Aortic aneurysm, familial thoracic 4, 132900
MYH14	98,4	94	100	100	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369 Deafness, autosomal dominant 4A, 600652
MYH2	99,9	99,4	100	100	Proximal myopathy and ophthalmoplegia, 605637
MYH3	99,9	99	100	100	Contractures, pterygia, and spondylocarpotarsal fusion syndrome 1B, 618469 Arthrogryposis, distal, type 2B3 (Sheldon-Hall), 618436 Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700 Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A, 178110
MYH6	99,4	97,1	100	100	Atrial septal defect 3, 614089 Cardiomyopathy, hypertrophic, 14, 613251 {Sick sinus syndrome 3}, 614090 Cardiomyopathy, dilated, 1EE, 613252
MYH7	99,6	97,3	100	100	Myopathy, myosin storage, autosomal recessive, 255160 Left ventricular noncompaction 5, 613426 Laing distal myopathy, 160500 Myopathy, myosin storage, autosomal dominant, 608358 Cardiomyopathy, dilated, 1S, 613426

					Scapuloperoneal syndrome, myopathic type, 181430 Cardiomyopathy, hypertrophic, 1, 192600
MYH7B	98,4	94,2	100	100	No OMIM disease ID
MYH8	100	99,6	100	100	Carney complex variant, 608837 Trismus-pseudocamptodactyly syndrome, 158300
MYH9	100	99,3	100	100	Deafness, autosomal dominant 17, 603622 Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100
MYL1	100	99,7	100	100	Myopathy, congenital, with fast-twitch (type II) fiber atrophy, 618414
MYL2	96,1	88,1	99	95,4	Cardiomyopathy, hypertrophic, 10, 608758
MYL3	100	100	100	100	Cardiomyopathy, hypertrophic, 8, 608751
MYL4	100	100	100	100	?Atrial fibrillation, familial, 18, 617280
MYL7	100	100	100	100	No OMIM disease ID
MYLK	100	99,9	100	100	Aortic aneurysm, familial thoracic 7, 613780 Megacystis-microcolon-intestinal hypoperistalsis syndrome, 249210
MYLK2	100	100	100	100	Cardiomyopathy, hypertrophic, 1, digenic, 192600
MYLK3	99,4	97,8	100	100	No OMIM disease ID
MYLPF	100	100	100	100	Arthrogryposis, distal, type 1C, 619110
MYMK	100	100	100	100	Carey-Fineman-Ziter syndrome, 254940
MYO15A	98,8	97	100	99,9	Deafness, autosomal recessive 3, 600316
MYO18B	100	99,1	100	100	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549
MYO1A	100	99,8	100	100	No OMIM disease ID
MYO1E	99,9	99,5	100	100	Glomerulosclerosis, focal segmental, 6, 614131
MYO3A	99,6	96,6	100	100	Deafness, autosomal recessive 30, 607101
MYO5A	99,8	98,9	100	100	Griscelli syndrome, type 1, 214450
MYO5B	99,1	96,2	100	100	Microvillus inclusion disease, 251850
MYO6	99,5	96,6	100	100	Deafness, autosomal recessive 37, 607821 Deafness, autosomal dominant 22, 606346 Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346

MYO7A	99,3	97,3	100	100	Deafness, autosomal recessive 2, 600060 Deafness, autosomal dominant 11, 601317 Usher syndrome, type 1B, 276900
MYO9A	99,9	99,1	100	100	Myasthenic syndrome, congenital, 24, presynaptic, 618198
MYOC	100	98,6	100	100	Glaucoma 1A, primary open angle, 137750
MYOM1	99,9	98,4	100	100	No OMIM disease ID
MYORG	100	100	100	100	Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317
MYOT	100	99,6	100	100	Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
MYOZ2	100	100	100	100	Cardiomyopathy, hypertrophic, 16, 613838
MYPN	100	99,7	100	100	Cardiomyopathy, dilated, 1KK, 615248 Cardiomyopathy, familial restrictive, 4, 615248 Nemaline myopathy 11, autosomal recessive, 617336 Cardiomyopathy, hypertrophic, 22, 615248
MYRF	99,3	98,5	100	100	Cardiac-urogenital syndrome, 618280 Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113
MYSM1	96,4	95,5	96,4	96,4	Bone marrow failure syndrome 4, 618116
MYT1L	87	86,2	90,2	90,2	Mental retardation, autosomal dominant 39, 616521
MZB1	100	100	100	100	No OMIM disease ID
NAA10	99,7	98,5	99,9	99,9	Ogden syndrome, 300855 Microphthalmia, syndromic 1, 309800
NAA15	95,8	91	96,8	96,7	Mental retardation, autosomal dominant 50, 617787
NACC1	100	99,8	100	100	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393
NADK2	99,9	97,2	99	96,3	2,4-dienoyl-CoA reductase deficiency, 616034
NADSYN1	100	100	100	100	Vertebral, cardiac, renal, and limb defects syndrome 3, 618845
NAGA	100	100	100	100	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241
NAGLU	92,9	89,9	99,9	99,2	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491
NAGS	99,7	95	100	100	N-acetylglutamate synthase deficiency, 237310

NALCN	100	99,5	99,8	99,8	Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419 Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266
NANOS1	99,6	95,1	95,2	88,8	Spermatogenic failure 12, 615413
NANS	100	99,9	100	100	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NARS2	98,3	97,4	100	100	?Deafness, autosomal recessive 94, 618434 Combined oxidative phosphorylation deficiency 24, 616239
NAT8L	99,5	94,2	95,6	90,8	?N-acetylaspartate deficiency, 614063
NAXD	100	100	100	100	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321
NAXE	100	99,8	100	100	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NBAS	100	99,6	100	100	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NBEA	92	90,6	100	100	No OMIM disease ID
NBEAL2	99,4	99,4	100	100	Gray platelet syndrome, 139090
NBN	99,9	98,6	100	100	Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260 Aplastic anemia, 609135
NCAPD2	100	99,7	100	100	?Microcephaly 21, primary, autosomal recessive, 617983
NCAPD3	99,9	98,9	100	100	Microcephaly 22, primary, autosomal recessive, 617984
NCAPG2	99,9	99,2	100	100	Khan-Khan-Katsanis syndrome, 618460
NCAPH	100	100	100	100	?Microcephaly 23, primary, autosomal recessive, 617985
NCF1	26	25,8	100	99,8	Chronic granulomatous disease 1, autosomal recessive, 233700
NCF2	99,9	98,3	100	100	Chronic granulomatous disease 2, autosomal recessive, 233710
NCF4	100	100	100	100	Chronic granulomatous disease 3, autosomal recessive, 613960
NCKAP1L	100	99,9	100	100	Immunodeficiency 72 with autoinflammation, 618982
NCOA3	99,6	97,4	100	100	No OMIM disease ID
NCOA4	96,4	93	100	100	No OMIM disease ID
NCSTN	100	99,8	100	100	Acne inversa, familial, 1, 142690

NDE1	100	100	100	100	Lissencephaly 4 (with microcephaly), 614019 ?Microhydranencephaly, 605013
NDN	98,7	89,1	100	100	Prader-Willi syndrome, 176270
NDP	100	99,7	100	100	Norrie disease, 310600 Exudative vitreoretinopathy 2, X-linked, 305390
NDRG1	100	100	100	100	Charcot-Marie-Tooth disease, type 4D, 601455
NDST1	100	100	100	100	Mental retardation, autosomal recessive 46, 616116
NDUFA1	99,9	99,3	100	100	Mitochondrial complex I deficiency, nuclear type 12, 301020
NDUFA10	99,8	98,6	100	100	Mitochondrial complex I deficiency, nuclear type 22, 618243
NDUFA11	100	100	100	99,8	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFA12	100	100	100	100	?Mitochondrial complex I deficiency, nuclear type 23, 618244
NDUFA13	92,2	89,2	100	100	?Mitochondrial complex I deficiency, nuclear type 28, 618249 {Thyroid carcinoma, Hurthle cell}, 607464
NDUFA2	100	100	100	100	Mitochondrial complex I deficiency, nuclear type 13, 618235
NDUFA3	89,1	87,9	88	87,9	No OMIM disease ID
NDUFA4	100	96,4	100	100	?Mitochondrial complex IV deficiency, nuclear type 21, 619065
NDUFA5	96,5	81,8	100	100	No OMIM disease ID
NDUFA6	100	100	100	100	Mitochondrial complex I deficiency, nuclear type 33, 618253
NDUFA7	100	99,9	100	100	No OMIM disease ID
NDUFA8	100	99	100	100	No OMIM disease ID
NDUFA9	99,9	96,5	100	100	Mitochondrial complex I deficiency, nuclear type 26, 618247
NDUFAB1	99,6	94,9	100	100	No OMIM disease ID
NDUFAF1	100	100	100	100	Mitochondrial complex I deficiency, nuclear type 11, 618234
NDUFAF2	95	83,4	100	99,9	Mitochondrial complex I deficiency, nuclear type 10, 618233
NDUFAF3	100	99,9	100	100	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF4	99,8	98,2	100	100	Mitochondrial complex I deficiency, nuclear type 15, 618237
NDUFAF5	100	99,5	100	100	Mitochondrial complex I deficiency, nuclear type 16, 618238

NDUFAF6	100	96,8	100	100	Fanconi renotubular syndrome 5, 618913 Mitochondrial complex I deficiency, nuclear type 17, 618239
NDUFAF7	100	99,8	100	100	No OMIM disease ID
NDUFAF8	62,6	61,1	100	99,6	Mitochondrial complex I deficiency, nuclear type 34, 618776
NDUFB1	78,2	57,8	100	100	No OMIM disease ID
NDUFB10	100	100	100	100	?Mitochondrial complex I deficiency, nuclear type 35, 619003
NDUFB11	99,5	96,5	100	99,5	Linear skin defects with multiple congenital anomalies 3, 300952 ?Mitochondrial complex I deficiency, nuclear type 30, 301021
NDUFB2	100	100	100	100	No OMIM disease ID
NDUFB3	95,8	80,5	100	100	Mitochondrial complex I deficiency, nuclear type 25, 618246
NDUFB4	90,2	85,2	100	100	No OMIM disease ID
NDUFB5	100	100	100	100	No OMIM disease ID
NDUFB6	98,4	88,4	100	100	No OMIM disease ID
NDUFB7	99,8	94,8	100	100	No OMIM disease ID
NDUFB8	100	99,9	100	100	Mitochondrial complex I deficiency, nuclear type 32, 618252
NDUFB9	96,5	92,5	98,7	98,7	?Mitochondrial complex I deficiency, nuclear type 24, 618245
NDUFC1	99,6	99,4	100	100	No OMIM disease ID
NDUFC2	99,8	96	100	100	No OMIM disease ID
NDUFS1	100	99,5	100	100	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	100	100	100	100	Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	90,7	90,6	91,9	90,7	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	100	99,4	100	100	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS5	100	100	100	100	No OMIM disease ID
NDUFS6	100	99,9	100	100	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFS7	100	99,2	100	100	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFS8	100	99,4	100	100	Mitochondrial complex I deficiency, nuclear type 2, 618222

NDUFV1	98	96,1	100	100	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	86,9	76,9	100	100	Mitochondrial complex I deficiency, nuclear type 7, 618229
NDUFV3	99,8	98	100	100	No OMIM disease ID
NEB	83	82,6	99,9	99,9	Nemaline myopathy 2, autosomal recessive, 256030
NEBL	99,2	97,1	100	100	No OMIM disease ID
NECAP1	100	100	100	100	Developmental and epileptic encephalopathy 21, 615833
NECTIN1	100	99,9	100	100	Orofacial cleft 7, 225060 Cleft lip/palate-ectodermal dysplasia syndrome, 225060
NECTIN4	100	100	100	100	Ectodermal dysplasia-syndactyly syndrome 1, 613573
NEDD4L	72	71,5	100	100	Periventricular nodular heterotopia 7, 617201
NEFH	93,4	84,5	100	100	Charcot-Marie-Tooth disease, axonal, type 2CC, 616924 ?{Amyotrophic lateral sclerosis, susceptibility to}, 105400
NEFL	99,9	98,2	100	100	Charcot-Marie-Tooth disease, type 1F, 607734 Charcot-Marie-Tooth disease, dominant intermediate G, 617882 Charcot-Marie-Tooth disease, type 2E, 607684
NEK1	99,8	98	100	100	{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892 Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NEK11	99,9	98,8	100	100	No OMIM disease ID
NEK2	99,7	95,5	96,1	96,1	?Retinitis pigmentosa 67, 615565
NEK8	100	99,9	100	100	?Nephronophthisis 9, 613824 Renal-hepatic-pancreatic dysplasia 2, 615415
NEK9	100	99,6	100	100	Lethal congenital contracture syndrome 10, 617022 Nevus comedonicus, somatic, 617025 ?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262
NEPRO	100	99,7	100	100	Anauxetic dysplasia 3, 618853
NEU1	99,7	97,7	100	100	Sialidosis, type II, 256550 Sialidosis, type I, 256550
NEUROD1	100	99,1	100	100	Maturity-onset diabetes of the young 6, 606394 {Type 2 diabetes mellitus, susceptibility to}, 125853
NEUROD2	100	99,9	100	100	Developmental and epileptic encephalopathy 72, 618374
NEUROG3	100	100	100	100	Diarrhea 4, malabsorptive, congenital, 610370

NEXMIF	100	99,5	100	100	Mental retardation, X-linked 98, 300912
NEXN	92	77,5	100	99,9	Cardiomyopathy, hypertrophic, 20, 613876 Cardiomyopathy, dilated, 1CC, 613122
NF1	92,6	90,2	100	100	Neurofibromatosis-Noonan syndrome, 601321 Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Watson syndrome, 193520 Neurofibromatosis, type 1, 162200
NF2	100	99,9	100	100	Meningioma, NF2-related, somatic, 607174 Schwannomatosis, somatic, 162091 Neurofibromatosis, type 2, 101000
NFASC	100	99,9	100	100	Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356
NFAT5	99,8	99,1	100	100	No OMIM disease ID
NFE2L2	100	99,9	100	100	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744
NFIA	99,2	98,8	99,2	99,2	Brain malformations with or without urinary tract defects, 613735
NFIB	97,4	96,5	100	100	Macrocephaly, acquired, with impaired intellectual development, 618286
NFIX	100	99,5	99,6	98,7	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753
NFKB1	100	99,4	100	100	Immunodeficiency, common variable, 12, 616576
NFKB2	98,8	95,6	100	100	Immunodeficiency, common variable, 10, 615577
NFKBIA	95,2	88	100	100	Ectodermal dysplasia and immunodeficiency 2, 612132
NFS1	87,9	84	89,5	89,5	No OMIM disease ID
NFU1	98,8	90,8	100	100	Multiple mitochondrial dysfunctions syndrome 1, 605711
NGF	100	100	100	100	Neuropathy, hereditary sensory and autonomic, type V, 608654
NGLY1	100	99,8	100	100	Congenital disorder of deglycosylation, 615273
NHEJ1	100	96,2	100	100	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
NHLRC1	100	98,7	100	100	Epilepsy, progressive myoclonic 2B (Lafora), 254780
NHLRC2	99,6	98,2	100	100	FINCA syndrome, 618278
NHP2	100	100	100	100	Dyskeratosis congenita, autosomal recessive 2, 613987

NHS	95,4	93,9	100	99,8	Nance-Horan syndrome, 302350 Cataract 40, X-linked, 302200
NIN	100	99,5	99,1	99,1	?Seckel syndrome 7, 614851
NIPA1	100	100	99,8	98,5	Spastic paraplegia 6, autosomal dominant, 600363
NIPAL4	100	99,1	100	100	Ichthyosis, congenital, autosomal recessive 6, 612281
NIPBL	98,9	97	100	100	Cornelia de Lange syndrome 1, 122470
NKAP	99,3	95,2	100	100	Intellectual developmental disorder, X-linked, syndromic, Hackman-Di Donato type, 301039
NKX2-1	98,6	85,6	100	100	{Thyroid cancer, nonmedullary, 1}, 188550 Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978
NKX2-5	100	99,7	100	100	Ventricular septal defect 3, 614432 Tetralogy of Fallot, 187500 Hypoplastic left heart syndrome 2, 614435 Conotruncal heart malformations, variable, 217095 Hypothyroidism, congenital nongoitrous, 5, 225250 Atrial septal defect 7, with or without AV conduction defects, 108900
NKX2-6	100	99,5	100	100	Persistent truncus arteriosus, 217095 Conotruncal heart malformations, 217095
NKX3-2	99,8	97	100	100	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330
NKX6-2	89	81,8	100	100	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560
NLGN2	92,9	88,5	100	100	No OMIM disease ID
NLGN3	99,9	99	100	100	{Autism susceptibility, X-linked 1}, 300425 {Asperger syndrome susceptibility, X-linked 1}, 300494
NLGN4X	99,9	98,9	100	99,9	Mental retardation, X-linked, 300495 {Asperger syndrome susceptibility, X-linked 2}, 300497 {Autism susceptibility, X-linked 2}, 300495
NLRC4	100	100	100	100	Autoinflammation with infantile enterocolitis, 616050 ?Familial cold autoinflammatory syndrome 4, 616115
NLRP1	99,6	98	100	100	Palmoplantar carcinoma, multiple self-healing, 615225 Autoinflammation with arthritis and dyskeratosis, 617388 {Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579 ?Respiratory papillomatosis, juvenile recurrent, congenital, 618803
NLRP12	100	99,9	100	100	Familial cold autoinflammatory syndrome 2, 611762

NLRP3	100	99,9	100	100	Familial cold inflammatory syndrome 1, 120100 Muckle-Wells syndrome, 191900 CINCA syndrome, 607115 Deafness, autosomal dominant 34, with or without inflammation, 617772 Keratoendothelitis fugax hereditaria, 148200
NLRP6	98,7	95,9	100	100	No OMIM disease ID
NLRP7	100	99,6	100	100	Hydatidiform mole, recurrent, 1, 231090
NME1	100	100	100	100	No OMIM disease ID
NME3	93,1	88,5	100	100	No OMIM disease ID
NME5	100	100	100	100	No OMIM disease ID
NME8	99,2	95,3	100	100	Ciliary dyskinesia, primary, 6, 610852
NMNAT1	100	99,2	98,3	95,6	Leber congenital amaurosis 9, 608553
NMNAT2	99,9	98,9	100	100	No OMIM disease ID
NNT	96,4	95,9	96,4	96,4	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736
NOBOX	99,9	98,4	100	99,8	Premature ovarian failure 5, 611548
NOD2	100	99,9	100	100	{Yao syndrome}, 617321 Blau syndrome, 186580 {Inflammatory bowel disease 1, Crohn disease}, 266600
NODAL	100	100	100	100	Heterotaxy, visceral, 5, 270100
NOG	100	100	100	100	Tarsal-carpal coalition syndrome, 186570 Symphalangism, proximal, 1A, 185800 Stapes ankylosis with broad thumbs and toes, 184460 Multiple synostoses syndrome 1, 186500 Brachydactyly, type B2, 611377
NOL3	93,7	84,2	100	100	?Myoclonus, familial, 1, 614937
NOMO3	16,7	16,5	99,8	99,7	No OMIM disease ID
NONO	100	98,4	100	100	Mental retardation, X-linked, syndromic 34, 300967
NOP10	100	99,8	100	100	Dyskeratosis congenita, autosomal recessive 1, 224230
NOP56	99,8	98,6	100	100	Spinocerebellar ataxia 36, 614153

NOS1AP	100	99,9	100	100	No OMIM disease ID
NOS3	95,8	93,3	94,1	94	{Alzheimer disease, late-onset, susceptibility to}, 104300 {Hypertension, susceptibility to}, 145500 {Ischemic stroke, susceptibility to}, 601367 {Hypertension, pregnancy-induced}, 189800 {Coronary artery spasm 1, susceptibility to}, 0 {Placental abruption}, 0
NOTCH1	99,2	97,2	100	100	Aortic valve disease 1, 109730 Adams-Oliver syndrome 5, 616028
NOTCH2	100	99,5	100	100	Hajdu-Cheney syndrome, 102500 Alagille syndrome 2, 610205
NOTCH3	94	90,2	99,9	99,4	?Myofibromatosis, infantile 2, 615293 Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310 Lateral meningocele syndrome, 130720
NOTCH4	99,5	98,1	100	100	No OMIM disease ID
NOVA2	99	94,6	96,8	93,3	Neurodevelopmental disorder with or without autistic features and/or structural brain abnormalities, 618859
NPAT	99,8	98,7	100	100	No OMIM disease ID
NPC1	99,6	98,7	100	100	Niemann-Pick disease, type D, 257220 Niemann-Pick disease, type C1, 257220
NPC2	100	99,6	100	100	Niemann-pick disease, type C2, 607625
NPHP1	100	99	100	100	Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900 Joubert syndrome 4, 609583
NPHP3	99,7	98,4	100	100	Meckel syndrome 7, 267010 Renal-hepatic-pancreatic dysplasia 1, 208540 Nephronophthisis 3, 604387
NPHP4	100	99,8	100	100	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
NPHS1	99,8	99,1	100	100	Nephrotic syndrome, type 1, 256300
NPHS2	100	99,5	100	100	Nephrotic syndrome, type 2, 600995
NPL	100	99,9	100	100	No OMIM disease ID
NPM1	98,2	85,3	100	100	Leukemia, acute myeloid, somatic, 601626

NPPA	100	100	100	100	Atrial standstill 2, 615745 Atrial fibrillation, familial, 6, 612201
NPPB	100	100	100	100	No OMIM disease ID
NPPC	100	99	100	100	No OMIM disease ID
NPR2	100	99,6	100	100	Short stature with nonspecific skeletal abnormalities, 616255 Epiphyseal chondrodysplasia, Miura type, 615923 Acromesomelic dysplasia, Maroteaux type, 602875
NPR3	100	100	100	100	?Hypertension, salt-resistant, 0
NPRL2	100	100	100	100	Epilepsy, familial focal, with variable foci 2, 617116
NPRL3	100	99,6	100	100	Epilepsy, familial focal, with variable foci 3, 617118
NR0B1	100	99,5	100	100	Adrenal hypoplasia, congenital, 300200 46XY sex reversal 2, dosage-sensitive, 300018
NR0B2	100	99,3	100	100	Obesity, mild, early-onset, 601665
NR1H4	99,8	98,5	100	100	Cholestasis, progressive familial intrahepatic, 5, 617049
NR2E3	100	99,6	100	100	Enhanced S-cone syndrome, 268100 Retinitis pigmentosa 37, 611131
NR2F1	100	100	99,1	95,1	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NR2F2	100	98,5	100	100	Congenital heart defects, multiple types, 4, 615779 46,XX sex reversal 5, 618901
NR3C1	100	99,9	100	100	Glucocorticoid resistance, 615962
NR3C2	100	99,7	100	100	Pseudohypoaldosteronism type I, autosomal dominant, 177735 Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115
NR4A2	100	100	100	100	No OMIM disease ID
NR4A3	99,9	98	100	100	Chondrosarcoma, extraskeletal myxoid, 612237
NR5A1	100	100	100	100	Adrenocortical insufficiency, 612964 46, XX sex reversal 4, 617480 Premature ovarian failure 7, 612964 Spermatogenic failure 8, 613957 46XY sex reversal 3, 612965
NRAS	100	100	100	100	Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200

					Colorectal cancer, somatic, 114500 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Thyroid carcinoma, follicular, somatic, 188470 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224
NRIP1	100	100	100	100	?Congenital anomalies of kidney and urinary tract 3, 618270
NRL	99,5	94,8	100	100	Retinitis pigmentosa 27, 613750 Retinal degeneration, autosomal recessive, clumped pigment type, 0
NRROS	100	100	100	100	Seizures, early-onset, with neurodegeneration and brain calcification, 618875
NRXN1	97,4	96,9	100	99,8	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332
NSD1	100	99,9	100	100	Sotos syndrome 1, 117550
NSD2	99,9	99,2	100	100	No OMIM disease ID
NSDHL	100	98,7	100	100	CHILD syndrome, 308050 CK syndrome, 300831
NSMCE2	99,7	98,2	100	100	Seckel syndrome 10, 617253
NSMCE3	100	100	100	100	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241
NSMF	96,1	95,6	100	100	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
NSUN2	96	95,1	100	100	Mental retardation, autosomal recessive 5, 611091
NSUN3	100	100	100	100	Combined oxidative phosphorylation deficiency 48, 619012
NT5C2	98	96,5	100	100	Spastic paraplegia 45, autosomal recessive, 613162
NT5C3A	97,8	88,2	100	100	Anemia, hemolytic, due to UMPH1 deficiency, 266120
NT5E	100	99,9	100	100	Calcification of joints and arteries, 211800
NTF4	99,6	93	100	100	Glaucoma 1, open angle, 1O, 613100
NTHL1	100	99,8	100	100	Familial adenomatous polyposis 3, 616415
NTM	100	100	100	100	No OMIM disease ID
NTN1	100	99,7	100	100	Mirror movements 4, 618264
NTNG2	98,5	96,7	99,9	99	Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718
NTRK1	99,8	98,2	100	100	Insensitivity to pain, congenital, with anhidrosis, 256800

NTRK2	100	99,9	100	100	Obesity, hyperphagia, and developmental delay, 613886 Developmental and epileptic encephalopathy 58, 617830
NUBPL	99,7	98,4	100	100	Mitochondrial complex I deficiency, nuclear type 21, 618242
NUMA1	100	99,8	100	100	Leukemia, acute promyelocytic, somatic, 612376
NUP107	99,8	98,5	100	100	Galloway-Mowat syndrome 7, 618348 ?Ovarian dysgenesis 6, 618078 Nephrotic syndrome, type 11, 616730
NUP133	99,7	98,3	100	100	Nephrotic syndrome, type 18, 618177 ?Galloway-Mowat syndrome 8, 618349
NUP155	99,2	97,4	100	100	?Atrial fibrillation 15, 615770
NUP160	100	99,9	100	100	?Nephrotic syndrome, type 19, 618178
NUP188	100	99,6	100	100	Sandestig-Stefanova syndrome, 618804
NUP205	99,9	99,4	100	100	?Nephrotic syndrome, type 13, 616893
NUP214	100	99,7	100	100	Leukemia, acute myeloid, somatic, 601626 {Encephalopathy, acute, infection-induced, susceptibility to, 9}, 618426 Leukemia, T-cell acute lymphoblastic, somatic, 613065
NUP37	100	100	100	100	?Microcephaly 24, primary, autosomal recessive, 618179
NUP62	100	100	100	100	Striatonigral degeneration, infantile, 271930
NUP85	100	100	100	100	Nephrotic syndrome, type 17, 618176
NUP88	100	100	100	100	Fetal akinesia deformation sequence 4, 618393
NUP93	98	94,2	95,5	95,5	Nephrotic syndrome, type 12, 616892
NUS1	60	44,5	100	100	Mental retardation, autosomal dominant 55, with seizures, 617831 ?Congenital disorder of glycosylation, type 1aa, 617082
NUTM2B-AS1					?Oculopharyngeal myopathy with leukoencephalopathy 1, 618637
NXN	100	100	99,9	99,5	Robinow syndrome, autosomal recessive 2, 618529
NYX	96,3	94,1	99,7	98,8	Night blindness, congenital stationary (complete), 1A, X-linked, 310500
OAS1	100	100	100	100	No OMIM disease ID
OAT	85,2	76,3	100	100	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OBSCN	99,3	98,1	100	99,9	No OMIM disease ID

OBSL1	100	99,3	100	100	3-M syndrome 2, 612921
OCA2	99,9	98,7	100	100	[Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 Albinism, oculocutaneous, type II, 203200 Albinism, brown oculocutaneous, 203200
OCLN	100	100	100	100	Pseudo-TORCH syndrome 1, 251290
OCRL	99,9	98,6	100	99,9	Lowe syndrome, 309000 Dent disease 2, 300555
CCDC114	100	100	100	100	Ciliary dyskinesia, primary, 20, 615067
ARMC4	92,1	90	96,3	96,3	Ciliary dyskinesia, primary, 23, 615451
CCDC151	100	99,7	100	100	Ciliary dyskinesia, primary, 30, 616037
TTC25	100	100	100	100	Ciliary dyskinesia, primary, 35, 617092
ODAM	99,8	98,7	100	100	No OMIM disease ID
ODAPH	100	100	100	100	Amelogenesis imperfecta, type IIA4, 614832
ODC1	100	99,8	100	100	Bachmann-Bupp syndrome, 619075
OFD1	88	73,7	100	99,9	Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Simpson-Golabi-Behmel syndrome, type 2, 300209
OGDH	100	99,9	100	100	Alpha-ketoglutarate dehydrogenase deficiency, 203740
OGG1	100	99,8	100	100	Renal cell carcinoma, clear cell, somatic, 144700
OGT	99,9	99	100	100	Mental retardation, X-linked 106, 300997
OPA1	99,6	97,6	100	100	{Glaucoma, normal tension, susceptibility to}, 606657 Behr syndrome, 210000 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896
OPA3	100	99	100	100	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPCML	99,6	99,6	100	100	Ovarian cancer, somatic, 167000
OPHN1	99,5	97,6	99,9	98,8	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486

OPLAH	100	99,8	100	100	5-oxoprolinase deficiency, 260005
OPN1LW	67,2	60,6	98,3	98,1	Blue cone monochromacy, 303700 Colorblindness, protan, 303900
OPN1MW	66,3	58,5	98,9	97,5	Colorblindness, deutan, 303800 Blue cone monochromacy, 303700
OPN1SW	100	100	100	100	Colorblindness, tritan, 190900
OPTN	100	99,9	100	100	{Glaucoma, normal tension, susceptibility to}, 606657 Glaucoma 1, open angle, E, 137760 Amyotrophic lateral sclerosis 12, 613435
ORAI1	99,1	96,4	99,6	97,1	Myopathy, tubular aggregate, 2, 615883 Immunodeficiency 9, 612782
ORC1	100	99,4	100	100	Meier-Gorlin syndrome 1, 224690
ORC4	98,7	93,6	100	100	Meier-Gorlin syndrome 2, 613800
ORC6	100	99,9	100	100	Meier-Gorlin syndrome 3, 613803
OSBPL2	100	100	100	100	Deafness, autosomal dominant 67, 616340
OSGEP	100	99,4	100	100	Galloway-Mowat syndrome 3, 617729
OSMR	100	99,7	100	100	Amyloidosis, primary localized cutaneous, 1, 105250
OSTM1	98,6	94	100	100	Osteopetrosis, autosomal recessive 5, 259720
OTC	100	100	100	100	Ornithine transcarbamylase deficiency, 311250
OTOA	99,4	97,6	100	99,9	Deafness, autosomal recessive 22, 607039
OTOF	100	99,9	100	100	Auditory neuropathy, autosomal recessive, 1, 601071 Deafness, autosomal recessive 9, 601071
OTOG	99,4	98,6	100	100	Deafness, autosomal recessive 18B, 614945
OTOGL	99,5	97,4	100	100	Deafness, autosomal recessive 84B, 614944
OTUD6B	99,9	98,8	100	100	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452
OTULIN	92,6	86,5	99,2	95	Autoinflammation, panniculitis, and dermatosis syndrome, 617099
OTX2	100	99,7	100	100	Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125 Microphthalmia, syndromic 5, 610125
OVOL2	95,7	89,5	100	100	Corneal dystrophy, posterior polymorphous, 1, 122000

OXA1L	100	99,8	100	100	No OMIM disease ID
OXCT1	99,8	98,1	100	100	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050
OXR1	99,4	97	100	100	Cerebellar hypoplasia/atrophy, epilepsy, and global developmental delay, 213000
P2RX2	100	100	100	100	Deafness, autosomal dominant 41, 608224
P2RY12	100	100	100	100	Bleeding disorder, platelet-type, 8, 609821
P3H1	100	100	100	100	Osteogenesis imperfecta, type VIII, 610915
P3H2	99,8	98	100	100	Myopia, high, with cataract and vitreoretinal degeneration, 614292
P4HA2	100	99,2	100	100	Myopia 25, autosomal dominant, 617238
P4HB	94,6	94	100	100	Cole-Carpenter syndrome 1, 112240
P4HTM	99	97,4	100	99,4	Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493
PABPN1	66,3	56,9	100	99,1	Oculopharyngeal muscular dystrophy, 164300
PACS1	98,8	96,9	100	100	Schuurs-Hoeijmakers syndrome, 615009
PACS2	99,3	96,2	100	99,8	Developmental and epileptic encephalopathy 66, 618067
PADI3	100	100	100	100	Uncombable hair syndrome, 191480
PADI6	100	99,6	100	100	Preimplantation embryonic lethality 2, 617234
PAFAH1B1	94,1	87,1	100	100	Subcortical laminar heterotopia, 607432 Lissencephaly 1, 607432
PAH	100	100	100	100	[Hyperphenylalaninemia, non-PKU mild], 261600 Phenylketonuria, 261600
PAK1	100	99,6	100	100	Intellectual developmental disorder with macrocephaly, seizures, and speech delay, 618158
PAK3	99,3	95,9	100	99,8	Mental retardation, X-linked 30/47, 300558
PALB2	100	100	100	100	{Pancreatic cancer, susceptibility to, 3}, 613348 Fanconi anemia, complementation group N, 610832 {Breast cancer, susceptibility to}, 114480
PAM16	65,3	65,2	82,9	82,9	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320
PANK2	100	99,3	100	100	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200

PANX1	100	100	100	100	Oocyte maturation defect 7, 618550
PAPPA2	100	99,9	100	100	No OMIM disease ID
PAPSS2	100	99,5	100	100	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847
PARK7	100	100	100	100	Parkinson disease 7, autosomal recessive early-onset, 606324
PARN	81,2	81,1	88,1	87,6	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371 Dyskeratosis congenita, autosomal recessive 6, 616353
PARS2	100	100	100	100	Developmental and epileptic encephalopathy 75, 618437
PATL2	100	99	100	100	Oocyte maturation defect 4, 617743
PAX1	92,4	87,9	100	99,6	Otofaciocervical syndrome 2, 615560
PAX2	100	99,9	100	100	Papillorenal syndrome, 120330 Glomerulosclerosis, focal segmental, 7, 616002
PAX3	100	99,9	100	100	Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220 Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820
PAX4	100	99,8	100	100	Maturity-onset diabetes of the young, type IX, 612225 Diabetes mellitus, type 2, 125853 {Diabetes mellitus, ketosis-prone, susceptibility to}, 612227
PAX5	98,7	96,1	100	100	{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545
PAX6	100	100	100	100	Optic nerve hypoplasia, 165550 ?Coloboma, ocular, 120200 Foveal hypoplasia 1, 136520 Aniridia, 106210 Keratitis, 148190 Anterior segment dysgenesis 5, multiple subtypes, 604229 Cataract with late-onset corneal dystrophy, 106210 ?Coloboma of optic nerve, 120430 ?Morning glory disc anomaly, 120430
PAX7	100	100	100	100	Myopathy, congenital, progressive, with scoliosis, 618578 Rhabdomyosarcoma 2, alveolar, 268220
PAX8	100	99,8	100	100	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700
PAX9	99,7	99,6	100	100	Tooth agenesis, selective, 3, 604625

PBX1	100	99,4	100	100	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PC	99,8	97,3	100	100	Pyruvate carboxylase deficiency, 266150
PCARE	99,6	98,5	100	100	Retinitis pigmentosa 54, 613428
PCBD1	100	99,6	100	99,7	Hyperphenylalaninemia, BH4-deficient, D, 264070
PCCA	99,5	96,7	100	100	Propionicacidemia, 606054
PCCB	97,9	96	98,7	96,2	Propionicacidemia, 606054
PCDH12	100	100	100	100	Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280
PCDH15	97,8	96,7	100	100	Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1F, 602083 Deafness, autosomal recessive 23, 609533
PCDH19	100	98,9	100	100	Developmental and epileptic encephalopathy 9, 300088
PCGF2	100	99,5	100	100	Turnpenny-Fry syndrome, 618371
PCIF1	100	100	100	100	No OMIM disease ID
PCK1	100	100	100	100	?Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680
PCK2	100	100	100	100	PEPCK deficiency, mitochondrial, 261650
PCLO	99,7	98,7	100	100	?Pontocerebellar hypoplasia, type 3, 608027
PCNA	100	98,4	100	100	?Ataxia-telangiectasia-like disorder 2, 615919
PCNT	99,6	97,1	100	100	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PCSK1	100	99,5	100	100	{Obesity, susceptibility to, BMIQ12}, 612362 Obesity with impaired prohormone processing, 600955
PCSK9	95	91,9	100	100	Hypercholesterolemia, familial, 3, 603776 {Low density lipoprotein cholesterol level QTL 1}, 603776
PCYT1A	98,9	95,5	100	100	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PCYT2	99,8	97,1	100	98,8	Spastic paraplegia 82, autosomal recessive, 618770
PDCD10	99,9	98,9	100	100	Cerebral cavernous malformations 3, 603285
PDE10A	65,5	64,5	86,9	84,1	Dyskinesia, limb and orofacial, infantile-onset, 616921 Striatal degeneration, autosomal dominant, 616922

PDE11A	99,9	99,7	100	100	Pigmented nodular adrenocortical disease, primary, 2, 610475
PDE1C	100	99,6	100	100	?Deafness, autosomal dominant 74, 618140
PDE2A	100	99,9	100	100	No OMIM disease ID
PDE3A	99,9	99,4	100	100	Hypertension and brachydactyly syndrome, 112410
PDE4D	95,7	93,5	100	99,8	Acrodysostosis 2, with or without hormone resistance, 614613
PDE6A	100	99,6	100	100	Retinitis pigmentosa 43, 613810
PDE6B	100	99,9	100	100	Retinitis pigmentosa-40, 613801 Night blindness, congenital stationary, autosomal dominant 2, 163500
PDE6C	99,9	97,8	100	100	Cone dystrophy 4, 613093
PDE6D	100	100	100	100	Joubert syndrome 22, 615665
PDE6G	100	100	100	100	Retinitis pigmentosa 57, 613582
PDE6H	100	97,9	100	100	Retinal cone dystrophy 3, 610024 Achromatopsia 6, 610024
PDE8B	99,9	99,7	100	100	Striatal degeneration, autosomal dominant, 609161 Pigmented nodular adrenocortical disease, primary, 3, 614190
PDGFB	100	99,3	100	100	Dermatofibrosarcoma protuberans, 607907 Basal ganglia calcification, idiopathic, 5, 615483 Meningioma, SIS-related, 607174
PDGFRA	100	100	100	100	Gastrointestinal stromal tumor/GIST-plus syndrome, somatic or familial, 175510 Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685
PDGFRB	99,2	97,5	100	100	Myeloproliferative disorder with eosinophilia, 131440 Basal ganglia calcification, idiopathic, 4, 615007 Kosaki overgrowth syndrome, 616592 Premature aging syndrome, Penttinen type, 601812 Myofibromatosis, infantile, 1, 228550
PDGFRL	100	100	100	100	Hepatocellular cancer, somatic, 114550 Colorectal cancer, somatic, 114500
PDHA1	99,4	97,1	100	100	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	99,1	97,5	100	100	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	99,9	99,4	100	100	Lacticacidemia due to PDX1 deficiency, 245349
PDK1	99,9	98,5	100	99,9	No OMIM disease ID

PDK2	100	100	100	100	No OMIM disease ID
PDK3	99,5	97,2	100	100	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905
PDK4	100	99,3	100	100	No OMIM disease ID
PDLIM3	100	99,7	100	100	No OMIM disease ID
PDLIM5	93,5	91,3	97,5	95,2	No OMIM disease ID
PDP1	100	100	100	100	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	94,7	87,6	97,3	96,6	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	99,8	97,1	100	100	Coenzyme Q10 deficiency, primary, 3, 614652
PDX1	93	82,4	100	100	{Diabetes mellitus, type II, susceptibility to}, 125853 Pancreatic agenesis 1, 260370 MODY, type IV, 606392
PDXK	79,3	76,6	99,4	96,7	Neuropathy, hereditary motor and sensory, type VIC, with optic atrophy, 618511
PDYN	100	100	100	100	Spinocerebellar ataxia 23, 610245
PDZD7	97	93	100	99,8	Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472 Deafness, autosomal recessive 57, 618003 {Retinal disease in Usher syndrome type IIA, modifier of}, 276901
PEPD	100	98,8	100	100	Prolidase deficiency, 170100
PER2	100	99,9	100	100	?Advanced sleep phase syndrome, familial, 1, 604348
PER3	99,9	98,8	100	100	?Advanced sleep phase syndrome, familial, 3, 616882
PERCC1	1,8	0	100	100	Diarrhea 11, malabsorptive, congenital, 618662
PERP	100	100	100	100	No OMIM disease ID
PET100	100	99,6	100	100	Mitochondrial complex IV deficiency, nuclear type 12, 619055
PET117	100	100	100	100	?Mitochondrial complex IV deficiency, nuclear type 19, 619063
PEX1	99,9	99,4	100	100	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	96,8	89,7	100	99,9	Peroxisome biogenesis disorder 6B, 614871 Peroxisome biogenesis disorder 6A (Zellweger), 614870
PEX11B	100	99,6	100	100	Peroxisome biogenesis disorder 14B, 614920

PEX12	100	100	100	100	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	100	100	100	100	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	96,7	90,8	100	100	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	97,9	94,2	100	100	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	99,9	98,5	100	100	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	100	100	100	100	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	100	100	100	100	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	100	99,3	100	100	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370
PEX5	99,9	99	100	100	Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716 Peroxisome biogenesis disorder 2A (Zellweger), 214110
PEX6	94,5	86,7	100	100	Peroxisome biogenesis disorder 4B, 614863 Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862
PEX7	87,8	80,7	91,3	91,3	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PFKM	100	99,5	100	100	Glycogen storage disease VII, 232800
PFN1	100	100	100	100	Amyotrophic lateral sclerosis 18, 614808
PGAM2	100	100	100	100	Glycogen storage disease X, 261670
PGAP1	99	94,4	100	100	Mental retardation, autosomal recessive 42, 615802
PGAP2	100	99,9	100	100	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGAP3	63,5	59,6	100	100	Hyperphosphatasia with mental retardation syndrome 4, 615716
PGK1	92,8	79,3	100	100	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	94,2	94,2	94,2	94,2	Congenital disorder of glycosylation, type It, 614921
PGM3	100	99,8	91,7	91,7	Immunodeficiency 23, 615816
PHACTR1	100	99,7	100	100	Developmental and epileptic encephalopathy 70, 618298

PHC1	99,9	99,2	100	100	?Microcephaly 11, primary, autosomal recessive, 615414
PHEX	100	99,6	99,9	99,2	Hypophosphatemic rickets, X-linked dominant, 307800
PHF21A	100	99,9	100	100	Intellectual developmental disorder with behavioral abnormalities and craniofacial dysmorphism with or without seizures, 618725
PHF6	97,8	88,3	99,9	98,9	Borjeson-Forssman-Lehmann syndrome, 301900
PHF8	99,7	96,8	100	100	Mental retardation syndrome, X-linked, Siderius type, 300263
PHGDH	99,9	98,8	100	100	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHIP	98,6	96,1	100	99,7	Chung-Jansen syndrome, 617991
PHKA1	99,2	95,3	100	99,9	Muscle glycogenosis, 300559
PHKA2	100	99,7	100	99,6	Glycogen storage disease, type IXa2, 306000 Glycogen storage disease, type IXa1, 306000
PHKB	99,9	99,2	100	100	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750
PHKG1	99,9	97,8	100	100	No OMIM disease ID
PHKG2	100	99,9	100	100	Glycogen storage disease IXc, 613027 Cirrhosis due to liver phosphorylase kinase deficiency, 0
PHOX2A	91,6	72,7	100	99,8	Fibrosis of extraocular muscles, congenital, 2, 602078
PHOX2B	100	99,7	99,5	97,8	Neuroblastoma with Hirschsprung disease, 613013 Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, 209880 {Neuroblastoma, susceptibility to, 2}, 613013
PHYH	100	99,6	100	100	Refsum disease, 266500
PI4K2A	91,9	86,4	100	100	No OMIM disease ID
PI4KA	92,6	88,8	99,9	99,9	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531
PIBF1	99,5	96,2	100	100	Joubert syndrome 33, 617767
PICALM	99,5	96,2	100	100	Leukemia, acute myeloid, somatic, 601626
PIEZO1	99,9	98,8	100	100	Lymphatic malformation 6, 616843 Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380
PIEZO2	100	99,5	100	100	Arthrogryposis, distal, with impaired proprioception and touch, 617146 Arthrogryposis, distal, type 5, 108145 ?Marden-Walker syndrome, 248700 Arthrogryposis, distal, type 3, 114300

PIGA	93,8	86,7	100	100	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGB	99,9	97,8	100	100	Developmental and epileptic encephalopathy 80, 618580
PIGC	99,2	90,9	100	100	Glycosylphosphatidylinositol biosynthesis defect 16, 617816
PIGG	100	99,7	100	100	Mental retardation, autosomal recessive 53, 616917
PIGH	82,1	68,1	75,2	74,4	Glycosylphosphatidylinositol biosynthesis defect 17, 618010
PIGK	99,2	95,1	100	100	Neurodevelopmental disorder with hypotonia and cerebellar atrophy, with or without seizures, 618879
PIGL	100	100	100	100	CHIME syndrome, 280000
PIGM	100	100	100	100	Glycosylphosphatidylinositol deficiency, 610293
PIGN	93,8	91,5	98,8	98,8	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	100	99,9	100	100	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGP	95,8	87,3	100	100	Developmental and epileptic encephalopathy 55, 617599
PIGQ	92,8	90,8	100	100	Developmental and epileptic encephalopathy 77, 618548
PIGS	100	100	100	100	Glycosylphosphatidylinositol biosynthesis defect 18, 618143
PIGT	98,1	98,1	100	100	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 ?Paroxysmal nocturnal hemoglobinuria 2, 615399
PIGU	100	99,1	100	100	Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis, 618590
PIGV	100	100	100	100	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIGW	100	99,8	100	100	Glycosylphosphatidylinositol biosynthesis defect 11, 616025
PIGY	100	99,9	100	100	Hyperphosphatasia with mental retardation syndrome 6, 616809
PIK3C2A	99,2	96,9	100	100	Oculoskeletodental syndrome, 618440
PIK3C3	99,3	98,9	99,8	99,7	No OMIM disease ID
PIK3CA	98	97,8	100	100	Ovarian cancer, somatic, 167000 Colorectal cancer, somatic, 114500 CLAPO syndrome, somatic, 613089 Cowden syndrome 5, 615108 Hepatocellular carcinoma, somatic, 114550 Breast cancer, somatic, 114480 Macrodactyly, somatic, 155500

					Keratosis, seborrheic, somatic, 182000 Gastric cancer, somatic, 613659 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Nevus, epidermal, somatic, 162900 CLOVE syndrome, somatic, 612918 Nonsmall cell lung cancer, somatic, 211980
PIK3CD	98,8	96,9	100	100	Immunodeficiency 14, 615513
PIK3CG	100	100	100	100	No OMIM disease ID
PIK3R1	99,8	99	100	100	?Agammaglobulinemia 7, autosomal recessive, 615214 SHORT syndrome, 269880 Immunodeficiency 36, 616005
PIK3R2	90,7	89,6	99,3	96,1	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PIK3R5	100	99,9	100	100	Ataxia-oculomotor apraxia 3, 615217
PIKFYVE	99,9	99,4	100	100	Corneal fleck dystrophy, 121850
PINK1	90,7	86,9	99,9	99,4	Parkinson disease 6, early onset, 605909
PIP5K1C	98	95,8	99,9	99,8	Lethal congenital contractural syndrome 3, 611369
PISD	100	100	100	100	Liberfarb syndrome, 618889
PITPNM3	99,5	98,7	100	100	Cone-rod dystrophy 5, 600977
PITRM1	98,4	96,1	100	100	No OMIM disease ID
PITX1	96,7	92	100	100	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800 Liebenberg syndrome, 186550
PITX2	99,9	97,7	100	100	Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550 Anterior segment dysgenesis 4, 137600
PITX3	100	98,4	100	100	Anterior segment dysgenesis 1, multiple subtypes, 107250 Cataract 11, syndromic, autosomal recessive, 610623 Cataract 11, multiple types, 610623
PJVK	100	99,7	100	100	Deafness, autosomal recessive 59, 610220
PKD1	39,2	30	99,2	98,9	Polycystic kidney disease 1, 173900
PKD1L1	100	99,8	100	100	Heterotaxy, visceral, 8, autosomal, 617205
PKD2	95,5	91,1	99,3	97,7	Polycystic kidney disease 2, 613095

PKDCC	90,6	81,5	97,8	94,7	Rhizomelic limb shortening with dysmorphic features, 618821
PKHD1	100	99,6	100	100	Polycystic kidney disease 4, with or without hepatic disease, 263200
PKLR	100	99,2	100	100	Pyruvate kinase deficiency, 266200 Adenosine triphosphate, elevated, of erythrocytes, 102900
PKP1	100	99,1	100	100	Ectodermal dysplasia/skin fragility syndrome, 604536
PKP2	95,4	88,6	95	95	Arrhythmogenic right ventricular dysplasia 9, 609040
PKP4	99,8	98,2	100	100	No OMIM disease ID
PLA2G4A	99,9	99,4	100	100	Gastrointestinal ulceration, recurrent, with dysfunctional platelets, 618372
PLA2G5	100	100	100	100	[Fleck retina, familial benign], 228980
PLA2G6	92,2	90,7	92,3	92,3	Infantile neuroaxonal dystrophy 1, 256600 Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217
PLA2G7	99,9	99	100	100	{Asthma, susceptibility to}, 600807 Platelet-activating factor acetylhydrolase deficiency, 614278 {Atopy, susceptibility to}, 147050
PLAA	100	99,2	100	100	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527
PLAG1	100	100	100	100	Silver-Russell syndrome 4, 618907 Adenomas, salivary gland pleomorphic, somatic, 181030
PLAT	100	99,1	100	100	Hyperfibrinolysis, familial, due to increased release of PLAT, 612348 Thrombophilia, familial, due to decreased release of PLAT, 612348
PLAU	100	99,8	100	100	Quebec platelet disorder, 601709 {Alzheimer disease, late-onset, susceptibility to}, 104300
PLAUR	100	99,8	100	100	No OMIM disease ID
PLCB1	100	99,8	100	100	Developmental and epileptic encephalopathy 12, 613722
PLCB3	100	99	100	100	Spondylometaphyseal dysplasia with corneal dystrophy, 618961
PLCB4	99,9	98,8	100	100	Auriculocondylar syndrome 2, 614669
PLCD1	99,9	97,8	100	100	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCE1	99,9	99,3	100	100	Nephrotic syndrome, type 3, 610725
PLCG2	100	99,8	100	100	Familial cold autoinflammatory syndrome 3, 614468 Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878

PLCZ1	99,9	96,9	100	100	Spermatogenic failure 17, 617214
PLD1	100	99,6	100	100	Cardiac valvular defect, developmental, 212093
PLD3	99,9	99,1	100	100	?Spinocerebellar ataxia 46, 617770
PLEC	100	99,8	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723 Epidermolysis bullosa simplex with pyloric atresia, 612138 Epidermolysis bullosa simplex with muscular dystrophy, 226670 ?Epidermolysis bullosa simplex with nail dystrophy, 616487 Epidermolysis bullosa simplex, Ogna type, 131950
PLEKHG2	100	99,3	100	100	Leukodystrophy and acquired microcephaly with or without dystonia, 616763
PLEKHG5	95,3	91,1	96,3	96,2	Spinal muscular atrophy, distal, autosomal recessive, 4, 611067 Charcot-Marie-Tooth disease, recessive intermediate C, 615376
PLEKHM1	100	99,8	100	100	Osteopetrosis, autosomal dominant 3, 618107 ?Osteopetrosis, autosomal recessive 6, 611497
PLEKHM2	100	100	100	100	No OMIM disease ID
PLG	87,8	87,5	100	100	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PLIN1	99,6	94,9	100	99,5	Lipodystrophy, familial partial, type 4, 613877
PLK4	99,9	98,2	100	100	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLN	100	100	100	100	Cardiomyopathy, hypertrophic, 18, 613874 Cardiomyopathy, dilated, 1P, 609909
PLOD1	100	98,4	100	100	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD2	99,3	97,3	100	100	Bruck syndrome 2, 609220
PLOD3	99,8	98	100	100	Lysyl hydroxylase 3 deficiency, 612394
PLP1	100	99,2	100	100	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PLPBP	98,2	90,1	100	100	Epilepsy, early-onset, vitamin B6-dependent, 617290
PLPP6	99,2	93,7	100	100	No OMIM disease ID
PLS1	100	99,1	100	100	Deafness, autosomal dominant 76, 618787
PLS3	97,7	96,1	97,2	97,2	Bone mineral density QTL18, osteoporosis, 300910
PLVAP	100	100	100	100	Diarrhea 10, protein-losing enteropathy type, 618183

PLXNA1	100	99,6	100	100	No OMIM disease ID
PLXND1	98,9	96,2	99,7	99,4	No OMIM disease ID
PMEPA1	100	99,2	100	99,9	No OMIM disease ID
PMFBP1	99,9	99,3	100	100	Spermatogenic failure 31, 618112
PML	100	99,8	100	100	Leukemia, acute promyelocytic, PML/RARA type, 0
PMM2	100	100	100	100	Congenital disorder of glycosylation, type Ia, 212065
PMP2	100	100	100	100	Charcot-Marie-Tooth disease, demyelinating, type 1G, 618279
PMP22	100	100	100	100	Dejerine-Sottas disease, 145900 ?Neuropathy, inflammatory demyelinating, 139393 Charcot-Marie-Tooth disease, type 1E, 118300 Roussy-Levy syndrome, 180800 Neuropathy, recurrent, with pressure palsies, 162500 Charcot-Marie-Tooth disease, type 1A, 118220
PMPCA	97,7	94,2	100	100	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMPCB	100	99,7	100	100	Multiple mitochondrial dysfunctions syndrome 6, 617954
PMS2	84,3	82,8	100	100	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 Mismatch repair cancer syndrome 4, 619101
PMS2CL					No OMIM disease ID
PMVK	100	100	100	100	Porokeratosis 1, multiple types, 175800
PNKD	100	99,9	100	100	Paroxysmal nonkinesigenic dyskinesia 1, 118800
PNKP	100	100	100	100	Microcephaly, seizures, and developmental delay, 613402 Ataxia-oculomotor apraxia 4, 616267 ?Charcot-Marie-Tooth disease, type 2B2, 605589
PNLIP	100	99,8	100	100	?Pancreatic lipase deficiency, 614338
PNMT	99,6	96,7	100	100	No OMIM disease ID
PNP	99,8	98,9	100	100	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA1	100	100	100	100	Ichthyosis, congenital, autosomal recessive 10, 615024
PNPLA2	99,7	96,1	100	100	Neutral lipid storage disease with myopathy, 610717

PNPLA6	100	99,7	100	100	Spastic paraplegia 39, autosomal recessive, 612020 Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 ?Laurence-Moon syndrome, 245800
PNPLA8	100	99,8	100	100	?Mitochondrial myopathy with lactic acidosis, 251950
PNPO	99,9	97,7	100	100	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
PNPT1	97,7	89,7	100	100	Deafness, autosomal recessive 70, 614934 Combined oxidative phosphorylation deficiency 13, 614932
POC1A	100	100	100	100	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POC1B	99,8	98,8	100	100	Cone-rod dystrophy 20, 615973
POC5	99,7	97,6	100	100	No OMIM disease ID
POF1B	95,6	86,9	100	99,8	?Premature ovarian failure 2B, 300604
POFUT1	100	99	100	100	Dowling-Degos disease 2, 615327
POGLUT1	99,4	94,6	100	100	?Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232 Dowling-Degos disease 4, 615696
POGZ	99,4	99	100	100	White-Sutton syndrome, 616364
POLA1	99,3	95,4	100	99,9	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 Van Esch-O'Driscoll syndrome, 301030
POLD1	98,5	95,2	100	100	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381 {Colorectal cancer, susceptibility to, 10}, 612591
POLE	100	99,8	100	100	FILS syndrome, 615139 IMAGE-I syndrome, 618336 {Colorectal cancer, susceptibility to, 12}, 615083
POLE2	97,3	89,8	100	100	No OMIM disease ID
POLG	100	99,3	100	100	Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
POLG2	99,6	98	100	99,9	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131 Mitochondrial DNA depletion syndrome 16 (hepatic type), 618528
POLH	100	99,6	100	100	Xeroderma pigmentosum, variant type, 278750

POLL	100	99,2	100	100	No OMIM disease ID
POLR1A	100	99,4	100	100	Acrofacial dysostosis, Cincinnati type, 616462
POLR1C	90,5	87	82,8	82,8	Treacher Collins syndrome 3, 248390 Leukodystrophy, hypomyelinating, 11, 616494
POLR1D	91,6	91,6	100	100	Treacher Collins syndrome 2, 613717
POLR2A	100	100	100	100	Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603
POLR3A	100	99,7	100	100	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 Wiedemann-Rautenstrauch syndrome, 264090
POLR3B	99,9	98,6	100	100	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMC	100	100	100	100	{Obesity, early-onset, susceptibility to}, 601665 Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734
POMGNT1	100	99,9	100	100	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 Retinitis pigmentosa 76, 617123 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280
POMGNT2	100	100	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830 Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135
POMK	100	100	100	100	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249
POMP	100	99,1	100	100	Proteasome-associated autoinflammatory syndrome 2, 618048 Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952
POMT1	99,3	97,5	100	100	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155
POMT2	99,4	96,4	100	100	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 mental retardation), type B, 2, 613156
POP1	100	99,7	100	100	Anauxetic dysplasia 2, 617396
POR	99,8	98,6	100	100	Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571 Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750
PORCN	100	99,1	100	100	Focal dermal hypoplasia, 305600
POT1	99,9	99	100	100	{Glioma susceptibility 9}, 616568 {Melanoma, cutaneous malignant, susceptibility to, 10}, 615848

POU1F1	100	99,2	100	100	Pituitary hormone deficiency, combined, 1, 613038
POU2AF1	100	99,3	100	100	No OMIM disease ID
POU3F3	73,2	59,6	94,9	83,8	Snijders Blok-Fisher syndrome, 618604
POU3F4	100	100	100	100	Deafness, X-linked 2, 304400
POU4F3	100	100	100	100	Deafness, autosomal dominant 15, 602459
POU6F2	95,2	95,2	100	100	{Wilms tumor susceptibility-5}, 601583
PPA2	98,7	94	100	100	?Sudden cardiac failure, alcohol-induced, 617223 Sudden cardiac failure, infantile, 617222
PPARG	100	100	98,3	98,3	Carotid intimal medial thickness 1, 609338 {Diabetes, type 2}, 125853 Insulin resistance, severe, digenic, 604367 Obesity, severe, 601665 Lipodystrophy, familial partial, type 3, 604367 [Obesity, resistance to], 0
PPARGC1A	100	99,9	100	100	No OMIM disease ID
PPCS	99,8	99,5	100	100	Cardiomyopathy, dilated, 2C, 618189
PPIB	100	100	100	100	Osteogenesis imperfecta, type IX, 259440
PPIP5K2	98,9	95,2	100	100	Deafness, autosomal recessive 100, 618422
PPM1D	100	99,9	100	100	Breast cancer, somatic, 114480 Jansen de Vries syndrome, 617450
PPM1K	100	100	100	100	?Maple syrup urine disease, mild variant, 615135
PPOX	99,7	96,8	100	100	Porphyria variegata, 176200
PPP1CB	99,9	99,3	100	100	Noonan syndrome-like disorder with loose anagen hair 2, 617506
PPP1R12A	97,7	95,3	100	100	Genitourinary and/or/brain malformation syndrome, 618820
PPP1R15B	100	99,6	100	100	Microcephaly, short stature, and impaired glucose metabolism 2, 616817
PPP1R21	99,6	96	100	100	No OMIM disease ID
PPP1R3A	99,7	99,2	100	100	Insulin resistance, severe, digenic, 125853
PPP2CA	100	100	100	100	Neurodevelopmental disorder and language delay with or without structural brain abnormalities, 618354

PPP2R1A	91,6	91,5	93,6	93,6	Mental retardation, autosomal dominant 36, 616362
PPP2R1B	100	99,9	100	100	Lung cancer, somatic, 211980
PPP2R2B	100	99,4	100	100	Spinocerebellar ataxia 12, 604326
PPP2R3C	99,6	94,9	100	100	Gonadal dysgenesis, dysmorphic facies, retinal dystrophy, and myopathy, 618419 Spermatogenic failure 36, 618420
PPP2R5B	100	100	100	100	No OMIM disease ID
PPP2R5C	97,7	93,1	100	100	No OMIM disease ID
PPP2R5D	100	100	100	100	Mental retardation, autosomal dominant 35, 616355
PPP3CA	99,8	98,4	100	100	Epileptic encephalopathy, infantile or early childhood, 1, 617711 Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265
PPT1	90,3	90,3	82,5	82,5	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	100	100	100	100	Renpenning syndrome, 309500
PRCC	99,9	98,5	100	100	Renal cell carcinoma, papillary, 605074
PRCD	100	100	100	100	Retinitis pigmentosa 36, 610599
PRDM12	90,8	88	93,4	91,7	Neuropathy, hereditary sensory and autonomic, type VIII, 616488
PRDM13	99,2	94,1	100	100	No OMIM disease ID
PRDM16	99,8	99,1	100	100	Left ventricular noncompaction 8, 615373 Cardiomyopathy, dilated, 1LL, 615373
PRDM5	99,9	99,2	100	100	Brittle cornea syndrome 2, 614170
PRDM6	95,8	87,8	100	100	Patent ductus arteriosus 3, 617039
PRDM8	92,9	88,6	100	99,8	?Epilepsy, progressive myoclonic, 10, 616640
PRDX1	100	100	100	100	Methylmalonic aciduria and homocystinuria, cblC type, digenic, 277400
PREPL	99,8	98,2	100	100	Myasthenic syndrome, congenital, 22, 616224
PRF1	91,2	90,8	100	100	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Aplastic anemia, 609135 Lymphoma, non-Hodgkin, 605027
PRG4	87,4	80,9	100	100	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250
PRICKLE1	100	100	100	100	Epilepsy, progressive myoclonic 1B, 612437

PRIMPOL	97,5	94,6	100	100	Myopia 22, autosomal dominant, 615420
PRKAA1	100	99,5	100	100	No OMIM disease ID
PRKACA	80,1	79,3	100	100	Cardioacrofacial dysplasia 1, 619142 Cushing syndrome, ACTH-independent adrenal, somatic, 615830
PRKACG	100	99,9	100	100	?Bleeding disorder, platelet-type, 19, 616176
PRKAG2	99,1	96,7	100	99,4	Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200 Cardiomyopathy, hypertrophic 6, 600858
PRKAR1A	99,3	93,5	100	100	Myxoma, intracardiac, 255960 Carney complex, type 1, 160980 Pigmented nodular adrenocortical disease, primary, 1, 610489 Acrodysostosis 1, with or without hormone resistance, 101800 Adrenocortical tumor, somatic, 0
PRKCA	100	100	100	100	Pituitary tumor, invasive, 0
PRKCB	100	100	100	100	No OMIM disease ID
PRKCD	100	100	100	100	Autoimmune lymphoproliferative syndrome, type III, 615559
PRKCG	99,9	98,4	100	100	Spinocerebellar ataxia 14, 605361
PRKCSH	99,8	95,4	100	100	Polycystic liver disease 1, 174050
PRKD1	99,6	98,7	100	100	Congenital heart defects and ectodermal dysplasia, 617364
PRKDC	99,7	98	100	100	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PRKG1	92,5	91,2	92,7	92,7	Aortic aneurysm, familial thoracic 8, 615436
PRKN	67	66,2	75,3	75,3	Parkinson disease, juvenile, type 2, 600116 Ovarian cancer, somatic, 167000 Adenocarcinoma of lung, somatic, 211980
PRKRA	100	99,4	100	100	Dystonia 16, 612067
PRLR	99,9	99,8	100	100	Hyperprolactinemia, 615555 Multiple fibroadenomas of the breast, 615554
PRMT7	100	99,9	100	100	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157
PRNP	100	100	100	100	Insomnia, fatal familial, 600072 {Kuru, susceptibility to}, 245300 Huntington disease-like 1, 603218

					Prion disease with protracted course, 606688 Cerebral amyloid angiopathy, PRNP-related, 137440 Creutzfeldt-Jakob disease, 123400 Gerstmann-Straussler disease, 137440
PROC	100	100	100	100	Thrombophilia due to protein C deficiency, autosomal dominant, 176860 Thrombophilia due to protein C deficiency, autosomal recessive, 612304
PRODH	85	80,6	100	100	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850
PROK2	99,9	98,5	100	100	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	100	100	100	100	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROM1	97,2	96,1	100	100	Retinitis pigmentosa 41, 612095 Stargardt disease 4, 603786 Cone-rod dystrophy 12, 612657 Macular dystrophy, retinal, 2, 608051
PROP1	92,6	82,6	100	100	Pituitary hormone deficiency, combined, 2, 262600
PRORP	100	99,5	100	100	No OMIM disease ID
PROS1	96,7	92,1	98,4	98,4	Thrombophilia due to protein S deficiency, autosomal recessive, 614514 Thrombophilia due to protein S deficiency, autosomal dominant, 612336
PROZ	100	99,8	100	100	[Protein Z deficiency], 614024
PRPF3	98,8	95,3	100	100	Retinitis pigmentosa 18, 601414
PRPF31	100	98,7	100	100	Retinitis pigmentosa 11, 600138
PRPF4	100	99,8	100	100	Retinitis pigmentosa 70, 615922
PRPF6	100	99,8	100	100	Retinitis pigmentosa 60, 613983
PRPF8	100	99,3	100	100	Retinitis pigmentosa 13, 600059
PRPH2	100	100	100	100	Macular dystrophy, patterned, 1, 169150 Retinitis punctata albescens, 136880 Choroidal dystrophy, central areolar 2, 613105 Retinitis pigmentosa 7 and digenic form, 608133 Leber congenital amaurosis 18, 608133 Macular dystrophy, vitelliform, 3, 608161
PRPS1	86,4	86,4	100	100	Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Phosphoribosylpyrophosphate synthetase superactivity, 300661 Deafness, X-linked 1, 304500

					Arts syndrome, 301835 Gout, PRPS-related, 300661
PRR12	98,7	97,2	100	100	No OMIM disease ID
PRRT2	100	99,6	100	100	Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751 Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066
PRRX1	100	99,7	100	100	Agnathia-otocephaly complex, 202650
PRSS1	100	100	100	100	Pancreatitis, hereditary, 167800
PRSS12	100	99,9	100	100	Mental retardation, autosomal recessive 1, 249500
PRSS56	99,9	96,5	100	100	Microphthalmia, isolated 6, 613517
PRUNE1	93,6	93,5	93,6	93,6	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481
PRX	96	95,5	96,5	96,1	Charcot-Marie-Tooth disease, type 4F, 614895 Dejerine-Sottas disease, 145900
PSAP	100	100	100	100	Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Combined SAP deficiency, 611721 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PSAT1	95,3	81,6	100	100	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992
PSEN1	100	100	100	100	Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 ?Acne inversa, familial, 3, 613737 Alzheimer disease, type 3, 607822 Dementia, frontotemporal, 600274 Pick disease, 172700 Cardiomyopathy, dilated, 1U, 613694
PSEN2	100	100	100	100	Alzheimer disease-4, 606889 Cardiomyopathy, dilated, 1V, 613697
PSENEN	100	100	100	100	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736
PSIP1	98,8	93,5	100	100	No OMIM disease ID
PSMA3	99,8	97,2	100	100	No OMIM disease ID
PSMB1	100	100	100	100	No OMIM disease ID

PSMB4	100	100	100	100	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591
PSMB8	99,9	98,5	100	100	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040
PSMB9	99,9	97,7	100	100	?Proteasome-associated autoinflammatory syndrome 3, digenic, 617591
PSMC3IP	100	100	100	100	Ovarian dysgenesis 3, 614324
PSMD12	98,6	92,9	100	100	Stankiewicz-Isidor syndrome, 617516
PSMG2	100	98,9	100	100	No OMIM disease ID
PSPH	100	100	100	100	Phosphoserine phosphatase deficiency, 614023
PSTPIP1	100	99,1	100	99,9	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416
PTCD3	99,2	97,6	100	100	?Combined oxidative phosphorylation deficiency 51, 619057
PTCH1	99,2	97,6	99,9	99,8	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly 7, 610828
PTCH2	99,9	99	100	100	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Medulloblastoma, somatic, 155255
PTCHD1	100	99,9	100	100	{Autism, susceptibility to, X-linked 4}, 300830
PTDSS1	100	100	100	100	Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	99,5	97	100	100	Prostate cancer, somatic, 176807 {Glioma susceptibility 2}, 613028 Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309 {Meningioma}, 607174
PTF1A	95,8	85,6	98,6	93,3	Pancreatic and cerebellar agenesis, 609069 Pancreatic agenesis 2, 615935
PTGIS	98,2	95,1	100	100	Hypertension, essential, 145500
PTGS1	100	99,8	100	100	No OMIM disease ID
PTH	99,7	97	100	100	Hypoparathyroidism, familial isolated 1, 146200
PTH1R	100	98,7	100	100	Metaphyseal chondrodysplasia, Murk Jansen type, 156400 Failure of tooth eruption, primary, 125350

					Eiken syndrome, 600002 Chondrodysplasia, Blomstrand type, 215045
PTHLH	99,7	98,4	100	100	Brachydactyly, type E2, 613382
PTPN11	99,1	93,7	100	100	LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Noonan syndrome 1, 163950 Leukemia, juvenile myelomonocytic, somatic, 607785
PTPN12	99,1	96,8	100	100	Colon cancer, somatic, 114500
PTPN14	99,7	97,4	100	100	Choanal atresia and lymphedema, 613611
PTPN22	99,5	97,1	100	100	{Diabetes, type 1, susceptibility to}, 222100 {Systemic lupus erythematosus susceptibility to}, 152700 {Rheumatoid arthritis, susceptibility to}, 180300
PTPN23	100	100	100	100	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity, 618890
PTPRC	99	95,1	100	100	{Hepatitis C virus, susceptibility to}, 609532 Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971
PTPRF	100	99,7	100	100	?Breasts and/or nipples, aplasia or hypoplasia of, 2, 616001
PTPRJ	97,7	97,2	99,9	99,6	Colon cancer, somatic, 114500
PTPRO	99,9	99,4	100	100	Nephrotic syndrome, type 6, 614196
PTPRQ	94,6	92,5	92,8	92,7	Deafness, autosomal dominant 73, 617663 Deafness, autosomal recessive 84A, 613391
PTRH2	100	100	100	100	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PTRHD1	100	100	100	100	No OMIM disease ID
PTS	99,9	99,1	100	100	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUF60	100	99,3	100	100	Verheij syndrome, 615583
PUM1	100	99,9	100	100	Spinocerebellar ataxia 47, 617931
PURA	99	95,2	100	99,8	Mental retardation, autosomal dominant 31, 616158
PUS1	100	99,5	99,6	97,2	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PUS3	100	100	100	100	Neurodevelopmental disorder with microcephaly and gray sclerae, 617051
PUS7	100	99,8	100	100	Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342

PXDN	100	99,6	100	100	Anterior segment dysgenesis 7, with sclerocornea, 269400
PYCR1	99,9	97,7	100	100	Cutis laxa, autosomal recessive, type IIIB, 614438 Cutis laxa, autosomal recessive, type IIB, 612940
PYCR2	100	99,1	100	100	Leukodystrophy, hypomyelinating, 10, 616420
PYGL	100	100	100	100	Glycogen storage disease VI, 232700
PYGM	100	99,9	100	100	McArdle disease, 232600
PYROXD1	95,2	83,9	100	100	Myopathy, myofibrillar, 8, 617258
QARS1	100	100	100	100	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	100	99,7	100	100	Hyperphenylalaninemia, BH4-deficient, C, 261630
QRICH1	100	99,9	100	100	Ververi-Brady syndrome, 617982
QRICH2	94,3	93,5	100	100	Spermatogenic failure 35, 618341
QRSL1	99,2	93,9	100	100	Combined oxidative phosphorylation deficiency 40, 618835
RAB11B	100	100	100	100	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807
RAB18	99,5	97,4	100	100	Warburg micro syndrome 3, 614222
RAB23	100	99,5	100	100	Carpenter syndrome, 201000
RAB27A	100	100	100	100	Griscelli syndrome, type 2, 607624
RAB28	99,7	96	100	100	Cone-rod dystrophy 18, 615374
RAB33B	85	85	100	100	Smith-McCort dysplasia 2, 615222
RAB39B	100	100	100	100	Waisman syndrome, 311510 Mental retardation, X-linked 72, 300271
RAB3GAP1	99,4	98,9	99,4	99,4	Warburg micro syndrome 1, 600118
RAB3GAP2	99,5	97	100	100	Warburg micro syndrome 2, 614225 Martsolf syndrome, 212720
RAB7A	100	99,9	100	100	Charcot-Marie-Tooth disease, type 2B, 600882
RAC1	99,9	96,2	100	99,9	Mental retardation, autosomal dominant 48, 617751
RAC2	99,9	98,3	100	100	?Immunodeficiency 73C with defective neutrophil chemotaxis and hypogammaglobulinemia, 618987 Immunodeficiency 73B with defective neutrophil chemotaxis and lymphopenia, 618986 Immunodeficiency 73A with defective neutrophil chemotaxix and leukocytosis, 608203

RAC3	97,3	94,4	99,7	98,2	Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577
RAD21	99,2	96,6	100	100	?Mungan syndrome, 611376 Cornelia de Lange syndrome 4, 614701
RAD50	97,5	91,6	100	100	Nijmegen breakage syndrome-like disorder, 613078
RAD51	89,4	89,4	89,4	89,4	{Breast cancer, susceptibility to}, 114480 Mirror movements 2, 614508 Fanconi anemia, complementation group R, 617244
RAD51C	100	99,8	100	100	{Breast-ovarian cancer, familial, susceptibility to, 3}, 613399 Fanconi anemia, complementation group O, 613390
RAD51D	100	99,9	100	100	{Breast-ovarian cancer, familial, susceptibility to, 4}, 614291
RAD54B	99,7	97,3	100	100	Colon cancer, somatic, 114500 Lymphoma, non-Hodgkin, somatic, 605027
RAD54L	100	98,9	100	100	{Breast cancer, invasive ductal}, 114480 Lymphoma, non-Hodgkin, somatic, 605027 Adenocarcinoma, colonic, somatic, 0
RAF1	100	100	100	100	LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553 Cardiomyopathy, dilated, 1NN, 615916
RAG1	100	100	100	100	Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457 Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889 Combined cellular and humoral immune defects with granulomas, 233650
RAG2	100	100	100	100	Severe combined immunodeficiency, B cell-negative, 601457 Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554
RAI1	100	100	100	100	Smith-Magenis syndrome, 182290
RALA	94,6	87,9	100	100	No OMIM disease ID
RALGAPA1	74,5	63,9	100	100	Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation, 618797
RANBP2	49,7	49,3	100	100	{Encephalopathy, acute, infection-induced, 3, susceptibility to}, 608033
RANGRF	100	99,9	100	100	No OMIM disease ID
RAP1GDS1	99,8	96,8	100	100	Lymphocytic leukemia, acute T-cell, 0

RAPGEF2	99,7	98,8	100	100	?Epilepsy, familial adult myoclonic, 7, 618075
RAPSN	100	99,7	100	100	Fetal akinesia deformation sequence 2, 618388 Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326
RARB	100	100	100	100	Microphthalmia, syndromic 12, 615524
RARS1	94,2	91,6	94,4	94,3	Leukodystrophy, hypomyelinating, 9, 616140
RARS2	100	99,8	100	100	Pontocerebellar hypoplasia, type 6, 611523
RASA1	98,8	96,3	100	100	Capillary malformation-arteriovenous malformation 1, 608354 Basal cell carcinoma, somatic, 605462
RASGRP1	100	99,6	100	100	Immunodeficiency 64, 618534
RASGRP2	99,7	97,3	100	100	?Bleeding disorder, platelet-type, 18, 615888
RAX	96	87	100	98,4	Microphthalmia, isolated 3, 611038
RAX2	100	92,3	100	100	?Macular degeneration, age-related, 6, 613757 Cone-rod dystrophy 11, 610381
RB1	96,8	92,1	100	100	Small cell cancer of the lung, somatic, 182280 Bladder cancer, somatic, 109800 Retinoblastoma, trilateral, 180200 Osteosarcoma, somatic, 259500 Retinoblastoma, 180200
RB1CC1	99,6	96,7	100	100	Breast cancer, somatic, 114480
RBBP6	97,8	95,9	100	100	No OMIM disease ID
RBBP8	100	99,7	100	100	Jawad syndrome, 251255 Seckel syndrome 2, 606744 Pancreatic carcinoma, somatic, 0
RBCK1	99,9	98,2	100	100	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RBFOX1	89,2	88,8	99,2	97,7	No OMIM disease ID
RBM10	99,5	97,1	100	100	TARP syndrome, 311900
RBM20	100	99,9	100	100	Cardiomyopathy, dilated, 1DD, 613172
RBM28	100	100	100	100	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RBM8A	99,8	97,9	100	100	Thrombocytopenia-absent radius syndrome, 274000
RBMX	94,6	84	100	100	?Mental retardation, X-linked, syndromic 11, Shashi type, 300238

RBP3	100	100	100	100	?Retinitis pigmentosa 66, 615233
RBP4	99,9	97,7	100	100	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RBPJ	98,4	92,8	100	100	Adams-Oliver syndrome 3, 614814
RC3H1	100	99,4	100	100	?Immune dysregulation and systemic hyperinflammation syndrome, 618998
RCBTB1	99,9	99,5	100	100	Retinal dystrophy with or without extraocular anomalies, 617175
RD3	100	100	100	100	Leber congenital amaurosis 12, 610612
RDH11	100	99	100	100	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108
RDH12	100	98,6	100	100	Leber congenital amaurosis 13, 612712
RDH5	100	99,9	100	100	Fundus albipunctatus, 136880
RDX	89,1	71,5	100	100	Deafness, autosomal recessive 24, 611022
RECQL4	99,8	98,1	100	99,9	RAPADILINO syndrome, 266280 Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2,, 268400
REEP1	78,7	76,1	100	100	Spastic paraplegia 31, autosomal dominant, 610250 ?Neuronopathy, distal hereditary motor, type VB, 614751
REEP2	99,9	98,6	100	100	?Spastic paraplegia 72, autosomal dominant, 615625 ?Spastic paraplegia 72, autosomal recessive, 615625
REEP6	100	100	91,5	87,4	Retinitis pigmentosa 77, 617304
RELA	99,6	98,8	100	100	?Mucocutaneous ulceration, chronic, 618287
RELB	98,8	88,7	100	100	?Immunodeficiency 53, 617585
RELN	100	99,8	100	100	{Epilepsy, familial temporal lobe, 7}, 616436 Lissencephaly 2 (Norman-Roberts type), 257320
RELT	100	99,9	100	100	Amelogenesis imperfecta, type IIIC, 618386
REN	100	100	100	100	Renal tubular dysgenesis, 267430 Hyperuricemic nephropathy, familial juvenile 2, 613092 [Hyperproreninemia], 0
REPS1	99,6	97,5	100	100	?Neurodegeneration with brain iron accumulation 7, 617916
RERE	96,1	91,3	99,9	99,9	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975

REST	98,5	98,2	98,6	98,6	{Wilms tumor 6, susceptibility to}, 616806 Fibromatosis, gingival, 5, 617626 ?Deafness, autosomal dominant 27, 612431
RET	99,9	99,1	100	100	Multiple endocrine neoplasia IIB, 162300 Pheochromocytoma, 171300 Multiple endocrine neoplasia IIA, 171400 Medullary thyroid carcinoma, 155240 {Hirschsprung disease, protection against}, 142623 Central hypoventilation syndrome, congenital, 209880 {Hirschsprung disease, susceptibility to, 1}, 142623
RETREG1	98,8	95,1	100	100	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
REV3L	97,6	97,2	97,6	97,6	No OMIM disease ID
RFC1	99,9	98,9	100	100	Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome, 614575
RFT1	99,8	99,6	100	100	Congenital disorder of glycosylation, type In, 612015
RFWD3	100	99,8	100	100	?Fanconi anemia, complementation group W, 617784
RFX3	100	100	100	100	No OMIM disease ID
RFX5	99,7	98,1	100	100	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFX6	100	99,6	100	100	Mitchell-Riley syndrome, 615710
RFXANK	100	99,5	100	100	MHC class II deficiency, complementation group B, 209920
RFXAP	99,3	97	100	99,9	Bare lymphocyte syndrome, type II, complementation group D, 209920
RGR	99	98,2	99	99	Retinitis pigmentosa 44, 613769
RGS9	98,5	97,1	100	100	Bradyopsia, 608415
RGS9BP	99,6	95	100	100	Bradyopsia, 608415
RHAG	100	99,7	100	100	Overhydrated hereditary stomatocytosis, 185000 Anemia, hemolytic, Rh-null, regulator type, 268150
RHBDF2	99,9	98,6	100	100	Tylosis with esophageal cancer, 148500
RHCE	98,1	98,1	97,6	97,5	Rh-null disease, amorph type, 617970 [Blood group, Rhesus], 0
RHEB	88,8	75,4	100	100	No OMIM disease ID

RHO	100	100	100	100	Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis punctata albescens, 136880 Retinitis pigmentosa 4, autosomal dominant or recessive, 613731
RHOA	81,2	80,7	80,7	80,7	Ectodermal dysplasia with facial dysmorphism and acral, ocular, and brain anomalies, somatic mosaic, 618727
RHOBTB2	100	100	100	100	Developmental and epileptic encephalopathy 64, 618004
RHOG	100	100	100	100	No OMIM disease ID
RHOH	100	100	100	100	{?Epidermodysplasia verruciformis, susceptibility to, 4}, 618307
RIC1	100	99,9	100	100	CATIFA syndrome, 618761
RIMS1	99,8	97,7	100	100	Cone-rod dystrophy 7, 603649
RIMS2	96,7	95,3	97,8	97,7	Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970
RIN2	100	100	100	100	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075
RINT1	99,9	98,6	100	100	Infantile liver failure syndrome 3, 618641
RIPK1	100	99	100	100	Immunodeficiency 57 with autoinflammation, 618108 Autoinflammation with episodic fever and lymphadenopathy, 618852
RIPK4	100	99,9	100	100	Popliteal pterygium syndrome, Bartsocas-Papas type, 263650 CHAND syndrome, 214350
RIPOR2	100	99,8	100	100	?Deafness, autosomal recessive 104, 616515
RIPPLY2	100	97,9	100	100	?Spondylocostal dysostosis 6, 616566
RIT1	100	100	100	100	Noonan syndrome 8, 615355
RLBP1	100	99,9	100	100	Retinitis punctata albescens, 136880 Bothnia retinal dystrophy, 607475 Newfoundland rod-cone dystrophy, 607476 Fundus albipunctatus, 136880
RLIM	100	99	100	100	Tonne-Kalscheuer syndrome, 300978
RMND1	100	98,6	100	100	Combined oxidative phosphorylation deficiency 11, 614922
RMRP					Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RNASEH1	98,5	95,3	100	100	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479

RNASEH2A	100	100	100	100	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	80,6	78,1	91	90,9	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	100	99,5	100	100	Aicardi-Goutieres syndrome 3, 610329
RNASEL	100	99,8	100	100	Prostate cancer 1, 601518
RNASET2	97,4	93,1	100	100	Leukoencephalopathy, cystic, without megalencephaly, 612951
RNF113A	100	100	100	100	Trichothiodystrophy 5, nonphotosensitive, 300953
RNF125	99,9	99,2	100	100	Tenorio syndrome, 616260
RNF13	95,2	81,6	100	100	Developmental and epileptic encephalopathy 73, 618379
RNF139	100	100	100	100	Renal cell carcinoma, 144700
RNF168	100	99,8	100	100	RIDDLE syndrome, 611943
RNF170	99,6	97,6	100	100	Ataxia, sensory, 1, autosomal dominant, 608984
RNF212	100	99,6	100	100	Recombination rate QTL 1, 612042
RNF213	99,8	99	100	100	{Moyamoya disease 2, susceptibility to}, 607151
RNF216	99,8	98,7	100	100	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840
RNF31	99,9	99	100	100	No OMIM disease ID
RNF43	99,9	99,1	100	100	Sessile serrated polyposis cancer syndrome, 617108
RNF6	100	99,7	100	100	Esophageal carcinoma, somatic, 133239
RNPC3	91,5	70,7	100	100	?Growth hormone deficiency, isolated, type V, 618160
RNU4ATAC					Lowry-Wood syndrome, 226960 Microcephalic osteodysplastic primordial dwarfism, type I, 210710 Roifman syndrome, 616651
ROBO1	100	99,9	100	100	No OMIM disease ID
ROBO2	99,4	97,8	100	100	Vesicoureteral reflux 2, 610878
ROBO3	98,9	96,1	100	100	Gaze palsy, familial horizontal, with progressive scoliosis, 1, 607313
ROBO4	99,9	98,6	100	100	Aortic valve disease 8, 618496
ROGDI	98,4	95,2	99,9	99,1	Kohlschutter-Tonz syndrome, 226750

ROM1	100	99,9	100	100	Retinitis pigmentosa 7, digenic form, 608133
ROR1	97	96,8	99,9	99,3	?Deafness, autosomal recessive 108, 617654
ROR2	100	99,9	97	97	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RORA	96,7	90,2	100	100	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060
RORC	100	100	100	100	Immunodeficiency 42, 616622
RP1	91,5	90,6	100	100	Retinitis pigmentosa 1, 180100
RP1L1	100	100	100	100	Retinitis pigmentosa 88, 618826 Occult macular dystrophy, 613587
RP2	100	99,8	100	100	Retinitis pigmentosa 2, 312600
RP9	80,8	75,9	100	99,3	?Retinitis pigmentosa 9, 180104
RPE65	99,8	97,8	100	100	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 87 with choroidal involvement, 618697 Retinitis pigmentosa 20, 613794
RPGR	76,5	72	100	99,9	Cone-rod dystrophy, X-linked, 1, 304020 Retinitis pigmentosa 3, 300029 Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 Macular degeneration, X-linked atrophic, 300834
RPGRIP1	100	99,9	100	100	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826
RPGRIP1L	96,7	95,7	100	99,5	?COACH syndrome 3, 619113 Meckel syndrome 5, 611561 Joubert syndrome 7, 611560
RPIA	98,6	94,9	100	100	Ribose 5-phosphate isomerase deficiency, 608611
RPL10	97,4	89,1	100	100	Mental retardation, X-linked, syndromic, 35, 300998 {Autism, susceptibility to, X-linked 5}, 300847
RPL11	100	100	100	100	Diamond-Blackfan anemia 7, 612562
RPL13	96,3	85,5	100	100	Spondyloepimetaphyseal dysplasia, Isidor-Toutain type, 618728
RPL15	86,8	78	100	100	?Diamond-Blackfan anemia 12, 615550
RPL18	100	100	100	100	?Diamond-Blackfan anemia 18, 618310
RPL21	88,8	71,7	100	100	Hypotrichosis 12, 615885

RPL26	97,2	84,4	100	100	?Diamond-Blackfan anemia 11, 614900
RPL27	73,6	56,5	100	100	?Diamond-Blackfan anemia 16, 617408
RPL31	99,3	94,6	100	100	No OMIM disease ID
RPL35	86,4	75	100	100	?Diamond-Blackfan anemia 19, 618312
RPL35A	97,1	88,7	100	100	Diamond-Blackfan anemia 5, 612528
RPL4	87,6	78,3	100	100	No OMIM disease ID
RPL5	86,2	70	100	100	Diamond-Blackfan anemia 6, 612561
RPL9	98,9	92	100	100	No OMIM disease ID
RPS10	98	92,5	100	100	Diamond-Blackfan anemia 9, 613308
RPS14	98,8	95	100	100	Macrocytic anemia, refractory, due to 5q deletion, somatic, 153550
RPS15A	96,9	86,7	80,5	80,4	?Diamond-Blackfan anemia 20, 618313
RPS17	84,2	69,8	100	100	Diamond-Blackfan anemia 4, 612527
RPS19	100	99,6	100	100	Diamond-Blackfan anemia 1, 105650
RPS20	98,6	93,6	100	100	No OMIM disease ID
RPS23	87,8	80,1	100	100	Brachycephaly, trichomegaly, and developmental delay, 617412
RPS24	98,4	93,1	100	100	Diamond-blackfan anemia 3, 610629
RPS26	95,7	84,9	100	100	Diamond-Blackfan anemia 10, 613309
RPS27	85,9	60,6	100	100	?Diamond-Blackfan anemia 17, 617409
RPS28	100	94,8	100	100	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164
RPS29	82	74,7	100	100	Diamond-Blackfan anemia 13, 615909
RPS6KA3	98,4	94,5	100	98,9	Mental retardation, X-linked 19, 300844 Coffin-Lowry syndrome, 303600
RPS7	80	68,7	100	100	Diamond-Blackfan anemia 8, 612563
RPSA	100	99,8	100	100	Asplenia, isolated congenital, 271400
RRAD	85,4	80,7	99,4	96,2	No OMIM disease ID

RRAGC	100	99,7	100	100	No OMIM disease ID
RRAS	99,8	95,7	100	100	No OMIM disease ID
RRAS2	96,8	88,7	100	100	Noonan syndrome 12, 618624 Ovarian carcinoma, 0
RREB1	99,9	99,2	100	100	No OMIM disease ID
RRM1	100	99,5	100	100	No OMIM disease ID
RRM2B	100	99,7	100	100	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075
RS1	99,8	93,9	100	100	Retinoschisis, 312700
RSPH1	100	100	100	100	Ciliary dyskinesia, primary, 24, 615481
RSPH3	99,9	98,8	100	100	Ciliary dyskinesia, primary, 32, 616481
RSPH4A	98,1	95,6	100	100	Ciliary dyskinesia, primary, 11, 612649
RSPH9	99,9	97,9	100	100	Ciliary dyskinesia, primary, 12, 612650
RSPO1	100	99,9	100	100	Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644 Palmoplantar hyperkeratosis and true hermaphroditism, 610644
RSPO2	97,1	90,7	100	100	Tetraamelia syndrome 2, 618021 ?Humerofemoral hypoplasia with radiotibial ray deficiency, 618022
RSPO4	100	100	100	100	Anonychia congenita, 206800
RSPRY1	100	100	100	100	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723
RSRC1	99,8	96,8	100	100	Intellectual developmental disorder, autosomal recessive 70, 618402
RTEL1	99,5	96,8	100	100	Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 Dyskeratosis congenita, autosomal dominant 4, 615190
RTN2	100	99,2	100	100	Spastic paraplegia 12, autosomal dominant, 604805
RTN4IP1	99,9	98,7	100	100	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
RTTN	98,9	98	100	100	Microcephaly, short stature, and polymicrogyria with seizures, 614833
RUBCN	99,4	97,5	100	100	Spinocerebellar ataxia, autosomal recessive 15, 615705
RUNX1	99,3	94,9	100	100	Leukemia, acute myeloid, 601626 Platelet disorder, familial, with associated myeloid malignancy, 601399

RUNX2	72,2	72,2	100	100	Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510 Cleidocranial dysplasia, 119600 Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600
RUNX3	97,2	93,3	100	100	No OMIM disease ID
RUSC2	100	100	100	100	Mental retardation, autosomal recessive 61, 617773
RXYLT1	99,5	96,8	100	99,9	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
RYR1	96,9	93,9	99,4	99	Central core disease, 117000 King-Denborough syndrome, 145600 {Malignant hyperthermia susceptibility 1}, 145600 Minicore myopathy with external ophthalmoplegia, 255320 Neuromuscular disease, congenital, with uniform type 1 fiber, 117000
RYR2	99,9	99	100	100	Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772 Arrhythmogenic right ventricular dysplasia 2, 600996
S1PR2	99,4	96,9	100	100	Deafness, autosomal recessive 68, 610419
SAA2	98,2	88,2	78,3	78,3	No OMIM disease ID
SACS	100	99,9	100	100	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAG	100	100	100	100	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758
SALL1	99,9	99	100	100	Townes-Brocks syndrome 1, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480
SALL2	100	100	100	100	?Coloboma, ocular, autosomal recessive, 216820
SALL4	98,6	96,7	100	100	Duane-radial ray syndrome, 607323 IVIC syndrome, 147750
SAMD11	90,2	81,6	100	100	No OMIM disease ID
SAMD12	100	100	100	100	Epilepsy, familial adult myoclonic, 1, 601068
SAMD9	100	99,8	100	100	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455 Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041
SAMD9L	100	100	100	100	Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270 Ataxia-pancytopenia syndrome, 159550
SAMHD1	98,7	98,4	100	100	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952

SAR1B	97	89,7	100	100	Chylomicron retention disease, 246700
SARDH	93,7	91,7	91,4	91,4	[Sarcosinemia], 268900
SARS1	100	99,3	100	100	?Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709
SARS2	95,8	94,6	100	100	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SART3	99,6	98,6	100	100	No OMIM disease ID
SASH1	99,9	98,7	100	100	?Cancer, alopecia, pigment dyscrasia, onychodystrophy, and keratoderma, 618373 Dyschromatosis universalis hereditaria 1, 127500
SASH3	99,9	97,6	100	100	No OMIM disease ID
SASS6	99,9	98,5	100	100	?Microcephaly 14, primary, autosomal recessive, 616402
SAT1	99,9	98,5	100	99,9	No OMIM disease ID
SATB2	99,7	97,4	100	100	Glass syndrome, 612313
SBDS	100	100	100	100	{Aplastic anemia, susceptibility to}, 609135 Shwachman-Diamond syndrome, 260400
SBF1	99	97,7	100	100	Charcot-Marie-Tooth disease, type 4B3, 615284
SBF2	99,9	99,4	100	100	Charcot-Marie-Tooth disease, type 4B2, 604563
SC5D	100	99,5	100	100	Lathosterolosis, 607330
SCAMP5	100	100	100	100	No OMIM disease ID
SCAPER	99,7	98,2	100	100	Intellectual developmental disorder and retinitis pigmentosa, 618195
SCARB2	100	99,8	100	100	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCARF2	95,4	86,2	99,8	99,2	Van den Ende-Gupta syndrome, 600920
SCD5	100	99,8	100	100	?Deafness, autosomal dominant 79, 619086
SCLT1	96	90,9	95,1	95,1	No OMIM disease ID
SCN10A	100	99,6	100	100	Episodic pain syndrome, familial, 2, 615551
SCN11A	99,8	98,3	100	100	Neuropathy, hereditary sensory and autonomic, type VII, 615548 Episodic pain syndrome, familial, 3, 615552
SCN1A	99,9	99,5	100	100	Febrile seizures, familial, 3A, 604403 Migraine, familial hemiplegic, 3, 609634

					Dravet syndrome, 607208 Epilepsy, generalized, with febrile seizures plus, type 2, 604403
SCN1B	98	96,4	99,8	99,3	Atrial fibrillation, familial, 13, 615377 Developmental and epileptic encephalopathy 52, 617350 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233 Brugada syndrome 5, 612838
SCN2A	99,6	97,6	100	100	Episodic ataxia, type 9, 618924 Developmental and epileptic encephalopathy 11, 613721 Seizures, benign familial infantile, 3, 607745
SCN2B	100	100	100	100	Atrial fibrillation, familial, 14, 615378
SCN3A	99,8	99,2	100	100	Epilepsy, familial focal, with variable foci 4, 617935 Developmental and epileptic encephalopathy 62, 617938
SCN3B	100	100	100	100	Brugada syndrome 7, 613120 Atrial fibrillation, familial, 16, 613120
SCN4A	100	99,6	100	100	Hyperkalemic periodic paralysis, type 2, 170500 Paramyotonia congenita, 168300 Myotonia congenita, atypical, acetazolamide-responsive, 608390 Myasthenic syndrome, congenital, 16, 614198 Hypokalemic periodic paralysis, type 2, 613345
SCN4B	100	99,6	100	100	Atrial fibrillation, familial, 17, 611819 Long QT syndrome 10, 611819
SCN5A	99	99	100	100	Atrial fibrillation, familial, 10, 614022 Sick sinus syndrome 1, 608567 Long QT syndrome 3, 603830 Heart block, nonprogressive, 113900 Cardiomyopathy, dilated, 1E, 601154 Brugada syndrome 1, 601144 Heart block, progressive, type IA, 113900 {Sudden infant death syndrome, susceptibility to}, 272120 Ventricular fibrillation, familial, 1, 603829
SCN7A	98,3	93,3	100	100	No OMIM disease ID
SCN8A	100	99,8	100	100	Seizures, benign familial infantile, 5, 617080 Developmental and epileptic encephalopathy 13, 614558 Cognitive impairment with or without cerebellar ataxia, 614306 ?Myoclonus, familial, 2, 618364

SCN9A	99,3	97,9	100	100	Neuropathy, hereditary sensory and autonomic, type IID, 243000 Generalized epilepsy with febrile seizures plus, type 7, 613863 Small fiber neuropathy, 133020 Paroxysmal extreme pain disorder, 167400 Insensitivity to pain, congenital, 243000 Erythermalgia, primary, 133020 Febrile seizures, familial, 3B, 613863
SCNN1A	99,7	98,2	100	100	Pseudohypoaldosteronism, type I, 264350 ?Liddle syndrome 3, 618126 Bronchiectasis with or without elevated sweat chloride 2, 613021
SCNN1B	100	99,7	100	100	Bronchiectasis with or without elevated sweat chloride 1, 211400 Pseudohypoaldosteronism, type I, 264350 Liddle syndrome 1, 177200
SCNN1G	99,8	98,3	100	100	Bronchiectasis with or without elevated sweat chloride 3, 613071 Liddle syndrome 2, 618114 Pseudohypoaldosteronism, type I, 264350
SCO1	97,1	93,8	100	100	Mitochondrial complex IV deficiency, nuclear type 4, 619048
SCO2	100	100	100	100	Mitochondrial complex IV deficiency, nuclear type 2, 604377 Myopia 6, 608908
SCP2	100	99,2	100	100	?Leukoencephalopathy with dystonia and motor neuropathy, 613724
SCYL1	100	100	100	100	Spinocerebellar ataxia, autosomal recessive 21, 616719
SDCCAG8	100	99,9	100	100	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SDHA	85,8	80,4	100	100	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Paragangliomas 5, 614165 Mitochondrial respiratory chain complex II deficiency, 252011
SDHAF1	99,9	93,2	100	100	Mitochondrial complex II deficiency, 252011
SDHAF2	94,6	94,2	98,9	95,4	Paragangliomas 2, 601650
SDHB	100	100	100	100	Pheochromocytoma, 171300 Paragangliomas 4, 115310 Gastrointestinal stromal tumor, 606764 Paraganglioma and gastric stromal sarcoma, 606864

SDHC	100	99,3	100	100	Paragangliomas 3, 605373 Paraganglioma and gastric stromal sarcoma, 606864 Gastrointestinal stromal tumor, 606764
SDHD	54	51,6	80,1	80,1	Paragangliomas 1, with or without deafness, 168000 Mitochondrial complex II deficiency, 252011 Paraganglioma and gastric stromal sarcoma, 606864 Pheochromocytoma, 171300
SDR9C7	100	100	100	100	Ichthyosis, congenital, autosomal recessive 13, 617574
SEC23A	99,7	98,2	100	100	Craniolenticulosutural dysplasia, 607812
SEC23B	99,9	99,3	100	100	?Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SEC24D	100	99,7	100	100	Cole-Carpenter syndrome 2, 616294
SEC31A	99,3	97,1	100	100	?Neurodevelopmental disorder with spastic quadriplegia, optic atrophy, seizures, and structural brain anomalies, 618651
SEC61A1	100	100	100	100	Hyperuricemic nephropathy, familial juvenile, 4, 617056
SEC61B	99,1	92,4	100	100	No OMIM disease ID
SEC63	91,2	83,3	100	100	Polycystic liver disease 2, 617004
SECISBP2	99,8	97,4	100	100	Thyroid hormone metabolism, abnormal, 609698
SELENBP1	100	100	100	100	Extraoral halitosis due to MTO deficiency, 618148
SELENON	84,5	84	87,7	85,1	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310
SELENOP	97,2	89,9	100	100	No OMIM disease ID
SEMA3A	100	99,9	100	100	{Hypogonadotropic hypogonadism 16 with or without anosmia}, 614897
SEMA3E	99,2	98,9	100	100	?CHARGE syndrome, 214800
SEMA4A	100	99,8	100	100	Cone-rod dystrophy 10, 610283 Retinitis pigmentosa 35, 610282
SEMA6B	80,6	73,6	100	100	Epilepsy, progressive myoclonic, 11, 618876
SEPSECS	100	100	100	100	Pontocerebellar hypoplasia type 2D, 613811
SEPTIN12	100	98,8	100	100	Spermatogenic failure 10, 614822
SEPTIN9	100	99,9	100	100	Amyotrophy, hereditary neuralgic, 162100

SERAC1	99,9	99,5	100	100	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPINA1	100	100	100	100	Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490 Emphysema-cirrhosis, due to AAT deficiency, 613490 Emphysema due to AAT deficiency, 613490
SERPINA3	100	100	100	100	Cerebrovascular disease, occlusive, 0 Alpha-1-antichymotrypsin deficiency, 0
SERPINA6	100	100	100	100	Corticosteroid-binding globulin deficiency, 611489
SERPINB6	93,4	93,4	100	100	?Deafness, autosomal recessive 91, 613453
SERPINB7	100	99,9	100	100	Palmoplantar keratoderma, Nagashima type, 615598
SERPINB8	95	95	100	100	Peeling skin syndrome 5, 617115
SERPINC1	100	100	100	100	Thrombophilia due to antithrombin III deficiency, 613118
SERPIND1	100	100	100	100	Thrombophilia due to heparin cofactor II deficiency, 612356
SERPINE1	100	100	100	100	Plasminogen activator inhibitor-1 deficiency, 613329 {Transcription of plasminogen activator inhibitor, modulator of}, 0
SERPINF1	100	100	100	100	Osteogenesis imperfecta, type VI, 613982
SERPINF2	100	99,8	100	100	Alpha-2-plasmin inhibitor deficiency, 262850
SERPING1	99,7	97,5	100	100	Angioedema, hereditary, types I and II, 106100 Complement component 4, partial deficiency of, 120790
SERPINH1	100	98,3	100	100	Osteogenesis imperfecta, type X, 613848 {Preterm premature rupture of the membranes, susceptibility to}, 610504
SERPINI1	99,9	99	100	100	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218
SET	98,3	90,5	98,9	97	Mental retardation, autosomal dominant 58, 618106
SETBP1	99,9	98,7	100	100	Mental retardation, autosomal dominant 29, 616078 Schinzel-Giedion midface retraction syndrome, 269150
SETD1A	100	99,8	100	100	Epilepsy, early-onset, with or without developmental delay, 618832 Neurodevelopmental disorder with speech impairment and dysmorphic facies, 619056
SETD1B	98,2	97,5	100	100	Intellectual developmental disorder with seizures and language delay, 619000
SETD2	100	99,9	100	100	Luscan-Lumish syndrome, 616831
SETD5	100	99,8	98	98	Mental retardation, autosomal dominant 23, 615761
SETX	100	99,8	100	100	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 Amyotrophic lateral sclerosis 4, juvenile, 602433

SF3B1	99,7	98,6	100	100	Myelodysplastic syndrome, somatic, 614286
SF3B4	99,9	97,3	100	100	Acrofacial dysostosis 1, Nager type, 154400
SFRP4	100	99,8	100	100	Pyle disease, 265900
SFTPA1	100	100	100	100	No OMIM disease ID
SFTPA2	100	100	100	100	Pulmonary fibrosis, idiopathic, 178500
SFTPB	99,4	96,7	100	100	Surfactant metabolism dysfunction, pulmonary, 1, 265120
SFTPC	99,2	95,7	100	100	Surfactant metabolism dysfunction, pulmonary, 2, 610913
SFXN4	99,9	98,9	100	100	Combined oxidative phosphorylation deficiency 18, 615578
SGCA	100	99,9	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099
SGCB	97,7	96,5	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286
SGCD	100	98,9	100	100	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287
SGCE	90,4	85	91,2	91,2	Dystonia-11, myoclonic, 159900
SGCG	100	99,2	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700
SGMS2	100	100	100	100	Calvarial doughnut lesions with bone fragility with or without spondylometaphyseal dysplasia, 126550
SGO1	99,9	98,9	100	100	Chronic atrial and intestinal dysrhythmia, 616201
SGPL1	100	100	100	100	Nephrotic syndrome, type 14, 617575
SGSH	94,4	94,1	100	100	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SH2B3	99,4	95,1	100	99,9	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950 Erythrocytosis, somatic, 133100
SH2D1A	97,2	94	100	100	Lymphoproliferative syndrome, X-linked, 1, 308240
SH3BP2	91,4	91,2	97	95,3	Cherubism, 118400
SH3KBP1	99,7	95,9	100	100	?Immunodeficiency 61, 300310
SH3PXD2B	100	100	100	100	Frank-ter Haar syndrome, 249420
SH3TC2	100	99,7	100	100	Charcot-Marie-Tooth disease, type 4C, 601596 Mononeuropathy of the median nerve, mild, 613353

SHANK2	97,7	97,6	98,9	98,9	{Autism susceptibility 17}, 613436
SHANK3	91,6	81,5	96	91,9	{Schizophrenia 15}, 613950 Phelan-McDermid syndrome, 606232
SHH	100	99,5	100	100	Schizencephaly, 269160 Microphthalmia with coloboma 5, 611638 Single median maxillary central incisor, 147250 Holoprosencephaly 3, 142945
SHOC2	99,9	99,4	100	100	Noonan syndrome-like with loose anagen hair 1, 607721
SHOX	70	59,7	95,1	95,1	Langer mesomelic dysplasia, 249700 Short stature, idiopathic familial, 300582 Leri-Weill dyschondrosteosis, 127300
SHROOM3	98,6	97,8	100	100	No OMIM disease ID
SHROOM4	100	99	100	100	Stocco dos Santos X-linked mental retardation syndrome, 300434
SI	99,2	96,1	100	100	Sucrase-isomaltase deficiency, congenital, 222900
SIAH1	100	100	100	100	No OMIM disease ID
SIGLEC7	100	99,8	100	100	No OMIM disease ID
SIGMAR1	100	100	100	100	?Amyotrophic lateral sclerosis 16, juvenile, 614373 ?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726
SIK1	98,7	94,4	100	100	Developmental and epileptic encephalopathy 30, 616341
SIK3	99,8	98,7	99,3	98,1	?Spondyloepimetaphyseal dysplasia, Krakow type, 618162
SIL1	99,2	96,7	100	100	Marinesco-Sjogren syndrome, 248800
SIN3A	100	99	100	100	Witteveen-Kolk syndrome, 613406
SIPA1L3	99,9	98,9	100	100	?Cataract 45, 616851
SIX1	100	99,2	100	100	Deafness, autosomal dominant 23, 605192 Branchiootic syndrome 3, 608389
SIX3	99,9	98,6	100	100	Holoprosencephaly 2, 157170 Schizencephaly, 269160
SIX5	95,4	88,2	100	100	Branchiootorenal syndrome 2, 610896
SIX6	100	100	100	100	Optic disc anomalies with retinal and/or macular dystrophy, 212550
SKI	99,3	94,9	100	99,4	Shprintzen-Goldberg syndrome, 182212

SKIV2L	100	99,8	100	100	Trichohepatoenteric syndrome 2, 614602
SLC10A2	100	100	100	100	Bile acid malabsorption, primary, 613291
SLC10A7	99,7	98	100	100	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363
SLC11A2	98,2	98	100	100	Anemia, hypochromic microcytic, with iron overload 1, 206100
SLC12A1	96,2	96,1	96,2	96,2	Bartter syndrome, type 1, 601678
SLC12A2	94	91,4	100	99,8	Delpire-McNeill syndrome, 619083 Kilquist syndrome, 619080 Deafness, autosomal dominant 78, 619081
SLC12A3	100	99,9	100	100	Gitelman syndrome, 263800
SLC12A5	83,9	83,8	97,4	97,4	Developmental and epileptic encephalopathy 34, 616645 {Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685
SLC12A6	100	100	100	100	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC13A3	99,4	97,5	100	100	Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate, 618384
SLC13A5	100	99,9	100	100	Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905
SLC16A1	100	99,3	100	100	Monocarboxylate transporter 1 deficiency, 616095 Hyperinsulinemic hypoglycemia, familial, 7, 610021 Erythrocyte lactate transporter defect, 245340
SLC16A12	100	99,9	100	100	Cataract 47, juvenile, with microcornea, 612018
SLC16A2	99,2	93,7	100	100	Allan-Herndon-Dudley syndrome, 300523
SLC17A5	99,6	97	100	100	Sialic acid storage disorder, infantile, 269920 Salla disease, 604369
SLC17A8	100	100	100	100	Deafness, autosomal dominant 25, 605583
SLC17A9	96,3	95,4	100	100	Porokeratosis 8, disseminated superficial actinic type, 616063
SLC18A2	100	99,7	100	100	?Parkinsonism-dystonia, infantile, 2, 618049
SLC18A3	100	100	100	100	Myasthenic syndrome, congenital, 21, presynaptic, 617239
SLC19A2	100	99,7	100	100	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	97,8	97,6	98,7	98,7	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A1	99,9	99,6	100	100	{?Schizophrenia susceptibility 18}, 615232 Dicarboxylic aminoaciduria, 222730

SLC1A2	96,1	95,4	100	100	Developmental and epileptic encephalopathy 41, 617105
SLC1A3	100	99,9	100	100	Episodic ataxia, type 6, 612656
SLC1A4	99	95,8	100	100	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC20A2	100	99,2	100	100	Basal ganglia calcification, idiopathic, 1, 213600
SLC22A12	100	99,8	100	100	Hypouricemia, renal, 220150
SLC22A18	100	98,1	100	100	Lung cancer, somatic, 211980 Breast cancer, somatic, 114480 Rhabdomyosarcoma, somatic, 268210
SLC22A4	100	99,6	100	100	{Rheumatoid arthritis, susceptibility to}, 180300
SLC22A5	100	100	100	100	Carnitine deficiency, systemic primary, 212140
SLC24A1	100	99,9	100	100	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
SLC24A4	100	99,8	100	100	[Skin/hair/eye pigmentation 6, blue/green eyes], 210750 Amelogenesis imperfecta, type IIA5, 615887 [Skin/hair/eye pigmentation 6, blond/brown hair], 210750
SLC24A5	99,9	99,1	100	100	[Skin/hair/eye pigmentation 4, fair/dark skin], 113750 Albinism, oculocutaneous, type VI, 113750
SLC25A1	95,8	88,6	99,5	97,8	Myasthenic syndrome, congenital, 23, presynaptic, 618197 Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A10	76,2	69,3	100	100	?Mitochondrial DNA depletion syndrome 19, 618972
SLC25A11	100	100	100	100	Paragangliomas 6, 618464
SLC25A12	99,9	99,5	100	100	Developmental and epileptic encephalopathy 39, 612949
SLC25A13	100	99,7	100	100	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A15	99,8	98,1	100	100	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A19	100	98,5	100	100	Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710 Microcephaly, Amish type, 607196
SLC25A20	100	100	100	100	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A21	100	99,7	100	100	?Mitochondrial DNA depletion syndrome 18, 618811
SLC25A22	98,6	95,8	100	100	Developmental and epileptic encephalopathy 3, 609304
SLC25A24	99,4	99,3	99,8	99,8	Fontaine progeroid syndrome, 612289

SLC25A26	100	99,5	100	100	Combined oxidative phosphorylation deficiency 28, 616794
SLC25A3	99,8	98	100	100	Mitochondrial phosphate carrier deficiency, 610773
SLC25A32	100	100	100	100	?Exercise intolerance, riboflavin-responsive, 616839
SLC25A37	100	100	100	100	No OMIM disease ID
SLC25A38	97,9	95,3	100	100	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC25A4	100	100	100	100	Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283
SLC25A42	96,5	93,2	100	100	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416
SLC25A46	99,7	97,3	100	100	Neuropathy, hereditary motor and sensory, type VIB, 616505
SLC26A1	100	99,6	100	100	?Nephrolithiasis, calcium oxalate, 167030
SLC26A2	100	100	100	100	De la Chapelle dysplasia, 256050 Atelosteogenesis, type II, 256050 Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 Diastrophic dysplasia, 222600 Achondrogenesis Ib, 600972 Epiphyseal dysplasia, multiple, 4, 226900
SLC26A3	100	99,5	100	100	Diarrhea 1, secretory chloride, congenital, 214700
SLC26A4	100	99,7	100	100	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791 Pendred syndrome, 274600
SLC26A5	99,1	96,8	100	100	?Deafness, autosomal recessive 61, 613865
SLC26A8	100	99,6	100	100	Spermatogenic failure 3, 606766
SLC27A4	100	99,8	100	100	Ichthyosis prematurity syndrome, 608649
SLC28A1	100	98,8	100	100	[Uridine-cytidineuria], 618477
SLC29A3	100	99,6	100	100	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC2A1	92,8	92,8	100	100	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 {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847

SLC2A10	97,7	97,7	100	100	Arterial tortuosity syndrome, 208050
SLC2A2	100	100	100	100	Fanconi-Bickel syndrome, 227810 {Diabetes mellitus, noninsulin-dependent}, 125853
SLC2A9	99,8	96,1	100	100	{Uric acid concentration, serum, QTL 2}, 612076 Hypouricemia, renal, 2, 612076
SLC30A10	100	100	100	100	Hypermanganesemia with dystonia 1, 613280
SLC30A2	100	99	100	100	Zinc deficiency, transient neonatal, 608118
SLC30A9	98,8	94,2	100	100	?Birk-Landau-Perez syndrome, 617595
SLC33A1	99,9	98,9	100	100	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC34A1	99,9	99,1	100	100	Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286 Hypercalcemia, infantile, 2, 616963 ?Fanconi renotubular syndrome 2, 613388
SLC34A2	100	100	100	100	Pulmonary alveolar microlithiasis, 265100
SLC34A3	100	99,4	100	100	Hypophosphatemic rickets with hypercalciuria, 241530
SLC35A1	100	99,7	100	100	Congenital disorder of glycosylation, type IIf, 603585
SLC35A2	99,9	98,4	100	100	Congenital disorder of glycosylation, type IIm, 300896
SLC35A3	80,7	78,6	81,1	81	?Arthrogryposis, mental retardation, and seizures, 615553
SLC35C1	99,9	98,7	100	100	Congenital disorder of glycosylation, type IIc, 266265
SLC35D1	100	97,7	100	100	Schneckenbecken dysplasia, 269250
SLC36A2	100	100	100	100	Iminoglycinuria, digenic, 242600 Hyperglycinuria, 138500
SLC37A4	100	99,2	100	100	Glycogen storage disease Ic, 232240 Glycogen storage disease Ib, 232220
SLC38A8	99,9	97,3	100	100	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218
SLC39A13	99,8	98,2	100	100	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350
SLC39A14	100	99,4	93,5	93,5	?Hyperostosis cranalis interna, 144755 Hypermanganesemia with dystonia 2, 617013
SLC39A4	99,5	95,5	100	100	Acrodermatitis enteropathica, 201100
SLC39A5	99,9	99	100	100	Myopia 24, autosomal dominant, 615946

SLC39A7	100	100	100	100	No OMIM disease ID
SLC39A8	100	99,7	100	100	Congenital disorder of glycosylation, type IIn, 616721
SLC3A1	100	99,8	96,6	96,6	Cystinuria, 220100
SLC40A1	100	99,5	100	100	Hemochromatosis, type 4, 606069
SLC41A1	100	100	100	100	No OMIM disease ID
SLC44A1	98,2	98,2	100	100	Neurodegeneration, childhood-onset, with ataxia, tremor, optic atrophy, and cognitive decline, 618868
SLC44A4	100	99,5	100	100	?Deafness, autosomal dominant 72, 617606
SLC45A1	100	99,6	100	100	Intellectual developmental disorder with neuropsychiatric features, 617532
SLC45A2	100	99,9	100	100	[Skin/hair/eye pigmentation 5, dark/fair skin], 227240 [Skin/hair/eye pigmentation 5, black/nonblack hair], 227240 Albinism, oculocutaneous, type IV, 606574 [Skin/hair/eye pigmentation 5, dark/light eyes], 227240
SLC46A1	99,9	98,5	100	100	Folate malabsorption, hereditary, 229050
SLC4A1	100	99,8	96,1	96,1	[Blood group, Swann], 601550 [Blood group, Froese], 601551 [Blood group, Waldner], 112010 Spherocytosis, type 4, 612653 Cryohydrocytosis, 185020 Distal renal tubular acidosis 4 with hemolytic anemia, 611590 Ovalocytosis, SA type, 166900 [Malaria, resistance to], 611162 [Blood group, Diego], 110500 Distal renal tubular acidosis 1, 179800 [Blood group, Wright], 112050
SLC4A11	100	99,9	100	100	Corneal endothelial dystrophy, autosomal recessive, 217700 Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy and perceptive deafness, 217400
SLC4A4	99,8	99,2	100	100	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC52A1	100	100	100	100	Riboflavin deficiency, 615026
SLC52A2	100	100	100	100	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	100	100	100	100	Brown-Vialetto-Van Laere syndrome 1, 211530 ?Fazio-Londe disease, 211500

SLC5A1	100	100	100	100	Glucose/galactose malabsorption, 606824
SLC5A2	100	100	100	100	Renal glucosuria, 233100
SLC5A5	100	99,8	100	100	Thyroid dyshormonogenesis 1, 274400
SLC5A6	100	100	100	100	Neurodegeneration, infantile-onset, biotin-responsive, 618973
SLC5A7	100	99,9	100	100	Neuronopathy, distal hereditary motor, type VIIA, 158580 Myasthenic syndrome, congenital, 20, presynaptic, 617143
SLC6A1	96,7	96,7	100	100	Myoclonic-atonic epilepsy, 616421
SLC6A17	100	100	100	100	Mental retardation, autosomal recessive 48, 616269
SLC6A19	100	100	100	100	Iminoglycinuria, digenic, 242600 Hartnup disorder, 234500 Hyperglycinuria, 138500
SLC6A2	100	99,9	100	100	?Orthostatic intolerance, 604715
SLC6A20	100	99,9	100	100	Iminoglycinuria, digenic, 242600 Hyperglycinuria, 138500
SLC6A3	100	100	100	100	{Nicotine dependence, protection against}, 188890 Parkinsonism-dystonia, infantile, 1, 613135
SLC6A5	100	100	100	100	Hyperekplexia 3, 614618
SLC6A8	93,5	81,6	100	99,8	Cerebral creatine deficiency syndrome 1, 300352
SLC6A9	100	100	100	100	Glycine encephalopathy with normal serum glycine, 617301
SLC7A14	100	100	100	100	Retinitis pigmentosa 68, 615725
SLC7A5	93,9	82,9	100	100	No OMIM disease ID
SLC7A7	100	99,9	100	100	Lysinuric protein intolerance, 222700
SLC7A9	100	99,9	100	100	Cystinuria, 220100
SLC8A1	99,9	99,2	100	100	No OMIM disease ID
SLC9A1	100	100	100	100	Lichtenstein-Knorr syndrome, 616291
SLC9A3	90,6	86	96,4	94,1	Diarrhea 8, secretory sodium, congenital, 616868
SLC9A3R1	100	98,7	100	100	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
SLC9A6	95,2	91,6	100	98,4	Mental retardation, X-linked syndromic, Christianson type, 300243

SLC9A7	97,6	90,3	99,9	99,5	Intellectual developmental disorder, X-linked 108, 301024
SLCO1B1	99,2	93,7	100	100	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO1B3	98,8	90,8	100	100	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO2A1	100	99,4	100	100	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
SLCO5A1	99,7	98,8	100	100	No OMIM disease ID
SLFN14	100	100	100	100	Bleeding disorder, platelet-type, 20, 616913
SLIT3	97,9	95,3	100	100	No OMIM disease ID
SLITRK1	100	100	100	100	?Trichotillomania, 613229 Tourette syndrome, 137580
SLITRK6	100	100	100	100	Deafness and myopia, 221200
SLMAP	99,2	94,6	100	100	No OMIM disease ID
SLN	100	97,6	100	100	No OMIM disease ID
SLURP1	100	99,3	100	100	Meleda disease, 248300
SLX4	100	99,8	100	100	Fanconi anemia, complementation group P, 613951
SMAD1	100	99,1	100	100	No OMIM disease ID
SMAD2	100	99,6	100	100	No OMIM disease ID
SMAD3	99,9	99	100	100	Loeys-Dietz syndrome 3, 613795
SMAD4	100	99,9	100	100	Polyposis, juvenile intestinal, 174900 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350
SMAD6	90,9	81	100	99,6	Aortic valve disease 2, 614823 {Radioulnar synostosis, nonsyndromic}, 179300 {Craniosynostosis 7, susceptibility to}, 617439
SMAD9	100	99,9	100	100	Pulmonary hypertension, primary, 2, 615342
SMARCA1	99,6	97,5	100	99,6	No OMIM disease ID
SMARCA2	96,7	96,2	97,4	96,8	Nicolaides-Baraitser syndrome, 601358
SMARCA4	99,9	99	100	100	{Rhabdoid tumor predisposition syndrome 2}, 613325 Coffin-Siris syndrome 4, 614609

SMARCAD1	99,3	95,8	100	100	Huriez syndrome, 181600 Basan syndrome, 129200 Adermatoglyphia, 136000
SMARCAL1	100	99,9	100	100	Schimke immunoosseous dysplasia, 242900
SMARCB1	100	100	100	100	Rhabdoid tumors, somatic, 609322 {Schwannomatosis-1, susceptibility to}, 162091 Coffin-Siris syndrome 3, 614608 {Rhabdoid tumor predisposition syndrome 1}, 609322
SMARCC2	99	96,6	100	100	Coffin-Siris syndrome 8, 618362
SMARCD1	94,2	89,3	100	100	Coffin-Siris syndrome 11, 618779
SMARCD2	87	85,9	99,6	97	Specific granule deficiency 2, 617475
SMARCE1	95,6	88,8	100	100	{Meningioma, familial, susceptibility to}, 607174 Coffin-Siris syndrome 5, 616938
SMC1A	100	98,7	100	99,8	Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044 Cornelia de Lange syndrome 2, 300590
SMC3	95,2	91	100	100	Cornelia de Lange syndrome 3, 610759
SMCHD1	99,5	96,3	100	100	Fascioscapulohumeral muscular dystrophy 2, digenic, 158901 Bosma arhinia microphthalmia syndrome, 603457
SMDT1	100	100	100	100	No OMIM disease ID
SMG9	100	100	100	100	Heart and brain malformation syndrome, 616920
SMN1	99,5	94,7	94,6	94,6	Spinal muscular atrophy-2, 253550 Spinal muscular atrophy-3, 253400 Spinal muscular atrophy-1, 253300 Spinal muscular atrophy-4, 271150
SMO	97,8	94,7	100	100	Curry-Jones syndrome, somatic mosaic, 601707 Pallister-Hall-like syndrome, 241800 Basal cell carcinoma, somatic, 605462
SMOC1	99,9	98,4	100	100	Microphthalmia with limb anomalies, 206920
SMOC2	76,8	76,6	100	100	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SMPD1	100	100	100	100	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SMPD4	99,4	94,2	100	100	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622

SMPX	100	97,6	100	100	Deafness, X-linked 4, 300066
SMS	91,5	78,5	100	99,9	Mental retardation, X-linked, Snyder-Robinson type, 309583
SMYD2	99,9	97,8	100	100	No OMIM disease ID
SNAI2	100	99,1	100	100	Waardenburg syndrome, type 2D, 608890 Piebaldism, 172800
SNAP25	100	99,9	100	100	?Myasthenic syndrome, congenital, 18, 616330
SNAP29	100	100	100	100	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNCA	79,1	79,1	79,1	79,1	Dementia, Lewy body, 127750 Parkinson disease 1, 168601 Parkinson disease 4, 605543
SNCB	100	99,9	100	100	Dementia, Lewy body, 127750
SNIP1	98,9	97,1	100	100	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501
SNORD118					Leukoencephalopathy, brain calcifications, and cysts, 614561
SNRNP200	99,9	99,1	100	100	Retinitis pigmentosa 33, 610359
SNRPB	100	99,3	100	100	Cerebrocostomandibular syndrome, 117650
SNRPE	99,5	92,6	100	100	Hypotrichosis 11, 615059
SNRPN	100	97	100	100	Prader-Willi syndrome, 176270
SNTA1	87	78,8	99,3	97,2	Long QT syndrome 12, 612955
SNX10	96,2	95,7	100	99,6	Osteopetrosis, autosomal recessive 8, 615085
SNX14	99,6	95,9	100	100	Spinocerebellar ataxia, autosomal recessive 20, 616354
SNX27	100	99,5	100	100	No OMIM disease ID
SOBP	97,5	92,9	97	95,3	Mental retardation, anterior maxillary protrusion, and strabismus, 613671
SOCS1	100	100	100	100	No OMIM disease ID
SOCS4	99,9	99,2	100	100	No OMIM disease ID
SOD1	100	99,9	100	100	Spastic tetraplegia and axial hypotonia, progressive, 618598 Amyotrophic lateral sclerosis 1, 105400
SOD2	100	100	100	100	{Microvascular complications of diabetes 6}, 612634

SOHLH1	99,7	96,5	100	100	Ovarian dysgenesis 5, 617690 Spermatogenic failure 32, 618115
SON	98,8	94,9	100	100	ZTTK syndrome, 617140
SORD	90,3	89,1	97	93,6	Sorbitol dehydrogenase deficiency with peripheral neuropathy, 618912
SORT1	89,5	87,5	100	100	[Low density lipoprotein cholesterol level QTL6], 613589
SOS1	99,8	98,4	100	100	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOS2	100	99,2	100	100	Noonan syndrome 9, 616559
SOST	100	99,5	100	100	Sclerosteosis 1, 269500 Van Buchem disease, 239100 Craniodiaphyseal dysplasia, autosomal dominant, 122860
SOX10	99,9	97,9	100	100	Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 PCWH syndrome, 609136 Waardenburg syndrome, type 4C, 613266
SOX11	100	100	100	100	Coffin-Siris syndrome 9, 615866
SOX17	100	99,5	100	100	Vesicoureteral reflux 3, 613674
SOX18	70,7	55,2	96,1	92,6	Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940 Hypotrichosis-lymphedema-telangiectasia syndrome, 607823
SOX2	100	100	100	100	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SOX3	91,4	75,2	100	99,5	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000
SOX4	97,3	90,9	99,9	98,2	Coffin-Siris syndrome 10, 618506
SOX5	99,9	98,9	100	100	Lamb-Shaffer syndrome, 616803
SOX6	99,9	99,4	100	100	Tolchin-Le Caignec syndrome, 618971
SOX9	100	98,6	100	100	Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290 Campomelic dysplasia with autosomal sex reversal, 114290
SP110	100	100	100	100	{Mycobacterium tuberculosis, susceptibility to}, 607948 Hepatic venoocclusive disease with immunodeficiency, 235550
SP7	100	99,8	100	100	Osteogenesis imperfecta, type XII, 613849
SPAG1	99,3	95,8	99,9	98,6	Ciliary dyskinesia, primary, 28, 615505

SPARC	100	100	100	100	Osteogenesis imperfecta, type XVII, 616507
SPART	99,7	96,8	100	100	Troyer syndrome, 275900
SPAST	99,8	98,7	100	100	Spastic paraplegia 4, autosomal dominant, 182601
SPATA16	100	99,5	100	100	?Spermatogenic failure 6, 102530
SPATA5	100	99,7	100	100	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPATA7	99,8	98,2	100	100	Retinitis pigmentosa, juvenile, autosomal recessive, 604232 Leber congenital amaurosis 3, 604232
SPECC1L	96	95,7	97,8	96,2	Hypertelorism, Teebi type, 145420 ?Facial clefting, oblique, 1, 600251 Opitz GBBB syndrome, type II, 145410
SPEG	96,1	88,7	99,7	99,7	Centronuclear myopathy 5, 615959
SPG11	100	99,3	100	100	Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360 Amyotrophic lateral sclerosis 5, juvenile, 602099
SPG21	99,4	96,8	100	100	Mast syndrome, 248900
SPG7	88,2	86,2	100	100	Spastic paraplegia 7, autosomal recessive, 607259
SPINK1	100	99,3	100	100	{Fibrocalculous pancreatic diabetes, susceptibility to}, 608189 Pancreatitis, hereditary, 167800 Tropical calcific pancreatitis, 608189
SPINK2	99,3	99,1	99,3	99,3	?Spermatogenic failure 29, 618091
SPINK5	99,9	99,5	100	100	Netherton syndrome, 256500
SPINT2	98,5	83,8	100	100	Diarrhea 3, secretory sodium, congenital, syndromic, 270420
SPNS2	92,1	89,3	97,6	95,7	?Deafness, autosomal recessive 115, 618457
SPOCK1	100	99,5	100	100	No OMIM disease ID
SPOP	100	100	100	100	Nabais Sa-de Vries syndrome, type 1, 618828 Nabais Sa-de Vries syndrome, type 2, 618829
SPP2	100	99,9	100	100	No OMIM disease ID
SPPL2A	85,9	74,6	100	100	No OMIM disease ID
SPR	99,8	96,3	100	100	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716

SPRED1	100	98,9	100	100	Legius syndrome, 611431
SPRTN	100	100	100	100	Ruijs-Aalfs syndrome, 616200
SPRY4	100	100	100	100	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
SPTA1	99,9	99,2	100	100	Pyropoikilocytosis, 266140 Elliptocytosis-2, 130600 Spherocytosis, type 3, 270970
SPTAN1	99,1	98,6	100	100	Developmental and epileptic encephalopathy 5, 613477
SPTB	100	100	100	100	Elliptocytosis-3, 617948 Spherocytosis, type 2, 616649 Anemia, neonatal hemolytic, fatal or near-fatal, 617948
SPTBN2	100	99,3	99,9	99,9	Spinocerebellar ataxia, autosomal recessive 14, 615386 Spinocerebellar ataxia 5, 600224
SPTBN4	97,3	91	100	100	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519
SPTLC1	99,2	95,4	100	100	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	100	100	100	100	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SPTLC3	100	99,9	100	100	No OMIM disease ID
SQOR	100	97,8	100	100	No OMIM disease ID
SQSTM1	98,8	95,5	100	100	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 Myopathy, distal, with rimmed vacuoles, 617158 Paget disease of bone 3, 167250
SRC	100	99,8	100	100	Colon cancer, advanced, somatic, 114500 ?Thrombocytopenia 6, 616937
SRCAP	99,4	98,9	100	100	Floating-Harbor syndrome, 136140
SRD5A2	99,9	99	100	100	Pseudovaginal perineoscrotal hypospadias, 264600
SRD5A3	99,9	99,1	100	100	Kahrizi syndrome, 612713 Congenital disorder of glycosylation, type Iq, 612379
SRF	100	99,4	100	99,9	No OMIM disease ID
SRI	99,9	97,8	100	100	No OMIM disease ID
SRP54	99,5	96,5	100	100	Neutropenia, severe congenital, 8, autosomal dominant, 618752

SRP72	97,6	89,7	100	100	Bone marrow failure syndrome 1, 614675
SRPK3	98,7	96,1	100	100	No OMIM disease ID
SRPX2	99,8	96,5	100	100	?Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643
SRY	50	50	60	60	46XY sex reversal 1, 400044 46XX sex reversal 1, 400045
SSBP1	99,8	97,6	100	100	Optic atrophy 13 with retinal and foveal abnormalities, 165510
SSR4	100	99,7	100	100	Congenital disorder of glycosylation, type Iy, 300934
SSTR5	100	99,9	100	100	No OMIM disease ID
SSX1	82,2	82	100	100	?Sarcoma, synovial, 300813
SSX2	65,3	62,6	100	100	?Sarcoma, synovial, 300813
ST14	99,9	98,6	100	100	Ichthyosis, congenital, autosomal recessive 11, 602400
ST3GAL3	68,8	68,6	95,3	95,2	Mental retardation, autosomal recessive 12, 611090 ?Developmental and epileptic encephalopathy 15, 615006
ST3GAL5	85	84,2	98,7	98,4	Salt and pepper developmental regression syndrome, 609056
STAC3	100	100	100	100	Myopathy, congenital, Baily-Bloch, 255995
STAG1	99,6	97,3	100	100	Mental retardation, autosomal dominant 47, 617635
STAG2	97,6	89,4	99,9	98,7	Mullegama-Klein-Martinez syndrome, 301022 Holoprosencephaly 13, X-linked, 301043
STAG3	93,5	93,2	100	100	Premature ovarian failure 8, 615723
STAMBP	100	99,4	100	100	Microcephaly-capillary malformation syndrome, 614261
STAR	100	100	100	100	Lipoid adrenal hyperplasia, 201710
STARD13	99,4	98,1	100	100	No OMIM disease ID
STARD7	98,2	93,1	100	100	Epilepsy, familial adult myoclonic, 2, 607876
STAT1	93,7	91,7	95,7	94,8	Immunodeficiency 31C, chronic mucocutaneous candidiasis, autosomal dominant, 614162 Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796
STAT2	100	99,9	100	100	Immunodeficiency 44, 616636 Pseudo-TORCH syndrome 3, 618886

STAT3	100	99,8	100	100	Hyper-IgE recurrent infection syndrome, 147060 Autoimmune disease, multisystem, infantile-onset, 1, 615952
STAT4	99,9	99,6	100	100	{Systemic lupus erythematosus, susceptibility to, 11}, 612253
STAT5B	100	98,5	100	100	Growth hormone insensitivity with immune dysregulation 2, autosomal dominant, 618985 Growth hormone insensitivity with immune dysregulation 1, autosomal recessive, 245590 Leukemia, acute promyelocytic, somatic, 102578
STAT6	100	99,9	100	100	No OMIM disease ID
STEAP3	100	99,7	100	100	?Anemia, hypochromic microcytic, with iron overload 2, 615234
CXorf56	99,8	96,7	100	100	?Mental retardation, X-linked 107, 301013
STIL	100	99,8	100	100	Microcephaly 7, primary, autosomal recessive, 612703
STIM1	99,8	98	100	100	Myopathy, tubular aggregate, 1, 160565 Immunodeficiency 10, 612783 Stormorken syndrome, 185070
STING1	99,7	95,3	100	100	STING-associated vasculopathy, infantile-onset, 615934
STK11	92,4	91,7	100	100	Testicular tumor, somatic, 273300 Peutz-Jeghers syndrome, 175200 Pancreatic cancer, somatic, 260350 Melanoma, malignant, somatic, 0
STK36	100	99,1	100	100	No OMIM disease ID
STK4	100	99,8	100	100	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STN1	100	100	100	100	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341
STOX1	80,5	80,5	94,8	90	Preeclampsia/eclampsia 4, 609404
STRA6	100	99,8	100	100	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186
STRADA	100	98,9	100	100	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087
STRC	99,9	98	100	100	Deafness, autosomal recessive 16, 603720
STS	97,1	95,5	97,4	97,3	Ichthyosis, X-linked, 308100
STT3A	100	100	100	100	Congenital disorder of glycosylation, type Iw, 615596
STT3B	100	99,6	100	100	?Congenital disorder of glycosylation, type Ix, 615597

STUB1	100	98,7	100	100	Spinocerebellar ataxia, autosomal recessive 16, 615768 ?Spinocerebellar ataxia 48, 618093
STX11	100	100	100	100	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STX16	100	98,6	100	100	Pseudohypoparathyroidism, type IB, 603233
STX1B	100	100	100	100	Generalized epilepsy with febrile seizures plus, type 9, 616172
STXBP1	96,8	96,5	100	100	Developmental and epileptic encephalopathy 4, 612164
STXBP2	82,1	79,7	99,3	97,1	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
SUCLA2	89,5	82,2	99,9	99,8	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	99,9	99,8	100	100	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUCLG2	96,7	86,3	100	100	No OMIM disease ID
SUFU	100	100	100	100	Basal cell nevus syndrome, 109400 Medulloblastoma, desmoplastic, 155255 {Meningioma, familial, susceptibility to}, 607174 Joubert syndrome 32, 617757
SUGCT	99,9	98,5	100	100	Glutaric aciduria III, 231690
SULF1	99,9	99,3	100	100	No OMIM disease ID
SULT2B1	100	100	100	100	Ichthyosis, congenital, autosomal recessive 14, 617571
SUMF1	97,5	90,8	100	100	Multiple sulfatase deficiency, 272200
SUMO1	67,2	49,9	69,4	69,4	?Orofacial cleft 10, 613705
SUN5	100	99,8	100	100	Spermatogenic failure 16, 617187
SUOX	100	100	100	100	Sulfite oxidase deficiency, 272300
SUPT16H	98,6	93,6	100	100	No OMIM disease ID
SURF1	89,4	88,2	100	100	Charcot-Marie-Tooth disease, type 4K, 616684 Mitochondrial complex IV deficiency, nuclear type 1, 220110
SUZ12	91,4	86,2	100	100	Imagawa-Matsumoto syndrome, 618786
SVBP	100	100	100	100	Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569
SYCE1	100	98,6	100	100	?Premature ovarian failure 12, 616947 ?Spermatogenic failure 15, 616950

SYCP3	99,7	98,2	100	100	Spermatogenic failure 4, 270960 Pregnancy loss, recurrent, 4, 270960
SYN1	81,9	73,2	100	99,6	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNCRIP	97,8	87,2	100	100	No OMIM disease ID
SYNE1	98,2	97,8	98,8	98,8	Arthrogryposis multiplex congenita 3, myogenic type, 618484 Spinocerebellar ataxia, autosomal recessive 8, 610743 Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998
SYNE2	99,7	98,1	100	99,9	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999
SYNE4	99,7	97	100	100	Deafness, autosomal recessive 76, 615540
SYNGAP1	99,4	98,1	100	100	Mental retardation, autosomal dominant 5, 612621
SYNJ1	99,9	99,4	100	100	Developmental and epileptic encephalopathy 53, 617389 Parkinson disease 20, early-onset, 615530
SYP	99,9	96,7	100	100	Mental retardation, X-linked 96, 300802
SYT1	99,8	98,5	100	100	Baker-Gordon syndrome, 618218
SYT14	61	60,5	100	100	?Spinocerebellar ataxia, autosomal recessive 11, 614229
SYT2	99,9	99	100	100	Myasthenic syndrome, congenital, 7, presynaptic, 616040
SZT2	99,6	99,5	100	99,9	Developmental and epileptic encephalopathy 18, 615476
TAB2	100	99,7	100	100	Congenital heart defects, nonsyndromic, 2, 614980
TAC3	100	99,6	100	100	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACO1	98,4	93	100	100	Mitochondrial complex IV deficiency, nuclear type 8, 619052
TACR3	100	100	100	100	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
TACSTD2	99	96,4	100	100	Corneal dystrophy, gelatinous drop-like, 204870
TAF1	99,8	97,7	100	100	Dystonia-Parkinsonism, X-linked, 314250 Mental retardation, X-linked, syndromic 33, 300966
TAF13	100	100	100	100	Mental retardation, autosomal recessive 60, 617432
TAF1C	100	100	100	100	No OMIM disease ID
TAF2	99,9	98,6	100	100	Mental retardation, autosomal recessive 40, 615599
TAF4B	97,4	93,1	100	100	?Spermatogenic failure 13, 615841

TAF6	99,8	98,9	100	100	Alazami-Yuan syndrome, 617126
TAL1	87,1	73,1	100	100	Leukemia, T-cell acute lymphocytic, somatic, 613065
TAL2	100	100	100	100	Leukemia, T-cell acute lymphocytic, somatic, 613065
TALDO1	100	97,9	100	100	Transaldolase deficiency, 606003
TANC2	100	99,5	100	100	Intellectual developmental disorder with autistic features and language delay, with or without seizures, 618906
TANGO2	100	99,3	100	100	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TAOK1	99,5	97,9	100	100	No OMIM disease ID
TAP1	100	99,2	100	100	Bare lymphocyte syndrome, type I, 604571
TAP2	99,9	99,3	100	100	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	96,5	95,5	96,6	96,6	Bare lymphocyte syndrome, type I, 604571
TAPT1	91,7	86,9	98,5	94,8	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897
TARDBP	100	100	100	100	Frontotemporal lobar degeneration, TARDBP-related, 612069 Amyotrophic lateral sclerosis 10, with or without FTD, 612069
TARS1	99,9	98,1	100	100	Trichothiodystrophy 7, nonphotosensitive, 618546
TARS2	100	99,3	100	100	?Combined oxidative phosphorylation deficiency 21, 615918
TASP1	99,7	98,8	100	100	Suleiman-El-Hattab syndrome, 618950
TAT	100	100	100	100	Tyrosinemia, type II, 276600
TAX1BP3	99,8	99,1	100	100	No OMIM disease ID
TAZ	99,1	95,5	100	100	Barth syndrome, 302060
TBC1D20	94,2	94,2	100	99,9	Warburg micro syndrome 4, 615663
TBC1D23	99,7	97,2	100	100	Pontocerebellar hypoplasia, type 11, 617695
TBC1D24	100	100	100	100	Developmental and epileptic encephalopathy 16, 615338 Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp, 608105 DOORS syndrome, 220500 Deafness, autosomal dominant 65, 616044 Myoclonic epilepsy, infantile, familial, 605021 Deafness , autosomal recessive 86, 614617

TBC1D2B	99,3	97,6	98,5	97,8	No OMIM disease ID
TBC1D32	99	95,8	100	100	No OMIM disease ID
TBC1D7	100	99,3	100	100	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000
TBC1D8B	98,5	93,2	100	100	Nephrotic syndrome, type 20, 301028
TBCD	96,2	94,4	100	100	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TBCE	99,8	97,5	100	100	Kenny-Caffey syndrome, type 1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
TBCK	99,1	96,8	100	100	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900
TBK1	99,7	97,2	100	100	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439 {Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 8}, 617900
TBL1X	96	90,7	100	100	Hypothyroidism, congenital, nongoitrous, 8, 301033
TBL1XR1	96,5	84,9	100	100	Pierpont syndrome, 602342 Mental retardation, autosomal dominant 41, 616944
TBL1Y	49,4	45,3	60	59,9	?Deafness, Y-linked 2, 400047
TBP	100	99,9	100	100	{Parkinson disease, susceptibility to}, 168600 Spinocerebellar ataxia 17, 607136
TBR1	99,9	97,9	100	100	Intellectual developmental disorder with autism and speech delay, 606053
TBX1	87	77,5	94	89,9	Conotruncal anomaly face syndrome, 217095 Velocardiofacial syndrome, 192430 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500
TBX15	100	99,9	100	100	Cousin syndrome, 260660
TBX18	99,5	97,1	100	100	Congenital anomalies of kidney and urinary tract 2, 143400
TBX19	100	100	100	100	Adrenocorticotropic hormone deficiency, 201400
TBX2	99,9	97,5	99	96,9	Vertebral anomalies and variable endocrine and T-cell dysfunction, 618223
TBX20	100	99,7	100	100	Atrial septal defect 4, 611363
TBX21	95,4	86,6	100	100	{Asthma, aspirin-induced, susceptibility to}, 208550 Asthma and nasal polyps, 208550
TBX22	99,2	95,7	100	100	?Abruzzo-Erickson syndrome, 302905 Cleft palate with ankyloglossia, 303400

TBX3	99,2	96,8	100	100	Ulnar-mammary syndrome, 181450
TBX4	97,6	95,1	100	100	Amelia, posterior, with pelvic and pulmonary hypoplasia syndrome, 601360 Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension, 147891
TBX5	100	100	100	100	Holt-Oram syndrome, 142900
TBX6	99,5	95,5	100	100	Spondylocostal dysostosis 5, 122600
TBXA2R	97,6	93,8	99,8	98	{Bleeding disorder, platelet-type, 13, susceptibility to}, 614009
TBXAS1	100	100	100	100	Ghosal hematodiaphyseal syndrome, 231095
TBXT	99,4	96,9	100	100	{Neural tube defects, susceptibility to}, 182940 Sacral agenesis with vertebral anomalies, 615709
TCAP	100	100	100	100	Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954
TCF12	100	99,9	100	100	Craniosynostosis 3, 615314
TCF20	100	100	100	100	Developmental delay with variable intellectual impairment and behavioral abnormalities, 618430
TCF3	97,1	94	100	100	Agammaglobulinemia 8, autosomal dominant, 616941
TCF4	100	99,8	100	100	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954
TCF7L2	99,9	98,8	100	100	{Diabetes mellitus, type 2, susceptibility to}, 125853
TCHH	100	98,8	100	100	?Uncombable hair syndrome 3, 617252
TCIRG1	97,6	90,1	100	100	Osteopetrosis, autosomal recessive 1, 259700
TCN2	100	100	100	100	Transcobalamin II deficiency, 275350
TCOF1	99,7	98,6	100	100	Treacher Collins syndrome 1, 154500
TCTN1	96,7	93	94,7	94,7	Joubert syndrome 13, 614173
TCTN2	100	99,5	100	100	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	100	100	100	100	Orofaciodigital syndrome IV, 258860 Joubert syndrome 18, 614815
TDGF1	99,9	96,7	100	100	Forebrain defects, 0
TDP1	99,9	99,5	100	100	?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250
TDP2	100	99,4	100	100	Spinocerebellar ataxia, autosomal recessive 23, 616949

TDRD7	99,9	99,1	100	100	Cataract 36, 613887
TDRD9	99,3	98,2	100	99,9	?Spermatogenic failure 30, 618110
TDRKH	94,7	94,7	100	100	No OMIM disease ID
TEAD1	100	99,9	100	100	Sveinsson chorioretinal atrophy, 108985
TECPR2	100	100	100	100	Spastic paraplegia 49, autosomal recessive, 615031
TECR	100	99	100	100	Mental retardation, autosomal recessive 14, 614020
TECRL	96,3	89,3	100	100	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021
TECTA	100	99,9	100	100	Deafness, autosomal dominant 8/12, 601543 Deafness, autosomal recessive 21, 603629
TEK	100	100	100	100	Glaucoma 3, primary congenital, E, 617272 Venous malformations, multiple cutaneous and mucosal, 600195
TELO2	99,7	96,2	100	100	You-Hoover-Fong syndrome, 616954
TENM3	100	99,7	100	100	Microphthalmia, syndromic 15, 615145 ?Microphthalmia, isolated, with coloboma 9, 615145
TENM4	100	99,6	100	100	Essential tremor, hereditary, 5, 616736
TENT5A	100	99,7	100	100	Osteogenesis imperfecta, type XVIII, 617952
TERC					{Aplastic anemia}, 614743 Dyskeratosis congenita, autosomal dominant 1, 127550 {Pulmonary fibrosis, idiopathic, susceptibility to}, 614743
TERF2IP	99,9	97,8	83,7	83,7	No OMIM disease ID
TERT	96,2	94,5	100	100	{Melanoma, cutaneous malignant, 9}, 615134 {Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1}, 614742 {Leukemia, acute myeloid}, 601626 {Dyskeratosis congenita, autosomal recessive 4}, 613989 {Dyskeratosis congenita, autosomal dominant 2}, 613989
TET2	100	100	100	100	Immunodeficiency 75, 619126 Myelodysplastic syndrome, somatic, 614286
TET3	94,4	94,4	100	100	Beck-Fahrner syndrome, 618798
TEX11	93,8	88,2	97,1	97	Spermatogenic failure, X-linked, 2, 309120
TEX14	99,9	98,9	100	100	Spermatogenic failure 23, 617707

TEX15	100	99,7	100	100	Spermatogenic failure 25, 617960
TF	100	100	100	100	Atransferrinemia, 209300
TFAM	97,5	83,5	100	100	?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156
TFAP2A	99,4	94,3	100	100	Branchiooculofacial syndrome, 113620
TFAP2B	99,9	98,6	100	100	Char syndrome, 169100 Patent ductus arteriosus 2, 617035
TFB2M	100	99,1	100	100	No OMIM disease ID
TFE3	99,3	94,2	100	100	Renal cell carcinoma, papillary, 1, 300854
TFG	96,9	96,3	100	100	?Spastic paraplegia 57, autosomal recessive, 615658 Hereditary motor and sensory neuropathy, Okinawa type, 604484
TFR2	99,1	97,8	100	100	Hemochromatosis, type 3, 604250
TFRC	100	99,8	100	100	Immunodeficiency 46, 616740
TG	100	99,4	100	100	{Autoimmune thyroid disease, susceptibility to, 3}, 608175 Thyroid dyshormonogenesis 3, 274700
TGDS	99,4	96,8	100	100	Catel-Manzke syndrome, 616145
TGFB1	100	99,9	100	100	{Cystic fibrosis lung disease, modifier of}, 219700 Camurati-Engelmann disease, 131300 Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213
TGFB2	100	100	100	100	Loeys-Dietz syndrome 4, 614816
TGFB3	100	100	100	100	Loeys-Dietz syndrome 5, 615582 Arrhythmogenic right ventricular dysplasia 1, 107970
TGFBI	99,5	94,6	100	100	Corneal dystrophy, lattice type IIIA, 608471 Corneal dystrophy, Groenouw type I, 121900 Corneal dystrophy, lattice type I, 122200 Corneal dystrophy, Reis-Bucklers type, 608470 Corneal dystrophy, Thiel-Behnke type, 602082 Corneal dystrophy, epithelial basement membrane, 121820 Corneal dystrophy, Avellino type, 607541
TGFBR1	93,7	93,6	99	96,3	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800
TGFBR2	100	100	100	100	Esophageal cancer, somatic, 133239 Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Loeys-Dietz syndrome 2, 610168

TGIF1	100	100	100	100	Holoprosencephaly 4, 142946
TGM1	100	99,9	100	100	Ichthyosis, congenital, autosomal recessive 1, 242300
TGM3	100	99,7	100	100	?Uncombable hair syndrome 2, 617251
TGM5	100	99,7	100	100	Peeling skin syndrome 2, 609796
TGM6	99,7	97,3	100	100	Spinocerebellar ataxia 35, 613908
TH	99,3	96,1	100	100	Segawa syndrome, recessive, 605407
THAP1	100	100	100	100	Dystonia 6, torsion, 602629
THBD	100	99,7	100	100	Thrombophilia due to thrombomodulin defect, 614486 {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
THBS4	99,9	99,4	100	100	No OMIM disease ID
THG1L	100	100	100	100	Spinocerebellar ataxia, autosomal recessive 28, 618800
THOC1	99,7	97,8	100	100	No OMIM disease ID
THOC2	98,8	93,7	100	100	Mental retardation, X-linked 12/35, 300957
THOC6	100	100	100	100	Beaulieu-Boycott-Innes syndrome, 613680
THPO	81,4	81	100	100	Thrombocythemia 1, 187950
THRA	100	99,6	100	100	Hypothyroidism, congenital, nongoitrous, 6, 614450
THRB	100	99,7	100	100	Thyroid hormone resistance, 188570 Thyroid hormone resistance, selective pituitary, 145650 Thyroid hormone resistance, autosomal recessive, 274300
TIA1	99,7	97,8	100	100	Welander distal myopathy, 604454
TICAM1	100	100	100	100	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 6}, 614850
TIMM22	100	99,7	100	100	?Combined oxidative phosphorylation deficiency 43, 618851
TIMM44	100	99,4	100	100	No OMIM disease ID
TIMM50	98,3	94,4	100	100	3-methylglutaconic aciduria, type IX, 617698
TIMM8A	98,1	90,6	100	100	Mohr-Tranebjaerg syndrome, 304700
TIMMDC1	100	100	100	100	Mitochondrial complex I deficiency, nuclear type 31, 618251

TIMP1	100	100	100	100	No OMIM disease ID
TIMP2	100	99	99,8	98,5	No OMIM disease ID
TIMP3	100	100	100	100	Sorsby fundus dystrophy, 136900
TINF2	100	100	100	100	Revesz syndrome, 268130 Dyskeratosis congenita, autosomal dominant 3, 613990
TIRAP	100	100	100	100	{Bacteremia, protection against}, 614382 {Malaria, protection against}, 611162 {Tuberculosis, protection against}, 607948
TJP1	100	99,7	100	100	No OMIM disease ID
TJP2	92,8	92,5	98,8	98,8	Cholestasis, progressive familial intrahepatic 4, 615878 Hypercholanemia, familial, 607748
TK2	99,2	96,3	100	100	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560 ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069
TKFC	100	99,5	100	100	Triokinase and FMN cyclase deficiency syndrome, 618805
TKT	98,7	97,8	98,7	98,7	Short stature, developmental delay, and congenital heart defects, 617044
TLE6	100	98,8	100	100	Preimplantation embryonic lethality, 616814
TLK2	99,1	95,1	100	100	Mental retardation, autosomal dominant 57, 618050
TLL1	100	100	100	100	Atrial septal defect 6, 613087
TLN1	100	99,3	100	100	No OMIM disease ID
TLR3	100	99,6	100	100	{HIV1 infection, resistance to}, 609423 {Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 2}, 613002
TLR4	100	99,8	100	100	No OMIM disease ID
TLR7	100	99,9	100	100	Immunodeficiency 74, COVID19-related, X-linked, 301051
TLR8	100	99,8	100	100	No OMIM disease ID
TMC1	99,7	97,3	100	100	Deafness, autosomal recessive 7, 600974 Deafness, autosomal dominant 36, 606705
TMC6	100	99,3	100	100	Epidermodysplasia verruciformis, 226400
TMC8	100	98,7	100	100	Epidermodysplasia verruciformis 2, 618231
TMCO1	88	87,4	88	88	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980

TMCO3	100	99,4	100	100	No OMIM disease ID
TMEM106B	99,9	98,8	100	100	Leukodystrophy, hypomyelinating, 16, 617964
TMEM107	100	100	100	100	Orofaciodigital syndrome XVI, 617563 Meckel syndrome 13, 617562 ?Joubert syndrome 29, 617562
TMEM126A	96,3	84,4	100	100	Optic atrophy 7, 612989
TMEM126B	99,8	97,4	100	100	Mitochondrial complex I deficiency, nuclear type 29, 618250
TMEM127	99,5	96,5	100	100	{Pheochromocytoma, susceptibility to}, 171300
TMEM132E	96,9	93,5	100	100	Deafness, autosomal recessive 99, 618481
TMEM138	100	99,1	100	100	Joubert syndrome 16, 614465
TMEM14C	100	99,8	100	100	No OMIM disease ID
TMEM165	100	100	100	100	Congenital disorder of glycosylation, type IIk, 614727
TMEM186	100	100	100	100	No OMIM disease ID
TMEM199	100	99,9	100	100	Congenital disorder of glycosylation, type IIp, 616829
TMEM216	99,9	98,1	100	100	Meckel syndrome 2, 603194 Joubert syndrome 2, 608091
TMEM218	100	99,9	100	100	No OMIM disease ID
TMEM231	100	99,6	100	100	Meckel syndrome 11, 615397 Joubert syndrome 20, 614970
TMEM237	100	99,9	100	100	Joubert syndrome 14, 614424
TMEM240	100	100	100	100	Spinocerebellar ataxia 21, 607454
TMEM260	97,5	93,4	100	100	Structural heart defects and renal anomalies syndrome, 617478
TMEM38B	100	99,9	100	100	Osteogenesis imperfecta, type XIV, 615066
TMEM43	99,9	98,9	100	100	Emery-Dreifuss muscular dystrophy 7, AD, 614302 Arrhythmogenic right ventricular dysplasia 5, 604400
TMEM63A	100	99,9	100	100	Leukodystrophy, hypomyelinating, 19, transient infantile, 618688
TMEM65	88	81,3	92,5	85,3	No OMIM disease ID

TMEM67	99,5	95	100	99,9	Meckel syndrome 3, 607361 COACH syndrome 1, 216360 ?RHYNS syndrome, 602152 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991 Joubert syndrome 6, 610688
TMEM70	98	93,9	100	100	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMEM94	100	100	100	100	Intellectual developmental disorder with cardiac defects and dysmorphic facies, 618316
TMEM98	99,3	97,8	100	100	Nanophthalmos 4, 615972
TMIE	99,2	95,1	100	100	Deafness, autosomal recessive 6, 600971
TMLHE	99,5	97,1	100	99,9	{Autism, susceptibility to, X-linked 6}, 300872
TMPO	98,4	94,7	100	100	No OMIM disease ID
TMPRSS15	98,5	95,2	100	100	Enterokinase deficiency, 226200
TMPRSS3	100	99,9	100	100	Deafness, autosomal recessive 8/10, 601072
TMPRSS6	99,9	99,1	100	100	Iron-refractory iron deficiency anemia, 206200
TMTC2	97,5	97,5	97,5	97,5	No OMIM disease ID
TMTC3	99,6	96,5	100	100	Lissencephaly 8, 617255
TMX2	100	99,8	100	100	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730
TNC	100	99,8	100	100	Deafness, autosomal dominant 56, 615629
TNFAIP3	100	100	100	100	Autoinflammatory syndrome, familial, Behcet-like, 616744
TNFRSF10B	100	100	100	100	Squamous cell carcinoma, head and neck, 275355
TNFRSF11A	94,6	93,3	99,2	98	Osteolysis, familial expansile, 174810 {Paget disease of bone 2, early-onset}, 602080 Osteopetrosis, autosomal recessive 7, 612301
TNFRSF11B	100	100	100	100	Paget disease of bone 5, juvenile-onset, 239000
TNFRSF13B	100	100	100	100	Immunoglobulin A deficiency 2, 609529 Immunodeficiency, common variable, 2, 240500
TNFRSF13C	80,1	75,4	100	99,9	Immunodeficiency, common variable, 4, 613494

TNFRSF1A	90,6	87,6	92,8	92,8	Periodic fever, familial, 142680 {Multiple sclerosis, susceptibility to, 5}, 614810
TNFRSF4	99,4	95,4	100	100	?Immunodeficiency 16, 615593
TNFRSF9	100	100	100	100	No OMIM disease ID
TNFSF11	100	99,9	100	100	Osteopetrosis, autosomal recessive 2, 259710
TNFSF12	98	93,6	100	100	No OMIM disease ID
TNIK	100	99,3	100	100	Mental retardation, autosomal recessive 54, 617028
TNNC1	100	100	100	100	Cardiomyopathy, hypertrophic, 13, 613243 Cardiomyopathy, dilated, 1Z, 611879
TNNI2	100	99,7	100	100	Arthrogryposis, distal, type 2B1, 601680
TNNI3	99,7	95,4	100	100	Cardiomyopathy, hypertrophic, 7, 613690 ?Cardiomyopathy, dilated, 2A, 611880 Cardiomyopathy, dilated, 1FF, 613286 Cardiomyopathy, familial restrictive, 1, 115210
TNNI3K	100	99,4	100	100	Cardiac conduction disease with or without dilated cardiomyopathy, 616117
TNNT1	99,9	97,6	100	100	Nemaline myopathy 5, Amish type, 605355
TNNT2	94,8	91,1	100	99,3	Cardiomyopathy, familial restrictive, 3, 612422 Cardiomyopathy, hypertrophic, 2, 115195 Left ventricular noncompaction 6, 601494 Cardiomyopathy, dilated, 1D, 601494
TNNT3	100	99,7	100	100	Arthrogryposis, distal, type 2B2, 618435
TNPO3	100	99,9	100	100	Muscular dystrophy, limb-girdle, autosomal dominant 2, 608423
TNRC6A	99,9	99,3	100	100	?Epilepsy, familial adult myoclonic, 6, 618074
TNRC6B	100	99,8	100	100	No OMIM disease ID
TNS2	100	99,9	100	100	No OMIM disease ID
TNXB	99,1	93,7	100	99,9	Ehlers-Danlos syndrome, classic-like, 1, 606408 Vesicoureteral reflux 8, 615963
TOE1	100	100	100	100	Pontocerebellar hypoplasia, type 7, 614969
TOGARAM1	99,6	98,1	100	100	No OMIM disease ID
TOMM70	100	99,8	100	100	No OMIM disease ID

TONSL	99,8	97,8	100	100	Spondyloepimetaphyseal dysplasia, sponastrime type, 271510
TOP1	99,9	98,6	100	100	DNA topoisomerase I, camptothecin-resistant, 0
TOP2A	100	99,3	100	100	DNA topoisomerase II, resistance to inhibition of, by amsacrine, 0
TOP2B	99,4	96,3	100	100	No OMIM disease ID
TOP3A	100	98,7	100	100	Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097 ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098
TOPORS	100	100	100	100	Retinitis pigmentosa 31, 609923
TOR1A	91,3	91,2	91,4	91,3	Dystonia-1, torsion, 128100 Arthrogryposis multiplex congenita 5, 618947 {Dystonia-1, modifier of}, 0
TOR1AIP1	99,9	98	100	100	?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072
TP53	99,9	97,7	91,7	91,7	{Adrenocortical carcinoma, pediatric}, 202300 {Glioma susceptibility 1}, 137800 {Basal cell carcinoma 7}, 614740 Bone marrow failure syndrome 5, 618165 {Colorectal cancer}, 114500 Nasopharyngeal carcinoma, somatic, 607107 Breast cancer, somatic, 114480 {Osteosarcoma}, 259500 {Choroid plexus papilloma}, 260500 Li-Fraumeni syndrome, 151623 Hepatocellular carcinoma, somatic, 114550 Pancreatic cancer, somatic, 260350
TP53RK	92,5	79,6	100	100	Galloway-Mowat syndrome 4, 617730
TP63	100	100	100	100	Limb-mammary syndrome, 603543 Orofacial cleft 8, 618149 Split-hand/foot malformation 4, 605289 Hay-Wells syndrome, 106260 Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 Rapp-Hodgkin syndrome, 129400 ADULT syndrome, 103285
TPCN2	95,1	92,4	100	100	[Skin/hair/eye pigmentation 10, blond/brown hair], 612267
TPI1	99,8	97,5	100	100	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512

TPK1	99,8	99	100	100	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TPM1	100	99,4	100	99,9	Left ventricular noncompaction 9, 611878 Cardiomyopathy, hypertrophic, 3, 115196 Cardiomyopathy, dilated, 1Y, 611878
TPM2	100	100	100	100	Nemaline myopathy 4, autosomal dominant, 609285 Arthrogryposis, distal, type 2B4, 108120 Arthrogryposis, distal, type 1A, 108120 CAP myopathy 2, 609285
TPM3	89,2	87,2	100	100	CAP myopathy 1, 609284 Nemaline myopathy 1, autosomal dominant or recessive, 609284 Myopathy, congenital, with fiber-type disproportion, 255310
TPM4	82,9	70,4	100	100	No OMIM disease ID
TPMT	99,1	90,1	100	100	{Thiopurines, poor metabolism of, 1}, 610460
TPO	99,9	98,2	100	100	Thyroid dyshormonogenesis 2A, 274500
TPP1	100	100	100	100	Spinocerebellar ataxia, autosomal recessive 7, 609270 Ceroid lipofuscinosis, neuronal, 2, 204500
TPP2	99,2	96,8	100	100	No OMIM disease ID
TPRKB	81,1	75,9	81,9	81,9	Galloway-Mowat syndrome 5, 617731
TPRN	87,9	79,3	94,4	89,8	Deafness, autosomal recessive 79, 613307
TRAC	100	100	100	100	Immunodeficiency 7, TCR-alpha/beta deficient, 615387
TRAF3	100	99,9	100	100	{?Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 5}, 614849
TRAF3IP1	99,6	97,6	100	100	Senior-Loken syndrome 9, 616629
TRAF3IP2	100	99,3	100	100	?Candidiasis, familial, 8, 615527 {Psoriasis susceptibility 13}, 614070
TRAF6	97,1	88,9	100	100	No OMIM disease ID
TRAF7	100	99,8	100	100	Cardiac, facial, and digital anomalies with developmental delay, 618164
TRAIP	100	100	100	100	Seckel syndrome 9, 616777
TRAK1	93,3	92,9	100	99,9	Developmental and epileptic encephalopathy 68, 618201
TRAP1	97,2	95,7	100	100	No OMIM disease ID
TRAPPC11	100	99,2	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356

TRAPPC12	100	99,6	100	100	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669
MAP11	100	99,4	100	100	?Microcephaly 25, primary, autosomal recessive, 618351
TRAPPC2	89,7	69,6	100	100	Spondyloepiphyseal dysplasia tarda, 313400
TRAPPC2L	100	100	100	100	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331
TRAPPC4	100	100	100	100	Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy, 618741
TRAPPC6B	99,9	98	100	100	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862
TRAPPC9	100	99,6	100	100	Mental retardation, autosomal recessive 13, 613192
TRDN	96,2	86,7	100	100	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441
TREH	96,9	92,1	100	100	Trehalase deficiency, 612119
TREM2	100	99,8	100	100	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193
TREX1	100	100	100	100	{Systemic lupus erythematosus, susceptibility to}, 152700 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
TRH	99,6	96,5	100	100	Thyrotropin-releasing hormone deficiency, 275120
TRHR	100	99,2	100	100	Hypothyroidism, congenital, nongoitrous, 7, 618573
TRIB1	99,5	91,6	100	100	No OMIM disease ID
TRIM2	93,9	93,3	93,9	93,9	Charcot-Marie-Tooth disease, type 2R, 615490
TRIM22	100	100	100	100	No OMIM disease ID
TRIM28	96,8	95,2	99,8	99,3	No OMIM disease ID
TRIM32	100	100	100	100	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRIM36	100	99,2	100	100	?Anencephaly, 206500
TRIM37	98,6	98,1	98,7	98,7	Mulibrey nanism, 253250
TRIM44	99,8	96,3	100	100	?Aniridia 3, 617142
TRIM63	100	100	100	100	No OMIM disease ID
TRIM8	99,3	97,2	100	100	No OMIM disease ID

TRIO	99,2	97,5	99,3	98,4	Intellectual developmental disorder, autosomal dominant 44, with microcephaly, 617061 Intellectual developmental disorder, autosomal dominant 63, with macrocephaly, 618825
TRIOBP	97,8	96,1	99,9	99,6	Deafness, autosomal recessive 28, 609823
TRIP11	98,4	94	100	100	Osteochondrodysplasia, 184260 Achondrogenesis, type IA, 200600
TRIP12	99,9	99,2	100	100	Mental retardation, autosomal dominant 49, 617752
TRIP13	100	100	100	100	Mosaic variegated aneuploidy syndrome 3, 617598 Oocyte maturation defect 9, 619011
TRIP4	100	99,1	100	100	Spinal muscular atrophy with congenital bone fractures 1, 616866 ?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066
TRIT1	100	100	100	100	Combined oxidative phosphorylation deficiency 35, 617873
TRMT1	99,4	96,2	100	100	Mental retardation, autosomal recessive 68, 618302
TRMT10A	100	99,7	100	100	Microcephaly, short stature, and impaired glucose metabolism 1, 616033
TRMT10C	100	100	100	99,9	Combined oxidative phosphorylation deficiency 30, 616974
TRMT5	100	99,3	100	100	Combined oxidative phosphorylation deficiency 26, 616539
TRMU	100	100	100	99,9	Liver failure, transient infantile, 613070 {Deafness, mitochondrial, modifier of}, 580000
TRNT1	99,5	96,5	100	100	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 Retinitis pigmentosa and erythrocytic microcytosis, 616959
TRPA1	96,1	89,8	100	100	?Episodic pain syndrome, familial, 1, 615040
TRPC3	99,7	98	100	100	?Spinocerebellar ataxia 41, 616410
TRPC6	98,2	96,1	100	100	Glomerulosclerosis, focal segmental, 2, 603965
TRPM1	100	99,8	100	100	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TRPM3	100	99,5	100	100	No OMIM disease ID
TRPM4	100	99,5	100	100	Erythrokeratodermia veriabilis et progressiva 6, 618531 Progressive familial heart block, type IB, 604559
TRPM6	99,9	99,5	100	100	Hypomagnesemia 1, intestinal, 602014
TRPM8	99,8	98,8	100	100	No OMIM disease ID
TRPS1	100	99,9	100	100	Trichorhinophalangeal syndrome, type I, 190350 Trichorhinophalangeal syndrome, type III, 190351

TRPV1	100	99,6	100	100	No OMIM disease ID
TRPV3	99,8	98,5	97,1	97,1	?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400 Olmsted syndrome, 614594
TRPV4	100	99,9	100	100	Spondylometaphyseal dysplasia, Kozlowski type, 184252 Parastremmatic dwarfism, 168400 SED, Maroteaux type, 184095 Neuronopathy, distal hereditary motor, type VIII, 600175 [Sodium serum level QTL 1], 613508 Scapuloperoneal spinal muscular atrophy, 181405 Metatropic dysplasia, 156530 Digital arthropathy-brachydactyly, familial, 606835 Hereditary motor and sensory neuropathy, type IIc, 606071 Brachyolmia type 3, 113500 ?Avascular necrosis of femoral head, primary, 2, 617383
TRPV6	100	99,5	99,9	98,9	Hyperparathyroidism, transient neonatal, 618188
TRRAP	99,9	99,5	100	100	Developmental delay with or without dysmorphic facies and autism, 618454 ?Deafness, autosomal dominant 75, 618778
TSC1	99,8	98,8	100	100	Tuberous sclerosis-1, 191100 Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690
TSC2	100	99,6	100	100	Tuberous sclerosis-2, 613254 ?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690
TSEN15	79	77,2	100	100	Pontocerebellar hypoplasia, type 2F, 617026
TSEN2	100	99,6	100	100	Pontocerebellar hypoplasia type 2B, 612389
TSEN34	90,8	86,4	100	100	?Pontocerebellar hypoplasia type 2C, 612390
TSEN54	96,3	94,3	99,9	98,9	Pontocerebellar hypoplasia type 4, 225753 Pontocerebellar hypoplasia type 2A, 277470 ?Pontocerebellar hypoplasia type 5, 610204
TSFM	100	99,5	94,9	94,9	Combined oxidative phosphorylation deficiency 3, 610505
TSGA10	89,5	88,8	100	100	?Spermatogenic failure 26, 617961
TSHB	100	100	100	100	Hypothyroidism, congenital, nongoitrous 4, 275100
TSHR	96	95,4	100	100	Hyperthyroidism, nonautoimmune, 609152 Hypothyroidism, congenital, nongoitrous, 1, 275200

					Hyperthyroidism, familial gestational, 603373 Thyroid adenoma, hyperfunctioning, somatic, 0 Thyroid carcinoma with thyrotoxicosis, 0
TSHZ1	98,8	98,8	100	100	Aural atresia, congenital, 607842
TSPAN12	100	99,8	100	100	Exudative vitreoretinopathy 5, 613310
TSPAN7	100	100	100	100	Mental retardation, X-linked 58, 300210
TSPEAR	100	99,2	100	100	Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180 ?Deafness, autosomal recessive 98, 614861
TSPYL1	100	100	100	100	Sudden infant death with dysgenesis of the testes syndrome, 608800
TSR2	100	100	100	99,9	?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946
TTBK2	99,8	97,6	100	100	Spinocerebellar ataxia 11, 604432
TTC19	81,5	73,8	100	99,2	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTC21A	100	100	100	100	Spermatogenic failure 37, 618429
TTC21B	99,9	99,3	100	100	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
TTC26	99,9	98,8	100	100	No OMIM disease ID
TTC37	100	99,3	100	100	Trichohepatoenteric syndrome 1, 222470
TTC5	100	99,9	100	100	No OMIM disease ID
TTC7A	99,3	95,4	100	100	Gastrointestinal defects and immunodeficiency syndrome, 243150
TTC8	99,6	98,1	100	100	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
TTI2	100	100	100	100	Mental retardation, autosomal recessive 39, 615541
TTLL5	100	99,7	100	100	Cone-rod dystrophy 19, 615860
TTN	98,6	98,1	100	100	Myopathy, myofibrillar, 9, with early respiratory failure, 603689 Cardiomyopathy, familial hypertrophic, 9, 613765 Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807 Cardiomyopathy, dilated, 1G, 604145 Tibial muscular dystrophy, tardive, 600334 Salih myopathy, 611705
TTPA	94,7	87,1	100	100	Ataxia with isolated vitamin E deficiency, 277460

TTR	94,6	94,6	94,6	94,6	Amyloidosis, hereditary, transthyretin-related, 105210 [Dystransthyretinemic hyperthyroxinemia], 145680 Carpal tunnel syndrome, familial, 115430
TUB	99,4	97,1	100	100	?Retinal dystrophy and obesity, 616188
TUBA1A	99,9	97	100	100	Lissencephaly 3, 611603
TUBA3D	100	99,2	100	100	Keratoconus 9, 617928
TUBA4A	100	100	100	100	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia, 616208
TUBA8	99,9	99,5	100	100	Cortical dysplasia, complex, with other brain malformations 8, 613180
TUBB	97,3	93,9	99,8	99,8	Symmetric circumferential skin creases, congenital, 1, 156610 Cortical dysplasia, complex, with other brain malformations 6, 615771
TUBB1	100	100	100	100	Macrothrombocytopenia, autosomal dominant, TUBB1-related, 613112
TUBB2A	97	95,7	100	100	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB2B	100	99,5	100	100	Cortical dysplasia, complex, with other brain malformations 7, 610031
TUBB3	98,3	96,9	100	100	Fibrosis of extraocular muscles, congenital, 3A, 600638 Cortical dysplasia, complex, with other brain malformations 1, 614039
TUBB4A	95,9	94	97,1	96	Leukodystrophy, hypomyelinating, 6, 612438 Dystonia 4, torsion, autosomal dominant, 128101
TUBB4B	99,9	96,9	100	100	Leber congenital amaurosis with early-onset deafness, 617879
TUBB6	90,6	90,1	100	100	?Facial palsy, congenitla, with ptosis and velopharyngeal dysfunction, 617732
TUBB8	83	55,3	100	100	Oocyte maturation defect 2, 616780
TUBG1	100	100	100	100	Cortical dysplasia, complex, with other brain malformations 4, 615412
TUBGCP2	99,7	96,2	97	97	Pachygyria, microcephaly, developmental delay, and dysmorphic facies, with or without seizures, 618737
TUBGCP4	99,2	96,4	100	100	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TUBGCP6	100	99,3	100	100	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
TUFM	100	99	100	100	Combined oxidative phosphorylation deficiency 4, 610678
TULP1	100	99,5	100	100	Retinitis pigmentosa 14, 600132 Leber congenital amaurosis 15, 613843
TUSC3	100	99,5	100	100	Mental retardation, autosomal recessive 7, 611093

TWIST1	100	98,9	97,2	92,3	Robinow-Sorauf syndrome, 180750 Craniosynostosis 1, 123100 Sweeney-Cox syndrome, 617746 Saethre-Chotzen syndrome with or without eyelid anomalies, 101400
TWIST2	100	100	100	100	Focal facial dermal dysplasia 3, Setleis type, 227260 Ablepharon-macrostomia syndrome, 200110 Barber-Say syndrome, 209885
TWNK	100	100	100	100	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 Perrault syndrome 5, 616138
TXN2	100	100	100	100	?Combined oxidative phosphorylation deficiency 29, 616811
TXNL4A	100	99,4	100	100	Burn-McKeown syndrome, 608572
TXNRD2	96,8	95,9	100	100	?Glucocorticoid deficiency 5, 617825
TYK2	99,9	99	100	100	Immunodeficiency 35, 611521
TYMP	100	97	100	100	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYMS	99,9	99,6	100	100	No OMIM disease ID
TYR	100	100	100	100	Waardenburg syndrome/albinism, digenic, 103470 Albinism, oculocutaneous, type IB, 606952 [Skin/hair/eye pigmentation 3, blue/green eyes], 601800 {Melanoma, cutaneous malignant, susceptibility to, 8}, 601800 Albinism, oculocutaneous, type IA, 203100 [Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800
TYROBP	100	100	100	100	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770
TYRP1	100	99,8	100	100	[Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271 Albinism, oculocutaneous, type III, 203290
U2AF2	99,9	98,3	100	100	No OMIM disease ID
UBA1	99,4	98,2	99,8	99	VEXAS syndrome, somatic, 301054 Spinal muscular atrophy, X-linked 2, infantile, 301830
UBA5	97,8	86,8	100	100	?Spinocerebellar ataxia, autosomal recessive 24, 617133 Developmental and epileptic encephalopathy 44, 617132
UBAP1	98,8	93,4	100	100	Spastic paraplegia 80, autosomal dominant, 618418
UBB	100	99,4	100	100	No OMIM disease ID

UBE2A	99,7	96	100	99,7	Mental retardation, X-linked syndromic, Nascimento-type, 300860
UBE2T	100	99,9	100	100	Fanconi anemia, complementation group T, 616435
UBE3A	99,1	94,8	100	100	Angelman syndrome, 105830
UBE3B	100	99,9	100	100	Kaufman oculocerebrofacial syndrome, 244450
UBIAD1	99,5	96	100	100	Corneal dystrophy, Schnyder type, 121800
UBQLN2	100	99,4	100	100	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857
UBR1	99,9	99,1	98	98	Johanson-Blizzard syndrome, 243800
UBTF	100	99,4	100	100	Neurodegeneration, childhood-onset, with brain atrophy, 617672
UCHL1	99,8	92,5	100	100	Spastic paraplegia 79, autosomal recessive, 615491 {?Parkinson disease 5, susceptibility to}, 613643
UFC1	100	100	100	100	Neurodevelopmental disorder with spasticity and poor growth, 618076
UFM1	74	69,4	100	100	Leukodystrophy, hypomyelinating, 14, 617899
UFSP2	100	99,6	100	100	?Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974 ?Hip dysplasia, Beukes type, 142669
UGDH	99,9	99,1	100	100	Developmental and epileptic encephalopathy 84, 618792
UGP2	99	98,6	96,3	96,3	Developmental and epileptic encephalopathy 83, 618744
UGT1A1	100	100	100	100	[Gilbert syndrome], 143500 Hyperbilirubinemia, familial transient neonatal, 237900 Crigler-Najjar syndrome, type I, 218800 Crigler-Najjar syndrome, type II, 606785 [Bilirubin, serum level of, QTL1], 601816
UMOD	97,7	96,2	100	100	Glomerulocystic kidney disease with hyperuricemia and isosthenuria, 609886 Hyperuricemic nephropathy, familial juvenile 1, 162000 Medullary cystic kidney disease 2, 603860
UMPS	100	99,4	97	97	Orotic aciduria, 258900
UNC119	100	99,7	100	100	?Immunodeficiency 13, 615518 ?Cone-rod dystrophy, 0
UNC13A	99,3	97,7	100	100	No OMIM disease ID
UNC13D	99,7	98,1	100	100	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC45B	99,3	98	100	100	?Cataract 43, 616279

UNC80	97,9	97,4	100	100	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801
UNC93B1	60,6	58,8	100	100	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 1}, 610551
UNG	100	98,8	99,9	99,3	Immunodeficiency with hyper IgM, type 5, 608106
UPB1	100	100	100	100	Beta-ureidopropionase deficiency, 613161
UPF3B	92,2	84,1	100	100	Mental retardation, X-linked, syndromic 14, 300676
UPK3A	100	99,5	100	100	No OMIM disease ID
UQCC1	100	99,9	100	100	No OMIM disease ID
UQCC2	100	99,7	100	100	Mitochondrial complex III deficiency, nuclear type 7, 615824
UQCC3	100	98,7	100	100	?Mitochondrial complex III deficiency, nuclear type 9, 616111
UQCR10	100	100	100	100	No OMIM disease ID
UQCR11	100	100	100	100	No OMIM disease ID
UQCRB	99,4	95,1	100	100	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC1	99,8	98,4	100	100	No OMIM disease ID
UQCRC2	99,9	99,3	100	100	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRFS1	91,9	84,9	100	100	Mitochondrial complex III deficiency, nuclear type 10, 618775
UQCRH	100	98,2	100	100	No OMIM disease ID
UQCRQ	100	100	100	100	Mitochondrial complex III deficiency, nuclear type 4, 615159
UROC1	100	100	100	100	?Urocanase deficiency, 276880
UROD	98,9	96,1	100	100	Porphyria, hepatoerythropoietic, 176100 Porphyria cutanea tarda, 176100
UROS	100	99,9	100	100	Porphyria, congenital erythropoietic, 263700
USB1	100	99,4	100	100	Poikiloderma with neutropenia, 604173
USH1C	100	99,8	100	100	Deafness, autosomal recessive 18A, 602092 Usher syndrome, type 1C, 276904
USH1G	99,6	97,9	100	100	Usher syndrome, type 1G, 606943
USH2A	100	99,8	99,5	99,5	Retinitis pigmentosa 39, 613809 Usher syndrome, type 2A, 276901

USP18	95,9	95,9	100	100	Pseudo-TORCH syndrome 2, 617397
USP27X	100	100	100	100	Mental retardation, X-linked 105, 300984
USP45	99,6	98,1	100	100	?Leber congenital amaurosis 19, 618513
USP48	99,9	99,3	100	100	No OMIM disease ID
USP7	91,3	87,9	94,8	94,8	Hao-Fountain syndrome, 616863
USP8	96,9	87,8	100	100	Pituitary adenoma 4, ACTH-secreting, somatic, 219090
USP9X	98,2	92,9	100	100	Mental retardation, X-linked 99, 300919 Mental retardation, X-linked 99, syndromic, female-restricted, 300968
USP9Y	48,6	43,2	60	60	Spermatogenic failure, Y-linked, 2, 415000
UST	100	98,9	100	100	No OMIM disease ID
UVSSA	100	100	100	100	UV-sensitive syndrome 3, 614640
VAC14	99,9	98,5	100	100	Striatonigral degeneration, childhood-onset, 617054
VAMP1	100	100	100	100	Spastic ataxia 1, autosomal dominant, 108600 Myasthenic syndrome, congenital, 25, 618323
VAMP2	99,5	97,7	100	100	Neurodevelopmental disorder with hypotonia and autistic features with or without hyperkinetic movements, 618760
VANGL1	100	100	100	100	Caudal regression syndrome, 600145 {Neural tube defects, susceptibility to}, 182940
VANGL2	99,9	99	100	100	Neural tube defects, 182940
VAPB	100	99,9	100	100	Spinal muscular atrophy, late-onset, Finkel type, 182980 Amyotrophic lateral sclerosis 8, 608627
VARS1	100	99,9	100	100	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802
VARS2	100	99,4	100	100	Combined oxidative phosphorylation deficiency 20, 615917
VAV1	98,5	97,1	97,1	97,1	No OMIM disease ID
VAX1	97,5	91,5	95,7	91,7	?Microphthalmia, syndromic 11, 614402
VCAN	100	100	100	100	Wagner syndrome 1, 143200
VCL	99,9	99	100	100	Cardiomyopathy, dilated, 1W, 611407 Cardiomyopathy, hypertrophic, 15, 613255

VCP	100	99,2	100	100	Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320 Charcot-Marie-Tooth disease, type 2Y, 616687 Frontotemporal dementia and/or amyotrophic lateral sclerosis 6, 613954
VDR	97,2	94,9	98,2	95,2	Rickets, vitamin D-resistant, type IIA, 277440
VEGFC	100	100	100	100	Lymphatic malformation 4, 615907
VHL	96,3	91,4	100	100	Pheochromocytoma, 171300 Erythrocytosis, familial, 2, 263400 von Hippel-Lindau syndrome, 193300 Renal cell carcinoma, somatic, 144700 Hemangioblastoma, cerebellar, somatic, 0
VIM	99,3	97	100	100	Cataract 30, pulverulent, 116300
VIPAS39	100	100	100	100	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VKORC1	100	100	93	93	Warfarin resistance, 122700 Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473
VLDLR	100	99,8	100	100	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VMA21	99	94,6	100	98,6	Myopathy, X-linked, with excessive autophagy, 310440
VPS11	94,9	93,6	100	100	Leukodystrophy, hypomyelinating, 12, 616683
VPS13A	99,4	95,6	100	100	Choreoacanthocytosis, 200150
VPS13B	99,5	98,2	99,5	99,4	Cohen syndrome, 216550
VPS13C	99,4	96,9	100	100	Parkinson disease 23, autosomal recessive, early onset, 616840
VPS13D	100	99,7	100	100	Spinocerebellar ataxia, autosomal recessive 4, 607317
VPS16	100	100	100	100	No OMIM disease ID
VPS33A	91,3	89,8	89,9	89,9	Mucopolysaccharidosis-plus syndrome, 617303
VPS33B	100	100	100	100	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
VPS35	97,3	91,3	100	100	{Parkinson disease 17}, 614203
VPS35L	100	99,9	100	100	Ritscher-Schinzel syndrome 3, 619135
VPS37A	91,3	78,2	100	100	Spastic paraplegia 53, autosomal recessive, 614898
VPS45	99,2	95,7	95,3	95,3	Neutropenia, severe congenital, 5, autosomal recessive, 615285

VPS51	95	83,2	100	100	Pontocerebellar hypoplasia, type 13, 618606
VPS53	91,5	90,7	100	99,3	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	99,7	98,5	100	100	Pontocerebellar hypoplasia type 1A, 607596
VSX1	84,7	80,5	100	100	Keratoconus 1, 148300 ?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195
VSX2	100	99,3	100	100	Microphthalmia with coloboma 3, 610092 Microphthalmia, isolated 2, 610093
VWA3B	100	99,7	100	100	?Spinocerebellar ataxia, autosomal recessive 22, 616948
VWF	99,8	98,6	100	100	von Willebrand disease, type 1, 193400 von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 von Willebrand disease, type 3, 277480
WAC	100	99,7	100	100	Desanto-Shinawi syndrome, 616708
WARS1	99,8	98,3	100	100	Neuronopathy, distal hereditary motor, type IX, 617721
WARS2	100	99,4	100	100	Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710
WAS	95,9	85,3	100	99,8	Thrombocytopenia, X-linked, intermittent, 313900 Thrombocytopenia, X-linked, 313900 Wiskott-Aldrich syndrome, 301000 Neutropenia, severe congenital, X-linked, 300299
WASF1	99,9	96,5	100	100	Neurodevelopmental disorder with absent language and variable seizures, 618707
WASHC4	99,1	95,5	100	100	?Mental retardation, autosomal recessive 43, 615817
WASHC5	100	99,8	100	100	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, autosomal dominant, 603563
WBP2	100	99,7	100	100	Deafness, autosomal recessive 107, 617639
WDFY3	100	99,6	100	100	?Microcephaly 18, primary, autosomal dominant, 617520
WDPCP	98,2	94,4	98,1	98,1	?Bardet-Biedl syndrome 15, 615992 Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR1	100	99,6	100	100	Periodic fever, immunodeficiency, and thrombocytopenia syndrome, 150550
WDR11	98	96,5	100	100	Hypogonadotropic hypogonadism 14 with or without anosmia, 614858
WDR13	99,9	98,6	100	100	No OMIM disease ID

WDR19	100	99,4	100	100	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WDR26	88,7	83,9	94,2	91,7	Skraban-Deardorff syndrome, 617616
WDR35	99,8	98,9	100	100	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 Cranioectodermal dysplasia 2, 613610
WDR36	99,7	97,6	100	100	Glaucoma 1, open angle, G, 609887
WDR37	86,5	86,2	86,5	86,5	Neurooculocardiogenitourinary syndrome, 618652
WDR4	100	100	100	100	Microcephaly, growth deficiency, seizures, and brain malformations, 618346 Galloway-Mowat syndrome 6, 618347
WDR45	98,1	92,4	100	100	Neurodegeneration with brain iron accumulation 5, 300894
WDR45B	98	89,2	100	100	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977
WDR62	100	99,5	100	100	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
WDR72	96,8	96,4	96,9	96,9	Amelogenesis imperfecta, type IIA3, 613211
WDR73	100	100	100	100	Galloway-Mowat syndrome 1, 251300
WDR81	100	100	100	100	Hydrocephalus, congenital, 3, with brain anomalies, 617967 Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185
WEE2	100	99,6	100	100	Oocyte maturation defect 5, 617996
WFS1	100	99,9	100	100	?Cataract 41, 116400 Deafness, autosomal dominant 6/14/38, 600965 {Diabetes mellitus, noninsulin-dependent, association with}, 125853 Wolfram-like syndrome, autosomal dominant, 614296 Wolfram syndrome 1, 222300
WHRN	99,8	98	100	100	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
WIPF1	100	99,9	100	100	Wiskott-Aldrich syndrome 2, 614493
WIPI2	100	99,3	100	100	?Intellectual developmental disorder with short stature and variable skeletal anomalies, 618453
WNK1	99,9	99,6	100	100	Pseudohypoaldosteronism, type IIC, 614492 Neuropathy, hereditary sensory and autonomic, type II, 201300
WNK4	99,9	99,3	100	100	Pseudohypoaldosteronism, type IIB, 614491

WNT1	99,3	95,3	100	100	{Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221 Osteogenesis imperfecta, type XV, 615220
WNT10A	100	99,4	100	100	Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400 Odontoonychodermal dysplasia, 257980
WNT10B	100	99,4	100	100	Split-hand/foot malformation 6, 225300 Tooth agenesis, selective, 8, 617073
WNT2B	98	91,3	100	100	Diarrhea 9, 618168
WNT3	100	99,6	100	100	?Tetra-amelia syndrome 1, 273395
WNT4	99,1	94,8	98,9	96,2	?SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330
WNT5A	100	100	100	100	Robinow syndrome, autosomal dominant 1, 180700
WNT6	100	98,7	100	100	No OMIM disease ID
WNT7A	100	100	100	100	Fuhrmann syndrome, 228930 Ulna and fibula, absence of, with severe limb deficiency, 276820
WRAP53	100	100	100	100	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	99,9	98,8	100	100	Werner syndrome, 277700
WT1	97,3	95,4	97,7	97,7	Mesothelioma, somatic, 156240 Wilms tumor, type 1, 194070 Frasier syndrome, 136680 Denys-Drash syndrome, 194080 Meacham syndrome, 608978 Nephrotic syndrome, type 4, 256370
WWOX	100	100	100	100	Spinocerebellar ataxia, autosomal recessive 12, 614322 Esophageal squamous cell carcinoma, somatic, 133239 Developmental and epileptic encephalopathy 28, 616211
XDH	100	99,9	100	100	Xanthinuria, type I, 278300
XIAP	93	88,8	100	100	Lymphoproliferative syndrome, X-linked, 2, 300635
XIRP2	100	99,9	100	99,9	No OMIM disease ID
XIST					X-inactivation, familial skewed, 300087
XK	99,8	98,1	100	100	McLeod syndrome with or without chronic granulomatous disease, 300842
XPA	99,6	95,6	100	100	Xeroderma pigmentosum, group A, 278700

XPC	100	100	100	100	Xeroderma pigmentosum, group C, 278720
XPNPEP3	100	100	100	100	Nephronophthisis-like nephropathy 1, 613159
XPO5	100	99,9	100	99,7	No OMIM disease ID
XPR1	100	99,9	100	100	Basal ganglia calcification, idiopathic, 6, 616413
XRCC1	100	98,8	100	100	?Spinocerebellar ataxia, autosomal recessive 26, 617633
XRCC2	99,8	97,4	100	100	Spermatogenic failure, 619145 ?Premature ovarian failure 17, 619146 ?Fanconi anemia, complementation group U, 617247
XRCC4	99,9	99,3	100	100	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	97,4	89,6	98,1	94,8	{Pseudoxanthoma elasticum, modifier of severity of}, 264800 Desbuquois dysplasia 2, 615777
XYLT2	100	98,3	96,7	96,7	{Pseudoxanthoma elasticum, modifier of severity of}, 264800 Spondyloocular syndrome, 605822
YAP1	96,4	89,4	100	100	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433
YARS1	100	99,9	100	100	Charcot-Marie-Tooth disease, dominant intermediate C, 608323
YARS2	100	99,8	100	100	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
YME1L1	99	95,2	100	100	?Optic atrophy 11, 617302
YWHAE	100	100	100	100	No OMIM disease ID
YWHAG	100	100	100	100	Developmental and epileptic encephalopathy 56, 617665
YWHAZ	81,2	71,7	100	100	No OMIM disease ID
YY1	100	99,8	100	100	Gabriele-de Vries syndrome, 617557
YY1AP1	99,3	98,2	100	100	Grange syndrome, 602531
ZAP70	100	99,3	100	100	Autoimmune disease, multisystem, infantile-onset, 2, 617006 Immunodeficiency 48, 269840
ZBTB11	99,9	99,6	100	100	Intellectual developmental disorder, autosomal recessive 69, 618383
ZBTB16	100	99,9	100	100	Skeletal defects, genital hypoplasia, and mental retardation, 612447 Leukemia, acute promyelocytic, PL2F/RARA type, 0
ZBTB17	100	100	100	100	No OMIM disease ID

ZBTB18	100	99,9	100	99,8	Mental retardation, autosomal dominant 22, 612337
ZBTB20	100	100	100	100	Primrose syndrome, 259050
ZBTB24	100	100	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069
ZBTB42	100	100	100	100	?Lethal congenital contracture syndrome 6, 616248
ZC3H14	99,9	98,9	100	100	Mental retardation, autosomal recessive 56, 617125
ZC4H2	100	99	100	100	Wieacker-Wolff syndrome, 314580 Wieacker-Wolff syndrome, female-restricted, 301041
ZCCHC8	99,8	98,7	100	100	?Pulmonary fibrosis and/or bone marrow failure, telomere-related, 5, 618674
ZDHHC9	99,9	93,8	100	100	Mental retardation, X-linked syndromic, Raymond type, 300799
ZEB1	100	99,4	100	100	Corneal dystrophy, posterior polymorphous, 3, 609141 Corneal dystrophy, Fuchs endothelial, 6, 613270
ZEB2	99,9	99,1	97,4	97,4	Mowat-Wilson syndrome, 235730
ZFHX2	100	99,6	100	100	?Marsili syndrome, 147430
ZFHX3	100	99,6	100	100	Prostate cancer, somatic, 176807
ZFP57	100	99,8	100	100	Diabetes mellitus, transient neonatal 1, 601410
ZFPM2	100	100	100	100	46XY sex reversal 9, 616067 Tetralogy of Fallot, 187500 Diaphragmatic hernia 3, 610187
ZFYVE26	100	99,1	100	100	Spastic paraplegia 15, autosomal recessive, 270700
ZFYVE27	100	100	100	100	Spastic paraplegia 33, autosomal dominant, 610244
ZIC1	100	100	100	100	Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736 ?Craniosynostosis 6, 616602
ZIC2	100	98,7	98,5	95,7	Holoprosencephaly 5, 609637
ZIC3	100	99,9	100	100	Congenital heart defects, nonsyndromic, 1, X-linked, 306955 Heterotaxy, visceral, 1, X-linked, 306955 VACTERL association, X-linked, 314390
ZMIZ1	99,4	98,4	100	100	Neurodevelopmental disorder with dysmorphic facies and distal skeletal anomalies, 618659
ZMPSTE24	100	99,9	100	100	Restrictive dermopathy, lethal, 275210 Mandibuloacral dysplasia with type B lipodystrophy, 608612
ZMYND10	100	100	100	100	Ciliary dyskinesia, primary, 22, 615444

ZMYND11	100	99,6	100	100	Mental retardation, autosomal dominant 30, 616083
ZMYND15	100	99,4	100	100	?Spermatogenic failure 14, 615842
ZNF141	100	100	100	100	?Polydactyly, postaxial, type A6, 615226
ZNF142	100	99,9	100	100	Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425
ZNF148	99,9	99,6	100	100	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260
ZNF292	99,6	98,3	99,6	99,6	No OMIM disease ID
ZNF335	100	99,9	100	100	Microcephaly 10, primary, autosomal recessive, 615095
ZNF341	97,2	95	100	100	Hyper-IgE recurrent infection syndrome 3, autosomal recessive, 618282
ZNF407	99,9	99,3	100	100	No OMIM disease ID
ZNF408	100	100	100	100	?Exudative vitreoretinopathy 6, 616468 Retinitis pigmentosa 72, 616469
ZNF41	100	99,6	100	100	No OMIM disease ID
ZNF423	100	100	100	100	Nephronophthisis 14, 614844 Joubert syndrome 19, 614844
ZNF462	100	99,9	100	100	Weiss-Kruszka syndrome, 618619
ZNF469	100	100	100	100	Brittle cornea syndrome 1, 229200
ZNF513	100	100	100	100	?Retinitis pigmentosa 58, 613617
ZNF592	100	99,6	100	100	No OMIM disease ID
ZNF644	100	100	100	100	Myopia 21, autosomal dominant, 614167
ZNF687	100	100	100	100	Paget disease of bone 6, 616833
ZNF711	99,8	98,2	100	100	Mental retardation, X-linked 97, 300803
ZNF750	100	100	100	100	Seborrhea-like dermatitis with psoriasiform elements, 610227
ZNHIT3	74,4	74,4	74,6	74,4	PEHO syndrome, 260565
ZP1	100	100	100	100	Oocyte maturation defect 1, 615774
ZP2	99,8	98,4	100	100	Oocyte maturation defect 6, 618353
ZP3	100	100	100	100	Oocyte maturation defect 3, 617712

ZSWIM6	95,5	91,9	94,9	92,1	Acromelic frontonasal dysostosis, 603671 Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865

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.
Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.
TWIST is the chemistry used for (in-house) TURBO/RAPID 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-DNA coding genes.
non-DNA coding genes are covered, but as coverage statistics are based on DNA coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : November 20th , 2020.
This list is accurate for panel version DG 3.0.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors