MENDELIOME GENE PANEL DG 2.16 (3741 genes)

Releasedate: 07-06-2019

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM disease ID*
A4GALT	176,9	100.0%	100.0%	NOR polyagglutination syndrome, 111400 [Blood group, P1Pk system, p phenotype], 111400 [Blood group, P1Pk system, P(2) phenotype], 111400
AAAS	102,3	100.0%	99.6%	Achalasia-addisonianism-alacrimia syndrome, 231550
AAGAB	134,7	99.9%	99.8%	Keratoderma, palmoplantar, punctate type IA, 148600
AARS	103,7	100.0%	99.5%	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 Epileptic encephalopathy, early infantile, 29, 616339
AARS2	122,7	100.0%	99.8%	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889
AASS	131,5	99.9%	99.4%	Hyperlysinemia, 238700 Saccharopinuria, 268700
ABAT	83,2	99.9%	98.3%	GABA-transaminase deficiency, 613163
ABCA1	97	99.9%	98.4%	HDL deficiency, type 2, 604091 Tangier disease, 205400 {Coronary artery disease in familial hypercholesterolemia, protection against}, 143890
ABCA12	129,3	99.6%	98.4%	Ichthyosis, congenital, autosomal recessive 4A, 601277 Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500
ABCA3	119,4	100.0%	99.5%	Surfactant metabolism dysfunction, pulmonary, 3, 610921
ABCA4	104,5	99.9%	98.9%	Cone-rod dystrophy 3, 604116 Fundus flavimaculatus, 248200 Retinal dystrophy, early-onset severe, 248200 Retinitis pigmentosa 19, 601718 Stargardt disease 1, 248200 {Macular degeneration, age-related, 2}, 153800
ABCA5	73,9	98.2%	91.7%	?Hypertrichosis, congenital generalized, with gingival hyperplasia, 135400
ABCB11	134,9	100.0%	99.2%	Cholestasis, benign recurrent intrahepatic, 2, 605479 Cholestasis, progressive familial intrahepatic 2, 601847
ABCB4	123,3	99.9%	99.2%	Cholestasis, intrahepatic, of pregnancy, 3, 614972 Cholestasis, progressive familial intrahepatic 3, 602347 Gallbladder disease 1, 600803

ABCB6	127,9	100.0%	99.9%	Dyschromatosis universalis hereditaria 3, 615402 Microphthalmia, isolated, with coloboma 7, 614497 Pseudohyperkalemia, familial, 2, due to red cell leak, 609153 [Blood group, Langereis system], 111600
ABCB7	126,2	99.9%	98.6%	Anemia, sideroblastic, with ataxia, 301310
ABCC2	110,7	100.0%	99.8%	Dubin-Johnson syndrome, 237500
ABCC6	109,1	93.6%	92.8%	Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850
ABCC8	125,8	100.0%	99.9%	Diabetes mellitus, noninsulin-dependent, 125853 Diabetes mellitus, permanent neonatal, 606176 Diabetes mellitus, transient neonatal 2, 610374 Hyperinsulinemic hypoglycemia, familial, 1, 256450 Hypoglycemia of infancy, leucine-sensitive, 240800
ABCC9	142,6	100.0%	99.7%	Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia, 239850
ABCD1	87,4	77.2%	75.0%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABCD3	108,1	99.5%	97.3%	?Bile acid synthesis defect, congenital, 5, 616278
ABCD4	129	99.8%	98.4%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	139,5	100.0%	99.9%	Sitosterolemia, 210250
ABCG8	133,9	99.7%	98.2%	Sitosterolemia, 210250 {Gallbladder disease 4}, 611465
ABHD12	93,1	100.0%	98.9%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ABHD5	180,9	100.0%	100.0%	Chanarin-Dorfman syndrome, 275630
ABL1	155,2	100.0%	99.9%	Congenital heart defects and skeletal malformations syndrome, 617602 Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 0
ACAD8	122,1	100.0%	99.9%	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	124,3	99.9%	99.1%	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADM	129,5	99.8%	99.2%	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	148,2	100.0%	100.0%	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	113,1	99.9%	97.3%	2-methylbutyrylglycinuria, 610006
ACADVL	115,8	99.8%	98.0%	VLCAD deficiency, 201475
ACAN	121,3	94.6%	89.1%	?Spondyloepiphyseal dysplasia, Kimberley type, 608361 Short stature and advanced bone age, with or without early-onset osteoarthritis and/or osteochondritis dissecans, 165800

				Spondyloepimetaphyseal dysplasia, aggrecan type, 612813
ACAT1	110,2	99.7%	98.3%	Alpha-methylacetoacetic aciduria, 203750
ACD	159,6	100.0%	100.0%	?Dyskeratosis congenita, autosomal dominant 6, 616553 ?Dyskeratosis congenita, autosomal recessive 7, 616553
ACE	120,1	99.9%	99.5%	Renal tubular dysgenesis, 267430 [Angiotensin I-converting enzyme, benign serum increase], 0 {Microvascular complications of diabetes 3}, 612624 {Myocardial infarction, susceptibility to}, 0 {SARS, progression of}, 0 {Stroke, hemorrhagic}, 614519
ACER3	115,7	99.6%	98.9%	?Leukodystrophy, progressive, early childhood-onset, 617762
ACO2	115,3	95.8%	89.5%	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559
ACOX1	123,7	100.0%	100.0%	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACOX2	111	100.0%	99.3%	Bile acid synthesis defect, congenital, 6, 617308
ACP4	91,2	96.8%	88.5%	Amelogenesis imperfecta, type IJ, 617297
ACP5	172,6	100.0%	99.6%	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACSF3	145,8	99.9%	99.1%	Combined malonic and methylmalonic aciduria, 614265
ACSL4	102,8	98.6%	93.5%	Mental retardation, X-linked 63, 300387
ACSL6	106	99.4%	97.7%	Myelodysplastic syndrome, 0 Myelogenous leukemia, acute, 0
ACTA1	95,3	99.8%	97.9%	?Myopathy, scapulohumeroperoneal, 616852 Myopathy, actin, congenital, with cores, 161800 Myopathy, actin, congenital, with excess of thin myofilaments, 161800 Myopathy, congenital, with fiber-type disproportion 1, 255310 Nemaline myopathy 3, autosomal dominant or recessive, 161800
ACTA2	87,3	99.9%	98.6%	Aortic aneurysm, familial thoracic 6, 611788 Moyamoya disease 5, 614042 Multisystemic smooth muscle dysfunction syndrome, 613834
ACTB	80,5	100.0%	99.7%	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACTC1	111,1	100.0%	98.9%	Atrial septal defect 5, 612794 Cardiomyopathy, dilated, 1R, 613424 Cardiomyopathy, hypertrophic, 11, 612098 Left ventricular noncompaction 4, 613424
ACTG1	116,3	100.0%	100.0%	Baraitser-Winter syndrome 2, 614583 Deafness, autosomal dominant 20/26, 604717

ACTG2	97,3	99.3%	96.0%	Visceral myopathy, 155310
ACTN1	131,5	100.0%	100.0%	Bleeding disorder, platelet-type, 15, 615193
ACTN2	128,9	100.0%	100.0%	Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158 Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158
ACTN4	130,5	100.0%	99.9%	Glomerulosclerosis, focal segmental, 1, 603278
ACVR1	136,9	100.0%	99.9%	Fibrodysplasia ossificans progressiva, 135100
ACVR1B	137,8	100.0%	99.4%	Pancreatic cancer, somatic, 0
ACVR2B	115,6	99.7%	97.0%	Heterotaxy, visceral, 4, autosomal, 613751
ACVRL1	113,6	100.0%	98.4%	Telangiectasia, hereditary hemorrhagic, type 2, 600376
ACY1	118,5	100.0%	98.6%	Aminoacylase 1 deficiency, 609924
ADA	104,6	100.0%	99.6%	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADA2	83,5	99.9%	97.6%	?Sneddon syndrome, 182410 Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688
ADAM10	121,4	94.8%	93.4%	Reticulate acropigmentation of Kitamura, 615537 {Alzheimer disease 18, susceptibility to}, 615590
ADAM17	119	99.8%	98.6%	?Inflammatory skin and bowel disease, neonatal, 1, 614328
ADAM22	133,9	99.9%	99.4%	?Epileptic encephalopathy, early infantile, 61, 617933
ADAM9	141,3	99.9%	99.0%	Cone-rod dystrophy 9, 612775
ADAMTS10	122,8	100.0%	99.8%	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS13	103,8	98.1%	95.2%	Thrombotic thrombocytopenic purpura, familial, 274150
ADAMTS17	109,2	97.6%	92.3%	Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTS18	129,7	100.0%	99.9%	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
ADAMTS2	126,3	100.0%	99.6%	Ehlers-Danlos syndrome, dermatosparaxis type, 225410
ADAMTS3	136,6	100.0%	99.8%	?Hennekam lymphangiectasia-lymphedema syndrome 3, 618154
ADAMTSL2	115,9	99.0%	96.3%	Geleophysic dysplasia 1, 231050
ADAMTSL4	122,7	100.0%	99.6%	Ectopia lentis et pupillae, 225200 Ectopia lentis, isolated, autosomal recessive, 225100
ADAR	109,2	99.9%	99.3%	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
ADAT3	128	100.0%	99.9%	Mental retardation, autosomal recessive 36, 615286
ADCY1	134,5	96.8%	95.4%	?Deafness, autosomal recessive 44, 610154
ADCY5	131,8	97.8%	94.7%	Dyskinesia, familial, with facial myokymia, 606703
ADCY6	162,8	100.0%	100.0%	?Lethal congenital contracture syndrome 8, 616287
ADD3	143,5	99.9%	99.6%	Cerebral palsy, spastic quadriplegic, 3, 617008
ADGRE2	137,9	96.6%	96.1%	Vibratory urticaria, 125630

ADGRG1	147,2	100.0%	100.0%	Polymicrogyria, bilateral frontoparietal, 606854 Polymicrogyria, bilateral perisylvian, 615752
ADGRG2	82,7	97.7%	90.9%	Congenital bilateral absence of vas deferens, X-linked, 300985
ADGRG6	135,7	99.8%	98.7%	Lethal congenital contracture syndrome 9, 616503
ADGRV1	126	99.7%	98.4%	?Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472
ADIPOQ	121,1	100.0%	100.0%	Adiponectin deficiency, 612556
ADK	102,4	99.8%	98.0%	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADNP	190,5	100.0%	100.0%	Helsmoortel-van der Aa syndrome, 615873
ADPRHL2	163,8	100.0%	100.0%	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
ADRA2B	214,3	100.0%	100.0%	Epilepsy, myoclonic, familial adult, 2, 607876
ADRB2	103,1	100.0%	100.0%	Beta-2-adrenoreceptor agonist, reduced response to, 0 {Asthma, nocturnal, susceptibility to}, 600807 {Obesity, susceptibility to}, 601665
ADSL	138,6	99.2%	98.6%	Adenylosuccinase deficiency, 103050
ADSSL1	110,7	95.5%	87.9%	Myopathy, distal, 5, 617030
AEBP1	147,7	100.0%	100.0%	Ehlers-Danlos syndrome, classic-like, 2, 618000
AFF2	107,8	99.8%	98.8%	Mental retardation, X-linked, FRAXE type, 309548
AFF4	99,8	99.9%	99.0%	CHOPS syndrome, 616368
AFG3L2	98,3	95.9%	86.1%	Spastic ataxia 5, autosomal recessive, 614487 Spinocerebellar ataxia 28, 610246
AFP	106,7	97.2%	90.6%	Alpha-fetoprotein deficiency, 615969 [Hereditary persistence of alpha-fetoprotein], 615970
AGA	142,7	100.0%	100.0%	Aspartylglucosaminuria, 208400
AGBL1	106,4	98.5%	98.4%	Corneal dystrophy, Fuchs endothelial, 8, 615523
AGBL5	104,9	100.0%	99.4%	Retinitis pigmentosa 75, 617023
AGK	108,5	99.5%	95.7%	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AGL	146,9	100.0%	99.4%	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGPAT2	162,6	99.1%	94.8%	Lipodystrophy, congenital generalized, type 1, 608594
AGPS	75,4	99.5%	97.8%	Rhizomelic chondrodysplasia punctata, type 3, 600121
AGRN	151,6	98.4%	94.5%	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120
AGT	192,1	100.0%	100.0%	Renal tubular dysgenesis, 267430 {Hypertension, essential, susceptibility to}, 145500

				{Preeclampsia, susceptibility to}, 0
AGTPBP1	116,9	98.7%	95.1%	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276
AGTR1	143,6	91.9%	91.6%	Renal tubular dysgenesis, 267430 {Hypertension, essential}, 145500
AGXT	160,8	100.0%	100.0%	Hyperoxaluria, primary, type 1, 259900
AHCY	111,6	99.9%	97.7%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHDC1	148,5	99.7%	98.3%	Xia-Gibbs syndrome, 615829
AHI1	129,8	99.7%	98.3%	Joubert syndrome 3, 608629
AHR	184	100.0%	99.6%	?Retinitis pigmentosa 85, 618345
AHSG	160,8	100.0%	99.8%	?Alopecia-mental retardation syndrome 1, 203650
AICDA	128,4	100.0%	99.5%	Immunodeficiency with hyper-IgM, type 2, 605258
AIFM1	90	99.8%	96.7%	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Deafness, X-linked 5, 300614
AIMP1	79,4	99.1%	92.4%	Leukodystrophy, hypomyelinating, 3, 260600
AIMP2	117,6	96.1%	89.3%	Leukodystrophy, hypomyelinating, 17, 618006
AIP	137,2	100.0%	99.6%	Pituitary adenoma 1, multiple types, 102200 Pituitary adenoma predisposition, 102200
AIPL1	113	100.0%	99.9%	Cone-rod dystrophy, 604393 Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393
AIRE	102,3	100.0%	99.9%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AK1	136,9	100.0%	99.9%	Hemolytic anemia due to adenylate kinase deficiency, 612631
AK2	96,1	98.7%	94.4%	Reticular dysgenesis, 267500
AK7	113,7	99.7%	96.8%	?Spermatogenic failure 27, 617965
AKAP9	98,4	99.1%	96.7%	?Long QT syndrome-11, 611820
AKR1C2	135,8	94.9%	87.9%	46XY sex reversal 8, 614279
AKR1D1	91,5	99.6%	95.7%	Bile acid synthesis defect, congenital, 2, 235555
AKT1	152,6	100.0%	99.4%	Breast cancer, somatic, 114480 Colorectal cancer, somatic, 114500 Cowden syndrome 6, 615109 Ovarian cancer, somatic, 167000 Proteus syndrome, somatic, 176920 {Schizophrenia, susceptibility to}, 181500
AKT2	162,7	100.0%	99.8%	Diabetes mellitus, type II, 125853 Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900

AKT3	82,3	99.2%	94.2%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937
ALAD	94,9	99.5%	94.7%	Porphyria, acute hepatic, 612740 {Lead poisoning, susceptibility to}, 612740
ALAS2	74,7	98.9%	94.7%	Anemia, sideroblastic, 1, 300751 Protoporphyria, erythropoietic, X-linked, 300752
ALB	156,5	100.0%	99.2%	Analbuminemia, 616000 [Dysalbuminemic hyperthyroxinemia], 615999
ALDH18A1	113,7	100.0%	99.8%	Cutis laxa, autosomal dominant 3, 616603 Cutis laxa, autosomal recessive, type IIIA, 219150 Spastic paraplegia 9A, autosomal dominant, 601162 Spastic paraplegia 9B, autosomal recessive, 616586
ALDH1A3	102,4	99.7%	97.0%	Microphthalmia, isolated 8, 615113
ALDH2	126,5	100.0%	100.0%	Alcohol sensitivity, acute, 610251 {Esophageal cancer, alcohol-related, susceptibility to}, 0 {Hangover, susceptibility to}, 610251 {Sublingual nitroglycerin, susceptibility to poor response to}, 0
ALDH3A2	113,5	95.3%	94.3%	Sjogren-Larsson syndrome, 270200
ALDH4A1	123,9	100.0%	99.8%	Hyperprolinemia, type II, 239510
ALDH5A1	91	99.3%	93.2%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	104,9	100.0%	99.6%	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	66,7	93.5%	86.1%	Epilepsy, pyridoxine-dependent, 266100
ALDOA	120,4	76.5%	74.5%	Glycogen storage disease XII, 611881
ALDOB	135,3	100.0%	99.3%	Fructose intolerance, hereditary, 229600
ALG1	46,5	53.2%	50.2%	Congenital disorder of glycosylation, type Ik, 608540
ALG11	129,3	96.8%	96.3%	Congenital disorder of glycosylation, type Ip, 613661
ALG12	155,7	100.0%	99.9%	Congenital disorder of glycosylation, type Ig, 607143
ALG13	77,3	98.5%	92.1%	?Congenital disorder of glycosylation, type Is, 300884 Epileptic encephalopathy, early infantile, 36, 300884
ALG14	199,4	100.0%	100.0%	?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227
ALG2	103,2	100.0%	100.0%	?Congenital disorder of glycosylation, type Ii, 607906 Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228
ALG3	106,5	100.0%	99.9%	Congenital disorder of glycosylation, type Id, 601110
ALG6	101,6	99.1%	95.6%	Congenital disorder of glycosylation, type Ic, 603147
ALG8	118,5	96.6%	96.2%	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	113	100.0%	99.6%	Congenital disorder of glycosylation, type Il, 608776

				Gillessen-Kaesbach-Nishimura syndrome, 263210
ALMS1	172,8	100.0%	99.7%	Alstrom syndrome, 203800
ALOX12B	125,6	100.0%	99.8%	Ichthyosis, congenital, autosomal recessive 2, 242100
ALOXE3	124,7	100.0%	99.4%	Ichthyosis, congenital, autosomal recessive 3, 606545
ALPK3	113,9	99.4%	97.2%	Cardiomyopathy, familial hypertrophic 27, 618052
ALPL	154,8	100.0%	99.7%	Hypophosphatasia, adult, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500 Odontohypophosphatasia, 146300
ALS2	145,1	100.0%	99.8%	Amyotrophic lateral sclerosis 2, juvenile, 205100 Primary lateral sclerosis, juvenile, 606353 Spastic paralysis, infantile onset ascending, 607225
ALX1	134,2	99.9%	98.5%	?Frontonasal dysplasia 3, 613456
ALX3	134,6	91.1%	79.0%	Frontonasal dysplasia 1, 136760
ALX4	157,1	100.0%	100.0%	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597 {Craniosynostosis 5, susceptibility to}, 615529
AMACR	157,7	100.0%	100.0%	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
AMBN	175,1	99.3%	97.1%	Amelogenesis imperfecta, type IF, 616270
AMELX	85,7	98.9%	93.5%	Amelogenesis imperfecta, type 1E, 301200
AMER1	98,2	99.8%	98.9%	Osteopathia striata with cranial sclerosis, 300373
AMH	82,2	99.9%	98.2%	Persistent Mullerian duct syndrome, type I, 261550
AMHR2	143,2	100.0%	99.4%	Persistent Mullerian duct syndrome, type II, 261550
AMMECR1	97,4	99.8%	98.9%	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
AMN	101,5	98.1%	90.6%	Megaloblastic anemia-1, Norwegian type, 261100
AMPD1	115,8	99.9%	98.6%	Myopathy due to myoadenylate deaminase deficiency, 615511
AMPD2	132,3	100.0%	99.9%	?Spastic paraplegia 63, 615686 Pontocerebellar hypoplasia, type 9, 615809
AMT	142,7	100.0%	100.0%	Glycine encephalopathy, 605899
AMTN	119,3	100.0%	99.1%	?Amelogenesis imperfecta, type IIIB, 617607
ANG	154,1	100.0%	100.0%	Amyotrophic lateral sclerosis 9, 611895
ANGPTL3	94	99.2%	95.9%	Hypobetalipoproteinemia, familial, 2, 605019
ANGPTL4	125,5	100.0%	98.0%	Plasma triglyceride level QTL, low, 615881
ANK1	132,5	100.0%	99.3%	Spherocytosis, type 1, 182900
ANK2	139,5	100.0%	100.0%	Cardiac arrhythmia, ankyrin-B-related, 600919

				Long QT syndrome 4, 600919
ANK3	139,9	99.4%	99.0%	?Mental retardation, autosomal recessive, 37, 615493
ANKH	111,6	100.0%	99.9%	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000
ANKLE2	144,7	100.0%	99.8%	?Microcephaly 16, primary, autosomal recessive, 616681
ANKRD11	119,6	99.2%	97.1%	KBG syndrome, 148050
ANKRD26	83,3	95.3%	90.1%	Thrombocytopenia 2, 188000
ANKS6	94,2	98.3%	94.4%	Nephronophthisis 16, 615382
ANLN	140,7	98.7%	97.7%	Focal segmental glomerulosclerosis 8, 616032
ANO10	106	98.9%	96.3%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANO3	118,3	99.5%	97.7%	Dystonia 24, 615034
ANO5	131	99.6%	97.3%	Gnathodiaphyseal dysplasia, 166260 Miyoshi muscular dystrophy 3, 613319 Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307
ANO6	133,3	99.8%	98.0%	Scott syndrome, 262890
ANOS1	76,7	91.7%	88.0%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
ANTXR1	108,3	99.0%	96.9%	GAPO syndrome, 230740 {?Hemangioma, capillary infantile, susceptibility to}, 602089
ANTXR2	119,3	99.9%	98.6%	Hyaline fibromatosis syndrome, 228600
ANXA11	86,8	99.9%	98.4%	Amytrophic lateral sclerosis 23, 617839
AP1S1	101	100.0%	99.8%	MEDNIK syndrome, 609313
AP1S2	55	75.3%	68.6%	Mental retardation, X-linked syndromic 5, 304340
AP2S1	110,7	90.4%	89.8%	Hypocalciuric hypercalcemia, type III, 600740
AP3B1	112,1	99.5%	96.5%	Hermansky-Pudlak syndrome 2, 608233
AP3B2	125,6	99.4%	97.6%	Epileptic encephalopathy, early infantile, 48, 617276
AP3D1	125,2	98.4%	97.9%	?Hermansky-Pudlak syndrome 10, 617050
AP4B1	121	99.9%	98.4%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	106,6	99.8%	98.8%	Spastic paraplegia 51, autosomal recessive, 613744 Stuttering, familial persistent, 1, 184450
AP4M1	129,3	99.7%	98.1%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	66,2	78.5%	71.3%	Spastic paraplegia 52, autosomal recessive, 614067
AP5Z1	121,3	100.0%	100.0%	Spastic paraplegia 48, autosomal recessive, 613647
APC	141,4	99.9%	99.6%	Adenoma, periampullary, somatic, 0 Adenomatous polyposis coli, 175100 Brain tumor-polyposis syndrome 2, 175100 Colorectal cancer, somatic, 114500

				Desmoid disease, hereditary, 135290 Gardner syndrome, 175100 Gastric cancer, somatic, 613659 Hepatoblastoma, somatic, 114550
APC2	122,4	99.9%	98.7%	?Sotos syndrome 3, 617169
APCDD1	171,4	100.0%	99.3%	Hypotrichosis 1, 605389
APOA1	140,5	100.0%	100.0%	Amyloidosis, 3 or more types, 105200 ApoA-I and apoC-III deficiency, combined, 0 Corneal clouding, autosomal recessive, 0 Hypoalphalipoproteinemia, 604091
APOA2	86,4	87.4%	81.4%	Apolipoprotein A-II deficiency, 0 {Hypercholesterolemia, familial, modifier of}, 143890
APOA5	187,8	100.0%	100.0%	Hyperchylomicronemia, late-onset, 144650 {Hypertriglyceridemia, susceptibility to}, 145750
APOB	154,7	100.0%	99.7%	Hypercholesterolemia, due to ligand-defective apo B, 144010 Hypobetalipoproteinemia, 615558
APOC2	103,7	100.0%	100.0%	Hyperlipoproteinemia, type Ib, 207750
APOC3	97,3	100.0%	100.0%	Apolipoprotein C-III deficiency, 614028
APOE	83,3	100.0%	99.9%	Alzheimer disease-2, 104310 Hyperlipoproteinemia, type III, 617347 Lipoprotein glomerulopathy, 611771 Sea-blue histiocyte disease, 269600 {?Macular degeneration, age-related}, 603075 {Coronary artery disease, severe, susceptibility to}, 617347
APOPT1	NC	NC	NC	Mitochondrial complex IV deficiency, 220110
APP	107,1	100.0%	99.8%	Alzheimer disease 1, familial, 104300 Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants, 605714
APRT	93,2	100.0%	100.0%	Adenine phosphoribosyltransferase deficiency, 614723
APTX	96,3	94.1%	91.3%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
AQP2	126,1	100.0%	99.9%	Diabetes insipidus, nephrogenic, 125800
AQP5	122,4	100.0%	99.5%	Palmoplantar keratoderma, Bothnian type, 600231
AR	90,8	98.1%	93.7%	Androgen insensitivity, 300068 Androgen insensitivity, partial, with or without breast cancer, 312300 Hypospadias 1, X-linked, 300633 Spinal and bulbar muscular atrophy of Kennedy, 313200 {Prostate cancer, susceptibility to}, 176807
ARCN1	141,9	96.7%	96.6%	Short stature, rhizomelic, with microcephaly, micrognathia, and developmental delay, 617164

ARF1	165,2	100.0%	100.0%	Periventricular nodular heterotopia 8, 618185
ARFGEF2	125,2	99.7%	98.7%	Periventricular heterotopia with microcephaly, 608097
ARG1	159,1	100.0%	100.0%	Argininemia, 207800
ARHGAP26	125,3	99.9%	99.6%	Leukemia, juvenile myelomonocytic, somatic, 607785
ARHGAP31	141,4	99.8%	98.7%	Adams-Oliver syndrome 1, 100300
ARHGDIA	202,4	100.0%	100.0%	Nephrotic syndrome, type 8, 615244
ARHGEF10	119,5	99.8%	98.1%	?Slowed nerve conduction velocity, AD, 608236
ARHGEF18	140,4	99.5%	97.3%	Retinitis pigmentosa 78, 617433
ARHGEF2	113,5	100.0%	99.8%	?Neurodevelopmental disorder with midbrain and hindbrain malformations, 617523
ARHGEF9	51,5	76.1%	71.3%	Epileptic encephalopathy, early infantile, 8, 300607
ARID1A	134,4	99.4%	98.4%	Coffin-Siris syndrome 2, 614607
ARID1B	139,6	99.5%	99.2%	Coffin-Siris syndrome 1, 135900
ARID2	156,4	99.8%	98.5%	Coffin-Siris syndrome 6, 617808
ARL13B	102,2	100.0%	99.4%	Joubert syndrome 8, 612291
ARL2BP	63,8	92.4%	83.1%	Retinitis pigmentosa with or without situs inversus, 615434
ARL3	73,5	99.8%	95.7%	Joubert syndrome 35, 618161 Retinitis pigmentosa 83, 618173
ARL6	100,3	99.8%	98.2%	?Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151 {Bardet-Biedl syndrome 1, modifier of}, 209900
ARL6IP1	62,7	94.8%	76.6%	?Spastic paraplegia 61, autosomal recessive, 615685
ARMC4	107,2	94.4%	93.5%	Ciliary dyskinesia, primary, 23, 615451
ARMC5	170	100.0%	99.6%	ACTH-independent macronodular adrenal hyperplasia 2, 615954
ARMC9	124,8	100.0%	99.3%	Joubert syndrome 30, 617622
ARNT2	120,9	100.0%	100.0%	?Webb-Dattani syndrome, 615926
ARPC1B	139,6	100.0%	100.0%	Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease, 617718
ARR3	83,7	99.9%	99.2%	Myopia 26, X-linked, female-limited, 301010
ARSA	138,5	100.0%	100.0%	Metachromatic leukodystrophy, 250100
ARSB	109,4	99.9%	98.9%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARSE	80,5	97.9%	89.2%	Chondrodysplasia punctata, X-linked recessive, 302950
ARSG	113,1	99.9%	98.6%	Usher syndrome, type IV, 618144
ARV1	108,9	100.0%	99.2%	Epileptic encephalopathy, early infantile, 38, 617020
ARX	49,3	87.3%	79.2%	Epileptic encephalopathy, early infantile, 1, 308350 Hydranencephaly with abnormal genitalia, 300215 Lissencephaly, X-linked 2, 300215 Mental retardation, X-linked 29 and others, 300419

				Partington syndrome, 309510 Proud syndrome, 300004
ASAH1	125,7	99.3%	97.2%	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASB10	106	99.8%	97.4%	Glaucoma 1, open angle, F, 603383
ASCC1	125,4	95.7%	92.0%	?Spinal muscular atrophy with congenital bone fractures 2, 616867 Barrett esophagus/esophageal adenocarcinoma, 614266
ASCL1	297,3	100.0%	100.0%	Central hypoventilation syndrome, congenital, 209880 Haddad syndrome, 209880
ASH1L	143,7	98.7%	98.5%	Mental retardation, autosomal dominant 52, 617796
ASL	123,6	100.0%	98.5%	Argininosuccinic aciduria, 207900
ASNS	82,8	98.6%	92.2%	Asparagine synthetase deficiency, 615574
ASPA	116,1	99.7%	96.9%	Canavan disease, 271900
ASPH	111,3	99.9%	98.8%	Traboulsi syndrome, 601552
ASPM	111,6	99.7%	98.0%	Microcephaly 5, primary, autosomal recessive, 608716
ASPSCR1	123	100.0%	99.8%	Alveolar soft-part sarcoma, 606243
ASS1	97,4	95.0%	87.1%	Citrullinemia, 215700
ASXL1	132,4	100.0%	99.5%	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ASXL2	140,9	99.7%	98.8%	Shashi-Pena syndrome, 617190
ASXL3	138,1	99.7%	99.1%	Bainbridge-Ropers syndrome, 615485
ATAD1	65,2	99.3%	89.8%	Hyperekplexia 4, 618011
ATAD3A	90,3	93.6%	87.5%	Harel-Yoon syndrome, 617183
ATCAY	152,7	100.0%	99.7%	Ataxia, cerebellar, Cayman type, 601238
ATF6	125,2	100.0%	99.3%	Achromatopsia 7, 616517
ATG5	126,9	99.1%	97.0%	?Spinocerebellar ataxia, autosomal recessive 25, 617584
ATIC	113,9	100.0%	99.7%	AICA-ribosiduria due to ATIC deficiency, 608688
ATL1	134,7	99.9%	99.0%	Neuropathy, hereditary sensory, type ID, 613708 Spastic paraplegia 3A, autosomal dominant, 182600
ATL3	115,4	99.8%	97.7%	Neuropathy, hereditary sensory, type IF, 615632
ATM	110,9	99.6%	97.2%	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic, 0 Lymphoma, mantle cell, somatic, 0 T-cell prolymphocytic leukemia, somatic, 0 {Breast cancer, susceptibility to}, 114480
ATN1	155,1	99.9%	99.1%	Dentatorubro-pallidoluysian atrophy, 125370

ATOH7	176,2	98.6%	97.0%	Persistent hyperplastic primary vitreous, autosomal recessive, 221900
ATP11C	77	98.6%	92.3%	?Hemolytic anemia, congenital, X-linked, 301015
ATP13A2	134,1	99.9%	99.7%	Kufor-Rakeb syndrome, 606693 Spastic paraplegia 78, autosomal recessive, 617225
ATP1A1	111,1	100.0%	99.6%	Charcot-Marie-Tooth disease, axonal, type 2DD, 618036 Hypomagnesemia, seizures, and mental retardation 2, 618314
ATP1A2	161,7	100.0%	99.5%	Alternating hemiplegia of childhood 1, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481
ATP1A3	159,8	100.0%	100.0%	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235
ATP2A1	146,4	100.0%	100.0%	Brody myopathy, 601003
ATP2A2	143,2	100.0%	99.8%	Acrokeratosis verruciformis, 101900 Darier disease, 124200
ATP2B3	123	99.6%	97.5%	?Spinocerebellar ataxia, X-linked 1, 302500
ATP2C1	111,6	99.9%	99.3%	Hailey-Hailey disease, 169600
ATP5A1	NC	NC	NC	?Combined oxidative phosphorylation deficiency 22, 616045 ?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4, 615228
ATP5D	NC	NC	NC	Mitochondrial complex V (ATP synthase) deficiency, 618120
ATP5E	NC	NC	NC	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053
ATP6AP1	105,6	99.8%	96.9%	Immunodeficiency 47, 300972
ATP6AP2	44,9	88.4%	64.2%	?Parkinsonism with spasticity, X-linked, 300911 Mental retardation, X-linked, syndromic, Hedera type, 300423
ATP6V0A2	117,4	99.9%	99.0%	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250
ATP6V0A4	105,4	100.0%	99.2%	Renal tubular acidosis, distal, autosomal recessive, 602722
ATP6V1A	133,4	99.6%	97.3%	Cutis laxa, autosomal recessive, type IID, 617403 Epileptic encephalopathy, infantile or early childhood, 3, 618012
ATP6V1B1	172,2	100.0%	100.0%	Renal tubular acidosis with deafness, 267300
ATP6V1B2	120,3	100.0%	99.1%	Deafness, congenital, with onychodystrophy, autosomal dominant, 124480 Zimmermann-Laband syndrome 2, 616455
ATP6V1E1	66,5	92.3%	85.9%	Cutis laxa, autosomal recessive, type IIC, 617402
ATP7A	111,2	99.5%	96.7%	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATP7B	128,7	99.9%	99.1%	Wilson disease, 277900

ATP8A2	115,2	99.9%	99.5%	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268
ATP8B1	114	97.5%	94.6%	Cholestasis, benign recurrent intrahepatic, 243300 Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, progressive familial intrahepatic 1, 211600
ATPAF2	103,5	100.0%	100.0%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
ATR	144,6	99.8%	98.6%	?Cutaneous telangiectasia and cancer syndrome, familial, 614564 Seckel syndrome 1, 210600
ATRX	89,2	99.1%	95.5%	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Alpha-thalassemia/mental retardation syndrome, 301040 Mental retardation-hypotonic facies syndrome, X-linked, 309580
ATXN1	170,2	100.0%	100.0%	Spinocerebellar ataxia 1, 164400
ATXN10	133,9	99.9%	99.8%	Spinocerebellar ataxia 10, 603516
ATXN2	83,1	94.3%	89.9%	Spinocerebellar ataxia 2, 183090 {Amyotrophic lateral sclerosis, susceptibility to, 13}, 183090 {Parkinson disease, late-onset, susceptibility to}, 168600
ATXN3	90,5	92.6%	87.9%	Machado-Joseph disease, 109150
ATXN7	110,9	99.5%	98.0%	Spinocerebellar ataxia 7, 164500
ATXN8OS	NC	NC	NC	Spinocerebellar ataxia 8, 608768 {Parkinson disease, susceptibility to}, 168600
AUH	127	100.0%	99.7%	3-methylglutaconic aciduria, type I, 250950
AURKC	69,1	99.7%	94.3%	Spermatogenic failure 5, 243060
AUTS2	130,6	99.5%	97.7%	Mental retardation, autosomal dominant 26, 615834
AVP	65,2	98.2%	83.8%	Diabetes insipidus, neurohypophyseal, 125700
AVPR2	133,1	100.0%	99.8%	Diabetes insipidus, nephrogenic, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539
AXIN1	140,8	99.7%	98.3%	?Caudal duplication anomaly, 607864 Hepatocellular carcinoma, somatic, 114550
AXIN2	124,2	100.0%	99.9%	Colorectal cancer, somatic, 114500 Oligodontia-colorectal cancer syndrome, 608615
B2M	198,9	100.0%	99.8%	?Amyloidosis, familial visceral, 105200 Immunodeficiency 43, 241600
B3GALNT2	93,9	92.9%	91.0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B3GALT6	81,7	82.6%	77.6%	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B3GAT3	121	99.6%	96.5%	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B3GLCT	96,6	99.7%	99.1%	Peters-plus syndrome, 261540

B4GALNT1	151,2	99.8%	97.9%	Spastic paraplegia 26, autosomal recessive, 609195
B4GALT1	112,2	99.8%	97.7%	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	123,9	99.8%	98.1%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
B4GAT1	136,9	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
B9D1	103,7	92.2%	92.1%	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120
B9D2	105,8	100.0%	100.0%	?Meckel syndrome 10, 614175 Joubert syndrome 34, 614175
BAAT	103	99.8%	97.3%	Hypercholanemia, familial, 607748
BACH2	159	100.0%	99.8%	Immunodeficiency 60, 618394
BAG3	171,4	100.0%	99.9%	Cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954
BANF1	51,1	96.6%	84.1%	Nestor-Guillermo progeria syndrome, 614008
BAP1	104,8	85.0%	82.9%	Tumor predisposition syndrome, 614327
BAX	108,4	100.0%	97.9%	Colorectal cancer, somatic, 114500 T-cell acute lymphoblastic leukemia, somatic, 613065
BBIP1	119,7	97.3%	90.3%	?Bardet-Biedl syndrome 18, 615995
BBS1	146,4	100.0%	100.0%	Bardet-Biedl syndrome 1, 209900
BBS10	158,1	100.0%	99.9%	Bardet-Biedl syndrome 10, 615987
BBS12	187,1	100.0%	100.0%	Bardet-Biedl syndrome 12, 615989
BBS2	150,7	99.9%	99.6%	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	110,2	99.9%	99.2%	Bardet-Biedl syndrome 4, 615982
BBS5	98,5	98.0%	93.3%	Bardet-Biedl syndrome 5, 615983
BBS7	142,9	99.1%	96.5%	Bardet-Biedl syndrome 7, 615984
BBS9	113,6	98.6%	94.4%	Bardet-Biedl syndrome 9, 615986
BCAP31	73,4	93.2%	78.3%	Deafness, dystonia, and cerebral hypomyelination, 300475
BCHE	179,2	100.0%	100.0%	Butyrylcholinesterase deficiency, 617936 {Apnea, postanesthetic, susceptibility to, due to BCHE deficiency}, 617936
BCKDHA	176,9	100.0%	99.8%	Maple syrup urine disease, type Ia, 248600
BCKDHB	123,3	98.6%	92.8%	Maple syrup urine disease, type Ib, 248600
BCKDK	203,5	100.0%	100.0%	Branched-chain ketoacid dehydrogenase kinase deficiency, 614923
BCL10	126,9	100.0%	100.0%	?Immunodeficiency 37, 616098 Lymphoma, MALT, somatic, 137245 {Lymphoma, follicular, somatic}, 605027 {Male germ cell tumor, somatic}, 273300

				{Mesothelioma, somatic}, 156240 {Sezary syndrome, somatic}, 0
BCL11A	145,5	99.5%	98.0%	Dias-Logan syndrome, 617101
BCL11B	127,4	99.9%	98.0%	Immunodeficiency 49, 617237 Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092
BCL2	204	100.0%	100.0%	Leukemia/lymphoma, B-cell, 2, 0
BCL7A	152,5	100.0%	100.0%	B-cell non-Hodgkin lymphoma, high-grade, 0
BCO1	129,2	100.0%	100.0%	?Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300
BCOR	102,7	98.8%	95.3%	Microphthalmia, syndromic 2, 300166
BCR	129,8	86.8%	84.5%	Leukemia, acute lymphocytic, somatic, 613065 Leukemia, chronic myeloid, somatic, 608232
BCS1L	147,9	100.0%	100.0%	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BDP1	135,3	98.2%	94.3%	?Deafness, autosomal recessive 112, 618257
BEAN1	144	99.9%	97.5%	Spinocerebellar ataxia 31, 117210
BEST1	127,2	99.6%	97.1%	Bestrophinopathy, autosomal recessive, 611809 Macular dystrophy, vitelliform, 2, 153700 Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma, 193220 Retinitis pigmentosa, concentric, 613194 Retinitis pigmentosa-50, 613194 Vitreoretinochoroidopathy, 193220
BFSP1	100,1	100.0%	99.1%	Cataract 33, multiple types, 611391
BFSP2	99,3	99.9%	98.2%	Cataract 12, multiple types, 611597
BGN	135,9	100.0%	99.8%	Meester-Loeys syndrome, 300989 Spondyloepimetaphyseal dysplasia, X-linked, 300106
BHLHA9	33,5	94.5%	73.0%	?Camptosynpolydactyly, complex, 607539 Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432
BICD2	150,5	100.0%	99.6%	Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290 Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291
BIN1	113,4	99.9%	98.4%	Centronuclear myopathy 2, 255200
BLK	127,9	100.0%	100.0%	Maturity-onset diabetes of the young, type 11, 613375
BLM	111	99.6%	98.0%	Bloom syndrome, 210900
BLNK	91,4	97.0%	93.1%	?Agammaglobulinemia 4, 613502
BLOC1S3	67,4	100.0%	99.9%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	101,1	99.2%	95.1%	?Hermansky-pudlak syndrome 9, 614171

BLVRA	111,9	100.0%	99.9%	Hyperbiliverdinemia, 614156
BMP1	152,7	100.0%	100.0%	Osteogenesis imperfecta, type XIII, 614856
BMP15	94,3	99.9%	97.9%	Ovarian dysgenesis 2, 300510 Premature ovarian failure 4, 300510
BMP2	163,4	100.0%	100.0%	Brachydactyly, type A2, 112600 Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 617877 {HFE hemochromatosis, modifier of}, 235200
BMP4	173,4	100.0%	100.0%	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625
BMPER	127,1	100.0%	99.5%	Diaphanospondylodysostosis, 608022
BMPR1A	78,2	99.5%	92.9%	Juvenile polyposis syndrome, infantile form, 174900 Polyposis syndrome, hereditary mixed, 2, 610069 Polyposis, juvenile intestinal, 174900
BMPR1B	139,4	100.0%	100.0%	Acromesomelic dysplasia, Demirhan type, 609441 Brachydactyly, type A1, D, 616849 Brachydactyly, type A2, 112600
BMPR2	153,7	99.9%	99.9%	Pulmonary hypertension, familial primary, 1, with or without HHT, 178600 Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600 Pulmonary venoocclusive disease 1, 265450
BMS1	76,4	66.8%	65.6%	?Aplasia cutis congenita, nonsyndromic, 107600
BOLA3	48,1	99.9%	92.5%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BPGM	101,3	100.0%	100.0%	Erythrocytosis, familial, 8, 222800
BPTF	143,6	96.3%	94.6%	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, 617755
BRAF	72,5	92.4%	80.2%	Adenocarcinoma of lung, somatic, 211980 Cardiofaciocutaneous syndrome, 115150 Colorectal cancer, somatic, 0 LEOPARD syndrome 3, 613707 Melanoma, malignant, somatic, 0 Nonsmall cell lung cancer, somatic, 0 Noonan syndrome 7, 613706
BRAT1	142	100.0%	99.3%	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056 Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BRCA1	161,4	99.1%	98.1%	Fanconi anemia, complementation group S, 617883 {Breast-ovarian cancer, familial, 1}, 604370 {Pancreatic cancer, susceptibility to, 4}, 614320
BRCA2	106,2	99.6%	98.7%	Fanconi anemia, complementation group D1, 605724 Wilms tumor, 194070

				{Breast cancer, male, susceptibility to}, 114480 {Breast-ovarian cancer, familial, 2}, 612555 {Glioblastoma 3}, 613029 {Medulloblastoma}, 155255 {Pancreatic cancer 2}, 613347 {Prostate cancer}, 176807
BRDT	107,1	97.2%	92.1%	?Spermatogenic failure 21, 617644
BRF1	109	99.8%	98.1%	Cerebellofaciodental syndrome, 616202
BRIP1	125,8	99.7%	98.8%	Fanconi anemia, complementation group J, 609054 {Breast cancer, early-onset, susceptibility to}, 114480
BRPF1	161,5	100.0%	100.0%	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333
BRWD3	106,4	99.2%	95.6%	Mental retardation, X-linked 93, 300659
BSCL2	105,2	100.0%	100.0%	Encephalopathy, progressive, with or without lipodystrophy, 615924 Lipodystrophy, congenital generalized, type 2, 269700 Neuropathy, distal hereditary motor, type VA, 600794 Silver spastic paraplegia syndrome, 270685
BSND	139,4	100.0%	99.9%	Bartter syndrome, type 4a, 602522 Sensorineural deafness with mild renal dysfunction, 602522
BTD	126,6	99.9%	99.7%	Biotinidase deficiency, 253260
BTK	98	99.9%	99.0%	Agammaglobulinemia, X-linked 1, 300755 Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200
BUB1	126,2	99.9%	98.6%	Colorectal cancer with chromosomal instability, somatic, 0
BUB1B	122	99.8%	98.7%	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430
BVES	112,6	99.7%	98.5%	Muscular dystrophy, limb-girdle, autosomal recessive 25, 616812
C11orf70	NC	NC	NC	Ciliary dyskinesia, primary, 38, 618063
C12orf4	128,2	99.9%	99.3%	Mental retardation, autosomal recessive 66, 618221
C12orf57	145,6	100.0%	100.0%	Temtamy syndrome, 218340
C12orf65	110,4	100.0%	99.6%	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035
C15orf41	122	100.0%	99.6%	Dyserythropoietic anemia, congenital, type Ib, 615631
C19orf12	104,2	100.0%	99.8%	?Spastic paraplegia 43, autosomal recessive, 615043 Neurodegeneration with brain iron accumulation 4, 614298
C19orf70	NC	NC	NC	Combined oxidative phosphorylation deficiency 37, 618329
C1GALT1C1	139,9	100.0%	99.0%	Tn polyagglutination syndrome, somatic, 300622
C1QA	196,1	100.0%	100.0%	C1q deficiency, 613652

C1QB	161,5	100.0%	100.0%	C1q deficiency, 613652
C1QBP	66,7	91.9%	79.5%	Combined oxidative phosphorylation deficiency 33, 617713
C1QC	187	100.0%	99.6%	C1q deficiency, 613652
C1QTNF5	154,2	97.2%	90.6%	Retinal degeneration, late-onset, autosomal dominant, 605670
C1R	151	100.0%	100.0%	Ehlers-Danlos syndrome, periodontal type, 1, 130080
C1S	96,8	99.8%	97.8%	C1s deficiency, 613783 Ehlers-Danlos syndrome, periodontal type, 2, 617174
C2	126,3	100.0%	100.0%	C2 deficiency, 217000 {Macular degeneration, age-related, 14, reduced risk of}, 615489
C21orf2	NC	NC	NC	Retinal dystrophy with macular staphyloma, 617547 Spondylometaphyseal dysplasia, axial, 602271
C21orf59	NC	NC	NC	Ciliary dyskinesia, primary, 26, 615500
C2CD3	116,9	95.8%	95.2%	Orofaciodigital syndrome XIV, 615948
C2orf71	NC	NC	NC	Retinitis pigmentosa 54, 613428
C3	141,6	100.0%	99.4%	C3 deficiency, 613779 {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 {Macular degeneration, age-related, 9}, 611378
C4A	86,4	98.5%	96.1%	C4a deficiency, 614380 [Blood group, Rodgers], 614374
C4B	83	98.7%	96.6%	C4B deficiency, 614379
C4orf26	NC	NC	NC	Amelogenesis imperfecta, type IIA4, 614832
C5	120,9	99.5%	97.7%	C5 deficiency, 609536 [Eculizumab, poor response to], 615749
C5orf42	NC	NC	NC	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
C6	139,1	100.0%	99.6%	C6 deficiency, 612446 Combined C6/C7 deficiency, 0
C7	113,1	99.7%	97.3%	C7 deficiency, 610102
C7orf43	NC	NC	NC	?Microcephaly 25, primary, autosomal recessive, 618351
C8A	105,6	100.0%	99.4%	C8 deficiency, type I, 613790
C8B	105,6	99.9%	98.7%	C8 deficiency, type II, 613789
C8orf37	144,1	99.8%	99.4%	Bardet-Biedl syndrome 21, 617406 Cone-rod dystrophy 16, 614500 Retinitis pigmentosa 64, 614500
C9	121	99.9%	99.3%	C9 deficiency, 613825 {Macular degeneration, age-related, 15, susceptibility to}, 615591

C9orf72	97,2	99.5%	96.8%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1, 105550
CA12	100,7	100.0%	100.0%	Hyperchlorhidrosis, isolated, 143860
CA2	137,4	100.0%	99.9%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA4	162,1	100.0%	100.0%	Retinitis pigmentosa 17, 600852
CA5A	93,2	99.6%	95.7%	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CA8	107,5	99.7%	97.6%	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CABP2	76,6	78.5%	71.0%	Deafness, autosomal recessive 93, 614899
CABP4	148,3	100.0%	100.0%	Cone-rod synaptic disorder, congenital nonprogressive, 610427
CACNA1A	92,4	97.8%	94.7%	Epileptic encephalopathy, early infantile, 42, 617106 Episodic ataxia, type 2, 108500 Migraine, familial hemiplegic, 1, 141500 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Spinocerebellar ataxia 6, 183086
CACNA1B	134,4	99.3%	97.3%	?Dystonia 23, 614860
CACNA1C	141	99.9%	99.1%	Brugada syndrome 3, 611875 Timothy syndrome, 601005
CACNA1D	127,4	98.0%	97.7%	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896
CACNA1E	120,9	99.8%	99.2%	Epileptic encephalopathy, early infantile, 69, 618285
CACNA1F	84,9	99.8%	97.1%	Aland Island eye disease, 300600 Cone-rod dystrophy, X-linked, 3, 300476 Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071
CACNA1G	148,6	100.0%	99.8%	Spinocerebellar ataxia 42, 616795 Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087
CACNA1H	139,3	99.4%	98.1%	Hyperaldosteronism, familial, type IV, 617027 {Epilepsy, childhood absence, susceptibility to, 6}, 611942 {Epilepsy, idiopathic generalized, susceptibility to, 6}, 611942
CACNA1S	120,9	100.0%	99.7%	Hypokalemic periodic paralysis, type 1, 170400 {Malignant hyperthermia susceptibility 5}, 601887 {Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580
CACNA2D4	95,9	99.0%	97.1%	Retinal cone dystrophy 4, 610478
CACNB2	130,1	99.9%	99.2%	Brugada syndrome 4, 611876
CACNB4	97,8	97.2%	95.5%	Episodic ataxia, type 5, 613855 {Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 {Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682
CACNG2	113,7	100.0%	99.4%	?Mental retardation, autosomal dominant 10, 614256
CAD	136,7	99.9%	99.2%	Epileptic encephalopathy, early infantile, 50, 616457

CALM1	95,8	99.9%	97.8%	Long QT syndrome 14, 616247 Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916
CALM2	44	66.7%	61.3%	Long QT syndrome 15, 616249
CALR	111,8	98.1%	91.7%	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
CAMK2A	117,3	100.0%	99.8%	?Mental retardation, autosomal recessive 63, 618095 Mental retardation, autosomal dominant 53, 617798
CAMK2B	111,8	100.0%	100.0%	Mental retardation, autosomal dominant 54, 617799
CAMTA1	179,5	100.0%	99.7%	Cerebellar ataxia, nonprogressive, with mental retardation, 614756
CANT1	144,9	100.0%	100.0%	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719
CAPN1	162,5	100.0%	100.0%	Spastic paraplegia 76, autosomal recessive, 616907
CAPN3	98,3	99.2%	97.0%	Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129 Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600
CAPN5	153,6	100.0%	99.8%	Vitreoretinopathy, neovascular inflammatory, 193235
CARD11	138,2	100.0%	99.6%	B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11A, 615206 Immunodeficiency 11B with atopic dermatitis, 617638
CARD14	124,5	100.0%	99.2%	Pityriasis rubra pilaris, 173200 Psoriasis 2, 602723
CARD9	133,2	100.0%	99.5%	Candidiasis, familial, 2, autosomal recessive, 212050
CARMIL2	137,3	98.4%	96.6%	Immunodeficiency 58, 618131
CARS2	128,2	100.0%	100.0%	Combined oxidative phosphorylation deficiency 27, 616672
CASK	85,1	99.5%	94.5%	FG syndrome 4, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 Mental retardation, with or without nystagmus, 300422
CASP10	106,2	99.8%	98.2%	Autoimmune lymphoproliferative syndrome, type II, 603909 Gastric cancer, somatic, 613659 Lymphoma, non-Hodgkin, somatic, 605027
CASP14	85,8	100.0%	99.9%	Ichthyosis, congenital, autosomal recessive 12, 617320
CASP8	128,1	95.6%	95.2%	?Autoimmune lymphoproliferative syndrome, type IIB, 607271 Hepatocellular carcinoma, somatic, 114550 {Breast cancer, protection against}, 114480 {Lung cancer, protection against}, 211980
CASQ1	93,5	99.8%	98.2%	Myopathy, vacuolar, with CASQ1 aggregates, 616231
CASQ2	113,5	100.0%	99.1%	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CASR	154,3	100.0%	99.7%	Hyperparathyroidism, neonatal, 239200

				Hypocalcemia, autosomal dominant, 601198 Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 Hypocalciuric hypercalcemia, type I, 145980 {Epilepsy idiopathic generalized, susceptibility to, 8}, 612899
CAST	112,4	99.8%	97.1%	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295
CAT	136,9	100.0%	100.0%	Acatalasemia, 614097
CATSPER1	115,6	100.0%	99.6%	Spermatogenic failure 7, 612997
CAV1	189,3	100.0%	100.0%	?Lipodystrophy, congenital generalized, type 3, 612526 Lipodystrophy, familial partial, type 7, 606721 Pulmonary hypertension, primary, 3, 615343
CAV3	220,8	100.0%	100.0%	Cardiomyopathy, familial hypertrophic, 192600 Creatine phosphokinase, elevated serum, 123320 Long QT syndrome 9, 611818 Myopathy, distal, Tateyama type, 614321 Rippling muscle disease 2, 606072
CAVIN1	174,1	100.0%	100.0%	Lipodystrophy, congenital generalized, type 4, 613327
CBL	126	97.3%	97.0%	?Juvenile myelomonocytic leukemia, 607785 Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563
CBS	123,3	99.9%	99.0%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CBX2	149,7	100.0%	100.0%	?46XY sex reversal 5, 613080
CC2D1A	135	100.0%	99.5%	Mental retardation, autosomal recessive 3, 608443
CC2D2A	111,7	99.0%	97.1%	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284
CCBE1	75,3	99.8%	99.1%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CCDC103	116,2	100.0%	99.8%	Ciliary dyskinesia, primary, 17, 614679
CCDC114	134,1	100.0%	99.8%	Ciliary dyskinesia, primary, 20, 615067
CCDC115	77,9	88.9%	87.1%	Congenital disorder of glycosylation, type IIo, 616828
CCDC151	127,2	100.0%	100.0%	Ciliary dyskinesia, primary, 30, 616037
CCDC174	120,5	99.7%	97.5%	Hypotonia, infantile, with psychomotor retardation, 616816
CCDC22	97,5	98.8%	94.5%	Ritscher-Schinzel syndrome 2, 300963
CCDC39	86,7	99.4%	96.8%	Ciliary dyskinesia, primary, 14, 613807
CCDC40	112	99.4%	98.4%	Ciliary dyskinesia, primary, 15, 613808
CCDC47	143,9	99.5%	96.6%	Trichohepatoneurodevelopmental syndrome, 618268
CCDC50	122,1	100.0%	99.7%	?Deafness, autosomal dominant 44, 607453

CCDC65	80,3	99.6%	97.1%	Ciliary dyskinesia, primary, 27, 615504
CCDC78	135,9	100.0%	100.0%	?Centronuclear myopathy 4, 614807
CCDC8	186,9	100.0%	100.0%	3-M syndrome 3, 614205
CCDC88A	92,8	99.3%	96.7%	?PEHO syndrome-like, 617507
CCDC88C	108,2	100.0%	99.4%	?Spinocerebellar ataxia 40, 616053 Hydrocephalus, congenital, 1, 236600
CCM2	133,4	99.2%	97.9%	Cerebral cavernous malformations-2, 603284
CCND2	135,7	100.0%	100.0%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938
CCNK	86,8	90.6%	87.0%	?Intellectual developmental disorder with hypertelorism and distinctive facies, 618147
CCNO	130,9	100.0%	99.8%	Ciliary dyskinesia, primary, 29, 615872
CCT5	117,9	99.9%	98.9%	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CD151	122,2	100.0%	100.0%	Nephropathy with pretibial epidermolysis bullosa and deafness, 609057 [Blood group, Raph], 179620
CD164	121,8	99.0%	94.5%	?Deafness, autosomal dominant 66, 616969
CD19	108,8	100.0%	99.9%	Immunodeficiency, common variable, 3, 613493
CD247	95,5	100.0%	99.5%	?Immunodeficiency 25, 610163
CD27	105,3	100.0%	100.0%	Lymphoproliferative syndrome 2, 615122
CD2AP	121,7	99.7%	98.3%	Glomerulosclerosis, focal segmental, 3, 607832
CD320	113,9	100.0%	99.9%	Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646
CD36	119,8	99.7%	99.1%	Platelet glycoprotein IV deficiency, 608404 [Macrothrombocytopenia], 0 {Coronary heart disease, susceptibility to, 7}, 610938 {Malaria, cerebral, reduced risk of}, 611162 {Malaria, cerebral, susceptibility to}, 611162
CD3D	138,9	100.0%	99.9%	Immunodeficiency 19, 615617
CD3E	125,9	100.0%	98.9%	Immunodeficiency 18, 615615 Immunodeficiency 18, SCID variant, 615615
CD3G	136,6	100.0%	100.0%	Immunodeficiency 17, CD3 gamma deficient, 615607
CD4	120,5	100.0%	99.6%	OKT4 epitope deficiency, 613949
CD40	147,8	100.0%	99.9%	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	109,6	97.2%	87.3%	Immunodeficiency, X-linked, with hyper-IgM, 308230
CD55	134,6	95.5%	90.4%	Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300 [Blood group Cromer], 613793
CD59	149,6	93.5%	85.8%	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD70	109	100.0%	99.2%	Lymphoproliferative syndrome 3, 618261
CD79A	133,5	100.0%	99.3%	Agammaglobulinemia 3, 613501

CD79B	194,3	100.0%	100.0%	Agammaglobulinemia 6, 612692
CD81	158,3	100.0%	100.0%	Immunodeficiency, common variable, 6, 613496
CD8A	150,8	100.0%	99.9%	CD8 deficiency, familial, 608957
CD96	140,9	99.9%	99.4%	C syndrome, 211750
CDAN1	112,4	100.0%	99.6%	Dyserythropoietic anemia, congenital, type Ia, 224120
CDC14A	150,1	99.6%	97.4%	Deafness, autosomal recessive 32, with or without immotile sperm, 608653
CDC42	90,2	97.6%	89.1%	Takenouchi-Kosaki syndrome, 616737
CDC45	138,9	99.6%	98.1%	Meier-Gorlin syndrome 7, 617063
CDC6	139,4	99.9%	99.8%	?Meier-Gorlin syndrome 5, 613805
CDC73	113,6	99.9%	98.8%	Hyperparathyroidism, familial primary, 145000 Hyperparathyroidism-jaw tumor syndrome, 145001 Parathyroid adenoma with cystic changes, 145001 Parathyroid carcinoma, 608266
CDCA7	112	100.0%	99.5%	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910
CDH1	104,5	99.2%	99.0%	Blepharocheilodontic syndrome 1, 119580 Endometrial carcinoma, somatic, 608089 Gastric cancer, hereditary diffuse, with or without cleft lip and/or palate, 137215 Ovarian cancer, somatic, 167000 {Breast cancer, lobular}, 114480 {Prostate cancer, susceptibility to}, 176807
CDH11	126,5	100.0%	100.0%	Elsahy-Waters syndrome, 211380
CDH15	157,7	100.0%	99.8%	Mental retardation, autosomal dominant 3, 612580
CDH23	172,7	100.0%	100.0%	Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D, 601067 Usher syndrome, type 1D/F digenic, 601067 {Pituitary adenoma 5, multiple types}, 617540
CDH3	140,5	100.0%	99.8%	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 Hypotrichosis, congenital, with juvenile macular dystrophy, 601553
CDHR1	143,6	99.9%	99.0%	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660
CDK10	131,3	100.0%	100.0%	Al Kaissi syndrome, 617694
CDK13	126,5	99.9%	98.2%	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360
CDK5	107,2	100.0%	99.8%	?Lissencephaly 7 with cerebellar hypoplasia, 616342
CDK5RAP2	107	99.8%	98.8%	Microcephaly 3, primary, autosomal recessive, 604804
CDK6	110	99.6%	97.4%	?Microcephaly 12, primary, autosomal recessive, 616080
CDKL5	100	95.1%	93.1%	Epileptic encephalopathy, early infantile, 2, 300672

CDKN1B	151,5	99.9%	99.4%	Multiple endocrine neoplasia, type IV, 610755
CDKN1C	100,1	89.8%	81.7%	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732
CDKN2A	121,7	92.3%	92.3%	Melanoma and neural system tumor syndrome, 155755 Orolaryngeal cancer, multiple, 0 Pancreatic cancer/melanoma syndrome, 606719 {Melanoma, cutaneous malignant, 2}, 155601
CDON	107	100.0%	99.0%	Holoprosencephaly 11, 614226
CDSN	131	100.0%	100.0%	Hypotrichosis 2, 146520 Peeling skin syndrome 1, 270300
CDT1	130,9	100.0%	99.9%	Meier-Gorlin syndrome 4, 613804
CEACAM16	130,5	100.0%	100.0%	Deafness, autosomal dominant 4B, 614614 Deafness, autosomal recessive 113, 618410
CEBPA	139,8	99.9%	99.1%	?Leukemia, acute myeloid, 601626 Leukemia, acute myeloid, somatic, 601626
CEBPE	99,1	100.0%	99.9%	Specific granule deficiency, 245480
CEL	146,5	94.0%	90.4%	Maturity-onset diabetes of the young, type VIII, 609812
CENPE	76,6	98.5%	93.2%	?Microcephaly 13, primary, autosomal recessive, 616051
CENPF	139,9	99.8%	98.7%	Stromme syndrome, 243605
CENPJ	136	99.9%	99.2%	?Seckel syndrome 4, 613676 Microcephaly 6, primary, autosomal recessive, 608393
CEP104	104	99.3%	97.5%	Joubert syndrome 25, 616781
CEP120	131,7	100.0%	99.4%	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP135	89,9	98.8%	92.6%	Microcephaly 8, primary, autosomal recessive, 614673
CEP152	144,4	99.7%	98.2%	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823
CEP164	89,3	99.8%	98.0%	Nephronophthisis 15, 614845
CEP19	181,7	100.0%	100.0%	Morbid obesity and spermatogenic failure, 615703
CEP250	99,8	99.9%	98.8%	Cone-rod dystrophy and hearing loss 2, 618358
CEP290	82,6	97.3%	91.7%	?Bardet-Biedl syndrome 14, 615991 Joubert syndrome 5, 610188 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Senior-Loken syndrome 6, 610189
CEP41	78,2	98.9%	94.4%	Joubert syndrome 15, 614464
CEP55	124,5	100.0%	100.0%	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500

CEP57	85,1	98.7%	91.6%	Mosaic variegated aneuploidy syndrome 2, 614114
CEP63	120,8	98.2%	94.7%	?Seckel syndrome 6, 614728
CEP78	120,1	99.7%	97.6%	Cone-rod dystrophy and hearing loss, 617236
CEP83	108,8	99.4%	96.6%	Nephronophthisis 18, 615862
CERKL	114,1	99.4%	97.2%	Retinitis pigmentosa 26, 608380
CERS1	70	92.6%	81.4%	?Epilepsy, progressive myoclonic, 8, 616230
CERS3	95,2	99.8%	98.2%	Ichthyosis, congenital, autosomal recessive 9, 615023
CES1	124,6	99.4%	97.2%	Drug metabolism, altered, CES1-related, 618057
CETP	120,6	100.0%	99.9%	Hyperalphalipoproteinemia, 143470 [High density lipoprotein cholesterol level QTL 10], 143470
CFAP43	119	99.7%	97.9%	Spermatogenic failure 19, 617592
CFAP44	110,2	99.5%	98.2%	?Spermatogenic failure 20, 617593
CFAP53	131,8	99.1%	97.0%	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFAP69	70	98.5%	92.8%	Spermatogenic failure 24, 617959
CFB	119,4	100.0%	99.9%	?Complement factor B deficiency, 615561 {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924 {Macular degeneration, age-related, 14, reduced risk of}, 615489
CFC1	125,8	91.0%	80.1%	Heterotaxy, visceral, 2, autosomal, 605376
CFD	113,8	96.9%	89.7%	Complement factor D deficiency, 613912
CFH	155,4	99.4%	97.9%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814 {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 {Macular degeneration, age-related, 4}, 610698
CFHR5	96,6	99.8%	97.5%	Nephropathy due to CFHR5 deficiency, 614809
CFI	139	99.5%	97.0%	Complement factor I deficiency, 610984 {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 {Macular degeneration, age-related, 13, susceptibility to}, 615439
CFL2	119,2	100.0%	99.2%	Nemaline myopathy 7, autosomal recessive, 610687
CFP	98,4	99.7%	97.4%	Properdin deficiency, X-linked, 312060
CFTR	113,5	99.4%	97.4%	Congenital bilateral absence of vas deferens, 277180 Cystic fibrosis, 219700 Sweat chloride elevation without CF, 0 {Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400 {Hypertrypsinemia, neonatal}, 0 {Pancreatitis, hereditary}, 167800
CHAMP1	172,9	100.0%	100.0%	Mental retardation, autosomal dominant 40, 616579

CHAT	117,1	95.4%	86.9%	Myasthenic syndrome, congenital, 6, presynaptic, 254210
CHCHD10	26,1	63.1%	38.4%	?Myopathy, isolated mitochondrial, autosomal dominant, 616209 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 Spinal muscular atrophy, Jokela type, 615048
CHCHD2	69,7	99.9%	93.7%	Parkinson disease 22, autosomal dominant, 616710
CHD1	108,1	98.2%	91.9%	Pilarowski-Bjornsson syndrome, 617682
CHD2	123,9	99.3%	99.0%	Epileptic encephalopathy, childhood-onset, 615369
CHD3	95	97.7%	94.0%	Snijders Blok-Campeau syndrome, 618205
CHD4	111,8	100.0%	99.9%	Sifrim-Hitz-Weiss syndrome, 617159
CHD7	137	99.9%	99.4%	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CHEK2	88,6	83.8%	80.1%	Li-Fraumeni syndrome, 609265 Osteosarcoma, somatic, 259500 {Breast and colorectal cancer, susceptibility to}, 0 {Breast cancer, susceptibility to}, 114480 {Prostate cancer, familial, susceptibility to}, 176807
CHKB	115,4	100.0%	100.0%	Muscular dystrophy, congenital, megaconial type, 602541
CHM	100,3	98.3%	92.0%	Choroideremia, 303100
CHMP1A	123,2	100.0%	99.8%	Pontocerebellar hypoplasia, type 8, 614961
CHMP2B	90,2	99.5%	97.7%	Amyotrophic lateral sclerosis 17, 614696 Dementia, familial, nonspecific, 600795
CHMP4B	139	100.0%	98.9%	Cataract 31, multiple types, 605387
CHN1	147,2	99.9%	99.4%	Duane retraction syndrome 2, 604356
CHRDL1	88,1	99.9%	98.8%	Megalocornea 1, X-linked, 309300
CHRM3	131	100.0%	100.0%	?Prune belly syndrome, 100100
CHRNA1	92,6	94.6%	93.3%	Multiple pterygium syndrome, lethal type, 253290 Myasthenic syndrome, congenital, 1A, slow-channel, 601462 Myasthenic syndrome, congenital, 1B, fast-channel, 608930
CHRNA2	174,4	100.0%	100.0%	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA4	109,8	99.9%	99.2%	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction, susceptibility to}, 188890
CHRNB1	128,2	100.0%	99.7%	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314 Myasthenic syndrome, congenital, 2A, slow-channel, 616313
CHRNB2	160,6	99.7%	98.0%	Epilepsy, nocturnal frontal lobe, 3, 605375
CHRND	140,4	99.8%	98.0%	?Myasthenic syndrome, congenital, 3A, slow-channel, 616321 ?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323 Multiple pterygium syndrome, lethal type, 253290

				Myasthenic syndrome, congenital, 3B, fast-channel, 616322
CHRNE	167,8	100.0%	100.0%	Myasthenic syndrome, congenital, 4A, slow-channel, 605809 Myasthenic syndrome, congenital, 4B, fast-channel, 616324 Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931
CHRNG	141,3	100.0%	100.0%	Escobar syndrome, 265000 Multiple pterygium syndrome, lethal type, 253290
CHST11	188,8	100.0%	100.0%	?Osteochondrodysplasia, brachydactyly, and overlapping malformed digits, 618167
CHST14	160,6	99.9%	98.9%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	133,8	100.0%	100.0%	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHST6	279,8	100.0%	100.0%	Macular corneal dystrophy, 217800
CHST8	257,9	100.0%	100.0%	?Peeling skin syndrome 3, 616265
CHSY1	125,9	99.3%	97.9%	Temtamy preaxial brachydactyly syndrome, 605282
CHUK	126,5	99.8%	99.2%	Cocoon syndrome, 613630
CIB1	122,3	99.3%	96.3%	Epidermodysplasia verruciformis 3, 618267
CIB2	198	99.9%	99.4%	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CIC	72,7	64.7%	63.3%	Mental retardation, autosomal dominant 45, 617600
CIDEC	83,6	99.9%	96.4%	?Lipodystrophy, familial partial, type 5, 615238
CIITA	148,8	100.0%	99.9%	Bare lymphocyte syndrome, type II, complementation group A, 209920 {Rheumatoid arthritis, susceptibility to}, 180300
CISD2	113,6	83.4%	83.3%	Wolfram syndrome 2, 604928
CIT	101,4	99.9%	98.5%	Microcephaly 17, primary, autosomal recessive, 617090
CITED2	149,7	99.2%	99.0%	Atrial septal defect 8, 614433 Ventricular septal defect 2, 614431
CKAP2L	156	99.9%	98.9%	Filippi syndrome, 272440
CLCF1	87,2	100.0%	99.6%	Cold-induced sweating syndrome 2, 610313
CLCN1	125,1	100.0%	99.8%	Myotonia congenita, dominant, 160800 Myotonia congenita, recessive, 255700 Myotonia levior, recessive, 0
CLCN2	115,1	100.0%	99.7%	Hyperaldosteronism, familial, type II, 605635 Leukoencephalopathy with ataxia, 615651 {Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628 {Epilepsy, juvenile absence, susceptibility to, 2}, 607628 {Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628
CLCN4	105,7	99.9%	98.9%	Raynaud-Claes syndrome, 300114
CLCN5	104,3	99.7%	96.5%	Dent disease, 300009

				Hypophosphatemic rickets, 300554 Nephrolithiasis, type I, 310468 Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990
CLCN7	146,7	99.8%	98.7%	Osteopetrosis, autosomal dominant 2, 166600 Osteopetrosis, autosomal recessive 4, 611490
CLCNKA	112	99.8%	97.3%	Bartter syndrome, type 4b, digenic, 613090
CLCNKB	99,4	99.7%	97.1%	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090
CLDN1	122,5	100.0%	100.0%	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626
CLDN10	138,2	100.0%	100.0%	HELIX syndrome, 617671
CLDN14	111,7	100.0%	99.9%	Deafness, autosomal recessive 29, 614035
CLDN16	126,6	100.0%	100.0%	Hypomagnesemia 3, renal, 248250
CLDN19	125,4	99.1%	95.1%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLEC7A	148,5	100.0%	99.9%	Candidiasis, familial, 4, autosomal recessive, 613108 {Aspergillosis, susceptibility to}, 614079
CLIC2	73,7	99.9%	96.8%	?Mental retardation, X-linked, syndromic 32, 300886
CLIC5	97,6	100.0%	99.9%	?Deafness, autosomal recessive 103, 616042
CLMP	86,2	100.0%	99.6%	Congenital short bowel syndrome, 615237
CLN3	114,7	92.6%	91.9%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	138,7	99.9%	98.8%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	132,3	100.0%	99.9%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	144,5	83.5%	83.5%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLP1	135,7	100.0%	100.0%	Pontocerebellar hypoplasia, type 10, 615803
CLPB	125,6	99.8%	97.9%	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CLPP	139,3	100.0%	99.2%	Perrault syndrome 3, 614129
CLPX	149,9	99.7%	98.4%	?Protoporphyria, erythropoietic, 2, 618015
CLRN1	135,3	100.0%	99.4%	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902
CLTC	153,1	100.0%	99.7%	Mental retardation, autosomal dominant 56, 617854
CNBP	117,6	100.0%	100.0%	Myotonic dystrophy 2, 602668
CNGA1	110,4	92.5%	86.5%	Retinitis pigmentosa 49, 613756
CNGA3	149,6	100.0%	99.7%	Achromatopsia 2, 216900
CNGB1	107,4	99.5%	98.0%	Retinitis pigmentosa 45, 613767
CNGB3	101,5	98.6%	93.9%	Achromatopsia 3, 262300

				Macular degeneration, juvenile, 248200
CNKSR2	89	98.5%	92.3%	Mental retardation, X-linked, syndromic, Houge type, 301008
CNNM2	199,8	100.0%	100.0%	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNNM4	161,6	100.0%	99.5%	Jalili syndrome, 217080
CNPY3	78,4	100.0%	100.0%	Epileptic encephalopathy, early infantile, 60, 617929
CNTN1	128	99.7%	98.7%	?Myopathy, congenital, Compton-North, 612540
CNTN2	123,7	92.7%	92.7%	?Epilepsy, myoclonic, familial adult, 5, 615400
CNTNAP1	157,2	99.9%	99.1%	Hypomyelinating neuropathy, congenital, 3, 618186 Lethal congenital contracture syndrome 7, 616286
CNTNAP2	127,1	100.0%	99.8%	Cortical dysplasia-focal epilepsy syndrome, 610042 Pitt-Hopkins like syndrome 1, 610042 {Autism susceptibility 15}, 612100
COA5	74,9	86.6%	83.5%	?Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 3, 616500
COA6	112,5	99.4%	97.0%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 4, 616501
COA7	122	100.0%	100.0%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387
COASY	172,7	100.0%	100.0%	Neurodegeneration with brain iron accumulation 6, 615643 Pontocerebellar hypoplasia, type 12, 618266
COCH	159,5	100.0%	99.9%	?Deafness, autosomal recessive 110, 618094 Deafness, autosomal dominant 9, 601369
COG1	108,4	100.0%	99.9%	Congenital disorder of glycosylation, type IIg, 611209
COG2	124,6	99.7%	98.1%	?Congenital disorder of glycosylation, type IIq, 617395
COG4	94,5	100.0%	99.6%	Congenital disorder of glycosylation, type IIj, 613489 Saul-Wilson syndrome, 618150
COG5	126,3	99.9%	98.4%	Congenital disorder of glycosylation, type IIi, 613612
COG6	90,4	99.1%	96.0%	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328
COG7	106,1	100.0%	99.7%	Congenital disorder of glycosylation, type IIe, 608779
COG8	145	100.0%	98.5%	Congenital disorder of glycosylation, type IIh, 611182
COL10A1	106,8	100.0%	99.9%	Metaphyseal chondrodysplasia, Schmid type, 156500
COL11A1	96,6	97.9%	94.0%	Fibrochondrogenesis 1, 228520 Marshall syndrome, 154780 Stickler syndrome, type II, 604841 {Lumbar disc herniation, susceptibility to}, 603932
COL11A2	111,6	100.0%	99.4%	Deafness, autosomal dominant 13, 601868 Deafness, autosomal recessive 53, 609706

				Fibrochondrogenesis 2, 614524 Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840 Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150
COL12A1	124,2	99.8%	99.1%	?Ullrich congenital muscular dystrophy 2, 616470 Bethlem myopathy 2, 616471
COL13A1	89,8	100.0%	99.3%	Myasthenic syndrome, congenital, 19, 616720
COL17A1	104,5	99.3%	96.9%	Epidermolysis bullosa, junctional, localisata variant, 226650 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epithelial recurrent erosion dystrophy, 122400
COL18A1	133,5	99.5%	96.9%	Knobloch syndrome, type 1, 267750
COL1A1	141	99.8%	98.4%	Caffey disease, 114000 Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060 Osteogenesis imperfecta, type I, 166200 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type III, 259420 Osteogenesis imperfecta, type IV, 166220 {Bone mineral density variation QTL, osteoporosis}, 166710
COL1A2	93,3	98.5%	94.6%	Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821 Ehlers-Danlos syndrome, cardiac valvular type, 225320 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type III, 259420 Osteogenesis imperfecta, type IV, 166220 {Osteoporosis, postmenopausal}, 166710
COL25A1	131,9	99.5%	98.9%	Fibrosis of extraocular muscles, congenital, 5, 616219
COL27A1	142,2	99.9%	99.2%	Steel syndrome, 615155
COL2A1	112,2	100.0%	99.7%	Achondrogenesis, type II or hypochondrogenesis, 200610 Avascular necrosis of the femoral head, 608805 Czech dysplasia, 609162 Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 Kniest dysplasia, 156550 Legg-Calve-Perthes disease, 150600 Osteoarthritis with mild chondrodysplasia, 604864 Platyspondylic skeletal dysplasia, Torrance type, 151210 SED congenita, 183900 SMED Strudwick type, 184250 Spondyloepiphyseal dysplasia, Stanescu type, 616583 Spondyloperipheral dysplasia, 271700 Stickler sydrome, type I, nonsyndromic ocular, 609508

				Stickler syndrome, type I, 108300 Vitreoretinopathy with phalangeal epiphyseal dysplasia, 0
COL3A1	99,2	99.3%	96.8%	Ehlers-Danlos syndrome, vascular type, 130050 Polymicrogyria with or without vascular-type EDS, 618343
COL4A1	95,7	99.6%	97.3%	?Retinal arteries, tortuosity of, 180000 Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Brain small vessel disease with or without ocular anomalies, 175780 Schizencephaly, 269160 {Hemorrhage, intracerebral, susceptibility to}, 614519
COL4A2	109	100.0%	99.1%	Brain small vessel disease 2, 614483 {Hemorrhage, intracerebral, susceptibility to}, 614519
COL4A3	90,5	99.6%	97.7%	Alport syndrome 2, autosomal recessive, 203780 Alport syndrome 3, autosomal dominant, 104200 Hematuria, benign familial, 141200
COL4A3BP	133,1	99.7%	97.8%	Mental retardation, autosomal dominant 34, 616351
COL4A4	92,4	99.8%	97.4%	Alport syndrome 2, autosomal recessive, 203780 Hematuria, familial benign, 141200
COL4A5	56,8	96.8%	85.7%	Alport syndrome 1, X-linked, 301050
COL4A6	81,4	97.1%	92.0%	?Deafness, X-linked 6, 300914
COL5A1	136,4	99.9%	98.9%	Ehlers-Danlos syndrome, classic type, 1, 130000
COL5A2	100,2	99.9%	99.4%	Ehlers-Danlos syndrome, classic type, 2, 130010
COL6A1	158,8	100.0%	99.8%	Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090
COL6A2	175,3	100.0%	99.8%	?Myosclerosis, congenital, 255600 Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090
COL6A3	154	100.0%	99.8%	Bethlem myopathy 1, 158810 Dystonia 27, 616411 Ullrich congenital muscular dystrophy 1, 254090
COL7A1	139,7	99.8%	98.9%	EBD inversa, 226600 EBD, Bart type, 132000 EBD, localisata variant, 0 Epidermolysis bullosa dystrophica, AD, 131750 Epidermolysis bullosa dystrophica, AR, 226600 Epidermolysis bullosa pruriginosa, 604129 Epidermolysis bullosa, pretibial, 131850 Toenail dystrophy, isolated, 607523

				Transient bullous of the newborn, 131705
COL8A2	119,3	100.0%	99.6%	Corneal dystrophy, Fuchs endothelial, 1, 136800 Corneal dystrophy, posterior polymorphous 2, 609140
COL9A1	132,3	100.0%	99.7%	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	95,2	99.9%	98.8%	?Stickler syndrome, type V, 614284 Epiphyseal dysplasia, multiple, 2, 600204
COL9A3	107,8	99.6%	96.8%	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 {Intervertebral disc disease, susceptibility to}, 603932
COLEC10	120,1	100.0%	99.9%	3MC syndrome 3, 248340
COLEC11	180,6	100.0%	100.0%	3MC syndrome 2, 265050
COLGALT1	149,5	97.8%	92.1%	Brain small vessel disease 3, 618360
COLQ	100,9	99.8%	97.5%	Myasthenic syndrome, congenital, 5, 603034
COMP	132	95.8%	92.8%	Epiphyseal dysplasia, multiple, 1, 132400 Pseudoachondroplasia, 177170
COPB2	139	99.8%	99.0%	?Microcephaly 19, primary, autosomal recessive, 617800
COQ2	103,5	97.6%	97.1%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ4	105	91.3%	90.2%	Coenzyme Q10 deficiency, primary, 7, 616276
COQ6	127,5	99.9%	98.6%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	138,3	99.9%	99.6%	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ8A	161,8	100.0%	99.9%	Coenzyme Q10 deficiency, primary, 4, 612016
COQ8B	99,5	100.0%	99.8%	Nephrotic syndrome, type 9, 615573
COQ9	73,8	100.0%	98.1%	Coenzyme Q10 deficiency, primary, 5, 614654
CORIN	140,2	100.0%	99.7%	Preeclampsia/eclampsia 5, 614595
CORO1A	150,8	99.9%	98.5%	Immunodeficiency 8, 615401
COX10	220,4	100.0%	99.9%	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 Mitochondrial complex IV deficiency, 220110
COX14	95,2	100.0%	100.0%	?Mitochondrial complex IV deficiency, 220110
COX15	87,7	99.9%	98.3%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000
COX20	66,2	96.4%	85.3%	Mitochondrial complex IV deficiency, 220110
COX4I2	116,5	100.0%	99.6%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX6A1	148,3	100.0%	99.9%	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
COX6B1	139,1	100.0%	100.0%	Mitochondrial complex IV deficiency, 220110
COX7B	38,6	62.3%	33.6%	Linear skin defects with multiple congenital anomalies 2, 300887

COX8A	109,4	100.0%	100.0%	?Mitochondrial complex IV deficiency, 220110
CP	100,6	93.1%	87.4%	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 [Hypoceruloplasminemia, hereditary], 604290
CPA6	109,1	99.4%	96.9%	Epilepsy, familial temporal lobe, 5, 614417 Febrile seizures, familial, 11, 614418
CPAMD8	101,3	98.3%	94.5%	Anterior segment dysgenesis 8, 617319
CPLX1	102,3	100.0%	100.0%	Epileptic encephalopathy, early infantile, 63, 617976
CPN1	97,9	99.8%	97.8%	Carboxypeptidase N deficiency, 212070
CPOX	134,1	99.5%	97.2%	Coproporphyria, 121300 Harderoporphyria, 121300
CPS1	133,8	100.0%	99.9%	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}, 0
CPT1A	131,5	99.9%	98.4%	CPT deficiency, hepatic, type IA, 255120
CPT1C	121,4	100.0%	100.0%	?Spastic paraplegia 73, autosomal dominant, 616282
CPT2	139,2	98.3%	98.2%	CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 CPT II deficiency, myopathic, stress-induced, 255110 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212
CR2	131,7	100.0%	99.9%	Immunodeficiency, common variable, 7, 614699 {Systemic lupus erythematosus, susceptibility to, 9}, 610927
CRADD	113,2	100.0%	98.2%	Mental retardation, autosomal recessive 34, with variant lissencephaly, 614499
CRAT	115,5	100.0%	99.9%	?Neurodegeneration with brain iron accumulation 8, 617917
CRB1	155	100.0%	100.0%	Leber congenital amaurosis 8, 613835 Pigmented paravenous chorioretinal atrophy, 172870 Retinitis pigmentosa-12, 600105
CRB2	116	99.8%	98.5%	Focal segmental glomerulosclerosis 9, 616220 Ventriculomegaly with cystic kidney disease, 219730
CRBN	125,2	88.1%	87.3%	Mental retardation, autosomal recessive 2, 607417
CREB1	120	99.6%	96.1%	Histiocytoma, angiomatoid fibrous, somatic, 612160
CREB3L1	135,8	100.0%	99.8%	Osteogenesis imperfecta, type XVI, 616229
CREBBP	110,7	99.4%	97.0%	Menke-Hennekam syndrome 1, 618332 Rubinstein-Taybi syndrome 1, 180849
CRELD1	98,9	99.8%	95.9%	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217 {Atrioventricular septal defect, susceptibility to, 2}, 606217
CRIPT	42,6	98.7%	91.2%	Short stature with microcephaly and distinctive facies, 615789

CRLF1	126,4	93.2%	90.7%	Cold-induced sweating syndrome 1, 272430
CRTAP	120,2	100.0%	99.1%	Osteogenesis imperfecta, type VII, 610682
CRTC1	166,1	99.8%	99.5%	Mucoepidermoid salivary gland carcinoma, 0
CRX	196,6	100.0%	100.0%	Cone-rod retinal dystrophy-2, 120970 Leber congenital amaurosis 7, 613829
CRYAA	131	95.8%	90.7%	Cataract 9, multiple types, 604219
CRYAB	94	99.7%	96.8%	Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, 2, 608810 Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869
CRYBA1	106	100.0%	99.7%	Cataract 10, multiple types, 600881
CRYBA2	168,4	100.0%	100.0%	?Cataract 42, 115900
CRYBA4	121,5	100.0%	100.0%	Cataract 23, 610425
CRYBB1	125,2	100.0%	99.8%	Cataract 17, multiple types, 611544
CRYBB2	137,6	100.0%	100.0%	Cataract 3, multiple types, 601547
CRYBB3	139,5	100.0%	100.0%	Cataract 22, 609741
CRYGB	96,8	99.8%	97.4%	Cataract 39, multiple types, autosomal dominant, 615188
CRYGC	127,3	100.0%	99.6%	Cataract 2, multiple types, 604307
CRYGD	103,2	100.0%	99.4%	Cataract 4, multiple types, 115700
CRYGS	79,8	93.9%	83.5%	Cataract 20, multiple types, 116100
CRYM	82,5	99.8%	97.2%	Deafness, autosomal dominant 40, 616357
CSF1R	113,3	99.9%	99.1%	Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
CSF2RA	53,7	90.0%	88.0%	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF2RB	123,2	99.8%	98.4%	Surfactant metabolism dysfunction, pulmonary, 5, 614370
CSF3R	105,4	99.7%	98.6%	Neutropenia, severe congenital, 7, autosomal recessive, 617014
CSNK1D	118,7	99.0%	95.4%	Advanced sleep-phase syndrome, familial, 2, 615224
CSNK2A1	104,5	93.7%	89.2%	Okur-Chung neurodevelopmental syndrome, 617062
CSPP1	119	100.0%	99.1%	Joubert syndrome 21, 615636
CSRP3	88,6	100.0%	98.3%	?Cardiomyopathy, dilated, 1M, 607482 Cardiomyopathy, hypertrophic, 12, 612124
CST3	79	99.7%	95.6%	Cerebral amyloid angiopathy, 105150 {Macular degeneration, age-related, 11}, 611953
CSTA	110,9	99.9%	99.1%	Peeling skin syndrome 4, 607936
CSTB	70	99.3%	90.9%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTBP1	101,2	94.3%	86.7%	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915
CTC1	105,5	100.0%	99.3%	Cerebroretinal microangiopathy with calcifications and cysts, 612199

CTCF	128,8	99.9%	98.9%	Mental retardation, autosomal dominant 21, 615502
CTDP1	128,1	95.1%	88.0%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTH	141	100.0%	99.8%	Cystathioninuria, 219500 Homocysteine, total plasma, elevated, 0
CTHRC1	100,7	99.5%	94.9%	Barrett esophagus/esophageal adenocarcinoma, 614266
CTLA4	141	100.0%	100.0%	Autoimmune lymphoproliferative syndrome, type V, 616100 {Celiac disease, susceptibility to, 3}, 609755 {Diabetes mellitus, insulin-dependent, 12}, 601388 {Hashimoto thyroiditis}, 140300 {Systemic lupus erythematosus, susceptibility to}, 152700
CTNNA1	109,6	99.1%	97.2%	Macular dystrophy, patterned, 2, 608970
CTNNA2	106	100.0%	99.6%	Cortical dysplasia, complex, with other brain malformations 9, 618174
CTNNA3	131,7	100.0%	99.8%	Arrhythmogenic right ventricular dysplasia, familial, 13, 615616
CTNNB1	127,4	100.0%	99.9%	Colorectal cancer, somatic, 114500 Exudative vitreoretinopathy 7, 617572 Hepatocellular carcinoma, somatic, 114550 Medulloblastoma, somatic, 155255 Neurodevelopmental disorder with spastic diplegia and visual defects, 615075 Ovarian cancer, somatic, 167000 Pilomatricoma, somatic, 132600
CTNND1	124,9	100.0%	99.9%	Blepharocheilodontic syndrome 2, 617681
CTNS	112,6	100.0%	99.5%	Cystinosis, atypical nephropathic, 219800 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750
CTPS1	108,8	100.0%	99.9%	Immunodeficiency 24, 615897
CTSA	132,9	100.0%	99.9%	Galactosialidosis, 256540
CTSC	116,2	100.0%	100.0%	Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000 Periodontitis 1, juvenile, 170650
CTSD	171	99.8%	97.8%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	107	91.3%	81.8%	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362
CTSK	86,3	100.0%	99.8%	Pycnodysostosis, 265800
CTU2	136,5	100.0%	99.8%	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142
CUBN	103,2	99.6%	97.6%	Megaloblastic anemia-1, Finnish type, 261100
CUL3	119	99.9%	98.9%	Pseudohypoaldosteronism, type IIE, 614496
CUL4B	78	97.6%	89.3%	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354

CUL7	129,2	100.0%	99.8%	3-M syndrome 1, 273750
CUX1	117	97.5%	95.1%	Global developmental delay with or without impaired intellectual development, 618330
CUX2	120,9	99.9%	99.3%	Epileptic encephalopathy, early infantile, 67, 618141
CWC27	84,5	99.8%	97.5%	Retinitis pigmentosa with or without skeletal anomalies, 250410
CWF19L1	103,2	99.9%	99.3%	Spinocerebellar ataxia, autosomal recessive 17, 616127
CXCR4	122,8	100.0%	100.0%	Myelokathexis, isolated, 0 WHIM syndrome, 193670
CXorf56	73,4	99.1%	92.7%	?Mental retardation, X-linked 107, 301013
CYB561	145	92.8%	92.7%	Orthostatic hypotension 2, 618182
CYB5A	132,5	100.0%	100.0%	Methemoglobinemia and ambiguous genitalia, 250790
CYB5R3	152,1	99.2%	98.3%	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYBA	110,5	96.7%	86.9%	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690
CYBB	99,1	99.9%	99.0%	Chronic granulomatous disease, X-linked, 306400 Immunodeficiency 34, mycobacteriosis, X-linked, 300645
CYC1	152,2	99.3%	95.6%	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYCS	61	99.1%	93.1%	Thrombocytopenia 4, 612004
CYFIP2	116,5	99.8%	98.5%	Epileptic encephalopathy, early infantile, 65, 618008
CYLD	109,2	99.7%	97.8%	Brooke-Spiegler syndrome, 605041 Cylindromatosis, familial, 132700 Trichoepithelioma, multiple familial, 1, 601606
CYP11A1	121,2	99.2%	95.0%	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	155,9	100.0%	100.0%	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 Aldosteronism, glucocorticoid-remediable, 103900
CYP11B2	156	100.0%	100.0%	Aldosterone to renin ratio raised, 0 Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 {Low renin hypertension, susceptibility to}, 0
CYP17A1	108,5	100.0%	99.6%	17,20-lyase deficiency, isolated, 202110 17-alpha-hydroxylase/17,20-lyase deficiency, 202110
CYP19A1	125,7	99.4%	97.3%	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300
CYP1B1	134,4	100.0%	100.0%	Anterior segment dysgenesis 6, multiple subtypes, 617315 Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300
CYP21A2	91,6	99.2%	93.4%	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910
CYP24A1	169,1	100.0%	100.0%	Hypercalcemia, infantile, 1, 143880

CYP26B1	168,7	100.0%	100.0%	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
CYP26C1	133	100.0%	99.9%	Focal facial dermal dysplasia 4, 614974
CYP27A1	173	100.0%	99.7%	Cerebrotendinous xanthomatosis, 213700
CYP27B1	147,1	100.0%	99.7%	Vitamin D-dependent rickets, type I, 264700
CYP2A6	140,1	100.0%	99.4%	Coumarin resistance, 122700 {Lung cancer, resistance to}, 211980 {Nicotine addiction, protection from}, 188890
CYP2B6	101,4	99.8%	97.2%	Efavirenz, poor metabolism of, 614546 {Efavirenz central nervous system toxicity, susceptibility to}, 614546
CYP2C19	148,1	99.7%	97.3%	Clopidogrel, impaired responsiveness to, 609535 Mephenytoin poor metabolizer, 609535 Omeprazole poor metabolizer, 609535 Proguanil poor metabolizer, 609535
CYP2C9	157	99.4%	96.5%	Tolbutamide poor metabolizer, 0 Warfarin sensitivity, 122700
CYP2R1	130,7	99.7%	96.8%	Rickets due to defect in vitamin D 25-hydroxylation, 600081
CYP2U1	134,3	98.4%	95.5%	Spastic paraplegia 56, autosomal recessive, 615030
CYP4F22	115,3	100.0%	98.8%	Ichthyosis, congenital, autosomal recessive 5, 604777
CYP4V2	137,2	99.9%	98.3%	Bietti crystalline corneoretinal dystrophy, 210370
CYP7B1	103,8	99.6%	96.6%	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, autosomal recessive, 270800
D2HGDH	142	100.0%	99.4%	D-2-hydroxyglutaric aciduria, 600721
DAB1	113,5	100.0%	100.0%	Spinocerebellar ataxia 37, 615945
DACT1	126,8	97.5%	93.8%	?Townes-Brocks syndrome 2, 617466
DAG1	189	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818
DARS	125,4	99.9%	99.0%	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2	126,8	100.0%	98.6%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBH	145,6	100.0%	99.9%	Orthostatic hypotension 1, due to DBH deficiency, 223360
DBT	109,9	99.6%	96.9%	Maple syrup urine disease, type II, 248600
DCAF17	90,4	99.9%	97.9%	Woodhouse-Sakati syndrome, 241080
DCAF8	110,7	100.0%	99.7%	?Giant axonal neuropathy 2, autosomal dominant, 610100
DCC	118,6	100.0%	99.8%	Colorectal cancer, somatic, 114500 Esophageal carcinoma, somatic, 133239 Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 Mirror movements 1 and/or agenesis of the corpus callosum, 157600

DCDC2	150	99.9%	99.8%	?Deafness, autosomal recessive 66, 610212 Nephronophthisis 19, 616217 Sclerosing cholangitis, neonatal, 617394
DCHS1	149,4	100.0%	99.9%	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390
DCLRE1C	138,9	99.9%	97.2%	Omenn syndrome, 603554 Severe combined immunodeficiency, Athabascan type, 602450
DCN	129,7	95.7%	95.2%	Corneal dystrophy, congenital stromal, 610048
DCPS	128	100.0%	99.6%	Al-Raqad syndrome, 616459
DCTN1	112,6	99.9%	99.2%	Neuropathy, distal hereditary motor, type VIIB, 607641 Perry syndrome, 168605 {Amyotrophic lateral sclerosis, susceptibility to}, 105400
DCX	90,5	99.9%	98.4%	Lissencephaly, X-linked, 300067 Subcortical laminal heterotopia, X-linked, 300067
DDB2	147,3	99.8%	98.4%	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DDC	97,9	99.5%	95.0%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	161,6	99.9%	98.4%	Spastic paraplegia 28, autosomal recessive, 609340
DDHD2	129,6	100.0%	99.5%	Spastic paraplegia 54, autosomal recessive, 615033
DDOST	114	100.0%	99.8%	?Congenital disorder of glycosylation, type Ir, 614507
DDR2	115,8	100.0%	99.3%	Spondylometaepiphyseal dysplasia, short limb-hand type, 271665 Warburg-Cinotti syndrome, 618175
DDRGK1	100	100.0%	99.8%	Spondyloepimetaphyseal dysplasia, Shohat type, 602557
DDX11	100,7	86.7%	81.2%	Warsaw breakage syndrome, 613398
DDX3X	73,5	86.2%	82.9%	Mental retardation, X-linked 102, 300958
DDX58	112,3	99.8%	99.1%	Singleton-Merten syndrome 2, 616298
DDX59	141,5	100.0%	99.8%	Orofaciodigital syndrome V, 174300
DEAF1	113,4	99.8%	97.6%	?Dyskinesia, seizures, and intellectual developmental disorder, 617171 Mental retardation, autosomal dominant 24, 615828
DEGS1	144,9	100.0%	100.0%	Leukodystrophy, hypomyelinating, 18, 618404
DENND5A	99,2	99.8%	98.9%	Epileptic encephalopathy, early infantile, 49, 617281
DEPDC5	124,5	99.9%	99.7%	Epilepsy, familial focal, with variable foci 1, 604364
DES	125	100.0%	100.0%	Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400
DFNA5	NC	NC	NC	Deafness, autosomal dominant 5, 600994
DFNB59	NC	NC	NC	Deafness, autosomal recessive 59, 610220

DGAT1	150,3	96.7%	92.0%	?Diarrhea 7, protein-losing enteropathy type, 615863
DGKE	127,8	99.8%	98.3%	Nephrotic syndrome, type 7, 615008 {Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008
DGUOK	119,4	99.9%	97.9%	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880 Portal hypertension, noncirrhotic, 617068 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070
DHCR24	155,8	100.0%	99.9%	Desmosterolosis, 602398
DHCR7	144,9	100.0%	100.0%	Smith-Lemli-Opitz syndrome, 270400
DHDDS	81	97.1%	93.8%	?Congenital disorder of glycosylation, type 1bb, 613861 Developmental delay and seizures with or without movement abnormalities, 617836 Retinitis pigmentosa 59, 613861
DHFR	50	94.1%	83.1%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHH	164,9	100.0%	100.0%	46XY partial gonadal dysgenesis, with minifascicular neuropathy, 607080 46XY sex reversal 7, 233420
DHODH	98,8	100.0%	100.0%	Miller syndrome, 263750
DHTKD1	122,4	99.9%	98.8%	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DHX30	161,5	100.0%	100.0%	Neurodevelopmental disorder with severe motor impairment and absent language, 617804
DHX38	104,9	100.0%	99.5%	Retinitis pigmentosa 84, 618220
DIABLO	174	100.0%	99.6%	Deafness, autosomal dominant 64, 614152
DIAPH1	101,7	99.9%	99.6%	Deafness, autosomal dominant 1, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632
DIAPH2	67	96.2%	88.1%	?Premature ovarian failure 2A, 300511
DIAPH3	82,7	99.7%	96.9%	Auditory neuropathy, autosomal dominant, 1, 609129
DICER1	137,9	99.8%	98.4%	GLOW syndrome, somatic mosaic, 618272 Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800 Pleuropulmonary blastoma, 601200 Rhabdomyosarcoma, embryonal, 2, 180295
DIP2B	128	100.0%	99.9%	Mental retardation, FRA12A type, 136630
DIS3L2	143,3	100.0%	99.8%	Perlman syndrome, 267000
DKC1	91,2	99.8%	97.7%	Dyskeratosis congenita, X-linked, 305000
DLAT	100,2	99.8%	99.2%	Pyruvate dehydrogenase E2 deficiency, 245348
DLC1	160	99.9%	99.7%	Colorectal cancer, somatic, 114500
DLD	119,2	99.9%	99.7%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLG3	79,3	99.2%	92.1%	Mental retardation, X-linked 90, 300850
DLL3	108,5	96.7%	92.5%	Spondylocostal dysostosis 1, autosomal recessive, 277300

DLL4	175,2	100.0%	99.7%	Adams-Oliver syndrome 6, 616589
DLX3	146,7	100.0%	99.0%	Amelogenesis imperfecta, type IV, 104510 Trichodontoosseous syndrome, 190320
DLX4	228,4	100.0%	100.0%	?Orofacial cleft 15, 616788
DLX5	145,3	100.0%	99.8%	?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600
DMD	108,2	99.4%	98.0%	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200
DMGDH	134,7	100.0%	99.8%	Dimethylglycine dehydrogenase deficiency, 605850
DMP1	133	100.0%	99.9%	Hypophosphatemic rickets, AR, 241520
DMPK	151,3	100.0%	99.9%	Myotonic dystrophy 1, 160900
DMXL2	154	99.8%	98.9%	?Deafness, autosomal dominant 71, 617605 ?Polyendocrine-polyneuropathy syndrome, 616113
DNA2	124,3	99.7%	97.3%	?Seckel syndrome 8, 615807 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156
DNAAF1	112,8	100.0%	99.5%	Ciliary dyskinesia, primary, 13, 613193
DNAAF2	150,1	99.9%	98.7%	Ciliary dyskinesia, primary, 10, 612518
DNAAF3	115	99.9%	98.5%	Ciliary dyskinesia, primary, 2, 606763
DNAAF4	94,1	99.9%	98.2%	Ciliary dyskinesia, primary, 25, 615482 {Dyslexia, susceptibility to, 1}, 127700
DNAAF5	107,6	95.4%	85.2%	Ciliary dyskinesia, primary, 18, 614874
DNAH1	158,1	100.0%	99.8%	?Ciliary dyskinesia, primary, 37, 617577 Spermatogenic failure 18, 617576
DNAH11	131,3	99.8%	98.7%	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH5	114,1	99.9%	99.1%	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAH9	110,7	99.9%	98.5%	Ciliary dyskinesia, primary, 40, 618300
DNAI1	115,4	100.0%	100.0%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	151,7	99.6%	96.6%	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJB11	107,4	99.8%	99.5%	Polycystic kidney disease 6 with or without polycystic liver disease, 618061
DNAJB13	116,3	100.0%	99.5%	Ciliary dyskinesia, primary, 34, 617091
DNAJB2	118,4	100.0%	100.0%	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881
DNAJB6	60,7	96.8%	84.4%	Muscular dystrophy, limb-girdle, autosomal dominant 1, 603511
DNAJC12	140,7	87.4%	87.4%	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC19	93,8	98.4%	92.3%	3-methylglutaconic aciduria, type V, 610198
DNAJC21	128,1	99.9%	99.5%	Bone marrow failure syndrome 3, 617052
DNAJC3	137,4	100.0%	99.7%	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192

DNAJC5	188,2	100.0%	100.0%	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350
DNAJC6	126,5	99.9%	99.0%	Parkinson disease 19a, juvenile-onset, 615528 Parkinson disease 19b, early-onset, 615528
DNAL1	104	99.3%	96.2%	Ciliary dyskinesia, primary, 16, 614017
DNAL4	69,9	99.9%	95.3%	?Mirror movements 3, 616059
DNASE1L3	113	100.0%	99.7%	Systemic lupus erythematosus 16, 614420
DNM1	139,8	94.7%	92.3%	Epileptic encephalopathy, early infantile, 31, 616346
DNM1L	119,5	99.9%	98.7%	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 Optic atrophy 5, 610708
DNM2	123,9	99.7%	96.7%	Centronuclear myopathy 1, 160150 Charcot-Marie-Tooth disease, axonal type 2M, 606482 Charcot-Marie-Tooth disease, dominant intermediate B, 606482 Lethal congenital contracture syndrome 5, 615368
DNMBP	129	99.8%	99.0%	Cataract 48, 618415
DNMT1	114,3	99.2%	98.7%	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 Neuropathy, hereditary sensory, type IE, 614116
DNMT3A	122,9	99.7%	98.2%	Acute myeloid leukemia, somatic, 601626 Tatton-Brown-Rahman syndrome, 615879
DNMT3B	116,4	100.0%	99.9%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK2	119,5	100.0%	99.5%	Immunodeficiency 40, 616433
DOCK3	113,1	100.0%	99.2%	Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292
DOCK6	121,5	99.6%	98.6%	Adams-Oliver syndrome 2, 614219
DOCK7	120,7	99.6%	97.8%	Epileptic encephalopathy, early infantile, 23, 615859
DOCK8	112,1	100.0%	99.6%	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
DOK7	135,1	94.0%	93.3%	?Fetal akinesia deformation sequence 3, 618389 Myasthenic syndrome, congenital, 10, 254300
DOLK	157,2	100.0%	100.0%	Congenital disorder of glycosylation, type Im, 610768
DONSON	90,2	99.0%	92.4%	Microcephaly, short stature, and limb abnormalities, 617604 Microcephaly-micromelia syndrome, 251230
DPAGT1	87,5	100.0%	99.9%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPF2	99,8	99.6%	96.7%	Coffin-Siris syndrome 7, 618027
DPH1	161,6	100.0%	100.0%	Developmental delay with short stature, dysmorphic features, and sparse hair, 616901
DPM1	134,7	95.2%	88.2%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	88,5	99.8%	97.6%	Congenital disorder of glycosylation, type Iu, 615042
DPM3	200,5	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937

DPP6	122,4	99.9%	98.6%	Mental retardation, autosomal dominant 33, 616311 {Ventricular fibrillation, paroxysmal familial, 2}, 612956
DPY19L2	85,9	74.4%	70.2%	Spermatogenic failure 9, 613958
DPYD	141,6	99.5%	96.4%	5-fluorouracil toxicity, 274270 Dihydropyrimidine dehydrogenase deficiency, 274270
DPYS	117,5	100.0%	99.8%	Dihydropyrimidinuria, 222748
DRAM2	130,4	100.0%	100.0%	Cone-rod dystrophy 21, 616502
DRC1	91,2	100.0%	99.3%	Ciliary dyskinesia, primary, 21, 615294
DRD4	107,9	96.4%	85.9%	Autonomic nervous system dysfunction, 0 [Novelty seeking personality], 601696 {Attention deficit-hyperactivity disorder}, 143465
DSC2	123,7	99.6%	97.2%	Arrhythmogenic right ventricular dysplasia 11, 610476 Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476
DSC3	95,6	99.3%	97.3%	?Hypotrichosis and recurrent skin vesicles, 613102
DSE	89,7	99.7%	97.2%	Ehlers-Danlos syndrome, musculocontractural type 2, 615539
DSG1	131	99.4%	97.7%	Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508 Keratosis palmoplantaris striata I, AD, 148700
DSG2	129,5	99.9%	99.2%	Arrhythmogenic right ventricular dysplasia 10, 610193 Cardiomyopathy, dilated, 1BB, 612877
DSG4	158,3	99.8%	99.0%	Hypotrichosis 6, 607903
DSP	140,6	100.0%	99.6%	Arrhythmogenic right ventricular dysplasia 8, 607450 Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821 Epidermolysis bullosa, lethal acantholytic, 609638 Keratosis palmoplantaris striata II, 612908 Skin fragility-woolly hair syndrome, 607655
DSPP	79	98.4%	93.8%	Deafness, autosomal dominant 39, with dentinogenesis, 605594 Dentin dysplasia, type II, 125420 Dentinogenesis imperfecta, Shields type II, 125490 Dentinogenesis imperfecta, Shields type III, 125500
DST	144,8	99.9%	99.2%	?Neuropathy, hereditary sensory and autonomic, type VI, 614653 Epidermolysis bullosa simplex, autosomal recessive 2, 615425
DSTYK	121,7	99.6%	97.7%	Congenital anomalies of kidney and urinary tract 1, 610805 Spastic paraplegia 23, 270750
DTNA	125,2	100.0%	99.9%	Left ventricular noncompaction 1, with or without congenital heart defects, 604169
DTNBP1	113,4	99.8%	97.9%	Hermansky-Pudlak syndrome 7, 614076
DUOX2	128,8	99.5%	96.4%	Thyroid dyshormonogenesis 6, 607200

DUOXA2	140,5	100.0%	100.0%	Thyroid dyshormonogenesis 5, 274900
DUSP6	164,1	100.0%	100.0%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
DVL1	140,8	98.6%	95.9%	Robinow syndrome, autosomal dominant 2, 616331
DVL3	186	100.0%	100.0%	Robinow syndrome, autosomal dominant 3, 616894
DYM	103,3	97.4%	95.5%	Dyggve-Melchior-Clausen disease, 223800 Smith-McCort dysplasia, 607326
DYNC1H1	140,6	100.0%	99.6%	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600
DYNC2H1	102,2	98.8%	95.5%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYNC2LI1	95,4	99.7%	97.0%	Short-rib thoracic dysplasia 15 with polydactyly, 617088
DYRK1A	130,4	100.0%	99.9%	Mental retardation, autosomal dominant 7, 614104
DYRK1B	110,5	98.8%	94.8%	Abdominal obesity-metabolic syndrome 3, 615812
DYSF	133,6	100.0%	99.9%	Miyoshi muscular dystrophy 1, 254130 Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601 Myopathy, distal, with anterior tibial onset, 606768
DZIP1L	96,7	99.7%	98.0%	Polycystic kidney disease 5, 617610
EARS2	99	99.7%	97.8%	Combined oxidative phosphorylation deficiency 12, 614924
EBF3	140,9	100.0%	99.8%	Hypotonia, ataxia, and delayed development syndrome, 617330
EBP	63,2	99.5%	95.2%	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
ECE1	145,6	98.0%	97.7%	?Hirschsprung disease, cardiac defects, and autonomic dysfunction, 613870 {Hypertension, essential, susceptibility to}, 145500
ECEL1	107,1	100.0%	97.4%	Arthrogryposis, distal, type 5D, 615065
ECHS1	103,8	100.0%	99.7%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ECM1	158,6	99.9%	99.0%	Urbach-Wiethe disease, 247100
EDA	102	95.6%	85.7%	Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100 Tooth agenesis, selective, X-linked 1, 313500
EDAR	126,6	100.0%	100.0%	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	89,8	99.7%	98.3%	Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940 Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941
EDC3	113,7	100.0%	99.0%	?Mental retardation, autosomal recessive 50, 616460
EDN1	156,5	100.0%	99.9%	Auriculocondylar syndrome 3, 615706 Question mark ears, isolated, 612798

				{High density lipoprotein cholesterol level QTL 7}, 0
EDN3	135,2	100.0%	100.0%	Central hypoventilation syndrome, congenital, 209880 Waardenburg syndrome, type 4B, 613265 {Hirschsprung disease, susceptibility to, 4}, 613712
EDNRA	150,7	100.0%	99.8%	Mandibulofacial dysostosis with alopecia, 616367 {Migraine, resistance to}, 157300
EDNRB	120,9	96.9%	92.5%	ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580 {Hirschsprung disease, susceptibility to, 2}, 600155
EED	85,3	99.5%	96.3%	Cohen-Gibson syndrome, 617561
EEF1A2	188,3	100.0%	100.0%	Epileptic encephalopathy, early infantile, 33, 616409 Mental retardation, autosomal dominant 38, 616393
EEF2	154,9	100.0%	100.0%	?Spinocerebellar ataxia 26, 609306
EFEMP1	147,8	99.9%	99.4%	Doyne honeycomb degeneration of retina, 126600
EFEMP2	129,4	100.0%	100.0%	Cutis laxa, autosomal recessive, type IB, 614437
EFL1	150,5	99.5%	98.1%	Shwachman-Diamond syndrome 2, 617941
EFNB1	116,7	100.0%	99.9%	Craniofrontonasal dysplasia, 304110
EFTUD2	103,2	100.0%	99.2%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EGF	110,5	100.0%	99.7%	Hypomagnesemia 4, renal, 611718
EGFR	135,2	100.0%	100.0%	?Inflammatory skin and bowel disease, neonatal, 2, 616069 Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980 Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980 {Nonsmall cell lung cancer, susceptibility to}, 211980
EGLN1	102	97.6%	85.9%	Erythrocytosis, familial, 3, 609820 [Hemoglobin, high altitude adaptation], 609070
EGR2	130	100.0%	100.0%	Charcot-Marie-Tooth disease, type 1D, 607678 Dejerine-Sottas disease, 145900 Hypomyelinating neuropathy, congenital, 1, 605253
EHHADH	133,4	100.0%	99.7%	?Fanconi renotubular syndrome 3, 615605
EHMT1	127,7	94.6%	94.2%	Kleefstra syndrome 1, 610253
EIF2AK3	134,2	99.5%	96.3%	Wolcott-Rallison syndrome, 226980
EIF2AK4	129,8	99.7%	98.6%	Pulmonary venoocclusive disease 2, 234810
EIF2B1	121,7	100.0%	99.9%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B2	109,7	99.4%	92.4%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B3	134,7	100.0%	100.0%	Leukoencephalopathy with vanishing white matter, 603896

EIF2B4	122	100.0%	99.6%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B5	103,1	100.0%	99.6%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2S3	81,4	96.7%	88.4%	MEHMO syndrome, 300148
EIF3F	63,1	98.7%	88.5%	Mental retardation, autosomal recessive 67, 618295
EIF4A3	87,5	100.0%	99.4%	Robin sequence with cleft mandible and limb anomalies, 268305
ELAC2	109,4	99.9%	99.0%	Combined oxidative phosphorylation deficiency 17, 615440 {Prostate cancer, hereditary, 2, susceptibility to}, 614731
ELANE	141,5	100.0%	99.3%	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
ELMO2	102,3	100.0%	98.8%	Vascular malformation, primary intraosseous, 606893
ELMOD3	135,8	100.0%	99.9%	?Deafness, autosomal recessive 88, 615429
ELN	103,1	100.0%	98.9%	Cutis laxa, autosomal dominant, 123700 Supravalvar aortic stenosis, 185500
ELOVL4	104,4	99.9%	99.1%	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110
ELOVL5	105,7	100.0%	99.1%	Spinocerebellar ataxia 38, 615957
ELP1	118,7	99.7%	98.4%	Dysautonomia, familial, 223900
ELP2	120,6	99.8%	98.0%	Mental retardation, autosomal recessive 58, 617270
ELP4	57,4	73.1%	69.7%	?Aniridia 2, 617141
EMC1	105,7	100.0%	98.9%	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EMD	138,2	100.0%	98.9%	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
EMG1	123,2	100.0%	99.9%	Bowen-Conradi syndrome, 211180
EML1	129,9	100.0%	99.6%	Band heterotopia, 600348
EMP2	78,4	99.5%	96.1%	Nephrotic syndrome, type 10, 615861
EMX2	155,2	100.0%	100.0%	Schizencephaly, 269160
ENAM	139,5	100.0%	100.0%	Amelogenesis imperfecta, type IB, 104500 Amelogenesis imperfecta, type IC, 204650
ENG	128,4	99.9%	98.7%	Telangiectasia, hereditary hemorrhagic, type 1, 187300
ENO3	173,2	100.0%	100.0%	?Glycogen storage disease XIII, 612932
ENPP1	129,2	97.5%	93.3%	Arterial calcification, generalized, of infancy, 1, 208000 Cole disease, 615522 Hypophosphatemic rickets, autosomal recessive, 2, 613312 {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853

				{Obesity, susceptibility to}, 601665
ENTPD1	125	100.0%	99.9%	Spastic paraplegia 64, autosomal recessive, 615683
EOGT	102,7	79.3%	78.1%	Adams-Oliver syndrome 4, 615297
EP300	165,5	99.7%	98.6%	Colorectal cancer, somatic, 114500 Menke-Hennekam syndrome 2, 618333 Rubinstein-Taybi syndrome 2, 613684
EPAS1	132	99.9%	98.2%	Erythrocytosis, familial, 4, 611783
EPB41	119,2	99.6%	97.4%	Elliptocytosis-1, 611804
EPB41L1	121,5	99.4%	95.3%	?Mental retardation, autosomal dominant 11, 614257
EPB42	128	100.0%	99.3%	Spherocytosis, type 5, 612690
EPCAM	76,5	99.7%	95.7%	Colorectal cancer, hereditary nonpolyposis, type 8, 613244 Diarrhea 5, with tufting enteropathy, congenital, 613217
EPG5	110,3	99.3%	97.9%	Vici syndrome, 242840
EPHA2	157	100.0%	99.8%	Cataract 6, multiple types, 116600
EPHB4	156,5	100.0%	99.8%	Capillary malformation-arteriovenous malformation 2, 618196 Lymphatic malformation 7, 617300
EPHX1	116	99.2%	96.0%	?Hypercholanemia, familial, 607748
EPM2A	116,5	90.9%	88.8%	Epilepsy, progressive myoclonic 2A (Lafora), 254780
EPO	98,6	100.0%	99.4%	?Diamond-Blackfan anemia-like, 617911 Erythrocytosis, familial, 5, 617907 {Microvascular complications of diabetes 2}, 612623
EPRS	139,4	99.9%	99.2%	Leukodystrophy, hypomyelinating, 15, 617951
EPS8	114,6	99.8%	96.6%	?Deafness, autosomal recessive 102, 615974
EPS8L2	159,7	99.0%	96.3%	Deafness autosomal recessive 106, 617637
ERAL1	156,5	100.0%	100.0%	Perrault syndrome 6, 617565
ERBB2	138,8	99.6%	98.0%	Adenocarcinoma of lung, somatic, 211980 Gastric cancer, somatic, 613659 Glioblastoma, somatic, 137800 Ovarian cancer, somatic, 0
ERBB3	113,3	99.9%	99.2%	?Lethal congenital contractural syndrome 2, 607598 {?Erythroleukemia, familial, susceptibility to}, 133180
ERBB4	127,8	99.9%	99.2%	Amyotrophic lateral sclerosis 19, 615515
ERCC1	85,7	100.0%	98.1%	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	128	100.0%	99.8%	?Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy 1, photosensitive, 601675 Xeroderma pigmentosum, group D, 278730

ERCC3	92	99.9%	98.4%	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651
ERCC4	132	100.0%	99.8%	Fanconi anemia, complementation group Q, 615272 Xeroderma pigmentosum, group F, 278760 Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 XFE progeroid syndrome, 610965
ERCC5	126,3	99.9%	99.5%	Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	158,2	100.0%	99.9%	Cerebrooculofacioskeletal syndrome 1, 214150 Cockayne syndrome, type B, 133540 De Sanctis-Cacchione syndrome, 278800 Premature ovarian failure 11, 616946 UV-sensitive syndrome 1, 600630 {Lung cancer, susceptibility to}, 211980 {Macular degeneration, age-related, susceptibility to, 5}, 613761
ERCC6L2	121,7	99.9%	99.0%	Bone marrow failure syndrome 2, 615715
ERCC8	82,8	98.9%	90.0%	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621
ERF	146,8	100.0%	99.2%	Chitayat syndrome, 617180 Craniosynostosis 4, 600775
ERGIC1	178	95.3%	94.5%	?Arthrogryposis multiplex congenita, neurogenic type, 208100
ERLIN1	141,4	100.0%	100.0%	Spastic paraplegia 62, 615681
ERLIN2	115,3	100.0%	99.2%	Spastic paraplegia 18, autosomal recessive, 611225
ERMARD	118,9	99.8%	98.5%	?Periventricular nodular heterotopia 6, 615544
ESCO2	115,6	99.4%	97.3%	Roberts syndrome, 268300 SC phocomelia syndrome, 269000
ESPN	30,8	53.5%	42.4%	Deafness, autosomal recessive 36, 609006 Deafness, neurosensory, without vestibular involvement, autosomal dominant, 0
ESR1	137	100.0%	100.0%	Breast cancer, somatic, 114480 Estrogen resistance, 615363 {Atherosclerosis, susceptibility to}, 0 {HDL response to hormone replacement, augmented}, 0 {Migraine, susceptibility to}, 157300 {Myocardial infarction, susceptibility to}, 608446
ESR2	111,6	100.0%	99.6%	?Ovarian dysgenesis 8, 618187
ESRP1	95,3	99.8%	98.4%	?Deafness, autosomal recessive 109, 618013

ESRRB	121,7	99.9%	99.1%	Deafness, autosomal recessive 35, 608565
ETFA	132,7	100.0%	99.8%	Glutaric acidemia IIA, 231680
ETFB	116,8	100.0%	100.0%	Glutaric acidemia IIB, 231680
ETFDH	114,4	100.0%	99.3%	Glutaric acidemia IIC, 231680
ETHE1	97,3	99.9%	97.8%	Ethylmalonic encephalopathy, 602473
ETV6	148,3	100.0%	99.4%	Leukemia, acute myeloid, somatic, 601626 Thrombocytopenia 5, 616216
EVC	106,3	95.9%	92.4%	?Weyers acrofacial dysostosis, 193530 Ellis-van Creveld syndrome, 225500
EVC2	110,2	99.4%	96.3%	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530
EWSR1	67,2	91.8%	82.8%	Ewing sarcoma, 612219 Neuroepithelioma, 612219
EXOC6B	107,7	98.7%	97.4%	Spondyloepimetaphyseal dysplasia with joint laxity, type 3, 618395
EXOSC2	110,3	100.0%	99.9%	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EXOSC3	125,1	96.4%	87.8%	Pontocerebellar hypoplasia, type 1B, 614678
EXOSC8	89	98.2%	90.3%	Pontocerebellar hypoplasia, type 1C, 616081
EXOSC9	132,5	99.6%	95.1%	Pontocerebellar hypoplasia, type 1D, 618065
EXPH5	163,6	100.0%	99.9%	Epidermolysis bullosa, nonspecific, autosomal recessive, 615028
EXT1	88,6	99.6%	98.0%	Chondrosarcoma, 215300 Exostoses, multiple, type 1, 133700
EXT2	118	99.9%	99.1%	?Seizures, scoliosis, and macrocephaly syndrome, 616682 Exostoses, multiple, type 2, 133701
EXTL3	184,1	100.0%	100.0%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
EYA1	120,2	99.9%	99.8%	?Otofaciocervical syndrome, 166780 Anterior segment anomalies with or without cataract, 602588 Branchiootic syndrome 1, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650
EYA4	136,9	100.0%	99.9%	?Cardiomyopathy, dilated, 1J, 605362 Deafness, autosomal dominant 10, 601316
EYS	132,5	99.7%	97.5%	Retinitis pigmentosa 25, 602772
EZH2	130	99.4%	97.6%	Weaver syndrome, 277590
F10	173,6	99.8%	99.1%	Factor X deficiency, 227600
F11	126,7	100.0%	99.9%	Factor XI deficiency, autosomal dominant, 612416 Factor XI deficiency, autosomal recessive, 612416
F12	151,3	99.9%	99.5%	Angioedema, hereditary, type III, 610618

				Factor XII deficiency, 234000
F13A1	112,9	100.0%	99.6%	Factor XIIIA deficiency, 613225 {Myocardial infarction, protection against}, 608446 {Venous thrombosis, protection against}, 188050
F13B	105,8	98.6%	92.9%	Factor XIIIB deficiency, 613235
F2	128,1	99.8%	97.1%	Dysprothrombinemia, 613679 Hypoprothrombinemia, 613679 Thrombophilia due to thrombin defect, 188050 {Pregnancy loss, recurrent, susceptibility to, 2}, 614390 {Stroke, ischemic, susceptibility to}, 601367
F5	145,5	99.5%	97.7%	Factor V deficiency, 227400 Thrombophilia due to activated protein C resistance, 188055 {Budd-Chiari syndrome}, 600880 {Pregnancy loss, recurrent, susceptibility to, 1}, 614389 {Stroke, ischemic, susceptibility to}, 601367 {Thrombophilia, susceptibility to, due to factor V Leiden}, 188055
F7	162	100.0%	100.0%	Factor VII deficiency, 227500 {Myocardial infarction, decreased susceptibility to}, 608446
F8	106	99.4%	97.2%	Hemophilia A, 306700
F9	109,5	99.9%	98.4%	Hemophilia B, 306900 Thrombophilia, X-linked, due to factor IX defect, 300807 {Deep venous thrombosis, protection against}, 300807 {Warfarin sensitivity}, 122700
FA2H	92,7	98.8%	92.5%	Spastic paraplegia 35, autosomal recessive, 612319
FADD	181,7	100.0%	100.0%	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations, 613759
FAH	128,4	100.0%	99.8%	Tyrosinemia, type I, 276700
FAM111A	232,1	100.0%	99.5%	Gracile bone dysplasia, 602361 Kenny-Caffey syndrome, type 2, 127000
FAM111B	157,9	99.9%	99.6%	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704
FAM126A	125,4	100.0%	99.4%	Leukodystrophy, hypomyelinating, 5, 610532
FAM161A	136,9	99.9%	99.1%	Retinitis pigmentosa 28, 606068
FAM20A	111,1	100.0%	99.4%	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM20C	145,2	100.0%	100.0%	Raine syndrome, 259775
FAM46A	NC	NC	NC	Osteogenesis imperfecta, type XVIII, 617952
FAM58A	NC	NC	NC	STAR syndrome, 300707
FAM83H	120,2	100.0%	99.9%	Amelogenesis imperfecta, type IIIA, 130900
FAM92A	79,4	87.6%	77.4%	?Polydactyly, postaxial, type A9, 618219

FAN1	132,2	100.0%	99.9%	Interstitial nephritis, karyomegalic, 614817
FANCA	112,4	99.9%	98.9%	Fanconi anemia, complementation group A, 227650
FANCB	76,4	98.6%	93.2%	Fanconi anemia, complementation group B, 300514
FANCC	100,8	99.7%	99.2%	Fanconi anemia, complementation group C, 227645
FANCD2	115,6	99.1%	96.6%	Fanconi anemia, complementation group D2, 227646
FANCE	118,2	96.6%	89.9%	Fanconi anemia, complementation group E, 600901
FANCF	244,4	100.0%	100.0%	Fanconi anemia, complementation group F, 603467
FANCG	140,7	100.0%	99.8%	Fanconi anemia, complementation group G, 614082
FANCI	136,2	99.9%	98.9%	Fanconi anemia, complementation group I, 609053
FANCL	105,8	99.7%	98.0%	Fanconi anemia, complementation group L, 614083
FANCM	100,6	99.3%	97.1%	?Premature ovarian failure 15, 618096 Spermatogenic failure 28, 618086
FAR1	73,7	97.2%	91.8%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154
FARS2	161,9	100.0%	100.0%	Combined oxidative phosphorylation deficiency 14, 614946 Spastic paraplegia 77, autosomal recessive, 617046
FARSB	78	97.4%	92.7%	Rajab interstitial lung disease with brain calcifications, 613658
FAS	226	99.9%	99.6%	Autoimmune lymphoproliferative syndrome, type IA, 601859 Squamous cell carcinoma, burn scar-related, somatic, 0 {Autoimmune lymphoproliferative syndrome}, 601859
FASLG	82	100.0%	99.0%	Autoimmune lymphoproliferative syndrome, type IB, 601859 {Lung cancer, susceptibility to}, 211980
FASTKD2	115,5	99.6%	97.9%	?Mitochondrial complex IV deficiency, 220110
FAT2	131,9	100.0%	99.8%	Spinocerebellar ataxia 45, 617769
FAT4	190,3	100.0%	99.9%	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 Van Maldergem syndrome 2, 615546
FBLN5	96,6	91.8%	91.5%	Cutis laxa, autosomal dominant 2, 614434 Cutis laxa, autosomal recessive, type IA, 219100 Macular degeneration, age-related, 3, 608895 Neuropathy, hereditary, with or without age-related macular degeneration, 608895
FBN1	137,1	100.0%	99.8%	Acromicric dysplasia, 102370 Ectopia lentis, familial, 129600 Geleophysic dysplasia 2, 614185 Marfan lipodystrophy syndrome, 616914 Marfan syndrome, 154700 MASS syndrome, 604308 Stiff skin syndrome, 184900 Weill-Marchesani syndrome 2, dominant, 608328

FBN2	142,2	100.0%	99.8%	Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118
FBP1	105,5	100.0%	99.3%	Fructose-1,6-bisphosphatase deficiency, 229700
FBXL3	188,9	100.0%	100.0%	Intellectual developmental disorder with short stature, facial anomalies, and speech defects, 606220
FBXL4	168,9	100.0%	100.0%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FBXO11	86	98.2%	93.7%	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities, 618089
FBXO31	111,5	99.6%	97.3%	?Mental retardation, autosomal recessive 45, 615979
FBXO38	159,8	99.7%	98.3%	Neuronopathy, distal hereditary motor, type IID, 615575
FBXO7	152,8	99.9%	99.6%	Parkinson disease 15, autosomal recessive, 260300
FCGR3A	163,2	99.6%	96.7%	Immunodeficiency 20, 615707
FCGR3B	137,4	98.8%	97.1%	Neutropenia, alloimmune neonatal, 0
FCN3	124,7	100.0%	100.0%	Immunodeficiency due to ficolin 3 deficiency, 613860
FDFT1	136,3	100.0%	99.6%	Squalene synthase deficiency, 618156
FDPS	58,4	97.9%	91.6%	Porokeratosis 9, multiple types, 616631
FDX2	147,2	100.0%	100.0%	Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900
FDXR	122,3	99.9%	99.1%	Auditory neuropathy and optic atrophy, 617717
FECH	104	100.0%	99.7%	Protoporphyria, erythropoietic, 1, 177000
FERMT1	90,8	99.6%	96.6%	Kindler syndrome, 173650
FERMT3	144,9	100.0%	99.9%	Leukocyte adhesion deficiency, type III, 612840
FEZF1	179,7	100.0%	100.0%	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FGA	137	99.3%	97.3%	Afibrinogenemia, congenital, 202400 Amyloidosis, familial visceral, 105200 Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, congenital, 616004
FGB	136,7	99.7%	98.2%	Afibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Hypofibrinogenemia, congenital, 202400
FGD1	86,7	98.4%	93.0%	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400
FGD4	104,3	99.7%	97.8%	Charcot-Marie-Tooth disease, type 4H, 609311
FGF10	120,5	100.0%	99.6%	Aplasia of lacrimal and salivary glands, 180920 LADD syndrome, 149730
FGF12	100,4	100.0%	99.9%	Epileptic encephalopathy, early infantile, 47, 617166
FGF14	214,2	100.0%	100.0%	Spinocerebellar ataxia 27, 609307
FGF16	96,3	99.3%	94.0%	Metacarpal 4-5 fusion, 309630
FGF17	167,3	100.0%	100.0%	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270

FGF20	125,3	100.0%	98.3%	?Renal hypodysplasia/aplasia 2, 615721
FGF23	122,3	99.7%	97.7%	Hypophosphatemic rickets, autosomal dominant, 193100 Osteomalacia, tumor-induced, 0 Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993
FGF3	139,5	100.0%	100.0%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGF5	155,5	99.9%	99.5%	Trichomegaly, 190330
FGF8	130	97.9%	86.8%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGF9	153,5	100.0%	100.0%	Multiple synostoses syndrome 3, 612961
FGFR1	122,6	100.0%	99.6%	Encephalocraniocutaneous lipomatosis, 613001 Hartsfield syndrome, 615465 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Jackson-Weiss syndrome, 123150 Osteoglophonic dysplasia, 166250 Pfeiffer syndrome, 101600 Trigonocephaly 1, 190440
FGFR2	113,1	97.7%	96.8%	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Apert syndrome, 101200 Beare-Stevenson cutis gyrata syndrome, 123790 Bent bone dysplasia syndrome, 614592 Craniofacial-skeletal-dermatologic dysplasia, 101600 Craniosynostosis, nonspecific, 0 Crouzon syndrome, 123500 Gastric cancer, somatic, 613659 Jackson-Weiss syndrome, 123150 LADD syndrome, 149730 Pfeiffer syndrome, 101600 Saethre-Chotzen syndrome, 101400 Scaphocephaly and Axenfeld-Rieger anomaly, 0 Scaphocephaly, maxillary retrusion, and mental retardation, 609579
FGFR3	138,5	100.0%	99.6%	Achondroplasia, 100800 Bladder cancer, somatic, 109800 CATSHL syndrome, 610474 Cervical cancer, somatic, 603956 Colorectal cancer, somatic, 114500 Crouzon syndrome with acanthosis nigricans, 612247 Hypochondroplasia, 146000 LADD syndrome, 149730 Muenke syndrome, 602849

				Nevus, epidermal, somatic, 162900 SADDAN, 616482 Spermatocytic seminoma, somatic, 273300 Thanatophoric dysplasia, type I, 187600 Thanatophoric dysplasia, type II, 187601
FGG	126,4	99.8%	98.0%	Afibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, 616004 Hypofibrinogenemia, congenital, 202400
FH	128	95.0%	88.5%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FHL1	64,1	98.3%	91.4%	?Uruguay faciocardiomusculoskeletal syndrome, 300280 Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 Myopathy, X-linked, with postural muscle atrophy, 300696 Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717 Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718 Scapuloperoneal myopathy, X-linked dominant, 300695
FIBP	124,7	100.0%	99.8%	Thauvin-Robinet-Faivre syndrome, 617107
FIG4	157,5	100.0%	99.6%	?Polymicrogyria, bilateral temporooccipital, 612691 Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228 Yunis-Varon syndrome, 216340
FIGLA	92	100.0%	99.6%	Premature ovarian failure 6, 612310
FKBP10	157,5	99.5%	97.3%	Bruck syndrome 1, 259450 Osteogenesis imperfecta, type XI, 610968
FKBP14	80,8	99.8%	97.9%	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557
FKRP	153,3	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155
FKTN	107,5	99.7%	96.1%	Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588
FLAD1	170,7	100.0%	99.6%	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100
FLCN	152,3	100.0%	100.0%	Birt-Hogg-Dube syndrome, 135150 Colorectal cancer, somatic, 114500 Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700

FLG	147,1	100.0%	99.9%	Ichthyosis vulgaris, 146700 {Dermatitis, atopic, susceptibility to, 2}, 605803
FLG2	352,3	99.9%	99.9%	Peeling skin syndrome 6, 618084
FLI1	153,8	99.3%	97.9%	Bleeding disorder, platelet-type, 21, 617443
FLNA	142,7	100.0%	99.9%	?FG syndrome 2, 300321 Cardiac valvular dysplasia, X-linked, 314400 Congenital short bowel syndrome, 300048 Frontometaphyseal dysplasia 1, 305620 Heterotopia, periventricular, 1, 300049 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Terminal osseous dysplasia, 300244
FLNB	123,6	99.7%	98.7%	Atelosteogenesis, type I, 108720 Atelosteogenesis, type III, 108721 Boomerang dysplasia, 112310 Larsen syndrome, 150250 Spondylocarpotarsal synostosis syndrome, 272460
FLNC	153,2	100.0%	99.6%	Cardiomyopathy, familial hypertrophic, 26, 0 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Myopathy, myofibrillar, 5, 609524
FLRT3	170,3	100.0%	100.0%	Hypogonadotropic hypogonadism 21 with anosmia, 615271
FLT3	110,2	99.9%	98.8%	Leukemia, acute lymphoblastic, somatic, 613065 Leukemia, acute myeloid, reduced survival in, somatic, 601626 Leukemia, acute myeloid, somatic, 601626
FLT4	160,3	99.2%	99.1%	Hemangioma, capillary infantile, somatic, 602089 Lymphatic malformation 1, 153100
FLVCR1	146,1	99.9%	99.2%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FLVCR2	124,8	100.0%	100.0%	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790
FMN2	109,6	86.6%	84.2%	Mental retardation, autosomal recessive 47, 616193
FMO3	131,9	100.0%	99.3%	Trimethylaminuria, 602079
FMR1	78,8	96.3%	91.0%	Fragile X syndrome, 300624 Fragile X tremor/ataxia syndrome, 300623 Premature ovarian failure 1, 311360
FN1	106	99.9%	98.9%	Glomerulopathy with fibronectin deposits 2, 601894

				Plasma fibronectin deficiency, 614101 Spondylometaphyseal dysplasia, corner fracture type, 184255
FOLR1	107,4	100.0%	99.9%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXC1	80,4	99.7%	97.5%	Anterior segment dysgenesis 3, multiple subtypes, 601631 Axenfeld-Rieger syndrome, type 3, 602482
FOXC2	122,3	100.0%	100.0%	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXE1	87,5	100.0%	99.7%	Bamforth-Lazarus syndrome, 241850 {Thyroid cancer, nonmedullary, 4}, 616534
FOXE3	88,3	89.7%	82.5%	Anterior segment dysgenesis 2, multiple subtypes, 610256 Cataract 34, multiple types, 612968 {Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349
FOXF1	153,8	100.0%	100.0%	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380
FOXG1	142	97.8%	88.7%	Rett syndrome, congenital variant, 613454
FOXI1	194,5	100.0%	100.0%	Enlarged vestibular aqueduct, 600791
FOXL2	117,7	100.0%	98.8%	Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 Premature ovarian failure 3, 608996
FOXN1	133	100.0%	99.5%	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXO1	107,9	100.0%	99.4%	Rhabdomyosarcoma, alveolar, 268220
FOXP1	114,1	100.0%	99.8%	Mental retardation with language impairment and with or without autistic features, 613670
FOXP2	129,2	99.5%	98.7%	Speech-language disorder-1, 602081
FOXP3	115,6	99.1%	94.8%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790
FOXRED1	121	99.8%	98.2%	Mitochondrial complex I deficiency, nuclear type 19, 618241
FRAS1	119,2	99.9%	99.2%	Fraser syndrome 1, 219000
FREM1	110,7	99.8%	98.4%	Bifid nose with or without anorectal and renal anomalies, 608980 Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485
FREM2	152,4	100.0%	99.5%	Cryptophthalmos, unilateral or bilateral, isolated, 123570 Fraser syndrome 2, 617666
FRMD4A	115,2	91.4%	90.5%	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819
FRMD7	101	99.8%	97.8%	Nystagmus 1, congenital, X-linked, 310700 Nystagmus, infantile periodic alternating, X-linked, 310700
FRMPD4	108,4	99.6%	97.2%	Mental retardation, X-linked 104, 300983
FRRS1L	99,1	85.5%	79.1%	Epileptic encephalopathy, early infantile, 37, 616981
FSCN2	164,3	100.0%	100.0%	Retinitis pigmentosa 30, 607921

FSHB	112	100.0%	100.0%	Hypogonadotropic hypogonadism 24 without anosmia, 229070
FSHR	96,6	99.9%	97.4%	Ovarian dysgenesis 1, 233300 Ovarian hyperstimulation syndrome, 608115 Ovarian response to FSH stimulation, 276400
FSIP2	96,8	99.9%	99.3%	Spermatogenic failure 34, 618153
FTCD	116,8	98.7%	95.2%	Glutamate formiminotransferase deficiency, 229100
FTH1	66,2	98.7%	87.9%	?Hemochromatosis, type 5, 615517
FTL	145,2	99.7%	96.7%	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159
FTO	95,8	83.8%	83.6%	Growth retardation, developmental delay, facial dysmorphism, 612938 {Obesity, susceptibility to, BMIQ14}, 612460
FTSJ1	122,9	99.2%	95.1%	Mental retardation, X-linked 9/44, 309549
FUCA1	125,9	100.0%	99.9%	Fucosidosis, 230000
FUK	NC	NC	NC	Congenital disorder of glycosylation with defective fucosylation 2, 618324
FUS	119,4	99.5%	97.0%	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 Essential tremor, hereditary, 4, 614782
FUT6	142,5	100.0%	100.0%	Fucosyltransferase 6 deficiency, 613852
FUT8	130,4	99.9%	99.2%	Congenital disorder of glycosylation with defective fucosylation 1, 618005
FUZ	128,5	100.0%	100.0%	{Neural tube defects, susceptibility to}, 182940
FXN	64,9	99.7%	96.8%	Friedreich ataxia, 229300 Friedreich ataxia with retained reflexes, 229300
FXYD2	108,1	100.0%	100.0%	Hypomagnesemia 2, renal, 154020
FYB1	96,3	99.5%	96.8%	Thrombocytopenia 3, 273900
FYCO1	123,2	100.0%	100.0%	Cataract 18, autosomal recessive, 610019
FZD2	179,4	99.7%	97.8%	Omodysplasia 2, 164745
FZD4	179,1	100.0%	100.0%	Exudative vitreoretinopathy 1, 133780 Retinopathy of prematurity, 133780
FZD6	186,4	100.0%	100.0%	Nail disorder, nonsyndromic congenital, 10, (claw-shaped nails), 614157
G6PC	146,8	100.0%	99.9%	Glycogen storage disease Ia, 232200
G6PC3	114,6	100.0%	100.0%	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
G6PD	114,4	99.5%	97.4%	Hemolytic anemia, G6PD deficient (favism), 300908 {Resistance to malaria due to G6PD deficiency}, 611162
GAA	160,8	100.0%	99.9%	Glycogen storage disease II, 232300
GAB1	155,1	100.0%	99.5%	?Deafness, autosomal recessive 26, 605428

GABBR2	109,2	98.2%	94.6%	Epileptic encephalopathy, early infantile, 59, 617904 Neurodevelopmental disorder with poor language and loss of hand skills, 617903 {Nicotine dependence, protection against}, 188890 {Nicotine dependence, susceptibility to}, 188890
GABRA1	164,2	100.0%	99.8%	Epileptic encephalopathy, early infantile, 19, 615744 {Epilepsy, childhood absence, susceptibility to, 4}, 611136 {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136
GABRB1	169	99.9%	99.8%	Epileptic encephalopathy, early infantile, 45, 617153
GABRB2	129,2	100.0%	99.9%	Epileptic encephalopathy, infantile or early childhood, 2, 617829
GABRB3	131,7	99.5%	97.8%	Epileptic encephalopathy, early infantile, 43, 617113 {Epilepsy, childhood absence, susceptibility to, 5}, 612269
GABRG2	126,4	91.1%	89.7%	Epilepsy, generalized, with febrile seizures plus, type 3, 611277 Epileptic encephalopathy, early infantile, 74, 618396 Febrile seizures, familial, 8, 611277 {Epilepsy, childhood absence, susceptibility to, 2}, 607681
GAD1	112,7	99.9%	99.7%	?Cerebral palsy, spastic quadriplegic, 1, 603513
GAL	169,5	100.0%	100.0%	?Epilepsy, familial temporal lobe, 8, 616461
GALC	102,9	99.8%	98.8%	Krabbe disease, 245200
GALE	140	100.0%	100.0%	Galactose epimerase deficiency, 230350
GALK1	165,2	100.0%	99.9%	Galactokinase deficiency with cataracts, 230200
GALNS	108,3	100.0%	99.3%	Mucopolysaccharidosis IVA, 253000
GALNT3	125,8	99.9%	98.7%	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GALT	152,6	100.0%	100.0%	Galactosemia, 230400
GAMT	112,5	98.3%	91.5%	Cerebral creatine deficiency syndrome 2, 612736
GAN	142,2	99.9%	99.4%	Giant axonal neuropathy-1, 256850
GANAB	107,1	99.9%	98.3%	Polycystic kidney disease 3, 600666
GARS	128,9	100.0%	99.7%	Charcot-Marie-Tooth disease, type 2D, 601472 Neuropathy, distal hereditary motor, type VA, 600794
GAS8	127,3	100.0%	99.4%	Ciliary dyskinesia, primary, 33, 616726
GATA1	92,9	99.9%	98.2%	Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835 Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 Thrombocytopenia with beta-thalassemia, X-linked, 314050 Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367
GATA2	115	100.0%	99.0%	Emberger syndrome, 614038 Immunodeficiency 21, 614172 {Leukemia, acute myeloid, susceptibility to}, 601626 {Myelodysplastic syndrome, susceptibility to}, 614286

GATA3	220,5	100.0%	100.0%	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GATA4	87,6	95.9%	86.7%	?Testicular anomalies with or without congenital heart disease, 615542 Atrial septal defect 2, 607941 Atrioventricular septal defect 4, 614430 Tetralogy of Fallot, 187500 Ventricular septal defect 1, 614429
GATA5	74	100.0%	99.2%	Congenital heart defects, multiple types, 5, 617912
GATA6	110,2	98.3%	92.5%	Atrial septal defect 9, 614475 Atrioventricular septal defect 5, 614474 Pancreatic agenesis and congenital heart defects, 600001 Persistent truncus arteriosus, 217095 Tetralogy of Fallot, 187500
GATAD1	136,1	100.0%	100.0%	?Cardiomyopathy, dilated, 2B, 614672
GATAD2B	97,4	100.0%	99.1%	Mental retardation, autosomal dominant 18, 615074
GATM	137,3	100.0%	100.0%	Cerebral creatine deficiency syndrome 3, 612718
GBA	169,8	100.0%	100.0%	Gaucher disease, perinatal lethal, 608013 Gaucher disease, type I, 230800 Gaucher disease, type II, 230900 Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 {Lewy body dementia, susceptibility to}, 127750 {Parkinson disease, late-onset, susceptibility to}, 168600
GBA2	141,5	100.0%	99.6%	Spastic paraplegia 46, autosomal recessive, 614409
GBE1	157,4	99.9%	99.7%	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GCDH	145,9	100.0%	99.1%	Glutaricaciduria, type I, 231670
GCH1	84,8	100.0%	99.5%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GCK	138,6	100.0%	100.0%	Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, permanent neonatal, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485 MODY, type II, 125851
GCLC	143,7	99.3%	95.8%	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 {Myocardial infarction, susceptibility to}, 608446
GCM2	135,1	100.0%	100.0%	Hyperparathyroidism 4, 617343 Hypoparathyroidism, familial isolated, 146200
GCNT2	151,2	99.5%	99.5%	Adult i phenotype without cataract, 110800

				Cataract 13 with adult i phenotype, 116700 [Blood group, Ii], 110800
GCSH	32,1	88.4%	69.8%	?Glycine encephalopathy, 605899
GDAP1	145,6	99.8%	99.0%	Charcot-Marie-Tooth disease, axonal, type 2K, 607831 Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706 Charcot-Marie-Tooth disease, recessive intermediate, A, 608340 Charcot-Marie-Tooth disease, type 4A, 214400
GDAP2	118,2	99.9%	99.3%	Spinocerebellar ataxia, autosomal recessive 27, 618369
GDF1	50,7	97.8%	84.7%	Congenital heart defects, multiple types, 6, 613854 Right atrial isomerism (Ivemark), 208530
GDF2	142,4	100.0%	100.0%	Telangiectasia, hereditary hemorrhagic, type 5, 615506
GDF3	127,9	100.0%	100.0%	Klippel-Feil syndrome 3, autosomal dominant, 613702 Microphthalmia with coloboma 6, 613703 Microphthalmia, isolated 7, 613704
GDF5	169,6	100.0%	100.0%	?Acromesomelic dysplasia, Hunter-Thompson type, 201250 Brachydactyly, type A1, C, 615072 Brachydactyly, type A2, 112600 Brachydactyly, type C, 113100 Chondrodysplasia, Grebe type, 200700 Du Pan syndrome, 228900 Multiple synostoses syndrome 2, 610017 Symphalangism, proximal, 1B, 615298 {Osteoarthritis-5}, 612400
GDF6	156,3	100.0%	100.0%	Klippel-Feil syndrome 1, autosomal dominant, 118100 Leber congenital amaurosis 17, 615360 Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094 Multiple synostoses syndrome 4, 617898
GDF9	141,5	100.0%	100.0%	?Premature ovarian failure 14, 618014
GDI1	136,4	99.5%	97.9%	Mental retardation, X-linked 41, 300849
GDNF	183,5	100.0%	100.0%	Central hypoventilation syndrome, 209880 {Hirschsprung disease, susceptibility to, 3}, 613711 {Pheochromocytoma, modifier of}, 171300
GEMIN4	148,4	100.0%	99.7%	Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities, 617913
GFAP	103,7	91.9%	91.4%	Alexander disease, 203450
GFER	90,6	100.0%	99.6%	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076
GFI1	105,7	100.0%	100.0%	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847

				?Neutropenia, severe congenital 2, autosomal dominant, 613107
GFI1B	170,9	99.9%	98.2%	Bleeding disorder, platelet-type, 17, 187900
GFM1	104,2	99.9%	99.0%	Combined oxidative phosphorylation deficiency 1, 609060
GFM2	121,3	98.9%	95.6%	Combined oxidative phosphorylation deficiency 39, 618397
GFPT1	146	99.9%	99.1%	Myasthenia, congenital, 12, with tubular aggregates, 610542
GGCX	101,2	100.0%	99.4%	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842 Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450
GGT1	12,1	20.3%	18.2%	?Glutathioninuria, 231950
GH1	159,5	100.0%	100.0%	Growth hormone deficiency, isolated, type IA, 262400 Growth hormone deficiency, isolated, type IB, 612781 Growth hormone deficiency, isolated, type II, 173100 Kowarski syndrome, 262650
GHR	150,6	99.5%	99.4%	Growth hormone insensitivity, partial, 604271 Increased responsiveness to growth hormone, 604271 Laron dwarfism, 262500 {Hypercholesterolemia, familial, modifier of}, 143890
GHRHR	108,1	96.0%	95.2%	Growth hormone deficiency, isolated, type IV, 618157
GHSR	173,6	98.7%	95.2%	Growth hormone deficiency, isolated partial, 615925
GIF	NC	NC	NC	Intrinsic factor deficiency, 261000
GINS1	122,7	98.1%	90.6%	Immunodeficiency 55, 617827
GIPC3	127,8	98.9%	95.2%	Deafness, autosomal recessive 15, 601869
GJA1	156,2	100.0%	100.0%	Atrioventricular septal defect 3, 600309 Craniometaphyseal dysplasia, autosomal recessive, 218400 Erythrokeratodermia variabilis et progressiva 3, 617525 Hypoplastic left heart syndrome 1, 241550 Oculodentodigital dysplasia, 164200 Oculodentodigital dysplasia, autosomal recessive, 257850 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100
GJA3	162,2	100.0%	100.0%	Cataract 14, multiple types, 601885
GJA5	207,8	100.0%	100.0%	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic (GJA5/SCN5A), 108770
GJA8	156,6	100.0%	100.0%	Cataract 1, multiple types, 116200
GJB1	150,9	100.0%	99.9%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GJB2	141,4	100.0%	100.0%	Bart-Pumphrey syndrome, 149200 Deafness, autosomal dominant 3A, 601544 Deafness, autosomal recessive 1A, 220290

				Hystrix-like ichthyosis with deafness, 602540 Keratitis-ichthyosis-deafness syndrome, 148210 Keratoderma, palmoplantar, with deafness, 148350 Vohwinkel syndrome, 124500
GJB3	228,5	100.0%	100.0%	Deafness, autosomal dominant 2B, 612644 Deafness, autosomal dominant, with peripheral neuropathy, 0 Deafness, autosomal recessive, 0 Deafness, digenic, GJB2/GJB3, 220290 Erythrokeratodermia variabilis et progressiva 1, 133200
GJB4	246,1	100.0%	100.0%	Erythrokeratodermia variabilis et progressiva 2, 617524
GJB6	140,9	100.0%	100.0%	Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290 Ectodermal dysplasia 2, Clouston type, 129500
GJC2	45,3	92.6%	75.4%	Leukodystrophy, hypomyelinating, 2, 608804 Lymphatic malformation 3, 613480 Spastic paraplegia 44, autosomal recessive, 613206
GK	44,1	84.9%	63.9%	Glycerol kinase deficiency, 307030
GLA	73,6	99.5%	95.8%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	82,6	99.7%	95.4%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLDC	59,2	90.6%	79.2%	Glycine encephalopathy, 605899
GLDN	106,6	99.8%	96.9%	Lethal congenital contracture syndrome 11, 617194
GLE1	97,1	100.0%	99.9%	Congenital arthrogryposis with anterior horn cell disease, 611890 Lethal congenital contracture syndrome 1, 253310
GLI1	122,9	100.0%	99.8%	Polydactyly, postaxial, type A8, 618123 Polydactyly, preaxial I, 174400
GLI2	158,2	100.0%	100.0%	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829
GLI3	139,5	100.0%	99.3%	Greig cephalopolysyndactyly syndrome, 175700 Pallister-Hall syndrome, 146510 Polydactyly, postaxial, types A1 and B, 174200 Polydactyly, preaxial, type IV, 174700 {Hypothalamic hamartomas, somatic}, 241800

GLIS2	129,6	100.0%	100.0%	Nephronophthisis 7, 611498
GLIS3	123,7	100.0%	99.5%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GLMN	70,2	99.0%	95.0%	Glomuvenous malformations, 138000
GLRA1	96,8	100.0%	99.7%	Hyperekplexia 1, 149400
GLRB	104,6	99.5%	94.5%	Hyperekplexia 2, 614619
GLRX5	137,6	99.6%	96.1%	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GLS	84,2	99.5%	96.0%	?Infantile cataract, skin abnormalities, glutamate excess, and impaired intellectual development, 618339 Epileptic encephalopathy, early infantile, 71, 618328 Global developmental delay, progressive ataxia, and elevated glutamine, 618312
GLUD1	65,5	98.1%	87.5%	Hyperinsulinism-hyperammonemia syndrome, 606762
GLUL	77	99.7%	96.5%	Glutamine deficiency, congenital, 610015
GLYCTK	161,3	100.0%	99.5%	D-glyceric aciduria, 220120
GM2A	122	100.0%	100.0%	GM2-gangliosidosis, AB variant, 272750
GMNN	124,4	99.7%	97.9%	Meier-Gorlin syndrome 6, 616835
GMPPA	147,2	100.0%	99.8%	Alacrima, achalasia, and mental retardation syndrome, 615510
GMPPB	211,8	100.0%	100.0%	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
GNA11	162,4	100.0%	99.5%	Hypocalcemia, autosomal dominant 2, 615361 Hypocalciuric hypercalcemia, type II, 145981
GNAI2	124,3	100.0%	100.0%	Pituitary ACTH-secreting adenoma, 0 Ventricular tachycardia, idiopathic, 192605
GNAI3	88,3	98.9%	94.5%	Auriculocondylar syndrome 1, 602483
GNAL	130,2	99.4%	96.5%	Dystonia 25, 615073
GNAO1	152,8	93.8%	93.8%	Epileptic encephalopathy, early infantile, 17, 615473 Neurodevelopmental disorder with involuntary movements, 617493
GNAQ	52,8	81.0%	64.3%	Capillary malformations, congenital, 1, somatic, mosaic, 163000 Sturge-Weber syndrome, somatic, mosaic, 185300
GNAS	211,3	100.0%	100.0%	ACTH-independent macronodular adrenal hyperplasia, 219080 McCune-Albright syndrome, somatic, mosaic, 174800 Osseous heteroplasia, progressive, 166350 Pituitary adenoma 3, multiple types, somatic, 617686 Pseudohypoparathyroidism Ia, 103580 Pseudohypoparathyroidism Ib, 603233 Pseudohypoparathyroidism Ic, 612462 Pseudopseudohypoparathyroidism, 612463

GNAS-AS1	NC	NC	NC	Pseudohypoparathyroidism, type IB, 603233
GNAT1	176,4	100.0%	100.0%	Night blindness, congenital stationary, autosomal dominant 3, 610444 Night blindness, congenital stationary, type 1G, 616389
GNAT2	100,7	99.8%	98.2%	Achromatopsia 4, 613856
GNB1	145,2	100.0%	100.0%	Leukemia, acute lymphoblastic, somatic, 613065 Mental retardation, autosomal dominant 42, 616973
GNB3	146	100.0%	99.9%	Night blindness, congenital stationary, type 1H, 617024 {Hypertension, essential, susceptibility to}, 145500
GNB4	150,3	100.0%	100.0%	Charcot-Marie-Tooth disease, dominant intermediate F, 615185
GNB5	113	99.9%	97.5%	Intellectual developmental disorder with cardiac arrhythmia, 617173 Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182
GNE	113,8	100.0%	99.3%	Nonaka myopathy, 605820 Sialuria, 269921
GNMT	126,9	99.9%	98.7%	Glycine N-methyltransferase deficiency, 606664
GNPAT	127,2	99.5%	96.8%	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	148	100.0%	99.3%	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GNPTG	177,6	100.0%	98.5%	Mucolipidosis III gamma, 252605
GNRH1	84,5	96.5%	78.0%	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841
GNRHR	145,4	100.0%	100.0%	Hypogonadotropic hypogonadism 7 without anosmia, 146110
GNS	94,5	99.6%	95.2%	Mucopolysaccharidosis type IIID, 252940
GORAB	165,7	100.0%	98.9%	Geroderma osteodysplasticum, 231070
GOSR2	102,6	95.8%	93.7%	Epilepsy, progressive myoclonic 6, 614018
GOT1	107,7	100.0%	99.4%	Aspartate aminotransferase, serum level of, QTL1, 614419
GP1BA	136,8	98.7%	95.7%	Bernard-Soulier syndrome, type A1 (recessive), 231200 Bernard-Soulier syndrome, type A2 (dominant), 153670 von Willebrand disease, platelet-type, 177820 {Nonarteritic anterior ischemic optic neuropathy, susceptibility to}, 258660
GP1BB	68,6	94.5%	83.1%	Bernard-Soulier syndrome, type B, 231200 Giant platelet disorder, isolated, 231200
GP6	123,2	100.0%	99.7%	Bleeding disorder, platelet-type, 11, 614201
GP9	134,3	99.9%	98.3%	Bernard-Soulier syndrome, type C, 231200
GPAA1	123,1	100.0%	98.3%	Glycosylphosphatidylinositol biosynthesis defect 15, 617810
GPC3	75,7	98.7%	92.7%	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPC6	126,6	100.0%	100.0%	Omodysplasia 1, 258315

GPD1	87,7	100.0%	99.4%	Hypertriglyceridemia, transient infantile, 614480
GPD1L	128	100.0%	99.9%	Brugada syndrome 2, 611777
GPHN	144,7	99.9%	98.8%	Molybdenum cofactor deficiency C, 615501
GPI	141,4	100.0%	99.8%	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470
GPIHBP1	149,5	100.0%	100.0%	Hyperlipoproteinemia, type 1D, 615947
GPNMB	150,5	100.0%	100.0%	Amyloidosis, primary localized cutaneous, 3, 617920
GPR101	111,7	100.0%	100.0%	Pituitary adenoma 2, GH-secreting, 300943
GPR143	59,5	91.0%	79.1%	Nystagmus 6, congenital, X-linked, 300814 Ocular albinism, type I, Nettleship-Falls type, 300500
GPR179	146,8	100.0%	100.0%	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
GPR68	165,4	99.9%	99.0%	Amelogenesis imperfecta, hypomaturation type, IIA6, 617217
GPR88	146,7	100.0%	99.6%	?Chorea, childhood-onset, with psychomotor retardation, 616939
GPRASP2	79,6	100.0%	99.5%	?Deafness, X-linked 7, 301018
GPSM2	119,6	99.8%	99.0%	Chudley-McCullough syndrome, 604213
GPT2	121,7	100.0%	99.4%	Mental retardation, autosomal recessive 49, 616281
GPX4	165,9	94.4%	90.7%	Spondylometaphyseal dysplasia, Sedaghatian type, 250220
GREB1L	128	99.9%	99.0%	Renal hypodysplasia/aplasia 3, 617805
GREM2	149,7	100.0%	100.0%	Tooth agenesis, selective, 9, 617275
GRHL2	116,8	100.0%	100.0%	Corneal dystrophy, posterior polymorphous, 4, 618031 Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029
GRHL3	133,2	100.0%	99.8%	Van der Woude syndrome 2, 606713
GRHPR	99,6	84.2%	81.7%	Hyperoxaluria, primary, type II, 260000
GRIA3	82,4	98.6%	92.1%	Mental retardation, X-linked 94, 300699
GRIA4	124	99.7%	98.1%	Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864
GRID2	146,8	99.9%	99.4%	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRIK2	121,7	96.2%	95.4%	Mental retardation, autosomal recessive, 6, 611092
GRIN1	166,1	100.0%	99.9%	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254 Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820
GRIN2A	131,2	100.0%	100.0%	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570
GRIN2B	158	99.8%	99.0%	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation, autosomal dominant 6, 613970
GRIN2D	82,8	91.9%	79.5%	Epileptic encephalopathy, early infantile, 46, 617162
GRIP1	111,1	100.0%	99.3%	Fraser syndrome 3, 617667

GRK1	135,7	100.0%	100.0%	Oguchi disease-2, 613411
GRM1	156,6	100.0%	99.9%	Spinocerebellar ataxia 44, 617691 Spinocerebellar ataxia, autosomal recessive 13, 614831
GRM6	138,3	98.0%	92.4%	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270
GRN	174,1	100.0%	100.0%	Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
GRXCR1	154,5	100.0%	99.7%	Deafness, autosomal recessive 25, 613285
GRXCR2	112,4	100.0%	99.9%	?Deafness, autosomal recessive 101, 615837
GSC	133,8	100.0%	98.9%	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471
GSN	115,5	95.6%	93.5%	Amyloidosis, Finnish type, 105120
GSS	93,3	100.0%	99.2%	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900
GTF2E2	85,2	100.0%	98.2%	Trichothiodystrophy 6, nonphotosensitive, 616943
GTF2H5	81,8	99.9%	95.9%	Trichothiodystrophy 3, photosensitive, 616395
GTPBP2	128,9	99.7%	98.6%	Jaberi-Elahi syndrome, 617988
GTPBP3	164,7	100.0%	100.0%	Combined oxidative phosphorylation deficiency 23, 616198
GUCA1A	172,9	100.0%	100.0%	Cone dystrophy-3, 602093 Cone-rod dystrophy 14, 602093
GUCA1B	130	100.0%	99.9%	Retinitis pigmentosa 48, 613827
GUCY1A3	NC	NC	NC	Moyamoya 6 with achalasia, 615750
GUCY2C	117,5	100.0%	99.4%	Diarrhea 6, 614616 Meconium ileus, 614665
GUCY2D	108,9	100.0%	99.9%	?Choroidal dystrophy, central areolar 1, 215500 Cone-rod dystrophy 6, 601777 Leber congenital amaurosis 1, 204000
GUF1	95,1	99.9%	98.4%	?Epileptic encephalopathy, early infantile, 40, 617065
GULOP	NC	NC	NC	Scurvy, 0
GUSB	99,5	92.5%	90.5%	Mucopolysaccharidosis VII, 253220
GYG1	125,6	100.0%	99.4%	?Glycogen storage disease XV, 613507 Polyglucosan body myopathy 2, 616199
GYS1	109,6	100.0%	98.6%	Glycogen storage disease 0, muscle, 611556
GYS2	120,1	99.7%	98.0%	Glycogen storage disease 0, liver, 240600
GZF1	187,8	100.0%	99.7%	Joint laxity, short stature, and myopia, 617662
H19	NC	NC	NC	Beckwith-Wiedemann syndrome, 130650 Silver-Russell syndrome, 180860

				Wilms tumor 2, 194071
H6PD	193,5	99.0%	99.0%	Cortisone reductase deficiency 1, 604931
HAAO	104,6	100.0%	99.8%	Vertebral, cardiac, renal, and limb defects syndrome 1, 617660
HACE1	136,2	99.9%	99.1%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADH	111,1	99.3%	98.8%	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HADHA	72,9	96.3%	89.3%	Fatty liver, acute, of pregnancy, 609016 HELLP syndrome, maternal, of pregnancy, 609016 LCHAD deficiency, 609016 Trifunctional protein deficiency, 609015
HADHB	77,6	96.7%	83.8%	Trifunctional protein deficiency, 609015
HAMP	169	100.0%	100.0%	Hemochromatosis, type 2B, 613313
HARS	134,8	100.0%	100.0%	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504
HARS2	136,6	100.0%	99.9%	?Perrault syndrome 2, 614926
HAVCR2	117,7	100.0%	99.9%	T-cell lymphoma, subcutaneous panniculitis-like, 618398
HAX1	137,4	100.0%	100.0%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HBA1	137,9	100.0%	100.0%	Erythrocytosis, 7, 617981 Heinz body anemias, alpha-, 140700 Hemoglobin H disease, nondeletional, 613978 Methemoglobinemia, alpha type, 617973 Thalassemias, alpha-, 604131
HBA2	127,4	98.7%	92.5%	Erythrocytosis 7, 617981 Heinz body anemia, 140700 Hemoglobin H disease, deletional and nondeletional, 613978 Thalassemia, alpha-, 604131
HBB	131,9	100.0%	100.0%	Delta-beta thalassemia, 141749 Erythrocytosis 6, 617980 Heinz body anemia, 140700 Hereditary persistence of fetal hemoglobin, 141749 Methmoglobinemia, beta type, 617971 Sickle cell anemia, 603903 Thalassemia, beta, 613985 Thalassemia-beta, dominant inclusion-body, 603902 {Malaria, resistance to}, 611162
HBD	178,4	100.0%	100.0%	Thalassemia due to Hb Lepore, 0 Thalassemia, delta-, 0

HBG1	134,8	96.9%	91.6%	Fetal hemoglobin quantitative trait locus 1, 141749
HBG2	242,1	100.0%	100.0%	Cyanosis, transient neonatal, 613977 Fetal hemoglobin quantitative trait locus 1, 141749
HCCS	92,4	99.2%	95.2%	Linear skin defects with multiple congenital anomalies 1, 309801
HCFC1	104,4	99.3%	95.8%	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541
HCN1	137,5	100.0%	99.7%	Epileptic encephalopathy, early infantile, 24, 615871
HCN4	96	100.0%	99.7%	Brugada syndrome 8, 613123 Sick sinus syndrome 2, 163800
HCRT	129,1	99.2%	93.0%	?Narcolepsy 1, 161400
HDAC6	113,9	99.7%	97.7%	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863
HDAC8	108,1	100.0%	99.2%	Cornelia de Lange syndrome 5, 300882
HECW2	109,3	99.9%	98.7%	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268
HELLS	102,8	98.8%	92.7%	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911
HEPACAM	118,3	95.5%	88.7%	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926
HERC1	142,1	100.0%	99.7%	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
HERC2	95,1	80.0%	76.1%	Mental retardation, autosomal recessive 38, 615516 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220
HES7	53,9	90.1%	72.7%	Spondylocostal dysostosis 4, autosomal recessive, 613686
HESX1	66,2	100.0%	98.7%	Growth hormone deficiency with pituitary anomalies, 182230 Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230
HEXA	106,3	93.7%	92.4%	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800 [Hex A pseudodeficiency], 272800
HEXB	163	99.7%	98.5%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HFE	108	100.0%	98.9%	Hemochromatosis, 235200 [Transferrin serum level QTL2], 614193 {Alzheimer disease, susceptibility to}, 104300 {Microvascular complications of diabetes 7}, 612635 {Porphyria cutanea tarda, susceptibility to}, 176100 {Porphyria variegata, susceptibility to}, 176200
HFE2	NC	NC	NC	Hemochromatosis, type 2A, 602390
HFM1	54,6	96.7%	90.6%	Premature ovarian failure 9, 615724
HGD	98,2	100.0%	99.8%	Alkaptonuria, 203500

HGF	138	99.9%	99.2%	Deafness, autosomal recessive 39, 608265
HGSNAT	98,3	87.2%	86.2%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544
HIBCH	69,9	96.3%	79.8%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HIKESHI	54,5	96.8%	84.3%	Leukodystrophy, hypomyelinating, 13, 616881
HINT1	63,3	91.2%	79.1%	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200
HIST1H1E	112,1	100.0%	100.0%	Rahman syndrome, 617537
HIVEP2	162,8	100.0%	100.0%	Mental retardation, autosomal dominant 43, 616977
HK1	116,5	100.0%	99.7%	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285 Retinitis pigmentosa 79, 617460
HLCS	142,3	100.0%	100.0%	Holocarboxylase synthetase deficiency, 253270
HMBS	97,3	100.0%	98.4%	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000
HMGB3	37,1	81.1%	62.4%	?Microphthalmia, syndromic 13, 300915
HMGCL	119,4	99.9%	98.7%	HMG-CoA lyase deficiency, 246450
HMGCS2	102,4	100.0%	99.4%	HMG-CoA synthase-2 deficiency, 605911
HMOX1	137,4	96.5%	90.7%	Heme oxygenase-1 deficiency, 614034 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963
HMX1	39,8	85.4%	63.9%	Oculoauricular syndrome, 612109
HNF1A	163,2	100.0%	99.9%	Diabetes mellitus, insulin-dependent, 20, 612520 Hepatic adenoma, somatic, 142330 MODY, type III, 600496 Renal cell carcinoma, 144700 {Diabetes mellitus, insulin-dependent}, 222100 {Diabetes mellitus, noninsulin-dependent, 2}, 125853
HNF1B	118,8	99.6%	96.8%	Diabetes mellitus, noninsulin-dependent, 125853 Renal cysts and diabetes syndrome, 137920 {Renal cell carcinoma}, 144700
HNF4A	129,8	99.9%	99.0%	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026 MODY, type I, 125850 {Diabetes mellitus, noninsulin-dependent}, 125853
HNMT	133,1	100.0%	99.9%	Mental retardation, autosomal recessive 51, 616739 {Asthma, susceptibility to}, 600807
HNRNPA1	61	98.3%	85.0%	?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3, 615424 Amyotrophic lateral sclerosis 20, 615426
HNRNPA2B1	139,1	99.8%	98.2%	?Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2, 615422

HNRNPDL	88,6	99.6%	93.8%	Muscular dystrophy, limb-girdle, autosomal dominant 3, 609115
HNRNPH2	126,2	100.0%	100.0%	Mental retardation, X-linked, syndromic, Bain type, 300986
HNRNPK	61,1	88.1%	79.9%	Au-Kline syndrome, 616580
HNRNPU	149,9	99.9%	99.3%	Epileptic encephalopathy, early infantile, 54, 617391
HOGA1	149,6	100.0%	99.1%	Hyperoxaluria, primary, type III, 613616
HOMER2	113,1	99.8%	97.8%	?Deafness, autosomal dominant 68, 616707
HOXA1	164	100.0%	100.0%	Athabaskan brainstem dysgenesis syndrome, 601536 Bosley-Salih-Alorainy syndrome, 601536
HOXA11	88,3	100.0%	98.0%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
HOXA13	77,7	90.9%	79.6%	Guttmacher syndrome, 176305 Hand-foot-uterus syndrome, 140000
HOXA2	83	100.0%	99.9%	?Microtia, hearing impairment, and cleft palate (AR), 612290 Microtia with or without hearing impairment (AD), 612290
HOXB1	147,8	100.0%	100.0%	Facial paresis, hereditary congenital, 3, 614744
HOXC13	172,8	100.0%	100.0%	Ectodermal dysplasia 9, hair/nail type, 614931
HOXD10	157,3	100.0%	100.0%	Charcot-Marie-Tooth disease, foot deformity of, 192950 Vertical talus, congenital, 192950
HOXD13	198,2	100.0%	100.0%	?Brachydactyly-syndactyly syndrome, 610713 Brachydactyly, type D, 113200 Brachydactyly, type E, 113300 Syndactyly, type V, 186300 Synpolydactyly 1, 186000
HPCA	253,9	100.0%	100.0%	Dystonia 2, torsion, autosomal recessive, 224500
HPD	148,1	100.0%	99.7%	Hawkinsinuria, 140350 Tyrosinemia, type III, 276710
HPGD	90,6	99.9%	98.9%	Cranioosteoarthropathy, 259100 Digital clubbing, isolated congenital, 119900 Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100
HPRT1	59,8	98.3%	88.2%	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322
HPS1	115,8	100.0%	99.9%	Hermansky-Pudlak syndrome 1, 203300
HPS3	132,7	99.9%	98.8%	Hermansky-Pudlak syndrome 3, 614072
HPS4	128,1	100.0%	99.9%	Hermansky-Pudlak syndrome 4, 614073
HPS5	122,8	99.9%	98.7%	Hermansky-Pudlak syndrome 5, 614074
HPS6	164,6	99.9%	97.8%	Hermansky-Pudlak syndrome 6, 614075
HPSE2	100,6	100.0%	99.7%	Urofacial syndrome 1, 236730

HR	117,4	99.6%	97.3%	Alopecia universalis, 203655 Atrichia with papular lesions, 209500 Hypotrichosis 4, 146550
HRAS	182,3	100.0%	100.0%	Bladder cancer, somatic, 109800 Congenital myopathy with excess of muscle spindles, 218040 Costello syndrome, 218040 Nevus sebaceous or woolly hair nevus, somatic, 162900 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Spitz nevus or nevus spilus, somatic, 137550 Thyroid carcinoma, follicular, somatic, 188470
HRG	126,1	95.0%	94.2%	Thrombophilia due to elevated HRG, 613116 Thrombophilia due to HRG deficiency, 613116
HSD11B1	113,8	100.0%	99.0%	Cortisone reductase deficiency 2, 614662
HSD11B2	165,6	94.3%	87.3%	Apparent mineralocorticoid excess, 218030
HSD17B10	92,4	100.0%	98.4%	HSD10 mitochondrial disease, 300438
HSD17B3	116,4	100.0%	99.9%	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	109,4	96.3%	93.6%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B2	131,8	100.0%	99.9%	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810
HSD3B7	143,7	99.5%	96.2%	Bile acid synthesis defect, congenital, 1, 607765
HSF4	148,7	99.8%	98.5%	Cataract 5, multiple types, 116800
HSPA9	82,6	89.5%	84.2%	Anemia, sideroblastic, 4, 182170 Even-plus syndrome, 616854
HSPB1	67,2	98.0%	91.9%	Charcot-Marie-Tooth disease, axonal, type 2F, 606595 Neuropathy, distal hereditary motor, type IIB, 608634
HSPB3	226,1	100.0%	100.0%	?Neuronopathy, distal hereditary motor, type IIC, 613376
HSPB8	213,9	100.0%	100.0%	Charcot-Marie-Tooth disease, axonal, type 2L, 608673 Neuropathy, distal hereditary motor, type IIA, 158590
HSPD1	74,3	98.1%	92.5%	Leukodystrophy, hypomyelinating, 4, 612233 Spastic paraplegia 13, autosomal dominant, 605280
HSPG2	119,8	99.5%	98.8%	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
HTR1A	184,7	100.0%	100.0%	Periodic fever, menstrual cycle dependent, 614674
HTRA1	89	95.2%	87.0%	CARASIL syndrome, 600142 Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779 {Macular degeneration, age-related, 7}, 610149 {Macular degeneration, age-related, neovascular type}, 610149

HTRA2	132,6	100.0%	99.6%	3-methylglutaconic aciduria, type VIII, 617248 {Parkinson disease 13}, 610297
HTT	125,2	98.9%	97.6%	Huntington disease, 143100 Lopes-Maciel-Rodan syndrome, 617435
HUWE1	79,3	99.1%	94.3%	Mental retardation, X-linked syndromic, Turner type, 300706
HYAL1	110,7	100.0%	100.0%	?Mucopolysaccharidosis type IX, 601492
HYDIN	106,8	99.8%	98.9%	Ciliary dyskinesia, primary, 5, 608647
HYLS1	156,6	100.0%	100.0%	Hydrolethalus syndrome, 236680
HYOU1	130,8	99.9%	99.5%	?Immunodeficiency 59 and hypoglycemia, 233600
IARS	125,4	99.9%	99.0%	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093
IARS2	142,9	100.0%	99.9%	?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
IBA57	137,4	99.3%	95.9%	?Spastic paraplegia 74, autosomal recessive, 616451 Multiple mitochondrial dysfunctions syndrome 3, 615330
ICK	110	99.8%	99.3%	Endocrine-cerebroosteodysplasia, 612651 {Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924
ICOS	156,4	100.0%	99.9%	Immunodeficiency, common variable, 1, 607594
IDH2	98,5	100.0%	99.6%	D-2-hydroxyglutaric aciduria 2, 613657
IDH3B	128,2	95.5%	95.4%	Retinitis pigmentosa 46, 612572
IDS	100,9	99.9%	97.1%	Mucopolysaccharidosis II, 309900
IDUA	148,1	98.9%	94.6%	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016
IER3IP1	106,3	94.3%	82.8%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	110,8	99.8%	98.2%	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFITM5	94,9	100.0%	99.1%	Osteogenesis imperfecta, type V, 610967
IFNAR2	129,5	99.5%	96.9%	?Immunodeficiency 45, 616669 {Hepatitis B virus, susceptibility to}, 610424
IFNGR1	145	99.9%	99.3%	Immunodeficiency 27A, mycobacteriosis, AR, 209950 Immunodeficiency 27B, mycobacteriosis, AD, 615978 {H. pylori infection, susceptibility to}, 600263 {Hepatitis B virus infection, susceptibility to}, 610424 {Tuberculosis infection, protection against}, 607948 {Tuberculosis, susceptibility to}, 607948
IFNGR2	125,2	97.0%	93.5%	Immunodeficiency 28, mycobacteriosis, 614889
IFT122	120,5	99.9%	99.0%	Cranioectodermal dysplasia 1, 218330

IFT140	117,6	99.9%	99.2%	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	94,5	100.0%	99.4%	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	115,8	100.0%	100.0%	?Bardet-Biedl syndrome 19, 615996
IFT43	112,4	100.0%	100.0%	?Cranioectodermal dysplasia 3, 614099 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866
IFT52	121	100.0%	99.7%	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102
IFT57	120,5	99.9%	99.3%	?Orofaciodigital syndrome XVIII, 617927
IFT74	84,7	99.1%	95.8%	?Bardet-Biedl syndrome 20, 617119
IFT80	64,9	96.7%	84.7%	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IFT81	92,3	93.6%	89.0%	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IGBP1	99,1	98.8%	93.2%	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472
IGF1	98	100.0%	99.8%	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IGF1R	114,9	100.0%	99.6%	Insulin-like growth factor I, resistance to, 270450
IGF2	119,6	100.0%	100.0%	?Growth restriction, severe, with distinctive facies, 616489
IGF2R	113,7	99.6%	97.9%	Hepatocellular carcinoma, somatic, 114550
IGFALS	108,2	100.0%	99.9%	Acid-labile subunit, deficiency of, 615961
IGFBP7	82,7	99.4%	95.3%	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis, 614224
IGHG2	26,2	72.5%	49.5%	IgG2 deficiency, selective, 0
IGHM	170	100.0%	100.0%	Agammaglobulinemia 1, 601495
IGHMBP2	108,3	99.6%	97.4%	Charcot-Marie-Tooth disease, axonal, type 2S, 616155 Neuronopathy, distal hereditary motor, type VI, 604320
IGKC	113,2	100.0%	100.0%	Kappa light chain deficiency, 614102
IGLL1	92,2	100.0%	99.6%	Agammaglobulinemia 2, 613500
IGSF1	69,9	99.2%	93.9%	Hypothyroidism, central, and testicular enlargement, 300888
IGSF3	97,5	96.3%	93.7%	?Lacrimal duct defect, 149700
IHH	171,9	100.0%	100.0%	Acrocapitofemoral dysplasia, 607778 Brachydactyly, type A1, 112500
IKBKB	110	99.3%	96.4%	Immunodeficiency 15A, 618204 Immunodeficiency 15B, 615592
IKBKG	60,1	88.1%	78.8%	Ectodermal dysplasia and immunodeficiency 1, 300291 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 Immunodeficiency 33, 300636 Immunodeficiency, isolated, 300584

				Incontinentia pigmenti, 308300 Invasive pneumococcal disease, recurrent isolated, 2, 300640
IKZF1	181,7	100.0%	100.0%	Immunodeficiency, common variable, 13, 616873
IL10RA	144,4	100.0%	100.0%	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	130,1	100.0%	99.3%	Inflammatory bowel disease 25, early onset, autosomal recessive, 612567 {Hepatitis B virus, susceptibility to}, 610424
IL11RA	131,5	100.0%	99.6%	Craniosynostosis and dental anomalies, 614188
IL12B	94,8	99.9%	97.0%	Immunodeficiency 29, mycobacteriosis, 614890
IL12RB1	112,9	98.0%	95.6%	Immunodeficiency 30, 614891
IL17F	73,1	99.2%	94.0%	?Candidiasis, familial, 6, autosomal dominant, 613956
IL17RA	149,1	100.0%	100.0%	Immunodeficiency 51, 613953
IL17RC	123,6	100.0%	100.0%	Candidiasis, familial, 9, 616445
IL17RD	134	99.9%	99.0%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
IL1RAPL1	99,7	99.9%	98.1%	Mental retardation, X-linked 21/34, 300143
IL1RN	139,3	100.0%	99.7%	Interleukin 1 receptor antagonist deficiency, 612852 {Gastric cancer risk after H. pylori infection}, 137215 {Microvascular complications of diabetes 4}, 612628
IL21	74,9	99.8%	95.0%	?Immunodeficiency, common variable, 11, 615767
IL21R	145	100.0%	100.0%	Immunodeficiency 56, 615207 [IgE, elevated level of], 147050
IL2RA	100,6	99.9%	98.7%	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367 {Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942
IL2RG	59,3	99.3%	94.0%	Combined immunodeficiency, X-linked, moderate, 312863 Severe combined immunodeficiency, X-linked, 300400
IL31RA	109,5	99.9%	99.6%	?Amyloidosis, primary localized cutaneous, 2, 613955
IL36RN	92,8	100.0%	100.0%	Psoriasis 14, pustular, 614204
IL7R	114,1	100.0%	99.9%	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
ILDR1	111,5	99.9%	98.7%	Deafness, autosomal recessive 42, 609646
IMPA1	72,1	96.7%	86.7%	Mental retardation, autosomal recessive 59, 617323
IMPAD1	170,4	100.0%	99.9%	Chondrodysplasia with joint dislocations, GPAPP type, 614078
IMPDH1	53,6	95.3%	84.1%	Leber congenital amaurosis 11, 613837 Retinitis pigmentosa 10, 180105
IMPG1	90,7	99.9%	98.6%	Macular dystrophy, vitelliform, 4, 616151
IMPG2	125,6	99.4%	97.6%	Macular dystrophy, vitelliform, 5, 616152 Retinitis pigmentosa 56, 613581
INF2	99,5	85.6%	83.5%	Charcot-Marie-Tooth disease, dominant intermediate E, 614455

				Glomerulosclerosis, focal segmental, 5, 613237
ING1	160,1	100.0%	100.0%	Squamous cell carcinoma, head and neck, somatic, 275355
INPP5E	116,8	100.0%	98.6%	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INPP5K	88,8	100.0%	99.3%	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404
INPPL1	127,9	99.8%	98.0%	Opsismodysplasia, 258480
INS	113,6	100.0%	99.9%	Diabetes mellitus, insulin-dependent, 2, 125852 Diabetes mellitus, permanent neonatal, 606176 Hyperproinsulinemia, 616214 Maturity-onset diabetes of the young, type 10, 613370
INSL3	67,1	81.3%	78.9%	Cryptorchidism, 219050
INSR	116,4	99.0%	95.1%	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968 Leprechaunism, 246200 Rabson-Mendenhall syndrome, 262190
INTU	115,4	99.8%	98.7%	?Orofaciodigital syndrome XVII, 617926 ?Short-rib thoracic dysplasia 20 with polydactyly, 617925
INVS	143,7	100.0%	99.9%	Nephronophthisis 2, infantile, 602088
IQCB1	93,3	91.6%	80.0%	Senior-Loken syndrome 5, 609254
IQCE	131,4	100.0%	98.9%	?Polydactyly, postaxial, type A7, 617642
IQSEC2	73,1	95.8%	87.9%	Mental retardation, X-linked 1/78, 309530
IRAK4	103,9	99.7%	96.5%	Invasive pneumococcal disease, recurrent isolated, 1, 610799 IRAK4 deficiency, 607676
IRF1	138,5	100.0%	99.9%	Gastric cancer, somatic, 613659 Myelodysplastic syndrome, preleukemic, 0 Myelogenous leukemia, acute, 0 Nonsmall cell lung cancer, somatic, 211980
IRF2BP2	86,8	100.0%	99.3%	?Immunodeficiency, common variable, 14, 617765
IRF2BPL	176,5	99.0%	96.7%	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088
IRF6	90,3	99.4%	95.0%	Popliteal pterygium syndrome 1, 119500 van der Woude syndrome, 119300 {Orofacial cleft 6}, 608864
IRF7	152,6	100.0%	99.8%	?Immunodeficiency 39, 616345
IRF8	111,8	99.9%	98.6%	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893 Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990
IRX5	134,9	100.0%	99.8%	Hamamy syndrome, 611174
ISCA1	67,2	93.5%	84.0%	Multiple mitochondrial dysfunctions syndrome 5, 617613

ISCA2	105,1	99.8%	95.8%	Multiple mitochondrial dysfunctions syndrome 4, 616370
ISCU	117,2	100.0%	99.9%	Myopathy with lactic acidosis, hereditary, 255125
ISG15	184,2	100.0%	100.0%	Immunodeficiency 38, 616126
ISPD	NC	NC	NC	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052
ITCH	116,3	95.4%	94.6%	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITGA2B	124,4	99.9%	98.9%	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Glanzmann thrombasthenia, 273800 Thrombocytopenia, neonatal alloimmune, BAK antigen related, 0
ITGA3	150,1	99.7%	98.0%	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748
ITGA6	138,9	99.9%	99.0%	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730
ITGA7	129,2	99.7%	98.2%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
ITGA8	115,5	100.0%	99.5%	Renal hypodysplasia/aplasia 1, 191830
ITGB2	156,8	100.0%	100.0%	Leukocyte adhesion deficiency, 116920
ITGB3	112,8	100.0%	99.8%	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Glanzmann thrombasthenia, 273800 Purpura, posttransfusion, 0 Thrombocytopenia, neonatal alloimmune, 0 {Myocardial infarction, susceptibility to}, 608446
ITGB4	152,1	99.2%	97.4%	Epidermolysis bullosa of hands and feet, 131800 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa, junctional, with pyloric atresia, 226730
ITGB6	127,5	96.7%	95.0%	Amelogenesis imperfecta, type IH, 616221
ITK	103,1	99.9%	99.1%	Lymphoproliferative syndrome 1, 613011
ITM2B	134,9	99.9%	99.7%	?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079 Dementia, familial British, 176500 Dementia, familial Danish, 117300
ITPA	130,2	100.0%	100.0%	Epileptic encephalopathy, early infantile, 35, 616647 [Inosine triphosphatase deficiency], 613850
ITPR1	131,2	100.0%	99.7%	Gillespie syndrome, 206700 Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360
ITPR2	128,9	99.9%	98.4%	?Anhidrosis, isolated, with normal sweat glands, 106190
IVD	100	100.0%	99.9%	Isovaleric acidemia, 243500
IYD	105,5	99.3%	94.7%	Thyroid dyshormonogenesis 4, 274800
JAG1	133,7	99.2%	97.1%	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Alagille syndrome 1, 118450

				Tetralogy of Fallot, 187500
JAGN1	118,5	100.0%	100.0%	Neutropenia, severe congenital, 6, autosomal recessive, 616022
JAK2	103,5	97.6%	95.0%	Erythrocytosis, somatic, 133100 Leukemia, acute myeloid, somatic, 601626 Myelofibrosis, somatic, 254450 Polycythemia vera, somatic, 263300 Thrombocythemia 3, 614521 {Budd-Chiari syndrome, somatic}, 600880
JAK3	122,1	98.8%	97.2%	SCID, autosomal recessive, T-negative/B-positive type, 600802
JAM3	126,9	100.0%	99.9%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
JPH1	168,8	100.0%	99.9%	?Charcot-Marie-Tooth disease, axonal, autosomal dominant, type 2K, 607831
JPH2	123,7	99.5%	95.5%	Cardiomyopathy, hypertrophic, 17, 613873
JPH3	196	100.0%	99.9%	Huntington disease-like 2, 606438
JUP	124,5	100.0%	99.8%	Arrhythmogenic right ventricular dysplasia 12, 611528 Naxos disease, 601214
KANK1	119,2	100.0%	99.8%	Cerebral palsy, spastic quadriplegic, 2, 612900
KANK2	163,4	100.0%	99.9%	Nephrotic syndrome, type 16, 617783 Palmoplantar keratoderma and woolly hair, 616099
KANSL1	139,1	99.8%	98.6%	Koolen-De Vries syndrome, 610443
KARS	104,1	100.0%	98.8%	?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness, autosomal recessive 89, 613916
KAT6A	148,4	100.0%	99.7%	Mental retardation, autosomal dominant 32, 616268
KAT6B	155,7	99.9%	99.1%	Genitopatellar syndrome, 606170 SBBYSS syndrome, 603736
KATNB1	154,2	100.0%	100.0%	Lissencephaly 6, with microcephaly, 616212
KBTBD13	177,3	100.0%	100.0%	Nemaline myopathy 6, autosomal dominant, 609273
KCNA1	150,1	100.0%	100.0%	Episodic ataxia/myokymia syndrome, 160120
KCNA2	126,3	100.0%	99.7%	Epileptic encephalopathy, early infantile, 32, 616366
KCNA4	123	100.0%	100.0%	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284
KCNA5	154,2	100.0%	100.0%	Atrial fibrillation, familial, 7, 612240
KCNB1	129,7	100.0%	99.7%	Epileptic encephalopathy, early infantile, 26, 616056
KCNC1	170,9	100.0%	100.0%	Epilepsy, progressive myoclonic 7, 616187
KCNC3	112,7	90.4%	72.9%	Spinocerebellar ataxia 13, 605259
KCND3	162	99.9%	99.2%	Brugada syndrome 9, 616399 Spinocerebellar ataxia 19, 607346
KCNE1	369,2	100.0%	100.0%	Jervell and Lange-Nielsen syndrome 2, 612347

				Long QT syndrome 5, 613695
KCNE2	126,7	100.0%	97.9%	Atrial fibrillation, familial, 4, 611493 Long QT syndrome 6, 613693
KCNE3	143,4	100.0%	100.0%	?Brugada syndrome 6, 613119
KCNH1	148,4	98.7%	98.3%	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500
KCNH2	106,9	98.6%	95.1%	Long QT syndrome 2, 613688 Short QT syndrome 1, 609620 {Long QT syndrome 2, acquired, susceptibility to}, 613688
KCNJ1	157,5	100.0%	100.0%	Bartter syndrome, type 2, 241200
KCNJ10	148,6	89.2%	88.1%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ11	199,7	100.0%	100.0%	Diabetes mellitus, transient neonatal, 3, 610582 Diabetes, permanent neonatal, with or without neurologic features, 606176 Hyperinsulinemic hypoglycemia, familial, 2, 601820 Maturity-onset diabetes of the young, type 13, 616329 {Diabetes mellitus, type 2, susceptibility to}, 125853
KCNJ13	138,9	100.0%	100.0%	Leber congenital amaurosis 16, 614186 Snowflake vitreoretinal degeneration, 193230
KCNJ2	154,8	100.0%	100.0%	Andersen syndrome, 170390 Atrial fibrillation, familial, 9, 613980 Short QT syndrome 3, 609622
KCNJ5	160,1	100.0%	99.8%	Hyperaldosteronism, familial, type III, 613677 Long QT syndrome 13, 613485
KCNJ6	157,2	100.0%	99.9%	Keppen-Lubinsky syndrome, 614098
KCNK3	161,7	99.8%	98.5%	Pulmonary hypertension, primary, 4, 615344
KCNK4	194,7	100.0%	99.9%	Facial dysmorphism, hypertrichosis, epilepsy, intellectual/developmental delay, and gingival overgrowth syndrome, 618381
KCNK9	171,2	100.0%	100.0%	Birk-Barel mental retardation dysmorphism syndrome, 612292
KCNMA1	102,3	94.8%	93.4%	?Cerebellar atrophy, developmental delay, and seizures, 617643 Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446
KCNN4	154,7	100.0%	99.5%	Dehydrated hereditary stomatocytosis 2, 616689
KCNQ1	135,8	97.9%	95.3%	Atrial fibrillation, familial, 3, 607554 Jervell and Lange-Nielsen syndrome, 220400 Long QT syndrome 1, 192500 Short QT syndrome 2, 609621 {Long QT syndrome 1, acquired, susceptibility to}, 192500

KCNQ1OT1	NC	NC	NC	Beckwith-Wiedemann syndrome, 130650
KCNQ2	118,3	91.5%	90.2%	Epileptic encephalopathy, early infantile, 7, 613720 Myokymia, 121200 Seizures, benign neonatal, 1, 121200
KCNQ3	110,4	99.9%	98.7%	Seizures, benign neonatal, 2, 121201
KCNQ4	165,1	99.2%	97.7%	Deafness, autosomal dominant 2A, 600101
KCNQ5	135	99.4%	97.7%	Mental retardation, autosomal dominant 46, 617601
KCNT1	131,2	96.0%	95.1%	Epilepsy, nocturnal frontal lobe, 5, 615005 Epileptic encephalopathy, early infantile, 14, 614959
KCNT2	106,2	99.5%	97.4%	?Epileptic encephalopathy, early infantile, 57, 617771
KCNV2	138,9	100.0%	100.0%	Retinal cone dystrophy 3B, 610356
KCTD1	93,9	100.0%	99.7%	Scalp-ear-nipple syndrome, 181270
KCTD17	113,4	100.0%	99.5%	Dystonia 26, myoclonic, 616398
KCTD7	154,9	95.0%	95.0%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDF1	110,3	100.0%	99.9%	?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/nail type, 617337
KDM1A	130,9	100.0%	98.8%	Cleft palate, psychomotor retardation, and distinctive facial features, 616728
KDM5B	121,6	99.0%	97.0%	Mental retardation, autosomal recessive 65, 618109
KDM5C	102,8	99.5%	97.0%	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
KDM6A	97,7	95.3%	87.8%	Kabuki syndrome 2, 300867
KDR	117,8	100.0%	99.6%	Hemangioma, capillary infantile, somatic, 602089 {Hemangioma, capillary infantile, susceptibility to}, 602089
KDSR	158,2	99.9%	99.5%	Erythrokeratodermia variabilis et progressiva 4, 617526
KERA	175,3	100.0%	100.0%	Cornea plana 2, autosomal recessive, 217300
KHDC3L	160	100.0%	99.7%	Hydatidiform mole, recurrent, 2, 614293
KIAA0556	126,6	100.0%	99.6%	Joubert syndrome 26, 616784
KIAA0586	117,7	97.0%	93.0%	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546
KIAA0753	113,2	99.9%	98.7%	?Orofaciodigital syndrome XV, 617127
KIAA1109	140,8	99.9%	98.9%	Alkuraya-Kucinskas syndrome, 617822
KIAA1161	NC	NC	NC	Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317
KIDINS220	137,5	100.0%	99.8%	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296
KIF11	92,1	97.8%	94.5%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF14	116,6	99.6%	97.9%	?Meckel syndrome 12, 616258 Microcephaly 20, primary, autosomal recessive, 617914
KIF1A	115	99.7%	97.6%	Mental retardation, autosomal dominant 9, 614255 Neuropathy, hereditary sensory, type IIC, 614213

				Spastic paraplegia 30, autosomal recessive, 610357
KIF1B	139,6	100.0%	99.6%	?Charcot-Marie-Tooth disease, type 2A1, 118210 Pheochromocytoma, 171300 {Neuroblastoma, susceptibility to, 1}, 256700
KIF1BP	161,1	96.1%	96.0%	Goldberg-Shprintzen megacolon syndrome, 609460
KIF1C	147,7	100.0%	99.4%	Spastic ataxia 2, autosomal recessive, 611302
KIF21A	120,7	99.9%	99.1%	Fibrosis of extraocular muscles, congenital, 1, 135700 Fibrosis of extraocular muscles, congenital, 3B, 135700
KIF22	173,8	100.0%	99.9%	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546
KIF2A	105,7	99.6%	96.8%	Cortical dysplasia, complex, with other brain malformations 3, 615411
KIF4A	77,4	98.1%	91.5%	?Mental retardation, X-linked 100, 300923
KIF5A	116	100.0%	99.9%	Myoclonus, intractable, neonatal, 617235 Spastic paraplegia 10, autosomal dominant, 604187 {Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921
KIF5C	109,9	99.9%	99.0%	Cortical dysplasia, complex, with other brain malformations 2, 615282
KIF7	105,2	98.2%	93.5%	?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990
KISS1	79,5	100.0%	98.1%	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842
KISS1R	156,4	100.0%	100.0%	?Precocious puberty, central, 1, 176400 Hypogonadotropic hypogonadism 8 with or without anosmia, 614837
KIT	136,2	100.0%	99.6%	Gastrointestinal stromal tumor, familial, 606764 Germ cell tumors, somatic, 273300 Leukemia, acute myeloid, 601626 Mastocytosis, cutaneous, 154800 Mastocytosis, systemic, somatic, 154800 Piebaldism, 172800
KITLG	83,3	99.6%	97.2%	Deafness, autosomal dominant 69, unilateral or asymmetric, 616697 Hyperpigmentation with or without hypopigmentation, 145250 [Skin/hair/eye pigmentation 7, blond/brown hair], 611664
KIZ	145	99.9%	98.1%	Retinitis pigmentosa 69, 615780
KL	171,8	99.2%	98.1%	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994
KLC2	127	99.9%	99.2%	Spastic paraplegia, optic atrophy, and neuropathy, 609541
KLF1	115,3	100.0%	99.9%	Blood group--Lutheran inhibitor, 111150 Dyserythropoietic anemia, congenital, type IV, 613673 [Hereditary persistence of fetal hemoglobin], 613566

KLF11	162,4	100.0%	99.8%	Maturity-onset diabetes of the young, type VII, 610508
KLF6	152,4	100.0%	100.0%	Gastric cancer, somatic, 613659 Prostate cancer, somatic, 176807
KLHL10	139,1	100.0%	99.9%	Spermatogenic failure 11, 615081
KLHL15	140,4	100.0%	99.7%	Mental retardation, X-linked 103, 300982
KLHL24	172,7	100.0%	100.0%	Epidermolysis bullosa simplex, generalized, with scarring and hair loss, 617294
KLHL3	105,6	99.9%	97.7%	Pseudohypoaldosteronism, type IID, 614495
KLHL40	130,6	100.0%	100.0%	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	172,8	100.0%	99.8%	Nemaline myopathy 9, 615731
KLHL7	116,2	100.0%	99.5%	Cold-induced sweating syndrome 3, 617055 Retinitis pigmentosa 42, 612943
KLK4	164,3	100.0%	100.0%	Amelogenesis imperfecta, type IIA1, 204700
KLKB1	132,3	100.0%	99.5%	Fletcher factor (prekallikrein) deficiency, 612423
KLLN	152,3	100.0%	100.0%	Cowden syndrome 4, 615107
KMT2A	133	100.0%	99.9%	Leukemia, myeloid/lymphoid or mixed-lineage, 159555 Wiedemann-Steiner syndrome, 605130
KMT2B	141,1	96.9%	93.5%	Dystonia 28, childhood-onset, 617284
KMT2C	138,9	91.8%	90.4%	Kleefstra syndrome 2, 617768
KMT2D	136,2	100.0%	99.7%	Kabuki syndrome 1, 147920
KMT5B	165,5	100.0%	99.6%	Mental retardation, autosomal dominant 51, 617788
KNL1	105,5	99.0%	97.2%	Microcephaly 4, primary, autosomal recessive, 604321
KPTN	145,7	100.0%	100.0%	Mental retardation, autosomal recessive 41, 615637
KRAS	67,2	99.4%	97.3%	Arteriovenous malformation of the brain, somatic, 108010 Bladder cancer, somatic, 109800 Breast cancer, somatic, 114480 Cardiofaciocutaneous syndrome 2, 615278 Gastric cancer, somatic, 137215 Leukemia, acute myeloid, 601626 Lung cancer, somatic, 211980 Noonan syndrome 3, 609942 Pancreatic carcinoma, somatic, 260350 RAS-associated autoimmune leukoproliferative disorder, 614470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200
KREMEN1	143,4	99.7%	97.3%	Ectodermal dysplasia 13, hair/tooth type, 617392
KRIT1	98,6	99.8%	98.8%	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	99.9%	98.8%	Epidermolytic hyperkeratosis, 113800 Ichthyosis histrix, Curth-Macklin type, 146590 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602 Keratosis palmoplantaris striata III, 607654 Palmoplantar keratoderma, epidermolytic, 144200 Palmoplantar keratoderma, nonepidermolytic, 600962
KRT10	128,8	99.9%	98.6%	Epidermolytic hyperkeratosis, 113800 Ichthyosis with confetti, 609165 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602
KRT12	134,2	99.7%	97.3%	Meesmann corneal dystrophy, 122100
KRT13	120,3	100.0%	99.3%	White sponge nevus 2, 615785
KRT14	42,6	89.0%	80.0%	Dermatopathia pigmentosa reticularis, 125595 Epidermolysis bullosa simplex, Dowling-Meara type, 131760 Epidermolysis bullosa simplex, Koebner type, 131900 Epidermolysis bullosa simplex, recessive 1, 601001 Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 Naegeli-Franceschetti-Jadassohn syndrome, 161000
KRT16	34,9	75.2%	53.2%	Pachyonychia congenita 1, 167200 Palmoplantar keratoderma, nonepidermolytic, focal, 613000
KRT17	17,6	46.8%	28.0%	Pachyonychia congenita 2, 167210 Steatocystoma multiplex, 184500
KRT18	34,2	89.8%	71.1%	Cirrhosis, cryptogenic, 215600 {Cirrhosis, noncryptogenic, susceptibility to}, 215600
KRT2	134,6	100.0%	99.5%	Ichthyosis bullosa of Siemens, 146800
KRT25	126,7	100.0%	100.0%	Woolly hair, autosomal recessive 3, 616760
KRT3	113,3	100.0%	99.7%	Meesmann corneal dystrophy, 122100
KRT4	121,3	100.0%	99.5%	White sponge nevus 1, 193900
KRT5	110,6	100.0%	100.0%	Dowling-Degos disease 1, 179850 Epidermolysis bullosa simplex, Dowling-Meara type, 131760 Epidermolysis bullosa simplex, Koebner type, 131900 Epidermolysis bullosa simplex, recessive 1, 601001 Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 Epidermolysis bullosa simplex-MCR, 609352 Epidermolysis bullosa simplex-MP, 131960
KRT6A	121,7	96.9%	89.2%	Pachyonychia congenita 3, 615726

KRT6B	118,2	98.2%	91.8%	Pachyonychia congenita 4, 615728
KRT6C	105,5	88.6%	79.8%	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735
KRT71	143,1	100.0%	99.9%	?Hypotrichosis 13, 615896
KRT74	138,6	100.0%	99.6%	?Ectodermal dysplasia 7, hair/nail type, 614929 ?Hypotrichosis 3, 613981 Woolly hair, autosomal dominant, 194300
KRT8	37	89.5%	67.8%	Cirrhosis, cryptogenic, 215600 {Cirrhosis, noncryptogenic, susceptibility to}, 215600
KRT81	83,7	99.9%	97.8%	Monilethrix, 158000
KRT83	67,6	98.6%	90.0%	Erythrokeratodermia variabilis et progressiva 5, 617756 Monilethrix, 158000
KRT85	101,3	99.0%	95.2%	Ectodermal dysplasia 4, hair/nail type, 602032
KRT86	85,3	100.0%	97.7%	Monilethrix, 158000
KRT9	68,1	99.4%	96.3%	Palmoplantar keratoderma, epidermolytic, 144200
KY	112,6	100.0%	99.8%	Myopathy, myofibrillar, 7, 617114
KYNU	104,7	98.8%	93.8%	?Hydroxykynureninuria, 236800 Vertebral, cardiac, renal, and limb defects syndrome 2, 617661
L1CAM	126,6	99.9%	98.6%	Corpus callosum, partial agenesis of, 304100 CRASH syndrome, 303350 Hydrocephalus due to aqueductal stenosis, 307000 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 Hydrocephalus with Hirschsprung disease, 307000 MASA syndrome, 303350
L2HGDH	124	99.0%	96.7%	L-2-hydroxyglutaric aciduria, 236792
LAGE3	68,3	98.1%	90.5%	Galloway-Mowat syndrome 2, X-linked, 301006
LAMA1	116	100.0%	99.5%	Poretti-Boltshauser syndrome, 615960
LAMA2	130,6	100.0%	99.5%	Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855 Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138
LAMA3	125,2	99.9%	99.6%	Epidermolysis bullosa, generalized atrophic benign, 226650 Epidermolysis bullosa, junctional, Herlitz type, 226700 Laryngoonychocutaneous syndrome, 245660
LAMA4	118,2	100.0%	99.7%	Cardiomyopathy, dilated, 1JJ, 615235
LAMB1	142,9	100.0%	99.7%	Lissencephaly 5, 615191
LAMB2	166,5	100.0%	99.6%	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 Pierson syndrome, 609049
LAMB3	116,9	100.0%	99.4%	Amelogenesis imperfecta, type IA, 104530 Epidermolysis bullosa, junctional, Herlitz type, 226700

				Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LAMC2	100,5	99.7%	98.3%	Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LAMC3	148,1	99.7%	98.6%	Cortical malformations, occipital, 614115
LAMP2	92,3	97.9%	92.8%	Danon disease, 300257
LAMTOR2	172,2	100.0%	100.0%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LARGE1	115,2	100.0%	99.7%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840
LARP7	75,5	88.8%	75.2%	Alazami syndrome, 615071
LARS	131,5	99.7%	98.0%	?Infantile liver failure syndrome 1, 615438
LARS2	122,8	100.0%	100.0%	?Hydrops, lactic acidosis, and sideroblastic anemia, 617021 Perrault syndrome 4, 615300
LAS1L	78,6	99.6%	95.7%	Wilson-Turner syndrome, 309585
LAT	114,4	100.0%	99.3%	Immunodeficiency 52, 617514
LBR	103	98.3%	91.5%	?Reynolds syndrome, 613471 Greenberg skeletal dysplasia, 215140 Pelger-Huet anomaly, 169400 Pelger-Huet anomaly with mild skeletal anomalies, 618019
LCA5	139,9	99.8%	98.9%	Leber congenital amaurosis 5, 604537
LCAT	140,7	99.4%	95.1%	Fish-eye disease, 136120 Norum disease, 245900
LCK	148,6	99.3%	97.3%	?Immunodeficiency 22, 615758
LCT	118,8	99.8%	97.9%	Lactase deficiency, congenital, 223000
LDB3	147,1	96.0%	94.7%	Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 Cardiomyopathy, hypertrophic, 24, 601493 Left ventricular noncompaction 3, 601493 Myopathy, myofibrillar, 4, 609452
LDHA	55,6	96.6%	88.0%	Glycogen storage disease XI, 612933
LDHD	143,8	100.0%	100.0%	D-lactic aciduria, 245450
LDLR	148,1	100.0%	98.9%	Hypercholesterolemia, familial, 143890 LDL cholesterol level QTL2, 143890
LDLRAP1	149	99.9%	99.1%	Hypercholesterolemia, familial, autosomal recessive, 603813
LEF1	106	100.0%	99.8%	Sebaceous tumors, somatic, 0
LEMD2	103,2	100.0%	99.6%	Cataract 46, juvenile-onset, 212500
LEMD3	122,5	99.8%	98.4%	Buschke-Ollendorff syndrome, 166700 Osteopoikilosis with or without melorheostosis, 166700

LEP	174,7	100.0%	99.9%	Obesity, morbid, due to leptin deficiency, 614962
LEPR	106,5	94.2%	91.4%	Obesity, morbid, due to leptin receptor deficiency, 614963
LFNG	117,6	92.8%	87.7%	Spondylocostal dysostosis 3, autosomal recessive, 609813
LGI1	133,3	98.3%	97.2%	Epilepsy, familial temporal lobe, 1, 600512
LGI4	99,9	99.4%	96.7%	Arthrogryposis multiplex congenita, neurogenic, with myelin defect, 617468
LHB	23,5	92.3%	52.6%	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300
LHCGR	137	97.9%	94.1%	Leydig cell adenoma, somatic, with precocious puberty, 176410 Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320 Leydig cell hypoplasia with pseudohermaphroditism, 238320 Luteinizing hormone resistance, female, 238320 Precocious puberty, male, 176410
LHFPL5	207,7	100.0%	100.0%	Deafness, autosomal recessive 67, 610265
LHX3	116,2	96.6%	96.4%	Pituitary hormone deficiency, combined, 3, 221750
LHX4	131,2	100.0%	100.0%	Pituitary hormone deficiency, combined, 4, 262700
LIAS	125,3	99.9%	98.7%	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIFR	110,3	99.7%	97.4%	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LIG4	173,4	100.0%	99.8%	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500
LIM2	112,9	100.0%	99.5%	Cataract 19, multiple types, 615277
LIMS2	118,5	95.7%	93.2%	?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827
LINGO1	201,9	100.0%	100.0%	Mental retardation, autosomal recessive 64, 618103
LINS1	133,6	100.0%	99.3%	Mental retardation, autosomal recessive 27, 614340
LIPA	105	96.5%	94.4%	Cholesteryl ester storage disease, 278000 Wolman disease, 278000
LIPC	98,5	100.0%	99.1%	Hepatic lipase deficiency, 614025 [High density lipoprotein cholesterol level QTL 12], 612797 {Diabetes mellitus, noninsulin-dependent}, 125853
LIPE	121,8	100.0%	99.5%	Lipodystrophy, familial partial, type 6, 615980
LIPH	120,2	100.0%	99.7%	Hypotrichosis 7, 604379 Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379
LIPN	113,9	100.0%	99.1%	Ichthyosis, congenital, autosomal recessive 8, 613943
LIPT1	203,2	100.0%	99.9%	Lipoyltransferase 1 deficiency, 616299
LIPT2	91,2	99.9%	99.3%	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668
LITAF	102,2	95.2%	91.1%	Charcot-Marie-Tooth disease, type 1C, 601098
LMAN1	144,3	99.8%	99.4%	Combined factor V and VIII deficiency, 227300
LMAN2L	109,3	100.0%	99.2%	?Mental retardation, autosomal recessive, 52, 616887

LMBR1	122,9	98.4%	96.3%	Acheiropody, 200500 Hypoplastic or aplastic tibia with polydactyly, 188740 Laurin-Sandrow syndrome, 135750 Polydactyly, preaxial type II, 174500 Syndactyly, type IV, 186200 Triphalangeal thumb, type I, 174500 Triphalangeal thumb-polysyndactyly syndrome, 174500
LMBRD1	100,1	98.9%	94.1%	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LMF1	136,8	100.0%	99.9%	Lipase deficiency, combined, 246650
LMNA	104,7	97.7%	91.9%	Cardiomyopathy, dilated, 1A, 115200 Charcot-Marie-Tooth disease, type 2B1, 605588 Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 Heart-hand syndrome, Slovenian type, 610140 Hutchinson-Gilford progeria, 176670 Lipodystrophy, familial partial, type 2, 151660 Malouf syndrome, 212112 Mandibuloacral dysplasia, 248370 Muscular dystrophy, congenital, 613205 Restrictive dermopathy, lethal, 275210
LMNB1	118,4	100.0%	99.7%	Leukodystrophy, adult-onset, autosomal dominant, 169500
LMNB2	140,4	99.2%	97.4%	?Epilepsy, progressive myoclonic, 9, 616540 {Lipodystrophy, partial, acquired, susceptibility to}, 608709
LMOD3	128,6	100.0%	99.8%	Nemaline myopathy 10, 616165
LMX1B	146,6	99.9%	98.5%	Nail-patella syndrome, 161200
LNPK	89,1	98.4%	92.3%	Neurodevelopmental disorder with epilepsy and hypoplasia of the corpus callosum, 618090
LONP1	148	100.0%	100.0%	CODAS syndrome, 600373
LOR	39,8	100.0%	93.8%	Vohwinkel syndrome with ichthyosis, 604117
LOX	158,5	99.9%	99.6%	Aortic aneurysm, familial thoracic 10, 617168
LOXHD1	113,3	99.9%	99.5%	Deafness, autosomal recessive 77, 613079
LPAR6	100,3	99.8%	98.4%	Hypotrichosis 8, 278150 Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150
LPIN1	123,4	99.1%	96.4%	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	97,8	100.0%	99.6%	Majeed syndrome, 609628
LPL	128,2	100.0%	99.9%	Combined hyperlipidemia, familial, 144250 Lipoprotein lipase deficiency, 238600 [High density lipoprotein cholesterol level QTL 11], 0

LPP	104,5	100.0%	99.9%	Leukemia, acute myeloid, 601626 Lipoma, 0
LRAT	240,7	100.0%	100.0%	Leber congenital amaurosis 14, 613341 Retinal dystrophy, early-onset severe, 613341 Retinitis pigmentosa, juvenile, 613341
LRBA	129,8	100.0%	99.6%	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRIG2	126,8	99.8%	99.1%	Urofacial syndrome 2, 615112
LRIT3	108	94.3%	93.1%	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRMDA	114,9	99.4%	97.6%	Albinism, oculocutaneous, type VII, 615179
LRP1	172,3	99.8%	99.3%	?Keratosis pilaris atrophicans, 604093
LRP2	139,2	100.0%	99.9%	Donnai-Barrow syndrome, 222448
LRP4	128	99.7%	99.0%	?Myasthenic syndrome, congenital, 17, 616304 Cenani-Lenz syndactyly syndrome, 212780 Sclerosteosis 2, 614305
LRP5	168,1	99.8%	98.7%	Exudative vitreoretinopathy 4, 601813 Hyperostosis, endosteal, 144750 Osteopetrosis, autosomal dominant 1, 607634 Osteoporosis-pseudoglioma syndrome, 259770 Osteosclerosis, 144750 Polycystic liver disease 4 with or without kidney cysts, 617875 van Buchem disease, type 2, 607636 [Bone mineral density variability 1], 601884 {Osteoporosis}, 166710
LRP6	136,8	99.9%	99.2%	Tooth agenesis, selective, 7, 616724 {Coronary artery disease, autosomal dominant, 2}, 610947
LRPAP1	141	100.0%	99.6%	Myopia 23, autosomal recessive, 615431
LRPPRC	129,3	100.0%	99.6%	Leigh syndrome, French-Canadian type, 220111
LRRC56	130,7	100.0%	99.2%	Ciliary dyskinesia, primary, 39, 618254
LRRC6	139,3	99.8%	97.3%	Ciliary dyskinesia, primary, 19, 614935
LRRC8A	223,3	100.0%	100.0%	?Agammaglobulinemia 5, 613506
LRSAM1	135,4	100.0%	99.9%	Charcot-Marie-Tooth disease, axonal, type 2P, 614436
LRTOMT	114,4	99.1%	94.4%	Deafness, autosomal recessive 63, 611451
LSS	127,6	100.0%	99.7%	Cataract 44, 616509 Hypotrichosis 14, 618275
LTBP2	112,9	99.9%	99.3%	?Weill-Marchesani syndrome 3, recessive, 614819 Glaucoma 3, primary congenital, D, 613086 Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750

LTBP3	147,5	100.0%	99.6%	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
LTBP4	148	100.0%	99.4%	Cutis laxa, autosomal recessive, type IC, 613177
LYRM4	77,5	67.4%	62.4%	?Combined oxidative phosphorylation deficiency 19, 615595
LYRM7	61,9	98.7%	91.3%	Mitochondrial complex III deficiency, nuclear type 8, 615838
LYST	136,3	99.4%	97.8%	Chediak-Higashi syndrome, 214500
LYZ	143	100.0%	100.0%	Amyloidosis, renal, 105200
LZTFL1	117	99.8%	99.2%	Bardet-Biedl syndrome 17, 615994
LZTR1	143,6	100.0%	99.7%	Noonan syndrome 10, 616564 Noonan syndrome 2, 605275 {Schwannomatosis-2, susceptibility to}, 615670
LZTS1	143,2	100.0%	100.0%	Esophageal squamous cell carcinoma, somatic, 133239
MAB21L2	237,8	100.0%	100.0%	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877
MACF1	128,6	99.9%	99.2%	Lissencephaly 9 with complex brainstem malformation, 618325
MAD1L1	106,6	99.8%	97.8%	Lymphoma, somatic, 0 Prostate cancer, somatic, 176807
MAD2L2	139,1	100.0%	99.8%	?Fanconi anemia, complementation group V, 617243
MAF	88,9	87.2%	83.0%	Ayme-Gripp syndrome, 601088 Cataract 21, multiple types, 610202
MAFA	58,9	99.8%	93.9%	Insulinomatosis and diabetes mellitus, 147630
MAFB	124	100.0%	99.9%	Duane retraction syndrome 3, 617041 Multicentric carpotarsal osteolysis syndrome, 166300
MAG	160,7	100.0%	100.0%	Spastic paraplegia 75, autosomal recessive, 616680
MAGED2	86,7	99.5%	97.6%	Bartter syndrome, type 5, antenatal, transient, 300971
MAGEL2	120,4	97.9%	93.1%	Schaaf-Yang syndrome, 615547
MAGI2	90,4	94.6%	91.7%	Nephrotic syndrome, type 15, 617609
MAGT1	96,8	98.2%	96.3%	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853
MAK	131,8	99.5%	97.4%	Retinitis pigmentosa 62, 614181
MALT1	129,4	93.0%	89.4%	Immunodeficiency 12, 615468
MAML2	110,9	100.0%	100.0%	Mucoepidermoid salivary gland carcinoma, 0
MAMLD1	125	99.8%	98.2%	Hypospadias 2, X-linked, 300758
MAN1B1	125,5	100.0%	99.8%	Mental retardation, autosomal recessive 15, 614202
MAN2B1	128,6	99.9%	98.6%	Mannosidosis, alpha-, types I and II, 248500
MANBA	118,3	99.5%	97.5%	Mannosidosis, beta, 248510
MAOA	98,9	100.0%	99.0%	Brunner syndrome, 300615 {Antisocial behavior}, 300615

MAP2K1	92,3	99.5%	96.3%	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	124,2	98.5%	94.1%	Cardiofaciocutaneous syndrome 4, 615280
MAP3K1	144	99.4%	97.0%	46XY sex reversal 6, 613762
MAP3K20	109,8	99.9%	98.9%	Centronuclear myopathy 6 with fiber-type disproportion, 617760 Split-foot malformation with mesoaxial polydactyly, 616890
MAP3K7	118,4	99.9%	99.3%	Cardiospondylocarpofacial syndrome, 157800 Frontometaphyseal dysplasia 2, 617137
MAP3K8	132,7	100.0%	99.9%	Lung cancer, somatic, 211980
MAPKAPK3	87,6	100.0%	99.6%	?Macular dystrophy, patterned, 3, 617111
MAPKBP1	132,5	100.0%	100.0%	Nephronophthisis 20, 617271
MAPRE2	158,4	99.7%	98.1%	Symmetric circumferential skin creases, congenital, 2, 616734
MAPT	151,6	99.9%	99.6%	Dementia, frontotemporal, with or without parkinsonism, 600274 Pick disease, 172700 Supranuclear palsy, progressive, 601104 Supranuclear palsy, progressive atypical, 260540 {Parkinson disease, susceptibility to}, 168600
MARK3	122,8	99.7%	97.6%	?Visual impairment and progressive phthisis bulbi, 618283
MARS	100,4	99.9%	98.0%	Charcot-Marie-Tooth disease, axonal, type 2U, 616280 Interstitial lung and liver disease, 615486
MARS2	178,4	100.0%	100.0%	?Combined oxidative phosphorylation deficiency 25, 616430 Spastic ataxia 3, autosomal recessive, 611390
MARVELD2	139	98.8%	95.9%	Deafness, autosomal recessive 49, 610153
MASP1	131,1	100.0%	99.3%	3MC syndrome 1, 257920
MASP2	121,9	100.0%	99.5%	MASP2 deficiency, 613791
MAST1	168,6	100.0%	99.8%	Mega-corpus-callosum syndrome with cerebellar hypoplasia and cortical malformations, 618273
MAT1A	144,1	99.8%	98.4%	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MATN3	99,6	86.5%	84.5%	?Spondyloepimetaphyseal dysplasia, 608728 Epiphyseal dysplasia, multiple, 5, 607078 {Osteoarthritis susceptibility 2}, 140600
MATR3	86,6	96.9%	92.7%	Amyotrophic lateral sclerosis 21, 606070
MBD5	147,7	99.9%	99.8%	Mental retardation, autosomal dominant 1, 156200
MBOAT7	110	100.0%	99.7%	Mental retardation, autosomal recessive 57, 617188
MBTPS1	112,5	99.7%	98.2%	?Spondyloepiphyseal dysplasia, Kondo-Fu type, 618392
MBTPS2	111,2	99.9%	98.6%	?Olmsted syndrome, X-linked, 300918

				IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800 Osteogenesis imperfecta, type XIX, 301014
MC2R	148,3	100.0%	99.2%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MC4R	186,5	100.0%	100.0%	Obesity (BMIQ20), 618406 {Obesity, resistence to (BMIQ20)}, 618306
MCC	120	100.0%	99.5%	Colorectal cancer, somatic, 114500
MCCC1	137,6	100.0%	99.4%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	119	100.0%	99.7%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	121,1	100.0%	99.9%	Methylmalonyl-CoA epimerase deficiency, 251120
MCFD2	91,4	97.6%	91.0%	Factor V and factor VIII, combined deficiency of, 613625
MCM2	151,9	100.0%	100.0%	?Deafness, autosomal dominant 70, 616968
MCM3AP	130,4	99.9%	99.2%	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124
MCM4	133,1	100.0%	99.5%	Immunodeficiency 54, 609981
MCM5	122,5	100.0%	100.0%	?Meier-Gorlin syndrome 8, 617564
MCM6	128,6	100.0%	100.0%	Lactase persistence/nonpersistence, 223100
MCM8	123,3	99.9%	99.1%	?Premature ovarian failure 10, 612885
MCM9	128,7	99.9%	99.7%	Ovarian dysgenesis 4, 616185
MCOLN1	157,1	99.9%	99.0%	Mucolipidosis IV, 252650
MCPH1	133,1	99.9%	98.5%	Microcephaly 1, primary, autosomal recessive, 251200
MDH2	109,4	98.0%	97.9%	Epileptic encephalopathy, early infantile, 51, 617339
MECOM	131,2	100.0%	99.6%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MECP2	124,8	100.0%	98.5%	Encephalopathy, neonatal severe, 300673 Mental retardation, X-linked syndromic, Lubs type, 300260 Mental retardation, X-linked, syndromic 13, 300055 Rett syndrome, 312750 Rett syndrome, atypical, 312750 Rett syndrome, preserved speech variant, 312750 {Autism susceptibility, X-linked 3}, 300496
MECR	108,2	100.0%	99.7%	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MED12	85,1	99.5%	95.5%	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Opitz-Kaveggia syndrome, 305450
MED13L	108,5	99.9%	99.6%	Mental retardation and distinctive facial features with or without cardiac defects, 616789 Transposition of the great arteries, dextro-looped 1, 608808
MED17	132,4	97.5%	94.7%	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668

MED23	133,6	99.8%	98.4%	Mental retardation, autosomal recessive 18, 614249
MED25	132,7	100.0%	99.7%	?Charcot-Marie-Tooth disease, type 2B2, 605589 Basel-Vanagait-Smirin-Yosef syndrome, 616449
MEF2C	127,9	99.4%	95.5%	Chromosome 5q14.3 deletion syndrome, 613443 Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
MEFV	126,8	98.6%	96.5%	Familial Mediterranean fever, AD, 134610 Familial Mediterranean fever, AR, 249100
MEGF10	125,9	100.0%	99.8%	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399
MEGF8	144	100.0%	99.5%	Carpenter syndrome 2, 614976
MEIOB	103,3	99.7%	98.3%	?Spermatogenic failure 22, 617706
MEIS2	123,6	100.0%	99.8%	Cleft palate, cardiac defects, and mental retardation, 600987
MEN1	132	100.0%	99.5%	Adrenal adenoma, somatic, 0 Angiofibroma, somatic, 0 Carcinoid tumor of lung, 0 Lipoma, somatic, 0 Multiple endocrine neoplasia 1, 131100 Parathyroid adenoma, somatic, 0
MEOX1	105	99.9%	97.4%	Klippel-Feil syndrome 2, 214300
MERTK	128,2	99.4%	98.7%	Retinitis pigmentosa 38, 613862
MESP2	128	97.0%	94.9%	Spondylocostal dysostosis 2, autosomal recessive, 608681
MET	151,3	99.9%	99.3%	?Deafness, autosomal recessive 97, 616705 Hepatocellular carcinoma, childhood type, somatic, 114550 Renal cell carcinoma, papillary, 1, familial and somatic, 605074 {Osteofibrous dysplasia, susceptibility to}, 607278
METTL23	116,9	100.0%	100.0%	Mental retardation, autosomal recessive 44, 615942
MFAP5	106,8	99.6%	94.8%	Aortic aneurysm, familial thoracic 9, 616166
MFF	86,2	93.7%	89.6%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFN2	122,8	100.0%	99.9%	Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260 Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 Hereditary motor and sensory neuropathy VIA, 601152
MFRP	128,1	100.0%	100.0%	Microphthalmia, isolated 5, 611040 Nanophthalmos 2, 609549
MFSD2A	114,3	100.0%	99.3%	Microcephaly 15, primary, autosomal recessive, 616486
MFSD8	121,3	100.0%	99.6%	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170
MGAT2	145,4	100.0%	100.0%	Congenital disorder of glycosylation, type IIa, 212066

MGME1	142,4	100.0%	99.9%	Mitochondrial DNA depletion syndrome 11, 615084
MGP	134,2	98.7%	94.6%	Keutel syndrome, 245150
MIB1	127,1	100.0%	99.9%	Left ventricular noncompaction 7, 615092
MICU1	103,3	98.8%	96.5%	Myopathy with extrapyramidal signs, 615673
MID1	124,1	99.8%	97.4%	Opitz GBBB syndrome, type I, 300000
MID2	108,7	99.5%	97.2%	?Mental retardation, X-linked 101, 300928
MINPP1	163,8	100.0%	99.5%	{Thyroid carcinoma, follicular}, 188470
MIP	117,1	99.7%	97.0%	Cataract 15, multiple types, 615274
MIPEP	99,3	99.4%	96.6%	Combined oxidative phosphorylation deficiency 31, 617228
MIR17HG	NC	NC	NC	Feingold syndrome 2, 614326
MIR184	NC	NC	NC	EDICT syndrome, 614303
MIR204	NC	NC	NC	?Retinal dystrophy and iris coloboma with or without cataract, 616722
MIR96	NC	NC	NC	Deafness, autosomal dominant 50, 613074
MITF	141,1	100.0%	99.8%	COMMAD syndrome, 617306 Tietz albinism-deafness syndrome, 103500 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456
MKKS	155,7	83.2%	83.2%	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKL1	NC	NC	NC	Megakaryoblastic leukemia, acute, 0
MKRN3	136	100.0%	100.0%	Precocious puberty, central, 2, 615346
MKS1	92,4	99.6%	97.8%	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000
MLC1	96,7	100.0%	99.9%	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MLH1	139,2	99.9%	99.3%	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MLH3	141,5	100.0%	99.9%	Colorectal cancer, hereditary nonpolyposis, type 7, 614385 Colorectal cancer, somatic, 114500 {Endometrial cancer, susceptibility to}, 608089
MLLT10	123,9	96.4%	94.2%	Leukemia, acute myeloid, 601626
MLPH	97,4	99.7%	97.2%	Griscelli syndrome, type 3, 609227
MLYCD	95,7	99.4%	96.5%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	168,5	100.0%	100.0%	Methylmalonic aciduria, vitamin B12-responsive, 251100

MMAB	94,6	100.0%	99.7%	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110
MMACHC	196	100.0%	100.0%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	81,2	92.7%	79.5%	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410
MME	112,7	99.7%	98.1%	?Spinocerebellar ataxia 43, 617018 Charcot-Marie-Tooth disease, axonal, type 2T, 617017
MMP1	133,2	99.8%	99.2%	COPD, rate of decline of lung function in, 606963 {Epidermolysis bullosa dystrophica, autosomal recessive, modifier of}, 226600
MMP13	112,5	93.4%	92.1%	Metaphyseal anadysplasia 1, 602111 Metaphyseal dysplasia, Spahr type, 250400 Spondyloepimetaphyseal dysplasia, Missouri type, 602111
MMP14	148	100.0%	99.7%	?Winchester syndrome, 277950
MMP19	118,1	99.9%	98.5%	Cavitary optic disc anomalies, 611543
MMP2	154,2	100.0%	100.0%	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP20	90,8	99.8%	97.6%	Amelogenesis imperfecta, type IIA2, 612529
MMP21	94,9	100.0%	98.0%	Heterotaxy, visceral, 7, autosomal, 616749
MMP9	143,9	100.0%	99.1%	Metaphyseal anadysplasia 2, 613073
MN1	143,4	100.0%	99.9%	Meningioma, 607174
MNX1	54,8	78.6%	70.8%	Currarino syndrome, 176450
MOCOS	147,2	99.9%	99.1%	Xanthinuria, type II, 603592
MOCS1	91,2	98.8%	95.7%	Molybdenum cofactor deficiency A, 252150
MOCS2	137,7	99.6%	99.5%	Molybdenum cofactor deficiency B, 252160
MOG	96,6	99.9%	99.3%	?Narcolepsy 7, 614250
MOGS	141	100.0%	100.0%	Congenital disorder of glycosylation, type IIb, 606056
MORC2	123,8	100.0%	99.5%	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688
MPC1	153,1	100.0%	99.6%	Mitochondrial pyruvate carrier deficiency, 614741
MPDU1	102,4	100.0%	99.6%	Congenital disorder of glycosylation, type If, 609180
MPDZ	128,3	99.6%	98.2%	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219
MPI	110,1	100.0%	99.9%	Congenital disorder of glycosylation, type Ib, 602579
MPIG6B	119	100.0%	99.8%	?Thrombocytopenia, anemia, and myelofibrosis, 617441
MPL	125,8	100.0%	99.8%	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498
MPLKIP	104,3	100.0%	99.9%	Trichothiodystrophy 4, nonphotosensitive, 234050

MPO	153,5	99.9%	99.4%	Myeloperoxidase deficiency, 254600 {Alzheimer disease, susceptibility to}, 104300 {Lung cancer, protection against, in smokers}, 0
MPV17	88,6	100.0%	98.9%	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MPZ	125	100.0%	98.9%	Charcot-Marie-Tooth disease, dominant intermediate D, 607791 Charcot-Marie-Tooth disease, type 1B, 118200 Charcot-Marie-Tooth disease, type 2I, 607677 Charcot-Marie-Tooth disease, type 2J, 607736 Dejerine-Sottas disease, 145900 Hypomyelinating neuropathy, congenital, 2, 618184 Roussy-Levy syndrome, 180800
MPZL2	91,7	100.0%	99.8%	Deafness, autosomal recessive 111, 618145
MRAP	163,4	100.0%	100.0%	Glucocorticoid deficiency 2, 607398
MRE11	49,7	97.3%	86.0%	Ataxia-telangiectasia-like disorder 1, 604391
MRPL3	63,4	93.2%	81.4%	Combined oxidative phosphorylation deficiency 9, 614582
MRPL44	126,3	100.0%	99.7%	?Combined oxidative phosphorylation deficiency 16, 615395
MRPS14	168,7	100.0%	100.0%	?Combined oxidative phosphorylation deficiency 38, 618378
MRPS16	127	99.9%	98.5%	Combined oxidative phosphorylation deficiency 2, 610498
MRPS2	158,6	100.0%	99.9%	Combined oxidative phosphorylation deficiency 36, 617950
MRPS22	134,6	99.9%	98.7%	Combined oxidative phosphorylation deficiency 5, 611719 Ovarian dysgenesis 7, 618117
MRPS34	169	100.0%	99.9%	Combined oxidative phosphorylation deficiency 32, 617664
MRPS7	153,2	100.0%	100.0%	?Combined oxidative phosphorylation deficiency 34, 617872
MS4A1	127,6	99.9%	98.8%	Immunodeficiency, common variable, 5, 613495
MSH2	111,7	99.4%	96.4%	Colorectal cancer, hereditary nonpolyposis, type 1, 120435 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MSH3	139,6	99.8%	99.2%	Endometrial carcinoma, somatic, 608089 Familial adenomatous polyposis 4, 617100
MSH5	104,2	100.0%	99.8%	?Premature ovarian failure 13, 617442
MSH6	165,1	100.0%	100.0%	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 Mismatch repair cancer syndrome, 276300 {Endometrial cancer, familial}, 608089
MSMO1	51,6	95.8%	88.5%	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MSN	66,9	97.5%	91.7%	Immunodeficiency 50, 300988
MSR1	144	100.0%	99.3%	Barrett esophagus/esophageal adenocarcinoma, 614266

MSRB3	131,9	99.9%	99.6%	Deafness, autosomal recessive 74, 613718
MSTN	155,6	100.0%	99.9%	Muscle hypertrophy, 614160
MSTO1	101,4	99.3%	96.1%	Myopathy, mitochondrial, and ataxia, 617675
MSX1	143,3	99.9%	98.6%	Ectodermal dysplasia 3, Witkop type, 189500 Orofacial cleft 5, 608874 Tooth agenesis, selective, 1, with or without orofacial cleft, 106600
MSX2	101,1	100.0%	100.0%	Craniosynostosis 2, 604757 Parietal foramina 1, 168500 Parietal foramina with cleidocranial dysplasia, 168550
MTAP	96	98.9%	93.4%	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250
MTFMT	132,5	100.0%	99.8%	Combined oxidative phosphorylation deficiency 15, 614947 Mitochondrial complex I deficiency, nuclear type 27, 618248
MTHFD1	115,4	99.8%	97.4%	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTHFR	114,9	98.2%	96.4%	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}, 0
MTHFS	89,4	75.4%	75.0%	Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367
MTM1	79,1	98.7%	91.9%	Myotubular myopathy, X-linked, 310400
MTMR2	99,1	99.9%	98.5%	Charcot-Marie-Tooth disease, type 4B1, 601382
MTO1	143,7	91.4%	89.6%	Combined oxidative phosphorylation deficiency 10, 614702
MTOR	112	99.9%	99.1%	Focal cortical dysplasia, type II, somatic, 607341 Smith-Kingsmore syndrome, 616638
MTPAP	122,3	99.0%	93.2%	?Spastic ataxia 4, autosomal recessive, 613672
MTR	131,4	99.9%	99.4%	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTRR	131,1	100.0%	99.0%	Homocystinuria-megaloblastic anemia, cbl E type, 236270 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTTP	114,7	99.9%	99.4%	Abetalipoproteinemia, 200100 {Metabolic syndrome, protection against}, 605552
MUC1	67,7	93.1%	84.2%	Medullary cystic kidney disease 1, 174000
MUSK	131,5	100.0%	100.0%	Fetal akinesia deformation sequence 1, 208150 Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325
MUT	NC	NC	NC	Methylmalonic aciduria, mut(0) type, 251000
MUTYH	152	100.0%	100.0%	Adenomas, multiple colorectal, 608456

				Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600 Gastric cancer, somatic, 613659
MVD	113	99.9%	98.4%	Porokeratosis 7, multiple types, 614714
MVK	121,4	91.0%	90.5%	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900
MXI1	102,7	98.3%	93.4%	Neurofibrosarcoma, somatic, 0 Prostate cancer, somatic, 176807
MYBPC1	127,8	99.9%	99.2%	Arthrogryposis, distal, type 1B, 614335 Lethal congenital contracture syndrome 4, 614915
MYBPC3	141,5	100.0%	98.6%	Cardiomyopathy, dilated, 1MM, 615396 Cardiomyopathy, hypertrophic, 4, 115197 Left ventricular noncompaction 10, 615396
MYC	132,2	66.0%	64.0%	Burkitt lymphoma, somatic, 113970
MYCN	173,8	100.0%	100.0%	Feingold syndrome 1, 164280
MYD88	194,7	100.0%	99.8%	Macroglobulinemia, Waldenstrom, somatic, 153600 Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260
MYF5	171,4	100.0%	100.0%	Ophthalmoplegia, external, with rib and vertebral anomalies, 618155
MYH11	122,1	100.0%	99.5%	Aortic aneurysm, familial thoracic 4, 132900
MYH14	109,8	99.0%	95.1%	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369 Deafness, autosomal dominant 4A, 600652
MYH2	108,2	100.0%	99.5%	Proximal myopathy and ophthalmoplegia, 605637
MYH3	94,1	99.9%	98.3%	Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700 Arthrogryposis, distal, type 2B (Sheldon-Hall), 601680 Arthrogryposis, distal, type 8, 178110
MYH6	95,5	99.0%	95.3%	Atrial septal defect 3, 614089 Cardiomyopathy, dilated, 1EE, 613252 Cardiomyopathy, hypertrophic, 14, 613251 {Sick sinus syndrome 3}, 614090
MYH7	92,2	99.5%	96.4%	Cardiomyopathy, dilated, 1S, 613426 Cardiomyopathy, hypertrophic, 1, 192600 Laing distal myopathy, 160500 Left ventricular noncompaction 5, 613426 Myopathy, myosin storage, autosomal dominant, 608358 Myopathy, myosin storage, autosomal recessive, 255160 Scapuloperoneal syndrome, myopathic type, 181430
MYH8	115,4	100.0%	99.4%	Carney complex variant, 608837

				Trismus-pseudocamptodactyly syndrome, 158300
MYH9	128,5	99.6%	98.5%	Deafness, autosomal dominant 17, 603622 Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100
MYL2	132,2	99.8%	95.9%	Cardiomyopathy, hypertrophic, 10, 608758
MYL3	98	100.0%	100.0%	Cardiomyopathy, hypertrophic, 8, 608751
MYL4	133,2	100.0%	100.0%	?Atrial fibrillation, familial, 18, 617280
MYLK	124,3	99.9%	99.5%	Aortic aneurysm, familial thoracic 7, 613780
MYLK2	132,9	100.0%	100.0%	Cardiomyopathy, hypertrophic, 1, digenic, 192600
MYMK	140,5	100.0%	100.0%	Carey-Fineman-Ziter syndrome, 254940
MYO15A	143,7	99.8%	98.8%	Deafness, autosomal recessive 3, 600316
MYO18B	122,1	99.9%	99.2%	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549
MYO1E	115,4	99.8%	98.7%	Glomerulosclerosis, focal segmental, 6, 614131
MYO3A	113	99.2%	95.3%	Deafness, autosomal recessive 30, 607101
MYO5A	109	99.7%	98.6%	Griscelli syndrome, type 1, 214450
MYO5B	108,3	97.9%	94.7%	Microvillus inclusion disease, 251850
MYO6	101,5	99.5%	96.4%	Deafness, autosomal dominant 22, 606346 Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346 Deafness, autosomal recessive 37, 607821
MYO7A	125,2	99.8%	98.5%	Deafness, autosomal dominant 11, 601317 Deafness, autosomal recessive 2, 600060 Usher syndrome, type 1B, 276900
MYO9A	139,8	99.9%	99.1%	Myasthenic syndrome, congenital, 24, presynaptic, 618198
MYOC	153,4	99.9%	98.6%	Glaucoma 1A, primary open angle, 137750
MYOT	138,6	100.0%	99.2%	Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
MYOZ2	142,4	100.0%	99.9%	Cardiomyopathy, hypertrophic, 16, 613838
MYPN	124,8	99.9%	99.0%	Cardiomyopathy, dilated, 1KK, 615248 Cardiomyopathy, familial restrictive, 4, 615248 Cardiomyopathy, hypertrophic, 22, 615248 Nemaline myopathy 11, autosomal recessive, 617336
MYRF	144,1	97.5%	96.5%	Cardiac-urogenital syndrome, 618280 Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113
MYSM1	111	99.8%	98.4%	Bone marrow failure syndrome 4, 618116
MYT1L	144,7	100.0%	99.8%	Mental retardation, autosomal dominant 39, 616521
NAA10	105	100.0%	98.8%	?Microphthalmia, syndromic 1, 309800

				Ogden syndrome, 300855
NAA15	95,7	97.6%	94.5%	Mental retardation, autosomal dominant 50, 617787
NACC1	169,5	100.0%	100.0%	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393
NADK2	163,3	99.9%	99.0%	?2,4-dienoyl-CoA reductase deficiency, 616034
NAGA	121,7	100.0%	100.0%	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241
NAGLU	117,7	97.1%	94.1%	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NAGS	104,3	100.0%	99.9%	N-acetylglutamate synthase deficiency, 237310
NALCN	117	99.7%	98.7%	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419
NANOS1	106,8	100.0%	99.3%	Spermatogenic failure 12, 615413
NANS	97,2	99.9%	98.4%	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NARS2	120,7	97.6%	97.2%	Combined oxidative phosphorylation deficiency 24, 616239
NAT8L	91,4	98.8%	94.6%	?N-acetylaspartate deficiency, 614063
NAXD	132,1	100.0%	99.9%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321
NAXE	81,4	99.7%	97.0%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NBAS	138,5	99.9%	99.1%	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NBEAL2	166	100.0%	99.5%	Gray platelet syndrome, 139090
NBN	93,8	99.8%	98.4%	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260
NCAPD2	118,1	100.0%	99.4%	?Microcephaly 21, primary, autosomal recessive, 617983
NCAPD3	100,1	99.8%	98.2%	Microcephaly 22, primary, autosomal recessive, 617984
NCAPH	119,6	100.0%	99.9%	?Microcephaly 23, primary, autosomal recessive, 617985
NCF1	23,1	27.8%	22.5%	Chronic granulomatous disease due to deficiency of NCF-1, 233700
NCF2	109,2	99.8%	98.2%	Chronic granulomatous disease due to deficiency of NCF-2, 233710
NCF4	148,6	100.0%	100.0%	?Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III, 613960
NCSTN	92,7	100.0%	99.6%	Acne inversa, familial, 1, 142690
NDE1	89,3	100.0%	99.6%	?Microhydranencephaly, 605013 Lissencephaly 4 (with microcephaly), 614019
NDN	125	99.5%	96.9%	Prader-Willi syndrome, 176270
NDP	91,6	100.0%	99.5%	Exudative vitreoretinopathy 2, X-linked, 305390

				Norrie disease, 310600
NDRG1	114	100.0%	100.0%	Charcot-Marie-Tooth disease, type 4D, 601455
NDST1	188,4	100.0%	100.0%	Mental retardation, autosomal recessive 46, 616116
NDUFA1	184,9	99.9%	99.2%	Mitochondrial complex I deficiency, nuclear type 12, 301020
NDUFA10	114,6	99.9%	98.9%	Mitochondrial complex I deficiency, nuclear type 22, 618243
NDUFA11	116	99.8%	97.4%	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFA12	160,8	100.0%	100.0%	?Mitochondrial complex I deficiency, nuclear type 23, 618244
NDUFA13	121,3	92.3%	91.7%	?Mitochondrial complex I deficiency, nuclear type 28, 618249 {Thyroid carcinoma, Hurthle cell}, 607464
NDUFA2	162,6	100.0%	99.6%	?Mitochondrial complex I deficiency, nuclear type 13, 618235
NDUFA6	201,9	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 33, 618253
NDUFA9	101,6	99.7%	96.5%	Mitochondrial complex I deficiency, nuclear type 26, 618247
NDUFAF1	98,5	100.0%	99.9%	Mitochondrial complex I deficiency, nuclear type 11, 618234
NDUFAF2	54,1	94.3%	82.0%	Mitochondrial complex I deficiency, nuclear type 10, 618233
NDUFAF3	141	100.0%	99.9%	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF4	98,3	99.2%	94.5%	Mitochondrial complex I deficiency, nuclear type 15, 618237
NDUFAF5	124,9	99.9%	99.1%	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFAF6	91,9	99.8%	98.5%	Mitochondrial complex I deficiency, nuclear type 17, 618239
NDUFB11	103,3	98.6%	95.0%	?Mitochondrial complex I deficiency, nuclear type 30, 301021 Linear skin defects with multiple congenital anomalies 3, 300952
NDUFB3	23,3	89.7%	62.5%	Mitochondrial complex I deficiency, nuclear type 25, 618246
NDUFB8	105,3	100.0%	99.8%	Mitochondrial complex I deficiency, nuclear type 32, 618252
NDUFB9	105,2	97.8%	93.3%	?Mitochondrial complex I deficiency, nuclear type 24, 618245
NDUFS1	143,5	99.9%	99.8%	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	100,1	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	124,8	90.7%	90.5%	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	144,5	100.0%	99.7%	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS6	111,9	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFS7	140,5	100.0%	99.9%	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFS8	156,8	100.0%	99.7%	Mitochondrial complex I deficiency, nuclear type 2, 618222
NDUFV1	141,7	99.9%	98.8%	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	74,2	92.4%	77.3%	Mitochondrial complex I deficiency, nuclear type 7, 618229
NEB	100,1	83.0%	82.4%	Nemaline myopathy 2, autosomal recessive, 256030
NECAP1	102,3	100.0%	100.0%	?Epileptic encephalopathy, early infantile, 21, 615833
NECTIN1	134	100.0%	99.9%	Cleft lip/palate-ectodermal dysplasia syndrome, 225060

				Orofacial cleft 7, 225060
NECTIN4	121,6	100.0%	99.9%	Ectodermal dysplasia-syndactyly syndrome 1, 613573
NEDD4L	93,7	72.3%	71.5%	Periventricular nodular heterotopia 7, 617201
NEFH	110,6	99.5%	97.6%	?{Amyotrophic lateral sclerosis, susceptibility to}, 105400 Charcot-Marie-Tooth disease, axonal, type 2CC, 616924
NEFL	178,7	99.8%	97.8%	Charcot-Marie-Tooth disease, dominant intermediate G, 617882 Charcot-Marie-Tooth disease, type 1F, 607734 Charcot-Marie-Tooth disease, type 2E, 607684
NEK1	115,9	99.7%	98.1%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520 {Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892
NEK2	86,4	98.6%	92.4%	?Retinitis pigmentosa 67, 615565
NEK8	141,3	100.0%	99.9%	?Nephronophthisis 9, 613824 Renal-hepatic-pancreatic dysplasia 2, 615415
NEK9	118,9	99.8%	98.2%	?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262 Lethal congenital contracture syndrome 10, 617022 Nevus comedonicus, somatic, 617025
NEU1	141,3	99.3%	96.4%	Sialidosis, type I, 256550 Sialidosis, type II, 256550
NEUROD1	154,5	100.0%	99.4%	Maturity-onset diabetes of the young 6, 606394 {Diabetes mellitus, noninsulin-dependent}, 125853
NEUROD2	145,4	100.0%	100.0%	Epileptic encephalopathy, early infantile, 72, 618374
NEUROG3	172,9	100.0%	100.0%	Diarrhea 4, malabsorptive, congenital, 610370
NEXMIF	132	100.0%	99.5%	Mental retardation, X-linked 98, 300912
NEXN	90,2	96.1%	85.9%	Cardiomyopathy, dilated, 1CC, 613122 Cardiomyopathy, hypertrophic, 20, 613876
NF1	106,2	92.5%	89.4%	Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321 Watson syndrome, 193520
NF2	94,2	100.0%	99.6%	Meningioma, NF2-related, somatic, 607174 Neurofibromatosis, type 2, 101000 Schwannomatosis, somatic, 162091
NFASC	121,2	100.0%	99.8%	Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356
NFE2L2	166,2	100.0%	99.9%	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744
NFIA	149,5	100.0%	98.8%	Brain malformations with or without urinary tract defects, 613735
NFIB	107,6	97.5%	96.9%	Macrocephaly, acquired, with impaired intellectual development, 618286

NFIX	174,4	100.0%	99.7%	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753
NFKB1	93	99.9%	98.7%	Immunodeficiency, common variable, 12, 616576
NFKB2	135	99.1%	96.5%	Immunodeficiency, common variable, 10, 615577
NFKBIA	134,6	95.3%	89.4%	Ectodermal dysplasia and immunodeficiency 2, 612132
NFU1	61,8	97.4%	82.1%	Multiple mitochondrial dysfunctions syndrome 1, 605711
NGF	199	100.0%	100.0%	Neuropathy, hereditary sensory and autonomic, type V, 608654
NGLY1	135,4	100.0%	99.7%	Congenital disorder of deglycosylation, 615273
NHEJ1	58,5	99.7%	92.8%	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
NHLRC1	169,7	100.0%	100.0%	Epilepsy, progressive myoclonic 2B (Lafora), 254780
NHLRC2	119	99.8%	98.7%	FINCA syndrome, 618278
NHP2	121,9	100.0%	99.2%	Dyskeratosis congenita, autosomal recessive 2, 613987
NHS	111	98.5%	96.0%	Cataract 40, X-linked, 302200 Nance-Horan syndrome, 302350
NIN	127	99.9%	99.4%	?Seckel syndrome 7, 614851
NIPA1	156,7	100.0%	99.9%	Spastic paraplegia 6, autosomal dominant, 600363
NIPAL4	126,7	100.0%	99.3%	Ichthyosis, congenital, autosomal recessive 6, 612281
NIPBL	124,9	98.8%	96.9%	Cornelia de Lange syndrome 1, 122470
NKX2-1	88,8	100.0%	99.7%	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 {Thyroid cancer, nonmedullary, 1}, 188550
NKX2-5	120,8	100.0%	99.9%	Atrial septal defect 7, with or without AV conduction defects, 108900 Conotruncal heart malformations, variable, 217095 Hypoplastic left heart syndrome 2, 614435 Hypothyroidism, congenital nongoitrous, 5, 225250 Tetralogy of Fallot, 187500 Ventricular septal defect 3, 614432
NKX2-6	139,9	100.0%	100.0%	Conotruncal heart malformations, 217095 Persistent truncus arteriosus, 217095
NKX3-2	138,4	100.0%	99.8%	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330
NKX6-2	122,9	98.1%	91.2%	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560
NLGN4X	147,9	99.5%	97.4%	Mental retardation, X-linked, 300495 {Asperger syndrome susceptibility, X-linked 2}, 300497 {Autism susceptibility, X-linked 2}, 300495
NLRC4	159,4	100.0%	99.9%	?Familial cold autoinflammatory syndrome 4, 616115 Autoinflammation with infantile enterocolitis, 616050

NLRP1	117,7	99.5%	97.6%	Autoinflammation with arthritis and dyskeratosis, 617388 Palmoplantar carcinoma, multiple self-healing, 615225 {Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579
NLRP12	161,7	100.0%	100.0%	Familial cold autoinflammatory syndrome 2, 611762
NLRP3	134,6	100.0%	99.9%	CINCA syndrome, 607115 Deafness, autosomal dominant 34, with or without inflammation, 617772 Familial cold inflammatory syndrome 1, 120100 Keratoendothelitis fugax hereditaria, 148200 Muckle-Wells syndrome, 191900
NLRP7	124,5	99.9%	98.8%	Hydatidiform mole, recurrent, 1, 231090
NME1	77,1	100.0%	99.9%	Neuroblastoma, 256700
NME8	104,7	98.6%	93.8%	Ciliary dyskinesia, primary, 6, 610852
NMNAT1	113,5	100.0%	98.5%	Leber congenital amaurosis 9, 608553
NNT	124,6	100.0%	98.5%	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736
NOBOX	98,9	99.8%	98.1%	Premature ovarian failure 5, 611548
NOD2	125,3	100.0%	99.9%	Blau syndrome, 186580 {Inflammatory bowel disease 1, Crohn disease}, 266600 {Psoriatic arthritis, susceptibility to}, 607507 {Yao syndrome}, 617321
NODAL	144,8	100.0%	100.0%	Heterotaxy, visceral, 5, 270100
NOG	233,6	100.0%	100.0%	Brachydactyly, type B2, 611377 Multiple synostoses syndrome 1, 186500 Stapes ankylosis with broad thumbs and toes, 184460 Symphalangism, proximal, 1A, 185800 Tarsal-carpal coalition syndrome, 186570
NOL3	112,8	99.6%	96.3%	?Myoclonus, familial, 1, 614937
NONO	78,1	99.1%	95.3%	Mental retardation, X-linked, syndromic 34, 300967
NOP10	120,5	100.0%	100.0%	Dyskeratosis congenita, autosomal recessive 1, 224230
NOP56	113,1	100.0%	98.8%	Spinocerebellar ataxia 36, 614153
NOTCH1	141,8	99.8%	98.9%	Adams-Oliver syndrome 5, 616028 Aortic valve disease 1, 109730
NOTCH2	123,7	100.0%	99.6%	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NOTCH3	106,3	98.4%	94.2%	?Myofibromatosis, infantile 2, 615293 Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310 Lateral meningocele syndrome, 130720
NPC1	117,8	100.0%	99.2%	Niemann-Pick disease, type C1, 257220

				Niemann-Pick disease, type D, 257220
NPC2	124,7	100.0%	99.9%	Niemann-pick disease, type C2, 607625
NPHP1	121,2	99.8%	98.5%	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NPHP3	121,4	99.8%	98.5%	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540
NPHP4	125,6	100.0%	99.7%	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
NPHS1	106,3	100.0%	99.5%	Nephrotic syndrome, type 1, 256300
NPHS2	114,5	100.0%	99.5%	Nephrotic syndrome, type 2, 600995
NPM1	65	94.5%	83.5%	Leukemia, acute myeloid, somatic, 601626
NPPA	158,5	100.0%	100.0%	Atrial fibrillation, familial, 6, 612201 Atrial standstill 2, 615745
NPR2	144,1	100.0%	99.4%	Acromesomelic dysplasia, Maroteaux type, 602875 Epiphyseal chondrodysplasia, Miura type, 615923 Short stature with nonspecific skeletal abnormalities, 616255
NPRL2	138,7	100.0%	100.0%	Epilepsy, familial focal, with variable foci 2, 617116
NPRL3	120,3	100.0%	99.8%	Epilepsy, familial focal, with variable foci 3, 617118
NR0B1	138,6	99.9%	99.2%	46XY sex reversal 2, dosage-sensitive, 300018 Adrenal hypoplasia, congenital, 300200
NR0B2	102,8	100.0%	99.7%	Obesity, mild, early-onset, 601665
NR1H4	122,3	99.8%	98.3%	Cholestasis, progressive familial intrahepatic, 5, 617049
NR2E3	105,1	99.9%	98.6%	Enhanced S-cone syndrome, 268100 Retinitis pigmentosa 37, 611131
NR2F1	222,3	100.0%	100.0%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NR2F2	236,6	100.0%	100.0%	Congenital heart defects, multiple types, 4, 615779
NR3C1	129,3	100.0%	99.9%	Glucocorticoid resistance, 615962
NR3C2	123,4	100.0%	98.1%	Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115 Pseudohypoaldosteronism type I, autosomal dominant, 177735
NR4A3	114,9	100.0%	99.9%	Chondrosarcoma, extraskeletal myxoid, 612237
NR5A1	111	100.0%	99.7%	46, XX sex reversal 4, 617480 46XY sex reversal 3, 612965 Adrenocortical insufficiency, 612964 Premature ovarian failure 7, 612964 Spermatogenic failure 8, 613957

NRAS	145,5	100.0%	100.0%	?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Colorectal cancer, somatic, 114500 Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Thyroid carcinoma, follicular, somatic, 188470
NRIP1	181,1	100.0%	100.0%	?Congenital anomalies of kidney and urinary tract 3, 618270
NRL	114,8	99.9%	98.3%	Retinal degeneration, autosomal recessive, clumped pigment type, 0 Retinitis pigmentosa 27, 613750
NRXN1	141,6	97.6%	97.3%	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332
NSD1	147	100.0%	99.8%	Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550
NSDHL	125,8	99.7%	97.1%	CHILD syndrome, 308050 CK syndrome, 300831
NSMCE2	81,4	99.9%	98.6%	Seckel syndrome 10, 617253
NSMCE3	194	100.0%	100.0%	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241
NSMF	109,8	99.4%	97.4%	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
NSUN2	94,7	97.6%	93.5%	Mental retardation, autosomal recessive 5, 611091
NT5C2	121,2	97.9%	96.3%	Spastic paraplegia 45, autosomal recessive, 613162
NT5C3A	64,1	97.2%	85.7%	Anemia, hemolytic, due to UMPH1 deficiency, 266120
NT5E	151,4	100.0%	99.9%	Calcification of joints and arteries, 211800
NTF4	151,2	99.8%	97.7%	Glaucoma 1, open angle, 1O, 613100
NTHL1	121,6	100.0%	100.0%	Familial adenomatous polyposis 3, 616415
NTN1	194,9	100.0%	100.0%	Mirror movements 4, 618264
NTRK1	133	100.0%	99.3%	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma, familial, 155240
NTRK2	136,2	100.0%	99.9%	Epileptic encephalopathy, early infantile, 58, 617830 Obesity, hyperphagia, and developmental delay, 613886
NUBPL	102	98.9%	95.5%	Mitochondrial complex I deficiency, nuclear type 21, 618242
NUMA1	132	100.0%	99.5%	Leukemia, acute promyelocytic, somatic, 612376
NUP107	126,6	99.8%	98.6%	?Ovarian dysgenesis 6, 618078 Galloway-Mowat syndrome 7, 618348 Nephrotic syndrome, type 11, 616730
NUP133	121,1	99.7%	97.8%	?Galloway-Mowat syndrome 8, 618349

				Nephrotic syndrome, type 18, 618177
NUP155	120,3	98.4%	96.9%	?Atrial fibrillation 15, 615770
NUP160	137,7	100.0%	99.9%	?Nephrotic syndrome, type 19, 618178
NUP205	133,5	99.6%	98.7%	?Nephrotic syndrome, type 13, 616893
NUP214	149,1	99.9%	99.4%	Leukemia, acute myeloid, somatic, 601626 Leukemia, T-cell acute lymphoblastic, somatic, 613065
NUP37	156,4	100.0%	99.7%	?Microcephaly 24, primary, autosomal recessive, 618179
NUP62	111,8	100.0%	100.0%	Striatonigral degeneration, infantile, 271930
NUP85	125,7	100.0%	100.0%	Nephrotic syndrome, type 17, 618176
NUP88	141,5	100.0%	100.0%	Fetal akinesia deformation sequence 4, 618393
NUP93	117,8	96.9%	93.8%	Nephrotic syndrome, type 12, 616892
NUS1	53,3	71.5%	44.1%	?Congenital disorder of glycosylation, type 1aa, 617082 Mental retardation, autosomal dominant 55, with seizures, 617831
NYX	131,7	99.6%	98.1%	Night blindness, congenital stationary (complete), 1A, X-linked, 310500
OAT	68,2	81.7%	70.1%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OBSL1	147,2	100.0%	99.8%	3-M syndrome 2, 612921
OCA2	116,8	99.7%	97.7%	Albinism, brown oculocutaneous, 203200 Albinism, oculocutaneous, type II, 203200 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220
OCLN	173,9	100.0%	100.0%	Pseudo-TORCH syndrome 1, 251290
OCRL	106,2	99.8%	98.3%	Dent disease 2, 300555 Lowe syndrome, 309000
OFD1	51,9	85.8%	70.8%	?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209
OGG1	118,7	100.0%	99.7%	Renal cell carcinoma, clear cell, somatic, 144700
OGT	106,5	99.9%	98.4%	Mental retardation, X-linked 106, 300997
OPA1	124,7	99.7%	97.4%	?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896 Behr syndrome, 210000 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250 {Glaucoma, normal tension, susceptibility to}, 606657
OPA3	156,6	100.0%	99.2%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300

OPCML	144,1	99.6%	99.5%	Ovarian cancer, somatic, 167000
OPHN1	78,3	98.9%	95.0%	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
OPLAH	141,5	100.0%	99.9%	5-oxoprolinase deficiency, 260005
OPN1LW	58,7	68.3%	61.3%	Blue cone monochromacy, 303700 Colorblindness, protan, 303900
OPN1MW	56,9	66.1%	57.9%	Blue cone monochromacy, 303700 Colorblindness, deutan, 303800
OPN1SW	95,9	100.0%	100.0%	Colorblindness, tritan, 190900
OPTN	104	100.0%	99.7%	Amyotrophic lateral sclerosis 12, 613435 Glaucoma 1, open angle, E, 137760 {Glaucoma, normal tension, susceptibility to}, 606657
ORAI1	198,9	99.8%	98.2%	Immunodeficiency 9, 612782 Myopathy, tubular aggregate, 2, 615883
ORC1	90,3	99.9%	98.7%	Meier-Gorlin syndrome 1, 224690
ORC4	73,6	98.1%	92.0%	Meier-Gorlin syndrome 2, 613800
ORC6	127,6	100.0%	99.9%	Meier-Gorlin syndrome 3, 613803
OSBPL2	138,2	100.0%	100.0%	Deafness, autosomal dominant 67, 616340
OSGEP	98,1	100.0%	97.3%	Galloway-Mowat syndrome 3, 617729
OSMR	131,7	100.0%	99.5%	Amyloidosis, primary localized cutaneous, 1, 105250
OSTM1	109,3	98.2%	92.5%	Osteopetrosis, autosomal recessive 5, 259720
OTC	111,4	100.0%	99.7%	Ornithine transcarbamylase deficiency, 311250
OTOA	97,2	99.7%	98.3%	Deafness, autosomal recessive 22, 607039
OTOF	135,6	100.0%	99.8%	Auditory neuropathy, autosomal recessive, 1, 601071 Deafness, autosomal recessive 9, 601071
OTOG	134,6	99.6%	98.8%	Deafness, autosomal recessive 18B, 614945
OTOGL	104,6	99.4%	97.0%	Deafness, autosomal recessive 84B, 614944
OTUD6B	117,7	99.8%	99.3%	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452
OTULIN	132,6	98.7%	95.2%	Autoinflammation, panniculitis, and dermatosis syndrome, 617099
OTX2	127,4	100.0%	99.3%	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125
OVOL2	120,8	99.8%	97.7%	Corneal dystrophy, posterior polymorphous, 1, 122000
OXCT1	125,5	99.7%	98.2%	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050
P2RX2	174,5	100.0%	100.0%	Deafness, autosomal dominant 41, 608224
P2RY12	186,2	100.0%	100.0%	Bleeding disorder, platelet-type, 8, 609821
P3H1	129,3	100.0%	100.0%	Osteogenesis imperfecta, type VIII, 610915

P3H2	98,8	99.9%	99.4%	Myopia, high, with cataract and vitreoretinal degeneration, 614292
P4HA2	114,8	99.9%	98.4%	Myopia 25, autosomal dominant, 617238
P4HB	108,7	94.6%	93.8%	Cole-Carpenter syndrome 1, 112240
PABPN1	76,7	76.7%	60.6%	Oculopharyngeal muscular dystrophy, 164300
PACS1	106,7	100.0%	99.4%	Schuurs-Hoeijmakers syndrome, 615009
PACS2	155,4	100.0%	99.3%	Epileptic encephalopathy, early infantile, 66, 618067
PADI3	139	100.0%	100.0%	Uncombable hair syndrome, 191480
PADI6	104,5	99.9%	99.0%	Preimplantation embryonic lethality 2, 617234
PAFAH1B1	77	92.0%	82.8%	Lissencephaly 1, 607432 Subcortical laminar heterotopia, 607432
PAH	126,4	100.0%	100.0%	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600
PAK1	103	99.9%	98.8%	Intellectual developmental disorder with macrocephaly, seizures, and speech delay, 618158
PAK3	85,3	98.6%	93.7%	Mental retardation, X-linked 30/47, 300558
PALB2	143,5	100.0%	99.9%	Fanconi anemia, complementation group N, 610832 {Breast cancer, susceptibility to}, 114480 {Pancreatic cancer, susceptibility to, 3}, 613348
PAM16	64,5	66.4%	65.3%	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320
PANK2	154,1	100.0%	100.0%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PAPSS2	103,8	99.7%	97.7%	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847
PARK7	83,5	100.0%	99.8%	Parkinson disease 7, autosomal recessive early-onset, 606324
PARN	127,3	99.9%	99.5%	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
PATL2	93,7	99.8%	96.2%	Oocyte maturation defect 4, 617743
PAX1	189,5	97.0%	92.1%	?Otofaciocervical syndrome 2, 615560
PAX2	184,1	100.0%	100.0%	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330
PAX3	106,9	100.0%	99.7%	Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220 Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820
PAX4	81,7	100.0%	99.0%	Diabetes mellitus, type 2, 125853 Maturity-onset diabetes of the young, type IX, 612225 {Diabetes mellitus, ketosis-prone, susceptibility to}, 612227
PAX6	116,5	100.0%	99.8%	?Coloboma of optic nerve, 120430

				?Coloboma, ocular, 120200 ?Morning glory disc anomaly, 120430 Aniridia, 106210 Anterior segment dysgenesis 5, multiple subtypes, 604229 Cataract with late-onset corneal dystrophy, 106210 Foveal hypoplasia 1, 136520 Keratitis, 148190 Optic nerve hypoplasia, 165550
PAX7	131,1	100.0%	100.0%	Rhabdomyosarcoma 2, alveolar, 268220
PAX8	94,4	100.0%	99.8%	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700
PAX9	236,1	99.8%	99.6%	Tooth agenesis, selective, 3, 604625
PBX1	111,7	99.9%	98.2%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PC	155,4	99.9%	98.7%	Pyruvate carboxylase deficiency, 266150
PCBD1	103,9	100.0%	99.7%	Hyperphenylalaninemia, BH4-deficient, D, 264070
PCCA	99,2	99.3%	95.5%	Propionicacidemia, 606054
PCCB	111,8	99.3%	96.9%	Propionicacidemia, 606054
PCDH12	182,1	100.0%	100.0%	Microcephaly, seizures, spasticity, and brain calcification, 251280
PCDH15	140,3	99.2%	99.0%	Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1F, 602083
PCDH19	176,6	99.9%	98.9%	Epileptic encephalopathy, early infantile, 9, 300088
PCGF2	95,1	99.4%	97.0%	Turnpenny-Fry syndrome, 618371
PCK1	119,4	100.0%	99.9%	?Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680
PCLO	142,1	99.8%	99.0%	?Pontocerebellar hypoplasia, type 3, 608027
PCNA	92	100.0%	98.2%	?Ataxia-telangiectasia-like disorder 2, 615919
PCNT	115,4	99.7%	97.7%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PCSK1	141,9	100.0%	99.2%	Obesity with impaired prohormone processing, 600955 {Obesity, susceptibility to, BMIQ12}, 612362
PCSK9	102,8	96.8%	92.9%	Hypercholesterolemia, familial, 3, 603776 {Low density lipoprotein cholesterol level QTL 1}, 603776
PCYT1A	95,6	97.9%	94.4%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDCD10	98,3	99.8%	99.0%	Cerebral cavernous malformations 3, 603285
PDE10A	107,6	81.4%	80.4%	Dyskinesia, limb and orofacial, infantile-onset, 616921 Striatal degeneration, autosomal dominant, 616922
PDE11A	152,1	99.9%	99.9%	Pigmented nodular adrenocortical disease, primary, 2, 610475

PDE1C	108	99.8%	99.4%	?Deafness, autosomal dominant 74, 618140
PDE3A	120,4	99.9%	99.2%	Hypertension and brachydactyly syndrome, 112410
PDE4D	102,7	95.8%	94.4%	Acrodysostosis 2, with or without hormone resistance, 614613
PDE6A	102,6	100.0%	99.2%	Retinitis pigmentosa 43, 613810
PDE6B	157,3	100.0%	100.0%	Night blindness, congenital stationary, autosomal dominant 2, 163500 Retinitis pigmentosa-40, 613801
PDE6C	116,2	99.5%	97.2%	Cone dystrophy 4, 613093
PDE6D	114,7	100.0%	99.9%	?Joubert syndrome 22, 615665
PDE6G	125,8	100.0%	99.5%	Retinitis pigmentosa 57, 613582
PDE6H	58,2	98.5%	76.0%	Achromatopsia 6, 610024 Retinal cone dystrophy 3, 610024
PDE8B	99,7	99.9%	98.9%	Pigmented nodular adrenocortical disease, primary, 3, 614190 Striatal degeneration, autosomal dominant, 609161
PDGFB	115,4	100.0%	100.0%	Basal ganglia calcification, idiopathic, 5, 615483 Dermatofibrosarcoma protuberans, 607907 Meningioma, SIS-related, 607174
PDGFRA	124,7	100.0%	100.0%	Gastrointestinal stromal tumor, somatic, 606764 Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685
PDGFRB	126,6	99.7%	98.0%	Basal ganglia calcification, idiopathic, 4, 615007 Kosaki overgrowth syndrome, 616592 Myeloproliferative disorder with eosinophilia, 131440 Myofibromatosis, infantile, 1, 228550 Premature aging syndrome, Penttinen type, 601812
PDGFRL	133,1	100.0%	99.5%	Colorectal cancer, somatic, 114500 Hepatocellular cancer, somatic, 114550
PDHA1	85,3	98.9%	95.4%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	111,4	99.2%	97.2%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	129	99.9%	99.5%	Lacticacidemia due to PDX1 deficiency, 245349
PDK3	108,4	97.4%	94.5%	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905
PDP1	129,1	100.0%	100.0%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	104,8	96.7%	87.7%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	112,9	99.6%	96.1%	Coenzyme Q10 deficiency, primary, 3, 614652
PDX1	72,9	99.1%	95.2%	MODY, type IV, 606392 Pancreatic agenesis 1, 260370 {Diabetes mellitus, type II, susceptibility to}, 125853
PDYN	121,8	100.0%	100.0%	Spinocerebellar ataxia 23, 610245

PDZD7	93,9	99.6%	97.8%	Deafness, autosomal recessive 57, 618003 Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472 {Retinal disease in Usher syndrome type IIA, modifier of}, 276901
PEPD	117,4	100.0%	99.6%	Prolidase deficiency, 170100
PER2	91,8	100.0%	99.2%	Advanced sleep phase syndrome, familial, 1, 604348
PER3	146,1	99.8%	98.1%	?Advanced sleep phase syndrome, familial, 3, 616882
PET100	87,9	98.0%	87.6%	Mitochondrial complex IV deficiency, 220110
PEX1	127,9	99.9%	99.3%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	113,3	99.9%	97.4%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX11B	87,9	100.0%	99.4%	?Peroxisome biogenesis disorder 14B, 614920
PEX12	120,6	100.0%	100.0%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	179,6	100.0%	100.0%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	130,5	99.8%	97.8%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	140,8	98.6%	94.8%	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	84,9	100.0%	98.9%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	134,9	100.0%	100.0%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	94,3	100.0%	99.6%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	113,9	99.9%	99.2%	?Peroxisome biogenesis disorder 10B, 617370 Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	107,9	100.0%	99.2%	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	106,5	98.5%	92.0%	Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863
PEX7	111	91.2%	89.3%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PFKM	113,7	100.0%	99.2%	Glycogen storage disease VII, 232800
PFN1	156,9	100.0%	100.0%	Amyotrophic lateral sclerosis 18, 614808

PGAM2	163,6	100.0%	100.0%	Glycogen storage disease X, 261670
PGAP1	110,9	99.1%	95.8%	Mental retardation, autosomal recessive 42, 615802
PGAP2	134,7	100.0%	99.5%	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGAP3	70,3	63.5%	59.9%	Hyperphosphatasia with mental retardation syndrome 4, 615716
PGK1	44,7	90.9%	75.9%	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	128,8	100.0%	99.8%	Congenital disorder of glycosylation, type It, 614921
PGM3	149,3	99.9%	99.6%	Immunodeficiency 23, 615816
PHACTR1	104,3	100.0%	99.6%	Epileptic encephalopathy, early infantile, 70, 618298
PHC1	178	100.0%	99.5%	?Microcephaly 11, primary, autosomal recessive, 615414
PHEX	107,9	99.8%	98.6%	Hypophosphatemic rickets, X-linked dominant, 307800
PHF6	60,3	98.2%	87.9%	Borjeson-Forssman-Lehmann syndrome, 301900
PHF8	74,4	99.2%	95.8%	Mental retardation syndrome, X-linked, Siderius type, 300263
PHGDH	106,6	100.0%	99.3%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHIP	130,8	99.3%	97.2%	Developmental delay, intellectual disability, obesity, and dysmorphic features, 617991
PHKA1	90,2	97.4%	91.6%	Muscle glycogenosis, 300559
PHKA2	93,7	99.9%	98.7%	Glycogen storage disease, type IXa1, 306000 Glycogen storage disease, type IXa2, 306000
PHKB	125,3	99.9%	99.1%	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750
PHKG2	155,6	100.0%	100.0%	Cirrhosis due to liver phosphorylase kinase deficiency, 0 Glycogen storage disease IXc, 613027
PHOX2A	56	98.6%	88.9%	Fibrosis of extraocular muscles, congenital, 2, 602078
PHOX2B	145,5	100.0%	100.0%	Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, 209880 Neuroblastoma with Hirschsprung disease, 613013 {Neuroblastoma, susceptibility to, 2}, 613013
PHYH	74	99.9%	96.9%	Refsum disease, 266500
PI4KA	91,8	93.7%	89.7%	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531
PIBF1	74,3	99.2%	94.4%	Joubert syndrome 33, 617767
PICALM	101,3	99.5%	95.9%	Leukemia, acute myeloid, somatic, 601626
PIEZO1	144,5	100.0%	99.5%	Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380 Lymphatic malformation 6, 616843
PIEZO2	104,2	99.9%	99.2%	?Marden-Walker syndrome, 248700 Arthrogryposis, distal, type 3, 114300 Arthrogryposis, distal, type 5, 108145 Arthrogryposis, distal, with impaired proprioception and touch, 617146
PIGA	70,9	92.9%	84.0%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868

				Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGC	85,9	99.3%	92.2%	Glycosylphosphatidylinositol biosynthesis defect 16, 617816
PIGG	143,4	100.0%	99.5%	Mental retardation, autosomal recessive 53, 616917
PIGH	93,8	78.8%	67.3%	Glycosylphosphatidylinositol biosynthesis defect 17, 618010
PIGL	122,1	99.7%	99.6%	CHIME syndrome, 280000
PIGM	148,9	100.0%	100.0%	Glycosylphosphatidylinositol deficiency, 610293
PIGN	106,3	93.6%	91.1%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	144,5	100.0%	99.9%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGP	89,3	94.8%	86.0%	?Epileptic encephalopathy, early infantile, 55, 617599
PIGS	90	100.0%	99.4%	Glycosylphosphatidylinositol biosynthesis defect 18, 618143
PIGT	159,3	98.1%	98.1%	?Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGV	124,4	100.0%	100.0%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIGW	145	100.0%	99.8%	Glycosylphosphatidylinositol biosynthesis defect 11, 616025
PIGY	90,3	100.0%	99.9%	Hyperphosphatasia with mental retardation syndrome 6, 616809
PIH1D3	74,7	98.4%	89.1%	Ciliary dyskinesia, primary, 36, X-linked, 300991
PIK3CA	127,7	100.0%	99.8%	Breast cancer, somatic, 114480 CLAPO syndrome, somatic, 613089 CLOVE syndrome, somatic, 612918 Colorectal cancer, somatic, 114500 Cowden syndrome 5, 615108 Gastric cancer, somatic, 613659 Hepatocellular carcinoma, somatic, 114550 Keratosis, seborrheic, somatic, 182000 Macrodactyly, somatic, 155500 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Nevus, epidermal, somatic, 162900 Nonsmall cell lung cancer, somatic, 211980 Ovarian cancer, somatic, 167000
PIK3CD	158,2	99.5%	97.8%	Immunodeficiency 14, 615513
PIK3R1	124,3	99.9%	98.9%	?Agammaglobulinemia 7, autosomal recessive, 615214 Immunodeficiency 36, 616005 SHORT syndrome, 269880
PIK3R2	104,3	93.9%	90.2%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PIK3R5	120,7	100.0%	99.9%	Ataxia-oculomotor apraxia 3, 615217
PIKFYVE	136,4	99.9%	99.7%	Corneal fleck dystrophy, 121850

PINK1	87,3	96.4%	90.7%	Parkinson disease 6, early onset, 605909
PIP5K1C	136,6	99.8%	97.6%	Lethal congenital contractural syndrome 3, 611369
PITPNM3	123,3	99.8%	98.6%	Cone-rod dystrophy 5, 600977
PITX1	174,5	98.9%	96.0%	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800 Liebenberg syndrome, 186550
PITX2	164,8	100.0%	99.5%	Anterior segment dysgenesis 4, 137600 Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550
PITX3	88,5	100.0%	99.5%	Anterior segment dysgenesis 1, multiple subtypes, 107250 Cataract 11, multiple types, 610623 Cataract 11, syndromic, autosomal recessive, 610623
PKD1	35,9	43.0%	35.0%	Polycystic kidney disease 1, 173900
PKD1L1	108,7	100.0%	99.3%	Heterotaxy, visceral, 8, autosomal, 617205
PKD2	102,3	98.7%	95.8%	Polycystic kidney disease 2, 613095
PKHD1	130,4	99.9%	99.4%	Polycystic kidney disease 4, with or without hepatic disease, 263200
PKLR	169,2	100.0%	99.7%	Adenosine triphosphate, elevated, of erythrocytes, 102900 Pyruvate kinase deficiency, 266200
PKP1	120,8	99.8%	98.4%	Ectodermal dysplasia/skin fragility syndrome, 604536
PKP2	91,9	96.7%	90.7%	Arrhythmogenic right ventricular dysplasia 9, 609040
PLA2G4A	124,5	99.8%	99.3%	Gastrointestinal ulceration, recurrent, with dysfunctional platelets, 618372
PLA2G6	111,9	99.8%	98.2%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953
PLA2G7	120,9	99.8%	98.6%	Platelet-activating factor acetylhydrolase deficiency, 614278 {Asthma, susceptibility to}, 600807 {Atopy, susceptibility to}, 147050
PLAA	163	99.7%	98.6%	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527
PLAG1	168,3	100.0%	100.0%	Adenomas, salivary gland pleomorphic, somatic, 181030
PLAU	99,6	100.0%	99.0%	Quebec platelet disorder, 601709 {Alzheimer disease, late-onset, susceptibility to}, 104300
PLCB1	134,9	100.0%	99.7%	Epileptic encephalopathy, early infantile, 12, 613722
PLCB4	102,5	99.8%	98.0%	Auriculocondylar syndrome 2, 614669
PLCD1	116,1	100.0%	99.3%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCE1	125	99.8%	99.0%	Nephrotic syndrome, type 3, 610725
PLCG2	105,8	100.0%	99.3%	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468

PLCZ1	75,7	99.1%	96.0%	?Spermatogenic failure 17, 617214
PLD1	116,4	99.9%	99.3%	Cardiac valvular defect, developmental, 212093
PLD3	172,6	100.0%	100.0%	?Spinocerebellar ataxia 46, 617770
PLEC	144,1	100.0%	100.0%	?Epidermolysis bullosa simplex with nail dystrophy, 616487 Epidermolysis bullosa simplex with muscular dystrophy, 226670 Epidermolysis bullosa simplex with pyloric atresia, 612138 Epidermolysis bullosa simplex, Ogna type, 131950 Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723
PLEKHG2	152,2	100.0%	99.0%	Leukodystrophy and acquired microcephaly with or without dystonia, 616763
PLEKHG5	101,6	99.8%	97.7%	Charcot-Marie-Tooth disease, recessive intermediate C, 615376 Spinal muscular atrophy, distal, autosomal recessive, 4, 611067
PLEKHM1	127,8	100.0%	99.9%	?Osteopetrosis, autosomal recessive 6, 611497 Osteopetrosis, autosomal dominant 3, 618107
PLG	93,4	87.8%	86.8%	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PLIN1	93,1	100.0%	99.3%	Lipodystrophy, familial partial, type 4, 613877
PLK4	149,7	99.8%	98.2%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLN	163,5	100.0%	100.0%	Cardiomyopathy, dilated, 1P, 609909 Cardiomyopathy, hypertrophic, 18, 613874
PLOD1	131,9	99.8%	97.3%	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD2	121,3	99.6%	97.3%	Bruck syndrome 2, 609220
PLOD3	109,7	100.0%	99.9%	Lysyl hydroxylase 3 deficiency, 612394
PLP1	112,8	99.7%	97.7%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PLPBP	95,3	99.6%	95.3%	Epilepsy, early-onset, vitamin B6-dependent, 617290
PLPP6	179	100.0%	99.4%	Phospholipid phosphatase 6, 611666
PLS3	116,9	96.9%	95.3%	Bone mineral density QTL18, osteoporosis, 300910
PLVAP	161	100.0%	100.0%	Diarrhea 10, protein-losing enteropathy type, 618183
PMFBP1	104,4	99.9%	99.0%	Spermatogenic failure 31, 618112
PML	153,4	100.0%	100.0%	Leukemia, acute promyelocytic, PML/RARA type, 0
PMM2	127,7	100.0%	99.7%	Congenital disorder of glycosylation, type Ia, 212065
PMP2	122,6	100.0%	99.7%	Charcot-Marie-Tooth disease, demyelinating, type 1G, 618279
PMP22	96,3	98.9%	94.3%	?Neuropathy, inflammatory demyelinating, 139393 Charcot-Marie-Tooth disease, type 1A, 118220 Charcot-Marie-Tooth disease, type 1E, 118300 Dejerine-Sottas disease, 145900

				Neuropathy, recurrent, with pressure palsies, 162500 Roussy-Levy syndrome, 180800
PMPCA	108,1	99.1%	95.9%	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMPCB	121,6	100.0%	99.2%	Multiple mitochondrial dysfunctions syndrome 6, 617954
PMS2	94,7	83.4%	81.0%	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 Mismatch repair cancer syndrome, 276300
PMVK	118,7	100.0%	99.9%	Porokeratosis 1, multiple types, 175800
PNKD	126,8	100.0%	99.8%	Paroxysmal nonkinesigenic dyskinesia 1, 118800
PNKP	109	100.0%	99.9%	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNLIP	133,9	100.0%	97.7%	?Pancreatic lipase deficiency, 614338
PNP	108,6	100.0%	99.5%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA1	164,8	100.0%	100.0%	Ichthyosis, congenital, autosomal recessive 10, 615024
PNPLA2	142,7	100.0%	99.8%	Neutral lipid storage disease with myopathy, 610717
PNPLA6	137,9	99.9%	99.5%	?Laurence-Moon syndrome, 245800 Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 Spastic paraplegia 39, autosomal recessive, 612020
PNPLA8	121,2	100.0%	99.7%	?Mitochondrial myopathy with lactic acidosis, 251950
PNPO	74,4	100.0%	99.3%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
PNPT1	56,1	96.2%	84.3%	Combined oxidative phosphorylation deficiency 13, 614932 Deafness, autosomal recessive 70, 614934
POC1A	112,9	100.0%	100.0%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POC1B	79,6	99.7%	97.9%	Cone-rod dystrophy 20, 615973
POF1B	71,7	94.6%	84.3%	?Premature ovarian failure 2B, 300604
POFUT1	134,6	99.9%	99.4%	Dowling-Degos disease 2, 615327
POGLUT1	101,2	100.0%	98.7%	?Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232 Dowling-Degos disease 4, 615696
POGZ	122,2	99.5%	99.0%	White-Sutton syndrome, 616364
POLA1	104	99.3%	95.4%	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220
POLD1	124,5	98.0%	93.9%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381 {Colorectal cancer, susceptibility to, 10}, 612591
POLE	126,9	99.9%	99.4%	FILS syndrome, 615139 IMAGE-I syndrome, 618336 {Colorectal cancer, susceptibility to, 12}, 615083
POLG	113,9	100.0%	99.6%	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700

				Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
POLG2	183,7	99.6%	98.0%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131
POLH	116,2	99.9%	98.6%	Xeroderma pigmentosum, variant type, 278750
POLR1A	103,6	99.9%	98.2%	Acrofacial dysostosis, Cincinnati type, 616462
POLR1C	98,3	98.9%	94.9%	Leukodystrophy, hypomyelinating, 11, 616494 Treacher Collins syndrome 3, 248390
POLR1D	183,1	91.6%	91.6%	Treacher Collins syndrome 2, 613717
POLR3A	116,8	100.0%	99.9%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 Wiedemann-Rautenstrauch syndrome, 264090
POLR3B	129,8	99.7%	98.2%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMC	148,2	100.0%	100.0%	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734 {Obesity, early-onset, susceptibility to}, 601665
POMGNT1	115,5	100.0%	99.6%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 Retinitis pigmentosa 76, 617123
POMGNT2	201,7	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830 Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135
POMK	138,7	100.0%	100.0%	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249
POMP	124,6	99.9%	97.6%	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952 Proteasome-associated autoinflammatory syndrome 2, 618048
POMT1	130,6	99.7%	97.8%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308
POMT2	103,3	100.0%	98.4%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158
POP1	120,1	100.0%	99.4%	Anauxetic dysplasia 2, 617396
POR	175,5	99.2%	97.1%	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571
PORCN	111,2	99.9%	98.8%	Focal dermal hypoplasia, 305600

POU1F1	109,1	99.9%	98.2%	Pituitary hormone deficiency, combined, 1, 613038
POU3F4	135,9	100.0%	100.0%	Deafness, X-linked 2, 304400
POU4F3	261,7	100.0%	100.0%	Deafness, autosomal dominant 15, 602459
PPA2	92,3	98.8%	91.7%	?Sudden cardiac failure, alcohol-induced, 617223 Sudden cardiac failure, infantile, 617222
PPARG	135,9	100.0%	99.9%	Carotid intimal medial thickness 1, 609338 Insulin resistance, severe, digenic, 604367 Lipodystrophy, familial partial, type 3, 604367 Obesity, severe, 601665 [Obesity, resistance to], 0 {Diabetes, type 2}, 125853
PPCS	148,5	100.0%	99.3%	Cardiomyopathy, dilated, 2C, 618189
PPIB	106,9	100.0%	100.0%	Osteogenesis imperfecta, type IX, 259440
PPM1D	170,2	100.0%	99.6%	Breast cancer, somatic, 114480 Intellectual developmental disorder with gastrointestinal difficulties and high pain threshold, 617450
PPM1K	132,5	100.0%	100.0%	?Maple syrup urine disease, mild variant, 615135
PPOX	95,2	99.8%	97.5%	Porphyria variegata, 176200
PPP1CB	113,1	100.0%	99.1%	Noonan syndrome-like disorder with loose anagen hair 2, 617506
PPP1R15B	124	100.0%	99.9%	Microcephaly, short stature, and impaired glucose metabolism 2, 616817
PPP1R3A	150,8	100.0%	99.7%	Insulin resistance, severe, digenic, 125853
PPP2CA	161	100.0%	99.9%	Neurodevelopmental disorder and language delay with or without structural brain abnormalities, 618354
PPP2R1A	129,1	91.6%	91.6%	Mental retardation, autosomal dominant 36, 616362
PPP2R1B	135,6	100.0%	99.8%	Lung cancer, somatic, 211980
PPP2R2B	124,2	99.9%	98.4%	Spinocerebellar ataxia 12, 604326
PPP2R5D	136,6	100.0%	100.0%	Mental retardation, autosomal dominant 35, 616355
PPP3CA	121,2	99.6%	97.5%	Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265 Epileptic encephalopathy, infantile or early childhood, 1, 617711
PPT1	136,6	90.2%	89.2%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	163,5	100.0%	100.0%	Renpenning syndrome, 309500
PRCC	152,6	100.0%	99.9%	Renal cell carcinoma, papillary, 605074
PRCD	95,6	100.0%	100.0%	Retinitis pigmentosa 36, 610599
PRDM12	138,9	92.6%	90.4%	Neuropathy, hereditary sensory and autonomic, type VIII, 616488
PRDM16	205,4	99.9%	99.1%	Cardiomyopathy, dilated, 1LL, 615373 Left ventricular noncompaction 8, 615373
PRDM5	137,8	99.6%	98.7%	Brittle cornea syndrome 2, 614170
PRDM6	110,6	99.9%	99.0%	Patent ductus arteriosus 3, 617039

PRDM8	116,4	99.7%	95.9%	?Epilepsy, progressive myoclonic, 10, 616640
PRDX1	95	100.0%	99.7%	Methylmalonic aciduria and homocystinuria, cblC type, digenic, 277400
PREPL	103,9	99.7%	97.7%	Myasthenic syndrome, congenital, 22, 616224
PRF1	138,1	91.2%	90.6%	Aplastic anemia, 609135 Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027
PRG4	131,1	99.2%	92.4%	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250
PRICKLE1	100	100.0%	99.8%	Epilepsy, progressive myoclonic 1B, 612437
PRIMPOL	118,1	97.7%	94.6%	Myopia 22, autosomal dominant, 615420
PRKACA	101,9	80.2%	79.5%	Cushing syndrome, ACTH-independent adrenal, somatic, 615830
PRKACG	204,5	100.0%	100.0%	?Bleeding disorder, platelet-type, 19, 616176
PRKAG2	129,9	99.7%	97.4%	Cardiomyopathy, hypertrophic 6, 600858 Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200
PRKAR1A	79,4	98.6%	92.6%	Acrodysostosis 1, with or without hormone resistance, 101800 Adrenocortical tumor, somatic, 0 Carney complex, type 1, 160980 Myxoma, intracardiac, 255960 Pigmented nodular adrenocortical disease, primary, 1, 610489
PRKCA	126,7	100.0%	100.0%	Pituitary tumor, invasive, 0
PRKCD	164,7	100.0%	100.0%	Autoimmune lymphoproliferative syndrome, type III, 615559
PRKCG	129,8	100.0%	99.3%	Spinocerebellar ataxia 14, 605361
PRKCSH	152,6	99.5%	94.8%	Polycystic liver disease 1, 174050
PRKD1	138,7	99.6%	99.5%	Congenital heart defects and ectodermal dysplasia, 617364
PRKDC	97,9	99.3%	96.5%	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PRKG1	125,4	99.7%	98.4%	Aortic aneurysm, familial thoracic 8, 615436
PRKN	82,1	79.9%	78.1%	Adenocarcinoma of lung, somatic, 211980 Ovarian cancer, somatic, 167000 Parkinson disease, juvenile, type 2, 600116
PRKRA	190,7	100.0%	100.0%	Dystonia 16, 612067
PRLR	129,5	100.0%	99.7%	Hyperprolactinemia, 615555 Multiple fibroadenomas of the breast, 615554
PRMT7	119,1	100.0%	99.9%	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157
PRNP	119	100.0%	100.0%	Cerebral amyloid angiopathy, PRNP-related, 137440 Creutzfeldt-Jakob disease, 123400 Gerstmann-Straussler disease, 137440

				Huntington disease-like 1, 603218 Insomnia, fatal familial, 600072 Prion disease with protracted course, 606688 {Kuru, susceptibility to}, 245300
PROC	142,6	100.0%	100.0%	Thrombophilia due to protein C deficiency, autosomal dominant, 176860 Thrombophilia due to protein C deficiency, autosomal recessive, 612304
PRODH	81,8	89.0%	81.7%	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850
PROK2	117,4	100.0%	99.9%	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	223	100.0%	100.0%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROM1	107,4	97.8%	95.8%	Cone-rod dystrophy 12, 612657 Macular dystrophy, retinal, 2, 608051 Retinitis pigmentosa 41, 612095 Stargardt disease 4, 603786
PROP1	96,9	92.5%	83.7%	Pituitary hormone deficiency, combined, 2, 262600
PROS1	95,8	97.7%	92.7%	Thrombophilia due to protein S deficiency, autosomal dominant, 612336 Thrombophilia due to protein S deficiency, autosomal recessive, 614514
PRPF3	73,2	98.4%	94.8%	Retinitis pigmentosa 18, 601414
PRPF31	117,4	99.8%	97.1%	Retinitis pigmentosa 11, 600138
PRPF4	124,9	100.0%	99.5%	Retinitis pigmentosa 70, 615922
PRPF6	112,3	100.0%	99.6%	Retinitis pigmentosa 60, 613983
PRPF8	103,4	99.9%	98.9%	Retinitis pigmentosa 13, 600059
PRPH2	203	100.0%	100.0%	Choroidal dystrophy, central areolar 2, 613105 Leber congenital amaurosis 18, 608133 Macular dystrophy, patterned, 1, 169150 Macular dystrophy, vitelliform, 3, 608161 Retinitis pigmentosa 7 and digenic form, 608133 Retinitis punctata albescens, 136880
PRPS1	111,6	100.0%	99.9%	Arts syndrome, 301835 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Deafness, X-linked 1, 304500 Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661
PRRT2	111,8	100.0%	99.0%	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751
PRRX1	100,4	100.0%	99.8%	Agnathia-otocephaly complex, 202650

PRSS1	141,4	100.0%	99.9%	Pancreatitis, hereditary, 167800 Trypsinogen deficiency, 614044
PRSS12	137,8	100.0%	99.6%	Mental retardation, autosomal recessive 1, 249500
PRSS56	95,4	100.0%	99.2%	Microphthalmia, isolated 6, 613517
PRUNE1	118	100.0%	99.3%	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481
PRX	156,5	100.0%	99.9%	Charcot-Marie-Tooth disease, type 4F, 614895 Dejerine-Sottas disease, 145900
PSAP	98,1	100.0%	99.3%	Combined SAP deficiency, 611721 Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PSAT1	42,8	90.3%	72.5%	?Phosphoserine aminotransferase deficiency, 610992 Neu-Laxova syndrome 2, 616038
PSEN1	131,5	100.0%	100.0%	?Acne inversa, familial, 3, 613737 Alzheimer disease, type 3, 607822 Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 Cardiomyopathy, dilated, 1U, 613694 Dementia, frontotemporal, 600274 Pick disease, 172700
PSEN2	109,9	100.0%	99.9%	Alzheimer disease-4, 606889 Cardiomyopathy, dilated, 1V, 613697
PSENEN	90,1	100.0%	100.0%	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736
PSMB4	119,2	100.0%	99.9%	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591
PSMB8	113,5	100.0%	98.8%	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040
PSMB9	81,5	99.9%	97.7%	?Proteasome-associated autoinflammatory syndrome 3, digenic, 617591
PSMC3IP	104,9	100.0%	100.0%	Ovarian dysgenesis 3, 614324
PSMD12	85,5	98.5%	90.1%	Stankiewicz-Isidor syndrome, 617516
PSPH	126,6	100.0%	99.8%	Phosphoserine phosphatase deficiency, 614023
PSTPIP1	103,8	99.9%	98.5%	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416
PTCH1	110,2	99.9%	98.4%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly 7, 610828
PTCH2	120,3	99.9%	98.7%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Medulloblastoma, somatic, 155255
PTDSS1	112	100.0%	99.9%	Lenz-Majewski hyperostotic dwarfism, 151050

PTEN	129,7	99.6%	97.0%	Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309 Prostate cancer, somatic, 176807 {Glioma susceptibility 2}, 613028 {Meningioma}, 607174
PTF1A	120,6	99.9%	98.2%	Pancreatic agenesis 2, 615935 Pancreatic and cerebellar agenesis, 609069
PTGIS	113,7	99.5%	96.4%	Hypertension, essential, 145500
PTH	94,4	99.7%	96.8%	Hypoparathyroidism, autosomal dominant, 146200 Hypoparathyroidism, autosomal recessive, 146200
PTH1R	106,6	100.0%	99.1%	Chondrodysplasia, Blomstrand type, 215045 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Metaphyseal chondrodysplasia, Murk Jansen type, 156400
PTHLH	127,2	98.4%	90.3%	Brachydactyly, type E2, 613382
PTPN11	78,3	98.6%	90.7%	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
PTPN12	148	99.2%	96.9%	Colon cancer, somatic, 114500
PTPN14	159,2	99.3%	96.8%	Choanal atresia and lymphedema, 613611
PTPRC	100,6	98.7%	93.9%	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971 {Hepatitis C virus, susceptibility to}, 609532
PTPRF	154,8	100.0%	99.9%	?Breasts and/or nipples, aplasia or hypoplasia of, 2, 616001
PTPRJ	137,4	97.7%	96.3%	Colon cancer, somatic, 114500
PTPRO	128,1	99.9%	99.4%	Nephrotic syndrome, type 6, 614196
PTPRQ	102,5	94.5%	92.3%	Deafness, autosomal dominant 73, 617663 Deafness, autosomal recessive 84A, 613391
PTRH2	200,8	100.0%	100.0%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PTS	101,5	99.8%	98.4%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUF60	163,3	100.0%	99.4%	Verheij syndrome, 615583
PUM1	126,9	100.0%	99.5%	Spinocerebellar ataxia 47, 617931
PURA	207,1	99.5%	96.9%	Mental retardation, autosomal dominant 31, 616158
PUS1	113,3	99.8%	97.5%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PUS3	158,3	100.0%	100.0%	Mental retardation, autosomal recessive 55, 617051
PUS7	135,4	100.0%	99.6%	Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342

PXDN	138,4	100.0%	99.6%	Anterior segment dysgenesis 7, with sclerocornea, 269400
PYCR1	96	99.7%	97.4%	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
PYCR2	116,5	99.7%	96.9%	Leukodystrophy, hypomyelinating, 10, 616420
PYGL	141	100.0%	100.0%	Glycogen storage disease VI, 232700
PYGM	121,1	100.0%	99.9%	McArdle disease, 232600
PYROXD1	48,6	93.0%	78.5%	Myopathy, myofibrillar, 8, 617258
QARS	129,2	100.0%	99.8%	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	97,9	100.0%	99.2%	Hyperphenylalaninemia, BH4-deficient, C, 261630
QRICH1	133	100.0%	99.4%	Ververi-Brady syndrome, 617982
QRICH2	113,4	95.2%	93.7%	Spermatogenic failure 35, 618341
RAB11B	212,7	100.0%	100.0%	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807
RAB18	83,4	99.7%	97.2%	Warburg micro syndrome 3, 614222
RAB23	107,4	100.0%	99.2%	Carpenter syndrome, 201000
RAB27A	126,1	100.0%	99.8%	Griscelli syndrome, type 2, 607624
RAB28	66,6	98.9%	92.3%	Cone-rod dystrophy 18, 615374
RAB33B	191,3	100.0%	100.0%	Smith-McCort dysplasia 2, 615222
RAB39B	102	100.0%	99.9%	?Waisman syndrome, 311510 Mental retardation, X-linked 72, 300271
RAB3GAP1	121,7	99.4%	98.9%	Warburg micro syndrome 1, 600118
RAB3GAP2	91,6	99.7%	96.9%	Martsolf syndrome, 212720 Warburg micro syndrome 2, 614225
RAB7A	115,3	100.0%	99.9%	Charcot-Marie-Tooth disease, type 2B, 600882
RAC1	101,1	99.6%	94.4%	Mental retardation, autosomal dominant 48, 617751
RAC2	100,6	100.0%	99.4%	Neutrophil immunodeficiency syndrome, 608203
RAD21	83	97.8%	93.4%	?Mungan syndrome, 611376 Cornelia de Lange syndrome 4, 614701
RAD50	102	97.5%	91.1%	Nijmegen breakage syndrome-like disorder, 613078
RAD51	100,6	89.4%	89.4%	?Fanconi anemia, complementation group R, 617244 Mirror movements 2, 614508 {Breast cancer, susceptibility to}, 114480
RAD51C	140,6	99.9%	99.5%	Fanconi anemia, complementation group O, 613390 {Breast-ovarian cancer, familial, susceptibility to, 3}, 613399
RAD54B	113,6	99.0%	96.1%	Colon cancer, somatic, 114500 Lymphoma, non-Hodgkin, somatic, 605027
RAD54L	97,8	100.0%	99.0%	Adenocarcinoma, colonic, somatic, 0

				Lymphoma, non-Hodgkin, somatic, 605027 {Breast cancer, invasive ductal}, 114480
RAF1	108,3	100.0%	99.9%	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553
RAG1	150,9	100.0%	100.0%	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 Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457
RAG2	186,2	100.0%	100.0%	Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457
RAI1	194,4	100.0%	100.0%	Smith-Magenis syndrome, 182290
RAP1GDS1	97,9	99.0%	94.4%	Lymphocytic leukemia, acute T-cell, 0
RAPGEF2	143	99.8%	98.9%	?Epilepsy, familial adult myoclonic, 7, 618075
RAPSN	149	99.8%	97.7%	Fetal akinesia deformation sequence 2, 618388 Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326
RARB	93,2	100.0%	100.0%	Microphthalmia, syndromic 12, 615524
RARS	93,4	93.6%	90.0%	Leukodystrophy, hypomyelinating, 9, 616140
RARS2	104	100.0%	99.4%	Pontocerebellar hypoplasia, type 6, 611523
RASA1	109,8	98.5%	95.7%	Basal cell carcinoma, somatic, 605462 Capillary malformation-arteriovenous malformation 1, 608354
RASGRP2	102,5	100.0%	99.7%	?Bleeding disorder, platelet-type, 18, 615888
RAX	135,9	99.9%	98.5%	Microphthalmia, isolated 3, 611038
RAX2	89,2	100.0%	99.9%	?Macular degeneration, age-related, 6, 613757 Cone-rod dystrophy 11, 610381
RB1	89,8	97.8%	93.1%	Bladder cancer, somatic, 109800 Osteosarcoma, somatic, 259500 Retinoblastoma, 180200 Retinoblastoma, trilateral, 180200 Small cell cancer of the lung, somatic, 182280
RB1CC1	109,3	99.4%	96.3%	Breast cancer, somatic, 114480
RBBP8	120,6	99.9%	99.3%	Jawad syndrome, 251255 Pancreatic carcinoma, somatic, 0 Seckel syndrome 2, 606744
RBCK1	107,9	100.0%	99.2%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895

RBM10	111,6	99.8%	97.6%	TARP syndrome, 311900
RBM20	177,6	100.0%	99.7%	Cardiomyopathy, dilated, 1DD, 613172
RBM28	130,1	100.0%	99.9%	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RBM8A	87,4	99.8%	97.4%	Thrombocytopenia-absent radius syndrome, 274000
RBMX	40,9	92.0%	74.4%	?Mental retardation, X-linked, syndromic 11, Shashi type, 300238
RBP3	150	100.0%	100.0%	?Retinitis pigmentosa 66, 615233
RBP4	137,9	99.2%	95.8%	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RBPJ	70,7	96.3%	87.0%	Adams-Oliver syndrome 3, 614814
RCBTB1	95,9	99.8%	99.3%	Retinal dystrophy with or without extraocular anomalies, 617175
RD3	165,7	100.0%	100.0%	Leber congenital amaurosis 12, 610612
RDH11	93	99.9%	98.5%	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108
RDH12	80,7	99.8%	97.2%	Leber congenital amaurosis 13, 612712
RDH5	167,7	100.0%	100.0%	Fundus albipunctatus, 136880
RDX	38,4	88.0%	69.1%	Deafness, autosomal recessive 24, 611022
RECQL4	159,9	100.0%	99.8%	Baller-Gerold syndrome, 218600 RAPADILINO syndrome, 266280 Rothmund-Thomson syndrome, 268400
REEP1	71,5	78.6%	76.2%	?Neuronopathy, distal hereditary motor, type VB, 614751 Spastic paraplegia 31, autosomal dominant, 610250
REEP2	173,9	100.0%	99.3%	?Spastic paraplegia 72, autosomal dominant, 615625 ?Spastic paraplegia 72, autosomal recessive, 615625
REEP6	206,2	100.0%	99.7%	Retinitis pigmentosa 77, 617304
RELA	97,8	99.8%	99.2%	?Mucocutaneous ulceration, chronic, 618287
RELB	108	99.2%	92.7%	?Immunodeficiency 53, 617585
RELN	130,1	100.0%	99.6%	Lissencephaly 2 (Norman-Roberts type), 257320 {Epilepsy, familial temporal lobe, 7}, 616436
RELT	141,4	100.0%	99.9%	Amelogenesis imperfecta, type IIIC, 618386
REN	127,5	100.0%	100.0%	Hyperuricemic nephropathy, familial juvenile 2, 613092 Renal tubular dysgenesis, 267430 [Hyperproreninemia], 0
REPS1	121,5	99.0%	96.4%	?Neurodegeneration with brain iron accumulation 7, 617916
RERE	77,6	96.5%	93.2%	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975
REST	115,5	98.5%	98.4%	Fibromatosis, gingival, 5, 617626 {Wilms tumor 6, susceptibility to}, 616806
RET	136,8	100.0%	99.2%	Central hypoventilation syndrome, congenital, 209880

				Medullary thyroid carcinoma, 155240 Multiple endocrine neoplasia IIA, 171400 Multiple endocrine neoplasia IIB, 162300 Pheochromocytoma, 171300 {Hirschsprung disease, protection against}, 142623 {Hirschsprung disease, susceptibility to, 1}, 142623
RETREG1	126,9	99.7%	98.8%	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
RFC1	125,7	99.9%	98.6%	Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome, 614575
RFT1	105,7	100.0%	99.2%	Congenital disorder of glycosylation, type In, 612015
RFWD3	103,7	100.0%	99.5%	?Fanconi anemia, complementation group W, 617784
RFX5	109	99.8%	97.5%	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFX6	147	99.9%	99.7%	Mitchell-Riley syndrome, 615710
RFXANK	123,1	100.0%	99.3%	MHC class II deficiency, complementation group B, 209920
RFXAP	116,5	100.0%	99.3%	Bare lymphocyte syndrome, type II, complementation group D, 209920
RGR	120,8	100.0%	99.1%	Retinitis pigmentosa 44, 613769
RGS9	107,8	98.7%	96.7%	Bradyopsia, 608415
RGS9BP	143,6	100.0%	100.0%	Bradyopsia, 608415
RHAG	122	100.0%	98.1%	Anemia, hemolytic, Rh-null, regulator type, 268150 Overhydrated hereditary stomatocytosis, 185000
RHBDF2	105,1	99.9%	98.9%	Tylosis with esophageal cancer, 148500
RHCE	151,2	98.1%	97.8%	Rh-null disease, amorph type, 617970 [Blood group, Rhesus], 0
RHO	165,3	100.0%	100.0%	Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 Retinitis punctata albescens, 136880
RHOBTB2	190,5	100.0%	99.9%	Epileptic encephalopathy, early infantile, 64, 618004
RIMS1	126	99.8%	98.6%	Cone-rod dystrophy 7, 603649
RIN2	119,5	100.0%	99.6%	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075
RIPK1	103,6	99.9%	98.7%	Immunodeficiency 57, 618108
RIPK4	167,5	100.0%	100.0%	CHAND syndrome, 214350 Popliteal pterygium syndrome, Bartsocas-Papas type, 263650
RIPOR2	111,5	100.0%	99.8%	?Deafness, autosomal recessive 104, 616515
RIPPLY2	78,7	100.0%	98.7%	?Spondylocostal dysostosis 6, 616566
RIT1	139,2	100.0%	100.0%	Noonan syndrome 8, 615355
RLBP1	120,4	100.0%	99.7%	Bothnia retinal dystrophy, 607475

				Fundus albipunctatus, 136880 Newfoundland rod-cone dystrophy, 607476 Retinitis punctata albescens, 136880
RLIM	98	99.5%	97.7%	Tonne-Kalscheuer syndrome, 300978
RMND1	132,6	100.0%	99.0%	Combined oxidative phosphorylation deficiency 11, 614922
RMRP	NC	NC	NC	Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RNASEH1	101,1	97.6%	92.5%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479
RNASEH2A	129,8	100.0%	99.7%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	100,8	98.9%	95.2%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	281,7	100.0%	100.0%	Aicardi-Goutieres syndrome 3, 610329
RNASEL	122,9	100.0%	99.7%	Prostate cancer 1, 601518
RNASET2	102,2	95.4%	90.2%	Leukoencephalopathy, cystic, without megalencephaly, 612951
RNF113A	134,9	100.0%	100.0%	?Trichothiodystrophy 5, nonphotosensitive, 300953
RNF125	175,4	99.9%	99.2%	Tenorio syndrome, 616260
RNF13	80,3	92.0%	79.0%	Epileptic encephalopathy, early infantile, 73, 618379
RNF139	172,1	100.0%	100.0%	Renal cell carcinoma, 144700
RNF168	182	100.0%	99.6%	RIDDLE syndrome, 611943
RNF170	126,6	99.7%	97.7%	Ataxia, sensory, 1, autosomal dominant, 608984
RNF212	107,5	99.9%	98.9%	Recombination rate QTL 1, 612042
RNF216	125	99.9%	98.1%	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840
RNF43	145,9	100.0%	98.8%	Sessile serrated polyposis cancer syndrome, 617108
RNF6	137,1	99.8%	98.6%	Esophageal carcinoma, somatic, 133239
RNPC3	41,6	88.8%	68.7%	?Growth hormone deficiency, isolated, type V, 618160
RNU4ATAC	NC	NC	NC	Microcephalic osteodysplastic primordial dwarfism, type I, 210710 Roifman syndrome, 616651
ROBO2	127,4	98.8%	97.5%	Vesicoureteral reflux 2, 610878
ROBO3	104,3	99.4%	96.9%	Gaze palsy, familial horizontal, with progressive scoliosis, 1, 607313
ROGDI	127,6	100.0%	99.4%	Kohlschutter-Tonz syndrome, 226750
ROM1	126,1	100.0%	99.9%	Retinitis pigmentosa 7, digenic form, 608133
ROR1	149,1	99.1%	97.2%	?Deafness, autosomal recessive 108, 617654
ROR2	160,6	100.0%	99.7%	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RORA	103,6	96.8%	91.2%	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060
RORC	118,6	100.0%	100.0%	Immunodeficiency 42, 616622

RP1	109,4	91.4%	90.8%	Retinitis pigmentosa 1, 180100
RP1L1	135,2	100.0%	100.0%	Occult macular dystrophy, 613587
RP2	155,8	100.0%	99.8%	Retinitis pigmentosa 2, 312600
RP9	64,5	91.4%	78.5%	?Retinitis pigmentosa 9, 180104
RPE65	131,9	100.0%	99.8%	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794
RPGR	81,3	82.4%	75.5%	Cone-rod dystrophy, X-linked, 1, 304020 Macular degeneration, X-linked atrophic, 300834 Retinitis pigmentosa 3, 300029 Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455
RPGRIP1	128	100.0%	99.7%	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826
RPGRIP1L	123,4	96.7%	95.4%	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561
RPIA	113	100.0%	98.8%	?Ribose 5-phosphate isomerase deficiency, 608611
RPL10	65,6	97.1%	86.9%	Mental retardation, X-linked, syndromic, 35, 300998 {Autism, susceptibility to, X-linked 5}, 300847
RPL11	85,4	100.0%	99.3%	Diamond-Blackfan anemia 7, 612562
RPL15	32	87.2%	72.1%	?Diamond-Blackfan anemia 12, 615550
RPL18	89,3	100.0%	99.5%	?Diamond-Blackfan anemia 18, 618310
RPL21	54,9	84.6%	64.0%	Hypotrichosis 12, 615885
RPL26	31	91.7%	68.9%	?Diamond-Blackfan anemia 11, 614900
RPL27	32,5	72.7%	54.6%	?Diamond-Blackfan anemia 16, 617408
RPL35	62,5	89.5%	75.8%	?Diamond-Blackfan anemia 19, 618312
RPL35A	75,4	96.4%	84.6%	Diamond-Blackfan anemia 5, 612528
RPL5	34,7	85.0%	67.7%	Diamond-Blackfan anemia 6, 612561
RPS10	91,8	98.8%	91.8%	Diamond-Blackfan anemia 9, 613308
RPS14	106	99.7%	93.8%	Macrocytic anemia, refractory, due to 5q deletion, somatic, 153550
RPS15A	58,3	97.1%	86.3%	?Diamond-Blackfan anemia 20, 618313
RPS17	38,2	87.0%	68.9%	Diamond-Blackfan anemia 4, 612527
RPS19	76,7	99.9%	96.6%	Diamond-Blackfan anemia 1, 105650
RPS23	59,4	88.9%	78.9%	Brachycephaly, trichomegaly, and developmental delay, 617412
RPS24	84,5	95.2%	89.7%	Diamond-blackfan anemia 3, 610629
RPS26	75,9	89.2%	75.8%	Diamond-Blackfan anemia 10, 613309
RPS27	34,4	89.5%	57.5%	?Diamond-Blackfan anemia 17, 617409

RPS28	54,1	99.7%	95.0%	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164
RPS29	90,8	98.7%	94.6%	Diamond-Blackfan anemia 13, 615909
RPS6KA3	87,8	98.3%	93.0%	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844
RPS7	76,6	84.8%	70.0%	Diamond-Blackfan anemia 8, 612563
RPSA	64	100.0%	99.4%	Asplenia, isolated congenital, 271400
RRAS2	82,6	92.4%	81.0%	Ovarian carcinoma, 0
RRM2B	143,9	99.9%	99.4%	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077
RS1	51,4	98.7%	87.3%	Retinoschisis, 312700
RSPH1	122,6	100.0%	99.9%	Ciliary dyskinesia, primary, 24, 615481
RSPH3	139,5	99.9%	99.3%	Ciliary dyskinesia, primary, 32, 616481
RSPH4A	146,9	98.1%	95.3%	Ciliary dyskinesia, primary, 11, 612649
RSPH9	131,2	99.7%	97.1%	Ciliary dyskinesia, primary, 12, 612650
RSPO1	103,8	100.0%	99.9%	Palmoplantar hyperkeratosis and true hermaphroditism, 610644 Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644
RSPO2	136,4	96.6%	90.1%	?Humerofemoral hypoplasia with radiotibial ray deficiency, 618022 Tetraamelia syndrome 2, 618021
RSPO4	144,4	100.0%	100.0%	Anonychia congenita, 206800
RSPRY1	142	100.0%	99.9%	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723
RSRC1	75,4	99.6%	95.4%	Intellectual developmental disorder, autosomal recessive 70, 618402
RTEL1	131,1	99.7%	97.7%	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373
RTN2	140,8	99.9%	98.7%	Spastic paraplegia 12, autosomal dominant, 604805
RTN4IP1	79,6	100.0%	98.0%	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
RTTN	117,9	98.8%	97.4%	Microcephaly, short stature, and polymicrogyria with seizures, 614833
RUBCN	99,3	99.9%	99.1%	?Spinocerebellar ataxia, autosomal recessive 15, 615705
RUNX1	84,6	99.6%	96.3%	Leukemia, acute myeloid, 601626 Platelet disorder, familial, with associated myeloid malignancy, 601399
RUNX2	102,8	73.4%	72.2%	Cleidocranial dysplasia, 119600 Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600 Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510
RUSC2	188,1	100.0%	100.0%	Mental retardation, autosomal recessive 61, 617773

RYR1	117,1	98.7%	95.7%	Central core disease, 117000 King-Denborough syndrome, 145600 Minicore myopathy with external ophthalmoplegia, 255320 Neuromuscular disease, congenital, with uniform type 1 fiber, 117000 {Malignant hyperthermia susceptibility 1}, 145600
RYR2	124,9	99.9%	98.9%	Arrhythmogenic right ventricular dysplasia 2, 600996 Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772
S1PR2	200,8	99.5%	96.8%	Deafness, autosomal recessive 68, 610419
SACS	150,4	100.0%	99.9%	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAG	127	100.0%	99.9%	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758
SALL1	113,3	99.9%	98.9%	Townes-Brocks branchiootorenal-like syndrome, 107480 Townes-Brocks syndrome 1, 107480
SALL2	128,4	100.0%	100.0%	?Coloboma, ocular, autosomal recessive, 216820
SALL4	135	99.9%	98.1%	Duane-radial ray syndrome, 607323 IVIC syndrome, 147750
SAMD12	139,2	100.0%	100.0%	Epilepsy, familial adult myoclonic, 1, 601068
SAMD9	163,9	100.0%	99.9%	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455
SAMD9L	171,8	100.0%	100.0%	Ataxia-pancytopenia syndrome, 159550
SAMHD1	133,4	99.8%	98.5%	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SAR1B	120,9	97.2%	90.5%	Chylomicron retention disease, 246700
SARS	107,6	99.9%	98.8%	?Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709
SARS2	117,9	95.1%	93.2%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SASH1	152,1	99.3%	97.8%	?Cancer, alopecia, pigment dyscrasia, onychodystrophy, and keratoderma, 618373 Dyschromatosis universalis hereditaria 1, 127500
SASS6	77,6	99.6%	97.6%	?Microcephaly 14, primary, autosomal recessive, 616402
SATB2	107,4	99.8%	97.7%	Glass syndrome, 612313
SBDS	166,2	100.0%	100.0%	Shwachman-Diamond syndrome, 260400 {Aplastic anemia, susceptibility to}, 609135
SBF1	122	99.4%	97.9%	Charcot-Marie-Tooth disease, type 4B3, 615284
SBF2	107,7	99.9%	99.0%	Charcot-Marie-Tooth disease, type 4B2, 604563
SC5D	153,6	99.8%	99.3%	Lathosterolosis, 607330
SCAPER	138,7	98.2%	96.4%	Intellectual developmental disorder and retinitis pigmentosa, 618195
SCARB2	105,8	99.8%	99.1%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900

SCARF2	104,8	99.5%	95.7%	Van den Ende-Gupta syndrome, 600920
SCN10A	133,3	100.0%	99.4%	Episodic pain syndrome, familial, 2, 615551
SCN11A	122,1	99.3%	97.1%	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN1A	121,4	100.0%	99.1%	Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Epileptic encephalopathy, early infantile, 6 (Dravet syndrome), 607208 Febrile seizures, familial, 3A, 604403 Migraine, familial hemiplegic, 3, 609634
SCN1B	169,7	99.9%	98.1%	Atrial fibrillation, familial, 13, 615377 Brugada syndrome 5, 612838 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233 Epileptic encephalopathy, early infantile, 52, 617350
SCN2A	132,4	99.6%	97.7%	Epileptic encephalopathy, early infantile, 11, 613721 Seizures, benign familial infantile, 3, 607745
SCN2B	176,4	100.0%	100.0%	Atrial fibrillation, familial, 14, 615378
SCN3A	138,7	99.9%	99.1%	Epilepsy, familial focal, with variable foci 4, 617935 Epileptic encephalopathy, early infantile, 62, 617938
SCN3B	137,7	100.0%	100.0%	Atrial fibrillation, familial, 16, 613120 Brugada syndrome 7, 613120
SCN4A	167,9	99.8%	99.3%	Hyperkalemic periodic paralysis, type 2, 170500 Hypokalemic periodic paralysis, type 2, 613345 Myasthenic syndrome, congenital, 16, 614198 Myotonia congenita, atypical, acetazolamide-responsive, 608390 Paramyotonia congenita, 168300
SCN4B	66	99.8%	97.7%	Atrial fibrillation, familial, 17, 611819 Long QT syndrome-10, 611819
SCN5A	141,1	99.0%	99.0%	Atrial fibrillation, familial, 10, 614022 Brugada syndrome 1, 601144 Cardiomyopathy, dilated, 1E, 601154 Heart block, nonprogressive, 113900 Heart block, progressive, type IA, 113900 Long QT syndrome-3, 603830 Sick sinus syndrome 1, 608567 Ventricular fibrillation, familial, 1, 603829 {Sudden infant death syndrome, susceptibility to}, 272120
SCN8A	154,3	100.0%	99.7%	?Myoclonus, familial, 2, 618364 Cognitive impairment with or without cerebellar ataxia, 614306

				Epileptic encephalopathy, early infantile, 13, 614558 Seizures, benign familial infantile, 5, 617080
SCN9A	128,4	99.1%	97.7%	Epilepsy, generalized, with febrile seizures plus, type 7, 613863 Erythermalgia, primary, 133020 Febrile seizures, familial, 3B, 613863 HSAN2D, autosomal recessive, 243000 Insensitivity to pain, congenital, 243000 Paroxysmal extreme pain disorder, 167400 Small fiber neuropathy, 133020 {Dravet syndrome, modifier of}, 607208
SCNN1A	127	99.8%	98.3%	?Liddle syndrome 3, 618126 Bronchiectasis with or without elevated sweat chloride 2, 613021 Pseudohypoaldosteronism, type I, 264350
SCNN1B	130,6	100.0%	100.0%	Bronchiectasis with or without elevated sweat chloride 1, 211400 Liddle syndrome 1, 177200 Pseudohypoaldosteronism, type I, 264350
SCNN1G	142,2	99.5%	97.2%	Bronchiectasis with or without elevated sweat chloride 3, 613071 Liddle syndrome 2, 618114 Pseudohypoaldosteronism, type I, 264350
SCO1	100,1	99.8%	98.1%	Mitochondrial complex IV deficiency, 220110
SCO2	115,7	100.0%	99.9%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908
SCP2	107,8	99.7%	96.4%	?Leukoencephalopathy with dystonia and motor neuropathy, 613724
SCYL1	146,2	100.0%	100.0%	Spinocerebellar ataxia, autosomal recessive 21, 616719
SDCCAG8	124,1	100.0%	99.7%	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SDHA	88,9	85.1%	77.7%	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165
SDHAF1	83	100.0%	100.0%	Mitochondrial complex II deficiency, 252011
SDHAF2	127,7	95.6%	94.6%	Paragangliomas 2, 601650
SDHB	114,8	100.0%	99.9%	Gastrointestinal stromal tumor, 606764 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 4, 115310 Pheochromocytoma, 171300
SDHC	85,8	99.6%	95.3%	Gastrointestinal stromal tumor, 606764

				Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 3, 605373
SDHD	43,7	52.7%	50.6%	Mitochondrial complex II deficiency, 252011 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 1, with or without deafness, 168000 Pheochromocytoma, 171300
SDR9C7	168	100.0%	99.9%	Ichthyosis, congenital, autosomal recessive 13, 617574
SEC23A	122,6	99.7%	97.9%	Craniolenticulosutural dysplasia, 607812
SEC23B	131	99.8%	99.0%	?Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SEC24D	126,3	99.9%	99.5%	Cole-Carpenter syndrome 2, 616294
SEC61A1	121	100.0%	99.9%	Hyperuricemic nephropathy, familial juvenile, 4, 617056
SEC63	78,7	86.5%	80.0%	Polycystic liver disease 2, 617004
SECISBP2	108	99.7%	97.1%	Thyroid hormone metabolism, abnormal, 609698
SELENBP1	121,6	100.0%	99.8%	Extraoral halitosis due to MTO deficiency, 618148
SELENON	131	84.9%	83.9%	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310
SEMA3E	130,9	100.0%	99.6%	?CHARGE syndrome, 214800
SEMA4A	124,3	100.0%	99.3%	Cone-rod dystrophy 10, 610283 Retinitis pigmentosa 35, 610282
SEPSECS	159,6	100.0%	99.6%	Pontocerebellar hypoplasia type 2D, 613811
SEPT12	NC	NC	NC	Spermatogenic failure 10, 614822
SEPT9	NC	NC	NC	Amyotrophy, hereditary neuralgic, 162100 Leukemia, acute myeloid, therapy-related, 0 Ovarian carcinoma, 0
SERAC1	111	99.7%	99.0%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPINA1	104,9	100.0%	99.7%	Emphysema due to AAT deficiency, 613490 Emphysema-cirrhosis, due to AAT deficiency, 613490 Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963
SERPINA3	114,4	100.0%	99.9%	Alpha-1-antichymotrypsin deficiency, 0 Cerebrovascular disease, occlusive, 0
SERPINA6	138,3	100.0%	100.0%	Corticosteroid-binding globulin deficiency, 611489
SERPINB6	138,9	95.9%	95.9%	?Deafness, autosomal recessive 91, 613453
SERPINB7	124,3	100.0%	99.6%	Palmoplantar keratoderma, Nagashima type, 615598
SERPINB8	125,8	95.0%	95.0%	Peeling skin syndrome 5, 617115

SERPINC1	117,9	100.0%	100.0%	Thrombophilia due to antithrombin III deficiency, 613118
SERPIND1	134,8	100.0%	99.9%	Thrombophilia due to heparin cofactor II deficiency, 612356
SERPINE1	134	100.0%	100.0%	Plasminogen activator inhibitor-1 deficiency, 613329 {Transcription of plasminogen activator inhibitor, modulator of}, 0
SERPINF1	104	100.0%	99.9%	Osteogenesis imperfecta, type VI, 613982
SERPINF2	151,6	100.0%	99.9%	Alpha-2-plasmin inhibitor deficiency, 262850
SERPING1	96,7	99.5%	96.7%	Angioedema, hereditary, types I and II, 106100 Complement component 4, partial deficiency of, 120790
SERPINH1	195,8	100.0%	99.6%	Osteogenesis imperfecta, type X, 613848 {Preterm premature rupture of the membranes, susceptibility to}, 610504
SERPINI1	101,2	99.8%	97.4%	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218
SET	58,3	97.2%	88.8%	Mental retardation, autosomal dominant 58, 618106
SETBP1	122,7	98.8%	97.7%	Mental retardation, autosomal dominant 29, 616078 Schinzel-Giedion midface retraction syndrome, 269150
SETD2	137,5	100.0%	99.7%	Luscan-Lumish syndrome, 616831
SETD5	147,6	100.0%	99.7%	Mental retardation, autosomal dominant 23, 615761
SETX	151,6	100.0%	99.6%	Amyotrophic lateral sclerosis 4, juvenile, 602433 Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002
SF3B1	130,9	99.6%	98.3%	Myelodysplastic syndrome, somatic, 614286
SF3B4	75,5	99.9%	98.3%	Acrofacial dysostosis 1, Nager type, 154400
SFRP4	125,4	99.9%	99.0%	Pyle disease, 265900
SFTPA2	141,1	100.0%	100.0%	Pulmonary fibrosis, idiopathic, 178500
SFTPB	94,1	100.0%	99.5%	Surfactant metabolism dysfunction, pulmonary, 1, 265120
SFTPC	98,4	100.0%	99.1%	Surfactant metabolism dysfunction, pulmonary, 2, 610913
SFXN4	124	100.0%	99.7%	Combined oxidative phosphorylation deficiency 18, 615578
SGCA	158,4	100.0%	100.0%	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099
SGCB	140,1	99.3%	96.7%	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286
SGCD	78	99.8%	97.2%	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287
SGCE	97,7	96.0%	92.3%	Dystonia-11, myoclonic, 159900
SGCG	114,5	100.0%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700
SGO1	107,8	99.9%	98.9%	Chronic atrial and intestinal dysrhythmia, 616201
SGPL1	132,3	100.0%	100.0%	Nephrotic syndrome, type 14, 617575
SGSH	140,2	97.6%	94.7%	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SH2B3	108,4	99.9%	97.6%	Erythrocytosis, somatic, 133100 Myelofibrosis, somatic, 254450

				Thrombocythemia, somatic, 187950
SH2D1A	108,9	97.8%	92.4%	Lymphoproliferative syndrome, X-linked, 1, 308240
SH3BP2	139,3	91.9%	91.4%	Cherubism, 118400
SH3KBP1	90,2	99.0%	93.7%	?Immunodeficiency 61, 300310
SH3PXD2B	161,4	100.0%	99.9%	Frank-ter Haar syndrome, 249420
SH3TC2	102,1	100.0%	99.4%	Charcot-Marie-Tooth disease, type 4C, 601596 Mononeuropathy of the median nerve, mild, 613353
SHANK3	123,9	97.5%	91.6%	Phelan-McDermid syndrome, 606232 {Schizophrenia 15}, 613950
SHH	147,1	100.0%	100.0%	Holoprosencephaly 3, 142945 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250
SHOC2	139,6	99.9%	99.4%	Noonan-like syndrome with loose anagen hair, 607721
SHOX	35,9	82.5%	67.2%	Langer mesomelic dysplasia, 249700 Leri-Weill dyschondrosteosis, 127300 Short stature, idiopathic familial, 300582
SHROOM4	96,2	99.9%	98.7%	Stocco dos Santos X-linked mental retardation syndrome, 300434
SI	118,7	99.4%	95.9%	Sucrase-isomaltase deficiency, congenital, 222900
SIGMAR1	146,6	100.0%	100.0%	?Amyotrophic lateral sclerosis 16, juvenile, 614373 ?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726
SIK1	118,5	99.6%	96.7%	Epileptic encephalopathy, early infantile, 30, 616341
SIK3	97,2	99.8%	98.5%	?Spondyloepimetaphyseal dysplasia, Krakow type, 618162
SIL1	129,5	98.9%	96.2%	Marinesco-Sjogren syndrome, 248800
SIN3A	109,7	99.9%	98.3%	Witteveen-Kolk syndrome, 613406
SIPA1L3	171,8	100.0%	99.5%	?Cataract 45, 616851
SIX1	131,1	99.9%	98.7%	Branchiootic syndrome 3, 608389 Deafness, autosomal dominant 23, 605192
SIX3	206	100.0%	99.9%	Holoprosencephaly 2, 157170 Schizencephaly, 269160
SIX5	76,3	99.9%	97.3%	Branchiootorenal syndrome 2, 610896
SIX6	278,5	100.0%	100.0%	Optic disc anomalies with retinal and/or macular dystrophy, 212550
SKI	132,9	100.0%	99.3%	Shprintzen-Goldberg syndrome, 182212
SKIV2L	138,4	100.0%	99.7%	Trichohepatoenteric syndrome 2, 614602
SLC10A2	122,1	100.0%	100.0%	Bile acid malabsorption, primary, 613291
SLC10A7	111	99.9%	99.2%	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363

SLC11A2	96,2	99.9%	98.7%	Anemia, hypochromic microcytic, with iron overload 1, 206100
SLC12A1	144,2	100.0%	99.8%	Bartter syndrome, type 1, 601678
SLC12A3	140	100.0%	100.0%	Gitelman syndrome, 263800
SLC12A5	111,9	86.1%	84.1%	Epileptic encephalopathy, early infantile, 34, 616645 {Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685
SLC12A6	118,9	100.0%	99.9%	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC13A3	89,5	100.0%	99.5%	Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate, 618384
SLC13A5	141,9	100.0%	99.9%	Epileptic encephalopathy, early infantile, 25, 615905
SLC16A1	138,1	100.0%	99.2%	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia, familial, 7, 610021 Monocarboxylate transporter 1 deficiency, 616095
SLC16A12	128,5	100.0%	99.9%	Cataract 47, juvenile, with microcornea, 612018
SLC16A2	63,3	98.7%	91.0%	Allan-Herndon-Dudley syndrome, 300523
SLC17A5	137,7	99.8%	96.1%	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC17A8	121,5	100.0%	99.8%	Deafness, autosomal dominant 25, 605583
SLC17A9	140,1	95.8%	95.4%	Porokeratosis 8, disseminated superficial actinic type, 616063
SLC18A2	106,8	100.0%	99.8%	?Parkinsonism-dystonia, infantile, 2, 618049
SLC18A3	256,7	100.0%	100.0%	Myasthenic syndrome, congenital, 21, presynaptic, 617239
SLC19A2	101,3	100.0%	99.6%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	134,6	100.0%	99.9%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A1	145,6	100.0%	99.6%	Dicarboxylic aminoaciduria, 222730 {?Schizophrenia susceptibility 18}, 615232
SLC1A2	97,1	99.3%	97.2%	Epileptic encephalopathy, early infantile, 41, 617105
SLC1A3	99,5	100.0%	99.8%	Episodic ataxia, type 6, 612656
SLC1A4	146,4	100.0%	99.5%	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC20A2	108,6	100.0%	98.5%	Basal ganglia calcification, idiopathic, 1, 213600
SLC22A12	117,7	100.0%	99.8%	Hypouricemia, renal, 220150
SLC22A18	113,5	100.0%	99.5%	Breast cancer, somatic, 114480 Lung cancer, somatic, 211980 Rhabdomyosarcoma, somatic, 268210
SLC22A5	129,7	100.0%	100.0%	Carnitine deficiency, systemic primary, 212140
SLC24A1	167	100.0%	100.0%	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
SLC24A4	103,5	100.0%	99.8%	Amelogenesis imperfecta, type IIA5, 615887 [Skin/hair/eye pigmentation 6, blond/brown hair], 210750 [Skin/hair/eye pigmentation 6, blue/green eyes], 210750

SLC24A5	104,1	99.9%	99.3%	Albinism, oculocutaneous, type VI, 113750 [Skin/hair/eye pigmentation 4, fair/dark skin], 113750
SLC25A1	103,2	99.3%	95.1%	?Myasthenic syndrome, congenital, 23, presynaptic, 618197 Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A12	150,9	99.9%	99.9%	Epileptic encephalopathy, early infantile, 39, 612949
SLC25A13	120,1	99.8%	98.9%	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A15	146,8	97.9%	93.6%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A19	77,4	99.9%	97.8%	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A20	90,7	100.0%	100.0%	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A22	123,8	100.0%	99.1%	Epileptic encephalopathy, early infantile, 3, 609304
SLC25A24	128,9	99.6%	99.1%	Fontaine progeroid syndrome, 612289
SLC25A26	98,4	99.9%	99.3%	Combined oxidative phosphorylation deficiency 28, 616794
SLC25A3	129,8	99.5%	96.9%	Mitochondrial phosphate carrier deficiency, 610773
SLC25A32	128,2	100.0%	99.9%	?Exercise intolerance, riboflavin-responsive, 616839
SLC25A38	94,5	99.1%	95.2%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC25A4	130,9	100.0%	99.9%	Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184 Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283
SLC25A46	173	99.8%	98.3%	Neuropathy, hereditary motor and sensory, type VIB, 616505
SLC26A1	149,3	100.0%	100.0%	?Nephrolithiasis, calcium oxalate, 167030
SLC26A2	205,1	100.0%	99.9%	Achondrogenesis Ib, 600972 Atelosteogenesis, type II, 256050 De la Chapelle dysplasia, 256050 Diastrophic dysplasia, 222600 Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 Epiphyseal dysplasia, multiple, 4, 226900
SLC26A3	132,5	99.9%	99.2%	Diarrhea 1, secretory chloride, congenital, 214700
SLC26A4	113	100.0%	99.5%	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791 Pendred syndrome, 274600
SLC26A5	130,3	98.3%	95.4%	?Deafness, autosomal recessive 61, 613865
SLC26A8	112,3	99.9%	99.4%	Spermatogenic failure 3, 606766
SLC27A4	150,9	100.0%	100.0%	Ichthyosis prematurity syndrome, 608649
SLC29A3	173,3	100.0%	99.5%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC2A1	148,9	92.8%	92.8%	Dystonia 9, 601042

				GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 GLUT1 deficiency syndrome 2, childhood onset, 612126 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847
SLC2A10	152,6	98.0%	97.5%	Arterial tortuosity syndrome, 208050
SLC2A2	158,1	100.0%	99.9%	Fanconi-Bickel syndrome, 227810 {Diabetes mellitus, noninsulin-dependent}, 125853
SLC2A9	104,8	100.0%	98.7%	Hypouricemia, renal, 2, 612076 {Uric acid concentration, serum, QTL 2}, 612076
SLC30A10	176,1	100.0%	100.0%	Hypermanganesemia with dystonia 1, 613280
SLC30A2	120,7	100.0%	99.2%	Zinc deficiency, transient neonatal, 608118
SLC30A9	88,6	98.7%	93.1%	?Birk-Landau-Perez syndrome, 617595
SLC33A1	132	99.7%	97.7%	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539
SLC34A1	149,8	100.0%	99.9%	?Fanconi renotubular syndrome 2, 613388 Hypercalcemia, infantile, 2, 616963 Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286
SLC34A2	140,3	100.0%	99.7%	Pulmonary alveolar microlithiasis, 265100
SLC34A3	141,1	99.9%	99.0%	Hypophosphatemic rickets with hypercalciuria, 241530
SLC35A1	127,1	100.0%	99.4%	Congenital disorder of glycosylation, type IIf, 603585
SLC35A2	104,8	99.8%	98.1%	Congenital disorder of glycosylation, type IIm, 300896
SLC35A3	66,6	80.6%	78.3%	?Arthrogryposis, mental retardation, and seizures, 615553
SLC35C1	187,8	100.0%	99.8%	Congenital disorder of glycosylation, type IIc, 266265
SLC35D1	125	99.5%	97.2%	Schneckenbecken dysplasia, 269250
SLC36A2	100,3	100.0%	99.9%	Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC37A4	114,3	100.0%	99.6%	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240
SLC38A8	71,5	99.4%	95.5%	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218
SLC39A13	145,1	100.0%	99.9%	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350
SLC39A14	95,4	99.9%	97.9%	?Hyperostosis cranalis interna, 144755 Hypermanganesemia with dystonia 2, 617013
SLC39A4	114,2	100.0%	99.0%	Acrodermatitis enteropathica, 201100
SLC39A5	130,2	100.0%	99.4%	Myopia 24, autosomal dominant, 615946
SLC39A8	140,9	100.0%	99.8%	Congenital disorder of glycosylation, type IIn, 616721
SLC3A1	144,5	100.0%	99.4%	Cystinuria, 220100

SLC40A1	120,8	100.0%	99.8%	Hemochromatosis, type 4, 606069
SLC44A4	113,9	100.0%	99.4%	?Deafness, autosomal dominant 72, 617606
SLC45A1	141	100.0%	100.0%	Intellectual developmental disorder with neuropsychiatric features, 617532
SLC45A2	115,2	100.0%	99.8%	Albinism, oculocutaneous, type IV, 606574 [Skin/hair/eye pigmentation 5, black/nonblack hair], 227240 [Skin/hair/eye pigmentation 5, dark/fair skin], 227240 [Skin/hair/eye pigmentation 5, dark/light eyes], 227240
SLC46A1	111,1	99.9%	98.4%	Folate malabsorption, hereditary, 229050
SLC4A1	139,2	100.0%	99.8%	Cryohydrocytosis, 185020 Ovalocytosis, SA type, 166900 Renal tubular acidosis, distal, AD, 179800 Renal tubular acidosis, distal, AR, 611590 Spherocytosis, type 4, 612653 [Blood group, Diego], 110500 [Blood group, Froese], 601551 [Blood group, Swann], 601550 [Blood group, Waldner], 112010 [Blood group, Wright], 112050 [Malaria, resistance to], 611162
SLC4A11	157,4	100.0%	100.0%	Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy and perceptive deafness, 217400 Corneal endothelial dystrophy, autosomal recessive, 217700
SLC4A4	113,9	99.8%	98.3%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC52A1	198,3	100.0%	100.0%	Riboflavin deficiency, 615026
SLC52A2	185,4	100.0%	100.0%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	118,8	100.0%	99.8%	?Fazio-Londe disease, 211500 Brown-Vialetto-Van Laere syndrome 1, 211530
SLC5A1	110,7	100.0%	99.3%	Glucose/galactose malabsorption, 606824
SLC5A2	135	100.0%	100.0%	Renal glucosuria, 233100
SLC5A5	105,7	100.0%	99.9%	Thyroid dyshormonogenesis 1, 274400
SLC5A7	100,2	100.0%	99.9%	Myasthenic syndrome, congenital, 20, presynaptic, 617143 Neuronopathy, distal hereditary motor, type VIIA, 158580
SLC6A1	126	100.0%	100.0%	Myoclonic-atonic epilepsy, 616421
SLC6A17	149,8	100.0%	100.0%	Mental retardation, autosomal recessive 48, 616269
SLC6A19	129,3	100.0%	100.0%	Hartnup disorder, 234500 Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600

SLC6A2	122,3	100.0%	99.9%	?Orthostatic intolerance, 604715
SLC6A20	151,9	100.0%	99.8%	Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC6A3	133	100.0%	99.9%	Parkinsonism-dystonia, infantile, 1, 613135 {Nicotine dependence, protection against}, 188890
SLC6A5	128,7	100.0%	100.0%	Hyperekplexia 3, 614618
SLC6A8	53,5	96.1%	83.8%	Cerebral creatine deficiency syndrome 1, 300352
SLC6A9	148,8	100.0%	100.0%	Glycine encephalopathy with normal serum glycine, 617301
SLC7A14	145,3	100.0%	100.0%	Retinitis pigmentosa 68, 615725
SLC7A7	105,5	100.0%	99.6%	Lysinuric protein intolerance, 222700
SLC7A9	119,8	100.0%	98.8%	Cystinuria, 220100
SLC9A1	142,4	100.0%	100.0%	?Lichtenstein-Knorr syndrome, 616291
SLC9A3	161,7	100.0%	99.8%	Diarrhea 8, secretory sodium, congenital, 616868
SLC9A3R1	142,8	100.0%	100.0%	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
SLC9A6	101	98.6%	94.3%	Mental retardation, X-linked syndromic, Christianson type, 300243
SLC9A7	83,6	97.9%	91.3%	Intellectual developmental disorder, X-linked 108, 301024
SLCO1B1	53,5	97.4%	89.9%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO1B3	50,9	97.5%	87.9%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO2A1	97,7	99.9%	98.2%	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
SLFN14	172	100.0%	100.0%	Bleeding disorder, platelet-type, 20, 616913
SLITRK1	131,8	100.0%	100.0%	?Trichotillomania, 613229 Tourette syndrome, 137580
SLITRK6	169,5	100.0%	100.0%	Deafness and myopia, 221200
SLURP1	100,1	100.0%	99.4%	Meleda disease, 248300
SLX4	124,2	100.0%	99.7%	Fanconi anemia, complementation group P, 613951
SMAD3	126,7	100.0%	99.8%	Loeys-Dietz syndrome 3, 613795
SMAD4	108,9	100.0%	99.9%	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Polyposis, juvenile intestinal, 174900
SMAD6	180,5	98.8%	89.1%	Aortic valve disease 2, 614823 {Craniosynostosis 7, susceptibility to}, 617439
SMAD9	110	100.0%	100.0%	Pulmonary hypertension, primary, 2, 615342
SMARCA2	105,9	96.8%	95.9%	Nicolaides-Baraitser syndrome, 601358
SMARCA4	150,9	100.0%	99.4%	Coffin-Siris syndrome 4, 614609 {Rhabdoid tumor predisposition syndrome 2}, 613325

SMARCAD1	93,5	99.5%	96.7%	Adermatoglyphia, 136000 Basan syndrome, 129200 Huriez syndrome, 181600
SMARCAL1	113,2	100.0%	99.6%	Schimke immunoosseous dysplasia, 242900
SMARCB1	179,1	100.0%	99.9%	Coffin-Siris syndrome 3, 614608 Rhabdoid tumors, somatic, 609322 {Rhabdoid tumor predisposition syndrome 1}, 609322 {Schwannomatosis-1, susceptibility to}, 162091
SMARCC2	98	99.8%	97.9%	Coffin-Siris syndrome 8, 618362
SMARCD2	92,3	87.3%	85.8%	Specific granule deficiency 2, 617475
SMARCE1	66,5	94.4%	84.2%	Coffin-Siris syndrome 5, 616938 {Meningioma, familial, susceptibility to}, 607174
SMC1A	87,8	99.9%	97.8%	Cornelia de Lange syndrome 2, 300590
SMC3	84	96.0%	89.7%	Cornelia de Lange syndrome 3, 610759
SMCHD1	100,1	99.7%	97.3%	Bosma arhinia microphthalmia syndrome, 603457 Fascioscapulohumeral muscular dystrophy 2, digenic, 158901
SMG9	94,5	100.0%	100.0%	Heart and brain malformation syndrome, 616920
SMN1	97,6	99.8%	96.9%	Spinal muscular atrophy-1, 253300 Spinal muscular atrophy-2, 253550 Spinal muscular atrophy-3, 253400 Spinal muscular atrophy-4, 271150
SMO	140,4	99.9%	98.3%	Basal cell carcinoma, somatic, 605462 Curry-Jones syndrome, somatic mosaic, 601707
SMOC1	115,1	99.8%	98.2%	Microphthalmia with limb anomalies, 206920
SMOC2	88,7	77.0%	75.7%	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SMPD1	146,4	100.0%	99.2%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SMPX	63,9	99.8%	97.0%	Deafness, X-linked 4, 300066
SMS	63	88.9%	73.1%	Mental retardation, X-linked, Snyder-Robinson type, 309583
SNAI2	102,7	99.9%	99.1%	Piebaldism, 172800 Waardenburg syndrome, type 2D, 608890
SNAP25	119	99.9%	99.7%	?Myasthenic syndrome, congenital, 18, 616330
SNAP29	168,4	100.0%	100.0%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNCA	105	100.0%	100.0%	Dementia, Lewy body, 127750 Parkinson disease 1, 168601 Parkinson disease 4, 605543
SNCB	97,5	100.0%	99.9%	Dementia, Lewy body, 127750

SNIP1	131	100.0%	99.2%	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501
SNORD118	NC	NC	NC	Leukoencephalopathy, brain calcifications, and cysts, 614561
SNRNP200	118,8	99.7%	98.3%	Retinitis pigmentosa 33, 610359
SNRPB	77,7	99.9%	97.6%	Cerebrocostomandibular syndrome, 117650
SNRPE	73	98.1%	89.9%	Hypotrichosis 11, 615059
SNRPN	91,5	100.0%	98.3%	Prader-Willi syndrome, 176270
SNTA1	94,2	97.5%	89.0%	Long QT syndrome 12, 612955
SNX10	131,4	96.2%	95.7%	Osteopetrosis, autosomal recessive 8, 615085
SNX14	84,1	99.0%	95.4%	Spinocerebellar ataxia, autosomal recessive 20, 616354
SOBP	182,7	98.8%	97.8%	Mental retardation, anterior maxillary protrusion, and strabismus, 613671
SOD1	123,6	100.0%	100.0%	Amyotrophic lateral sclerosis 1, 105400
SOHLH1	101,7	99.9%	97.5%	Ovarian dysgenesis 5, 617690 Spermatogenic failure 32, 618115
SON	126,8	98.9%	95.2%	ZTTK syndrome, 617140
SOS1	102	99.6%	97.4%	?Fibromatosis, gingival, 1, 135300 Noonan syndrome 4, 610733
SOS2	99,7	99.7%	97.9%	Noonan syndrome 9, 616559
SOST	182,9	100.0%	99.6%	Craniodiaphyseal dysplasia, autosomal dominant, 122860 Sclerosteosis 1, 269500 Van Buchem disease, 239100
SOX10	88,2	100.0%	99.1%	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266
SOX11	196,2	100.0%	100.0%	Mental retardation, autosomal dominant 27, 615866
SOX17	124,8	100.0%	100.0%	Vesicoureteral reflux 3, 613674
SOX18	50	91.5%	76.2%	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823 Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940
SOX2	230	100.0%	100.0%	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SOX3	74	97.7%	92.9%	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000
SOX5	89,4	99.7%	97.6%	Lamb-Shaffer syndrome, 616803
SOX9	159,9	100.0%	100.0%	Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290 Campomelic dysplasia with autosomal sex reversal, 114290
SP110	109,3	100.0%	99.8%	Hepatic venoocclusive disease with immunodeficiency, 235550

				{Mycobacterium tuberculosis, susceptibility to}, 607948
SP7	148,4	100.0%	99.3%	Osteogenesis imperfecta, type XII, 613849
SPAG1	101,6	99.1%	95.3%	Ciliary dyskinesia, primary, 28, 615505
SPARC	134,3	100.0%	100.0%	Osteogenesis imperfecta, type XVII, 616507
SPART	132,6	99.8%	98.2%	Troyer syndrome, 275900
SPAST	95,4	99.8%	97.7%	Spastic paraplegia 4, autosomal dominant, 182601
SPATA16	136,8	100.0%	99.2%	?Spermatogenic failure 6, 102530
SPATA5	139,5	100.0%	99.8%	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPATA7	122,7	99.4%	97.4%	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, autosomal recessive, 604232
SPECC1L	127,5	100.0%	99.8%	?Facial clefting, oblique, 1, 600251 Hypertelorism, Teebi type, 145420 Opitz GBBB syndrome, type II, 145410
SPEG	128,6	99.2%	97.1%	Centronuclear myopathy 5, 615959
SPG11	116,1	99.7%	98.4%	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360
SPG21	120,6	99.7%	96.8%	Mast syndrome, 248900
SPG7	115,2	99.3%	96.4%	Spastic paraplegia 7, autosomal recessive, 607259
SPINK1	85	100.0%	99.4%	Pancreatitis, hereditary, 167800 Tropical calcific pancreatitis, 608189 {Fibrocalculous pancreatic diabetes, susceptibility to}, 608189
SPINK2	106,2	99.3%	98.9%	?Spermatogenic failure 29, 618091
SPINK5	128	99.9%	99.5%	Netherton syndrome, 256500
SPINT2	68,8	99.7%	90.0%	Diarrhea 3, secretory sodium, congenital, syndromic, 270420
SPR	145,7	100.0%	99.8%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRED1	146,5	99.8%	98.8%	Legius syndrome, 611431
SPRTN	160,6	100.0%	100.0%	Ruijs-Aalfs syndrome, 616200
SPRY4	164,5	100.0%	99.6%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
SPTA1	105,2	99.9%	98.6%	Elliptocytosis-2, 130600 Pyropoikilocytosis, 266140 Spherocytosis, type 3, 270970
SPTAN1	112	99.1%	98.3%	Epileptic encephalopathy, early infantile, 5, 613477
SPTB	142,9	100.0%	100.0%	Anemia, neonatal hemolytic, fatal or near-fatal, 617948 Elliptocytosis-3, 617948 Spherocytosis, type 2, 616649

SPTBN2	126,2	100.0%	99.7%	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia, autosomal recessive 14, 615386
SPTBN4	103,8	99.8%	98.1%	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519
SPTLC1	108,6	98.5%	93.4%	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	142,8	100.0%	99.9%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SQSTM1	117,8	99.9%	99.2%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 Myopathy, distal, with rimmed vacuoles, 617158 Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 Paget disease of bone 3, 167250
SRC	124,1	100.0%	99.5%	?Thrombocytopenia 6, 616937 Colon cancer, advanced, somatic, 114500
SRCAP	153	100.0%	99.6%	Floating-Harbor syndrome, 136140
SRD5A2	85,6	100.0%	98.1%	Pseudovaginal perineoscrotal hypospadias, 264600
SRD5A3	139,9	99.8%	98.3%	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713
SRP72	69,2	95.7%	85.6%	Bone marrow failure syndrome 1, 614675
SRPX2	61,6	99.4%	93.6%	?Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643
SRY	31,6	50.0%	50.0%	46XX sex reversal 1, 400045 46XY sex reversal 1, 400044
SSR4	109,5	100.0%	99.7%	Congenital disorder of glycosylation, type Iy, 300934
SSTR5	173,7	100.0%	100.0%	Somatostatin analog, resistance to, 0
SSX1	84,7	82.0%	79.7%	?Sarcoma, synovial, 300813
SSX2	57,2	62.9%	58.4%	?Sarcoma, synovial, 300813
ST14	154,8	100.0%	99.9%	Ichthyosis, congenital, autosomal recessive 11, 602400
ST3GAL3	134,7	100.0%	99.5%	?Epileptic encephalopathy, early infantile, 15, 615006 Mental retardation, autosomal recessive 12, 611090
ST3GAL5	101,8	89.0%	84.9%	Salt and pepper developmental regression syndrome, 609056
STAC3	114,7	100.0%	100.0%	Myopathy, congenital, Baily-Bloch, 255995
STAG1	111,1	99.6%	97.2%	Mental retardation, autosomal dominant 47, 617635
STAG2	74	96.8%	89.0%	Neurodevelopmental disorder, X-linked, with craniofacial abnormalities, 301022
STAG3	99,7	93.5%	92.5%	Premature ovarian failure 8, 615723
STAMBP	93,7	99.8%	97.9%	Microcephaly-capillary malformation syndrome, 614261
STAR	135	100.0%	100.0%	Lipoid adrenal hyperplasia, 201710
STAT1	117,8	99.6%	97.7%	Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796 Immunodeficiency 31C, autosomal dominant, 614162

STAT2	110	100.0%	99.8%	Immunodeficiency 44, 616636
STAT3	103,2	100.0%	99.0%	Autoimmune disease, multisystem, infantile-onset, 1, 615952 Hyper-IgE recurrent infection syndrome, 147060
STAT5B	114,1	99.8%	97.8%	Growth hormone insensitivity with immunodeficiency, 245590 Leukemia, acute promyelocytic, somatic, 102578
STEAP3	166,9	100.0%	99.4%	?Anemia, hypochromic microcytic, with iron overload 2, 615234
STIL	154,1	100.0%	99.8%	Microcephaly 7, primary, autosomal recessive, 612703
STIM1	120,7	99.8%	96.8%	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070
STK11	131	100.0%	100.0%	Melanoma, malignant, somatic, 0 Pancreatic cancer, somatic, 260350 Peutz-Jeghers syndrome, 175200 Testicular tumor, somatic, 273300
STK4	122,5	100.0%	99.7%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STN1	82,2	100.0%	99.6%	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341
STOX1	146,5	89.6%	89.5%	Preeclampsia/eclampsia 4, 609404
STRA6	117,6	100.0%	99.8%	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186
STRADA	108,6	100.0%	98.8%	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087
STRC	99	99.9%	98.1%	Deafness, autosomal recessive 16, 603720
STS	78,7	99.3%	95.2%	Ichthyosis, X-linked, 308100
STT3A	123,1	100.0%	99.9%	?Congenital disorder of glycosylation, type Iw, 615596
STT3B	127,4	99.9%	99.6%	?Congenital disorder of glycosylation, type Ix, 615597
STUB1	173,9	100.0%	99.5%	?Spinocerebellar ataxia 48, 618093 Spinocerebellar ataxia, autosomal recessive 16, 615768
STX11	298,3	100.0%	100.0%	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STX16	111	99.9%	98.5%	Pseudohypoparathyroidism, type IB, 603233
STX1B	157,7	100.0%	100.0%	Generalized epilepsy with febrile seizures plus, type 9, 616172
STXBP1	103,7	96.8%	96.4%	Epileptic encephalopathy, early infantile, 4, 612164
STXBP2	100,2	83.7%	80.4%	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
SUCLA2	58,8	91.7%	82.6%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	102,9	99.9%	99.6%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUFU	132,8	100.0%	99.9%	Basal cell nevus syndrome, 109400 Joubert syndrome 32, 617757

				Medulloblastoma, desmoplastic, 155255 {Meningioma, familial, susceptibility to}, 607174
SUGCT	127,1	99.4%	95.8%	Glutaric aciduria III, 231690
SULT2B1	124,7	100.0%	100.0%	Ichthyosis, congenital, autosomal recessive 14, 617571
SUMF1	89,7	99.7%	96.8%	Multiple sulfatase deficiency, 272200
SUMO1	20	62.3%	46.0%	?Orofacial cleft 10, 613705
SUN5	98,3	100.0%	99.8%	Spermatogenic failure 16, 617187
SUOX	167,2	100.0%	100.0%	Sulfite oxidase deficiency, 272300
SURF1	84,8	91.3%	88.4%	Charcot-Marie-Tooth disease, type 4K, 616684 Leigh syndrome, due to COX IV deficiency, 256000
SYCE1	114	99.9%	98.2%	?Premature ovarian failure 12, 616947 ?Spermatogenic failure 15, 616950
SYCP3	84,4	99.7%	97.9%	Pregnancy loss, recurrent, 4, 270960 Spermatogenic failure 4, 270960
SYN1	66,6	90.6%	79.1%	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNE1	121,6	98.3%	97.8%	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 Spinocerebellar ataxia, autosomal recessive 8, 610743
SYNE2	110,7	99.6%	98.0%	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999
SYNE4	83,7	99.9%	98.5%	Deafness, autosomal recessive 76, 615540
SYNGAP1	140,7	98.4%	97.7%	Mental retardation, autosomal dominant 5, 612621
SYNJ1	126,6	99.9%	98.5%	Epileptic encephalopathy, early infantile, 53, 617389 Parkinson disease 20, early-onset, 615530
SYP	79,9	99.9%	98.1%	Mental retardation, X-linked 96, 300802
SYT1	149,6	99.9%	99.1%	Baker-Gordon syndrome, 618218
SYT14	104,1	60.3%	58.0%	?Spinocerebellar ataxia, autosomal recessive 11, 614229
SYT2	96	99.8%	98.4%	Myasthenic syndrome, congenital, 7, presynaptic, 616040
SZT2	135,6	99.6%	99.4%	Epileptic encephalopathy, early infantile, 18, 615476
T	NC	NC	NC	Sacral agenesis with vertebral anomalies, 615709 {Neural tube defects, susceptibility to}, 182940
TAB2	170,6	99.9%	99.5%	Congenital heart defects, nonsyndromic, 2, 614980
TAC3	61,8	99.9%	95.4%	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACO1	93,9	99.5%	95.3%	Mitochondrial complex IV deficiency, 220110
TACR3	146,1	100.0%	99.7%	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
TACSTD2	268,7	100.0%	99.9%	Corneal dystrophy, gelatinous drop-like, 204870
TAF1	86,8	99.1%	95.5%	Dystonia-Parkinsonism, X-linked, 314250 Mental retardation, X-linked, syndromic 33, 300966

TAF13	100,1	100.0%	99.9%	Mental retardation, autosomal recessive 60, 617432
TAF2	113,9	99.8%	98.5%	Mental retardation, autosomal recessive 40, 615599
TAF4B	122,5	99.0%	94.4%	?Spermatogenic failure 13, 615841
TAF6	127,3	100.0%	99.3%	Alazami-Yuan syndrome, 617126
TAL1	58,5	95.1%	85.9%	Leukemia, T-cell acute lymphocytic, somatic, 613065
TAL2	108	100.0%	100.0%	Leukemia, T-cell acute lymphocytic, somatic, 613065
TALDO1	148,2	100.0%	99.6%	Transaldolase deficiency, 606003
TANGO2	127,3	100.0%	100.0%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TAP1	117,7	99.9%	97.3%	Bare lymphocyte syndrome, type I, 604571
TAP2	93	99.6%	98.4%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	116,6	96.6%	96.5%	Bare lymphocyte syndrome, type I, 604571
TAPT1	89,2	97.9%	92.2%	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897
TARDBP	123,8	100.0%	99.9%	Amyotrophic lateral sclerosis 10, with or without FTD, 612069 Frontotemporal lobar degeneration, TARDBP-related, 612069
TARS2	89	99.7%	96.4%	?Combined oxidative phosphorylation deficiency 21, 615918
TAT	115	100.0%	100.0%	Tyrosinemia, type II, 276600
TAZ	114,5	99.3%	95.8%	Barth syndrome, 302060
TBC1D20	115,7	96.3%	93.8%	Warburg micro syndrome 4, 615663
TBC1D23	92,7	99.2%	95.4%	Pontocerebellar hypoplasia, type 11, 617695
TBC1D24	177,7	100.0%	100.0%	Deafness , autosomal recessive 86, 614617 Deafness, autosomal dominant 65, 616044 DOORS syndrome, 220500 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021
TBC1D7	99,6	99.8%	99.3%	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000
TBCD	136,2	98.2%	94.3%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TBCE	116,4	98.7%	94.7%	Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Kenny-Caffey syndrome, type 1, 244460
TBCK	101,6	99.5%	96.1%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900
TBK1	101,6	99.5%	97.6%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439 {Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 8}, 617900
TBL1XR1	62,9	94.5%	80.8%	Mental retardation, autosomal dominant 41, 616944 Pierpont syndrome, 602342
TBP	99,5	99.9%	99.7%	Spinocerebellar ataxia 17, 607136

				{Parkinson disease, susceptibility to}, 168600
TBR1	166,2	100.0%	100.0%	Intellectual developmental disorder with autism and speech delay, 606053
TBX1	101,2	93.0%	86.9%	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Velocardiofacial syndrome, 192430
TBX15	106,3	100.0%	99.7%	Cousin syndrome, 260660
TBX18	103,3	99.6%	97.6%	Congenital anomalies of kidney and urinary tract 2, 143400
TBX19	156,9	100.0%	99.9%	Adrenocorticotropic hormone deficiency, 201400
TBX2	158,2	100.0%	100.0%	Vertebral anomalies and variable endocrine and T-cell dysfunction, 618223
TBX20	108,2	100.0%	99.9%	Atrial septal defect 4, 611363
TBX21	110,5	99.9%	97.9%	Asthma and nasal polyps, 208550 {Asthma, aspirin-induced, susceptibility to}, 208550
TBX22	105	99.4%	94.8%	?Abruzzo-Erickson syndrome, 302905 Cleft palate with ankyloglossia, 303400
TBX3	100,7	99.8%	98.2%	Ulnar-mammary syndrome, 181450
TBX4	175,9	99.5%	97.5%	Ischiocoxopodopatellar syndrome, 147891
TBX5	135,3	100.0%	100.0%	Holt-Oram syndrome, 142900
TBX6	124,4	99.7%	96.5%	Spondylocostal dysostosis 5, 122600
TBXAS1	128,8	100.0%	100.0%	?Thromboxane synthase deficiency, 614158 Ghosal hematodiaphyseal syndrome, 231095
TCAP	100,1	100.0%	100.0%	Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954
TCF12	137,7	99.9%	99.9%	Craniosynostosis 3, 615314
TCF3	91,1	99.1%	95.6%	Agammaglobulinemia 8, autosomal dominant, 616941
TCF4	109,2	100.0%	99.8%	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954
TCHH	162,8	100.0%	99.9%	?Uncombable hair syndrome 3, 617252
TCIRG1	131,4	99.2%	96.6%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	148,5	100.0%	100.0%	Transcobalamin II deficiency, 275350
TCOF1	111,6	99.9%	99.1%	Treacher Collins syndrome 1, 154500
TCTEX1D2	123,6	100.0%	99.4%	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405
TCTN1	94,8	95.6%	92.3%	Joubert syndrome 13, 614173
TCTN2	122,4	99.9%	99.0%	?Meckel syndrome 8, 613885 Joubert syndrome 24, 616654
TCTN3	116,3	100.0%	99.9%	Joubert syndrome 18, 614815

				Orofaciodigital syndrome IV, 258860
TDGF1	120,9	99.7%	94.8%	Forebrain defects, 0
TDP1	103,9	99.9%	99.5%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250
TDP2	173	99.9%	99.4%	Spinocerebellar ataxia, autosomal recessive 23, 616949
TDRD7	136,7	99.8%	98.7%	Cataract 36, 613887
TDRD9	112,8	99.9%	98.4%	?Spermatogenic failure 30, 618110
TEAD1	132,3	100.0%	99.6%	Sveinsson chorioretinal atrophy, 108985
TECPR2	137,2	100.0%	100.0%	Spastic paraplegia 49, autosomal recessive, 615031
TECR	124,9	100.0%	99.6%	Mental retardation, autosomal recessive 14, 614020
TECRL	75	97.1%	90.3%	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021
TECTA	164,9	100.0%	99.9%	Deafness, autosomal dominant 8/12, 601543 Deafness, autosomal recessive 21, 603629
TEK	148	100.0%	99.7%	Glaucoma 3, primary congenital, E, 617272 Venous malformations, multiple cutaneous and mucosal, 600195
TELO2	122,6	99.8%	97.5%	You-Hoover-Fong syndrome, 616954
TENM3	148,6	99.8%	99.3%	Microphthalmia, isolated, with coloboma 9, 615145
TENM4	119,9	100.0%	99.6%	Essential tremor, hereditary, 5, 616736
TERC	NC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550 {Aplastic anemia}, 614743 {Pulmonary fibrosis, idiopathic, susceptibility to}, 614743
TET2	169,7	100.0%	99.9%	Myelodysplastic syndrome, somatic, 614286
TEX11	72,7	92.6%	85.2%	Spermatogenic failure, X-linked, 2, 309120
TEX14	97	99.8%	97.8%	?Spermatogenic failure 23, 617707
TEX15	116,5	99.9%	99.3%	Spermatogenic failure 25, 617960
TF	101,6	100.0%	99.7%	Atransferrinemia, 209300
TFAM	67,1	94.2%	74.5%	?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156
TFAP2A	112,7	99.8%	98.0%	Branchiooculofacial syndrome, 113620
TFAP2B	168,3	99.2%	96.8%	Char syndrome, 169100 Patent ductus arteriosus 2, 617035
TFE3	88,1	99.7%	96.3%	Renal cell carcinoma, papillary, 1, 300854
TFG	106,2	96.8%	95.3%	?Spastic paraplegia 57, autosomal recessive, 615658 Hereditary motor and sensory neuropathy, Okinawa type, 604484
TFR2	124	99.6%	98.3%	Hemochromatosis, type 3, 604250
TFRC	132	99.9%	99.0%	Immunodeficiency 46, 616740
TG	117	99.9%	99.1%	Thyroid dyshormonogenesis 3, 274700 {Autoimmune thyroid disease, susceptibility to, 3}, 608175

TGDS	88,3	99.4%	96.6%	Catel-Manzke syndrome, 616145
TGFB1	102,1	100.0%	99.6%	Camurati-Engelmann disease, 131300 Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213 {Cystic fibrosis lung disease, modifier of}, 219700
TGFB2	173,2	99.9%	99.0%	Loeys-Dietz syndrome 4, 614816
TGFB3	140,4	100.0%	100.0%	Arrhythmogenic right ventricular dysplasia 1, 107970 Loeys-Dietz syndrome 5, 615582
TGFBI	114,1	99.9%	98.6%	Corneal dystrophy, Avellino type, 607541 Corneal dystrophy, epithelial basement membrane, 121820 Corneal dystrophy, Groenouw type I, 121900 Corneal dystrophy, lattice type I, 122200 Corneal dystrophy, lattice type IIIA, 608471 Corneal dystrophy, Reis-Bucklers type, 608470 Corneal dystrophy, Thiel-Behnke type, 602082
TGFBR1	156,4	95.4%	93.8%	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800
TGFBR2	156,8	100.0%	100.0%	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome 2, 610168
TGIF1	141,2	100.0%	100.0%	Holoprosencephaly 4, 142946
TGM1	141	100.0%	99.9%	Ichthyosis, congenital, autosomal recessive 1, 242300
TGM3	137,1	99.9%	98.4%	?Uncombable hair syndrome 2, 617251
TGM5	144,8	100.0%	99.9%	Peeling skin syndrome 2, 609796
TGM6	130,2	99.9%	98.7%	Spinocerebellar ataxia 35, 613908
TH	96,3	100.0%	98.2%	Segawa syndrome, recessive, 605407
THAP1	141,3	100.0%	100.0%	Dystonia 6, torsion, 602629
THBD	181,1	100.0%	100.0%	Thrombophilia due to thrombomodulin defect, 614486 {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
THOC2	83,9	98.9%	93.6%	Mental retardation, X-linked 12/35, 300957
THOC6	228,9	100.0%	100.0%	Beaulieu-Boycott-Innes syndrome, 613680
THPO	97,3	100.0%	99.7%	Thrombocythemia 1, 187950
THRA	170,6	100.0%	100.0%	Hypothyroidism, congenital, nongoitrous, 6, 614450
THRB	141,2	99.9%	99.3%	Thyroid hormone resistance, 188570 Thyroid hormone resistance, autosomal recessive, 274300 Thyroid hormone resistance, selective pituitary, 145650
TIA1	131,8	99.7%	97.2%	Welander distal myopathy, 604454
TIMM50	122,9	99.9%	98.7%	3-methylglutaconic aciduria, type IX, 617698

TIMM8A	46,3	94.6%	79.9%	Mohr-Tranebjaerg syndrome, 304700
TIMMDC1	161,4	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 31, 618251
TIMP3	137,5	100.0%	100.0%	Sorsby fundus dystrophy, 136900
TINF2	177,1	100.0%	100.0%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TJP2	109,3	94.0%	93.4%	Cholestasis, progressive familial intrahepatic 4, 615878 Hypercholanemia, familial, 607748
TK2	103,8	100.0%	99.2%	?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069 Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560
TKT	115,6	98.7%	98.1%	Short stature, developmental delay, and congenital heart defects, 617044
TLE6	116,7	99.9%	98.6%	Preimplantation embryonic lethality, 616814
TLK2	89,3	97.9%	92.3%	Mental retardation, autosomal dominant 57, 618050
TLL1	129,8	100.0%	99.9%	Atrial septal defect 6, 613087
TMC1	110,1	99.8%	97.5%	Deafness, autosomal dominant 36, 606705 Deafness, autosomal recessive 7, 600974
TMC6	91,1	100.0%	99.7%	Epidermodysplasia verruciformis, 226400
TMC8	133	100.0%	99.7%	Epidermodysplasia verruciformis 2, 618231
TMCO1	81,7	88.0%	87.5%	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980
TMEM106B	121,4	99.7%	98.9%	Leukodystrophy, hypomyelinating, 16, 617964
TMEM107	148,7	100.0%	100.0%	?Joubert syndrome 29, 617562 Meckel syndrome 13, 617562 Orofaciodigital syndrome XVI, 617563
TMEM126A	104,8	96.2%	82.8%	Optic atrophy 7, 612989
TMEM126B	87,8	99.5%	95.9%	Mitochondrial complex I deficiency, nuclear type 29, 618250
TMEM138	82,7	100.0%	99.2%	Joubert syndrome 16, 614465
TMEM165	148,2	100.0%	99.8%	Congenital disorder of glycosylation, type IIk, 614727
TMEM173	95,3	99.1%	94.0%	STING-associated vasculopathy, infantile-onset, 615934
TMEM199	118,4	100.0%	99.8%	Congenital disorder of glycosylation, type IIp, 616829
TMEM216	88	99.7%	95.7%	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	101,1	100.0%	99.3%	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	117,7	100.0%	99.2%	Joubert syndrome 14, 614424
TMEM240	163,9	100.0%	100.0%	Spinocerebellar ataxia 21, 607454
TMEM260	117,5	99.6%	97.6%	Structural heart defects and renal anomalies syndrome, 617478
TMEM38B	107,9	100.0%	99.0%	Osteogenesis imperfecta, type XIV, 615066

TMEM43	131	99.9%	98.0%	Arrhythmogenic right ventricular dysplasia 5, 604400 Emery-Dreifuss muscular dystrophy 7, AD, 614302
TMEM5	NC	NC	NC	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
TMEM67	83,1	99.1%	94.6%	?RHYNS syndrome, 602152 COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991
TMEM70	117,9	99.8%	97.6%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMEM94	164,7	100.0%	100.0%	Intellectual developmental disorder with cardiac defects and dysmorphic facies, 618316
TMEM98	124,4	99.2%	96.5%	Nanophthalmos 4, 615972
TMIE	104,5	100.0%	99.7%	Deafness, autosomal recessive 6, 600971
TMPRSS15	106,6	98.5%	95.1%	Enterokinase deficiency, 226200
TMPRSS3	98,5	100.0%	99.4%	Deafness, autosomal recessive 8/10, 601072
TMPRSS6	107	100.0%	99.4%	Iron-refractory iron deficiency anemia, 206200
TMTC3	92,8	99.5%	97.6%	Lissencephaly 8, 617255
TNC	143,1	100.0%	99.7%	Deafness, autosomal dominant 56, 615629
TNFAIP3	149,4	100.0%	100.0%	Autoinflammatory syndrome, familial, Behcet-like, 616744
TNFRSF10B	109,1	100.0%	99.9%	Squamous cell carcinoma, head and neck, 275355
TNFRSF11A	131	96.1%	95.2%	Osteolysis, familial expansile, 174810 Osteopetrosis, autosomal recessive 7, 612301 {Paget disease of bone 2, early-onset}, 602080
TNFRSF11B	172,4	100.0%	100.0%	Paget disease of bone 5, juvenile-onset, 239000
TNFRSF13B	100,8	100.0%	99.7%	Immunodeficiency, common variable, 2, 240500 Immunoglobulin A deficiency 2, 609529
TNFRSF13C	98,4	96.2%	82.4%	Immunodeficiency, common variable, 4, 613494
TNFRSF1A	106,8	92.5%	89.7%	Periodic fever, familial, 142680 {Multiple sclerosis, susceptibility to, 5}, 614810
TNFRSF4	79,8	99.5%	96.8%	?Immunodeficiency 16, 615593
TNFSF11	129,8	100.0%	100.0%	Osteopetrosis, autosomal recessive 2, 259710
TNIK	106,3	99.9%	98.7%	Mental retardation, autosomal recessive 54, 617028
TNNC1	150,1	100.0%	100.0%	Cardiomyopathy, dilated, 1Z, 611879 Cardiomyopathy, hypertrophic, 13, 613243
TNNI2	150,5	100.0%	100.0%	Arthrogryposis multiplex congenita, distal, type 2B, 601680
TNNI3	107,3	99.6%	94.9%	?Cardiomyopathy, dilated, 2A, 611880

				Cardiomyopathy, dilated, 1FF, 613286 Cardiomyopathy, familial restrictive, 1, 115210 Cardiomyopathy, hypertrophic, 7, 613690
TNNI3K	105,8	99.9%	99.3%	Cardiac conduction disease with or without dilated cardiomyopathy, 616117
TNNT1	104,4	99.9%	98.5%	Nemaline myopathy 5, Amish type, 605355
TNNT2	106,4	100.0%	100.0%	Cardiomyopathy, dilated, 1D, 601494 Cardiomyopathy, familial restrictive, 3, 612422 Cardiomyopathy, hypertrophic, 2, 115195 Left ventricular noncompaction 6, 601494
TNNT3	146,3	100.0%	99.8%	Arthrogryposis, distal, type 2B, 601680
TNPO3	115,2	100.0%	99.7%	Muscular dystrophy, limb-girdle, autosomal dominant 2, 608423
TNRC6A	127,5	100.0%	99.5%	?Epilepsy, familial adult myoclonic, 6, 618074
TNXB	105,6	99.5%	95.8%	Ehlers-Danlos syndrome, classic-like, 1, 606408 Vesicoureteral reflux 8, 615963
TOE1	141,1	100.0%	99.8%	Pontocerebellar hypoplasia, type 7, 614969
TOP1	95,1	99.7%	97.6%	DNA topoisomerase I, camptothecin-resistant, 0
TOP2A	120,9	100.0%	99.2%	DNA topoisomerase II, resistance to inhibition of, by amsacrine, 0
TOP3A	121	99.8%	97.5%	?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098 Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097
TOPORS	182,2	100.0%	100.0%	Retinitis pigmentosa 31, 609923
TOR1A	142,4	100.0%	100.0%	Dystonia-1, torsion, 128100 {Dystonia-1, modifier of}, 0
TOR1AIP1	133,3	99.5%	97.2%	?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072
TP53	89,2	99.8%	98.5%	Bone marrow failure syndrome 5, 618165 Breast cancer, somatic, 114480 Hepatocellular carcinoma, somatic, 114550 Li-Fraumeni syndrome, 151623 Nasopharyngeal carcinoma, somatic, 607107 Pancreatic cancer, somatic, 260350 {Adrenocortical carcinoma, pediatric}, 202300 {Basal cell carcinoma 7}, 614740 {Choroid plexus papilloma}, 260500 {Colorectal cancer}, 114500 {Glioma susceptibility 1}, 137800 {Osteosarcoma}, 259500
TP53RK	81,3	99.7%	96.1%	Galloway-Mowat syndrome 4, 617730
TP63	162,8	100.0%	100.0%	ADULT syndrome, 103285

				Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 Hay-Wells syndrome, 106260 Limb-mammary syndrome, 603543 Orofacial cleft 8, 618149 Rapp-Hodgkin syndrome, 129400 Split-hand/foot malformation 4, 605289
TPI1	112,1	99.9%	96.4%	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512
TPK1	94	100.0%	98.7%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TPM1	113	100.0%	99.2%	Cardiomyopathy, dilated, 1Y, 611878 Cardiomyopathy, hypertrophic, 3, 115196 Left ventricular noncompaction 9, 611878
TPM2	105,2	100.0%	99.7%	Arthrogryposis multiplex congenita, distal, type 1, 108120 Arthrogryposis, distal, type 2B, 601680 CAP myopathy 2, 609285 Nemaline myopathy 4, autosomal dominant, 609285
TPM3	74,3	89.5%	88.0%	CAP myopathy 1, 609284 Myopathy, congenital, with fiber-type disproportion, 255310 Nemaline myopathy 1, autosomal dominant or recessive, 609284
TPO	137,6	100.0%	99.6%	Thyroid dyshormonogenesis 2A, 274500
TPP1	123,7	100.0%	99.9%	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TPRKB	59,4	80.3%	73.9%	Galloway-Mowat syndrome 5, 617731
TPRN	98,7	91.5%	86.7%	Deafness, autosomal recessive 79, 613307
TRAC	127,5	100.0%	100.0%	Immunodeficiency 7, TCR-alpha/beta deficient, 615387
TRAF3IP1	84,2	99.4%	97.1%	Senior-Loken syndrome 9, 616629
TRAF3IP2	111,1	100.0%	98.3%	?Candidiasis, familial, 8, 615527 {Psoriasis susceptibility 13}, 614070
TRAF7	161,7	99.8%	98.1%	Cardiac, facial, and digital anomalies with developmental delay, 618164
TRAIP	123,4	100.0%	100.0%	Seckel syndrome 9, 616777
TRAK1	149,3	100.0%	99.6%	Epileptic encephalopathy, early infantile, 68, 618201
TRAPPC11	125,6	99.9%	99.0%	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356
TRAPPC12	162,3	100.0%	100.0%	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669
TRAPPC2	59,2	86.5%	67.4%	Spondyloepiphyseal dysplasia tarda, 313400
TRAPPC2L	198,9	100.0%	100.0%	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331
TRAPPC6B	79	99.9%	98.2%	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862
TRAPPC9	125,2	100.0%	99.7%	Mental retardation, autosomal recessive 13, 613192

TRDN	82,7	97.5%	88.4%	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441
TREH	141,3	98.2%	93.2%	Trehalase deficiency, 612119
TREM2	127	100.0%	99.9%	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193
TREX1	233,4	100.0%	100.0%	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700
TRHR	174,8	99.9%	99.1%	Thyrotropin-releasing hormone resistance, generalized, 0
TRIM2	136,6	93.9%	93.6%	Charcot-Marie-Tooth disease, type 2R, 615490
TRIM32	123	100.0%	100.0%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRIM36	136	99.9%	99.0%	?Anencephaly, 206500
TRIM37	113,8	98.5%	97.4%	Mulibrey nanism, 253250
TRIM44	113,5	100.0%	100.0%	?Aniridia 3, 617142
TRIO	121,9	99.0%	96.9%	Mental retardation, autosomal dominant 44, 617061
TRIOBP	156,9	99.1%	97.5%	Deafness, autosomal recessive 28, 609823
TRIP11	90,9	97.5%	92.6%	Achondrogenesis, type IA, 200600 Osteochondrodysplasia, 184260
TRIP12	132,2	99.9%	99.4%	Mental retardation, autosomal dominant 49, 617752
TRIP13	127,4	100.0%	99.9%	Mosaic variegated aneuploidy syndrome 3, 617598
TRIP4	103,3	99.8%	98.5%	?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066 Spinal muscular atrophy with congenital bone fractures 1, 616866
TRIT1	107,9	100.0%	99.9%	Combined oxidative phosphorylation deficiency 35, 617873
TRMT1	117,1	99.7%	97.6%	Mental retardation, autosomal recessive 68, 618302
TRMT10A	119,5	99.9%	99.2%	Microcephaly, short stature, and impaired glucose metabolism 1, 616033
TRMT10C	138,5	100.0%	100.0%	Combined oxidative phosphorylation deficiency 30, 616974
TRMT5	175,7	99.8%	98.7%	Combined oxidative phosphorylation deficiency 26, 616539
TRMU	100	100.0%	99.4%	Liver failure, transient infantile, 613070 {Deafness, mitochondrial, modifier of}, 580000
TRNT1	101,5	99.2%	96.5%	Retinitis pigmentosa and erythrocytic microcytosis, 616959 Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084
TRPA1	83,8	95.5%	88.7%	?Episodic pain syndrome, familial, 1, 615040
TRPC3	146,8	99.8%	98.5%	?Spinocerebellar ataxia 41, 616410
TRPC6	91,4	98.0%	96.0%	Glomerulosclerosis, focal segmental, 2, 603965
TRPM1	128,8	100.0%	99.0%	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TRPM4	137,4	100.0%	99.9%	Progressive familial heart block, type IB, 604559

TRPM6	126,6	99.9%	99.1%	Hypomagnesemia 1, intestinal, 602014
TRPS1	154	100.0%	99.9%	Trichorhinophalangeal syndrome, type I, 190350 Trichorhinophalangeal syndrome, type III, 190351
TRPV3	122,8	99.9%	98.5%	?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400 Olmsted syndrome, 614594
TRPV4	138,4	100.0%	99.8%	?Avascular necrosis of femoral head, primary, 2, 617383 Brachyolmia type 3, 113500 Digital arthropathy-brachydactyly, familial, 606835 Hereditary motor and sensory neuropathy, type IIc, 606071 Metatropic dysplasia, 156530 Parastremmatic dwarfism, 168400 Scapuloperoneal spinal muscular atrophy, 181405 SED, Maroteaux type, 184095 Spinal muscular atrophy, distal, congenital nonprogressive, 600175 Spondylometaphyseal dysplasia, Kozlowski type, 184252 [Sodium serum level QTL 1], 613508
TRPV6	144,3	100.0%	99.9%	Hyperparathyroidism, transient neonatal, 618188
TSC1	112,5	99.6%	98.2%	Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100
TSC2	140,5	100.0%	99.9%	?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254
TSEN15	89,8	99.7%	96.4%	Pontocerebellar hypoplasia, type 2F, 617026
TSEN2	95,6	99.9%	98.9%	Pontocerebellar hypoplasia type 2B, 612389
TSEN34	77,6	98.7%	93.6%	?Pontocerebellar hypoplasia type 2C, 612390
TSEN54	114,4	99.4%	96.8%	?Pontocerebellar hypoplasia type 5, 610204 Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753
TSFM	120	100.0%	99.2%	Combined oxidative phosphorylation deficiency 3, 610505
TSGA10	112,8	99.8%	99.2%	?Spermatogenic failure 26, 617961
TSHB	229,7	100.0%	100.0%	Hypothyroidism, congenital, nongoitrous 4, 275100
TSHR	153	100.0%	99.0%	Hyperthyroidism, familial gestational, 603373 Hyperthyroidism, nonautoimmune, 609152 Hypothyroidism, congenital, nongoitrous, 1, 275200 Thyroid adenoma, hyperfunctioning, somatic, 0 Thyroid carcinoma with thyrotoxicosis, 0

TSHZ1	147,6	98.9%	98.7%	Aural atresia, congenital, 607842
TSPAN12	135,3	100.0%	99.4%	Exudative vitreoretinopathy 5, 613310
TSPAN7	107,3	100.0%	99.0%	Mental retardation, X-linked 58, 300210
TSPEAR	139,3	100.0%	99.8%	?Deafness, autosomal recessive 98, 614861 Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180
TSPYL1	144,9	100.0%	100.0%	Sudden infant death with dysgenesis of the testes syndrome, 608800
TSR2	76,1	100.0%	99.4%	?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946
TTBK2	108,4	99.9%	96.8%	Spinocerebellar ataxia 11, 604432
TTC19	83,4	97.0%	82.6%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTC21B	119,5	99.7%	98.8%	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
TTC25	93,7	100.0%	99.6%	Ciliary dyskinesia, primary, 35, 617092
TTC37	135,1	99.9%	99.2%	Trichohepatoenteric syndrome 1, 222470
TTC7A	115	99.8%	98.0%	Gastrointestinal defects and immunodeficiency syndrome, 243150
TTC8	115,2	99.8%	98.8%	?Retinitis pigmentosa 51, 613464 Bardet-Biedl syndrome 8, 615985
TTI2	96,2	100.0%	99.9%	Mental retardation, autosomal recessive 39, 615541
TTLL5	136,1	100.0%	98.7%	Cone-rod dystrophy 19, 615860
TTN	163	98.6%	98.1%	Cardiomyopathy, dilated, 1G, 604145 Cardiomyopathy, familial hypertrophic, 9, 613765 Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807 Myopathy, myofibrillar, 9, with early respiratory failure, 603689 Salih myopathy, 611705 Tibial muscular dystrophy, tardive, 600334
TTPA	109,2	97.6%	92.5%	Ataxia with isolated vitamin E deficiency, 277460
TTR	122,6	94.6%	94.6%	Amyloidosis, hereditary, transthyretin-related, 105210 Carpal tunnel syndrome, familial, 115430 [Dystransthyretinemic hyperthyroxinemia], 145680
TUB	103,4	100.0%	99.4%	?Retinal dystrophy and obesity, 616188
TUBA1A	77,6	99.8%	97.1%	Lissencephaly 3, 611603
TUBA3D	103,6	99.3%	95.9%	Keratoconus 9, 617928
TUBA4A	159,8	100.0%	100.0%	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia, 616208
TUBA8	126,1	100.0%	99.5%	Cortical dysplasia, complex, with other brain malformations 8, 613180
TUBB	112,9	98.2%	94.2%	Cortical dysplasia, complex, with other brain malformations 6, 615771 Symmetric circumferential skin creases, congenital, 1, 156610
TUBB1	150	100.0%	100.0%	Macrothrombocytopenia, autosomal dominant, TUBB1-related, 613112

TUBB2A	77,1	99.7%	97.2%	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB2B	78,2	100.0%	99.7%	Cortical dysplasia, complex, with other brain malformations 7, 610031
TUBB3	121,3	99.8%	98.4%	Cortical dysplasia, complex, with other brain malformations 1, 614039 Fibrosis of extraocular muscles, congenital, 3A, 600638
TUBB4A	101,2	97.1%	95.6%	Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438
TUBB4B	86,1	100.0%	99.9%	Leber congenital amaurosis with early-onset deafness, 617879
TUBB6	88,6	91.9%	90.0%	?Facial palsy, congenitla, with ptosis and velopharyngeal dysfunction, 617732
TUBB8	21,9	88.0%	51.9%	Oocyte maturation defect 2, 616780
TUBG1	154,3	100.0%	100.0%	Cortical dysplasia, complex, with other brain malformations 4, 615412
TUBGCP4	104,6	98.0%	94.7%	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TUBGCP6	155,1	100.0%	99.5%	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
TUFM	130,6	100.0%	99.2%	Combined oxidative phosphorylation deficiency 4, 610678
TULP1	117,9	100.0%	99.6%	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132
TUSC3	155,1	99.9%	99.5%	Mental retardation, autosomal recessive 7, 611093
TWIST1	160,2	100.0%	99.6%	Craniosynostosis 1, 123100 Robinow-Sorauf syndrome, 180750 Saethre-Chotzen syndrome with or without eyelid anomalies, 101400 Sweeney-Cox syndrome, 617746
TWIST2	132,6	100.0%	100.0%	Ablepharon-macrostomia syndrome, 200110 Barber-Say syndrome, 209885 Focal facial dermal dysplasia 3, Setleis type, 227260
TWNK	159,6	100.0%	100.0%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Perrault syndrome 5, 616138 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286
TXN2	61,4	100.0%	99.8%	?Combined oxidative phosphorylation deficiency 29, 616811
TXNL4A	122,6	99.5%	98.3%	Burn-McKeown syndrome, 608572
TXNRD2	111,6	96.7%	95.2%	?Glucocorticoid deficiency 5, 617825
TYK2	129,9	100.0%	99.5%	Immunodeficiency 35, 611521
TYMP	120,9	100.0%	100.0%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYR	147,9	100.0%	99.9%	Albinism, oculocutaneous, type IA, 203100 Albinism, oculocutaneous, type IB, 606952 Waardenburg syndrome/albinism, digenic, 103470 [Skin/hair/eye pigmentation 3, blue/green eyes], 601800 [Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 {Melanoma, cutaneous malignant, susceptibility to, 8}, 601800

TYROBP	83,3	100.0%	100.0%	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770
TYRP1	152,3	100.0%	100.0%	Albinism, oculocutaneous, type III, 203290 [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271
UBA1	130,5	99.6%	98.1%	Spinal muscular atrophy, X-linked 2, infantile, 301830
UBA5	79,9	97.7%	86.6%	?Spinocerebellar ataxia, autosomal recessive 24, 617133 Epileptic encephalopathy, early infantile, 44, 617132
UBE2A	117,9	99.9%	96.4%	Mental retardation, X-linked syndromic, Nascimento-type, 300860
UBE2T	91,1	100.0%	99.3%	Fanconi anemia, complementation group T, 616435
UBE3A	81,5	98.6%	93.3%	Angelman syndrome, 105830
UBE3B	113,5	100.0%	99.7%	Kaufman oculocerebrofacial syndrome, 244450
UBIAD1	187,7	99.8%	97.4%	Corneal dystrophy, Schnyder type, 121800
UBQLN2	124,1	100.0%	99.6%	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857
UBR1	119,9	99.8%	99.0%	Johanson-Blizzard syndrome, 243800
UBTF	116,6	100.0%	99.6%	Neurodegeneration, childhood-onset, with brain atrophy, 617672
UCHL1	98,7	99.5%	95.4%	Spastic paraplegia 79, autosomal recessive, 615491 {?Parkinson disease 5, susceptibility to}, 613643
UFC1	121,3	100.0%	100.0%	Neurodevelopmental disorder with spasticity and poor growth, 618076
UFM1	104,9	73.2%	69.6%	Leukodystrophy, hypomyelinating, 14, 617899
UFSP2	130,7	100.0%	99.5%	?Hip dysplasia, Beukes type, 142669 ?Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974
UGT1A1	184,1	100.0%	100.0%	Crigler-Najjar syndrome, type I, 218800 Crigler-Najjar syndrome, type II, 606785 Hyperbilirubinemia, familial transient neonatal, 237900 [Bilirubin, serum level of, QTL1], 601816 [Gilbert syndrome], 143500
UMOD	110,4	97.6%	94.8%	Glomerulocystic kidney disease with hyperuricemia and isosthenuria, 609886 Hyperuricemic nephropathy, familial juvenile 1, 162000 Medullary cystic kidney disease 2, 603860
UMPS	149,3	100.0%	98.8%	Orotic aciduria, 258900
UNC119	115,7	100.0%	99.7%	?Cone-rod dystrophy, 0 ?Immunodeficiency 13, 615518
UNC13D	108,2	99.8%	98.7%	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC45B	114,7	99.9%	99.2%	?Cataract 43, 616279
UNC80	111,2	100.0%	99.5%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801
UNG	116,8	99.5%	95.6%	Immunodeficiency with hyper IgM, type 5, 608106
UPB1	143	100.0%	100.0%	Beta-ureidopropionase deficiency, 613161

UPF3B	61,6	95.3%	84.8%	Mental retardation, X-linked, syndromic 14, 300676
UQCC2	132,2	100.0%	98.1%	Mitochondrial complex III deficiency, nuclear type 7, 615824
UQCC3	123	100.0%	99.9%	?Mitochondrial complex III deficiency, nuclear type 9, 616111
UQCRB	106	99.2%	95.4%	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC2	105,3	99.6%	97.2%	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRQ	158,9	100.0%	100.0%	Mitochondrial complex III deficiency, nuclear type 4, 615159
UROC1	132,8	100.0%	99.9%	?Urocanase deficiency, 276880
UROD	130,8	98.9%	95.6%	Porphyria cutanea tarda, 176100 Porphyria, hepatoerythropoietic, 176100
UROS	103,8	100.0%	99.7%	Porphyria, congenital erythropoietic, 263700
USB1	118,2	99.8%	97.2%	Poikiloderma with neutropenia, 604173
USH1C	92,5	99.9%	99.2%	Deafness, autosomal recessive 18A, 602092 Usher syndrome, type 1C, 276904
USH1G	191,3	99.9%	98.8%	Usher syndrome, type 1G, 606943
USH2A	129,3	100.0%	99.7%	Retinitis pigmentosa 39, 613809 Usher syndrome, type 2A, 276901
USP18	144,4	95.9%	95.9%	Pseudo-TORCH syndrome 2, 617397
USP27X	155,1	100.0%	100.0%	Mental retardation, X-linked 105, 300984
USP8	64,9	98.0%	89.8%	Pituitary adenoma 4, ACTH-secreting, somatic, 219090
USP9X	93,9	98.3%	92.3%	Mental retardation, X-linked 99, 300919 Mental retardation, X-linked 99, syndromic, female-restricted, 300968
UVSSA	122,6	99.2%	98.9%	UV-sensitive syndrome 3, 614640
VAC14	98,9	99.8%	98.6%	Striatonigral degeneration, childhood-onset, 617054
VAMP1	135,4	100.0%	100.0%	Myasthenic syndrome, congenital, 25, 618323 Spastic ataxia 1, autosomal dominant, 108600
VANGL1	149,8	100.0%	99.9%	Caudal regression syndrome, 600145 {Neural tube defects, susceptibility to}, 182940
VANGL2	159,2	100.0%	99.6%	Neural tube defects, 182940
VAPB	92,2	99.9%	99.0%	Amyotrophic lateral sclerosis 8, 608627 Spinal muscular atrophy, late-onset, Finkel type, 182980
VARS	129,8	100.0%	99.9%	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802
VARS2	120,1	100.0%	99.8%	Combined oxidative phosphorylation deficiency 20, 615917
VAX1	95,1	99.5%	95.7%	?Microphthalmia, syndromic 11, 614402
VCAN	153,1	100.0%	100.0%	Wagner syndrome 1, 143200
VCL	100,5	99.9%	98.9%	Cardiomyopathy, dilated, 1W, 611407 Cardiomyopathy, hypertrophic, 15, 613255

VCP	100,3	100.0%	99.2%	Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954 Charcot-Marie-Tooth disease, type 2Y, 616687 Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320
VDR	108,8	99.1%	96.0%	?Osteoporosis, involutional, 166710 Rickets, vitamin D-resistant, type IIA, 277440
VEGFC	161,7	100.0%	100.0%	Lymphatic malformation 4, 615907
VHL	169,6	100.0%	98.3%	Erythrocytosis, familial, 2, 263400 Hemangioblastoma, cerebellar, somatic, 0 Pheochromocytoma, 171300 Renal cell carcinoma, somatic, 144700 von Hippel-Lindau syndrome, 193300
VIM	129,2	99.7%	98.3%	Cataract 30, pulverulent, 116300
VIPAS39	114,7	100.0%	99.9%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VKORC1	146,5	100.0%	99.9%	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473 Warfarin resistance, 122700
VLDLR	141,4	100.0%	99.9%	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VMA21	89,6	99.5%	93.0%	Myopathy, X-linked, with excessive autophagy, 310440
VPS11	119,6	95.4%	93.1%	Leukodystrophy, hypomyelinating, 12, 616683
VPS13A	78,2	99.2%	95.3%	Choreoacanthocytosis, 200150
VPS13B	134,5	99.3%	98.0%	Cohen syndrome, 216550
VPS13C	110,2	99.5%	97.0%	Parkinson disease 23, autosomal recessive, early onset, 616840
VPS13D	138	100.0%	99.7%	Spinocerebellar ataxia, autosomal recessive 4, 607317
VPS33A	103,9	96.2%	94.4%	Mucopolysaccharidosis-plus syndrome, 617303
VPS33B	107,2	100.0%	99.9%	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
VPS37A	64,3	91.3%	79.3%	Spastic paraplegia 53, autosomal recessive, 614898
VPS45	126,5	97.3%	94.4%	Neutropenia, severe congenital, 5, autosomal recessive, 615285
VPS53	111,3	91.1%	89.6%	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	129,6	99.8%	98.7%	Pontocerebellar hypoplasia type 1A, 607596
VSX1	66,6	97.8%	89.5%	?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195 Keratoconus 1, 148300
VSX2	120,2	100.0%	99.8%	Microphthalmia with coloboma 3, 610092 Microphthalmia, isolated 2, 610093
VWA3B	124,9	99.9%	98.9%	?Spinocerebellar ataxia, autosomal recessive 22, 616948
VWF	98,2	99.9%	99.1%	von Willebrand disease, type 1, 193400 von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 von Willibrand disease, type 3, 277480

WAC	146,3	100.0%	99.2%	Desanto-Shinawi syndrome, 616708
WARS	97,6	99.5%	97.0%	Neuronopathy, distal hereditary motor, type IX, 617721
WARS2	132,3	99.9%	99.1%	Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710
WAS	70,4	94.2%	83.6%	Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, 313900 Thrombocytopenia, X-linked, intermittent, 313900 Wiskott-Aldrich syndrome, 301000
WASHC4	108,9	99.3%	95.9%	?Mental retardation, autosomal recessive 43, 615817
WASHC5	134,2	99.9%	99.5%	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, autosomal dominant, 603563
WBP2	94,3	100.0%	99.9%	Deafness, autosomal recessive 107, 617639
WDFY3	124,6	99.9%	99.3%	?Microcephaly 18, primary, autosomal dominant, 617520
WDPCP	106,7	97.8%	94.9%	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR11	115	97.5%	96.2%	Hypogonadotropic hypogonadism 14 with or without anosmia, 614858
WDR19	126,8	100.0%	99.2%	?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307
WDR26	98,2	99.5%	97.4%	Skraban-Deardorff syndrome, 617616
WDR34	116,1	100.0%	100.0%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
WDR35	141,8	99.7%	98.4%	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091
WDR36	125,1	99.8%	97.6%	Glaucoma 1, open angle, G, 609887
WDR4	142,2	100.0%	100.0%	Galloway-Mowat syndrome 6, 618347 Microcephaly, growth deficiency, seizures, and brain malformations, 618346
WDR45	68,7	96.8%	88.9%	Neurodegeneration with brain iron accumulation 5, 300894
WDR45B	72,7	97.5%	90.1%	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977
WDR60	108,1	99.7%	98.1%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WDR62	152,6	100.0%	99.8%	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
WDR66	118,9	100.0%	99.8%	Spermatogenic failure 33, 618152
WDR72	123,8	96.8%	96.1%	Amelogenesis imperfecta, type IIA3, 613211
WDR73	153,2	100.0%	99.9%	Galloway-Mowat syndrome 1, 251300
WDR81	184,8	100.0%	100.0%	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185

				Hydrocephalus, congenital, 3, with brain anomalies, 617967
WEE2	97,9	99.9%	98.7%	Oocyte maturation defect 5, 617996
WFS1	189,9	100.0%	99.9%	?Cataract 41, 116400 Deafness, autosomal dominant 6/14/38, 600965 Wolfram syndrome 1, 222300 Wolfram-like syndrome, autosomal dominant, 614296 {Diabetes mellitus, noninsulin-dependent, association with}, 125853
WHRN	132,8	99.9%	99.0%	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
WIPF1	89,1	100.0%	99.1%	?Wiskott-Aldrich syndrome 2, 614493
WISP3	NC	NC	NC	Arthropathy, progressive pseudorheumatoid, of childhood, 208230 Spondyloepiphyseal dysplasia tarda with progressive arthropathy, 208230
WNK1	134,4	100.0%	99.5%	Neuropathy, hereditary sensory and autonomic, type II, 201300 Pseudohypoaldosteronism, type IIC, 614492
WNK4	143,3	99.9%	99.5%	Pseudohypoaldosteronism, type IIB, 614491
WNT1	255,8	100.0%	99.8%	Osteogenesis imperfecta, type XV, 615220 {Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221
WNT10A	141,8	100.0%	99.9%	Odontoonychodermal dysplasia, 257980 Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400
WNT10B	157	100.0%	100.0%	Split-hand/foot malformation 6, 225300 Tooth agenesis, selective, 8, 617073
WNT2B	132	99.6%	97.1%	Diarrhea 9, 618168
WNT3	169,3	100.0%	99.8%	?Tetra-amelia syndrome 1, 273395
WNT4	226,5	99.5%	97.3%	?SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330
WNT5A	159	100.0%	100.0%	Robinow syndrome, autosomal dominant 1, 180700
WNT7A	195,8	100.0%	100.0%	Fuhrmann syndrome, 228930 Ulna and fibula, absence of, with severe limb deficiency, 276820
WRAP53	162,8	100.0%	100.0%	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	124,8	99.7%	98.8%	Werner syndrome, 277700
WT1	90,1	100.0%	99.3%	Denys-Drash syndrome, 194080 Frasier syndrome, 136680 Meacham syndrome, 608978 Mesothelioma, somatic, 156240 Nephrotic syndrome, type 4, 256370 Wilms tumor, type 1, 194070

WWOX	116,1	100.0%	99.9%	Epileptic encephalopathy, early infantile, 28, 616211 Esophageal squamous cell carcinoma, somatic, 133239 Spinocerebellar ataxia, autosomal recessive 12, 614322
XDH	93,8	100.0%	99.7%	Xanthinuria, type I, 278300
XIAP	89,4	93.3%	87.9%	Lymphoproliferative syndrome, X-linked, 2, 300635
XIST	NC	NC	NC	X-inactivation, familial skewed, 300087
XK	85,4	100.0%	99.4%	McLeod syndrome with or without chronic granulomatous disease, 300842
XPA	74,7	99.7%	98.2%	Xeroderma pigmentosum, group A, 278700
XPC	143,5	100.0%	99.8%	Xeroderma pigmentosum, group C, 278720
XPNPEP3	99,9	100.0%	99.4%	Nephronophthisis-like nephropathy 1, 613159
XPR1	126	100.0%	99.6%	Basal ganglia calcification, idiopathic, 6, 616413
XRCC1	111,4	99.9%	99.1%	?Spinocerebellar ataxia, autosomal recessive 26, 617633
XRCC2	171,8	99.8%	96.5%	?Fanconi anemia, complementation group U, 617247
XRCC4	143	99.9%	99.0%	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	128,1	99.9%	98.2%	Desbuquois dysplasia 2, 615777 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
XYLT2	147,5	99.7%	98.1%	Spondyloocular syndrome, 605822 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
YAP1	91,1	98.2%	92.3%	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433
YARS	105,5	100.0%	99.2%	Charcot-Marie-Tooth disease, dominant intermediate C, 608323
YARS2	175,2	99.9%	99.6%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
YME1L1	103,9	98.2%	93.5%	?Optic atrophy 11, 617302
YWHAG	167,1	100.0%	100.0%	Epileptic encephalopathy, early infantile, 56, 617665
YY1	128,4	100.0%	98.4%	Gabriele-de Vries syndrome, 617557
YY1AP1	143,6	98.6%	97.1%	Grange syndrome, 602531
ZAP70	186,1	100.0%	99.9%	Autoimmune disease, multisystem, infantile-onset, 2, 617006 Immunodeficiency 48, 269840
ZBTB11	164,8	100.0%	99.7%	Intellectual developmental disorder, autosomal recessive 69, 618383
ZBTB16	148,4	100.0%	100.0%	Leukemia, acute promyelocytic, PL2F/RARA type, 0 Skeletal defects, genital hypoplasia, and mental retardation, 612447
ZBTB18	177,3	99.9%	99.2%	Mental retardation, autosomal dominant 22, 612337
ZBTB20	180,2	100.0%	100.0%	Primrose syndrome, 259050
ZBTB24	155,5	100.0%	100.0%	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069
ZBTB42	131,4	100.0%	100.0%	?Lethal congenital contracture syndrome 6, 616248
ZC3H14	152,9	99.8%	98.5%	Mental retardation, autosomal recessive 56, 617125
ZC4H2	72,4	99.8%	95.9%	Wieacker-Wolff syndrome, 314580

ZDHHC15	85,9	98.9%	95.1%	?Mental retardation, X-linked 91, 300577
ZDHHC9	48,8	97.7%	87.0%	Mental retardation, X-linked syndromic, Raymond type, 300799
ZEB1	151,3	100.0%	99.8%	Corneal dystrophy, Fuchs endothelial, 6, 613270 Corneal dystrophy, posterior polymorphous, 3, 609141
ZEB2	140,1	99.7%	98.4%	Mowat-Wilson syndrome, 235730
ZFHX2	129,9	100.0%	99.8%	?Marsili syndrome, 147430
ZFHX3	119,1	100.0%	99.8%	Prostate cancer, somatic, 176807
ZFP57	106,5	99.9%	98.9%	Diabetes mellitus, transient neonatal, 1, 601410
ZFPM2	155,6	100.0%	99.8%	46XY sex reversal 9, 616067 Diaphragmatic hernia 3, 610187 Tetralogy of Fallot, 187500
ZFYVE26	104,8	99.9%	98.7%	Spastic paraplegia 15, autosomal recessive, 270700
ZFYVE27	110,4	100.0%	100.0%	Spastic paraplegia 33, autosomal dominant, 610244
ZIC1	279,6	100.0%	100.0%	Craniosynostosis 6, 616602
ZIC2	165,6	97.5%	95.4%	Holoprosencephaly 5, 609637
ZIC3	140,9	100.0%	99.8%	Congenital heart defects, nonsyndromic, 1, X-linked, 306955 Heterotaxy, visceral, 1, X-linked, 306955 VACTERL association, X-linked, 314390
ZMPSTE24	128,7	100.0%	99.6%	Mandibuloacral dysplasia with type B lipodystrophy, 608612 Restrictive dermopathy, lethal, 275210
ZMYND10	123	100.0%	100.0%	Ciliary dyskinesia, primary, 22, 615444
ZMYND11	119,9	99.9%	99.5%	Mental retardation, autosomal dominant 30, 616083
ZMYND15	148,8	100.0%	99.8%	?Spermatogenic failure 14, 615842
ZNF141	120,8	100.0%	100.0%	?Polydactyly, postaxial, type A6, 615226
ZNF148	157,2	100.0%	99.8%	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260
ZNF335	134,2	100.0%	99.8%	Microcephaly 10, primary, autosomal recessive, 615095
ZNF341	118,8	98.9%	96.8%	Hyper-IgE recurrent infection syndrome 3, autosomal recessive, 618282
ZNF408	144,5	100.0%	100.0%	?Exudative vitreoretinopathy 6, 616468 Retinitis pigmentosa 72, 616469
ZNF423	192,9	100.0%	100.0%	Joubert syndrome 19, 614844 Nephronophthisis 14, 614844
ZNF469	157,6	100.0%	100.0%	Brittle cornea syndrome 1, 229200
ZNF513	135,3	100.0%	100.0%	?Retinitis pigmentosa 58, 613617
ZNF644	155,4	99.9%	99.8%	Myopia 21, autosomal dominant, 614167
ZNF687	169,9	100.0%	100.0%	Paget disease of bone 6, 616833
ZNF711	114,4	99.7%	98.1%	Mental retardation, X-linked 97, 300803

ZNF750	176,9	100.0%	99.9%	Seborrhea-like dermatitis with psoriasiform elements, 610227
ZNHIT3	136	74.4%	74.4%	PEHO syndrome, 260565
ZP1	178,3	100.0%	100.0%	Oocyte maturation defect 1, 615774
ZP2	115,2	99.8%	98.8%	Oocyte maturation defect 6, 618353
ZP3	144,7	100.0%	100.0%	Oocyte maturation defect 3, 617712
ZSWIM6	120,9	96.4%	93.7%	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.
Median Coverage describes the average number of reads seen across 50 exomes.
% 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 Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : May 8th, 2019.
This list is accurate for panel version DG 2.16
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors