MENDELIOME GENE PANEL DG 3.2.0 (4878 genes)

Releasedate: 16-09-2021

Gene	Agilent V5 covered >10x	Agilent V5 covered >20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype Description and OMIM disease ID
A2M	100	99,3	100	100	No OMIM disease ID
A2ML1	99,9	99,3	100	100	No OMIM disease ID
A4GALT	100	100	100	100	NOR polyagglutination syndrome, 111400
AAAS	100	99,4	100	100	Achalasia-addisonianism-alacrimia syndrome, 231550
AAGAB	100	99,8	100	100	Keratoderma, palmoplantar, punctate type IA, 148600
AARS1	100	99,7	100	99,9	Developmental and epileptic encephalopathy 29, 616339 Charcot-Marie-Tooth disease, axonal, type 2N, 613287
AARS2	100	99,4	100	100	Leukoencephalopathy, progressive, with ovarian failure, 615889 Combined oxidative phosphorylation deficiency 8, 614096
AASS	99,9	99,8	100	99,9	Hyperlysinemia, 238700
ABAT	99,9	97,8	100	100	GABA-transaminase deficiency, 613163
ABCA1	99,8	97,8	100	100	Tangier disease, 205400 HDL deficiency, familial, 1, 604091
ABCA12	99,4	98,7	100	100	Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500 Ichthyosis, congenital, autosomal recessive 4A, 601277
ABCA2	99,9	99,2	100	99,9	Intellectual developmental disorder with poor growth and with or without seizures or ataxia, 618808
ABCA3	99,8	99	100	100	Surfactant metabolism dysfunction, pulmonary, 3, 610921
ABCA4	99,9	99	96,5	96,5	Retinal dystrophy, early-onset severe, 248200 Retinitis pigmentosa 19, 601718 Cone-rod dystrophy 3, 604116 Fundus flavimaculatus, 248200 Stargardt disease 1, 248200
ABCA5	97,5	90,2	100	99,7	?Hypertrichosis, congenital generalized, with gingival hyperplasia, 135400
ABCB10	77,6	68,9	99,4	96,8	No OMIM disease ID

ABCB11	*100*	*99,4*	*100*	*100*	*Cholestasis, benign recurrent intrahepatic, 2, 605479* *Cholestasis, progressive familial intrahepatic 2, 601847*
ABCB4	*99,9*	*99,3*	*100*	*100*	*Gallbladder disease 1, 600803* *Cholestasis, intrahepatic, of pregnancy, 3, 614972* *Cholestasis, progressive familial intrahepatic 3, 602347*
ABCB6	*100*	*100*	*100*	*100*	*Microphthalmia, isolated, with coloboma 7, 614497* *Dyschromatosis universalis hereditaria 3, 615402* *Pseudohyperkalemia, familial, 2, due to red cell leak, 609153*
ABCB7	*99,5*	*97,1*	*99,5*	*98,7*	*Anemia, sideroblastic, with ataxia, 301310*
ABCC1	*98,9*	*97,1*	*100*	*100*	*?Deafness, autosomal dominant 77, 618915*
ABCC2	*100*	*99,9*	*100*	*100*	*Dubin-Johnson syndrome, 237500*
ABCC6	*93,6*	*92,5*	*100*	*100*	*Pseudoxanthoma elasticum, 264800* *Arterial calcification, generalized, of infancy, 2, 614473* *Pseudoxanthoma elasticum, forme fruste, 177850*
ABCC8	*100*	*99,5*	*100*	*100*	*Diabetes mellitus, permanent neonatal 3, with or without neurologic features, 618857* *Diabetes mellitus, transient neonatal 2, 610374* *Diabetes mellitus, noninsulin-dependent, 125853* *Hypoglycemia of infancy, leucine-sensitive, 240800* *Hyperinsulinemic hypoglycemia, familial, 1, 256450*
ABCC9	*100*	*99,8*	*100*	*100*	*Cardiomyopathy, dilated, 1O, 608569* *Hypertrichotic osteochondrodysplasia, 239850* *?Atrial fibrillation, familial, 12, 614050*
ABCD1	*76*	*72,6*	*100*	*100*	*Adrenoleukodystrophy, 300100* *Adrenomyeloneuropathy, adult, 300100*
ABCD2	*100*	*99,8*	*100*	*100*	*No OMIM disease ID*
ABCD3	*99,4*	*98*	*100*	*100*	*?Bile acid synthesis defect, congenital, 5, 616278*
ABCD4	*99,8*	*97,7*	*100*	*100*	*Methylmalonic aciduria and homocystinuria, cblJ type, 614857*
ABCG5	*99,9*	*99,9*	*100*	*100*	*Sitosterolemia 2, 618666*
ABCG8	*99*	*97,1*	*100*	*100*	*Sitosterolemia 1, 210250*
ABHD12	*91,7*	*86*	*100*	*99,8*	*Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674*
ABHD5	*100*	*100*	*100*	*100*	*Chanarin-Dorfman syndrome, 275630*
ABL1	*100*	*100*	*100*	*100*	*Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 608232* *Congenital heart defects and skeletal malformations syndrome, 617602*
ACACA	*98,4*	*97,9*	*100*	*100*	*No OMIM disease ID*

ACAD8	*100*	*100*	*100*	*100*	*Isobutyryl-CoA dehydrogenase deficiency, 611283*
ACAD9	*100*	*99,8*	*100*	*100*	*Mitochondrial complex I deficiency, nuclear type 20, 611126*
ACADM	*99,8*	*97,9*	*100*	*100*	*Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450*
ACADS	*100*	*99,4*	*100*	*100*	*Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470*
ACADSB	*99,8*	*97,9*	*100*	*100*	*2-methylbutyrylglycinuria, 610006*
ACADVL	*99,7*	*96,6*	*100*	*100*	*VLCAD deficiency, 201475*
ACAN	*96,9*	*92,5*	*98,9*	*98,7*	*?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	*99,6*	*97,9*	*100*	*99,7*	*Alpha-methylacetoacetic aciduria, 203750*
ACAT2	*100*	*99,9*	*100*	*100*	*No OMIM disease ID*
ACBD5	*100*	*98,4*	*100*	*99,9*	*Retinal dystrophy with leukodystrophy, 618863*
ACD	*100*	*99,9*	*100*	*100*	*?Dyskeratosis congenita, autosomal recessive 7, 616553* *?Dyskeratosis congenita, autosomal dominant 6, 616553*
ACE	*99,9*	*98,4*	*100*	*99,9*	*Renal tubular dysgenesis, 267430*
ACER3	*99,5*	*99*	*100*	*100*	*?Leukodystrophy, progressive, early childhood-onset, 617762*
ACKR3	*100*	*100*	*100*	*100*	*?Oculomotor-abducens synkinesis, 619215*
ACO2	*94,1*	*86,3*	*100*	*100*	*?Optic atrophy 9, 616289* *Infantile cerebellar-retinal degeneration, 614559*
ACOX1	*100*	*99,3*	*100*	*100*	*Mitchell syndrome, 618960* *Peroxisomal acyl-CoA oxidase deficiency, 264470*
ACOX2	*100*	*99*	*100*	*100*	*Bile acid synthesis defect, congenital, 6, 617308*
ACP4	*98,4*	*91,4*	*100*	*100*	*Amelogenesis imperfecta, type IJ, 617297*
ACP5	*99,9*	*98,9*	*100*	*100*	*Spondyloenchondrodysplasia with immune dysregulation, 607944*
ACSF3	*100*	*99,5*	*100*	*100*	*Combined malonic and methylmalonic aciduria, 614265*
ACSL4	*98,3*	*94,2*	*100*	*99,6*	*Intellectual developmental disorder, X-linked 63, 300387*
ACSL6	*95,8*	*94,4*	*97,1*	*97,1*	*Myelodysplastic syndrome,* *Myelogenous leukemia, acute,*
ACTA1	*98,2*	*89,5*	*100*	*100*	*?Myopathy, scapulohumeroperoneal, 616852* *Nemaline myopathy 3, autosomal dominant or recessive, 161800* *Myopathy, actin, congenital, with excess of thin myofilaments, 161800*

					Myopathy, actin, congenital, with cores, 161800 *Myopathy, congenital, with fiber-type disproportion 1, 255310*
ACTA2	*100*	*98,9*	*100*	*100*	*Multisystemic smooth muscle dysfunction syndrome, 613834* *Aortic aneurysm, familial thoracic 6, 611788* *Moyamoya disease 5, 614042*
ACTB	*99,9*	*97,2*	*100*	*100*	*Baraitser-Winter syndrome 1, 243310* *?Dystonia, juvenile-onset, 607371*
ACTC1	*99,9*	*98,9*	*100*	*100*	*Left ventricular noncompaction 4, 613424* *Cardiomyopathy, hypertrophic, 11, 612098* *Atrial septal defect 5, 612794* *Cardiomyopathy, dilated, 1R, 613424*
ACTG1	*100*	*100*	*100*	*100*	*Deafness, autosomal dominant 20/26, 604717* *Baraitser-Winter syndrome 2, 614583*
ACTG2	*99,7*	*97,4*	*100*	*100*	*Megacystis-microcolon-intestinal hypoperistalsis syndrome 5, 619431* *Visceral myopathy 1, 155310*
ACTL6A	*99,8*	*98,9*	*100*	*99,9*	*No OMIM disease ID*
ACTL6B	*100*	*100*	*100*	*100*	*Developmental and epileptic encephalopathy 76, 618468* *Intellectual developmental disorder with severe speech and ambulation defects, 618470*
ACTL9	*100*	*100*	*100*	*100*	*Spermatogenic failure 53, 619258*
ACTN1	*100*	*99,9*	*100*	*100*	*Bleeding disorder, platelet-type, 15, 615193*
ACTN2	*100*	*100*	*100*	*100*	*Myopathy, distal, 6, adult onset, 618655* *Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158* *Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158* *Myopathy, congenital with structured cores and Z-line abnormalities, 618654*
ACTN4	*99,9*	*98*	*100*	*100*	*Glomerulosclerosis, focal segmental, 1, 603278*
ACVR1	*100*	*99,9*	*100*	*100*	*Fibrodysplasia ossificans progressiva, 135100*
ACVR1B	*99,1*	*96*	*100*	*100*	*Pancreatic cancer, somatic,*
ACVR2B	*98,6*	*95,1*	*100*	*100*	*Heterotaxy, visceral, 4, autosomal, 613751*
ACVRL1	*99,9*	*98,1*	*100*	*100*	*Telangiectasia, hereditary hemorrhagic, type 2, 600376*
ACY1	*100*	*99,7*	*100*	*100*	*Aminoacylase 1 deficiency, 609924*
ADA	*99,7*	*96,1*	*100*	*100*	*Adenosine deaminase deficiency, partial, 102700* *Severe combined immunodeficiency due to ADA deficiency, 102700*
ADA2	*99,9*	*97,3*	*100*	*100*	*Sneddon syndrome, 182410* *Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688*
ADAM10	*94,7*	*93,8*	*100*	*100*	*Reticulate acropigmentation of Kitamura, 615537*

ADAM17	*99,6*	*98*	*100*	*99,9*	*?Inflammatory skin and bowel disease, neonatal, 1, 614328*
ADAM22	*99,8*	*99,6*	*100*	*100*	*Developmental and epileptic encephalopathy 61, 617933*
ADAM9	*99,7*	*98*	*100*	*100*	*Cone-rod dystrophy 9, 612775*
ADAMTS1	*100*	*100*	*100*	*100*	*No OMIM disease ID*
ADAMTS10	*100*	*99,9*	*100*	*100*	*Weill-Marchesani syndrome 1, recessive, 277600*
ADAMTS13	*97*	*94,3*	*99,8*	*98,9*	*Thrombotic thrombocytopenic purpura, hereditary, 274150*
ADAMTS17	*92,9*	*88,8*	*97,9*	*95,9*	*Weill-Marchesani 4 syndrome, recessive, 613195*
ADAMTS18	*100*	*99,6*	*100*	*100*	*Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458*
ADAMTS19	*95,7*	*92,6*	*100*	*100*	*No OMIM disease ID*
ADAMTS2	*99,8*	*97,5*	*98*	*97,7*	*Ehlers-Danlos syndrome, dermatosparaxis type, 225410*
ADAMTS3	*100*	*100*	*100*	*100*	*Hennekam lymphangiectasia-lymphedema syndrome 3, 618154*
ADAMTS9	*99,4*	*98,4*	*100*	*100*	*No OMIM disease ID*
ADAMTSL2	*98*	*94,7*	*99,9*	*99,8*	*Geleophysic dysplasia 1, 231050*
ADAMTSL4	*99,8*	*98,4*	*100*	*100*	*Ectopia lentis et pupillae, 225200* *Ectopia lentis, isolated, autosomal recessive, 225100*
ADAR	*100*	*99,4*	*100*	*100*	*Dyschromatosis symmetrica hereditaria, 127400* *Aicardi-Goutieres syndrome 6, 615010*
ADARB1	*97,2*	*95,3*	*95,1*	*95,1*	*Neurodevelopmental disorder with hypotonia, microcephaly, and seizures, 618862*
ADAT3	*100*	*100*	*100*	*100*	*Neurodevelopmental disorder with brain abnormalities, poor growth, and dysmorphic facies, 615286*
ADCK2	*100*	*99,7*	*100*	*100*	*No OMIM disease ID*
ADCK5	*100*	*100*	*100*	*100*	*No OMIM disease ID*
ADCY1	*95,7*	*94,3*	*98,4*	*97,7*	*?Deafness, autosomal recessive 44, 610154*
ADCY10	*100*	*99,6*	*100*	*100*	*No OMIM disease ID*
ADCY3	*100*	*99*	*100*	*100*	*No OMIM disease ID*
ADCY5	*95,9*	*92,5*	*99,2*	*97,9*	*Dyskinesia, familial, with facial myokymia, 606703*
ADCY6	*100*	*100*	*100*	*100*	*Lethal congenital contracture syndrome 8, 616287*
ADD3	*99,8*	*98,8*	*100*	*100*	*Cerebral palsy, spastic quadriplegic, 3, 617008*
ADGRE2	*96,8*	*95,8*	*99,1*	*98,6*	*Vibratory urticaria, 125630*
ADGRG1	*100*	*100*	*100*	*100*	*Polymicrogyria, bilateral frontoparietal, 606854* *Polymicrogyria, bilateral perisylvian, 615752*

ADGRG2	*97,2*	*90,2*	*100*	*99,9*	*Congenital bilateral absence of vas deferens, X-linked, 300985*
ADGRG6	*99,7*	*98,7*	*100*	*100*	*Lethal congenital contracture syndrome 9, 616503*
ADGRV1	*99,5*	*98,4*	*100*	*100*	*Usher syndrome, type 2C, 605472* *Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472* *?Febrile seizures, familial, 4, 604352*
ADH5	*99,7*	*98,9*	*100*	*100*	*AMED syndrome, digenic, 619151*
ADIPOQ	*100*	*100*	*100*	*100*	*Adiponectin deficiency, 612556*
ADIPOR1	*99,2*	*94,4*	*100*	*100*	*No OMIM disease ID*
ADK	*83,3*	*79,7*	*84,5*	*84,5*	*Hypermethioninemia due to adenosine kinase deficiency, 614300*
ADNP	*90,5*	*90,5*	*95,4*	*95,4*	*Helsmoortel-van der Aa syndrome, 615873*
ADPRS	*100*	*99,9*	*100*	*100*	*Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170*
ADRB2	*100*	*99,3*	*100*	*100*	*Beta-2-adrenoreceptor agonist, reduced response to,*
ADSL	*99,2*	*98,6*	*100*	*100*	*Adenylosuccinase deficiency, 103050*
ADSS1	*90,8*	*87*	*100*	*100*	*Myopathy, distal, 5, 617030*
AEBP1	*100*	*100*	*100*	*100*	*Ehlers-Danlos syndrome, classic-like, 2, 618000*
AFF2	*99,8*	*98,8*	*100*	*99,6*	*Intellectual developmental disorder, X-linked 109, 309548*
AFF3	*98,6*	*97,9*	*100*	*100*	*KINSSHIP syndrome, 619297*
AFF4	*99,8*	*98,2*	*100*	*100*	*CHOPS syndrome, 616368*
AFG3L2	*94,6*	*86,3*	*100*	*100*	*Spastic ataxia 5, autosomal recessive, 614487* *Optic atrophy 12, 618977* *Spinocerebellar ataxia 28, 610246*
AFP	*96,6*	*89*	*100*	*99,9*	*Alpha-fetoprotein deficiency, 615969*
AGA	*100*	*99,9*	*100*	*100*	*Aspartylglucosaminuria, 208400*
AGAP1	*97,1*	*91*	*100*	*99,5*	*No OMIM disease ID*
AGBL1	*98,5*	*98,1*	*100*	*100*	*Corneal dystrophy, Fuchs endothelial, 8, 615523*
AGBL5	*99,6*	*98,4*	*100*	*100*	*Retinitis pigmentosa 75, 617023*
AGK	*90,4*	*87,9*	*91,2*	*91,1*	*Cataract 38, autosomal recessive, 614691* *Sengers syndrome, 212350*
AGL	*99,8*	*99,5*	*100*	*100*	*Glycogen storage disease IIIa, 232400* *Glycogen storage disease IIIb, 232400*
AGMO	*99*	*92,4*	*100*	*99,9*	*No OMIM disease ID*

AGO2	*99,1*	*99,1*	*99,7*	*99,3*	*Lessel-Kreienkamp syndrome, 619149*
AGPAT2	*99*	*94*	*100*	*100*	*Lipodystrophy, congenital generalized, type 1, 608594*
AGPS	*98,8*	*95,2*	*100*	*99,4*	*Rhizomelic chondrodysplasia punctata, type 3, 600121*
AGRN	*97,6*	*92,6*	*100*	*99,9*	*Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120*
AGT	*100*	*99,9*	*100*	*100*	*Renal tubular dysgenesis, 267430*
AGTPBP1	*96,3*	*94,2*	*100*	*100*	*Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276*
AGTR1	*91,9*	*91,8*	*100*	*100*	*Renal tubular dysgenesis, 267430*
AGXT	*100*	*100*	*100*	*100*	*Hyperoxaluria, primary, type 1, 259900*
AHCY	*99,9*	*98,8*	*100*	*100*	*Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752*
AHDC1	*100*	*98,9*	*100*	*100*	*Xia-Gibbs syndrome, 615829*
AHI1	*99,4*	*97,4*	*100*	*100*	*Joubert syndrome 3, 608629*
AHNAK2	*97,3*	*96,9*	*96,7*	*96,5*	*No OMIM disease ID*
AHR	*99,2*	*98,6*	*100*	*100*	*?Retinitis pigmentosa 85, 618345*
AHSG	*100*	*99,8*	*100*	*100*	*?Alopecia-mental retardation syndrome 1, 203650*
AICDA	*100*	*99,9*	*100*	*100*	*Immunodeficiency with hyper-IgM, type 2, 605258*
AIFM1	*99,9*	*97,8*	*100*	*100*	*Combined oxidative phosphorylation deficiency 6, 300816* *Cowchock syndrome, 310490* *Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232* *Deafness, X-linked 5, 300614*
AIMP1	*99,2*	*92,5*	*100*	*99,9*	*Leukodystrophy, hypomyelinating, 3, 260600*
AIMP2	*89,4*	*86*	*100*	*99,9*	*Leukodystrophy, hypomyelinating, 17, 618006*
AIP	*100*	*99,6*	*100*	*100*	*Pituitary adenoma 1, multiple types, 102200* *Pituitary adenoma predisposition, 102200*
AIPL1	*100*	*99,2*	*100*	*100*	*Leber congenital amaurosis 4, 604393* *Retinitis pigmentosa, juvenile, 604393* *Cone-rod dystrophy, 604393*
AIRE	*100*	*99,9*	*100*	*100*	*Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300*
AK1	*100*	*100*	*100*	*100*	*Hemolytic anemia due to adenylate kinase deficiency, 612631*
AK2	*98,7*	*95,2*	*100*	*99,7*	*Reticular dysgenesis, 267500*
AK7	*99,4*	*96,3*	*100*	*99,9*	*?Spermatogenic failure 27, 617965*
AKAP9	*98,4*	*94,6*	*100*	*100*	*?Long QT syndrome 11, 611820*

AKR1C1	*94,5*	*87,4*	*100*	*100*	*No OMIM disease ID*
AKR1C2	*94,5*	*88,3*	*100*	*100*	*46XY sex reversal 8, 614279*
AKR1D1	*99,8*	*98,6*	*100*	*100*	*Bile acid synthesis defect, congenital, 2, 235555*
AKT1	*100*	*99,9*	*100*	*100*	*Breast cancer, somatic, 114480* *Cowden syndrome 6, 615109* *Colorectal cancer, somatic, 114500* *Proteus syndrome, somatic, 176920* *Ovarian cancer, somatic, 167000*
AKT2	*100*	*99,7*	*100*	*100*	*Diabetes mellitus, type II, 125853* *Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900*
AKT3	*97,4*	*92,4*	*100*	*100*	*Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937*
ALAD	*97,8*	*92,5*	*100*	*100*	*Porphyria, acute hepatic, 612740*
ALAS2	*98,7*	*93,2*	*100*	*100*	*Anemia, sideroblastic, 1, 300751* *Protoporphyria, erythropoietic, X-linked, 300752*
ALB	*99,8*	*99,4*	*100*	*100*	*Analbuminemia, 616000*
ALDH18A1	*100*	*99,9*	*100*	*100*	*Spastic paraplegia 9A, autosomal dominant, 601162* *Cutis laxa, autosomal recessive, type IIIA, 219150* *Spastic paraplegia 9B, autosomal recessive, 616586* *Cutis laxa, autosomal dominant 3, 616603*
ALDH1A2	*100*	*99*	*100*	*100*	*No OMIM disease ID*
ALDH1A3	*97,6*	*94,6*	*100*	*100*	*Microphthalmia, isolated 8, 615113*
ALDH1B1	*100*	*100*	*100*	*100*	*No OMIM disease ID*
ALDH2	*100*	*100*	*100*	*100*	*Alcohol sensitivity, acute, 610251*
ALDH3A2	*88,8*	*88,4*	*93,2*	*93,2*	*Sjogren-Larsson syndrome, 270200*
ALDH4A1	*100*	*99,7*	*100*	*100*	*Hyperprolinemia, type II, 239510*
ALDH5A1	*92,4*	*83,5*	*100*	*100*	*Succinic semialdehyde dehydrogenase deficiency, 271980*
ALDH6A1	*100*	*99,6*	*100*	*100*	*Methylmalonate semialdehyde dehydrogenase deficiency, 614105*
ALDH7A1	*91,1*	*84,5*	*100*	*100*	*Epilepsy, pyridoxine-dependent, 266100*
ALDOA	*98,9*	*96,3*	*100*	*100*	*Glycogen storage disease XII, 611881*
ALDOB	*98,8*	*95,7*	*100*	*100*	*Fructose intolerance, hereditary, 229600*
ALG1	*53,6*	*46,9*	*100*	*100*	*Congenital disorder of glycosylation, type Ik, 608540*
ALG10	*100*	*99,7*	*100*	*100*	*No OMIM disease ID*

ALG11	*96,8*	*96,8*	*96,8*	*96,8*	*Congenital disorder of glycosylation, type Ip, 613661*
ALG12	*100*	*99,9*	*100*	*100*	*Congenital disorder of glycosylation, type Ig, 607143*
ALG13	*97,4*	*90*	*99,9*	*99,4*	*?Congenital disorder of glycosylation, type Is, 300884* *Developmental and epileptic encephalopathy 36, 300884*
ALG14	*100*	*99,9*	*100*	*100*	*Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies, 619031* *Myopathy, epilepsy, and progressive cerebral atrophy, 619036* *?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227*
ALG2	*100*	*100*	*100*	*100*	*?Congenital disorder of glycosylation, type Ii, 607906* *Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228*
ALG3	*100*	*99,5*	*100*	*100*	*Congenital disorder of glycosylation, type Id, 601110*
ALG6	*98,2*	*93,7*	*100*	*99,9*	*Congenital disorder of glycosylation, type Ic, 603147*
ALG8	*96,6*	*95,9*	*96,6*	*96,6*	*Congenital disorder of glycosylation, type Ih, 608104* *Polycystic liver disease 3 with or without kidney cysts, 617874*
ALG9	*99,9*	*99,3*	*100*	*100*	*Gillessen-Kaesbach-Nishimura syndrome, 263210* *Congenital disorder of glycosylation, type Il, 608776*
ALK	*100*	*99,4*	*100*	*100*	*No OMIM disease ID*
ALKBH1	*100*	*99,7*	*100*	*100*	*No OMIM disease ID*
ALKBH8	*99,7*	*98,6*	*100*	*100*	*Intellectual developmental disorder, autosomal recessive 71, 618504*
ALMS1	*99,7*	*99,5*	*100*	*100*	*Alstrom syndrome, 203800*
ALOX12B	*100*	*99,7*	*100*	*100*	*Ichthyosis, congenital, autosomal recessive 2, 242100*
ALOXE3	*99,9*	*99*	*100*	*100*	*Ichthyosis, congenital, autosomal recessive 3, 606545*
ALPI	*100*	*99,7*	*100*	*100*	*No OMIM disease ID*
ALPK1	*99,9*	*99,3*	*100*	*100*	*ROSAH syndrome, 614979*
ALPK3	*98,1*	*95,1*	*100*	*100*	*Cardiomyopathy, familial hypertrophic 27, 618052*
ALPL	*100*	*99,4*	*100*	*100*	*Odontohypophosphatasia, 146300* *Hypophosphatasia, infantile, 241500* *Hypophosphatasia, childhood, 241510* *Hypophosphatasia, adult, 146300*
ALS2	*99,9*	*99,8*	*100*	*100*	*Primary lateral sclerosis, juvenile, 606353* *Spastic paralysis, infantile onset ascending, 607225* *Amyotrophic lateral sclerosis 2, juvenile, 205100*
ALX1	*99,6*	*95,2*	*100*	*100*	*Frontonasal dysplasia 3, 613456*
ALX3	*80,2*	*72,8*	*100*	*100*	*Frontonasal dysplasia 1, 136760*

ALX4	*100*	*99,9*	*100*	*100*	*Parietal foramina 2, 609597* *Frontonasal dysplasia 2, 613451*
AMACR	*100*	*100*	*100*	*100*	*Alpha-methylacyl-CoA racemase deficiency, 614307* *Bile acid synthesis defect, congenital, 4, 214950*
AMBN	*99,6*	*97,7*	*100*	*99,9*	*Amelogenesis imperfecta, type IF, 616270*
AMELX	*99,4*	*94*	*100*	*100*	*Amelogenesis imperfecta, type 1E, 301200*
AMER1	*99,6*	*96,6*	*100*	*100*	*Osteopathia striata with cranial sclerosis, 300373*
AMH	*99,4*	*92,9*	*100*	*100*	*Persistent Mullerian duct syndrome, type I, 261550*
AMHR2	*100*	*99,6*	*100*	*100*	*Persistent Mullerian duct syndrome, type II, 261550*
AMMECR1	*99,9*	*98,4*	*100*	*100*	*Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990*
AMN	*92,5*	*82,9*	*100*	*100*	*Imerslund-Grasbeck syndrome 2, 618882*
AMPD1	*100*	*99,5*	*100*	*100*	*Myopathy due to myoadenylate deaminase deficiency, 615511*
AMPD2	*99,8*	*99*	*100*	*100*	*?Spastic paraplegia 63, 615686* *Pontocerebellar hypoplasia, type 9, 615809*
AMPD3	*100*	*99,2*	*100*	*100*	*No OMIM disease ID*
AMT	*100*	*100*	*100*	*100*	*Glycine encephalopathy, 605899*
AMTN	*99,9*	*99,1*	*100*	*100*	*?Amelogenesis imperfecta, type IIIB, 617607*
ANAPC1	*58,8*	*56,8*	*100*	*99,9*	*Rothmund-Thomson syndrome, type 1, 618625*
ANG	*100*	*99,7*	*100*	*100*	*Amyotrophic lateral sclerosis 9, 611895*
ANGPT1	*99,7*	*98,7*	*100*	*100*	*?Angioedema, hereditary, 5, 619361*
ANGPT2	*99,9*	*99,8*	*100*	*99,9*	*Lymphatic malformation 10, 619369*
ANGPTL3	*98,6*	*92*	*100*	*100*	*Hypobetalipoproteinemia, familial, 2, 605019*
ANGPTL4	*100*	*99,3*	*100*	*100*	*Plasma triglyceride level QTL, low, 615881*
ANK1	*99,9*	*98,7*	*100*	*100*	*Spherocytosis, type 1, 182900*
ANK2	*100*	*99,9*	*100*	*100*	*Long QT syndrome 4, 600919* *Cardiac arrhythmia, ankyrin-B-related, 600919*
ANK3	*99,3*	*99*	*100*	*100*	*Mental retardation, autosomal recessive, 37, 615493*
ANKFY1	*100*	*98,7*	*100*	*100*	*No OMIM disease ID*
ANKH	*100*	*99,9*	*100*	*100*	*Chondrocalcinosis 2, 118600* *Craniometaphyseal dysplasia, 123000*
ANKLE2	*100*	*99,3*	*100*	*99,7*	*Microcephaly 16, primary, autosomal recessive, 616681*

ANKRD1	*99,9*	*98,3*	*100*	*100*	*No OMIM disease ID*
ANKRD11	*97*	*94*	*100*	*100*	*KBG syndrome, 148050*
ANKRD17	*99,4*	*98,2*	*100*	*100*	*Chopra-Amiel-Gordon syndrome, 619504*
ANKRD26	*94,6*	*88,5*	*97,2*	*97*	*Thrombocytopenia 2, 188000*
ANKS1B	*99,9*	*99,3*	*100*	*100*	*No OMIM disease ID*
ANKS6	*94,2*	*89,7*	*97*	*95*	*Nephronophthisis 16, 615382*
ANLN	*98,4*	*97*	*100*	*100*	*Focal segmental glomerulosclerosis 8, 616032*
ANO10	*99,2*	*96,6*	*100*	*100*	*Spinocerebellar ataxia, autosomal recessive 10, 613728*
ANO3	*91,8*	*90,4*	*100*	*100*	*Dystonia 24, 615034*
ANO5	*99,2*	*96,9*	*100*	*100*	*Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307* *Miyoshi muscular dystrophy 3, 613319* *Gnathodiaphyseal dysplasia, 166260*
ANO6	*99,2*	*96,8*	*100*	*100*	*Scott syndrome, 262890*
ANOS1	*89,8*	*88,3*	*99,9*	*99,4*	*Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700*
ANTXR1	*99,9*	*99,1*	*100*	*100*	*GAPO syndrome, 230740*
ANTXR2	*99,5*	*97,2*	*100*	*100*	*Hyaline fibromatosis syndrome, 228600*
ANXA11	*99,8*	*97,6*	*100*	*100*	*Amyotrophic lateral sclerosis 23, 617839*
AP1B1	*100*	*99,4*	*100*	*100*	*Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150*
AP1S1	*99,9*	*99,4*	*100*	*100*	*MEDNIK syndrome, 609313*
AP1S2	*73,8*	*66,8*	*100*	*98,8*	*Pettigrew syndrome, 304340*
AP1S3	*90,4*	*90,1*	*90,5*	*90,5*	*No OMIM disease ID*
AP2M1	*100*	*99,9*	*100*	*100*	*Intellectual developmental disorder 60 with seizures, 618587*
AP2S1	*90,4*	*90*	*100*	*100*	*Hypocalciuric hypercalcemia, type III, 600740*
AP3B1	*99,2*	*96,4*	*100*	*99,9*	*Hermansky-Pudlak syndrome 2, 608233*
AP3B2	*93,3*	*89,8*	*99,9*	*99*	*Developmental and epileptic encephalopathy 48, 617276*
AP3D1	*99,6*	*98,4*	*100*	*100*	*?Hermansky-Pudlak syndrome 10, 617050*
AP4B1	*99,8*	*98,5*	*100*	*100*	*Spastic paraplegia 47, autosomal recessive, 614066*
AP4E1	*99,5*	*98,6*	*100*	*99,9*	*Stuttering, familial persistent, 1, 184450* *Spastic paraplegia 51, autosomal recessive, 613744*
AP4M1	*99,8*	*97,8*	*100*	*100*	*Spastic paraplegia 50, autosomal recessive, 612936*

AP4S1	*78,9*	*71*	*87,9*	*87,9*	*Spastic paraplegia 52, autosomal recessive, 614067*
AP5Z1	*100*	*99,9*	*100*	*100*	*Spastic paraplegia 48, autosomal recessive, 613647*
APC	*99,9*	*99,7*	*100*	*100*	*Colorectal cancer, somatic, 114500* *Brain tumor-polyposis syndrome 2, 175100* *Desmoid disease, hereditary, 135290* *Adenoma, periampullary, somatic, 175100* *Hepatoblastoma, somatic, 114550* *Gastric cancer, somatic, 613659* *Gastric adenocarcinoma and proximal polyposis of the stomach, 619182* *Gardner syndrome, 175100* *Adenomatous polyposis coli, 175100*
APC2	*98,3*	*94,8*	*99,7*	*98,5*	*Cortical dysplasia, complex, with other brain malformations 10, 618677* *?Sotos syndrome 3, 617169*
APCDD1	*100*	*99,7*	*100*	*100*	*Hypotrichosis 1, 605389*
APOA1	*100*	*100*	*100*	*100*	*Hypoalphalipoproteinemia, primary, 2, with or without corneal clouding, 618463* *Amyloidosis, 3 or more types, 105200* *ApoA-I and apoC-III deficiency, combined, 618463*
APOA2	*84,5*	*81,6*	*100*	*100*	*Apolipoprotein A-II deficiency,*
APOA5	*100*	*99,9*	*100*	*99,8*	*Hyperchylomicronemia, late-onset, 144650*
APOB	*99,9*	*99,5*	*100*	*100*	*Hypercholesterolemia, familial, 2, 144010* *Hypobetalipoproteinemia, 615558*
APOC2	*100*	*100*	*100*	*100*	*Hyperlipoproteinemia, type Ib, 207750*
APOC3	*100*	*100*	*100*	*100*	*Apolipoprotein C-III deficiency, 614028*
APOE	*100*	*98,8*	*100*	*100*	*Alzheimer disease 2, 104310* *Sea-blue histiocyte disease, 269600* *Lipoprotein glomerulopathy, 611771* *Hyperlipoproteinemia, type III, 617347*
APOL1	*100*	*100*	*100*	*100*	*No OMIM disease ID*
APOO	*81,1*	*71,1*	*100*	*100*	*No OMIM disease ID*
APP	*100*	*99,9*	*100*	*100*	*Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants, 605714* *Alzheimer disease 1, familial, 104300*
APRT	*100*	*100*	*100*	*100*	*Adenine phosphoribosyltransferase deficiency, 614723*
APTX	*94,1*	*90,6*	*100*	*100*	*Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920*
AQP2	*100*	*99,2*	*100*	*100*	*Diabetes insipidus, nephrogenic, 2, 125800*

AQP5	*99,9*	*97,5*	*100*	*100*	*Palmoplantar keratoderma, Bothnian type, 600231*
AR	*98,1*	*93,6*	*99,9*	*99,5*	*Androgen insensitivity, partial, with or without breast cancer, 312300* *Androgen insensitivity, 300068* *Spinal and bulbar muscular atrophy of Kennedy, 313200* *Hypospadias 1, X-linked, 300633*
ARCN1	*96,8*	*96,6*	*96,7*	*96,6*	*Short stature, rhizomelic, with microcephaly, micrognathia, and developmental delay, 617164*
ARF1	*100*	*99,9*	*100*	*100*	*Periventricular nodular heterotopia 8, 618185*
ARFGEF2	*99,7*	*98,7*	*100*	*100*	*Periventricular heterotopia with microcephaly, 608097*
ARG1	*92,9*	*92,9*	*92,9*	*92,7*	*Argininemia, 207800*
ARHGAP24	*100*	*100*	*100*	*100*	*No OMIM disease ID*
ARHGAP26	*90,4*	*90,2*	*100*	*100*	*Leukemia, juvenile myelomonocytic, somatic, 607785*
ARHGAP29	*98,9*	*97,6*	*100*	*99,9*	*No OMIM disease ID*
ARHGAP31	*99,7*	*98,2*	*100*	*100*	*Adams-Oliver syndrome 1, 100300*
ARHGAP35	*100*	*100*	*100*	*100*	*No OMIM disease ID*
ARHGDIA	*100*	*100*	*100*	*100*	*Nephrotic syndrome, type 8, 615244*
ARHGEF1	*99,9*	*98,4*	*100*	*100*	*?Immunodeficiency 62, 618459*
ARHGEF10	*99,6*	*97*	*100*	*100*	*?Slowed nerve conduction velocity, AD, 608236*
ARHGEF18	*98,8*	*93,9*	*100*	*100*	*Retinitis pigmentosa 78, 617433*
ARHGEF2	*93*	*92,6*	*100*	*100*	*?Neurodevelopmental disorder with midbrain and hindbrain malformations, 617523*
ARHGEF28	*99,3*	*95,3*	*100*	*100*	*No OMIM disease ID*
ARHGEF6	*99*	*94,8*	*100*	*99,8*	*No OMIM disease ID*
ARHGEF9	*76,3*	*72,5*	*97,2*	*97,1*	*Developmental and epileptic encephalopathy 8, 300607*
ARID1A	*98,3*	*96*	*100*	*100*	*Coffin-Siris syndrome 2, 614607*
ARID1B	*96,2*	*94,6*	*97,8*	*96,9*	*Coffin-Siris syndrome 1, 135900*
ARID2	*99,7*	*98,2*	*100*	*100*	*Coffin-Siris syndrome 6, 617808*
ARIH1	*99,7*	*99,3*	*100*	*100*	*No OMIM disease ID*
ARL13B	*100*	*99,3*	*100*	*100*	*Joubert syndrome 8, 612291*
ARL2	*100*	*99,7*	*100*	*100*	*?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 1, 619082*
ARL2BP	*94,9*	*87,3*	*100*	*100*	*Retinitis pigmentosa with or without situs inversus, 615434*

ARL3	*99,7*	*96,5*	*100*	*100*	*Retinitis pigmentosa 83, 618173* *Joubert syndrome 35, 618161*
ARL6	*99,1*	*98,4*	*100*	*100*	*Retinitis pigmentosa 55, 613575* *Bardet-Biedl syndrome 3, 600151*
ARL6IP1	*98,5*	*81,5*	*100*	*100*	*?Spastic paraplegia 61, autosomal recessive, 615685*
ARMC2	*99,8*	*99,1*	*100*	*99,9*	*Spermatogenic failure 38, 618433*
ARMC5	*100*	*99,1*	*100*	*100*	*ACTH-independent macronodular adrenal hyperplasia 2, 615954*
ARMC9	*99,9*	*99,4*	*100*	*100*	*Joubert syndrome 30, 617622*
ARNT2	*100*	*100*	*100*	*99,5*	*?Webb-Dattani syndrome, 615926*
ARPC1B	*100*	*100*	*100*	*100*	*Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia, 617718*
ARR3	*100*	*99,6*	*100*	*100*	*Myopia 26, X-linked, female-limited, 301010*
ARSA	*100*	*99,8*	*100*	*100*	*Metachromatic leukodystrophy, 250100*
ARSB	*98,8*	*91*	*100*	*100*	*Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200*
ARSG	*99,9*	*98*	*100*	*100*	*Usher syndrome, type IV, 618144*
ARSL	*98,9*	*92,4*	*100*	*99,8*	*Chondrodysplasia punctata, X-linked recessive, 302950*
ARV1	*99,9*	*98,8*	*100*	*99,9*	*Developmental and epileptic encephalopathy 38, 617020*
ARX	*82,1*	*67,5*	*91,4*	*86,6*	*Proud syndrome, 300004* *Hydranencephaly with abnormal genitalia, 300215* *Partington syndrome, 309510* *Developmental and epileptic encephalopathy 1, 308350* *Lissencephaly, X-linked 2, 300215* *Intellectual developmental disorder, X-linked 29, 300419*
ASAH1	*99,1*	*97,3*	*100*	*100*	*Spinal muscular atrophy with progressive myoclonic epilepsy, 159950* *Farber lipogranulomatosis, 228000*
ASB10	*99,4*	*95,7*	*100*	*100*	*Glaucoma 1, open angle, F, 603383*
ASCC1	*92,5*	*89,4*	*87,1*	*87*	*Spinal muscular atrophy with congenital bone fractures 2, 616867* *Barrett esophagus/esophageal adenocarcinoma, 614266*
ASCL1	*100*	*99,4*	*100*	*100*	*No OMIM disease ID*
ASH1L	*98,7*	*98,6*	*98,7*	*98,7*	*Mental retardation, autosomal dominant 52, 617796*
ASIP	*100*	*100*	*100*	*100*	*No OMIM disease ID*
ASL	*100*	*99,7*	*100*	*100*	*Argininosuccinic aciduria, 207900*
ASNS	*98,1*	*91,2*	*100*	*100*	*Asparagine synthetase deficiency, 615574*

ASPA	*99,9*	*99,1*	*100*	*100*	*Canavan disease, 271900*
ASPH	*99,6*	*98,6*	*100*	*99,9*	*Traboulsi syndrome, 601552*
ASPM	*99,4*	*97,9*	*100*	*99,9*	*Microcephaly 5, primary, autosomal recessive, 608716*
ASPRV1	*100*	*99,4*	*100*	*100*	*Ichthyosis, lamellar, autosomal dominant, 146750*
ASPSCR1	*99,8*	*98,3*	*100*	*100*	*Alveolar soft-part sarcoma, 606243*
ASRGL1	*100*	*99,9*	*100*	*100*	*No OMIM disease ID*
ASS1	*93,2*	*83,2*	*100*	*100*	*Citrullinemia, 215700*
ASXL1	*99,8*	*98,9*	*100*	*100*	*Myelodysplastic syndrome, somatic, 614286* *Bohring-Opitz syndrome, 605039*
ASXL2	*99,9*	*99,5*	*100*	*100*	*Shashi-Pena syndrome, 617190*
ASXL3	*99,9*	*99,6*	*100*	*100*	*Bainbridge-Ropers syndrome, 615485*
ATAD1	*99,1*	*91,6*	*100*	*100*	*Hyperekplexia 4, 618011*
ATAD3A	*91,4*	*86,7*	*100*	*100*	*Harel-Yoon syndrome, 617183* *Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810*
ATAD3B	*91,1*	*83,2*	*100*	*100*	*No OMIM disease ID*
ATCAY	*100*	*100*	*100*	*100*	*Ataxia, cerebellar, Cayman type, 601238*
ATF3	*99,9*	*97,3*	*100*	*100*	*No OMIM disease ID*
ATF6	*99,9*	*99,5*	*100*	*100*	*Achromatopsia 7, 616517*
ATG4A	*99*	*94,3*	*99,9*	*97,9*	*No OMIM disease ID*
ATG5	*98,4*	*96,4*	*100*	*100*	*?Spinocerebellar ataxia, autosomal recessive 25, 617584*
ATG7	*100*	*99,7*	*100*	*100*	*Spinocerebellar ataxia, autosomal recessive 31, 619422*
ATIC	*99,8*	*99,1*	*100*	*100*	*AICA-ribosiduria due to ATIC deficiency, 608688*
ATL1	*99,9*	*99,5*	*100*	*99,8*	*Spastic paraplegia 3A, autosomal dominant, 182600* *Neuropathy, hereditary sensory, type ID, 613708*
ATL3	*99,6*	*97,6*	*100*	*99,9*	*Neuropathy, hereditary sensory, type IF, 615632*
ATM	*99,4*	*97,1*	*100*	*100*	*Ataxia-telangiectasia, 208900* *Lymphoma, B-cell non-Hodgkin, somatic,* *T-cell prolymphocytic leukemia, somatic,* *Lymphoma, mantle cell, somatic,*
ATN1	*99,8*	*98*	*100*	*100*	*Dentatorubral-pallidoluysian atrophy, 125370* *Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494*
ATOH1	*100*	*100*	*100*	*100*	*No OMIM disease ID*

ATOH7	*97,6*	*92,6*	*99,2*	*94,1*	*Persistent hyperplastic primary vitreous, autosomal recessive, 221900*
ATP11C	*97,8*	*91,5*	*99,9*	*98,9*	*?Hemolytic anemia, congenital, X-linked, 301015*
ATP13A2	*100*	*99,6*	*100*	*100*	*Spastic paraplegia 78, autosomal recessive, 617225* *Kufor-Rakeb syndrome, 606693*
ATP1A1	*100*	*99,8*	*100*	*100*	*Hypomagnesemia, seizures, and mental retardation 2, 618314* *Charcot-Marie-Tooth disease, axonal, type 2DD, 618036*
ATP1A2	*100*	*99,8*	*100*	*100*	*Alternating hemiplegia of childhood 1, 104290* *Migraine, familial basilar, 602481* *Migraine, familial hemiplegic, 2, 602481*
ATP1A3	*100*	*99,9*	*100*	*100*	*Alternating hemiplegia of childhood 2, 614820* *Dystonia-12, 128235* *CAPOS syndrome, 601338*
ATP2A1	*100*	*100*	*100*	*100*	*Brody myopathy, 601003*
ATP2A2	*100*	*99,6*	*100*	*100*	*Acrokeratosis verruciformis, 101900* *Darier disease, 124200*
ATP2B2	*100*	*99,8*	*100*	*100*	*No OMIM disease ID*
ATP2B3	*99,7*	*97,4*	*100*	*100*	*?Spinocerebellar ataxia, X-linked 1, 302500*
ATP2C1	*99,8*	*99,5*	*100*	*99,9*	*Hailey-Hailey disease, 169600*
ATP4A	*100*	*98,6*	*100*	*100*	*No OMIM disease ID*
ATP5F1A	*92,2*	*83*	*100*	*100*	*?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4, 615228* *?Combined oxidative phosphorylation deficiency 22, 616045*
ATP5F1B	*99,8*	*97,7*	*100*	*100*	*No OMIM disease ID*
ATP5F1C	*95,5*	*89,3*	*100*	*100*	*No OMIM disease ID*
ATP5F1D	*97,4*	*91,8*	*100*	*100*	*Mitochondrial complex V (ATP synthase) deficiency, 618120*
ATP5F1E	*100*	*100*	*100*	*100*	*?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053*
ATP5IF1	*100*	*100*	*100*	*100*	*No OMIM disease ID*
ATP5MC1	*100*	*99,6*	*100*	*100*	*No OMIM disease ID*
ATP5MC2	*99,3*	*92,4*	*100*	*100*	*No OMIM disease ID*
ATP5MC3	*100*	*100*	*100*	*100*	*No OMIM disease ID*
ATP5ME	*100*	*100*	*100*	*100*	*No OMIM disease ID*
ATP5MF	*99,6*	*94,8*	*100*	*100*	*No OMIM disease ID*
ATP5MG	*100*	*100*	*100*	*100*	*No OMIM disease ID*

ATP5MGL	*100*	*100*	*100*	*100*	*No OMIM disease ID*
ATP5MD	*83,8*	*35,7*	*100*	*100*	*Mitochondrial complex V (ATP synthase) deficiency, nuclear type 6, 618683*
ATP5PB	*97*	*83,3*	*100*	*100*	*No OMIM disease ID*
ATP5PD	*89,2*	*67,8*	*100*	*100*	*No OMIM disease ID*
ATP5PF	*99,9*	*92,4*	*100*	*100*	*No OMIM disease ID*
ATP5PO	*100*	*98,1*	*100*	*99,7*	*No OMIM disease ID*
ATP6AP1	*98,2*	*93*	*100*	*100*	*Immunodeficiency 47, 300972*
ATP6AP2	*89,9*	*69,7*	*100*	*99,8*	*Intellectual developmental disorder, X-linked, syndromic, Hedera type, 300423* *?Parkinsonism with spasticity, X-linked, 300911* *Congenital disorder of glycosylation, type IIr, 301045*
ATP6V0A1	*99,8*	*98,6*	*100*	*100*	*No OMIM disease ID*
ATP6V0A2	*99,9*	*98,7*	*100*	*100*	*Wrinkly skin syndrome, 278250* *Cutis laxa, autosomal recessive, type IIA, 219200*
ATP6V0A4	*100*	*99,3*	*100*	*100*	*Distal renal tubular acidosis 3, with or without sensorineural hearing loss, 602722*
ATP6V0C	*100*	*100*	*100*	*100*	*No OMIM disease ID*
ATP6V1A	*99,8*	*98,4*	*100*	*100*	*Cutis laxa, autosomal recessive, type IID, 617403* *Developmental and epileptic encephalopathy 93, 618012*
ATP6V1B1	*100*	*100*	*100*	*100*	*Distal renal tubular acidosis 2 with progressive sensorineural hearing loss, 267300*
ATP6V1B2	*99,9*	*99,3*	*100*	*100*	*Zimmermann-Laband syndrome 2, 616455* *Deafness, congenital, with onychodystrophy, autosomal dominant, 124480*
ATP6V1E1	*92,5*	*86,1*	*100*	*100*	*Cutis laxa, autosomal recessive, type IIC, 617402*
ATP7A	*98,7*	*96*	*100*	*99,9*	*Occipital horn syndrome, 304150* *Spinal muscular atrophy, distal, X-linked 3, 300489* *Menkes disease, 309400*
ATP7B	*99,9*	*99,2*	*100*	*100*	*Wilson disease, 277900*
ATP8A2	*100*	*99,6*	*100*	*100*	*?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268*
ATP8B1	*96,6*	*93,5*	*100*	*100*	*Cholestasis, progressive familial intrahepatic 1, 211600* *Cholestasis, intrahepatic, of pregnancy, 1, 147480* *Cholestasis, benign recurrent intrahepatic, 243300*
ATPAF1	*83,7*	*71,4*	*100*	*100*	*No OMIM disease ID*
ATPAF2	*100*	*99,9*	*100*	*100*	*?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273*
ATR	*99,7*	*99*	*100*	*100*	*Seckel syndrome 1, 210600* *?Cutaneous telangiectasia and cancer syndrome, familial, 614564*

ATRX	*98,7*	*95,2*	*100*	*99,9*	*Alpha-thalassemia/mental retardation syndrome, 301040* *Alpha-thalassemia myelodysplasia syndrome, somatic, 300448* *Mental retardation-hypotonic facies syndrome, X-linked, 309580*
ATXN1	*100*	*99,8*	*100*	*100*	*Spinocerebellar ataxia 1, 164400*
ATXN10	*99,9*	*99,3*	*100*	*100*	*Spinocerebellar ataxia 10, 603516*
ATXN2	*93,3*	*84,9*	*99,3*	*96,8*	*Spinocerebellar ataxia 2, 183090*
ATXN2L	*98,3*	*94,7*	*100*	*100*	*No OMIM disease ID*
ATXN3	*94,2*	*88,2*	*95,8*	*95,7*	*Machado-Joseph disease, 109150*
ATXN7	*99,6*	*96,1*	*98,8*	*97,2*	*Spinocerebellar ataxia 7, 164500*
ATXN8OS	*NC*	*NC*	*NC*	*NC*	*Spinocerebellar ataxia 8, 608768*
AUH	*99,7*	*99,4*	*100*	*99,9*	*3-methylglutaconic aciduria, type I, 250950*
AURKC	*99,9*	*97,2*	*100*	*100*	*Spermatogenic failure 5, 243060*
AUTS2	*98,8*	*96,9*	*100*	*100*	*Mental retardation, autosomal dominant 26, 615834*
AVIL	*100*	*99,8*	*100*	*100*	*Nephrotic syndrome, type 21, 618594*
AVP	*90,2*	*66,6*	*100*	*100*	*Diabetes insipidus, neurohypophyseal, 125700*
AVPR2	*100*	*99,8*	*100*	*100*	*Diabetes insipidus, nephrogenic, 1, 304800* *Nephrogenic syndrome of inappropriate antidiuresis, 300539*
AXIN1	*99,9*	*98,9*	*100*	*100*	*Hepatocellular carcinoma, somatic, 114550* *?Caudal duplication anomaly, 607864*
AXIN2	*100*	*99,8*	*100*	*100*	*Colorectal cancer, somatic, 114500* *Oligodontia-colorectal cancer syndrome, 608615*
AXL	*100*	*98,9*	*100*	*100*	*No OMIM disease ID*
B2M	*100*	*100*	*100*	*100*	*?Amyloidosis, familial visceral, 105200* *Immunodeficiency 43, 241600*
B3GALNT1	*100*	*100*	*100*	*100*	*No OMIM disease ID*
B3GALNT2	*94,3*	*89,8*	*92,5*	*92,5*	*Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181*
B3GALT6	*77*	*73*	*91,7*	*81*	*Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349* *Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640* *Al-Gazali syndrome, 609465*
B3GAT3	*99,4*	*96,6*	*95,4*	*94,8*	*Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600*
B3GLCT	*99,7*	*98,2*	*100*	*99,4*	*Peters-plus syndrome, 261540*
B4GALNT1	*98,3*	*93,5*	*100*	*100*	*Spastic paraplegia 26, autosomal recessive, 609195*

B4GALT1	*100*	*99,3*	*100*	*100*	*Congenital disorder of glycosylation, type IId, 607091*
B4GALT7	*99,7*	*96,8*	*100*	*99,4*	*Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070*
B4GAT1	*100*	*100*	*100*	*100*	*Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287*
B9D1	*85,2*	*85,2*	*95,8*	*94*	*?Meckel syndrome 9, 614209* *Joubert syndrome 27, 617120*
B9D2	*100*	*100*	*100*	*100*	*?Meckel syndrome 10, 614175* *Joubert syndrome 34, 614175*
BAAT	*99,5*	*97,5*	*100*	*100*	*Hypercholanemia, familial, 607748* *Bile acid conjugation defect 1, 619232*
BACH2	*100*	*99,9*	*100*	*100*	*Immunodeficiency 60, 618394*
BAG3	*100*	*99,7*	*100*	*100*	*Cardiomyopathy, dilated, 1HYPOGONADOTROPIC HYPOGONADISM, 613881* *Myopathy, myofibrillar, 6, 612954*
BANF1	*95,3*	*78,1*	*100*	*100*	*Nestor-Guillermo progeria syndrome, 614008*
BAP1	*83,9*	*82,4*	*100*	*100*	*Tumor predisposition syndrome, 614327*
BARD1	*100*	*99,8*	*100*	*100*	*No OMIM disease ID*
BAX	*98,3*	*95,6*	*100*	*100*	*Colorectal cancer, somatic, 114500* *T-cell acute lymphoblastic leukemia, somatic, 613065*
BAZ2B	*99,6*	*98,8*	*100*	*99,9*	*No OMIM disease ID*
BBIP1	*95,7*	*87,4*	*100*	*100*	*?Bardet-Biedl syndrome 18, 615995*
BBS1	*100*	*100*	*100*	*100*	*Bardet-Biedl syndrome 1, 209900*
BBS10	*100*	*99,9*	*100*	*100*	*Bardet-Biedl syndrome 10, 615987*
BBS12	*100*	*100*	*100*	*100*	*Bardet-Biedl syndrome 12, 615989*
BBS2	*99,4*	*98*	*100*	*100*	*Retinitis pigmentosa 74, 616562* *Bardet-Biedl syndrome 2, 615981*
BBS4	*99,9*	*98,9*	*100*	*99,9*	*Bardet-Biedl syndrome 4, 615982*
BBS5	*98,4*	*94,7*	*100*	*100*	*Bardet-Biedl syndrome 5, 615983*
BBS7	*99*	*96,5*	*100*	*99,9*	*Bardet-Biedl syndrome 7, 615984*
BBS9	*92*	*89*	*95,8*	*95,8*	*Bardet-Biedl syndrome 9, 615986*
BCAP31	*92,1*	*79,1*	*100*	*99,6*	*Deafness, dystonia, and cerebral hypomyelination, 300475*
BCAS3	*99,1*	*98,9*	*100*	*100*	*No OMIM disease ID*
BCAT1	*100*	*100*	*100*	*100*	*No OMIM disease ID*
BCAT2	*100*	*100*	*100*	*100*	*?Hypervalinemia or hyperleucine-isoleucinemia, 618850*

BCHE	*100*	*99,8*	*100*	*100*	*Butyrylcholinesterase deficiency, 617936*
BCKDHA	*99,8*	*97,9*	*100*	*100*	*Maple syrup urine disease, type Ia, 248600*
BCKDHB	*99,8*	*95,4*	*100*	*100*	*Maple syrup urine disease, type Ib, 248600*
BCKDK	*100*	*100*	*100*	*100*	*Branched-chain ketoacid dehydrogenase kinase deficiency, 614923*
BCL10	*100*	*100*	*100*	*100*	*?Immunodeficiency 37, 616098* *Lymphoma, MALT, somatic, 137245*
BCL11A	*97,3*	*96*	*100*	*100*	*Dias-Logan syndrome, 617101*
BCL11B	*99,6*	*96,5*	*99,4*	*97,9*	*Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092* *Immunodeficiency 49, 617237*
BCL2	*100*	*100*	*100*	*100*	*Leukemia/lymphoma, B-cell, 2,*
BCL7A	*100*	*100*	*100*	*100*	*B-cell non-Hodgkin lymphoma, high-grade,*
BCO1	*100*	*100*	*100*	*100*	*?Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300*
BCOR	*99,2*	*95,8*	*100*	*100*	*Microphthalmia, syndromic 2, 300166*
BCORL1	*99,4*	*97,7*	*100*	*100*	*Shukla-Vernon syndrome, 301029*
BCS1L	*100*	*100*	*100*	*100*	*GRACILE syndrome, 603358* *Mitochondrial complex III deficiency, nuclear type 1, 124000* *Bjornstad syndrome, 262000*
BDP1	*97,4*	*93,1*	*100*	*100*	*?Deafness, autosomal recessive 112, 618257*
BEAN1	*99,2*	*96,5*	*92,2*	*92,2*	*Spinocerebellar ataxia 31, 117210*
BEST1	*98,7*	*95*	*100*	*99,6*	*Macular dystrophy, vitelliform, 2, 153700* *?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 2, 193220* *Retinitis pigmentosa-50, 613194* *Retinitis pigmentosa, concentric, 613194* *Vitreoretinochoroidopathy, 193220* *Bestrophinopathy, autosomal recessive, 611809*
BFSP1	*99,8*	*94*	*100*	*100*	*Cataract 33, multiple types, 611391*
BFSP2	*99,7*	*97,2*	*100*	*100*	*Cataract 12, multiple types, 611597*
BGN	*100*	*99,9*	*100*	*100*	*Meester-Loeys syndrome, 300989* *Spondyloepimetaphyseal dysplasia, X-linked, 300106*
BHLHA9	*72,5*	*53,5*	*99,6*	*96,9*	*?Camptosynpolydactyly, complex, 607539* *Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432*
BICC1	*100*	*100*	*100*	*100*	*No OMIM disease ID*

BICD2	*99,9*	*99,1*	*100*	*100*	*Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291* *Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290*
BICRA	*99,8*	*98,4*	*100*	*100*	*Coffin-Siris syndrome 12, 619325*
BIN1	*99,7*	*96*	*100*	*100*	*Centronuclear myopathy 2, 255200*
BLK	*100*	*100*	*100*	*100*	*Maturity-onset diabetes of the young, type 11, 613375*
BLM	*99,3*	*97,7*	*100*	*100*	*Bloom syndrome, 210900*
BLNK	*96,9*	*92,6*	*100*	*100*	*?Agammaglobulinemia 4, 613502*
BLOC1S1	*100*	*99,1*	*100*	*100*	*No OMIM disease ID*
BLOC1S3	*99,9*	*90,3*	*100*	*100*	*Hermansky-Pudlak syndrome 8, 614077*
BLOC1S5	*99,7*	*98,7*	*100*	*100*	*Hermansky-Pudlak syndrome 11, 619172*
BLOC1S6	*99,3*	*97,6*	*100*	*100*	*?Hermansky-Pudlak syndrome 9, 614171*
BLVRA	*99,8*	*97,8*	*100*	*100*	*Hyperbiliverdinemia, 614156*
BMP1	*100*	*100*	*100*	*100*	*Osteogenesis imperfecta, type XIII, 614856*
BMP15	*100*	*98,7*	*100*	*100*	*Premature ovarian failure 4, 300510* *Ovarian dysgenesis 2, 300510*
BMP2	*100*	*100*	*100*	*100*	*Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 1, 617877* *Brachydactyly, type A2, 112600*
BMP4	*100*	*100*	*100*	*100*	*Orofacial cleft 11, 600625* *Microphthalmia, syndromic 6, 607932*
BMP6	*96,3*	*94*	*99,2*	*96,2*	*No OMIM disease ID*
BMP7	*99,8*	*98,4*	*100*	*100*	*No OMIM disease ID*
BMPER	*100*	*99,6*	*100*	*100*	*Diaphanospondylodysostosis, 608022*
BMPR1A	*99,5*	*94*	*100*	*100*	*Polyposis syndrome, hereditary mixed, 2, 610069* *Polyposis, juvenile intestinal, 174900*
BMPR1B	*99,9*	*99,9*	*100*	*100*	*Acromesomelic dysplasia, Demirhan type, 609441* *Brachydactyly, type A2, 112600* *Brachydactyly, type A1, D, 616849*
BMPR2	*99,9*	*99,9*	*99,9*	*99,9*	*Pulmonary hypertension, familial primary, 1, with or without HYPOGONADOTROPIC HYPOGONADISMT, 178600* *Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600* *Pulmonary venoocclusive disease 1, 265450*
BMS1	*66,7*	*66,1*	*100*	*100*	*?Aplasia cutis congenita, nonsyndromic, 107600*
BNC1	*98,3*	*97,1*	*98,8*	*97,7*	*?Premature ovarian failure 16, 618723*
BNC2	*99,1*	*99,1*	*100*	*100*	*Lower urinary tract obstruction, congenital, 618612*

BOLA1	*100*	*99,9*	*100*	*100*	*No OMIM disease ID*
BOLA2	*100*	*100*	*100*	*100*	*No OMIM disease ID*
BOLA3	*99*	*86,7*	*100*	*100*	*Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299*
BPGM	*100*	*100*	*100*	*100*	*Erythrocytosis, familial, 8, 222800*
IMPAD1	*100*	*99,9*	*100*	*100*	*Chondrodysplasia with joint dislocations, GPAPP type, 614078*
BPTF	*96,1*	*94,3*	*99,6*	*98,4*	*Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, 617755*
BRAF	*89,4*	*77,6*	*100*	*100*	*Melanoma, malignant, somatic, 155600* *LEOPARD syndrome 3, 613707* *Cardiofaciocutaneous syndrome, 115150* *Adenocarcinoma of lung, somatic, 211980* *Noonan syndrome 7, 613706* *Colorectal cancer, somatic, 114500* *Nonsmall cell lung cancer, somatic, 211980*
BRAT1	*99,9*	*98,9*	*100*	*100*	*Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056* *Rigidity and multifocal seizure syndrome, lethal neonatal, 614498*
BRCA1	*99,4*	*98,4*	*100*	*100*	*Fanconi anemia, complementation group S, 617883*
BRCA2	*99,1*	*98,2*	*100*	*100*	*Fanconi anemia, complementation group D1, 605724* *Wilms tumor, 194070*
BRDT	*97,7*	*91,1*	*100*	*100*	*?Spermatogenic failure 21, 617644*
BRF1	*99,8*	*98,4*	*100*	*100*	*Cerebellofaciodental syndrome, 616202*
BRIP1	*99,4*	*98,5*	*100*	*100*	*Fanconi anemia, complementation group J, 609054*
BRPF1	*100*	*100*	*100*	*100*	*Intellectual developmental disorder with dysmorphic facies and ptosis, 617333*
BRSK2	*99,4*	*96,9*	*100*	*100*	*No OMIM disease ID*
BRWD3	*98,7*	*95,3*	*100*	*99,8*	*Intellectual developmental disorder, X-linked 93, 300659*
BSCL2	*100*	*99,9*	*100*	*100*	*Lipodystrophy, congenital generalized, type 2, 269700* *Neuropathy, distal hereditary motor, type VC, 619112* *Silver spastic paraplegia syndrome, 270685* *Encephalopathy, progressive, with or without lipodystrophy, 615924*
BSND	*100*	*99,9*	*100*	*100*	*Sensorineural deafness with mild renal dysfunction, 602522* *Bartter syndrome, type 4a, 602522*
BTD	*83*	*82,9*	*83,1*	*83,1*	*Biotinidase deficiency, 253260*
BTG4	*99,1*	*95,6*	*100*	*100*	*Oocyte maturation defect 8, 619009*

BTK	*100*	*99,7*	*100*	*99,8*	*Agammaglobulinemia, X-linked 1, 300755* *Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200*
BTRC	*97,6*	*97,2*	*100*	*100*	*No OMIM disease ID*
BUB1	*99,7*	*98,4*	*100*	*99,9*	*Colorectal cancer with chromosomal instability, somatic, 114500*
BUB1B	*99,3*	*98,3*	*100*	*100*	*Colorectal cancer, somatic, 114500* *Mosaic variegated aneuploidy syndrome 1, 257300*
BUB3	*99,1*	*97,9*	*100*	*100*	*No OMIM disease ID*
BVES	*99,4*	*98,6*	*100*	*100*	*Muscular dystrophy, limb-girdle, autosomal recessive 25, 616812*
C11orf80	*98,2*	*95*	*91,9*	*91,9*	*Hydatidiform mole, recurrent, 4, 618432*
C12orf4	*99,6*	*99,5*	*100*	*100*	*Mental retardation, autosomal recessive 66, 618221*
C12orf57	*100*	*98,6*	*100*	*100*	*Temtamy syndrome, 218340*
C14orf39	*97,3*	*89,8*	*100*	*99,7*	*Spermatogenic failure 52, 619202* *?Premature ovarian failure 18, 619203*
C19orf12	*100*	*99,8*	*100*	*100*	*Neurodegeneration with brain iron accumulation 4, 614298* *?Spastic paraplegia 43, autosomal recessive, 615043*
C1GALT1C1	*100*	*99,1*	*100*	*100*	*Tn polyagglutination syndrome, somatic, 300622*
C1QA	*100*	*100*	*100*	*100*	*C1q deficiency, 613652*
C1QB	*100*	*100*	*100*	*100*	*C1q deficiency, 613652*
C1QBP	*84,3*	*70,6*	*100*	*100*	*Combined oxidative phosphorylation deficiency 33, 617713*
C1QC	*100*	*99,7*	*100*	*100*	*C1q deficiency, 613652*
C1QTNF5	*91,1*	*78,9*	*100*	*100*	*Retinal degeneration, late-onset, autosomal dominant, 605670*
C1R	*100*	*100*	*99,7*	*98*	*Ehlers-Danlos syndrome, periodontal type, 1, 130080*
C1S	*99,9*	*98,8*	*99,7*	*97,7*	*C1s deficiency, 613783* *Ehlers-Danlos syndrome, periodontal type, 2, 617174*
C2	*100*	*100*	*100*	*100*	*C2 deficiency, 217000*
C2CD3	*95,8*	*95,4*	*95,9*	*95,9*	*Orofaciodigital syndrome XIV, 615948*
C2orf69	*96,7*	*85,6*	*100*	*100*	*Combined oxidative phosphorylation deficiency 53, 619423*
C3	*99,9*	*98,5*	*100*	*100*	*C3 deficiency, 613779*
C4A	*98,3*	*94,9*	*99,5*	*99,1*	*C4a deficiency, 614380*
C4B	*98,8*	*95,6*	*100*	*99,9*	*C4B deficiency, 614379*
C5	*99,6*	*98,2*	*100*	*99,8*	*C5 deficiency, 609536*

C6	*100*	*99,8*	*100*	*100*	*C6 deficiency, 612446* *Combined C6/C7 deficiency,*
C7	*99,8*	*97,4*	*100*	*100*	*C7 deficiency, 610102*
C8A	*100*	*99,4*	*100*	*100*	*C8 deficiency, type I, 613790*
C8B	*99,9*	*98,6*	*100*	*100*	*C8 deficiency, type II, 613789*
C8G	*100*	*100*	*100*	*100*	*No OMIM disease ID*
C9	*99,7*	*99,2*	*100*	*100*	*C9 deficiency, 613825*
C9orf72	*97,4*	*95,5*	*100*	*100*	*Frontotemporal dementia and/or amyotrophic lateral sclerosis 1, 105550*
CA12	*100*	*99,9*	*100*	*100*	*Hyperchlorhidrosis, isolated, 143860*
CA2	*100*	*100*	*100*	*100*	*Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730*
CA4	*100*	*100*	*100*	*100*	*No OMIM disease ID*
CA5A	*87,6*	*85,6*	*87,7*	*87,7*	*Hyperammonemia due to carbonic anhydrase VA deficiency, 615751*
CA8	*99,4*	*96,6*	*100*	*100*	*Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227*
CABIN1	*100*	*99,4*	*100*	*99,9*	*No OMIM disease ID*
CABP2	*75,4*	*64,9*	*100*	*100*	*Deafness, autosomal recessive 93, 614899*
CABP4	*100*	*99,6*	*100*	*100*	*Cone-rod synaptic disorder, congenital nonprogressive, 610427*
CACNA1A	*93,1*	*88,4*	*100*	*99,9*	*Developemental and epileptic encephalopathy 42, 617106* *Spinocerebellar ataxia 6, 183086* *Episodic ataxia, type 2, 108500* *Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500* *Migraine, familial hemiplegic, 1, 141500*
CACNA1B	*98*	*96,1*	*99,3*	*98,2*	*Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497*
CACNA1C	*99,9*	*99,2*	*100*	*100*	*Timothy syndrome, 601005* *Long QT syndrome 8, 618447* *Brugada syndrome 3, 611875*
CACNA1D	*97,9*	*97,7*	*100*	*100*	*Primary aldosteronism, seizures, and neurologic abnormalities, 615474* *Sinoatrial node dysfunction and deafness, 614896*
CACNA1E	*100*	*99,8*	*100*	*100*	*Developmental and epileptic encephalopathy 69, 618285*
CACNA1F	*99,7*	*97,2*	*100*	*100*	*Cone-rod dystrophy, X-linked, 3, 300476* *Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071* *Aland Island eye disease, 300600*
CACNA1G	*100*	*99,2*	*100*	*100*	*Spinocerebellar ataxia 42, 616795* *Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087*

CACNA1H	*98,9*	*96,3*	*100*	*99,9*	*Hyperaldosteronism, familial, type IV, 617027*
CACNA1S	*100*	*99,8*	*100*	*100*	*Hypokalemic periodic paralysis, type 1, 170400*
CACNA2D1	*99*	*96*	*100*	*99,8*	*No OMIM disease ID*
CACNA2D2	*93,8*	*93,1*	*99,6*	*98,4*	*Cerebellar atrophy with seizures and variable developmental delay, 618501*
CACNA2D4	*98,6*	*96,9*	*100*	*100*	*Retinal cone dystrophy 4, 610478*
CACNB2	*98,5*	*98,4*	*100*	*100*	*Brugada syndrome 4, 611876*
CACNB4	*95,8*	*94,4*	*100*	*100*	*Episodic ataxia, type 5, 613855*
CACNG2	*100*	*100*	*100*	*100*	*?Mental retardation, autosomal dominant 10, 614256*
CAD	*99,8*	*98,5*	*100*	*100*	*Developmental and epileptic encephalopathy 50, 616457*
CADM3	*100*	*99,7*	*100*	*100*	*No OMIM disease ID*
CALCRL	*98,9*	*92,8*	*100*	*100*	*?Lymphatic malformation 8, 618773*
CALM1	*99,9*	*97,3*	*100*	*99,9*	*Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916* *Long QT syndrome 14, 616247*
CALM2	*66,5*	*59,7*	*72*	*72*	*Long QT syndrome 15, 616249*
CALM3	*100*	*99,2*	*100*	*100*	*Long QT syndrome 16, 618782* *?Ventricular tachycardia, catecholaminergic polymorphic 6, 618782*
CALR	*94,5*	*87,5*	*100*	*100*	*Myelofibrosis, somatic, 254450* *Thrombocythemia, somatic, 187950*
CAMK2A	*99,8*	*98,4*	*99,9*	*99,6*	*Mental retardation, autosomal dominant 53, 617798* *?Mental retardation, autosomal recessive 63, 618095*
CAMK2B	*100*	*99,4*	*100*	*99,7*	*Mental retardation, autosomal dominant 54, 617799*
CAMK2G	*99,9*	*98,6*	*100*	*100*	*Mental retardation, autosomal dominant 59, 618522*
CAMTA1	*99,6*	*99*	*100*	*100*	*Cerebellar ataxia, nonprogressive, with mental retardation, 614756*
CANT1	*100*	*100*	*100*	*100*	*Desbuquois dysplasia 1, 251450* *Epiphyseal dysplasia, multiple, 7, 617719*
CAPN1	*100*	*100*	*100*	*100*	*Spastic paraplegia 76, autosomal recessive, 616907*
CAPN10	*100*	*99,4*	*100*	*100*	*No OMIM disease ID*
CAPN12	*95*	*87,9*	*100*	*100*	*No OMIM disease ID*
CAPN15	*99,8*	*98,1*	*100*	*100*	*Oculogastrointestinal neurodevelopmental syndrome, 619318*
CAPN3	*97,7*	*96,3*	*97,9*	*97,9*	*Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600* *Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129*
CAPN5	*100*	*99,9*	*100*	*100*	*Vitreoretinopathy, neovascular inflammatory, 193235*

CARD11	*100*	*99,9*	*100*	*100*	*B-cell expansion with NFKB and T-cell anergy, 616452* *Immunodeficiency 11B with atopic dermatitis, 617638* *Immunodeficiency 11A, 615206*
CARD14	*100*	*99,5*	*100*	*100*	*Psoriasis 2, 602723* *Pityriasis rubra pilaris, 173200*
CARD8	*100*	*100*	*100*	*100*	*?Inflammatory bowel disease (Crohn disease) 30, 619079*
CARD9	*100*	*99*	*100*	*100*	*Candidiasis, familial, 2, autosomal recessive, 212050*
CARMIL2	*96,4*	*95,2*	*99,8*	*98,8*	*Immunodeficiency 58, 618131*
CARS1	*100*	*99,4*	*100*	*100*	*Microcephaly, developmental delay, and brittle hair syndrome, 618891*
CARS2	*100*	*100*	*100*	*99,5*	*Combined oxidative phosphorylation deficiency 27, 616672*
CASK	*97,2*	*93,9*	*100*	*99,9*	*Mental retardation, with or without nystagmus, 300422* *Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia, 300749* *FG syndrome 4, 300422*
CASP10	*99,3*	*96,8*	*100*	*100*	*Autoimmune lymphoproliferative syndrome, type II, 603909* *Gastric cancer, somatic, 613659* *Lymphoma, non-Hodgkin, somatic, 605027*
CASP14	*100*	*99,9*	*100*	*100*	*Ichthyosis, congenital, autosomal recessive 12, 617320*
CASP8	*95,6*	*95,4*	*95,6*	*95,6*	*Hepatocellular carcinoma, somatic, 114550* *?Autoimmune lymphoproliferative syndrome, type IIB, 607271*
CASQ1	*99,8*	*96,5*	*100*	*100*	*Myopathy, vacuolar, with CASQ1 aggregates, 616231*
CASQ2	*100*	*99,8*	*100*	*100*	*Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938*
CASR	*100*	*99,5*	*100*	*100*	*Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198* *Hyperparathyroidism, neonatal, 239200* *Hypocalcemia, autosomal dominant, 601198* *Hypocalciuric hypercalcemia, type I, 145980*
CAST	*99*	*95,4*	*100*	*100*	*Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295*
CASZ1	*96,5*	*93,2*	*97,8*	*95,9*	*No OMIM disease ID*
CAT	*100*	*100*	*100*	*100*	*Acatalasemia, 614097*
CATIP	*100*	*99,9*	*100*	*100*	*?Spermatogenic failure 54, 619379*
CATSPER1	*100*	*99,8*	*100*	*100*	*Spermatogenic failure 7, 612997*
CATSPER2	*100*	*98,6*	*100*	*100*	*No OMIM disease ID*
CAV1	*100*	*100*	*100*	*100*	*?Lipodystrophy, congenital generalized, type 3, 612526* *Pulmonary hypertension, primary, 3, 615343* *Lipodystrophy, familial partial, type 7, 606721*

CAV3	*100*	*100*	*100*	*100*	*Myopathy, distal, Tateyama type, 614321* *Creatine phosphokinase, elevated serum, 123320* *Cardiomyopathy, familial hypertrophic, 192600* *Rippling muscle disease 2, 606072* *Long QT syndrome 9, 611818*
CAVIN1	*100*	*100*	*100*	*100*	*Lipodystrophy, congenital generalized, type 4, 613327*
CBL	*97,3*	*96,9*	*100*	*100*	*Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563* *?Juvenile myelomonocytic leukemia, 607785*
CBLB	*99,8*	*99,1*	*100*	*100*	*No OMIM disease ID*
CBLIF	*100*	*99,6*	*100*	*100*	*Intrinsic factor deficiency, 261000*
CBS	*99,9*	*98,5*	*100*	*100*	*Thrombosis, hyperhomocysteinemic, 236200* *Homocystinuria, B6-responsive and nonresponsive types, 236200*
CBWD1	*20,5*	*19,1*	*99,4*	*98,5*	*No OMIM disease ID*
CBX2	*100*	*100*	*100*	*100*	*?46XY sex reversal 5, 613080*
CBY1	*82,2*	*82*	*100*	*100*	*No OMIM disease ID*
CC2D1A	*100*	*99,6*	*100*	*100*	*Mental retardation, autosomal recessive 3, 608443*
CC2D2A	*98,3*	*96,6*	*97,1*	*97*	*COACH syndrome 2, 619111* *Meckel syndrome 6, 612284* *Joubert syndrome 9, 612285*
CCBE1	*99,9*	*98,8*	*100*	*100*	*Hennekam lymphangiectasia-lymphedema syndrome 1, 235510*
CCDC103	*100*	*100*	*100*	*100*	*Ciliary dyskinesia, primary, 17, 614679*
CCDC115	*95,8*	*90,2*	*100*	*100*	*Congenital disorder of glycosylation, type IIo, 616828*
CCDC134	*100*	*100*	*100*	*100*	*No OMIM disease ID*
CCDC141	*99,8*	*99,5*	*100*	*99,9*	*No OMIM disease ID*
CCDC174	*99,5*	*96,7*	*100*	*100*	*Hypotonia, infantile, with psychomotor retardation, 616816*
CCDC186	*99*	*95,6*	*100*	*99,8*	*No OMIM disease ID*
CCDC22	*99*	*95,4*	*100*	*100*	*Ritscher-Schinzel syndrome 2, 300963*
CCDC28B	*99,9*	*98,2*	*100*	*100*	*No OMIM disease ID*
CCDC32	*99,9*	*98,5*	*100*	*100*	*Cardiofacioneurodevelopmental syndrome, 619123*
CCDC39	*99*	*96,2*	*100*	*99,9*	*Ciliary dyskinesia, primary, 14, 613807*
CCDC40	*99,1*	*98,2*	*100*	*100*	*Ciliary dyskinesia, primary, 15, 613808*
CCDC47	*99,9*	*97,4*	*100*	*100*	*Trichohepatoneurodevelopmental syndrome, 618268*

CCDC50	*100*	*99,7*	*100*	*100*	*?Deafness, autosomal dominant 44, 607453*
CCDC65	*97*	*92,5*	*100*	*100*	*Ciliary dyskinesia, primary, 27, 615504*
CCDC78	*100*	*100*	*100*	*100*	*?Centronuclear myopathy 4, 614807*
CCDC8	*100*	*100*	*100*	*100*	*3-M syndrome 3, 614205*
CCDC88A	*95,9*	*91,8*	*97,5*	*97,3*	*?PEHO syndrome-like, 617507*
CCDC88C	*99,9*	*99,3*	*100*	*100*	*?Spinocerebellar ataxia 40, 616053* *Hydrocephalus, congenital, 1, 236600*
CCL2	*100*	*100*	*100*	*100*	*No OMIM disease ID*
CCM2	*98,4*	*97,8*	*100*	*100*	*Cerebral cavernous malformations-2, 603284*
CCN6	*84,6*	*84,6*	*84,9*	*84,6*	*Progressive pseudorheumatoid dysplasia, 208230*
CCND2	*100*	*100*	*100*	*100*	*Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938*
CCNF	*99,5*	*97,6*	*100*	*100*	*Frontotemporal dementia and/or amyotrophic lateral sclerosis 5, 619141*
CCNK	*92,4*	*89*	*100*	*99,2*	*?Intellectual developmental disorder with hypertelorism and distinctive facies, 618147*
CCNO	*100*	*99*	*100*	*100*	*Ciliary dyskinesia, primary, 29, 615872*
CCNQ	*82,9*	*78,3*	*99,8*	*98,2*	*STAR syndrome, 300707*
CCT2	*100*	*99,6*	*100*	*100*	*No OMIM disease ID*
CCT5	*99,9*	*99,3*	*100*	*100*	*Neuropathy, hereditary sensory, with spastic paraplegia, 256840*
CD151	*100*	*100*	*100*	*100*	*Nephropathy with pretibial epidermolysis bullosa and deafness, 609057*
CD164	*98,1*	*91,8*	*100*	*100*	*?Deafness, autosomal dominant 66, 616969*
CD19	*100*	*100*	*100*	*100*	*Immunodeficiency, common variable, 3, 613493*
CD247	*100*	*99,4*	*100*	*100*	*?Immunodeficiency 25, 610163*
CD27	*99,9*	*98,3*	*100*	*100*	*Lymphoproliferative syndrome 2, 615122*
CD28	*100*	*99,9*	*100*	*100*	*No OMIM disease ID*
CD2AP	*99,6*	*98,8*	*100*	*99,9*	*Glomerulosclerosis, focal segmental, 3, 607832*
CD320	*100*	*100*	*100*	*99,9*	*Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646*
CD36	*99,6*	*99,2*	*100*	*100*	*Platelet glycoprotein IV deficiency, 608404*
CD3D	*100*	*99,9*	*100*	*100*	*Immunodeficiency 19, 615617*
CD3E	*100*	*98,8*	*100*	*100*	*Immunodeficiency 18, 615615* *Immunodeficiency 18, SCID variant, 615615*
CD3G	*100*	*100*	*100*	*100*	*Immunodeficiency 17, CD3 gamma deficient, 615607*

CD4	*100*	*99,7*	*100*	*100*	*Immunodeficiency 79, 619238* *OKT4 epitope deficiency, 613949*
CD40	*100*	*100*	*100*	*100*	*Immunodeficiency with hyper-IgM, type 3, 606843*
CD40LG	*92,4*	*81*	*100*	*100*	*Immunodeficiency, X-linked, with hyper-IgM, 308230*
CD46	*99,7*	*98,9*	*100*	*99,9*	*No OMIM disease ID*
CD55	*91,5*	*82,6*	*95*	*92,6*	*Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300*
CD59	*75,5*	*67*	*64,5*	*64,5*	*Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300*
CD70	*100*	*96,8*	*100*	*100*	*Lymphoproliferative syndrome 3, 618261*
CD79A	*100*	*100*	*100*	*100*	*Agammaglobulinemia 3, 613501*
CD79B	*100*	*100*	*100*	*100*	*Agammaglobulinemia 6, 612692*
CD81	*100*	*99,6*	*100*	*99,8*	*Immunodeficiency, common variable, 6, 613496*
CD8A	*100*	*100*	*100*	*100*	*CD8 deficiency, familial, 608957*
CD96	*99,9*	*99,5*	*100*	*100*	*C syndrome, 211750*
CDAN1	*100*	*99,9*	*100*	*100*	*Dyserythropoietic anemia, congenital, type Ia, 224120*
CDC14A	*99,9*	*99,2*	*100*	*100*	*Deafness, autosomal recessive 32, with or without immotile sperm, 608653*
CDC40	*99,6*	*98,9*	*100*	*99,8*	*?Pontocerebellar hypoplasia, type 15, 619302*
CDC42	*96,3*	*87,9*	*100*	*100*	*Takenouchi-Kosaki syndrome, 616737*
CDC42BPB	*99,9*	*98,5*	*100*	*100*	*No OMIM disease ID*
CDC45	*99,8*	*98,5*	*100*	*100*	*Meier-Gorlin syndrome 7, 617063*
CDC6	*100*	*99,9*	*100*	*100*	*?Meier-Gorlin syndrome 5, 613805*
CDC73	*99,8*	*98,3*	*100*	*100*	*Hyperparathyroidism, familial primary, 145000* *Parathyroid adenoma with cystic changes, 145001* *Parathyroid carcinoma, 608266* *Hyperparathyroidism-jaw tumor syndrome, 145001*
CDCA7	*100*	*99,6*	*100*	*100*	*Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910*
CDH1	*99,2*	*98,6*	*99,2*	*99,1*	*Ovarian cancer, somatic, 167000* *Blepharocheilodontic syndrome 1, 119580* *Endometrial carcinoma, somatic, 608089* *Gastric cancer, hereditary diffuse, with or without cleft lip and/or palate, 137215*
CDH11	*100*	*100*	*100*	*100*	*Elsahy-Waters syndrome, 211380*
CDH15	*100*	*99,2*	*100*	*100*	*Mental retardation, autosomal dominant 3, 612580*

CDH2	*99,4*	*97,1*	*100*	*100*	*Arrhythmogenic right ventricular dysplasia, familial, 14, 618920* *Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929*
CDH23	*100*	*100*	*100*	*100*	*Usher syndrome, type 1D, 601067* *Usher syndrome, type 1D/F digenic, 601067* *Deafness, autosomal recessive 12, 601386*
CDH3	*100*	*99,1*	*100*	*100*	*Hypotrichosis, congenital, with juvenile macular dystrophy, 601553* *Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280*
CDHR1	*99,5*	*98,1*	*100*	*100*	*Cone-rod dystrophy 15, 613660* *Retinitis pigmentosa 65, 613660*
C15orf41	*85,9*	*85,7*	*100*	*100*	*Dyserythropoietic anemia, congenital, type Ib, 615631*
CDK10	*100*	*99,5*	*100*	*100*	*Al Kaissi syndrome, 617694*
CDK13	*97,7*	*91,7*	*100*	*99,9*	*Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360*
CDK19	*99,8*	*99,3*	*100*	*100*	*Developmental and epileptic encephalopathy 87, 618916*
CDK4	*99,9*	*99*	*100*	*100*	*No OMIM disease ID*
CDK5	*100*	*99,5*	*100*	*100*	*?Lissencephaly 7 with cerebellar hypoplasia, 616342*
CDK5RAP2	*99,6*	*98,5*	*100*	*100*	*Microcephaly 3, primary, autosomal recessive, 604804*
CDK6	*99,9*	*99,1*	*100*	*100*	*?Microcephaly 12, primary, autosomal recessive, 616080*
CDK8	*99,6*	*96,7*	*100*	*100*	*Intellectual developmental disorder with hypotonia and behavioral abnormalities, 618748*
CDKL5	*91,4*	*89*	*92,2*	*91,1*	*Developmental and epileptic encephalopathy 2, 300672*
CDKN1A	*100*	*100*	*100*	*100*	*No OMIM disease ID*
CDKN1B	*99,9*	*99,3*	*100*	*100*	*Multiple endocrine neoplasia, type IV, 610755*
CDKN1C	*89,9*	*81,6*	*98,9*	*95,8*	*IMAGE syndrome, 614732* *Beckwith-Wiedemann syndrome, 130650*
CDKN2A	*92,3*	*92,3*	*100*	*100*	*No OMIM disease ID*
CDKN2B	*100*	*99,7*	*100*	*100*	*No OMIM disease ID*
CDKN2C	*100*	*100*	*100*	*100*	*No OMIM disease ID*
CDON	*99,9*	*98,6*	*100*	*99,9*	*Holoprosencephaly 11, 614226*
CDSN	*100*	*99,8*	*100*	*100*	*Hypotrichosis 2, 146520* *Peeling skin syndrome 1, 270300*
CDT1	*99,7*	*97,8*	*99,9*	*98*	*Meier-Gorlin syndrome 4, 613804*
CEACAM16	*100*	*99,2*	*100*	*100*	*Deafness, autosomal dominant 4B, 614614* *Deafness, autosomal recessive 113, 618410*

CEBPA	*95,9*	*80,1*	*99,7*	*97,3*	*Leukemia, acute myeloid, somatic, 601626* *?Leukemia, acute myeloid, 601626*
CEBPE	*100*	*100*	*100*	*100*	*Specific granule deficiency, 245480*
CEL	*90,8*	*89,2*	*100*	*99,9*	*Maturity-onset diabetes of the young, type VIII, 609812*
CELA2A	*98*	*94,6*	*100*	*100*	*Abdominal obesity-metabolic syndrome 4, 618620*
CELF2	*94,9*	*94,4*	*100*	*100*	*No OMIM disease ID*
CELSR1	*95,7*	*93,2*	*99,3*	*98,4*	*Lymphatic malformation 9, 619319*
CENPE	*96,7*	*90*	*100*	*99,7*	*?Microcephaly 13, primary, autosomal recessive, 616051*
CENPF	*99,4*	*96,9*	*100*	*100*	*Stromme syndrome, 243605*
CENPJ	*99,8*	*98,7*	*100*	*100*	*Microcephaly 6, primary, autosomal recessive, 608393* *?Seckel syndrome 4, 613676*
CENPS	*100*	*98*	*100*	*100*	*No OMIM disease ID*
CENPT	*100*	*100*	*100*	*100*	*?Short stature and microcephaly with genital anomalies, 618702*
CEP104	*99,9*	*98*	*100*	*100*	*Joubert syndrome 25, 616781*
CEP112	*99,1*	*96,6*	*100*	*100*	*Spermatogenic failure 44, 619044*
CEP120	*99,9*	*99,6*	*100*	*100*	*Short-rib thoracic dysplasia 13 with or without polydactyly, 616300* *Joubert syndrome 31, 617761*
CEP135	*98,3*	*90,1*	*100*	*99,9*	*Microcephaly 8, primary, autosomal recessive, 614673*
CEP152	*99,5*	*98*	*100*	*100*	*Microcephaly 9, primary, autosomal recessive, 614852* *Seckel syndrome 5, 613823*
CEP164	*99,8*	*98,2*	*100*	*100*	*Nephronophthisis 15, 614845*
CEP19	*100*	*100*	*100*	*100*	*Morbid obesity and spermatogenic failure, 615703*
CEP250	*99,9*	*98,4*	*100*	*100*	*Cone-rod dystrophy and hearing loss 2, 618358*
CEP290	*96,2*	*90,8*	*100*	*99,9*	*Leber congenital amaurosis 10, 611755* *Joubert syndrome 5, 610188* *Senior-Loken syndrome 6, 610189* *?Bardet-Biedl syndrome 14, 615991* *Meckel syndrome 4, 611134*
CEP41	*98,8*	*93,4*	*100*	*100*	*Joubert syndrome 15, 614464*
CEP55	*100*	*99,8*	*100*	*100*	*Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500*
CEP57	*97,6*	*89,3*	*100*	*100*	*Mosaic variegated aneuploidy syndrome 2, 614114*
CEP63	*98,6*	*94,9*	*100*	*100*	*?Seckel syndrome 6, 614728*

CEP78	*98,8*	*95,6*	*100*	*99,9*	*Cone-rod dystrophy and hearing loss, 617236*
CEP83	*99*	*96,6*	*100*	*99,9*	*Nephronophthisis 18, 615862*
CEP85L	*99*	*96,5*	*100*	*99,9*	*Lissencephaly 10, 618873*
CEP89	*95,8*	*94,5*	*100*	*100*	*No OMIM disease ID*
CERKL	*99,3*	*96,5*	*100*	*100*	*Retinitis pigmentosa 26, 608380*
CERS1	*79,1*	*65,2*	*93,2*	*86,5*	*?Epilepsy, progressive myoclonic, 8, 616230*
CERS3	*99,7*	*98*	*100*	*100*	*Ichthyosis, congenital, autosomal recessive 9, 615023*
CERT1	*89,8*	*86,5*	*100*	*100*	*Mental retardation, autosomal dominant 34, 616351*
CES1	*99,7*	*97,6*	*99,9*	*99,8*	*Drug metabolism, altered, CES1-related, 618057*
CETP	*100*	*99,5*	*100*	*100*	*Hyperalphalipoproteinemia, 143470*
WDR66	*100*	*99,9*	*100*	*100*	*Spermatogenic failure 33, 618152*
C1orf194	*100*	*98,4*	*100*	*100*	*No OMIM disease ID*
CFAP298	*99,6*	*96,7*	*100*	*100*	*Ciliary dyskinesia, primary, 26, 615500*
CFAP300	*99,2*	*98,3*	*100*	*99,9*	*Ciliary dyskinesia, primary, 38, 618063*
CFAP410	*100*	*99,6*	*100*	*100*	*Retinal dystrophy with macular staphyloma, 617547* *Spondylometaphyseal dysplasia, axial, 602271*
C8orf37	*99,7*	*99,6*	*100*	*100*	*Retinitis pigmentosa 64, 614500* *Cone-rod dystrophy 16, 614500* *Bardet-Biedl syndrome 21, 617406*
CFAP43	*99,5*	*98*	*100*	*100*	*Hydrocephalus, normal pressure, 1, 236690* *Spermatogenic failure 19, 617592*
CFAP44	*99,5*	*98,6*	*100*	*100*	*?Spermatogenic failure 20, 617593*
CFAP47	*75,8*	*71*	*99,8*	*97,8*	*Spermatogenic failure, X-linked, 3, 301059*
CFAP53	*99,3*	*96,6*	*100*	*100*	*Heterotaxy, visceral, 6, autosomal recessive, 614779*
CFAP58	*99,7*	*97,1*	*100*	*100*	*Spermatogenic failure 49, 619144*
CFAP65	*99,9*	*98,7*	*100*	*100*	*Spermatogenic failure 40, 618664*
CFAP69	*98,7*	*94,9*	*100*	*100*	*Spermatogenic failure 24, 617959*
CFAP70	*99,9*	*99,7*	*100*	*100*	*?Spermatogenic failure 41, 618670*
MAATS1	*99,8*	*98,8*	*100*	*100*	*Spermatogenic failure 51, 619177*
CFB	*100*	*99,6*	*100*	*100*	*?Complement factor B deficiency, 615561*

CFC1	*85*	*78*	*100*	*100*	*Heterotaxy, visceral, 2, autosomal, 605376*
CFD	*90,9*	*84,6*	*100*	*100*	*Complement factor D deficiency, 613912*
CFH	*99,8*	*98,5*	*100*	*99,9*	*Basal laminar drusen, 126700* *Complement factor H deficiency, 609814*
CFHR1	*91,7*	*89,6*	*96,3*	*94,1*	*No OMIM disease ID*
CFHR2	*75,7*	*74,4*	*76,1*	*76,1*	*No OMIM disease ID*
CFHR3	*89*	*87,8*	*97,7*	*96*	*No OMIM disease ID*
CFHR4	*99,8*	*99,5*	*100*	*99,7*	*No OMIM disease ID*
CFHR5	*99,8*	*97,6*	*100*	*100*	*Nephropathy due to CFHR5 deficiency, 614809*
CFI	*99,3*	*96*	*100*	*99,9*	*Complement factor I deficiency, 610984*
CFL2	*99,5*	*99*	*100*	*99,9*	*Nemaline myopathy 7, autosomal recessive, 610687*
CFP	*100*	*99*	*100*	*100*	*Properdin deficiency, X-linked, 312060*
CFTR	*99,5*	*97,9*	*100*	*100*	*Cystic fibrosis, 219700* *Congenital bilateral absence of vas deferens, 277180* *Sweat chloride elevation without CF,*
CHAMP1	*100*	*100*	*100*	*100*	*Mental retardation, autosomal dominant 40, 616579*
CHAT	*93,1*	*85,1*	*100*	*99,9*	*Myasthenic syndrome, congenital, 6, presynaptic, 254210*
CHCHD10	*57,8*	*42*	*100*	*100*	*?Myopathy, isolated mitochondrial, autosomal dominant, 616209* *Spinal muscular atrophy, Jokela type, 615048* *Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911*
CHCHD2	*93,7*	*78,7*	*100*	*100*	*Parkinson disease 22, autosomal dominant, 616710*
CHD1	*98,6*	*93*	*100*	*99,9*	*Pilarowski-Bjornsson syndrome, 617682*
CHD2	*99,3*	*98,6*	*100*	*100*	*Develomental and epileptic encephalopathy 94, 615369*
CHD3	*96,3*	*92*	*99,7*	*99,4*	*Snijders Blok-Campeau syndrome, 618205*
CHD4	*100*	*99,6*	*100*	*100*	*Sifrim-Hitz-Weiss syndrome, 617159*
CHD5	*99,6*	*96,1*	*100*	*99,9*	*No OMIM disease ID*
CHD7	*100*	*99,2*	*100*	*100*	*Hypogonadotropic hypogonadism 5 with or without anosmia, 612370* *CHARGE syndrome, 214800*
CHD8	*100*	*99,7*	*100*	*100*	*No OMIM disease ID*
CHEK2	*84,9*	*80,7*	*100*	*100*	*Osteosarcoma, somatic, 259500* *Li-Fraumeni syndrome 2, 609265*
CHIT1	*99,9*	*98,2*	*100*	*100*	*No OMIM disease ID*

CHKB	*100*	*99,6*	*100*	*100*	*Muscular dystrophy, congenital, megaconial type, 602541*
CHM	*98,2*	*91,6*	*98,6*	*96,4*	*Choroideremia, 303100*
CHMP1A	*100*	*99,6*	*100*	*100*	*Pontocerebellar hypoplasia, type 8, 614961*
CHMP2B	*99,6*	*98,5*	*100*	*100*	*Frontotemporal dementia and/or amyotrophic lateral sclerosis 7, 600795*
CHMP4B	*100*	*99,2*	*100*	*100*	*Cataract 31, multiple types, 605387*
CHN1	*99,8*	*99,2*	*97*	*96,9*	*Duane retraction syndrome 2, 604356*
CHP1	*97,1*	*85,7*	*100*	*100*	*?Spastic ataxia 9, autosomal recessive, 618438*
CHRDL1	*100*	*99,5*	*100*	*100*	*Megalocornea 1, X-linked, 309300*
CHRM2	*100*	*100*	*100*	*100*	*No OMIM disease ID*
CHRM3	*100*	*100*	*100*	*100*	*Prune belly syndrome, 100100*
CHRNA1	*100*	*99,6*	*100*	*100*	*Myasthenic syndrome, congenital, 1B, fast-channel, 608930* *Myasthenic syndrome, congenital, 1A, slow-channel, 601462* *Multiple pterygium syndrome, lethal type, 253290*
CHRNA2	*100*	*100*	*100*	*100*	*Epilepsy, nocturnal frontal lobe, type 4, 610353*
CHRNA3	*100*	*99,2*	*100*	*100*	*Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT, 191800*
CHRNA4	*99*	*96,5*	*100*	*100*	*Epilepsy, nocturnal frontal lobe, 1, 600513*
CHRNB1	*100*	*99,5*	*100*	*100*	*?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314* *Myasthenic syndrome, congenital, 2A, slow-channel, 616313*
CHRNB2	*99,3*	*95,5*	*100*	*100*	*Epilepsy, nocturnal frontal lobe, 3, 605375*
CHRND	*99,4*	*97,4*	*100*	*100*	*?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323* *Multiple pterygium syndrome, lethal type, 253290* *Myasthenic syndrome, congenital, 3B, fast-channel, 616322* *?Myasthenic syndrome, congenital, 3A, slow-channel, 616321*
CHRNE	*100*	*100*	*100*	*100*	*Myasthenic syndrome, congenital, 4A, slow-channel, 605809* *Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931* *Myasthenic syndrome, congenital, 4B, fast-channel, 616324*
CHRNG	*100*	*100*	*100*	*100*	*Multiple pterygium syndrome, lethal type, 253290* *Escobar syndrome, 265000*
CHST11	*100*	*100*	*100*	*100*	*?Osteochondrodysplasia, brachydactyly, and overlapping malformed digits, 618167*
CHST14	*99,9*	*98,8*	*100*	*100*	*Ehlers-Danlos syndrome, musculocontractural type 1, 601776*
CHST3	*100*	*99,9*	*100*	*100*	*Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095*
CHST6	*100*	*100*	*100*	*100*	*Macular corneal dystrophy, 217800*

CHST8	*100*	*100*	*100*	*100*	*?Peeling skin syndrome 3, 616265*
CHSY1	*97,8*	*96,3*	*99,3*	*96,9*	*Temtamy preaxial brachydactyly syndrome, 605282*
CHUK	*99,7*	*98,1*	*100*	*100*	*?Popliteal pterygium syndrome, Bartsocas-Papas type 2, 619339* *Cocoon syndrome, 613630*
CIB1	*98,1*	*94,5*	*100*	*100*	*Epidermodysplasia verruciformis 3, 618267*
CIB2	*99,3*	*96,2*	*100*	*99,9*	*Deafness, autosomal recessive 48, 609439* *Usher syndrome, type IJ, 614869*
FAM92A	*89,9*	*81,7*	*100*	*100*	*?Polydactyly, postaxial, type A9, 618219*
CIC	*63,4*	*63,3*	*100*	*100*	*Mental retardation, autosomal dominant 45, 617600*
CIDEC	*99,9*	*98,3*	*100*	*100*	*?Lipodystrophy, familial partial, type 5, 615238*
CIITA	*100*	*99,4*	*100*	*100*	*Bare lymphocyte syndrome, type II, complementation group A, 209920*
CILK1	*99,6*	*98*	*100*	*99,8*	*Endocrine-cerebroosteodysplasia, 612651*
CISD2	*83,4*	*83,4*	*100*	*100*	*Wolfram syndrome 2, 604928*
CIT	*99,8*	*98,2*	*100*	*100*	*Microcephaly 17, primary, autosomal recessive, 617090*
CITED2	*99,2*	*99,1*	*100*	*100*	*Atrial septal defect 8, 614433* *Ventricular septal defect 2, 614431*
CKAP2L	*99,5*	*98,3*	*100*	*100*	*Filippi syndrome, 272440*
CLCC1	*99,5*	*95,6*	*100*	*100*	*Retinitis pigmentosa 32, 609913*
CLCF1	*100*	*99,5*	*100*	*100*	*Cold-induced sweating syndrome 2, 610313*
CLCN1	*99,9*	*98,8*	*100*	*100*	*Myotonia congenita, recessive, 255700* *Myotonia congenita, dominant, 160800* *Myotonia levior, recessive,*
CLCN2	*100*	*99,3*	*100*	*100*	*Leukoencephalopathy with ataxia, 615651* *Hyperaldosteronism, familial, type II, 605635*
CLCN3	*98*	*94,2*	*96,7*	*96,7*	*Neurodevelopmental disorder with seizures and brain abnormalities, 619517* *Neurodevelopmental disorder with hypotonia and brain abnormalities, 619512*
CLCN4	*99,9*	*97,7*	*100*	*100*	*Raynaud-Claes syndrome, 300114*
CLCN5	*99,7*	*97,1*	*100*	*99,9*	*Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990* *Hypophosphatemic rickets, 300554* *Dent disease 1, 300009* *Nephrolithiasis, type I, 310468*
CLCN6	*99,9*	*98,9*	*100*	*100*	*Neurodegeneration, childhood-onset, hypotonia, respiratory insufficiency and brain imaging abnormalities, 619173*

CLCN7	*99,4*	*97,8*	*100*	*100*	*Hypopigmentation, organomegaly, and delayed myelination and development, 618541* *Osteopetrosis, autosomal recessive 4, 611490* *Osteopetrosis, autosomal dominant 2, 166600*
CLCNKA	*99,6*	*97,1*	*100*	*100*	*Bartter syndrome, type 4b, digenic, 613090*
CLCNKB	*98,7*	*95,3*	*100*	*100*	*Bartter syndrome, type 3, 607364* *Bartter syndrome, type 4b, digenic, 613090*
CLDN1	*100*	*100*	*100*	*100*	*Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626*
CLDN10	*100*	*100*	*100*	*100*	*HELIX syndrome, 617671*
CLDN11	*100*	*99,9*	*100*	*100*	*Leukodystrophy, hypomyelinating, 22, 619328*
CLDN14	*100*	*99,5*	*100*	*100*	*Deafness, autosomal recessive 29, 614035*
CLDN16	*100*	*100*	*100*	*100*	*Hypomagnesemia 3, renal, 248250*
CLDN19	*98,3*	*92,9*	*100*	*100*	*Hypomagnesemia 5, renal, with ocular involvement, 248190*
CLDN2	*100*	*99,7*	*100*	*100*	*?Azoospermia, obstructive, with nephrolithiasis, 301060*
CLDN9	*100*	*100*	*100*	*100*	*?Deafness, autosomal recessive 116, 619093*
CLEC4D	*100*	*99,9*	*100*	*100*	*No OMIM disease ID*
CLEC7A	*100*	*99,7*	*100*	*100*	*Candidiasis, familial, 4, autosomal recessive, 613108*
CLIC2	*99,3*	*94,9*	*100*	*100*	*?Intellectual developmental disorder, X-linked syndromic 32, 300886*
CLIC5	*89,5*	*87,3*	*100*	*100*	*?Deafness, autosomal recessive 103, 616042*
CLIP1	*99,8*	*98,7*	*100*	*100*	*No OMIM disease ID*
CLMP	*100*	*99,5*	*100*	*100*	*Congenital short bowel syndrome, 615237*
CLN3	*92,5*	*92,4*	*92,5*	*92,5*	*Ceroid lipofuscinosis, neuronal, 3, 204200*
CLN5	*69*	*66,3*	*71,8*	*71,6*	*Ceroid lipofuscinosis, neuronal, 5, 256731*
CLN6	*99,9*	*98,9*	*100*	*100*	*Ceroid lipofuscinosis, neuronal, 6, 601780* *Ceroid lipofuscinosis, neuronal, 4A (Kufs type), autosomal recessive, 204300*
CLN8	*83,5*	*83,5*	*100*	*100*	*Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003* *Ceroid lipofuscinosis, neuronal, 8, 600143*
CLP1	*100*	*100*	*100*	*100*	*Pontocerebellar hypoplasia, type 10, 615803*
CLPB	*94,9*	*94*	*100*	*100*	*3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271*
CLPP	*100*	*99,5*	*100*	*100*	*Perrault syndrome 3, 614129*
CLPX	*99,8*	*99,5*	*100*	*99,9*	*?Protoporphyria, erythropoietic, 2, 618015*

CLRN1	*100*	*99,7*	*100*	*100*	*Usher syndrome, type 3A, 276902* *Retinitis pigmentosa 61, 614180*
CLRN2	*99,9*	*98,2*	*100*	*100*	*?Deafness, autosomal recessive 117, 619174*
CLTC	*99,9*	*99,7*	*100*	*100*	*Mental retardation, autosomal dominant 56, 617854*
CLTCL1	*98,6*	*97,5*	*100*	*100*	*No OMIM disease ID*
CLUAP1	*99,9*	*99,7*	*100*	*100*	*No OMIM disease ID*
CMAS	*99,3*	*96,7*	*100*	*100*	*No OMIM disease ID*
CNBP	*100*	*100*	*100*	*100*	*Myotonic dystrophy 2, 602668*
CNGA1	*91*	*85,6*	*91*	*90,9*	*Retinitis pigmentosa 49, 613756*
CNGA3	*100*	*99,8*	*100*	*100*	*Achromatopsia 2, 216900*
CNGB1	*99*	*97,1*	*100*	*100*	*Retinitis pigmentosa 45, 613767*
CNGB3	*97,8*	*90,6*	*100*	*100*	*Achromatopsia 3, 262300*
CNKSR2	*95,1*	*89*	*100*	*99,8*	*Intellectual developmental disorder, X-linked, syndromic, Houge type, 301008*
CNNM2	*100*	*99,9*	*100*	*100*	*Hypomagnesemia 6, renal, 613882* *Hypomagnesemia, seizures, and mental retardation, 616418*
CNNM4	*99,9*	*99*	*99,7*	*99*	*Jalili syndrome, 217080*
CNOT1	*100*	*99,8*	*100*	*100*	*Vissers-Bodmer syndrome, 619033* *Holoprosencephaly 12, with or without pancreatic agenesis, 618500*
CNOT2	*99,9*	*99,6*	*100*	*99,9*	*Intellectual developmental disorder with nasal speech, dysmorphic facies, and variable skeletal anomalies, 618608*
CNOT3	*100*	*100*	*100*	*100*	*Intellectual developmental disorder with speech delay, autism, and dysmorphic facies, 618672*
CNP	*100*	*100*	*100*	*100*	*?Leukodystrophy, hypomyelinating, 20, 619071*
CNPY3	*100*	*99,7*	*100*	*100*	*Developmental and epileptic encephalopathy 60, 617929*
CNTN1	*99,7*	*98,6*	*100*	*100*	*?Myopathy, congenital, Compton-North, 612540*
CNTN2	*92,7*	*92,7*	*100*	*100*	*?Epilepsy, myoclonic, familial adult, 5, 615400*
CNTNAP1	*100*	*99,8*	*100*	*100*	*Lethal congenital contracture syndrome 7, 616286* *Hypomyelinating neuropathy, congenital, 3, 618186*
CNTNAP2	*100*	*99,5*	*100*	*100*	*Pitt-Hopkins like syndrome 1, 610042* *Cortical dysplasia-focal epilepsy syndrome, 610042*
COA1	*100*	*100*	*100*	*100*	*No OMIM disease ID*
COA3	*100*	*100*	*100*	*100*	*?Mitochondrial complex IV deficiency, nuclear type 14, 619058*
COA5	*94,4*	*83*	*85,2*	*85,2*	*?Mitochondrial complex IV, deficiency, nuclear type 9, 616500*

COA6	*99,6*	*96,3*	*100*	*100*	*Mitochondrial complex IV deficiency, nuclear type 13, 616501*
COA7	*100*	*100*	*100*	*100*	*Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387*
COA8	*81,9*	*80,8*	*93,7*	*93,4*	*Mitochondrial complex IV deficiency, nuclear type 17, 619061*
COASY	*100*	*100*	*100*	*100*	*Pontocerebellar hypoplasia, type 12, 618266* *Neurodegeneration with brain iron accumulation 6, 615643*
COCH	*95*	*93,5*	*100*	*99,9*	*Deafness, autosomal dominant 9, 601369* *?Deafness, autosomal recessive 110, 618094*
COG1	*100*	*99,9*	*100*	*100*	*Congenital disorder of glycosylation, type IIg, 611209*
COG2	*99,8*	*98,9*	*100*	*100*	*?Congenital disorder of glycosylation, type IIq, 617395*
COG4	*100*	*100*	*100*	*100*	*Congenital disorder of glycosylation, type IIj, 613489* *Saul-Wilson syndrome, 618150*
COG5	*99,1*	*96,8*	*100*	*100*	*Congenital disorder of glycosylation, type IIi, 613612*
COG6	*98,5*	*93,1*	*100*	*100*	*Shaheen syndrome, 615328* *Congenital disorder of glycosylation, type III, 614576*
COG7	*100*	*99,4*	*100*	*100*	*Congenital disorder of glycosylation, type IIe, 608779*
COG8	*98,6*	*95,3*	*100*	*100*	*Congenital disorder of glycosylation, type IIh, 611182*
COL10A1	*100*	*99,6*	*100*	*100*	*Metaphyseal chondrodysplasia, Schmid type, 156500*
COL11A1	*96*	*92,7*	*100*	*99,9*	*Fibrochondrogenesis 1, 228520* *Stickler syndrome, type II, 604841* *Marshall syndrome, 154780* *Deafness, autosomal dominant 37, 618533*
COL11A2	*100*	*99,6*	*100*	*100*	*Deafness, autosomal dominant 13, 601868* *Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150* *Fibrochondrogenesis 2, 614524* *Deafness, autosomal recessive 53, 609706* *Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840*
COL12A1	*99,9*	*99,3*	*100*	*100*	*Bethlem myopathy 2, 616471* *?Ullrich congenital muscular dystrophy 2, 616470*
COL13A1	*93,9*	*93,5*	*100*	*100*	*Myasthenic syndrome, congenital, 19, 616720*
COL14A1	*99,9*	*99,3*	*100*	*100*	*No OMIM disease ID*
COL17A1	*99*	*96,3*	*100*	*100*	*Epithelial recurrent erosion dystrophy, 122400* *Epidermolysis bullosa, junctional, localisata variant, 226650* *Epidermolysis bullosa, junctional, non-Herlitz type, 226650*

COL18A1	*98,2*	*95,7*	*100*	*100*	*Knobloch syndrome, type 1, 267750* *Glaucoma, primary closed-angle, 618880*
COL1A1	*99,8*	*98,2*	*100*	*100*	*Osteogenesis imperfecta, type II, 166210* *Caffey disease, 114000* *Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060* *Osteogenesis imperfecta, type I, 166200* *Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1, 619115* *Osteogenesis imperfecta, type IV, 166220* *Osteogenesis imperfecta, type III, 259420*
COL1A2	*98,7*	*95,7*	*100*	*100*	*Osteogenesis imperfecta, type III, 259420* *Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821* *Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2, 619120* *Ehlers-Danlos syndrome, cardiac valvular type, 225320* *Osteogenesis imperfecta, type IV, 166220* *Osteogenesis imperfecta, type II, 166210*
COL25A1	*95,6*	*95,3*	*99,9*	*99,9*	*Fibrosis of extraocular muscles, congenital, 5, 616219*
COL27A1	*99,8*	*99,4*	*100*	*100*	*Steel syndrome, 615155*
COL2A1	*100*	*99,8*	*100*	*100*	*?Vitreoretinopathy with phalangeal epiphyseal dysplasia, 619248* *Czech dysplasia, 609162* *Achondrogenesis, type II or hypochondrogenesis, 200610* *Spondyloperipheral dysplasia, 271700* *SMED Strudwick type, 184250* *Stickler sydrome, type I, nonsyndromic ocular, 609508* *?Epiphyseal dysplasia, multiple, with myopia and deafness, 132450* *SED congenita, 183900* *Kniest dysplasia, 156550* *Osteoarthritis with mild chondrodysplasia, 604864* *Stickler syndrome, type I, 108300* *Platyspondylic skeletal dysplasia, Torrance type, 151210* *Spondyloepiphyseal dysplasia, Stanescu type, 616583* *Avascular necrosis of the femoral head, 608805* *Legg-Calve-Perthes disease, 150600*
COL3A1	*99,6*	*96,2*	*100*	*100*	*Ehlers-Danlos syndrome, vascular type, 130050* *Polymicrogyria with or without vascular-type EDS, 618343*
COL4A1	*99*	*97*	*100*	*100*	*?Retinal arteries, tortuosity of, 180000* *Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773*

					Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564 *Brain small vessel disease with or without ocular anomalies, 175780*
COL4A2	*100*	*99,6*	*100*	*100*	*Brain small vessel disease 2, 614483*
COL4A3	*98,9*	*97,4*	*100*	*100*	*Hematuria, benign familial, 141200* *Alport syndrome 3, autosomal dominant, 104200* *Alport syndrome 2, autosomal recessive, 203780*
COL4A4	*99,6*	*97,4*	*100*	*100*	*Hematuria, familial benign, 141200* *Alport syndrome 2, autosomal recessive, 203780*
COL4A5	*97,6*	*86,6*	*100*	*99,8*	*Alport syndrome 1, X-linked, 301050*
COL4A6	*96,9*	*90,8*	*100*	*99,8*	*?Deafness, X-linked 6, 300914*
COL5A1	*98,8*	*97,7*	*100*	*99,8*	*Ehlers-Danlos syndrome, classic type, 1, 130000* *Fibromuscular dysplasia, multifocal, 619329*
COL5A2	*100*	*98,4*	*100*	*100*	*Ehlers-Danlos syndrome, classic type, 2, 130010*
COL6A1	*100*	*99,7*	*100*	*100*	*Bethlem myopathy 1, 158810* *Ullrich congenital muscular dystrophy 1, 254090*
COL6A2	*100*	*99,8*	*100*	*100*	*Bethlem myopathy 1, 158810* *?Myosclerosis, congenital, 255600* *Ullrich congenital muscular dystrophy 1, 254090*
COL6A3	*100*	*99,7*	*100*	*100*	*Ullrich congenital muscular dystrophy 1, 254090* *Dystonia 27, 616411* *Bethlem myopathy 1, 158810*
COL6A5	*99,9*	*99,2*	*100*	*100*	*No OMIM disease ID*
COL7A1	*99,6*	*98,5*	*100*	*100*	*Epidermolysis bullosa, pretibial, 131850* *Transient bullous of the newborn, 131705* *EBD, Bart type, 132000* *Epidermolysis bullosa dystrophica, AD, 131750* *Epidermolysis bullosa pruriginosa, 604129* *EBD inversa, 226600* *Epidermolysis bullosa dystrophica, AR, 226600* *Toenail dystrophy, isolated, 607523* *EBD, localisata variant,*
COL8A2	*100*	*99,8*	*100*	*100*	*Corneal dystrophy, posterior polymorphous 2, 609140* *Corneal dystrophy, Fuchs endothelial, 1, 136800*
COL9A1	*99,9*	*98,6*	*100*	*100*	*Stickler syndrome, type IV, 614134* *?Epiphyseal dysplasia, multiple, 6, 614135*

COL9A2	*99,9*	*98,9*	*100*	*100*	*Epiphyseal dysplasia, multiple, 2, 600204* *?Stickler syndrome, type V, 614284*
COL9A3	*98,9*	*95,5*	*99,9*	*99,3*	*Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969*
COLEC10	*100*	*99,9*	*100*	*100*	*3MC syndrome 3, 248340*
COLEC11	*100*	*100*	*100*	*100*	*3MC syndrome 2, 265050*
COLGALT1	*94,3*	*90,3*	*99,1*	*97,9*	*Brain small vessel disease 3, 618360*
COLQ	*99,8*	*97,1*	*100*	*100*	*Myasthenic syndrome, congenital, 5, 603034*
COMP	*93,8*	*92,4*	*100*	*100*	*Pseudoachondroplasia, 177170* *Carpal tunnel syndrome 2, 619161* *Epiphyseal dysplasia, multiple, 1, 132400*
COMT	*100*	*100*	*100*	*100*	*No OMIM disease ID*
COPA	*99,9*	*99,2*	*100*	*100*	*No OMIM disease ID*
COPB1	*98,4*	*94,5*	*100*	*99,9*	*Baralle-Macken syndrome, 619255*
COPB2	*99,6*	*98,2*	*100*	*100*	*?Microcephaly 19, primary, autosomal recessive, 617800*
COQ2	*97,6*	*96,7*	*97,2*	*97,2*	*Coenzyme Q10 deficiency, primary, 1, 607426*
COQ4	*91*	*89,7*	*100*	*100*	*Coenzyme Q10 deficiency, primary, 7, 616276*
COQ5	*100*	*100*	*100*	*100*	*?Coenzyme Q10 deficiency, primary, 9, 619028*
COQ6	*99,9*	*98,5*	*100*	*100*	*Coenzyme Q10 deficiency, primary, 6, 614650*
COQ7	*100*	*99,6*	*100*	*100*	*?Coenzyme Q10 deficiency, primary, 8, 616733*
COQ8A	*100*	*99,6*	*100*	*100*	*Coenzyme Q10 deficiency, primary, 4, 612016*
COQ8B	*100*	*99,2*	*100*	*100*	*Nephrotic syndrome, type 9, 615573*
COQ9	*100*	*98,7*	*100*	*100*	*Coenzyme Q10 deficiency, primary, 5, 614654*
CORIN	*99,9*	*99,6*	*100*	*100*	*Preeclampsia/eclampsia 5, 614595*
CORO1A	*99,9*	*98,9*	*100*	*99,9*	*Immunodeficiency 8, 615401*
COX10	*100*	*99,9*	*100*	*100*	*Mitochondrial complex IV deficiency, nuclear type 3, 619046*
COX14	*100*	*100*	*100*	*100*	*?Mitochondrial complex IV deficiency, nuclear type 10, 619053*
COX15	*99,9*	*97,8*	*100*	*100*	*Mitochondrial complex IV deficiency, nuclear type 6, 615119*
COX16	*99,3*	*96,9*	*100*	*99,9*	*Mitochondrial complex IV deficiency, nuclear type 22, 619355*
COX20	*95,7*	*82,4*	*100*	*99,9*	*Mitochondrial complex IV deficiency, nuclear type 11, 619054*
COX4I1	*100*	*100*	*100*	*100*	*Mitochondrial complex IV deficiency, nuclear type 16, 619060*

COX4I2	*100*	*99,9*	*100*	*100*	*Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714*
COX5A	*66*	*36*	*100*	*100*	*?Mitochondrial complex IV deficiency, nuclear type 20, 619064*
COX5B	*100*	*100*	*100*	*100*	*No OMIM disease ID*
COX6A1	*100*	*99,9*	*100*	*100*	*Charcot-Marie-Tooth disease, recessive intermediate D, 616039*
COX6A2	*99,8*	*94*	*100*	*100*	*Mitochondrial complex IV deficiency, nuclear type 18, 619062*
COX6B1	*100*	*100*	*100*	*100*	*Mitochondrial complex IV deficiency, nuclear type 7, 619051*
COX6B2	*100*	*99,9*	*100*	*100*	*No OMIM disease ID*
COX6C	*99,5*	*94,3*	*100*	*99,9*	*No OMIM disease ID*
COX7A1	*100*	*99,9*	*100*	*100*	*No OMIM disease ID*
COX7A2	*100*	*98,8*	*100*	*99,9*	*No OMIM disease ID*
COX7B	*69,1*	*35,9*	*100*	*100*	*Linear skin defects with multiple congenital anomalies 2, 300887*
COX7B2	*100*	*100*	*100*	*100*	*No OMIM disease ID*
COX7C	*98,4*	*78,9*	*100*	*100*	*No OMIM disease ID*
COX8A	*100*	*100*	*100*	*100*	*?Mitochondrial complex IV deficiency, nuclear type 15, 619059*
COX8C	*100*	*99,9*	*100*	*100*	*No OMIM disease ID*
CP	*92,6*	*85,2*	*100*	*99,9*	*Cerebellar ataxia, 604290* *Hemosiderosis, systemic, due to aceruloplasminemia, 604290*
CPA6	*99,2*	*96,1*	*100*	*100*	*Febrile seizures, familial, 11, 614418* *Epilepsy, familial temporal lobe, 5, 614417*
CPAMD8	*95,9*	*92,6*	*99,9*	*99,6*	*Anterior segment dysgenesis 8, 617319*
CPE	*99,8*	*98,8*	*100*	*100*	*Intellectual developmental disorder and hypogonadotropic hypogonadism, 619326*
CPLANE1	*99,4*	*98,2*	*100*	*100*	*Orofaciodigital syndrome VI, 277170* *Joubert syndrome 17, 614615*
CPLX1	*100*	*100*	*100*	*100*	*Developmental and epileptic encephalopathy 63, 617976*
CPN1	*99,9*	*98,7*	*100*	*100*	*Carboxypeptidase N deficiency, 212070*
CPOX	*99,8*	*97,2*	*100*	*100*	*Coproporphyria, 121300* *Harderoporphyria, 618892*
CPS1	*100*	*100*	*100*	*100*	*Carbamoylphosphate synthetase I deficiency, 237300*
CPSF1	*98,5*	*97,1*	*100*	*100*	*Myopia 27, 618827*
CPT1A	*99,8*	*97,6*	*100*	*100*	*CPT deficiency, hepatic, type IA, 255120*
CPT1C	*99,8*	*99,2*	*100*	*100*	*?Spastic paraplegia 73, autosomal dominant, 616282*

CPT2	*98,2*	*97,4*	*100*	*100*	*CPT II deficiency, infantile, 600649* *CPT II deficiency, lethal neonatal, 608836* *CPT II deficiency, myopathic, stress-induced, 255110*
CR2	*100*	*99,9*	*100*	*100*	*Immunodeficiency, common variable, 7, 614699*
CRADD	*99,9*	*97,5*	*100*	*100*	*Mental retardation, autosomal recessive 34, with variant lissencephaly, 614499*
CRAT	*100*	*99,9*	*100*	*100*	*?Neurodegeneration with brain iron accumulation 8, 617917*
CRB1	*100*	*99,9*	*100*	*100*	*Leber congenital amaurosis 8, 613835* *Retinitis pigmentosa-12, 600105* *Pigmented paravenous chorioretinal atrophy, 172870*
CRB2	*98,9*	*94,2*	*100*	*100*	*Focal segmental glomerulosclerosis 9, 616220* *Ventriculomegaly with cystic kidney disease, 219730*
CRBN	*87,9*	*87,8*	*96,3*	*91,8*	*Mental retardation, autosomal recessive 2, 607417*
CREB1	*99,9*	*96,3*	*100*	*100*	*Histiocytoma, angiomatoid fibrous, somatic, 612160*
CREB3L1	*100*	*99,9*	*100*	*100*	*Osteogenesis imperfecta, type XVI, 616229*
CREB3L3	*100*	*100*	*100*	*100*	*Hypertriglyceridemia 2, 619324*
CREBBP	*99,6*	*97,8*	*100*	*100*	*Menke-Hennekam syndrome 1, 618332* *Rubinstein-Taybi syndrome 1, 180849*
CRELD1	*99,5*	*94*	*100*	*100*	*Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217*
CRIPT	*98,9*	*93,5*	*100*	*100*	*Short stature with microcephaly and distinctive facies, 615789*
CRLF1	*91,1*	*90,3*	*97,9*	*95,7*	*Cold-induced sweating syndrome 1, 272430*
CRPPA	*98,4*	*94,7*	*100*	*99,8*	*Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052* *Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643*
CRTAP	*100*	*99,4*	*100*	*100*	*Osteogenesis imperfecta, type VII, 610682*
CRTC1	*99,8*	*99,8*	*100*	*100*	*Mucoepidermoid salivary gland carcinoma,*
CRX	*100*	*100*	*100*	*100*	*Leber congenital amaurosis 7, 613829* *Cone-rod retinal dystrophy-2, 120970*
CRYAA	*99,9*	*98*	*100*	*100*	*Cataract 9, multiple types, 604219*
CRYAB	*100*	*98,2*	*100*	*100*	*Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869* *Myopathy, myofibrillar, 2, 608810* *Cataract 16, multiple types, 613763* *Cardiomyopathy, dilated, 1II, 615184*
CRYBA1	*100*	*99,6*	*100*	*100*	*Cataract 10, multiple types, 600881*
CRYBA2	*100*	*100*	*100*	*100*	*?Cataract 42, 115900*

CRYBA4	*100*	*100*	*100*	*100*	*Cataract 23, 610425*
CRYBB1	*100*	*99,9*	*100*	*100*	*Cataract 17, multiple types, 611544*
CRYBB2	*100*	*99,9*	*100*	*100*	*Cataract 3, multiple types, 601547*
CRYBB3	*100*	*99,9*	*100*	*100*	*Cataract 22, 609741*
CRYGB	*100*	*98,7*	*100*	*100*	*Cataract 39, multiple types, autosomal dominant, 615188*
CRYGC	*99*	*95,2*	*100*	*100*	*Cataract 2, multiple types, 604307*
CRYGD	*100*	*98,9*	*100*	*100*	*Cataract 4, multiple types, 115700*
CRYGS	*92,8*	*82,1*	*100*	*100*	*Cataract 20, multiple types, 116100*
CRYL1	*100*	*99,9*	*100*	*100*	*No OMIM disease ID*
CRYM	*100*	*97,9*	*100*	*100*	*Deafness, autosomal dominant 40, 616357*
CSDE1	*100*	*99,7*	*100*	*100*	*No OMIM disease ID*
CSF1R	*100*	*99,6*	*100*	*100*	*Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476* *Leukoencephalopathy, diffuse hereditary, with spheroids, 221820*
CSF2RA	*89,6*	*84,4*	*94,6*	*90,9*	*Surfactant metabolism dysfunction, pulmonary, 4, 300770*
CSF2RB	*99,9*	*99*	*100*	*100*	*Surfactant metabolism dysfunction, pulmonary, 5, 614370*
CSF3R	*99,8*	*98,5*	*100*	*100*	*Neutropenia, severe congenital, 7, autosomal recessive, 617014*
CSGALNACT1	*100*	*100*	*100*	*100*	*Skeletal dysplasia, mild, with joint laxity and advanced bone age, 618870*
CSNK1D	*97,4*	*94,2*	*100*	*100*	*Advanced sleep-phase syndrome, familial, 2, 615224*
CSNK1G1	*98,4*	*97,6*	*100*	*100*	*No OMIM disease ID*
CSNK2A1	*81,1*	*76,6*	*94*	*94*	*Okur-Chung neurodevelopmental syndrome, 617062*
CSNK2B	*100*	*100*	*100*	*100*	*Poirier-Bienvenu neurodevelopmental syndrome, 618732*
CSPP1	*99,7*	*98,1*	*100*	*100*	*Joubert syndrome 21, 615636*
CSRP3	*98,9*	*93,9*	*100*	*100*	*?Cardiomyopathy, dilated, 1M, 607482* *Cardiomyopathy, hypertrophic, 12, 612124*
CST3	*96,2*	*70,8*	*100*	*100*	*Cerebral amyloid angiopathy, 105150*
CST6	*99,1*	*94,4*	*100*	*100*	*?Ectodermal dysplasia 15, hypohidrotic/hair type, 618535*
CSTA	*99,7*	*99,3*	*100*	*100*	*Peeling skin syndrome 4, 607936*
CSTB	*99,6*	*90,5*	*100*	*100*	*Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800*
CTBP1	*94,3*	*86,9*	*99,4*	*98,4*	*Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915*
CTC1	*100*	*99,1*	*100*	*100*	*Cerebroretinal microangiopathy with calcifications and cysts, 612199*

CTCF	*99,7*	*98,4*	*100*	*100*	*Mental retardation, autosomal dominant 21, 615502*
CTDP1	*88,7*	*85*	*100*	*99,8*	*Congenital cataracts, facial dysmorphism, and neuropathy, 604168*
CTH	*100*	*99,9*	*100*	*100*	*Cystathioninuria, 219500*
CTHRC1	*94,2*	*88*	*100*	*100*	*Barrett esophagus/esophageal adenocarcinoma, 614266*
CTLA4	*100*	*100*	*100*	*100*	*Autoimmune lymphoproliferative syndrome, type V, 616100*
CTNNA1	*98,8*	*97,2*	*100*	*100*	*Macular dystrophy, patterned, 2, 608970*
CTNNA2	*99,9*	*99,7*	*100*	*100*	*Cortical dysplasia, complex, with other brain malformations 9, 618174*
CTNNA3	*99,9*	*99,8*	*100*	*100*	*Arrhythmogenic right ventricular dysplasia, familial, 13, 615616*
CTNNB1	*100*	*100*	*100*	*100*	*Exudative vitreoretinopathy 7, 617572* *Pilomatricoma, somatic, 132600* *Colorectal cancer, somatic, 114500* *Neurodevelopmental disorder with spastic diplegia and visual defects, 615075* *Medulloblastoma, somatic, 155255* *Ovarian cancer, somatic, 167000* *Hepatocellular carcinoma, somatic, 114550*
CTNNBL1	*99,8*	*98,7*	*100*	*100*	*No OMIM disease ID*
CTNND1	*100*	*99,8*	*100*	*100*	*Blepharocheilodontic syndrome 2, 617681*
CTNND2	*93,7*	*89,7*	*97,3*	*95,2*	*No OMIM disease ID*
CTNS	*100*	*99,3*	*100*	*100*	*Cystinosis, nephropathic, 219800* *Cystinosis, ocular nonnephropathic, 219750* *Cystinosis, late-onset juvenile or adolescent nephropathic, 219900* *Cystinosis, atypical nephropathic, 219800*
CTPS1	*93*	*93*	*93*	*93*	*Immunodeficiency 24, 615897*
CTR9	*99,9*	*99,8*	*100*	*100*	*No OMIM disease ID*
CTSA	*100*	*99,6*	*100*	*100*	*Galactosialidosis, 256540*
CTSB	*100*	*100*	*100*	*100*	*No OMIM disease ID*
CTSC	*100*	*100*	*100*	*100*	*Periodontitis 1, juvenile, 170650* *Haim-Munk syndrome, 245010* *Papillon-Lefevre syndrome, 245000*
CTSD	*98,4*	*95*	*100*	*100*	*Ceroid lipofuscinosis, neuronal, 10, 610127*
CTSF	*83,9*	*78,9*	*100*	*100*	*Ceroid lipofuscinosis, neuronal, 13 (Kufs type), autosomal dominant, 615362*
CTSH	*100*	*100*	*100*	*100*	*No OMIM disease ID*

CTSK	*100*	*99,2*	*100*	*100*	*Pycnodysostosis, 265800*
CTTNBP2	*99,3*	*96,4*	*100*	*100*	*No OMIM disease ID*
CTU2	*100*	*98,7*	*100*	*100*	*Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142*
CUBN	*99,2*	*97,1*	*100*	*100*	*Imerslund-Grasbeck syndrome 1, 261100*
CUL3	*99,4*	*97,4*	*100*	*100*	*Neurodevelopmental disorder with or without autism or seizures, 619239* *Pseudohypoaldosteronism, type IIE, 614496*
CUL4B	*96,9*	*88,8*	*99,9*	*98,5*	*Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354*
CUL7	*100*	*99,1*	*100*	*100*	*3-M syndrome 1, 273750*
CUX1	*96,5*	*94,6*	*99,5*	*98,7*	*Global developmental delay with or without impaired intellectual development, 618330*
CUX2	*99,9*	*99,3*	*100*	*100*	*Developmental and epileptic encephalopathy 67, 618141*
CWC27	*99,5*	*95,9*	*100*	*100*	*Retinitis pigmentosa with or without skeletal anomalies, 250410*
CWF19L1	*100*	*99,6*	*100*	*100*	*Spinocerebellar ataxia, autosomal recessive 17, 616127*
CXCR2	*100*	*100*	*100*	*100*	*?WHIM syndrome 2, 619407*
CXCR4	*100*	*100*	*100*	*100*	*WHIM syndrome 1, 193670* *Myelokathexis, isolated, 193670*
CYB561	*92,8*	*92,7*	*100*	*100*	*Orthostatic hypotension 2, 618182*
CYB5A	*100*	*100*	*100*	*100*	*Methemoglobinemia and ambiguous genitalia, 250790*
CYB5R3	*99,1*	*98,1*	*99,6*	*98,5*	*Methemoglobinemia, type I, 250800* *Methemoglobinemia, type II, 250800*
CYBA	*96*	*82,5*	*100*	*100*	*Chronic granulomatous disease 4, autosomal recessive, 233690*
CYBB	*99,8*	*98,8*	*100*	*100*	*Immunodeficiency 34, mycobacteriosis, X-linked, 300645* *Chronic granulomatous disease, X-linked, 306400*
CYBC1	*100*	*99,3*	*100*	*100*	*Chronic granulomatous disease 5, autosomal recessive, 618935*
CYBRD1	*100*	*99,7*	*100*	*100*	*No OMIM disease ID*
CYC1	*98,3*	*89,5*	*100*	*99,4*	*Mitochondrial complex III deficiency, nuclear type 6, 615453*
CYCS	*99,4*	*96,9*	*100*	*100*	*Thrombocytopenia 4, 612004*
CYFIP2	*99,9*	*98,7*	*100*	*100*	*Developmental and epileptic encephalopathy 65, 618008*
CYLD	*99,6*	*98,8*	*100*	*100*	*Brooke-Spiegler syndrome, 605041* *Cylindromatosis, familial, 132700* *Trichoepithelioma, multiple familial, 1, 601606* *?Frontotemporal dementia and/or amyotrophic lateral sclerosis 8, 619132*
CYP11A1	*99,2*	*94,5*	*100*	*100*	*Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743*

CYP11B1	*100*	*99,9*	*100*	*100*	*Aldosteronism, glucocorticoid-remediable, 103900* *Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010*
CYP11B2	*100*	*99,9*	*100*	*100*	*Hypoaldosteronism, congenital, due to CMO I deficiency, 203400* *Hypoaldosteronism, congenital, due to CMO II deficiency, 610600* *Aldosterone to renin ratio raised,*
CYP17A1	*99,9*	*98,5*	*100*	*100*	*17,20-lyase deficiency, isolated, 202110* *17-alpha-hydroxylase/17,20-lyase deficiency, 202110*
CYP19A1	*98,3*	*95,7*	*100*	*100*	*Aromatase deficiency, 613546* *Aromatase excess syndrome, 139300*
CYP1B1	*100*	*100*	*100*	*100*	*Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300* *Anterior segment dysgenesis 6, multiple subtypes, 617315*
CYP21A2	*97,4*	*91,1*	*100*	*100*	*Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910* *Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910*
CYP24A1	*100*	*100*	*100*	*100*	*Hypercalcemia, infantile, 1, 143880*
CYP26B1	*100*	*100*	*100*	*100*	*Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416*
CYP26C1	*99,8*	*98,5*	*100*	*99,9*	*Focal facial dermal dysplasia 4, 614974*
CYP27A1	*99,7*	*98,1*	*100*	*100*	*Cerebrotendinous xanthomatosis, 213700*
CYP27B1	*100*	*99,8*	*100*	*100*	*Vitamin D-dependent rickets, type I, 264700*
CYP2A6	*99,9*	*99,2*	*100*	*100*	*Coumarin resistance, 122700*
CYP2B6	*99,3*	*95,7*	*100*	*100*	*Efavirenz, poor metabolism of, 614546*
CYP2C19	*99,7*	*96,2*	*100*	*100*	*Proguanil poor metabolizer, 609535* *Mephenytoin poor metabolizer, 609535* *Clopidogrel, impaired responsiveness to, 609535* *Omeprazole poor metabolizer, 609535*
CYP2C8	*99,9*	*97,5*	*100*	*100*	*No OMIM disease ID*
CYP2C9	*99,7*	*97,9*	*100*	*100*	*Warfarin sensitivity, 122700* *Tolbutamide poor metabolizer,*
CYP2R1	*99,5*	*96*	*100*	*100*	*Rickets due to defect in vitamin D 25-hydroxylation deficiency, 600081*
CYP2U1	*95,3*	*92*	*100*	*99,9*	*Spastic paraplegia 56, autosomal recessive, 615030*
CYP3A4	*94*	*88,4*	*99,7*	*98,3*	*Vitamin D-dependent rickets, type 3, 619073*
CYP4F22	*100*	*98,8*	*100*	*100*	*Ichthyosis, congenital, autosomal recessive 5, 604777*
CYP4V2	*99,6*	*97*	*100*	*100*	*Bietti crystalline corneoretinal dystrophy, 210370*
CYP7B1	*98,1*	*92,7*	*100*	*100*	*Spastic paraplegia 5A, autosomal recessive, 270800* *Bile acid synthesis defect, congenital, 3, 613812*

D2HGDH	*99,7*	*98,2*	*100*	*100*	*D-2-hydroxyglutaric aciduria, 600721*
DAAM2	*99,1*	*98,2*	*100*	*100*	*Nephrotic syndrome, type 24, 619263*
DAB1	*100*	*100*	*100*	*100*	*Spinocerebellar ataxia 37, 615945*
DACT1	*93,9*	*90,3*	*100*	*100*	*?Townes-Brocks syndrome 2, 617466*
DAG1	*100*	*99,9*	*100*	*100*	*Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538* *Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818*
DALRD3	*98,3*	*94,8*	*100*	*100*	*?Developmental and epileptic encephalopathy 86, 618910*
DAO	*100*	*99,9*	*100*	*100*	*No OMIM disease ID*
DARS1	*99,5*	*99,3*	*100*	*99,9*	*Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281*
DARS2	*94,8*	*93,8*	*100*	*100*	*Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105*
DBF4	*96*	*88,9*	*100*	*99,9*	*No OMIM disease ID*
DBH	*100*	*100*	*100*	*100*	*Orthostatic hypotension 1, due to DBH deficiency, 223360*
DBR1	*99,8*	*99*	*100*	*100*	*No OMIM disease ID*
DBT	*99,1*	*96,1*	*100*	*100*	*Maple syrup urine disease, type II, 248600*
DCAF17	*98,5*	*93,4*	*100*	*100*	*Woodhouse-Sakati syndrome, 241080*
DCAF8	*100*	*99,4*	*100*	*100*	*?Giant axonal neuropathy 2, autosomal dominant, 610100*
DCC	*100*	*100*	*100*	*100*	*Mirror movements 1 and/or agenesis of the corpus callosum, 157600* *Esophageal carcinoma, somatic, 133239* *Colorectal cancer, somatic, 114500* *Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542*
DCDC2	*100*	*99,9*	*100*	*100*	*Nephronophthisis 19, 616217* *?Deafness, autosomal recessive 66, 610212* *Sclerosing cholangitis, neonatal, 617394*
DCHS1	*99,9*	*99,4*	*100*	*100*	*Mitral valve prolapse 2, 607829* *Van Maldergem syndrome 1, 601390*
DCLRE1C	*99,8*	*98,2*	*100*	*99,9*	*Severe combined immunodeficiency, Athabascan type, 602450* *Omenn syndrome, 603554*
DCN	*95,7*	*95*	*95,7*	*95,7*	*Corneal dystrophy, congenital stromal, 610048*
DCPS	*91,3*	*91,2*	*100*	*100*	*Al-Raqad syndrome, 616459*
DCT	*99,9*	*99,9*	*100*	*100*	*Oculocutaneous albinism, type VIII, 619165*
DCTN1	*99,8*	*98,4*	*100*	*100*	*Neuronopathy, distal hereditary motor, type VIIB, 607641* *Perry syndrome, 168605*

DCTN2	*99,9*	*97,9*	*100*	*100*	*No OMIM disease ID*
DCX	*100*	*98,9*	*100*	*100*	*Subcortical laminal heterotopia, X-linked, 300067* *Lissencephaly, X-linked, 300067*
DCXR	*99,1*	*94,4*	*100*	*100*	*No OMIM disease ID*
DDB1	*100*	*99,5*	*100*	*100*	*White-Kernohan syndrome, 619426*
DDB2	*99,7*	*97,7*	*100*	*100*	*Xeroderma pigmentosum, group E, DDB-negative subtype, 278740*
DDC	*99,2*	*95*	*100*	*100*	*Aromatic L-amino acid decarboxylase deficiency, 608643*
DDHD1	*98,5*	*96,5*	*100*	*100*	*Spastic paraplegia 28, autosomal recessive, 609340*
DDHD2	*99,7*	*99,5*	*100*	*100*	*Spastic paraplegia 54, autosomal recessive, 615033*
DDOST	*100*	*99,8*	*100*	*100*	*?Congenital disorder of glycosylation, type Ir, 614507*
DDR2	*100*	*99,6*	*100*	*100*	*Warburg-Cinotti syndrome, 618175* *Spondylometaepiphyseal dysplasia, short limb-hand type, 271665*
DDRGK1	*100*	*100*	*100*	*100*	*Spondyloepimetaphyseal dysplasia, Shohat type, 602557*
DDX11	*84,9*	*80*	*100*	*100*	*Warsaw breakage syndrome, 613398*
DDX23	*99,7*	*97,6*	*100*	*100*	*No OMIM disease ID*
DDX3X	*81,1*	*78,6*	*98,3*	*96,1*	*Intellectual developmental disorder, X-linked, syndrome, Snijders Blok type, 300958*
DDX41	*100*	*100*	*100*	*100*	*No OMIM disease ID*
DDX58	*99,6*	*98,3*	*100*	*99,9*	*Singleton-Merten syndrome 2, 616298*
DDX59	*100*	*99,8*	*100*	*100*	*Orofaciodigital syndrome V, 174300*
DDX6	*95,8*	*81,5*	*100*	*100*	*Intellectual developmental disorder with impaired language and dysmorphic facies, 618653*
DEAF1	*99*	*94,5*	*99,9*	*98,2*	*Vulto-van Silfout-de Vries syndrome, 615828* *Neurodevelopmental disorder with hypotonia, impaired expressive language, and with or without seizures, 617171*
DEF6	*96,4*	*92,9*	*100*	*100*	*No OMIM disease ID*
DEGS1	*100*	*100*	*100*	*100*	*Leukodystrophy, hypomyelinating, 18, 618404*
DENND5A	*99,8*	*98,7*	*100*	*100*	*Developmental and epileptic encephalopathy 49, 617281*
DEPDC5	*99,9*	*99,7*	*100*	*100*	*Epilepsy, familial focal, with variable foci 1, 604364*
DES	*100*	*99,6*	*100*	*100*	*Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400* *Cardiomyopathy, dilated, 1I, 604765* *Myopathy, myofibrillar, 1, 601419*
DGAT1	*91,8*	*87,6*	*99,9*	*98,9*	*?Diarrhea 7, protein-losing enteropathy type, 615863*

DGAT2	*98,5*	*94,7*	*100*	*100*	*No OMIM disease ID*
DGKE	*99,7*	*98,5*	*100*	*100*	*Nephrotic syndrome, type 7, 615008*
DGUOK	*99,9*	*98,8*	*100*	*100*	*Portal hypertension, noncirrhotic, 1, 617068* *Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070* *Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880*
DHCR24	*97,7*	*97,7*	*97,7*	*97,7*	*Desmosterolosis, 602398*
DHCR7	*100*	*100*	*100*	*100*	*Smith-Lemli-Opitz syndrome, 270400*
DHDDS	*99,4*	*95,6*	*95,2*	*95,2*	*Developmental delay and seizures with or without movement abnormalities, 617836* *?Congenital disorder of glycosylation, type 1bb, 613861* *Retinitis pigmentosa 59, 613861*
DHFR	*88,9*	*76,3*	*100*	*100*	*Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839*
DHH	*100*	*100*	*100*	*100*	*46XY gonadal dysgenesis with minifascicular neuropathy, 607080* *46XY sex reversal 7, 233420*
DHODH	*100*	*99,9*	*100*	*100*	*Miller syndrome, 263750*
DHPS	*100*	*99,8*	*93,3*	*93,2*	*Neurodevelopmental disorder with seizures and speech and walking impairment, 618480*
DHTKD1	*99,8*	*98,8*	*100*	*100*	*?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025* *Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750*
DHX16	*100*	*99,7*	*100*	*100*	*Neuromuscular disease and ocular or auditory anomalies with or without seizures, 618733*
DHX30	*100*	*100*	*100*	*100*	*Neurodevelopmental disorder with severe motor impairment and absent language, 617804*
DHX37	*99,8*	*97*	*100*	*100*	*Neurodevelopmental disorder with brain anomalies and with or without vertebral or cardiac anomalies, 618731* *46, XY sex reversal 11, 273250*
DHX38	*100*	*99,1*	*100*	*100*	*Retinitis pigmentosa 84, 618220*
DIABLO	*99,9*	*97,9*	*100*	*100*	*Deafness, autosomal dominant 64, 614152*
DIAPH1	*99,8*	*98,4*	*99,9*	*99*	*Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900* *Seizures, cortical blindness, microcephaly syndrome, 616632*
DIAPH2	*95,3*	*86,8*	*99,8*	*97,9*	*?Premature ovarian failure 2A, 300511*
DIAPH3	*99*	*96,4*	*100*	*99,9*	*Auditory neuropathy, autosomal dominant, 1, 609129*
DICER1	*99,5*	*98,5*	*100*	*100*	*Pleuropulmonary blastoma, 601200* *Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800* *GLOW syndrome, somatic mosaic, 618272* *Rhabdomyosarcoma, embryonal, 2, 180295*
DIP2B	*99,8*	*99*	*100*	*100*	*Mental retardation, FRA12A type, 136630*
DIS3L2	*100*	*100*	*100*	*100*	*Perlman syndrome, 267000*

DISP1	*99,9*	*99,9*	*100*	*100*	*No OMIM disease ID*
DKC1	*99,7*	*97,2*	*100*	*99,6*	*Dyskeratosis congenita, X-linked, 305000*
DLAT	*99,8*	*99,3*	*100*	*99,9*	*Pyruvate dehydrogenase E2 deficiency, 245348*
DLC1	*100*	*99,9*	*100*	*100*	*Colorectal cancer, somatic, 114500*
DLD	*99,9*	*99,7*	*100*	*99,9*	*Dihydrolipoamide dehydrogenase deficiency, 246900*
DLG3	*98,9*	*92,5*	*100*	*100*	*Intellectual developmental disorder, X-linked 90, 300850*
DLG4	*99,1*	*98,7*	*98,8*	*98,8*	*Intellectual developmental disorder 62, 618793*
DLL1	*100*	*98,8*	*100*	*100*	*Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizures, 618709*
DLL3	*93*	*87,8*	*100*	*99,5*	*Spondylocostal dysostosis 1, autosomal recessive, 277300*
DLL4	*100*	*99,4*	*100*	*100*	*Adams-Oliver syndrome 6, 616589*
DLST	*95,7*	*87,7*	*100*	*100*	*Paragangliomas 7, 618475*
DLX3	*99,8*	*97,6*	*100*	*100*	*Trichodontoosseous syndrome, 190320* *Amelogenesis imperfecta, type IV, 104510*
DLX4	*100*	*100*	*100*	*100*	*?Orofacial cleft 15, 616788*
DLX5	*99,9*	*98,2*	*100*	*100*	*Split-hand/foot malformation 1, 183600* *?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600*
DLX6	*100*	*100*	*100*	*100*	*No OMIM disease ID*
DMAC1	*100*	*99,9*	*100*	*100*	*No OMIM disease ID*
DMAC2	*98,3*	*98,3*	*100*	*100*	*No OMIM disease ID*
DMAC2L	*99,8*	*99,8*	*100*	*99,9*	*No OMIM disease ID*
DMC1	*99,6*	*97,4*	*100*	*100*	*No OMIM disease ID*
DMD	*99,5*	*98,1*	*100*	*99,9*	*Becker muscular dystrophy, 300376* *Cardiomyopathy, dilated, 3B, 302045* *Duchenne muscular dystrophy, 310200*
DMGDH	*99,9*	*99,7*	*100*	*100*	*Dimethylglycine dehydrogenase deficiency, 605850*
DMP1	*99,9*	*99,9*	*100*	*100*	*Hypophosphatemic rickets, AR, 241520*
DMPK	*99,8*	*98,4*	*100*	*100*	*Myotonic dystrophy 1, 160900*
DMRT1	*100*	*98,9*	*100*	*100*	*No OMIM disease ID*
DMRT2	*98,9*	*91,6*	*100*	*100*	*No OMIM disease ID*

DMXL2	*99,7*	*98,9*	*100*	*99,9*	*Developmental and epileptic encephalopathy 81, 618663* *?Deafness, autosomal dominant 71, 617605* *?Polyendocrine-polyneuropathy syndrome, 616113*
DNA2	*99,6*	*96,9*	*100*	*100*	*?Seckel syndrome 8, 615807* *Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156*
DNAAF1	*100*	*99,4*	*100*	*100*	*Ciliary dyskinesia, primary, 13, 613193*
LRRC6	*99,3*	*96,9*	*100*	*100*	*Ciliary dyskinesia, primary, 19, 614935*
DNAAF2	*99,7*	*98,4*	*100*	*100*	*Ciliary dyskinesia, primary, 10, 612518*
DNAAF3	*99,5*	*96,3*	*100*	*100*	*Ciliary dyskinesia, primary, 2, 606763*
DNAAF4	*99,4*	*94,7*	*100*	*99,7*	*Ciliary dyskinesia, primary, 25, 615482*
DNAAF5	*85,6*	*78,8*	*98,8*	*96,9*	*Ciliary dyskinesia, primary, 18, 614874*
PIH1D3	*97,9*	*86*	*99,9*	*99,6*	*Ciliary dyskinesia, primary, 36, X-linked, 300991*
DNAH1	*99,9*	*99,6*	*100*	*100*	*Spermatogenic failure 18, 617576* *?Ciliary dyskinesia, primary, 37, 617577*
DNAH10	*99,9*	*99,1*	*100*	*100*	*Spermatogenic failure 56, 619515*
DNAH11	*99,8*	*98,8*	*100*	*100*	*Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884*
DNAH17	*99,9*	*99,2*	*100*	*100*	*Spermatogenic failure 39, 618643*
DNAH2	*98,8*	*97,8*	*99,6*	*99*	*Spermatogenic failure 45, 619094*
DNAH5	*99,9*	*98,9*	*100*	*100*	*Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644*
DNAH8	*99,7*	*98,6*	*100*	*99,9*	*Spermatogenic failure 46, 619095*
DNAH9	*99,5*	*97,8*	*100*	*100*	*Ciliary dyskinesia, primary, 40, 618300*
DNAI1	*100*	*99,9*	*100*	*100*	*Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400*
DNAI2	*98,2*	*95,8*	*100*	*100*	*Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444*
DNAJA3	*98,5*	*96,1*	*100*	*100*	*No OMIM disease ID*
DNAJB11	*99,9*	*99,6*	*100*	*100*	*Polycystic kidney disease 6 with or without polycystic liver disease, 618061*
DNAJB13	*100*	*99,6*	*100*	*100*	*Ciliary dyskinesia, primary, 34, 617091*
DNAJB2	*100*	*100*	*100*	*100*	*Spinal muscular atrophy, distal, autosomal recessive, 5, 614881*
DNAJB5	*95,5*	*89,4*	*100*	*100*	*No OMIM disease ID*
DNAJB6	*95,9*	*84,6*	*100*	*100*	*Muscular dystrophy, limb-girdle, autosomal dominant 1, 603511*
DNAJC12	*87,4*	*87,3*	*100*	*100*	*Hyperphenylalaninemia, mild, non-BH4-deficient, 617384*

DNAJC19	*99,3*	*92,2*	*100*	*100*	*3-methylglutaconic aciduria, type V, 610198*
DNAJC21	*99,5*	*97,4*	*100*	*100*	*Bone marrow failure syndrome 3, 617052*
DNAJC3	*99,7*	*99,7*	*100*	*99,9*	*?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192*
DNAJC30	*100*	*100*	*100*	*100*	*Leber hereditary optic neuropathy, autosomal recessive, 619382*
DNAJC5	*100*	*100*	*100*	*100*	*Ceroid lipofuscinosis, neuronal, 4B (Kufs type), autosomal dominant, 162350*
DNAJC6	*99,8*	*98,7*	*100*	*100*	*Parkinson disease 19a, juvenile-onset, 615528* *Parkinson disease 19b, early-onset, 615528*
DNAL1	*99,6*	*98,3*	*100*	*99,2*	*Ciliary dyskinesia, primary, 16, 614017*
DNAL4	*99,8*	*95,2*	*100*	*100*	*?Mirror movements 3, 616059*
DNASE1	*100*	*100*	*100*	*100*	*No OMIM disease ID*
DNASE1L3	*100*	*100*	*100*	*100*	*Systemic lupus erythematosus 16, 614420*
DNASE2	*98,4*	*95,1*	*100*	*100*	*No OMIM disease ID*
DNM1	*92,7*	*89*	*97,5*	*97,4*	*Developmental and epileptic encephalopathy 31, 616346*
DNM1L	*99,6*	*98,3*	*100*	*100*	*Optic atrophy 5, 610708* *Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388*
DNM2	*98,6*	*93,9*	*100*	*100*	*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	*99,9*	*98,9*	*100*	*100*	*Cataract 48, 618415*
DNMT1	*99,2*	*98,8*	*99,9*	*99,4*	*Neuropathy, hereditary sensory, type IE, 614116* *Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121*
DNMT3A	*99,8*	*98,2*	*100*	*100*	*Tatton-Brown-Rahman syndrome, 615879* *Acute myeloid leukemia, somatic, 601626* *Heyn-Sproul-Jackson syndrome, 618724*
DNMT3B	*100*	*99,9*	*100*	*100*	*Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860* *Facioscapulohumeral muscular dystrophy 4, digenic, 619478*
DOCK2	*99,8*	*98,8*	*100*	*100*	*Immunodeficiency 40, 616433*
DOCK3	*99,9*	*98,9*	*100*	*100*	*Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292*
DOCK6	*99,4*	*98,7*	*100*	*100*	*Adams-Oliver syndrome 2, 614219*
DOCK7	*99,6*	*98,3*	*100*	*99,9*	*Developmental and epileptic encephalopathy 23, 615859*
DOCK8	*100*	*99*	*100*	*100*	*Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700*

DOK7	*94,9*	*92*	*100*	*100*	*Fetal akinesia deformation sequence 3, 618389* *Myasthenic syndrome, congenital, 10, 254300*
DOLK	*100*	*100*	*100*	*100*	*Congenital disorder of glycosylation, type Im, 610768*
DONSON	*93,8*	*85,8*	*100*	*100*	*Microcephaly, short stature, and limb abnormalities, 617604* *Microcephaly-micromelia syndrome, 251230*
DOT1L	*100*	*99,4*	*100*	*100*	*No OMIM disease ID*
DPAGT1	*100*	*99,8*	*100*	*100*	*Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750* *Congenital disorder of glycosylation, type Ij, 608093*
DPCD	*100*	*100*	*100*	*100*	*No OMIM disease ID*
DPF2	*99,5*	*96,4*	*100*	*100*	*Coffin-Siris syndrome 7, 618027*
DPH1	*100*	*99,9*	*100*	*100*	*Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901*
DPM1	*97,4*	*90,9*	*98,6*	*94,6*	*Congenital disorder of glycosylation, type Ie, 608799*
DPM2	*100*	*97,7*	*100*	*100*	*Congenital disorder of glycosylation, type Iu, 615042*
DPM3	*100*	*100*	*100*	*100*	*?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992* *Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937*
DPP6	*99,7*	*97,9*	*98,8*	*96,8*	*Mental retardation, autosomal dominant 33, 616311*
DPY19L2	*73,7*	*69,3*	*100*	*99,9*	*Spermatogenic failure 9, 613958*
DPYD	*99,5*	*96,5*	*100*	*100*	*Dihydropyrimidine dehydrogenase deficiency, 274270* *5-fluorouracil toxicity, 274270*
DPYS	*100*	*100*	*100*	*100*	*Dihydropyrimidinuria, 222748*
DPYSL5	*100*	*99,9*	*100*	*100*	*Ritscher-Schinzel syndrome 4, 619435*
DRAM2	*100*	*99,9*	*100*	*100*	*Cone-rod dystrophy 21, 616502*
DRC1	*99,9*	*98,3*	*100*	*100*	*Ciliary dyskinesia, primary, 21, 615294*
DRD4	*94,8*	*83,6*	*100*	*100*	*Autonomic nervous system dysfunction,*
DRP2	*98,3*	*93,6*	*100*	*100*	*No OMIM disease ID*
DSC2	*99,4*	*97,4*	*100*	*99,9*	*Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476* *Arrhythmogenic right ventricular dysplasia 11, 610476*
DSC3	*98,9*	*96,7*	*100*	*100*	*Hypotrichosis and recurrent skin vesicles, 613102*
DSE	*98,4*	*95,4*	*100*	*100*	*Ehlers-Danlos syndrome, musculocontractural type 2, 615539*
DSG1	*99,3*	*97,4*	*100*	*99,9*	*Keratosis palmoplantaris striata I, AD, 148700* *Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508*

DSG2	*99,8*	*99,6*	*100*	*100*	*Cardiomyopathy, dilated, 1BB, 612877* *Arrhythmogenic right ventricular dysplasia 10, 610193*
DSG3	*99,9*	*99,7*	*100*	*100*	*Blistering, acantholytic, of oral and laryngeal mucosa, 619226*
DSG4	*99,8*	*99*	*100*	*100*	*Hypotrichosis 6, 607903*
DSP	*99,9*	*99,4*	*100*	*100*	*Arrhythmogenic right ventricular dysplasia 8, 607450* *Skin fragility-woolly hair syndrome, 607655* *Epidermolysis bullosa, lethal acantholytic, 609638* *Keratosis palmoplantaris striata II, 612908* *Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821* *Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676*
DSPP	*97,8*	*93,5*	*100*	*100*	*Dentinogenesis imperfecta, Shields type III, 125500* *Dentinogenesis imperfecta, Shields type II, 125490* *Dentin dysplasia, type II, 125420* *Deafness, autosomal dominant 39, with dentinogenesis, 605594*
DST	*95,3*	*94,5*	*95,6*	*95,6*	*Epidermolysis bullosa simplex, autosomal recessive 2, 615425* *?Neuropathy, hereditary sensory and autonomic, type VI, 614653*
DSTYK	*99,9*	*98,8*	*100*	*100*	*Congenital anomalies of kidney and urinary tract 1, 610805* *Spastic paraplegia 23, 270750*
DTNA	*99,9*	*99,9*	*100*	*100*	*Left ventricular noncompaction 1, with or without congenital heart defects, 604169*
DTNBP1	*99,7*	*98*	*100*	*99,9*	*Hermansky-Pudlak syndrome 7, 614076*
DTYMK	*100*	*100*	*100*	*100*	*No OMIM disease ID*
DUOX2	*97,2*	*94,8*	*100*	*100*	*Thyroid dyshormonogenesis 6, 607200*
DUOXA2	*100*	*100*	*100*	*100*	*Thyroid dyshormonogenesis 5, 274900*
DUSP6	*100*	*100*	*100*	*100*	*Hypogonadotropic hypogonadism 19 with or without anosmia, 615269*
DVL1	*97,2*	*95,1*	*100*	*100*	*Robinow syndrome, autosomal dominant 2, 616331*
DVL3	*100*	*100*	*100*	*100*	*Robinow syndrome, autosomal dominant 3, 616894*
DYM	*97*	*95,6*	*100*	*100*	*Smith-McCort dysplasia, 607326* *Dyggve-Melchior-Clausen disease, 223800*
DYNC1H1	*99,9*	*99,3*	*100*	*100*	*Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600* *Charcot-Marie-Tooth disease, axonal, type 20, 614228* *Mental retardation, autosomal dominant 13, 614563*
DYNC1I2	*84*	*66*	*100*	*100*	*Neurodevelopmental disorder with microcephaly and structural brain anomalies, 618492*
DYNC2H1	*98,6*	*95,2*	*100*	*99,8*	*Short-rib thoracic dysplasia 3 with or without polydactyly, 613091*
WDR60	*99,3*	*95,8*	*100*	*100*	*Short-rib thoracic dysplasia 8 with or without polydactyly, 615503*

WDR34	*100*	*99,8*	*100*	*100*	*Short-rib thoracic dysplasia 11 with or without polydactyly, 615633*
DYNC2LI1	*99,6*	*98,4*	*100*	*100*	*Short-rib thoracic dysplasia 15 with polydactyly, 617088*
TCTEX1D2	*100*	*99,8*	*100*	*100*	*Short-rib thoracic dysplasia 17 with or without polydactyly, 617405*
DYRK1A	*100*	*100*	*100*	*100*	*Mental retardation, autosomal dominant 7, 614104*
DYRK1B	*97,9*	*91,7*	*100*	*100*	*Abdominal obesity-metabolic syndrome 3, 615812*
DYSF	*100*	*99,8*	*100*	*100*	*Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601* *Miyoshi muscular dystrophy 1, 254130* *Myopathy, distal, with anterior tibial onset, 606768*
DZIP1	*98,3*	*95,9*	*100*	*100*	*Spermatogenic failure 47, 619102* *?Mitral valve prolapse 3, 610840*
DZIP1L	*99,8*	*98*	*100*	*100*	*Polycystic kidney disease 5, 617610*
E2F1	*80*	*78,3*	*98,3*	*93,2*	*No OMIM disease ID*
EARS2	*99,8*	*98*	*100*	*100*	*Combined oxidative phosphorylation deficiency 12, 614924*
EBF3	*100*	*100*	*100*	*100*	*Hypotonia, ataxia, and delayed development syndrome, 617330*
EBP	*99,5*	*94,3*	*100*	*100*	*MEND syndrome, 300960* *Chondrodysplasia punctata, X-linked dominant, 302960*
ECE1	*88,8*	*88,2*	*90,2*	*90,2*	*?Hirschsprung disease, cardiac defects, and autonomic dysfunction, 613870*
ECEL1	*95,9*	*91,8*	*100*	*100*	*Arthrogryposis, distal, type 5D, 615065*
ECHS1	*100*	*99,4*	*100*	*100*	*Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277*
ECM1	*100*	*99,6*	*100*	*100*	*Urbach-Wiethe disease, 247100*
ECSIT	*100*	*100*	*100*	*100*	*No OMIM disease ID*
EDA	*98,3*	*89,7*	*100*	*99,8*	*Tooth agenesis, selective, X-linked 1, 313500* *Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100*
EDAR	*100*	*99,7*	*100*	*100*	*Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490* *Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900*
EDARADD	*99,7*	*98,9*	*100*	*100*	*Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941* *Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940*
EDC3	*100*	*99,5*	*100*	*100*	*?Mental retardation, autosomal recessive 50, 616460*
EDEM3	*99,3*	*96,9*	*100*	*99,8*	*Congenital disorder of glycosylation, type 2V, 619493*
EDN1	*100*	*99,4*	*100*	*100*	*Question mark ears, isolated, 612798* *Auriculocondylar syndrome 3, 615706*
EDN3	*98,8*	*98,8*	*100*	*100*	*Waardenburg syndrome, type 4B, 613265*

EDNRA	*99,8*	*99,8*	*100*	*99,9*	*Mandibulofacial dysostosis with alopecia, 616367*
EDNRB	*96,3*	*92,5*	*100*	*100*	*ABCD syndrome, 600501* *Waardenburg syndrome, type 4A, 277580*
EED	*95,6*	*91,7*	*100*	*100*	*Cohen-Gibson syndrome, 617561*
EEF1A2	*100*	*100*	*100*	*99,4*	*Mental retardation, autosomal dominant 38, 616393* *Developmental and epileptic encephalopathy 33, 616409*
EEF2	*100*	*100*	*100*	*100*	*?Spinocerebellar ataxia 26, 609306*
EFEMP1	*100*	*100*	*100*	*99,9*	*Doyne honeycomb degeneration of retina, 126600*
EFEMP2	*100*	*100*	*100*	*100*	*Cutis laxa, autosomal recessive, type IB, 614437*
EFHC1	*92,8*	*91*	*98*	*98*	*No OMIM disease ID*
EFL1	*99,3*	*97,7*	*100*	*100*	*Shwachman-Diamond syndrome 2, 617941*
EFNA4	*100*	*100*	*100*	*100*	*No OMIM disease ID*
EFNB1	*100*	*100*	*100*	*100*	*Craniofrontonasal dysplasia, 304110*
EFNB2	*100*	*99,7*	*100*	*100*	*No OMIM disease ID*
EFTUD2	*100*	*99,2*	*100*	*100*	*Mandibulofacial dysostosis, Guion-Almeida type, 610536*
EGF	*99,9*	*99,8*	*100*	*100*	*?Hypomagnesemia 4, renal, 611718*
EGFR	*100*	*100*	*100*	*100*	*?Inflammatory skin and bowel disease, neonatal, 2, 616069* *Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980* *Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980*
EGLN1	*89,1*	*80,1*	*100*	*100*	*Erythrocytosis, familial, 3, 609820*
EGLN2	*100*	*100*	*100*	*100*	*No OMIM disease ID*
EGR2	*100*	*100*	*100*	*100*	*Dejerine-Sottas disease, 145900* *Charcot-Marie-Tooth disease, type 1D, 607678* *Hypomyelinating neuropathy, congenital, 1, 605253*
EHHADH	*100*	*100*	*100*	*100*	*?Fanconi renotubular syndrome 3, 615605*
EHMT1	*94,5*	*93,6*	*99,6*	*99,5*	*Kleefstra syndrome 1, 610253*
EIF2AK1	*98,1*	*94,5*	*100*	*100*	*?Leukoencephalopathy, motor delay, spasticity, and dysarthria syndrome, 618878*
EIF2AK2	*99,7*	*98,9*	*100*	*99,9*	*Leukoencephalopathy, developmental delay, and episodic neurologic regression syndrome, 618877*
EIF2AK3	*98,2*	*95,5*	*100*	*99,8*	*Wolcott-Rallison syndrome, 226980*
EIF2AK4	*99,6*	*97,8*	*100*	*100*	*Pulmonary venoocclusive disease 2, 234810*
EIF2B1	*100*	*99,8*	*100*	*100*	*Leukoencephalopathy with vanishing white matter, 603896*

EIF2B2	*100*	*98,1*	*100*	*100*	*Leukoencephalopathy with vanishing white matter, 603896* *Ovarioleukodystrophy, 603896*
EIF2B3	*100*	*100*	*100*	*100*	*Leukoencephalopathy with vanishing white matter, 603896*
EIF2B4	*100*	*99,5*	*100*	*100*	*Ovarioleukodystrophy, 603896* *Leukoencephalopathy with vanishing white matter, 603896*
EIF2B5	*99,8*	*98,5*	*100*	*100*	*Ovarioleukodystrophy, 603896* *Leukoencephalopathy with vanishing white matter, 603896*
EIF2S3	*95*	*86,8*	*100*	*100*	*MEHMO syndrome, 300148*
EIF3F	*97,1*	*82,5*	*100*	*100*	*Mental retardation, autosomal recessive 67, 618295*
EIF4A3	*100*	*99,2*	*100*	*100*	*Robin sequence with cleft mandible and limb anomalies, 268305*
EIF5A	*99,8*	*96,4*	*100*	*100*	*Faundes-Banka syndrome, 619376*
ELAC2	*100*	*99,2*	*100*	*100*	*Combined oxidative phosphorylation deficiency 17, 615440*
ELANE	*99,9*	*98,8*	*100*	*100*	*Neutropenia, cyclic, 162800* *Neutropenia, severe congenital 1, autosomal dominant, 202700*
ELF2	*99*	*94,8*	*100*	*100*	*No OMIM disease ID*
ELF4	*99,9*	*98,7*	*100*	*100*	*No OMIM disease ID*
ELMO2	*100*	*99*	*100*	*100*	*Vascular malformation, primary intraosseous, 606893*
ELMOD3	*100*	*100*	*100*	*100*	*?Deafness, autosomal recessive 88, 615429* *?Deafness, autosomal dominant 81, 619500*
ELN	*99,8*	*98,3*	*100*	*100*	*Cutis laxa, autosomal dominant, 123700* *Supravalvar aortic stenosis, 185500*
ELOVL1	*99,6*	*96,5*	*100*	*100*	*Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527*
ELOVL4	*99,7*	*98,9*	*100*	*99,9*	*Spinocerebellar ataxia 34, 133190* *Stargardt disease 3, 600110* *Ichthyosis, spastic quadriplegia, and mental retardation, 614457*
ELOVL5	*100*	*99,4*	*100*	*100*	*Spinocerebellar ataxia 38, 615957*
ELP1	*99,8*	*98,9*	*100*	*100*	*Dysautonomia, familial, 223900*
ELP2	*99,8*	*98,3*	*100*	*99,9*	*Mental retardation, autosomal recessive 58, 617270*
ELP4	*72,1*	*69,5*	*87,1*	*87*	*?Aniridia 2, 617141*
EMC1	*99,9*	*98*	*100*	*100*	*Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875*
EMC10	*97,1*	*92*	*100*	*100*	*Neurodevelopmental disorder with dysmorphic facies and variable seizures, 619264*
EMD	*99,8*	*97,9*	*100*	*99,8*	*Emery-Dreifuss muscular dystrophy 1, X-linked, 310300*

EMG1	*100*	*100*	*100*	*100*	*Bowen-Conradi syndrome, 211180*
EMILIN1	*98,3*	*90,3*	*100*	*100*	*No OMIM disease ID*
EML1	*99,6*	*98,1*	*100*	*100*	*Band heterotopia, 600348*
EMP2	*98,9*	*93*	*100*	*100*	*Nephrotic syndrome, type 10, 615861*
EMX2	*100*	*100*	*100*	*100*	*Schizencephaly, 269160*
EN1	*99,4*	*93,1*	*99,6*	*97,4*	*?ENDOVE syndrome, limb-brain type, 619218*
ENAM	*100*	*100*	*100*	*100*	*Amelogenesis imperfecta, type IC, 204650* *Amelogenesis imperfecta, type IB, 104500*
ENG	*99,8*	*97*	*100*	*100*	*Telangiectasia, hereditary hemorrhagic, type 1, 187300*
ENO3	*100*	*100*	*100*	*100*	*?Glycogen storage disease XIII, 612932*
ENPP1	*96,5*	*90,6*	*98,8*	*97,8*	*Hypophosphatemic rickets, autosomal recessive, 2, 613312* *Arterial calcification, generalized, of infancy, 1, 208000* *Cole disease, 615522*
ENTPD1	*100*	*99,8*	*100*	*100*	*Spastic paraplegia 64, autosomal recessive, 615683*
EOGT	*79,3*	*77,8*	*91,8*	*88,3*	*Adams-Oliver syndrome 4, 615297*
EP300	*99,9*	*98,9*	*100*	*100*	*Menke-Hennekam syndrome 2, 618333* *Colorectal cancer, somatic, 114500* *Rubinstein-Taybi syndrome 2, 613684*
EPAS1	*99,8*	*98,1*	*100*	*100*	*Erythrocytosis, familial, 4, 611783*
EPB41	*85,4*	*83,8*	*100*	*100*	*Elliptocytosis-1, 611804*
EPB41L1	*99,4*	*96,7*	*97,8*	*97,8*	*?Intellectual developmental disorder, autosomal dominant 11, 614257*
EPB42	*99,9*	*98,5*	*100*	*100*	*Spherocytosis, type 5, 612690*
EPCAM	*97,5*	*89,6*	*100*	*100*	*Colorectal cancer, hereditary nonpolyposis, type 8, 613244* *Diarrhea 5, with tufting enteropathy, congenital, 613217*
EPG5	*99,2*	*97,8*	*100*	*100*	*Vici syndrome, 242840*
EPHA2	*100*	*99,7*	*100*	*100*	*Cataract 6, multiple types, 116600*
EPHA7	*100*	*99,4*	*100*	*100*	*No OMIM disease ID*
EPHB2	*98,1*	*98,1*	*99,4*	*98,7*	*?Bleeding disorder, platelet-type, 22, 618462*
EPHB4	*100*	*99,6*	*100*	*100*	*Capillary malformation-arteriovenous malformation 2, 618196* *Lymphatic malformation 7, 617300*
EPHX1	*99,8*	*97,8*	*100*	*100*	*No OMIM disease ID*
EPHX2	*99,5*	*96,5*	*100*	*99,9*	*No OMIM disease ID*

EPM2A	*93,9*	*91,2*	*99,6*	*96,3*	*Epilepsy, progressive myoclonic 2A (Lafora), 254780*
EPO	*99,9*	*97,8*	*100*	*100*	*Erythrocytosis, familial, 5, 617907* *?Diamond-Blackfan anemia-like, 617911*
EPRS1	*99,8*	*99,4*	*100*	*100*	*Leukodystrophy, hypomyelinating, 15, 617951*
EPS8	*96,9*	*96*	*100*	*100*	*?Deafness, autosomal recessive 102, 615974*
EPS8L2	*84,7*	*82,3*	*88*	*88*	*Deafness autosomal recessive 106, 617637*
EPS8L3	*99,1*	*97,5*	*100*	*100*	*?Hypotrichosis 5, 612841*
ERAL1	*100*	*99,6*	*100*	*100*	*Perrault syndrome 6, 617565*
ERBB2	*98,5*	*97,2*	*100*	*100*	*Gastric cancer, somatic, 613659* *Adenocarcinoma of lung, somatic, 211980* *Ovarian cancer, somatic, 167000* *?Visceral neuropathy, familial, 2, autosomal recessive, 619465* *Glioblastoma, somatic, 137800*
ERBB3	*100*	*99,3*	*100*	*100*	*?Lethal congenital contractural syndrome 2, 607598* *Visceral neuropathy, familial, 1, autosomal recessive, 243180*
ERBB4	*99,9*	*99,5*	*100*	*100*	*Amyotrophic lateral sclerosis 19, 615515*
ERCC1	*100*	*96,4*	*100*	*100*	*Cerebrooculofacioskeletal syndrome 4, 610758*
ERCC2	*100*	*99,4*	*100*	*100*	*Xeroderma pigmentosum, group D, 278730* *Trichothiodystrophy 1, photosensitive, 601675* *?Cerebrooculofacioskeletal syndrome 2, 610756*
ERCC3	*96,8*	*95,6*	*100*	*100*	*Trichothiodystrophy 2, photosensitive, 616390* *Xeroderma pigmentosum, group B, 610651*
ERCC4	*100*	*99,9*	*100*	*100*	*Xeroderma pigmentosum, type F/Cockayne syndrome, 278760* *XFE progeroid syndrome, 610965* *Xeroderma pigmentosum, group F, 278760* *Fanconi anemia, complementation group Q, 615272*
ERCC5	*99,9*	*99*	*100*	*100*	*Xeroderma pigmentosum, group G, 278780* *Cerebrooculofacioskeletal syndrome 3, 616570* *Xeroderma pigmentosum, group G/Cockayne syndrome, 278780*
ERCC6	*100*	*100*	*100*	*100*	*UV-sensitive syndrome 1, 600630* *Cerebrooculofacioskeletal syndrome 1, 214150* *Cockayne syndrome, type B, 133540* *De Sanctis-Cacchione syndrome, 278800* *Premature ovarian failure 11, 616946*
ERCC6L2	*99,6*	*98,6*	*100*	*100*	*Bone marrow failure syndrome 2, 615715*

ERCC8	*99*	*94,8*	*100*	*100*	*UV-sensitive syndrome 2, 614621* *Cockayne syndrome, type A, 216400*
ERF	*100*	*98,9*	*100*	*100*	*Craniosynostosis 4, 600775* *Chitayat syndrome, 617180*
ERGIC1	*95,3*	*94,6*	*98,4*	*98,4*	*?Arthrogryposis multiplex congenita 2, neurogenic type, 208100*
ERLIN1	*100*	*100*	*100*	*100*	*Spastic paraplegia 62, 615681*
ERLIN2	*100*	*99,1*	*100*	*99,9*	*Spastic paraplegia 18, autosomal recessive, 611225*
ERMARD	*99,8*	*98,5*	*100*	*100*	*?Periventricular nodular heterotopia 6, 615544*
ESCO2	*98,5*	*94,6*	*100*	*99,7*	*Juberg-Hayward syndrome, 216100* *Roberts-SC phocomelia syndrome, 268300*
ESPN	*48,5*	*38,4*	*100*	*99,9*	*Deafness, neurosensory, without vestibular involvement, autosomal dominant, 609006* *Deafness, autosomal recessive 36, 609006* *?Usher syndrome, type 1M, 618632*
ESR1	*100*	*99,8*	*100*	*100*	*Breast cancer, somatic, 114480* *Estrogen resistance, 615363*
ESR2	*99,9*	*98,9*	*100*	*100*	*?Ovarian dysgenesis 8, 618187*
ESRP1	*99,9*	*98,4*	*100*	*100*	*?Deafness, autosomal recessive 109, 618013*
ESRRB	*96,2*	*93*	*100*	*100*	*Deafness, autosomal recessive 35, 608565*
ETFA	*99,8*	*99,6*	*100*	*99,9*	*Glutaric acidemia IIA, 231680*
ETFB	*100*	*99,9*	*100*	*100*	*Glutaric acidemia IIB, 231680*
ETFDH	*99,8*	*99,4*	*100*	*100*	*Glutaric acidemia IIC, 231680*
ETHE1	*99,3*	*93,3*	*100*	*100*	*Ethylmalonic encephalopathy, 602473*
ETV6	*100*	*99,3*	*100*	*100*	*Thrombocytopenia 5, 616216* *Leukemia, acute myeloid, somatic, 601626*
EVC	*94,2*	*91,4*	*97,5*	*95,1*	*Ellis-van Creveld syndrome, 225500* *?Weyers acrofacial dysostosis, 193530*
EVC2	*98*	*96,2*	*100*	*100*	*Ellis-van Creveld syndrome, 225500* *Weyers acrofacial dysostosis, 193530*
EWSR1	*90*	*82*	*100*	*100*	*Neuroepithelioma, 612219* *Ewing sarcoma, 612219*
EXOC2	*99,8*	*99,5*	*100*	*100*	*Neurodevelopmental disorder with dysmorphic facies and cerebellar hypoplasia, 619306*
EXOC6	*98,5*	*95,7*	*100*	*99,9*	*No OMIM disease ID*
EXOC6B	*98,1*	*97,1*	*99,9*	*99,5*	*Spondyloepimetaphyseal dysplasia with joint laxity, type 3, 618395*

EXOC7	*100*	*99,6*	*100*	*100*	*Neurodevelopmental disorder with seizures and brain atrophy, 619072*
EXOC8	*100*	*100*	*100*	*100*	*?Neurodevelopmental disorder with microcephaly, seizures, and brain atrophy, 619076*
EXOSC1	*100*	*100*	*100*	*99,9*	*?Pontocerebellar hypoplasia, type 1F, 619304*
EXOSC2	*100*	*99,9*	*100*	*100*	*Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763*
EXOSC3	*98,1*	*90,5*	*100*	*100*	*Pontocerebellar hypoplasia, type 1B, 614678*
EXOSC5	*100*	*100*	*100*	*100*	*No OMIM disease ID*
EXOSC8	*98,7*	*90*	*100*	*100*	*Pontocerebellar hypoplasia, type 1C, 616081*
EXOSC9	*99,3*	*94,7*	*100*	*99,9*	*Pontocerebellar hypoplasia, type 1D, 618065*
EXPH5	*100*	*99,9*	*100*	*100*	*Epidermolysis bullosa, nonspecific, autosomal recessive, 615028*
EXT1	*99,6*	*97,1*	*100*	*100*	*Exostoses, multiple, type 1, 133700* *Chondrosarcoma, 215300*
EXT2	*99,9*	*99*	*100*	*100*	*Seizures, scoliosis, and macrocephaly syndrome, 616682* *Exostoses, multiple, type 2, 133701*
EXTL3	*100*	*100*	*100*	*100*	*Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425*
EYA1	*99,9*	*99,5*	*100*	*100*	*Branchiootic syndrome 1, 602588* *Branchiootorenal syndrome 1, with or without cataracts, 113650* *Anterior segment anomalies with or without cataract, 602588* *?Otofaciocervical syndrome, 166780*
EYA4	*99,9*	*99,9*	*100*	*100*	*?Cardiomyopathy, dilated, 1J, 605362* *Deafness, autosomal dominant 10, 601316*
EYS	*99,5*	*97,5*	*100*	*100*	*Retinitis pigmentosa 25, 602772*
EZH2	*99,7*	*98*	*100*	*100*	*Weaver syndrome, 277590*
F10	*99,8*	*98,4*	*100*	*100*	*Factor X deficiency, 227600*
F11	*100*	*100*	*100*	*100*	*Factor XI deficiency, autosomal dominant, 612416* *Factor XI deficiency, autosomal recessive, 612416*
F12	*100*	*98,6*	*100*	*100*	*Angioedema, hereditary, 3, 610618* *Factor XII deficiency, 234000*
F13A1	*100*	*100*	*100*	*100*	*Factor XIIIA deficiency, 613225*
F13B	*98,3*	*92,8*	*100*	*99,9*	*Factor XIIIB deficiency, 613235*
F2	*99,9*	*97,8*	*100*	*100*	*Hypoprothrombinemia, 613679* *Dysprothrombinemia, 613679* *Thrombophilia due to thrombin defect, 188050*
F2RL3	*100*	*100*	*100*	*100*	*No OMIM disease ID*

F5	*99,9*	*98,4*	*100*	*100*	*Thrombophilia due to activated protein C resistance, 188055* *Factor V deficiency, 227400*
F7	*100*	*100*	*100*	*100*	*Factor VII deficiency, 227500*
F8	*97,2*	*95,7*	*100*	*99,9*	*Hemophilia A, 306700*
F9	*99,7*	*98,3*	*100*	*97,9*	*Thrombophilia, X-linked, due to factor IX defect, 300807* *Hemophilia B, 306900*
FA2H	*92,4*	*82,6*	*100*	*100*	*Spastic paraplegia 35, autosomal recessive, 612319*
FAAH	*94,3*	*90,3*	*100*	*100*	*No OMIM disease ID*
FAAP24	*98,3*	*94,8*	*100*	*100*	*No OMIM disease ID*
FADD	*100*	*100*	*100*	*100*	*Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations, 613759*
FAH	*100*	*99,5*	*100*	*99,9*	*Tyrosinemia, type I, 276700*
FAM111A	*100*	*99,4*	*100*	*100*	*Kenny-Caffey syndrome, type 2, 127000* *Gracile bone dysplasia, 602361*
FAM111B	*99,9*	*99,7*	*100*	*100*	*Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704*
FAM126A	*99,5*	*99,4*	*100*	*100*	*Leukodystrophy, hypomyelinating, 5, 610532*
FAM149B1	*98,2*	*94,3*	*100*	*100*	*Joubert syndrome 36, 618763*
FAM161A	*99,8*	*99,5*	*100*	*100*	*Retinitis pigmentosa 28, 606068*
FAM20A	*99,6*	*94,4*	*100*	*100*	*Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690*
FAM20B	*100*	*99,4*	*100*	*100*	*No OMIM disease ID*
FAM20C	*100*	*100*	*100*	*99,7*	*Raine syndrome, 259775*
FAM50A	*99,8*	*97*	*100*	*99,6*	*Intellectual developmental disorder, X-linked, syndromic, Armfield type, 300261*
FAM83G	*100*	*100*	*100*	*100*	*No OMIM disease ID*
FAM83H	*85,5*	*83,1*	*100*	*100*	*Amelogenesis imperfecta, type IIIA, 130900*
FAN1	*100*	*99,8*	*100*	*100*	*Interstitial nephritis, karyomegalic, 614817*
FANCA	*99,9*	*98,7*	*100*	*100*	*Fanconi anemia, complementation group A, 227650*
FANCB	*98*	*91,7*	*100*	*99,6*	*Fanconi anemia, complementation group B, 300514*
FANCC	*96,9*	*95,7*	*97,3*	*97,3*	*Fanconi anemia, complementation group C, 227645*
FANCD2	*98,7*	*95,9*	*98,8*	*98,8*	*Fanconi anemia, complementation group D2, 227646*
FANCE	*90,7*	*85,5*	*100*	*100*	*Fanconi anemia, complementation group E, 600901*
FANCF	*100*	*100*	*100*	*100*	*Fanconi anemia, complementation group F, 603467*

FANCG	*100*	*99,9*	*100*	*100*	*Fanconi anemia, complementation group G, 614082*
FANCI	*99,8*	*98,6*	*100*	*100*	*Fanconi anemia, complementation group I, 609053*
FANCL	*99,4*	*97,6*	*100*	*100*	*Fanconi anemia, complementation group L, 614083*
FANCM	*98,9*	*96,3*	*100*	*100*	*?Premature ovarian failure 15, 618096* *Spermatogenic failure 28, 618086*
FAR1	*97,4*	*94*	*100*	*100*	*Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154* *Cataracts, spastic paraparesis, and speech delay, 619338*
FARS2	*100*	*100*	*100*	*100*	*Combined oxidative phosphorylation deficiency 14, 614946* *Spastic paraplegia 77, autosomal recessive, 617046*
FARSA	*95,6*	*91,2*	*100*	*100*	*?Rajab interstitial lung disease with brain calcifications 2, 619013*
FARSB	*98*	*92,9*	*100*	*100*	*Rajab interstitial lung disease with brain calcifications 1, 613658*
FAS	*100*	*99,6*	*100*	*100*	*Autoimmune lymphoproliferative syndrome, type IA, 601859* *Squamous cell carcinoma, burn scar-related, somatic,*
FASLG	*100*	*99,1*	*100*	*100*	*Autoimmune lymphoproliferative syndrome, type IB, 601859*
FASTKD2	*99,6*	*98,6*	*100*	*100*	*Combined oxidative phosphorylation deficiency 44, 618855*
FAT1	*100*	*99,9*	*100*	*100*	*No OMIM disease ID*
FAT2	*100*	*99,6*	*100*	*100*	*Spinocerebellar ataxia 45, 617769*
FAT4	*100*	*100*	*100*	*100*	*Van Maldergem syndrome 2, 615546* *Hennekam lymphangiectasia-lymphedema syndrome 2, 616006*
FBLN1	*99,7*	*96,6*	*100*	*99,9*	*No OMIM disease ID*
FBLN5	*91,8*	*91,7*	*91,8*	*91,8*	*Cutis laxa, autosomal recessive, type IA, 219100* *Macular degeneration, age-related, 3, 608895* *Neuropathy, hereditary, with or without age-related macular degeneration, 608895* *?Cutis laxa, autosomal dominant 2, 614434*
FBN1	*100*	*99,7*	*100*	*100*	*Geleophysic dysplasia 2, 614185* *Weill-Marchesani syndrome 2, dominant, 608328* *Ectopia lentis, familial, 129600* *MASS syndrome, 604308* *Marfan lipodystrophy syndrome, 616914* *Acromicric dysplasia, 102370* *Marfan syndrome, 154700* *Stiff skin syndrome, 184900*
FBN2	*100*	*99,8*	*100*	*100*	*Macular degeneration, early-onset, 616118* *Contractural arachnodactyly, congenital, 121050*

FBP1	*93,6*	*91,3*	*93,7*	*93,7*	*Fructose-1,6-bisphosphatase deficiency, 229700*
FBRSL1	*56,4*	*50,8*	*96,5*	*92,4*	*No OMIM disease ID*
FBXL3	*100*	*100*	*100*	*100*	*Intellectual developmental disorder with short stature, facial anomalies, and speech defects, 606220*
FBXL4	*100*	*100*	*100*	*100*	*Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471*
FBXO11	*97,3*	*91,2*	*100*	*100*	*Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities, 618089*
FBXO31	*97*	*94,2*	*100*	*100*	*?Mental retardation, autosomal recessive 45, 615979*
FBXO32	*100*	*100*	*100*	*100*	*No OMIM disease ID*
FBXO38	*99,8*	*98,9*	*100*	*100*	*Neuronopathy, distal hereditary motor, type IID, 615575*
FBXO7	*99,8*	*98,3*	*100*	*100*	*Parkinson disease 15, autosomal recessive, 260300*
FBXW11	*99,5*	*95,9*	*100*	*100*	*Neurodevelopmental, jaw, eye, and digital syndrome, 618914*
FBXW4	*82,2*	*79,8*	*88,8*	*84,2*	*No OMIM disease ID*
FBXW7	*99,9*	*98,2*	*100*	*99,9*	*No OMIM disease ID*
FCGR1A	*46,5*	*41,8*	*100*	*99,9*	*No OMIM disease ID*
FCGR2A	*100*	*100*	*100*	*100*	*No OMIM disease ID*
FCGR2B	*99,5*	*96,1*	*100*	*100*	*No OMIM disease ID*
FCGR2C	*98,2*	*98,1*	*99,4*	*99,3*	*No OMIM disease ID*
FCGR3A	*98,6*	*96,8*	*100*	*99,9*	*Immunodeficiency 20, 615707*
FCGR3B	*99,1*	*97,3*	*99,5*	*97,9*	*No OMIM disease ID*
FCHO1	*99,3*	*97,5*	*100*	*100*	*Immunodeficiency 76, 619164*
FCN3	*100*	*99,3*	*100*	*100*	*Immunodeficiency due to ficolin 3 deficiency, 613860*
FCSK	*98*	*96,1*	*100*	*100*	*Congenital disorder of glycosylation with defective fucosylation 2, 618324*
FDFT1	*98,5*	*96,7*	*100*	*100*	*Squalene synthase deficiency, 618156*
FDPS	*97,4*	*90,8*	*100*	*100*	*Porokeratosis 9, multiple types, 616631*
FDX2	*100*	*100*	*100*	*100*	*Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900*
FDXR	*100*	*98,6*	*100*	*100*	*Auditory neuropathy and optic atrophy, 617717*
FECH	*99,9*	*99,8*	*100*	*100*	*Protoporphyria, erythropoietic, 1, 177000*
FERMT1	*99,1*	*95,8*	*100*	*99,9*	*Kindler syndrome, 173650*
FERMT3	*100*	*100*	*100*	*100*	*Leukocyte adhesion deficiency, type III, 612840*
FEZF1	*100*	*100*	*100*	*100*	*Hypogonadotropic hypogonadism 22, with or without anosmia, 616030*

FGA	*99,1*	*96,8*	*100*	*100*	*Hypodysfibrinogenemia, congenital, 616004* *Dysfibrinogenemia, congenital, 616004* *Amyloidosis, familial visceral, 105200* *Afibrinogenemia, congenital, 202400*
FGB	*99,8*	*98,8*	*100*	*100*	*Hypofibrinogenemia, congenital, 202400* *Dysfibrinogenemia, congenital, 616004* *Afibrinogenemia, congenital, 202400*
FGD1	*97,2*	*91,2*	*100*	*100*	*Mental retardation, X-linked syndromic 16, 305400* *Aarskog-Scott syndrome, 305400*
FGD4	*99,8*	*98,8*	*100*	*100*	*Charcot-Marie-Tooth disease, type 4H, 609311*
FGF10	*100*	*99,5*	*100*	*100*	*Aplasia of lacrimal and salivary glands, 180920* *LADD syndrome, 149730*
FGF12	*100*	*99,1*	*100*	*100*	*Developmental and epileptic encephalopathy 47, 617166*
FGF13	*99,1*	*95,1*	*100*	*100*	*Developmental and epileptic encephalopathy 90, 301058*
FGF14	*100*	*100*	*100*	*100*	*Spinocerebellar ataxia 27, 609307*
FGF16	*100*	*98,7*	*100*	*99,8*	*Metacarpal 4-5 fusion, 309630*
FGF17	*100*	*100*	*100*	*100*	*Hypogonadotropic hypogonadism 20 with or without anosmia, 615270*
FGF20	*95,8*	*88,4*	*100*	*100*	*?Renal hypodysplasia/aplasia 2, 615721*
FGF23	*99,4*	*96,7*	*100*	*100*	*Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993* *Hypophosphatemic rickets, autosomal dominant, 193100*
FGF3	*100*	*98,7*	*100*	*100*	*Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706*
FGF5	*100*	*99,7*	*100*	*100*	*Trichomegaly, 190330*
FGF8	*97,1*	*87,2*	*100*	*99,9*	*Hypogonadotropic hypogonadism 6 with or without anosmia, 612702*
FGF9	*100*	*100*	*100*	*100*	*Multiple synostoses syndrome 3, 612961*
FGFR1	*100*	*99,3*	*100*	*100*	*Pfeiffer syndrome, 101600* *Hypogonadotropic hypogonadism 2 with or without anosmia, 147950* *Jackson-Weiss syndrome, 123150* *Hartsfield syndrome, 615465* *Trigonocephaly 1, 190440* *Osteoglophonic dysplasia, 166250* *Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001*
FGFR2	*97,6*	*97*	*100*	*100*	*Bent bone dysplasia syndrome, 614592* *LADD syndrome, 149730* *Scaphocephaly, maxillary retrusion, and mental retardation, 609579*

					Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 *Jackson-Weiss syndrome, 123150* *Gastric cancer, somatic, 613659* *Craniofacial-skeletal-dermatologic dysplasia, 101600* *Apert syndrome, 101200* *Pfeiffer syndrome, 101600* *Beare-Stevenson cutis gyrata syndrome, 123790* *Crouzon syndrome, 123500* *Saethre-Chotzen syndrome, 101400* *Scaphocephaly and Axenfeld-Rieger anomaly,* *Craniosynostosis, nonspecific,*
FGFR3	*99,8*	*98*	*100*	*100*	*Muenke syndrome, 602849* *SADDAN, 616482* *Hypochondroplasia, 146000* *LADD syndrome, 149730* *Thanatophoric dysplasia, type II, 187601* *Nevus, epidermal, somatic, 162900* *CATSHL syndrome, 610474* *Thanatophoric dysplasia, type I, 187600* *Spermatocytic seminoma, somatic, 273300* *Bladder cancer, somatic, 109800* *Achondroplasia, 100800* *Cervical cancer, somatic, 603956* *Colorectal cancer, somatic, 114500* *Crouzon syndrome with acanthosis nigricans, 612247*
FGG	*99,4*	*97,5*	*100*	*100*	*Dysfibrinogenemia, congenital, 616004* *Hypodysfibrinogenemia, 616004* *Hypofibrinogenemia, congenital, 202400* *Afibrinogenemia, congenital, 202400*
FH	*93,2*	*87,2*	*100*	*100*	*Leiomyomatosis and renal cell cancer, 150800* *Fumarase deficiency, 606812*
FHL1	*99,4*	*93,8*	*100*	*100*	*Myopathy, X-linked, with postural muscle atrophy, 300696* *Emery-Dreifuss muscular dystrophy 6, X-linked, 300696* *?Uruguay faciocardiomusculoskeletal syndrome, 300280* *Scapuloperoneal myopathy, X-linked dominant, 300695* *Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718* *Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717*
FHL2	*99,8*	*98,1*	*100*	*100*	*No OMIM disease ID*

FHOD3	*100*	*99,4*	*100*	*100*	*Cardiomyopathy, familial hypertrophic, 28, 619402*
FIBP	*100*	*99,8*	*100*	*100*	*Thauvin-Robinet-Faivre syndrome, 617107*
FIG4	*99,8*	*99,7*	*100*	*100*	*Yunis-Varon syndrome, 216340* *?Polymicrogyria, bilateral temporooccipital, 612691* *Amyotrophic lateral sclerosis 11, 612577* *Charcot-Marie-Tooth disease, type 4J, 611228*
FIGLA	*99,8*	*96,6*	*100*	*100*	*Premature ovarian failure 6, 612310*
FIGN	*100*	*100*	*100*	*100*	*No OMIM disease ID*
FITM2	*100*	*100*	*100*	*100*	*Siddiqi syndrome, 618635*
FKBP10	*98,9*	*97,3*	*100*	*100*	*Osteogenesis imperfecta, type XI, 610968* *Bruck syndrome 1, 259450*
FKBP14	*99,8*	*98,7*	*100*	*100*	*Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557*
FKRP	*100*	*100*	*100*	*100*	*Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612* *Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155* *Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153*
FKTN	*99,8*	*95,2*	*100*	*100*	*Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588* *Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800* *Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152* *Cardiomyopathy, dilated, 1X, 611615*
FLAD1	*100*	*99,7*	*100*	*100*	*Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100*
FLCN	*100*	*100*	*100*	*100*	*Birt-Hogg-Dube syndrome, 135150* *Colorectal cancer, somatic, 114500* *Pneumothorax, primary spontaneous, 173600* *Renal carcinoma, chromophobe, somatic, 144700*
FLG	*99,9*	*99,9*	*100*	*100*	*Ichthyosis vulgaris, 146700*
FLG2	*100*	*100*	*99,9*	*99,9*	*Peeling skin syndrome 6, 618084*
FLI1	*99,4*	*97,9*	*100*	*100*	*Bleeding disorder, platelet-type, 21, 617443*
FLNA	*100*	*99,9*	*100*	*100*	*Otopalatodigital syndrome, type II, 304120* *Intestinal pseudoobstruction, neuronal, 300048* *Cardiac valvular dysplasia, X-linked, 314400* *?FG syndrome 2, 300321* *Melnick-Needles syndrome, 309350* *Terminal osseous dysplasia, 300244* *Congenital short bowel syndrome, 300048* *Otopalatodigital syndrome, type I, 311300*

					Heterotopia, periventricular, 1, 300049 *Frontometaphyseal dysplasia 1, 305620*
FLNB	*99,4*	*98,7*	*100*	*100*	*Larsen syndrome, 150250* *Atelosteogenesis, type I, 108720* *Atelosteogenesis, type III, 108721* *Spondylocarpotarsal synostosis syndrome, 272460* *Boomerang dysplasia, 112310*
FLNC	*100*	*99,4*	*100*	*100*	*Cardiomyopathy, familial hypertrophic, 26, 617047* *Cardiomyopathy, familial restrictive 5, 617047* *Myopathy, distal, 4, 614065* *Myopathy, myofibrillar, 5, 609524*
FLRT3	*100*	*100*	*100*	*100*	*Hypogonadotropic hypogonadism 21 with anosmia, 615271*
FLT3	*99,8*	*98,6*	*100*	*100*	*Leukemia, acute lymphoblastic, somatic, 613065* *Leukemia, acute myeloid, reduced survival in, somatic, 601626* *Leukemia, acute myeloid, somatic, 601626*
FLT4	*99,2*	*98,9*	*100*	*100*	*Hemangioma, capillary infantile, somatic, 602089* *Lymphatic malformation 1, 153100* *Congenital heart defects, multiple types, 7, 618780*
FLVCR1	*99,7*	*98,3*	*100*	*99,9*	*Ataxia, posterior column, with retinitis pigmentosa, 609033*
FLVCR2	*100*	*100*	*100*	*100*	*Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790*
FMN1	*97,1*	*95,7*	*100*	*100*	*No OMIM disease ID*
FMN2	*85,5*	*82,8*	*100*	*100*	*Mental retardation, autosomal recessive 47, 616193*
FMO3	*99,9*	*99,7*	*100*	*100*	*Trimethylaminuria, 602079*
FMR1	*95,2*	*90,1*	*100*	*99,9*	*Fragile X tremor/ataxia syndrome, 300623* *Fragile X syndrome, 300624* *Premature ovarian failure 1, 311360*
FN1	*99,9*	*98,7*	*100*	*100*	*Spondylometaphyseal dysplasia, corner fracture type, 184255* *Glomerulopathy with fibronectin deposits 2, 601894*
FNIP1	*99,9*	*99,9*	*100*	*100*	*No OMIM disease ID*
FOLR1	*100*	*99,9*	*100*	*100*	*Neurodegeneration due to cerebral folate transport deficiency, 613068*
FOXC1	*98,7*	*93,7*	*99,6*	*97,3*	*Axenfeld-Rieger syndrome, type 3, 602482* *Anterior segment dysgenesis 3, multiple subtypes, 601631*
FOXC2	*100*	*98,1*	*100*	*99,6*	*Lymphedema-distichiasis syndrome, 153400* *Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400*
FOXD4	*22,4*	*5*	*100*	*100*	*No OMIM disease ID*

FOXE1	*97,9*	*82,2*	*100*	*98,9*	*Bamforth-Lazarus syndrome, 241850*
FOXE3	*87,6*	*79*	*95,8*	*89,2*	*Anterior segment dysgenesis 2, multiple subtypes, 610256* *Cataract 34, multiple types, 612968*
FOXF1	*100*	*99,6*	*100*	*100*	*Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380*
FOXF2	*94,6*	*89,3*	*95,6*	*93,9*	*No OMIM disease ID*
FOXG1	*87,2*	*79,4*	*98,3*	*94,8*	*Rett syndrome, congenital variant, 613454*
FOXH1	*100*	*98,7*	*100*	*100*	*No OMIM disease ID*
FOXI1	*100*	*100*	*100*	*100*	*Enlarged vestibular aqueduct, 600791*
FOXJ1	*100*	*98,9*	*100*	*100*	*Ciliary dyskinesia, primary, 43, 618699*
FOXL1	*97*	*88,8*	*100*	*100*	*No OMIM disease ID*
FOXL2	*99,4*	*94,7*	*99,9*	*99*	*Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100* *Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100* *Premature ovarian failure 3, 608996*
FOXN1	*100*	*99,1*	*100*	*100*	*T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806* *T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705*
FOXO1	*100*	*97,3*	*99,4*	*97,4*	*Rhabdomyosarcoma, alveolar, 268220*
FOXP1	*99,9*	*99,1*	*100*	*100*	*Mental retardation with language impairment and with or without autistic features, 613670*
FOXP2	*99,3*	*98,7*	*100*	*100*	*Speech-language disorder-1, 602081*
FOXP3	*98,5*	*94,8*	*100*	*100*	*Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790*
FOXRED1	*100*	*99,6*	*100*	*100*	*Mitochondrial complex I deficiency, nuclear type 19, 618241*
FPR1	*100*	*100*	*100*	*100*	*No OMIM disease ID*
FRAS1	*100*	*99,2*	*100*	*100*	*Fraser syndrome 1, 219000*
FREM1	*99,8*	*98,4*	*100*	*100*	*Manitoba oculotrichoanal syndrome, 248450* *Bifid nose with or without anorectal and renal anomalies, 608980* *Trigonocephaly 2, 614485*
FREM2	*99,8*	*98,7*	*100*	*100*	*Fraser syndrome 2, 617666* *Cryptophthalmos, unilateral or bilateral, isolated, 123570*
FRMD4A	*91,3*	*89,4*	*96,6*	*96,6*	*?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819*
FRMD7	*99,9*	*98,1*	*100*	*99,3*	*Nystagmus, infantile periodic alternating, X-linked, 310700* *Nystagmus 1, congenital, X-linked, 310700*
FRMPD4	*97*	*94,2*	*98,3*	*98,2*	*Intellectual developmental disorder, X-linked 104, 300983*
FRRS1L	*82,5*	*73,7*	*98,8*	*93,9*	*Developmental and epileptic encephalopathy 37, 616981*

FSCN2	*100*	*100*	*100*	*100*	*Retinitis pigmentosa 30, 607921*
FSHB	*100*	*100*	*100*	*100*	*Hypogonadotropic hypogonadism 24 without anosmia, 229070*
FSHR	*99,2*	*97*	*100*	*100*	*Ovarian response to FSH stimulation, 276400* *Ovarian hyperstimulation syndrome, 608115* *Ovarian dysgenesis 1, 233300*
FSIP2	*99,3*	*98,3*	*100*	*100*	*Spermatogenic failure 34, 618153*
FTCD	*97,7*	*93,2*	*100*	*100*	*Glutamate formiminotransferase deficiency, 229100*
FTH1	*91,9*	*73,8*	*100*	*100*	*?Hemochromatosis, type 5, 615517*
FTL	*98,6*	*88,5*	*100*	*100*	*Hyperferritinemia-cataract syndrome, 600886* *L-ferritin deficiency, dominant and recessive, 615604* *Neurodegeneration with brain iron accumulation 3, 606159*
FTO	*83,8*	*83,7*	*94,2*	*94,2*	*Growth retardation, developmental delay, facial dysmorphism, 612938*
FTSJ1	*98,3*	*93,8*	*100*	*100*	*Intellectual developmental disorder, X-linked 9, 309549*
FUCA1	*100*	*100*	*100*	*100*	*Fucosidosis, 230000*
FURIN	*100*	*100*	*100*	*100*	*No OMIM disease ID*
FUS	*98,4*	*95,3*	*100*	*100*	*Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030* *Essential tremor, hereditary, 4, 614782*
FUT2	*100*	*100*	*100*	*100*	*No OMIM disease ID*
FUT6	*100*	*100*	*100*	*100*	*No OMIM disease ID*
FUT8	*99,8*	*98,9*	*100*	*100*	*Congenital disorder of glycosylation with defective fucosylation 1, 618005*
FUZ	*100*	*100*	*100*	*100*	*No OMIM disease ID*
FXN	*98,3*	*84,7*	*100*	*100*	*Friedreich ataxia with retained reflexes, 229300* *Friedreich ataxia, 229300*
FXR1	*99,4*	*96,8*	*100*	*99,9*	*?Myopathy, congenital, with respiratory insufficiency and bone fractures, 618822* *?Myopathy, congenital proximal, with minicore lesions, 618823*
FXYD2	*100*	*100*	*100*	*100*	*Hypomagnesemia 2, renal, 154020*
FYB1	*99,4*	*95,9*	*100*	*100*	*Thrombocytopenia 3, 273900*
FYCO1	*100*	*100*	*100*	*100*	*Cataract 18, autosomal recessive, 610019*
FZD2	*100*	*97,8*	*100*	*100*	*Omodysplasia 2, 164745*
FZD4	*100*	*100*	*100*	*100*	*Retinopathy of prematurity, 133780* *Exudative vitreoretinopathy 1, 133780*
FZD6	*100*	*100*	*100*	*100*	*Nail disorder, nonsyndromic congenital, 1, 161050*

G6PC	*100*	*100*	*100*	*100*	*Glycogen storage disease Ia, 232200*
G6PC3	*100*	*99,9*	*100*	*100*	*Dursun syndrome, 612541* *Neutropenia, severe congenital 4, autosomal recessive, 612541*
G6PD	*99,1*	*97,4*	*100*	*100*	*Hemolytic anemia, G6PD deficient (favism), 300908*
GAA	*100*	*99,9*	*100*	*100*	*Glycogen storage disease II, 232300*
GAB1	*99,9*	*98,9*	*100*	*100*	*?Deafness, autosomal recessive 26, 605428*
GABBR2	*96,1*	*90,9*	*98,8*	*98*	*Developmental and epileptic encephalopathy 59, 617904* *Neurodevelopmental disorder with poor language and loss of hand skills, 617903*
GABRA1	*100*	*100*	*100*	*100*	*Developmental and epileptic encephalopathy 19, 615744*
GABRA2	*99,5*	*96,9*	*100*	*100*	*Developmental and epileptic encephalopathy 78, 618557*
GABRA3	*98*	*94*	*100*	*99,3*	*No OMIM disease ID*
GABRA5	*100*	*99,4*	*100*	*100*	*Developmental and epileptic encephalopathy 79, 618559*
GABRB1	*100*	*100*	*100*	*100*	*Developmental and epileptic encephalopathy 45, 617153*
GABRB2	*100*	*100*	*100*	*100*	*Developmental and epileptic encephalopathy 92, 617829*
GABRB3	*99,8*	*98,1*	*100*	*100*	*Developmental and epileptic encephalopathy 43, 617113*
GABRG2	*89,9*	*88,4*	*93*	*93*	*Developmental and epileptic encephalopathy 74, 618396* *Febrile seizures, familial, 8, 607681* *Generalized epilepsy with febrile seizures plus, type 3, 607681*
GAD1	*100*	*99,3*	*100*	*100*	*Developmental and epileptic encephalopathy 89, 619124*
GAL	*100*	*99,8*	*100*	*100*	*?Epilepsy, familial temporal lobe, 8, 616461*
GALC	*99,7*	*97,6*	*100*	*100*	*Krabbe disease, 245200*
GALE	*100*	*100*	*100*	*100*	*Galactose epimerase deficiency, 230350*
GALK1	*100*	*99,2*	*100*	*100*	*Galactokinase deficiency with cataracts, 230200*
GALM	*100*	*99,5*	*100*	*100*	*Galactosemia IV, 618881*
GALNS	*100*	*99,3*	*100*	*100*	*Mucopolysaccharidosis IVA, 253000*
GALNT12	*86,4*	*82,6*	*97,9*	*94,7*	*No OMIM disease ID*
GALNT2	*99,8*	*97,1*	*100*	*100*	*Congenital disorder of glycosylation, type IIt, 618885*
GALNT3	*99,8*	*98,7*	*100*	*100*	*Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900*
GALNTL5	*99,8*	*99,4*	*100*	*99,8*	*No OMIM disease ID*
GALT	*100*	*99,6*	*100*	*100*	*Galactosemia, 230400*

GAMT	*95*	*82,7*	*100*	*100*	*Cerebral creatine deficiency syndrome 2, 612736*
GAN	*99,9*	*98,8*	*100*	*100*	*Giant axonal neuropathy-1, 256850*
GANAB	*99,8*	*97,8*	*100*	*100*	*Polycystic kidney disease 3, 600666*
GAPVD1	*99,9*	*98,9*	*100*	*100*	*No OMIM disease ID*
GARS1	*99,9*	*99,4*	*100*	*100*	*Spinal muscular atrophy, infantile, James type, 619042* *Neuronopathy, distal hereditary motor, type VA, 600794* *Charcot-Marie-Tooth disease, type 2D, 601472*
GAS2	*100*	*100*	*100*	*100*	*No OMIM disease ID*
GAS2L2	*100*	*99,9*	*100*	*100*	*?Ciliary dyskinesia, primary, 41, 618449*
GAS8	*99,9*	*99,6*	*100*	*100*	*Ciliary dyskinesia, primary, 33, 616726*
GATA1	*99,9*	*98,5*	*100*	*100*	*Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685* *Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367* *Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835* *Thrombocytopenia with beta-thalassemia, X-linked, 314050*
GATA2	*99,8*	*97*	*100*	*100*	*Emberger syndrome, 614038* *Immunodeficiency 21, 614172*
GATA3	*100*	*100*	*100*	*100*	*Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255*
GATA4	*87,4*	*78,5*	*100*	*100*	*Tetralogy of Fallot, 187500* *Atrial septal defect 2, 607941* *Ventricular septal defect 1, 614429* *Atrioventricular septal defect 4, 614430* *?Testicular anomalies with or without congenital heart disease, 615542*
GATA5	*99,9*	*95,5*	*100*	*100*	*Congenital heart defects, multiple types, 5, 617912*
GATA6	*91,5*	*84,5*	*99,7*	*98,4*	*Atrial septal defect 9, 614475* *Persistent truncus arteriosus, 217095* *Pancreatic agenesis and congenital heart defects, 600001* *Atrioventricular septal defect 5, 614474* *Tetralogy of Fallot, 187500*
GATAD1	*100*	*98,7*	*99,9*	*98,4*	*?Cardiomyopathy, dilated, 2B, 614672*
GATAD2B	*100*	*99,1*	*100*	*100*	*GAND syndrome, 615074*
GATB	*100*	*99*	*100*	*100*	*?Combined oxidative phosphorylation deficiency 41, 618838*
GATC	*100*	*100*	*100*	*100*	*Combined oxidative phosphorylation deficiency 42, 618839*
GATM	*100*	*100*	*100*	*100*	*Cerebral creatine deficiency syndrome 3, 612718* *Fanconi renotubular syndrome 1, 134600*

GBA	*100*	*100*	*100*	*100*	*Gaucher disease, type II, 230900* *Gaucher disease, type IIIC, 231005* *Gaucher disease, type III, 231000* *Gaucher disease, type I, 230800* *Gaucher disease, perinatal lethal, 608013*
GBA2	*100*	*99,5*	*100*	*100*	*Spastic paraplegia 46, autosomal recessive, 614409*
GBE1	*99,9*	*99,7*	*100*	*100*	*Glycogen storage disease IV, 232500* *Polyglucosan body disease, adult form, 263570*
GBF1	*98,3*	*97,7*	*100*	*100*	*No OMIM disease ID*
GCDH	*100*	*99,2*	*100*	*100*	*Glutaricaciduria, type I, 231670*
GCGR	*100*	*100*	*100*	*100*	*Mahvash disease, 619290*
GCH1	*99,9*	*97,3*	*100*	*100*	*Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230* *Hyperphenylalaninemia, BH4-deficient, B, 233910*
GCK	*95,4*	*95,4*	*95,2*	*92,6*	*MODY, type II, 125851* *Diabetes mellitus, permanent neonatal 1, 606176* *Hyperinsulinemic hypoglycemia, familial, 3, 602485* *Diabetes mellitus, noninsulin-dependent, late onset, 125853*
GCLC	*99,4*	*97,1*	*100*	*99,9*	*Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450*
GCLM	*99,5*	*95,4*	*100*	*100*	*No OMIM disease ID*
GCM2	*100*	*100*	*100*	*100*	*Hypoparathyroidism, familial isolated 2, 618883* *Hyperparathyroidism 4, 617343*
GCNT2	*99,5*	*99,5*	*100*	*100*	*Adult i phenotype without cataract, 110800* *Cataract 13 with adult i phenotype, 116700*
GCSH	*75,7*	*64,4*	*100*	*100*	*?Glycine encephalopathy, 605899*
GDAP1	*99,7*	*98,4*	*100*	*100*	*Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706* *Charcot-Marie-Tooth disease, recessive intermediate, A, 608340* *Charcot-Marie-Tooth disease, axonal, type 2K, 607831* *Charcot-Marie-Tooth disease, type 4A, 214400*
GDAP2	*99,7*	*99*	*100*	*100*	*Spinocerebellar ataxia, autosomal recessive 27, 618369*
GDF1	*80,8*	*59*	*98,5*	*92*	*Congenital heart defects, multiple types, 6, 613854* *Right atrial isomerism (Ivemark), 208530*
GDF11	*96,9*	*92,6*	*97,7*	*92,4*	*?Vertebral hypersegmentation and orofacial anomalies, 619122*
GDF2	*100*	*100*	*100*	*100*	*Telangiectasia, hereditary hemorrhagic, type 5, 615506*

GDF3	*100*	*100*	*100*	*100*	*Klippel-Feil syndrome 3, autosomal dominant, 613702* *Microphthalmia with coloboma 6, 613703* *Microphthalmia, isolated 7, 613704*
GDF5	*100*	*100*	*100*	*100*	*Du Pan syndrome, 228900* *Multiple synostoses syndrome 2, 610017* *Symphalangism, proximal, 1B, 615298* *?Acromesomelic dysplasia, Hunter-Thompson type, 201250* *Brachydactyly, type A2, 112600* *Brachydactyly, type C, 113100* *Chondrodysplasia, Grebe type, 200700* *Brachydactyly, type A1, C, 615072*
GDF6	*100*	*100*	*100*	*99,6*	*Microphthalmia with coloboma 6, digenic, 613703* *Microphthalmia, isolated 4, 613094* *Leber congenital amaurosis 17, 615360* *Multiple synostoses syndrome 4, 617898* *Klippel-Feil syndrome 1, autosomal dominant, 118100*
GDF9	*100*	*100*	*100*	*100*	*?Premature ovarian failure 14, 618014*
GDI1	*99,8*	*98,6*	*100*	*100*	*Intellectual developmental disorder, X-linked 41, 300849*
GDNF	*100*	*100*	*100*	*100*	*No OMIM disease ID*
GDPD1	*99,3*	*96,2*	*100*	*100*	*No OMIM disease ID*
GEMIN4	*99,9*	*99,2*	*100*	*100*	*Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities, 617913*
GEMIN5	*99,9*	*98,7*	*100*	*100*	*Neurodevelopmental disorder with cerebellar atrophy and motor dysfunction, 619333*
GFAP	*91,7*	*89,5*	*100*	*100*	*Alexander disease, 203450*
GFER	*99,8*	*97,6*	*100*	*100*	*Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076*
GFI1	*100*	*99,9*	*100*	*100*	*?Neutropenia, nonimmune chronic idiopathic, of adults, 607847* *Neutropenia, severe congenital 2, autosomal dominant, 613107*
GFI1B	*99*	*97,3*	*100*	*100*	*Bleeding disorder, platelet-type, 17, 187900*
GFM1	*99,7*	*98,7*	*100*	*100*	*Combined oxidative phosphorylation deficiency 1, 609060*
GFM2	*98,1*	*93,7*	*100*	*100*	*Combined oxidative phosphorylation deficiency 39, 618397*
GFPT1	*99,9*	*99,4*	*100*	*100*	*Myasthenia, congenital, 12, with tubular aggregates, 610542*
GFRA1	*100*	*99,9*	*100*	*100*	*No OMIM disease ID*
GGCX	*100*	*99,6*	*100*	*100*	*Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450* *Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842*
GGPS1	*99,8*	*99,8*	*100*	*100*	*Muscular dystrophy, congenital hearing loss, and ovarian insufficiency syndrome, 619518*

GGT1	*19,7*	*18,2*	*100*	*100*	*?Glutathioninuria, 231950*
GH1	*100*	*100*	*100*	*100*	*Kowarski syndrome, 262650* *Growth hormone deficiency, isolated, type II, 173100* *Growth hormone deficiency, isolated, type IB, 612781* *Growth hormone deficiency, isolated, type IA, 262400*
GHR	*99,5*	*99,5*	*99,5*	*99,5*	*Laron dwarfism, 262500* *Increased responsiveness to growth hormone, 604271* *Growth hormone insensitivity, partial, 604271*
GHRHR	*96,5*	*96,4*	*100*	*99,9*	*Growth hormone deficiency, isolated, type IV, 618157*
GHSR	*98,7*	*95,6*	*100*	*100*	*Growth hormone deficiency, isolated partial, 615925*
GIGYF1	*99,1*	*95,1*	*100*	*100*	*No OMIM disease ID*
GIMAP5	*100*	*100*	*100*	*100*	*Portal hypertension, noncirrhotic, 2, 619463*
GINS1	*98,4*	*93,4*	*100*	*100*	*Immunodeficiency 55, 617827*
GINS2	*100*	*97,5*	*100*	*100*	*No OMIM disease ID*
GIPC1	*95,1*	*86,3*	*100*	*100*	*Oculopharyngodistal myopathy 2, 618940*
GIPC3	*24,9*	*23,2*	*100*	*99,6*	*Deafness, autosomal recessive 15, 601869*
GJA1	*100*	*100*	*100*	*100*	*Erythrokeratodermia variabilis et progressiva 3, 617525* *Craniometaphyseal dysplasia, autosomal recessive, 218400* *Oculodentodigital dysplasia, 164200* *Hypoplastic left heart syndrome 1, 241550* *Palmoplantar keratoderma with congenital alopecia, 104100* *Syndactyly, type III, 186100* *Oculodentodigital dysplasia, autosomal recessive, 257850* *Atrioventricular septal defect 3, 600309*
GJA3	*100*	*99,8*	*100*	*100*	*Cataract 14, multiple types, 601885*
GJA5	*100*	*100*	*100*	*100*	*Atrial fibrillation, familial, 11, 614049* *Atrial standstill, digenic (GJA5/SCN5A), 108770*
GJA8	*100*	*100*	*100*	*100*	*Cataract 1, multiple types, 116200*
GJB1	*100*	*100*	*100*	*100*	*Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800*
GJB2	*100*	*100*	*100*	*100*	*Keratoderma, palmoplantar, with deafness, 148350* *Deafness, autosomal recessive 1A, 220290* *Deafness, autosomal dominant 3A, 601544* *Hystrix-like ichthyosis with deafness, 602540* *Bart-Pumphrey syndrome, 149200*

					Keratitis-ichthyosis-deafness syndrome, 148210 *Vohwinkel syndrome, 124500*
GJB3	*100*	*100*	*100*	*100*	*Deafness, digenic, GJB2/GJB3, 220290* *Deafness, autosomal dominant 2B, 612644* *Erythrokeratodermia variabilis et progressiva 1, 133200* *Deafness, autosomal recessive,* *Deafness, autosomal dominant, with peripheral neuropathy,*
GJB4	*100*	*100*	*100*	*100*	*Erythrokeratodermia variabilis et progressiva 2, 617524*
GJB6	*100*	*100*	*100*	*100*	*Ectodermal dysplasia 2, Clouston type, 129500* *Deafness, autosomal dominant 3B, 612643* *Deafness, autosomal recessive 1B, 612645* *Deafness, digenic GJB2/GJB6, 220290*
GJC2	*82,3*	*64,5*	*97,8*	*93,2*	*Lymphatic malformation 3, 613480* *Spastic paraplegia 44, autosomal recessive, 613206* *Leukodystrophy, hypomyelinating, 2, 608804*
GK	*84,2*	*61,8*	*100*	*99,6*	*Glycerol kinase deficiency, 307030*
GLA	*91*	*85,9*	*91,3*	*91,3*	*Fabry disease, cardiac variant, 301500* *Fabry disease, 301500*
GLB1	*99,2*	*92,8*	*100*	*100*	*GM1-gangliosidosis, type I, 230500* *GM1-gangliosidosis, type III, 230650* *Mucopolysaccharidosis type IVB (Morquio), 253010* *GM1-gangliosidosis, type II, 230600*
GLDC	*88,9*	*77,8*	*100*	*99,9*	*Glycine encephalopathy, 605899*
GLDN	*95,8*	*91,5*	*100*	*100*	*Lethal congenital contracture syndrome 11, 617194*
GLE1	*100*	*99,9*	*100*	*100*	*Lethal congenital contracture syndrome 1, 253310* *Congenital arthrogryposis with anterior horn cell disease, 611890*
GLI1	*100*	*99,9*	*100*	*100*	*Polydactyly, preaxial I, 174400* *Polydactyly, postaxial, type A8, 618123*
GLI2	*99,8*	*98,6*	*100*	*99,9*	*Culler-Jones syndrome, 615849* *Holoprosencephaly 9, 610829*
GLI3	*98,5*	*97,7*	*100*	*100*	*Greig cephalopolysyndactyly syndrome, 175700* *Polydactyly, postaxial, types A1 and B, 174200* *Pallister-Hall syndrome, 146510* *Polydactyly, preaxial, type IV, 174700*
GLIS2	*100*	*99,9*	*100*	*100*	*Nephronophthisis 7, 611498*
GLIS3	*98,5*	*97,4*	*100*	*100*	*Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199*

GLMN	*98,6*	*94,1*	*100*	*100*	*Glomuvenous malformations, 138000*
GLRA1	*100*	*99,8*	*100*	*100*	*Hyperekplexia 1, 149400*
GLRB	*99,1*	*95,3*	*100*	*100*	*Hyperekplexia 2, 614619*
GLRX5	*97,2*	*89,6*	*99,3*	*95,2*	*Anemia, sideroblastic, 3, pyridoxine-refractory, 616860* *Spasticity, childhood-onset, with hyperglycinemia, 616859*
GLS	*96,9*	*88,5*	*100*	*99,9*	*Global developmental delay, progressive ataxia, and elevated glutamine, 618412* *?Infantile cataract, skin abnormalities, glutamate excess, and impaired intellectual development, 618339* *Developmental and epileptic encephalopathy 71, 618328*
GLUD1	*96,4*	*84,4*	*100*	*100*	*Hyperinsulinism-hyperammonemia syndrome, 606762*
GLUL	*73*	*69*	*100*	*100*	*Glutamine deficiency, congenital, 610015*
GLYCTK	*98,7*	*97,3*	*100*	*100*	*D-glyceric aciduria, 220120*
GM2A	*100*	*100*	*100*	*100*	*GM2-gangliosidosis, AB variant, 272750*
GMNN	*99,6*	*96,3*	*100*	*99,8*	*Meier-Gorlin syndrome 6, 616835*
GMPPA	*100*	*100*	*100*	*100*	*Alacrima, achalasia, and mental retardation syndrome, 615510*
GMPPB	*100*	*100*	*100*	*100*	*Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352* *Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351* *Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350*
GMPR	*100*	*99,9*	*100*	*100*	*No OMIM disease ID*
GMPS	*98,2*	*94,5*	*100*	*99,9*	*No OMIM disease ID*
GNA11	*98,4*	*93,1*	*100*	*100*	*Hypocalciuric hypercalcemia, type II, 145981* *Hypocalcemia, autosomal dominant 2, 615361*
GNA14	*100*	*100*	*100*	*100*	*No OMIM disease ID*
GNAI1	*97,2*	*89,7*	*100*	*100*	*No OMIM disease ID*
GNAI2	*100*	*100*	*100*	*100*	*Ventricular tachycardia, idiopathic, 192605* *Pituitary adenoma, ACTH-secreting, somatic,*
GNAI3	*98,4*	*93,2*	*100*	*100*	*Auriculocondylar syndrome 1, 602483*
GNAL	*96,8*	*93,3*	*100*	*100*	*Dystonia 25, 615073*
GNAO1	*93,8*	*93,8*	*100*	*100*	*Developmental and epileptic encephalopathy 17, 615473* *Neurodevelopmental disorder with involuntary movements, 617493*
GNAQ	*82,5*	*70,1*	*100*	*100*	*Capillary malformations, congenital, 1, somatic, mosaic, 163000* *Sturge-Weber syndrome, somatic, mosaic, 185300*
GNAS	*86,7*	*84,4*	*81,8*	*81,7*	*ACTH-independent macronodular adrenal hyperplasia, 219080* *Pituitary adenoma 3, multiple types, somatic, 617686*

					Pseudohypoparathyroidism Ic, 612462 *Pseudohypoparathyroidism Ia, 103580* *Osseous heteroplasia, progressive, 166350* *Pseudohypoparathyroidism Ib, 603233* *McCune-Albright syndrome, somatic, mosaic, 174800* *Pseudopseudohypoparathyroidism, 612463*
GNAS-AS1	*NC*	*NC*	*NC*	*NC*	*Pseudohypoparathyroidism, type IB, 603233*
GNAT1	*100*	*100*	*100*	*100*	*Night blindness, congenital stationary, autosomal dominant 3, 610444* *Night blindness, congenital stationary, type 1G, 616389*
GNAT2	*99,9*	*97,9*	*100*	*100*	*Achromatopsia 4, 613856*
GNB1	*100*	*100*	*100*	*100*	*Myelodysplastic syndrome, somatic, 614286* *Leukemia, acute lymphoblastic, somatic, 613065* *Mental retardation, autosomal dominant 42, 616973*
GNB2	*100*	*100*	*100*	*100*	*Neurodevelopmental disorder with hypotonia and dysmorphic facies, 619503* *?Sick sinus syndrome 4, 619464*
GNB3	*100*	*100*	*100*	*100*	*Night blindness, congenital stationary, type 1H, 617024*
GNB4	*99,8*	*99,6*	*100*	*100*	*Charcot-Marie-Tooth disease, dominant intermediate F, 615185*
GNB5	*99,9*	*96,5*	*100*	*100*	*Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182* *Intellectual developmental disorder with cardiac arrhythmia, 617173*
GNE	*100*	*99,5*	*100*	*100*	*Sialuria, 269921* *Nonaka myopathy, 605820*
GNMT	*100*	*100*	*100*	*100*	*Glycine N-methyltransferase deficiency, 606664*
GNPAT	*99,5*	*95,6*	*100*	*100*	*Rhizomelic chondrodysplasia punctata, type 2, 222765*
GNPNAT1	*69,3*	*46,4*	*100*	*100*	*No OMIM disease ID*
GNPTAB	*99,9*	*99,7*	*100*	*100*	*Mucolipidosis III alpha/beta, 252600* *Mucolipidosis II alpha/beta, 252500*
GNPTG	*99,8*	*96,6*	*100*	*100*	*Mucolipidosis III gamma, 252605*
GNRH1	*99,5*	*89,5*	*100*	*100*	*?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841*
GNRHR	*100*	*100*	*100*	*100*	*Hypogonadotropic hypogonadism 7 without anosmia, 146110*
GNS	*99,2*	*94,6*	*100*	*100*	*Mucopolysaccharidosis type IIID, 252940*
GORAB	*99,7*	*97,2*	*100*	*100*	*Geroderma osteodysplasticum, 231070*
GOSR2	*96*	*95,1*	*100*	*100*	*Epilepsy, progressive myoclonic 6, 614018*
GOT1	*100*	*98,6*	*100*	*100*	*Aspartate aminotransferase, serum level of, QTL1, 614419*

GOT2	*94,6*	*87*	*100*	*100*	*Developmental and epileptic encephalopathy 82, 618721*
GP1BA	*97,6*	*94,3*	*100*	*100*	*Bernard-Soulier syndrome, type A1 (recessive), 231200* *Bernard-Soulier syndrome, type A2 (dominant), 153670* *von Willebrand disease, platelet-type, 177820*
GP1BB	*77,8*	*66,9*	*100*	*99,5*	*Giant platelet disorder, isolated, 231200* *Bernard-Soulier syndrome, type B, 231200*
GP6	*100*	*99,9*	*97,7*	*93,5*	*Bleeding disorder, platelet-type, 11, 614201*
GP9	*98,1*	*91,8*	*100*	*100*	*Bernard-Soulier syndrome, type C, 231200*
GPAA1	*98,6*	*95,5*	*100*	*100*	*Glycosylphosphatidylinositol biosynthesis defect 15, 617810*
GPC3	*98,8*	*92,9*	*100*	*99,9*	*Wilms tumor, somatic, 194070* *Simpson-Golabi-Behmel syndrome, type 1, 312870*
GPC4	*99,9*	*98,3*	*100*	*100*	*Keipert syndrome, 301026*
GPC6	*100*	*99,9*	*100*	*100*	*Omodysplasia 1, 258315*
GPD1	*100*	*99,9*	*100*	*100*	*Hypertriglyceridemia, transient infantile, 614480*
GPD1L	*100*	*98,8*	*100*	*100*	*Brugada syndrome 2, 611777*
GPHN	*99,9*	*99,1*	*100*	*100*	*Molybdenum cofactor deficiency C, 615501*
GPI	*100*	*99,3*	*100*	*100*	*Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470*
GPIHBP1	*100*	*99,9*	*100*	*100*	*Hyperlipoproteinemia, type 1D, 615947*
GPNMB	*95,5*	*95,5*	*95,5*	*95,5*	*Amyloidosis, primary localized cutaneous, 3, 617920*
GPR101	*100*	*100*	*100*	*100*	*Pituitary adenoma 2, GH-secreting, 300943*
GPR143	*86,3*	*74,8*	*99,9*	*98,8*	*Ocular albinism, type I, Nettleship-Falls type, 300500* *Nystagmus 6, congenital, X-linked, 300814*
GPR161	*100*	*100*	*100*	*100*	*No OMIM disease ID*
GPR179	*100*	*100*	*100*	*100*	*Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565*
GPR68	*99,6*	*97,1*	*100*	*100*	*Amelogenesis imperfecta, hypomaturation type, IIA6, 617217*
GPR88	*99,8*	*97,1*	*98,7*	*95,2*	*?Chorea, childhood-onset, with psychomotor retardation, 616939*
GPRASP2	*100*	*98,9*	*100*	*100*	*?Deafness, X-linked 7, 301018*
GPSM2	*99,9*	*99,3*	*100*	*100*	*Chudley-McCullough syndrome, 604213*
GPT2	*99,4*	*95,3*	*100*	*100*	*Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281*
GPX1	*97,4*	*88,7*	*100*	*100*	*No OMIM disease ID*
GPX4	*91,3*	*88,4*	*98,8*	*96,1*	*Spondylometaphyseal dysplasia, Sedaghatian type, 250220*

GRAP	*81,6*	*77,3*	*100*	*100*	*Deafness, autosomal recessive 114, 618456*
GREB1L	*100*	*99,9*	*100*	*100*	*Deafness, autosomal dominant 80, 619274* *Renal hypodysplasia/aplasia 3, 617805*
GREM1	*100*	*100*	*100*	*100*	*No OMIM disease ID*
GREM2	*100*	*100*	*100*	*100*	*Tooth agenesis, selective, 9, 617275*
GRHL2	*100*	*99,9*	*100*	*100*	*Deafness, autosomal dominant 28, 608641* *Ectodermal dysplasia/short stature syndrome, 616029* *Corneal dystrophy, posterior polymorphous, 4, 618031*
GRHL3	*100*	*99,9*	*100*	*100*	*Van der Woude syndrome 2, 606713*
GRHPR	*83,3*	*79,2*	*100*	*99,3*	*Hyperoxaluria, primary, type II, 260000*
GRIA2	*99,7*	*96,6*	*100*	*100*	*Neurodevelopmental disorder with language impairment and behavioral abnormalities, 618917*
GRIA3	*99,5*	*94*	*99,9*	*98,6*	*Intellectual developmental disorder, X-linked, syndromic, Wu type, 300699*
GRIA4	*99,8*	*98,9*	*100*	*100*	*Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864*
GRID2	*100*	*99,8*	*100*	*100*	*Spinocerebellar ataxia, autosomal recessive 18, 616204*
GRIK2	*96,1*	*95,3*	*96,3*	*96,3*	*Mental retardation, autosomal recessive, 6, 611092*
GRIN1	*100*	*99,9*	*100*	*100*	*Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820* *Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254*
GRIN2A	*100*	*100*	*100*	*100*	*Epilepsy, focal, with speech disorder and with or without impaired intellectual development, 245570*
GRIN2B	*99,6*	*98,7*	*100*	*100*	*Developmental and epileptic encephalopathy 27, 616139* *Intellectual developmental disorder, autosomal dominant 6, with or without seizures, 613970*
GRIN2D	*83,8*	*69,4*	*93,7*	*87,4*	*Developmental and epileptic encephalopathy 46, 617162*
GRIP1	*100*	*99,3*	*100*	*100*	*Fraser syndrome 3, 617667*
GRK1	*100*	*100*	*100*	*100*	*Oguchi disease-2, 613411*
GRM1	*100*	*99,5*	*100*	*100*	*Spinocerebellar ataxia, autosomal recessive 13, 614831* *Spinocerebellar ataxia 44, 617691*
GRM6	*93,1*	*83,6*	*98,4*	*96,3*	*Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270*
GRM7	*99,9*	*99,1*	*100*	*100*	*Neurodevelopmental disorder with seizures, hypotonia, and brain abnormalities, 618922*
GRN	*100*	*100*	*100*	*100*	*Aphasia, primary progressive, 607485* *Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485* *Ceroid lipofuscinosis, neuronal, 11, 614706*
GRXCR1	*99,8*	*99,5*	*100*	*100*	*Deafness, autosomal recessive 25, 613285*

GRXCR2	*100*	*100*	*100*	*100*	*?Deafness, autosomal recessive 101, 615837*
GSC	*98,9*	*93*	*100*	*100*	*Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471*
GSDME	*99,9*	*98,3*	*100*	*100*	*Deafness, autosomal dominant 5, 600994*
GSE1	*100*	*99,4*	*100*	*100*	*No OMIM disease ID*
GSN	*95,7*	*93,5*	*100*	*99,7*	*Amyloidosis, Finnish type, 105120*
GSR	*95,8*	*92,7*	*100*	*99,9*	*Hemolytic anemia due to glutathione reductase deficiency, 618660*
GSS	*96,5*	*96,3*	*100*	*100*	*Hemolytic anemia due to glutathione synthetase deficiency, 231900* *Glutathione synthetase deficiency, 266130*
GSX2	*100*	*100*	*100*	*100*	*Diencephalic-mesencephalic junction dysplasia syndrome 2, 618646*
GTF2E2	*99,6*	*99,5*	*100*	*99,9*	*Trichothiodystrophy 6, nonphotosensitive, 616943*
GTF2H5	*72,2*	*71,7*	*72,5*	*72,5*	*Trichothiodystrophy 3, photosensitive, 616395*
GTPBP2	*99,8*	*98,5*	*100*	*100*	*Jaberi-Elahi syndrome, 617988*
GTPBP3	*100*	*99,9*	*100*	*100*	*Combined oxidative phosphorylation deficiency 23, 616198*
GUCA1A	*100*	*100*	*100*	*100*	*Cone-rod dystrophy 14, 602093* *Cone dystrophy-3, 602093*
GUCA1B	*100*	*100*	*100*	*100*	*Retinitis pigmentosa 48, 613827*
GUCY1A1	*99,9*	*99,8*	*100*	*100*	*Moyamoya 6 with achalasia, 615750*
GUCY2C	*99,9*	*99,4*	*100*	*100*	*Diarrhea 6, 614616* *Meconium ileus, 614665*
GUCY2D	*100*	*98,7*	*100*	*100*	*Cone-rod dystrophy 6, 601777* *?Choroidal dystrophy, central areolar 1, 215500* *Leber congenital amaurosis 1, 204000* *Night blindness, congenital stationary, type 1I, 618555*
GUF1	*99,3*	*97,9*	*100*	*100*	*?Developmental and epileptic encephalopathy 40, 617065*
GULOP	*NC*	*NC*	*NC*	*NC*	*Scurvy,*
GUSB	*92,5*	*90,1*	*100*	*100*	*Mucopolysaccharidosis VII, 253220*
GYG1	*99,6*	*97,4*	*100*	*100*	*?Glycogen storage disease XV, 613507* *Polyglucosan body myopathy 2, 616199*
GYS1	*100*	*98*	*100*	*100*	*Glycogen storage disease 0, muscle, 611556*
GYS2	*99,9*	*99,4*	*100*	*100*	*Glycogen storage disease 0, liver, 240600*
GZF1	*100*	*99,7*	*100*	*100*	*Joint laxity, short stature, and myopia, 617662*
H1-4	*100*	*100*	*100*	*100*	*Rahman syndrome, 617537*

H19	*NC*	*NC*	*NC*	*NC*	*No OMIM disease ID*
H4C3	*100*	*100*	*100*	*100*	*No OMIM disease ID*
H6PD	*99*	*99*	*100*	*100*	*Cortisone reductase deficiency 1, 604931*
HAAO	*100*	*100*	*100*	*100*	*Vertebral, cardiac, renal, and limb defects syndrome 1, 617660*
HABP2	*100*	*99,7*	*100*	*100*	*No OMIM disease ID*
HACE1	*99,7*	*99,3*	*100*	*99,9*	*Spastic paraplegia and psychomotor retardation with or without seizures, 616756*
HADH	*99,2*	*97,7*	*100*	*100*	*Hyperinsulinemic hypoglycemia, familial, 4, 609975* *3-hydroxyacyl-CoA dehydrogenase deficiency, 231530*
HADHA	*95,5*	*88,3*	*100*	*100*	*HELLP syndrome, maternal, of pregnancy, 609016* *Mitochondrial trifunctional protein deficiency, 609015* *LCHAD deficiency, 609016* *Fatty liver, acute, of pregnancy, 609016*
HADHB	*97,7*	*87*	*100*	*99,9*	*Trifunctional protein deficiency, 609015*
HAGH	*100*	*100*	*99,1*	*96,7*	*No OMIM disease ID*
HAMP	*100*	*100*	*100*	*100*	*Hemochromatosis, type 2B, 613313*
HAND1	*100*	*100*	*100*	*100*	*No OMIM disease ID*
HAND2	*99,7*	*94,9*	*100*	*100*	*No OMIM disease ID*
HARS1	*100*	*100*	*100*	*100*	*Charcot-Marie-Tooth disease, axonal, type 2W, 616625* *Usher syndrome type 3B, 614504*
HARS2	*100*	*99,5*	*100*	*99,8*	*Perrault syndrome 2, 614926*
HAVCR2	*100*	*99,8*	*100*	*100*	*T-cell lymphoma, subcutaneous panniculitis-like, 618398*
HAX1	*100*	*100*	*100*	*100*	*Neutropenia, severe congenital 3, autosomal recessive, 610738*
HBA1	*100*	*100*	*100*	*100*	*Hemoglobin H disease, nondeletional, 613978* *Thalassemias, alpha-, 604131* *Heinz body anemias, alpha-, 140700* *Methemoglobinemia, alpha type, 617973* *Erythrocytosis 7, 617981*
HBA2	*99,8*	*96,6*	*100*	*100*	*Heinz body anemia, 140700* *Erythrocytosis 7, 617981* *Thalassemia, alpha-, 604131* *Hemoglobin H disease, deletional and nondeletional, 613978*
HBB	*100*	*100*	*100*	*100*	*Methemoglobinemia, beta type, 617971* *Thalassemia-beta, dominant inclusion-body, 603902* *Sickle cell anemia, 603903*

					Thalassemia, beta, 613985 *Delta-beta thalassemia, 141749* *Hereditary persistence of fetal hemoglobin, 141749* *Heinz body anemia, 140700* *Erythrocytosis 6, 617980*
HBD	*100*	*100*	*100*	*100*	*Thalassemia due to Hb Lepore,* *Thalassemia, delta-,*
HBG1	*98,3*	*94,7*	*98,2*	*95,8*	*Fetal hemoglobin quantitative trait locus 1, 141749*
HBG2	*100*	*100*	*100*	*100*	*Fetal hemoglobin quantitative trait locus 1, 141749* *Cyanosis, transient neonatal, 613977*
HCCS	*99,3*	*96,1*	*100*	*100*	*Linear skin defects with multiple congenital anomalies 1, 309801*
HCFC1	*98,1*	*93*	*100*	*100*	*Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541*
HCN1	*98,4*	*98,3*	*98,5*	*98,4*	*Developmental and epileptic encephalopathy 24, 615871* *Generalized epilepsy with febrile seizures plus, type 10, 618482*
HCN2	*59,8*	*47,7*	*84*	*76,9*	*Febrile seizures, familial, 2, 602477* *Generalized epilepsy with febrile seizures plus, type 11, 602477*
HCN3	*99,9*	*98,5*	*100*	*100*	*No OMIM disease ID*
HCN4	*100*	*99,2*	*100*	*100*	*Sick sinus syndrome 2, 163800* *Brugada syndrome 8, 613123*
HCRT	*91,9*	*83,1*	*100*	*100*	*?Narcolepsy 1, 161400*
HDAC4	*100*	*99,9*	*100*	*100*	*No OMIM disease ID*
HDAC6	*99,5*	*97,1*	*100*	*99,9*	*?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863*
HDAC8	*85,7*	*83,7*	*96,4*	*95,2*	*Cornelia de Lange syndrome 5, 300882*
HEATR5B	*100*	*99,3*	*100*	*100*	*No OMIM disease ID*
HECW2	*99,8*	*98,2*	*100*	*100*	*Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268*
HELLS	*98,2*	*91,9*	*100*	*99,9*	*Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911*
HEPACAM	*86,8*	*78,5*	*100*	*100*	*Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925* *Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926*
HEPH	*98*	*89*	*100*	*100*	*No OMIM disease ID*
HEPHL1	*100*	*99,5*	*100*	*100*	*?Abnormal hair, joint laxity, and developmental delay, 261990*
HERC1	*100*	*99,9*	*100*	*100*	*Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011*
HERC2	*79,7*	*76,7*	*100*	*100*	*Mental retardation, autosomal recessive 38, 615516*

HES7	*75,6*	*44,1*	*100*	*100*	*Spondylocostal dysostosis 4, autosomal recessive, 613686*
HESX1	*99,3*	*97,3*	*100*	*100*	*Pituitary hormone deficiency, combined, 5, 182230* *Septooptic dysplasia, 182230* *Growth hormone deficiency with pituitary anomalies, 182230*
HEXA	*93,8*	*93,1*	*100*	*100*	*GM2-gangliosidosis, several forms, 272800* *Tay-Sachs disease, 272800*
HEXB	*99,4*	*96,6*	*100*	*100*	*Sandhoff disease, infantile, juvenile, and adult forms, 268800*
HEY2	*99,8*	*98,7*	*100*	*100*	*No OMIM disease ID*
HFE	*99,9*	*97,8*	*100*	*100*	*Hemochromatosis, 235200*
HFM1	*95,4*	*89,8*	*100*	*99,9*	*Premature ovarian failure 9, 615724*
HGD	*100*	*99,7*	*100*	*100*	*Alkaptonuria, 203500*
HGF	*99,7*	*99,6*	*100*	*100*	*Deafness, autosomal recessive 39, 608265*
HGSNAT	*86,4*	*86,2*	*91,3*	*89,1*	*Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930* *Retinitis pigmentosa 73, 616544*
HHAT	*99*	*96,8*	*100*	*100*	*Nivelon-Nivelon-Mabille syndrome, 600092*
HIBADH	*93,8*	*91,3*	*100*	*100*	*No OMIM disease ID*
HIBCH	*98,2*	*84,5*	*100*	*100*	*3-hydroxyisobutryl-CoA hydrolase deficiency, 250620*
HIKESHI	*98,9*	*91,5*	*100*	*100*	*Leukodystrophy, hypomyelinating, 13, 616881*
HINT1	*95,2*	*82,5*	*100*	*100*	*Neuromyotonia and axonal neuropathy, autosomal recessive, 137200*
HIVEP2	*100*	*99,9*	*100*	*100*	*Mental retardation, autosomal dominant 43, 616977*
HJV	*100*	*100*	*100*	*100*	*Hemochromatosis, type 2A, 602390*
HK1	*100*	*99,9*	*100*	*100*	*Retinitis pigmentosa 79, 617460* *Neuropathy, hereditary motor and sensory, Russe type, 605285* *Neurodevelopmental disorder with visual defects and brain anomalies, 618547* *Hemolytic anemia due to hexokinase deficiency, 235700*
HLCS	*100*	*100*	*100*	*100*	*Holocarboxylase synthetase deficiency, 253270*
HMBS	*100*	*98,4*	*100*	*100*	*Porphyria, acute intermittent, nonerythroid variant, 176000* *Porphyria, acute intermittent, 176000*
HMGA2	*81*	*76,6*	*89,6*	*80,1*	*Silver-Russell syndrome 5, 618908*
HMGB3	*77,6*	*63,1*	*100*	*100*	*?Microphthalmia, syndromic 13, 300915*
HMGCL	*100*	*99,4*	*100*	*100*	*HMG-CoA lyase deficiency, 246450*
HMGCS2	*100*	*99,7*	*100*	*100*	*HMG-CoA synthase-2 deficiency, 605911*

HMOX1	*97,7*	*90,1*	*100*	*100*	*Heme oxygenase-1 deficiency, 614034*
HMX1	*64*	*43,1*	*99,8*	*96,8*	*Oculoauricular syndrome, 612109*
HNF1A	*100*	*99,8*	*100*	*100*	*Hepatic adenoma, somatic, 142330* *Diabetes mellitus, insulin-dependent, 20, 612520* *MODY, type III, 600496* *Renal cell carcinoma, 144700*
HNF1B	*99*	*95,7*	*100*	*100*	*Type 2 diabetes mellitus, 125853* *Renal cysts and diabetes syndrome, 137920*
HNF4A	*99,9*	*98,6*	*100*	*100*	*Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026* *MODY, type I, 125850*
HNMT	*99,9*	*99,5*	*100*	*100*	*Mental retardation, autosomal recessive 51, 616739*
HNRNPA1	*97,1*	*84,8*	*100*	*100*	*?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3, 615424* *Amyotrophic lateral sclerosis 20, 615426*
HNRNPA2B1	*99,9*	*98,9*	*100*	*100*	*?Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2, 615422*
HNRNPD	*86,3*	*79,8*	*100*	*100*	*No OMIM disease ID*
HNRNPDL	*96,5*	*86,3*	*100*	*100*	*Muscular dystrophy, limb-girdle, autosomal dominant 3, 609115*
HNRNPH1	*99,1*	*94,2*	*100*	*100*	*No OMIM disease ID*
HNRNPH2	*100*	*100*	*100*	*100*	*Intellectual developmental disorder, X-linked, syndromic, Bain type, 300986*
HNRNPK	*88,8*	*78,1*	*100*	*100*	*Au-Kline syndrome, 616580*
HNRNPU	*99,9*	*98,7*	*100*	*100*	*Developmental and epileptic encephalopathy 54, 617391*
HOGA1	*99,5*	*95,5*	*100*	*100*	*Hyperoxaluria, primary, type III, 613616*
HOMER2	*99,5*	*98,6*	*100*	*100*	*?Deafness, autosomal dominant 68, 616707*
HOXA1	*100*	*100*	*100*	*100*	*Bosley-Salih-Alorainy syndrome, 601536* *Athabaskan brainstem dysgenesis syndrome, 601536*
HOXA11	*97,1*	*88,3*	*100*	*100*	*Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432*
HOXA13	*76,4*	*67,3*	*90,3*	*81,4*	*Hand-foot-uterus syndrome, 140000* *?Guttmacher syndrome, 176305*
HOXA2	*99,9*	*99,3*	*100*	*100*	*Microtia with or without hearing impairment (AD), 612290* *?Microtia, hearing impairment, and cleft palate (AR), 612290*
HOXB1	*100*	*100*	*100*	*100*	*Facial paresis, hereditary congenital, 3, 614744*
HOXB13	*100*	*99,6*	*100*	*100*	*No OMIM disease ID*
HOXC13	*100*	*100*	*100*	*100*	*Ectodermal dysplasia 9, hair/nail type, 614931*

HOXD10	*100*	*99,6*	*100*	*100*	*Vertical talus, congenital, 192950* *Charcot-Marie-Tooth disease, foot deformity of, 192950*
HOXD13	*100*	*98,8*	*100*	*100*	*Syndactyly, type V, 186300* *Synpolydactyly 1, 186000* *Brachydactyly, type E, 113300* *Brachydactyly, type D, 113200* *?Brachydactyly-syndactyly syndrome, 610713*
HPCA	*100*	*100*	*100*	*100*	*Dystonia 2, torsion, autosomal recessive, 224500*
HPD	*100*	*99,8*	*100*	*100*	*Hawkinsinuria, 140350* *Tyrosinemia, type III, 276710*
HPDL	*100*	*100*	*100*	*100*	*Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026* *Spastic paraplegia 83, autosomal recessive, 619027*
HPGD	*99,5*	*99,3*	*100*	*99,7*	*?Digital clubbing, isolated congenital, 119900* *Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100* *Cranioosteoarthropathy, 259100*
HPRT1	*98,6*	*90,6*	*99,5*	*98,4*	*Hyperuricemia, HRPT-related, 300323* *Lesch-Nyhan syndrome, 300322*
HPS1	*100*	*100*	*100*	*100*	*Hermansky-Pudlak syndrome 1, 203300*
HPS3	*99,8*	*97,1*	*100*	*100*	*Hermansky-Pudlak syndrome 3, 614072*
HPS4	*100*	*100*	*100*	*100*	*Hermansky-Pudlak syndrome 4, 614073*
HPS5	*99,9*	*99,3*	*100*	*100*	*Hermansky-Pudlak syndrome 5, 614074*
HPS6	*97,7*	*88,7*	*100*	*100*	*Hermansky-Pudlak syndrome 6, 614075*
HPSE2	*100*	*99,5*	*100*	*100*	*Urofacial syndrome 1, 236730*
HR	*98,9*	*96,2*	*100*	*100*	*Atrichia with papular lesions, 209500* *Alopecia universalis, 203655*
HRAS	*100*	*100*	*100*	*100*	*Bladder cancer, somatic, 109800* *Thyroid carcinoma, follicular, somatic, 188470* *Congenital myopathy with excess of muscle spindles, 218040* *Nevus sebaceous or woolly hair nevus, somatic, 162900* *Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200* *Spitz nevus or nevus spilus, somatic, 137550* *Costello syndrome, 218040*
HRG	*95,4*	*93,9*	*100*	*100*	*Thrombophilia due to HRG deficiency, 613116*
HS2ST1	*99,6*	*99,2*	*100*	*99,5*	*Neurofacioskeletal syndrome with or without renal agenesis, 619194*
HS3ST6	*87,4*	*84,4*	*93,8*	*87*	*?Angioedema, hereditary, 8, 619367*

HS6ST1	*93,6*	*86,7*	*100*	*100*	*No OMIM disease ID*
HS6ST2	*97,4*	*96*	*100*	*100*	*?Paganini-Miozzo syndrome, 301025*
HSCB	*99,8*	*97,6*	*100*	*100*	*No OMIM disease ID*
HSD11B1	*100*	*99,7*	*100*	*100*	*Cortisone reductase deficiency 2, 614662*
HSD11B2	*87,6*	*83,8*	*99,9*	*97,6*	*Apparent mineralocorticoid excess, 218030*
HSD17B10	*99,9*	*98,3*	*100*	*100*	*HSD10 mitochondrial disease, 300438*
HSD17B3	*97,8*	*97,8*	*100*	*100*	*Pseudohermaphroditism, male, with gynecomastia, 264300*
HSD17B4	*95,3*	*92,8*	*96,6*	*96,6*	*D-bifunctional protein deficiency, 261515* *Perrault syndrome 1, 233400*
HSD3B2	*100*	*99,7*	*100*	*100*	*Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810*
HSD3B7	*98,9*	*95*	*100*	*100*	*Bile acid synthesis defect, congenital, 1, 607765*
HSF2	*99*	*93,2*	*100*	*99,8*	*No OMIM disease ID*
HSF2BP	*100*	*98,3*	*100*	*100*	*Premature ovarian failure 19, 619245*
HSF4	*99*	*96,3*	*100*	*100*	*Cataract 5, multiple types, 116800*
HSPA9	*87,1*	*82,8*	*100*	*100*	*Even-plus syndrome, 616854* *Anemia, sideroblastic, 4, 182170*
HSPB1	*99,1*	*92,1*	*100*	*100*	*Neuronopathy, distal hereditary motor, type IIB, 608634* *Charcot-Marie-Tooth disease, axonal, type 2F, 606595*
HSPB3	*100*	*100*	*100*	*100*	*?Neuronopathy, distal hereditary motor, type IIC, 613376*
HSPB6	*92,7*	*83*	*100*	*100*	*No OMIM disease ID*
HSPB8	*100*	*100*	*100*	*100*	*Neuronopathy, distal hereditary motor, type IIA, 158590* *Charcot-Marie-Tooth disease, axonal, type 2L, 608673*
HSPD1	*96,7*	*90*	*100*	*100*	*Spastic paraplegia 13, autosomal dominant, 605280* *Leukodystrophy, hypomyelinating, 4, 612233*
HSPG2	*99,2*	*97,5*	*100*	*99,8*	*Dyssegmental dysplasia, Silverman-Handmaker type, 224410* *Schwartz-Jampel syndrome, type 1, 255800*
HTR1A	*100*	*100*	*100*	*100*	*Periodic fever, menstrual cycle dependent, 614674*
HTRA1	*77,9*	*73,2*	*87,5*	*83,2*	*CARASIL syndrome, 600142* *Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779*
HTRA2	*100*	*99,6*	*100*	*100*	*3-methylglutaconic aciduria, type VIII, 617248*
HTT	*98,9*	*96,6*	*100*	*99,9*	*Lopes-Maciel-Rodan syndrome, 617435* *Huntington disease, 143100*
HUWE1	*98,6*	*93,2*	*100*	*100*	*Intellectual developmental disorder, X-linked, Turner type, 309590*

HYAL1	*100*	*100*	*100*	*100*	*?Mucopolysaccharidosis type IX, 601492*
HYAL2	*100*	*99,9*	*100*	*100*	*No OMIM disease ID*
HYDIN	*99,8*	*98,7*	*100*	*100*	*Ciliary dyskinesia, primary, 5, 608647*
HYLS1	*100*	*100*	*100*	*100*	*Hydrolethalus syndrome, 236680*
HYOU1	*99,9*	*99*	*100*	*100*	*?Immunodeficiency 59 and hypoglycemia, 233600*
IARS1	*99,9*	*99,4*	*100*	*100*	*Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093*
IARS2	*99,9*	*99,8*	*100*	*100*	*Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007*
IBA57	*95,4*	*91,7*	*100*	*100*	*Multiple mitochondrial dysfunctions syndrome 3, 615330* *?Spastic paraplegia 74, autosomal recessive, 616451*
ICOS	*99,9*	*99,8*	*100*	*99,9*	*Immunodeficiency, common variable, 1, 607594*
ICOSLG	*99,6*	*99,1*	*100*	*100*	*No OMIM disease ID*
ID4	*90,7*	*85,3*	*98,8*	*90,6*	*No OMIM disease ID*
IDH1	*90,6*	*75,5*	*100*	*100*	*No OMIM disease ID*
IDH2	*99,8*	*97,4*	*100*	*100*	*D-2-hydroxyglutaric aciduria 2, 613657*
IDH3A	*98,9*	*97,3*	*100*	*100*	*Retinitis pigmentosa 90, 619007*
IDH3B	*95,4*	*95,4*	*100*	*100*	*Retinitis pigmentosa 46, 612572*
IDI1	*99*	*96,4*	*100*	*100*	*No OMIM disease ID*
IDS	*99,6*	*95,3*	*100*	*100*	*Mucopolysaccharidosis II, 309900*
IDUA	*94,6*	*87,4*	*100*	*100*	*Mucopolysaccharidosis Is, 607016* *Mucopolysaccharidosis Ih/s, 607015* *Mucopolysaccharidosis Ih, 607014*
IER3IP1	*92*	*80,2*	*100*	*100*	*Microcephaly, epilepsy, and diabetes syndrome, 614231*
IFIH1	*99,5*	*97,3*	*100*	*100*	*Aicardi-Goutieres syndrome 7, 615846* *Singleton-Merten syndrome 1, 182250*
IFITM5	*99,7*	*97*	*100*	*100*	*Osteogenesis imperfecta, type V, 610967*
IFNAR1	*97,4*	*96,6*	*97,8*	*97,7*	*No OMIM disease ID*
IFNAR2	*99,8*	*98,8*	*100*	*100*	*?Immunodeficiency 45, 616669*
IFNG	*100*	*99,9*	*100*	*100*	*?Immunodeficiency 69, mycobacteriosis, 618963*
IFNGR1	*98*	*97,3*	*100*	*100*	*Immunodeficiency 27A, mycobacteriosis, AR, 209950* *Immunodeficiency 27B, mycobacteriosis, AD, 615978*

IFNGR2	*93,7*	*93,2*	*100*	*99,5*	*Immunodeficiency 28, mycobacteriosis, 614889*
IFNLR1	*99,8*	*98,2*	*100*	*100*	*No OMIM disease ID*
IFRD1	*99,9*	*99,2*	*100*	*100*	*No OMIM disease ID*
IFT122	*99,9*	*99,2*	*100*	*100*	*Cranioectodermal dysplasia 1, 218330*
IFT140	*99,9*	*99,2*	*100*	*100*	*Short-rib thoracic dysplasia 9 with or without polydactyly, 266920* *Retinitis pigmentosa 80, 617781*
IFT172	*99,6*	*98,6*	*100*	*100*	*Retinitis pigmentosa 71, 616394* *Bardet-Biedl syndrome 20, 619471* *Short-rib thoracic dysplasia 10 with or without polydactyly, 615630*
IFT27	*100*	*100*	*100*	*100*	*Bardet-Biedl syndrome 19, 615996*
IFT43	*100*	*100*	*100*	*100*	*?Cranioectodermal dysplasia 3, 614099* *?Retinitis pigmentosa 81, 617871* *Short-rib thoracic dysplasia 18 with polydactyly, 617866*
IFT52	*100*	*99,9*	*100*	*99,9*	*Short-rib thoracic dysplasia 16 with or without polydactyly, 617102*
IFT57	*99,9*	*99*	*100*	*99,7*	*?Orofaciodigital syndrome XVIII, 617927*
IFT74	*98,6*	*96,2*	*100*	*99,9*	*?Bardet-Biedl syndrome 22, 617119*
IFT80	*97,2*	*85,7*	*100*	*99,9*	*Short-rib thoracic dysplasia 2 with or without polydactyly, 611263*
IFT81	*92,9*	*89,6*	*94,9*	*94,6*	*Short-rib thoracic dysplasia 19 with or without polydactyly, 617895*
IFT88	*99,3*	*96,9*	*100*	*99,9*	*No OMIM disease ID*
IGBP1	*98,8*	*93,5*	*100*	*100*	*?Corpus callosum, agenesis of, with impaired intellectual development, ocular coloboma and micrognathia, 300472*
IGF1	*99,8*	*99,8*	*100*	*100*	*Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747*
IGF1R	*100*	*99,7*	*100*	*100*	*Insulin-like growth factor I, resistance to, 270450*
IGF2	*100*	*100*	*100*	*100*	*Silver-Russell syndrome 3, 616489*
IGF2R	*99,3*	*96,6*	*99,8*	*99,5*	*Hepatocellular carcinoma, somatic, 114550*
IGFALS	*100*	*100*	*100*	*100*	*Acid-labile subunit, deficiency of, 615961*
IGFBP7	*93,6*	*88,5*	*100*	*100*	*Retinal arterial macroaneurysm with supravalvular pulmonic stenosis, 614224*
IGHG2	*65,9*	*47,5*	*100*	*100*	*IgG2 deficiency, selective,*
IGHM	*100*	*100*	*100*	*100*	*Agammaglobulinemia 1, 601495*
IGHMBP2	*99,3*	*96,9*	*100*	*100*	*Neuronopathy, distal hereditary motor, type VI, 604320* *Charcot-Marie-Tooth disease, axonal, type 2S, 616155*
IGKC	*100*	*100*	*100*	*100*	*Kappa light chain deficiency, 614102*

IGLL1	*100*	*99,7*	*100*	*100*	*Agammaglobulinemia 2, 613500*
IGSF1	*99,3*	*94*	*100*	*100*	*Hypothyroidism, central, and testicular enlargement, 300888*
IGSF10	*100*	*99,9*	*100*	*100*	*No OMIM disease ID*
IGSF3	*95,3*	*94*	*100*	*100*	*?Lacrimal duct defect, 149700*
IHH	*100*	*100*	*100*	*100*	*Acrocapitofemoral dysplasia, 607778* *Brachydactyly, type A1, 112500*
IKBKB	*99*	*96,1*	*100*	*100*	*Immunodeficiency 15B, 615592* *Immunodeficiency 15A, 618204*
IKBKG	*84,6*	*75,2*	*100*	*100*	*Incontinentia pigmenti, 308300* *Ectodermal dysplasia and immunodeficiency 1, 300291* *Immunodeficiency 33, 300636*
IKZF1	*99,3*	*99,3*	*100*	*100*	*Immunodeficiency, common variable, 13, 616873*
IKZF3	*100*	*100*	*100*	*100*	*?Immunodeficiency 84, 619437*
IKZF5	*100*	*100*	*100*	*100*	*Thrombocytopenia, autosomal dominant, 7, 619130*
IL10	*100*	*98,1*	*100*	*100*	*No OMIM disease ID*
IL10RA	*100*	*99,9*	*100*	*100*	*Inflammatory bowel disease 28, early onset, autosomal recessive, 613148*
IL10RB	*99,9*	*97,8*	*100*	*100*	*Inflammatory bowel disease 25, early onset, autosomal recessive, 612567*
IL11RA	*100*	*99,6*	*100*	*100*	*Craniosynostosis and dental anomalies, 614188*
IL12B	*100*	*99,1*	*100*	*100*	*Immunodeficiency 29, mycobacteriosis, 614890*
IL12RB1	*98,7*	*96,1*	*94,1*	*94,1*	*Immunodeficiency 30, 614891*
IL17F	*99,9*	*97,6*	*100*	*100*	*?Candidiasis, familial, 6, autosomal dominant, 613956*
IL17RA	*100*	*99,8*	*100*	*100*	*Immunodeficiency 51, 613953*
IL17RC	*100*	*99,9*	*100*	*100*	*Candidiasis, familial, 9, 616445*
IL17RD	*99,9*	*99*	*100*	*100*	*Hypogonadotropic hypogonadism 18 with or without anosmia, 615267*
IL18BP	*100*	*100*	*100*	*100*	*No OMIM disease ID*
IL1RAPL1	*99,8*	*98,5*	*100*	*100*	*Intellectual developmental disorder, X-linked 21, 300143*
IL1RN	*100*	*99,9*	*100*	*100*	*Interleukin 1 receptor antagonist deficiency, 612852*
IL2	*96,2*	*88,8*	*100*	*100*	*No OMIM disease ID*
IL21	*99,2*	*93,5*	*100*	*100*	*?Immunodeficiency, common variable, 11, 615767*
IL21R	*100*	*100*	*100*	*100*	*Immunodeficiency 56, 615207*

IL2RA	*100*	*99,1*	*100*	*100*	*Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367*
IL2RB	*100*	*99,8*	*100*	*100*	*Immunodeficiency 63 with lymphoproliferation and autoimmunity, 618495*
IL2RG	*99,8*	*92,8*	*100*	*100*	*Combined immunodeficiency, X-linked, moderate, 312863* *Severe combined immunodeficiency, X-linked, 300400*
IL31RA	*99,8*	*99,8*	*100*	*100*	*?Amyloidosis, primary localized cutaneous, 2, 613955*
IL36RN	*100*	*99,9*	*100*	*100*	*Psoriasis 14, pustular, 614204*
IL37	*99,9*	*97,1*	*100*	*100*	*?Inflammatory bowel disease (infantile ulcerative colitis) 31, 619398*
IL6R	*99,1*	*94,2*	*92,7*	*92,7*	*Hyper-IgE recurrent infection syndrome 5, autosomal recessive, 618944*
IL6ST	*94,9*	*89,4*	*100*	*100*	*Hyper-IgE recurrent infection syndrome 4, autosomal recessive, 618523*
IL7R	*99,9*	*99,3*	*100*	*100*	*Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971*
ILDR1	*99,1*	*97,1*	*100*	*100*	*Deafness, autosomal recessive 42, 609646*
ILK	*100*	*99,9*	*100*	*100*	*No OMIM disease ID*
IMPA1	*96,1*	*86,4*	*100*	*99,8*	*Mental retardation, autosomal recessive 59, 617323*
IMPDH1	*89*	*81,7*	*100*	*99,9*	*Retinitis pigmentosa 10, 180105* *Leber congenital amaurosis 11, 613837*
IMPG1	*99,6*	*98,3*	*100*	*99,9*	*Macular dystrophy, vitelliform, 4, 616151* *Retinitis pigmentosa 91, 153870*
IMPG2	*99,4*	*97,9*	*100*	*100*	*Retinitis pigmentosa 56, 613581* *Macular dystrophy, vitelliform, 5, 616152*
INF2	*87,2*	*84,8*	*100*	*100*	*Glomerulosclerosis, focal segmental, 5, 613237* *Charcot-Marie-Tooth disease, dominant intermediate E, 614455*
ING1	*100*	*100*	*100*	*100*	*Squamous cell carcinoma, head and neck, somatic, 275355*
INO80	*99,9*	*98*	*100*	*100*	*No OMIM disease ID*
INPP5E	*96,9*	*93,2*	*100*	*100*	*Joubert syndrome 1, 213300* *Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156*
INPP5K	*100*	*99,7*	*100*	*100*	*Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404*
INPPL1	*98,6*	*94,4*	*100*	*99,9*	*Opsismodysplasia, 258480*
INS	*100*	*99,2*	*100*	*100*	*Diabetes mellitus, insulin-dependent, 2, 125852* *Maturity-onset diabetes of the young, type 10, 613370* *Hyperproinsulinemia, 616214* *Diabetes mellitus, permanent neonatal 4, 618858*
INSL3	*80,6*	*77,6*	*80,7*	*80,7*	*Cryptorchidism, 219050*

INSR	*97,3*	*93*	*100*	*99,6*	*Rabson-Mendenhall syndrome, 262190* *Leprechaunism, 246200* *Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549* *Hyperinsulinemic hypoglycemia, familial, 5, 609968*
INTS1	*99,8*	*98,6*	*100*	*100*	*Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies, 618571*
INTS8	*99,6*	*98,8*	*100*	*99,9*	*?Neurodevelopmental disorder with cerebellar hypoplasia and spasticity, 618572*
INTU	*99,9*	*98,6*	*100*	*100*	*?Orofaciodigital syndrome XVII, 617926* *?Short-rib thoracic dysplasia 20 with polydactyly, 617925*
INVS	*100*	*99,8*	*100*	*100*	*Nephronophthisis 2, infantile, 602088*
IPMK	*98,9*	*89,9*	*100*	*100*	*No OMIM disease ID*
IPO8	*99,5*	*97*	*100*	*99,9*	*VISS syndrome, 619472*
IQCB1	*92,8*	*82,8*	*100*	*100*	*Senior-Loken syndrome 5, 609254*
IQCE	*100*	*98,1*	*100*	*100*	*Polydactyly, postaxial, type A7, 617642*
IQSEC1	*88,6*	*86,1*	*97,6*	*94,8*	*Intellectual developmental disorder with short stature and behavioral abnormalities, 618687*
IQSEC2	*94,6*	*84*	*99,5*	*98,3*	*Intellectual developmental disorder, X-linked 1, 309530*
IRAK1	*99,4*	*95,9*	*99,7*	*98,9*	*No OMIM disease ID*
IRAK4	*99,5*	*95,7*	*100*	*99,8*	*Immunodeficiency 67, 607676*
IREB2	*99,9*	*99,8*	*100*	*100*	*Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451*
IRF1	*100*	*100*	*100*	*100*	*Nonsmall cell lung cancer, somatic, 211980* *Gastric cancer, somatic, 613659* *Myelodysplastic syndrome, preleukemic,* *Myelogenous leukemia, acute,*
IRF2BP2	*97,2*	*83,5*	*100*	*100*	*?Immunodeficiency, common variable, 14, 617765*
IRF2BPL	*100*	*97,9*	*99,9*	*99*	*Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088*
IRF3	*100*	*99,9*	*100*	*100*	*No OMIM disease ID*
IRF4	*100*	*100*	*100*	*100*	*No OMIM disease ID*
IRF6	*99,4*	*93*	*100*	*100*	*Popliteal pterygium syndrome 1, 119500* *van der Woude syndrome, 119300*
IRF7	*100*	*99,8*	*100*	*100*	*?Immunodeficiency 39, 616345*
IRF8	*98,7*	*96*	*100*	*100*	*Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893* *Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990*
IRF9	*100*	*99,9*	*100*	*100*	*Immunodeficiency 65, susceptibility to viral infections, 618648*

IRGM	*100*	*100*	*100*	*100*	*No OMIM disease ID*
IRS4	*100*	*100*	*100*	*100*	*Hypothyroidism, congenital, nongoitrous, 9, 301035*
IRX1	*87,1*	*81,1*	*98,7*	*94,7*	*No OMIM disease ID*
IRX5	*100*	*98,8*	*100*	*99,9*	*Hamamy syndrome, 611174*
ISCA1	*89,5*	*76,2*	*95,1*	*95,1*	*Multiple mitochondrial dysfunctions syndrome 5, 617613*
ISCA2	*99,8*	*96,5*	*100*	*100*	*Multiple mitochondrial dysfunctions syndrome 4, 616370*
ISCU	*100*	*100*	*100*	*100*	*Myopathy with lactic acidosis, hereditary, 255125*
ISG15	*100*	*100*	*100*	*100*	*Immunodeficiency 38, 616126*
ITCH	*91,5*	*90,8*	*95,3*	*93,1*	*Autoimmune disease, multisystem, with facial dysmorphism, 613385*
ITGA2	*99,1*	*96,8*	*100*	*100*	*No OMIM disease ID*
ITGA2B	*99,7*	*97,2*	*100*	*100*	*Glanzmann thrombasthenia 1, 273800* *Bleeding disorder, platelet-type, 16, autosomal dominant, 187800* *Thrombocytopenia, neonatal alloimmune, BAK antigen related,*
ITGA3	*99,6*	*97,9*	*100*	*100*	*Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748*
ITGA6	*99,8*	*98*	*100*	*100*	*Epidermolysis bullosa, junctional, with pyloric stenosis, 226730*
ITGA7	*99,7*	*97,9*	*100*	*100*	*Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204*
ITGA8	*99,9*	*99,3*	*100*	*100*	*Renal hypodysplasia/aplasia 1, 191830*
ITGB2	*97,2*	*97,2*	*97,2*	*97,2*	*Leukocyte adhesion deficiency, 116920*
ITGB3	*100*	*99*	*100*	*100*	*Bleeding disorder, platelet-type, 24, autosomal dominant, 619271* *Glanzmann thrombasthenia 2, 619267* *Thrombocytopenia, neonatal alloimmune,* *Purpura, posttransfusion,*
ITGB4	*98,7*	*96,4*	*100*	*100*	*Epidermolysis bullosa of hands and feet, 131800* *Epidermolysis bullosa, junctional, with pyloric atresia, 226730* *Epidermolysis bullosa, junctional, non-Herlitz type, 226650*
ITGB6	*97,4*	*95,7*	*100*	*100*	*Amelogenesis imperfecta, type IH, 616221*
ITK	*99,8*	*98,6*	*100*	*100*	*Lymphoproliferative syndrome 1, 613011*
ITM2B	*99,9*	*99,5*	*100*	*99,9*	*?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079* *Dementia, familial British, 176500* *Dementia, familial Danish, 117300*
ITPA	*100*	*100*	*100*	*100*	*Developmental and epileptic encephalopathy 35, 616647*

ITPR1	*100*	*99,5*	*100*	*100*	*Gillespie syndrome, 206700* *Spinocerebellar ataxia 29, congenital nonprogressive, 117360* *Spinocerebellar ataxia 15, 606658*
ITPR2	*99,7*	*98,3*	*100*	*100*	*?Anhidrosis, isolated, with normal sweat glands, 106190*
ITPR3	*100*	*99,6*	*100*	*100*	*No OMIM disease ID*
ITSN1	*98,7*	*96*	*100*	*100*	*No OMIM disease ID*
ITSN2	*97,8*	*95,4*	*100*	*100*	*No OMIM disease ID*
IVD	*100*	*99,9*	*100*	*100*	*Isovaleric acidemia, 243500*
IVNS1ABP	*99,1*	*96,4*	*100*	*100*	*Immunodeficiency 70, 618969*
IYD	*99,4*	*94,5*	*100*	*100*	*Thyroid dyshormonogenesis 4, 274800*
JAG1	*97,8*	*96,7*	*100*	*100*	*?Deafness, congenital heart defects, and posterior embryotoxon, 617992* *Alagille syndrome 1, 118450* *Tetralogy of Fallot, 187500*
JAGN1	*100*	*100*	*100*	*99,2*	*Neutropenia, severe congenital, 6, autosomal recessive, 616022*
JAK1	*99,9*	*99,4*	*100*	*100*	*Autoinflammation, immune dysregulation, and eosinophilia, 618999*
JAK2	*97,6*	*95,2*	*100*	*99,9*	*Myelofibrosis, somatic, 254450* *Erythrocytosis, somatic, 133100* *Leukemia, acute myeloid, somatic, 601626* *Thrombocythemia 3, 614521* *Polycythemia vera, somatic, 263300*
JAK3	*99,5*	*97,6*	*100*	*100*	*SCID, autosomal recessive, T-negative/B-positive type, 600802*
JAM2	*99,9*	*99,7*	*92,3*	*92,3*	*Basal ganglia calcification, idiopathic, 8, autosomal recessive, 618824*
JAM3	*100*	*100*	*100*	*100*	*Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730*
JARID2	*100*	*100*	*100*	*100*	*No OMIM disease ID*
JMJD1C	*99,5*	*98,9*	*100*	*100*	*No OMIM disease ID*
JPH1	*100*	*99,8*	*100*	*100*	*?Charcot-Marie-Tooth disease, axonal, autosomal dominant, type 2K, 607831*
JPH2	*96*	*85,6*	*100*	*100*	*Cardiomyopathy, dilated, 2E, 619492* *Cardiomyopathy, hypertrophic, 17, 613873*
JPH3	*99,9*	*99,3*	*100*	*100*	*Huntington disease-like 2, 606438*
JUP	*100*	*99,8*	*100*	*100*	*Naxos disease, 601214* *?Arrhythmogenic right ventricular dysplasia 12, 611528*
KALRN	*99,9*	*99,4*	*100*	*100*	*No OMIM disease ID*

KANK1	*100*	*99,9*	*100*	*100*	*Cerebral palsy, spastic quadriplegic, 2, 612900*
KANK2	*100*	*100*	*100*	*100*	*Nephrotic syndrome, type 16, 617783* *Palmoplantar keratoderma and woolly hair, 616099*
KANSL1	*99,8*	*98,2*	*100*	*100*	*Koolen-De Vries syndrome, 610443*
KARS1	*99,9*	*98,9*	*100*	*100*	*Deafness, autosomal recessive 89, 613916* *Leukoencephalopathy, progressive, infantile-onset, with or without deafness, 619147* *?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641* *Deafness, congenital, and adult-onset progressive leukoencephalopathy, 619196*
KAT5	*99,8*	*97,8*	*100*	*100*	*Neurodevelopmental disorder wtih dysmorphic facies, sleep disturbance, and brain abnormalities, 619103*
KAT6A	*100*	*99,2*	*100*	*100*	*Arboleda-Tham syndrome, 616268*
KAT6B	*99,4*	*98*	*100*	*100*	*SBBYSS syndrome, 603736* *Genitopatellar syndrome, 606170*
KAT8	*100*	*98,6*	*100*	*100*	*Li-Ghorgani-Weisz-Hubshman syndrome, 618974*
KATNB1	*100*	*100*	*100*	*100*	*Lissencephaly 6, with microcephaly, 616212*
KIAA0556	*100*	*99,6*	*100*	*100*	*Joubert syndrome 26, 616784*
KBTBD13	*99,9*	*96,7*	*100*	*100*	*Nemaline myopathy 6, autosomal dominant, 609273*
KCNA1	*100*	*100*	*100*	*100*	*Episodic ataxia/myokymia syndrome, 160120*
KCNA2	*100*	*99,6*	*100*	*100*	*Developmental and epileptic encephalopathy 32, 616366*
KCNA4	*100*	*100*	*100*	*100*	*Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284*
KCNA5	*100*	*99,3*	*100*	*100*	*Atrial fibrillation, familial, 7, 612240*
KCNB1	*100*	*99,7*	*100*	*100*	*Developmental and epileptic encephalopathy 26, 616056*
KCNC1	*100*	*100*	*100*	*100*	*Epilepsy, progressive myoclonic 7, 616187*
KCNC3	*76,9*	*64,2*	*94,7*	*88,4*	*Spinocerebellar ataxia 13, 605259*
KCND2	*100*	*100*	*100*	*100*	*No OMIM disease ID*
KCND3	*100*	*98,6*	*100*	*100*	*Spinocerebellar ataxia 19, 607346* *Brugada syndrome 9, 616399*
KCNE1	*100*	*100*	*100*	*100*	*Jervell and Lange-Nielsen syndrome 2, 612347* *Long QT syndrome 5, 613695*
KCNE2	*100*	*96,1*	*100*	*100*	*Long QT syndrome 6, 613693* *Atrial fibrillation, familial, 4, 611493*
KCNE3	*100*	*100*	*100*	*100*	*?Brugada syndrome 6, 613119*
KCNE4	*80,5*	*80,5*	*100*	*100*	*No OMIM disease ID*

KCNE5	*98,5*	*93,5*	*100*	*100*	*No OMIM disease ID*
KCNH1	*98,7*	*98,5*	*98,7*	*98,7*	*Zimmermann-Laband syndrome 1, 135500* *Temple-Baraitser syndrome, 611816*
KCNH2	*95,9*	*92,1*	*100*	*100*	*Short QT syndrome 1, 609620* *Long QT syndrome 2, 613688*
KCNJ1	*100*	*100*	*100*	*100*	*Bartter syndrome, type 2, 241200*
KCNJ10	*89,2*	*88,5*	*100*	*100*	*Enlarged vestibular aqueduct, digenic, 600791* *SESAME syndrome, 612780*
KCNJ11	*100*	*100*	*100*	*100*	*Diabetes, permanent neonatal 2, with or without neurologic features, 618856* *Maturity-onset diabetes of the young, type 13, 616329* *Diabetes mellitus, transient neonatal 3, 610582* *Hyperinsulinemic hypoglycemia, familial, 2, 601820*
KCNJ13	*100*	*100*	*100*	*100*	*Snowflake vitreoretinal degeneration, 193230* *Leber congenital amaurosis 16, 614186*
KCNJ16	*98,3*	*98,3*	*100*	*100*	*Hypokalemic tubulopathy and deafness, 619406*
KCNJ2	*100*	*100*	*100*	*100*	*Atrial fibrillation, familial, 9, 613980* *Andersen syndrome, 170390* *Short QT syndrome 3, 609622*
KCNJ5	*100*	*100*	*100*	*100*	*Long QT syndrome 13, 613485* *Hyperaldosteronism, familial, type III, 613677*
KCNJ6	*100*	*100*	*100*	*100*	*Keppen-Lubinsky syndrome, 614098*
KCNJ8	*100*	*100*	*100*	*100*	*No OMIM disease ID*
KCNK3	*97,9*	*95,1*	*100*	*100*	*Pulmonary hypertension, primary, 4, 615344*
KCNK4	*99,6*	*98,1*	*100*	*100*	*Facial dysmorphism, hypertrichosis, epilepsy, intellectual/developmental delay, and gingival overgrowth syndrome, 618381*
KCNK9	*97,3*	*97,3*	*97,3*	*97,3*	*Birk-Barel syndrome, 612292*
KCNMA1	*94*	*93*	*100*	*100*	*Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446* *Cerebellar atrophy, developmental delay, and seizures, 617643* *Liang-Wang syndrome, 618729*
KCNN2	*73,7*	*72,7*	*100*	*100*	*No OMIM disease ID*
KCNN3	*100*	*99,7*	*100*	*100*	*Zimmermann-Laband syndrome 3, 618658*
KCNN4	*100*	*99,8*	*100*	*100*	*Dehydrated hereditary stomatocytosis 2, 616689*
KCNQ1	*93,5*	*90,6*	*99,9*	*99,4*	*Short QT syndrome 2, 609621* *Atrial fibrillation, familial, 3, 607554*

					Long QT syndrome 1, 192500 *Jervell and Lange-Nielsen syndrome, 220400*
KCNQ1OT1	*NC*	*NC*	*NC*	*NC*	*Beckwith-Wiedemann syndrome, 130650*
KCNQ2	*91,2*	*89,1*	*100*	*100*	*Developmental and epileptic encephalopathy 7, 613720* *Seizures, benign neonatal, 1, 121200* *Myokymia, 121200*
KCNQ3	*99,8*	*97,2*	*99,5*	*98,7*	*Seizures, benign neonatal, 2, 121201*
KCNQ4	*97,3*	*96,2*	*97,1*	*95,1*	*Deafness, autosomal dominant 2A, 600101*
KCNQ5	*97,4*	*95,3*	*100*	*100*	*Mental retardation, autosomal dominant 46, 617601*
KCNT1	*95,8*	*95*	*98,6*	*97,1*	*Developmental and epileptic encephalopathy 14, 614959* *Epilepsy nocturnal frontal lobe, 5, 615005*
KCNT2	*99,1*	*96,9*	*100*	*99,9*	*Developmental and epileptic encephalopathy 57, 617771*
KCNV2	*100*	*99,7*	*100*	*100*	*Retinal cone dystrophy 3B, 610356*
KCTD1	*97,5*	*89,6*	*99,9*	*99,2*	*Scalp-ear-nipple syndrome, 181270*
KCTD17	*100*	*97,6*	*100*	*100*	*Dystonia 26, myoclonic, 616398*
KCTD3	*99,7*	*99,4*	*100*	*100*	*No OMIM disease ID*
KCTD7	*95*	*95*	*100*	*100*	*Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726*
KDELR2	*100*	*100*	*100*	*100*	*Osteogenesis imperfecta 21, 619131*
KDF1	*99,8*	*98*	*100*	*100*	*?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/nail type, 617337*
KDM1A	*97,7*	*95,1*	*100*	*100*	*Cleft palate, psychomotor retardation, and distinctive facial features, 616728*
KDM3B	*97,8*	*96,2*	*100*	*100*	*Diets-Jongmans syndrome, 618846*
KDM4B	*99,8*	*98,6*	*100*	*100*	*Intellectual developmental disorder, autosomal dominant 65, 619320*
KDM5B	*93,4*	*90,7*	*94,7*	*93,3*	*Mental retardation, autosomal recessive 65, 618109*
KDM5C	*99,7*	*97,7*	*100*	*100*	*Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type, 300534*
KDM6A	*94,2*	*85,9*	*100*	*99,9*	*Kabuki syndrome 2, 300867*
KDM6B	*98,6*	*97,4*	*100*	*100*	*Neurodevelopmental disorder with coarse facies and mild distal skeletal abnormalities, 618505*
KDR	*100*	*99,7*	*100*	*100*	*Hemangioma, capillary infantile, somatic, 602089*
KDSR	*99,8*	*99,5*	*100*	*100*	*Erythrokeratodermia variabilis et progressiva 4, 617526*
KERA	*100*	*100*	*100*	*100*	*Cornea plana 2, autosomal recessive, 217300*
KHDC3L	*100*	*99,8*	*100*	*100*	*Hydatidiform mole, recurrent, 2, 614293*

KIAA0586	*97,1*	*92*	*95,8*	*95,7*	*Short-rib thoracic dysplasia 14 with polydactyly, 616546* *Joubert syndrome 23, 616490*
KIAA0753	*99,9*	*98,9*	*100*	*100*	*?Orofaciodigital syndrome XV, 617127* *?Joubert syndrome 38, 619476* *Short-rib thoracic dysplasia 21 without polydactyly, 619479*
KIAA0825	*99*	*96,8*	*100*	*100*	*Polydactyly, postaxial, type A10, 618498*
KIAA1109	*99,8*	*99*	*100*	*100*	*Alkuraya-Kucinskas syndrome, 617822*
KIAA1549	*97,8*	*96,3*	*99*	*98,4*	*Retinitis pigmentosa 86, 618613*
KIDINS220	*100*	*99,9*	*100*	*100*	*Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296* *Ventriculomegaly and arthrogryposis, 619501*
KIF11	*96,8*	*93,1*	*100*	*100*	*Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950*
KIF14	*99,2*	*96,8*	*100*	*99,9*	*Microcephaly 20, primary, autosomal recessive, 617914* *?Meckel syndrome 12, 616258*
KIF1A	*97,4*	*95,3*	*98*	*98*	*NESCAV syndrome, 614255* *Neuropathy, hereditary sensory, type IIC, 614213* *Spastic paraplegia 30, autosomal dominant, 610357* *Spastic paraplegia 30, autosomal recessive, 610357*
KIF1B	*99,9*	*99,2*	*100*	*100*	*Pheochromocytoma, 171300* *Charcot-Marie-Tooth disease, type 2A1, 118210*
KIF1C	*100*	*99,7*	*100*	*100*	*Spastic ataxia 2, autosomal recessive, 611302*
KIF20A	*100*	*99,1*	*100*	*100*	*?Cardiomyopathy, familial restrictive, 6, 619433*
KIF21A	*99,7*	*98,9*	*100*	*100*	*Fibrosis of extraocular muscles, congenital, 3B, 135700* *Fibrosis of extraocular muscles, congenital, 1, 135700*
KIF21B	*98,1*	*96,8*	*100*	*100*	*No OMIM disease ID*
KIF22	*100*	*99,6*	*100*	*100*	*Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546*
KIF23	*98,2*	*94*	*100*	*99,9*	*No OMIM disease ID*
KIF2A	*99*	*95,3*	*100*	*99,8*	*Cortical dysplasia, complex, with other brain malformations 3, 615411*
KIF3B	*99,9*	*98,9*	*100*	*100*	*Retinitis pigmentosa 89, 618955*
KIF4A	*98,4*	*92,2*	*100*	*100*	*?Intellectual developmental disorder, X-linked 100, 300923*
KIF5A	*100*	*99,8*	*100*	*100*	*Myoclonus, intractable, neonatal, 617235* *Spastic paraplegia 10, autosomal dominant, 604187*
KIF5C	*99,6*	*97*	*99,8*	*99,8*	*Cortical dysplasia, complex, with other brain malformations 2, 615282*
KIF7	*93,6*	*91,9*	*99,7*	*98,6*	*Joubert syndrome 12, 200990* *Acrocallosal syndrome, 200990*

					?Hydrolethalus syndrome 2, 614120 *?Al-Gazali-Bakalinova syndrome, 607131*
KIFBP	*96,1*	*96*	*96,1*	*96,1*	*Goldberg-Shprintzen megacolon syndrome, 609460*
KIRREL1	*100*	*99,9*	*100*	*100*	*Nephrotic syndrome, type 23, 619201*
KIRREL3	*99,9*	*98,8*	*100*	*100*	*No OMIM disease ID*
KISS1	*100*	*98,2*	*100*	*100*	*?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842*
KISS1R	*100*	*99,6*	*100*	*100*	*Hypogonadotropic hypogonadism 8 with or without anosmia, 614837* *?Precocious puberty, central, 1, 176400*
KIT	*100*	*99,4*	*100*	*100*	*Gastrointestinal stromal tumor, familial, 606764* *Mastocytosis, cutaneous, 154800* *Piebaldism, 172800* *Germ cell tumors, somatic, 273300* *Mastocytosis, systemic, somatic, 154800* *Leukemia, acute myeloid, somatic, 601626*
KITLG	*99,6*	*98,1*	*100*	*100*	*Hyperpigmentation with or without hypopigmentation, 145250* *Deafness, autosomal dominant 69, unilateral or asymmetric, 616697*
KIZ	*99,8*	*98,4*	*100*	*100*	*Retinitis pigmentosa 69, 615780*
KL	*98,5*	*97,5*	*98,7*	*97,9*	*?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994*
KLB	*100*	*99,9*	*100*	*100*	*No OMIM disease ID*
KLC2	*99,2*	*98*	*100*	*100*	*Spastic paraplegia, optic atrophy, and neuropathy, 609541*
KLF1	*100*	*99,6*	*100*	*100*	*Blood group--Lutheran inhibitor, 111150* *Dyserythropoietic anemia, congenital, type IV, 613673*
KLF10	*100*	*99,9*	*100*	*100*	*No OMIM disease ID*
KLF11	*100*	*98,6*	*100*	*100*	*Maturity-onset diabetes of the young, type VII, 610508*
KLF6	*100*	*100*	*100*	*100*	*Gastric cancer, somatic, 613659* *Prostate cancer, somatic, 176807*
KLF7	*100*	*98,9*	*100*	*100*	*No OMIM disease ID*
KLHL10	*100*	*100*	*100*	*100*	*Spermatogenic failure 11, 615081*
KLHL15	*99,9*	*99,1*	*100*	*100*	*Intellectual developmental disorder, X-linked 103, 300982*
KLHL24	*100*	*100*	*100*	*100*	*Epidermolysis bullosa simplex, generalized, with scarring and hair loss, 617294*
KLHL3	*100*	*98,9*	*100*	*100*	*Pseudohypoaldosteronism, type IID, 614495*
KLHL40	*100*	*100*	*100*	*100*	*Nemaline myopathy 8, autosomal recessive, 615348*
KLHL41	*99,9*	*99,4*	*100*	*100*	*Nemaline myopathy 9, 615731*

KLHL7	*99,8*	*99,7*	*100*	*100*	*Retinitis pigmentosa 42, 612943* *PERCHING syndrome, 617055*
KLHL9	*100*	*99,9*	*100*	*100*	*No OMIM disease ID*
KLK4	*100*	*100*	*100*	*100*	*Amelogenesis imperfecta, type IIA1, 204700*
KLKB1	*99,8*	*99,3*	*100*	*99,9*	*Fletcher factor (prekallikrein) deficiency, 612423*
KLLN	*100*	*100*	*100*	*100*	*Cowden syndrome 4, 615107*
KMT2A	*100*	*99,7*	*100*	*99,8*	*Wiedemann-Steiner syndrome, 605130*
KMT2B	*96,2*	*94*	*98,5*	*97,8*	*Dystonia 28, childhood-onset, 617284*
KMT2C	*91,9*	*90,3*	*100*	*100*	*Kleefstra syndrome 2, 617768*
KMT2D	*99,9*	*99*	*100*	*100*	*Kabuki syndrome 1, 147920*
KMT2E	*99,6*	*98,1*	*100*	*100*	*O'Donnell-Luria-Rodan syndrome, 618512*
KMT5B	*99,7*	*98,5*	*100*	*100*	*Mental retardation, autosomal dominant 51, 617788*
KNG1	*100*	*100*	*100*	*100*	*Angioedema, hereditary, 6, 619363*
KNL1	*99,1*	*97,3*	*98,9*	*98,7*	*Microcephaly 4, primary, autosomal recessive, 604321*
KNSTRN	*100*	*100*	*100*	*100*	*?Roifman-Chitayat syndrome, digenic, 613328*
KPTN	*100*	*100*	*100*	*100*	*Mental retardation, autosomal recessive 41, 615637*
KRAS	*99*	*97,8*	*100*	*100*	*Gastric cancer, somatic, 137215* *Oculoectodermal syndrome, somatic, 600268* *Breast cancer, somatic, 114480* *Noonan syndrome 3, 609942* *RAS-associated autoimmune leukoproliferative disorder, 614470* *Arteriovenous malformation of the brain, somatic, 108010* *Lung cancer, somatic, 211980* *Pancreatic carcinoma, somatic, 260350* *Leukemia, acute myeloid, somatic, 601626* *Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200* *Cardiofaciocutaneous syndrome 2, 615278* *Bladder cancer, somatic, 109800*
KREMEN1	*99,3*	*96,1*	*99,4*	*97,7*	*Ectodermal dysplasia 13, hair/tooth type, 617392*
KRIT1	*99,5*	*98,8*	*100*	*99,9*	*Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations, 116860* *Cerebral cavernous malformations-1, 116860* *Cavernous malformations of CNS and retina, 116860*

KRT1	*97,8*	*94,2*	*100*	*100*	*Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602* *Epidermolytic hyperkeratosis, 113800* *Palmoplantar keratoderma, nonepidermolytic, 600962* *Keratosis palmoplantaris striata III, 607654* *Palmoplantar keratoderma, epidermolytic, 144200* *Ichthyosis histrix, Curth-Macklin type, 146590*
KRT10	*99,9*	*99,1*	*100*	*100*	*Epidermolytic hyperkeratosis, 113800* *Ichthyosis with confetti, 609165* *Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602*
KRT12	*99,9*	*98,8*	*100*	*100*	*Meesmann corneal dystrophy 1, 122100*
KRT13	*100*	*99,5*	*100*	*100*	*White sponge nevus 2, 615785*
KRT14	*89,6*	*81,6*	*100*	*100*	*Epidermolysis bullosa simplex, recessive 1, 601001* *Epidermolysis bullosa simplex, Weber-Cockayne type, 131800* *Dermatopathia pigmentosa reticularis, 125595* *Epidermolysis bullosa simplex, Koebner type, 131900* *Naegeli-Franceschetti-Jadassohn syndrome, 161000* *Epidermolysis bullosa simplex, Dowling-Meara type, 131760*
KRT16	*75,4*	*55,8*	*100*	*100*	*Palmoplantar keratoderma, nonepidermolytic, focal, 613000* *Pachyonychia congenita 1, 167200*
KRT17	*39,3*	*24,1*	*100*	*100*	*Steatocystoma multiplex, 184500* *Pachyonychia congenita 2, 167210*
KRT18	*83,2*	*66,1*	*100*	*100*	*Cirrhosis, cryptogenic, 215600*
KRT2	*100*	*99,4*	*100*	*100*	*Ichthyosis bullosa of Siemens, 146800*
KRT25	*100*	*100*	*100*	*100*	*Woolly hair, autosomal recessive 3, 616760*
KRT3	*100*	*99,7*	*100*	*100*	*Meesmann corneal dystrophy 2, 618767*
KRT4	*100*	*98,8*	*100*	*100*	*White sponge nevus 1, 193900*
KRT5	*100*	*99,7*	*100*	*100*	*Dowling-Degos disease 1, 179850* *Epidermolysis bullosa simplex-MP, 131960* *Epidermolysis bullosa simplex, Koebner type, 131900* *Epidermolysis bullosa simplex-MCR, 609352* *Epidermolysis bullosa simplex, Weber-Cockayne type, 131800* *Epidermolysis bullosa simplex, recessive 1, 601001* *Epidermolysis bullosa simplex, Dowling-Meara type, 131760*
KRT6A	*92,8*	*87,9*	*100*	*100*	*Pachyonychia congenita 3, 615726*
KRT6B	*93,8*	*88,5*	*100*	*100*	*Pachyonychia congenita 4, 615728*

KRT6C	*88,3*	*80,9*	*99,9*	*99,8*	*Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735*
KRT71	*100*	*99,9*	*100*	*100*	*?Hypotrichosis 13, 615896*
KRT74	*100*	*100*	*100*	*100*	*Woolly hair, autosomal dominant, 194300* *?Hypotrichosis 3, 613981* *?Ectodermal dysplasia 7, hair/nail type, 614929*
KRT75	*100*	*99,9*	*100*	*100*	*No OMIM disease ID*
KRT8	*90,3*	*69,5*	*100*	*100*	*Cirrhosis, cryptogenic, 215600*
KRT81	*99,7*	*95,5*	*100*	*100*	*Monilethrix, 158000*
KRT83	*95,9*	*84,3*	*100*	*100*	*Monilethrix, 158000* *Erythrokeratodermia variabilis et progressiva 5, 617756*
KRT85	*99,1*	*94,5*	*100*	*100*	*Ectodermal dysplasia 4, hair/nail type, 602032*
KRT86	*99,6*	*96,2*	*100*	*100*	*Monilethrix, 158000*
KRT9	*99,7*	*96,8*	*100*	*100*	*Palmoplantar keratoderma, epidermolytic, 144200*
KY	*100*	*99,6*	*100*	*100*	*Myopathy, myofibrillar, 7, 617114*
KYNU	*99,5*	*96,7*	*100*	*100*	*?Hydroxykynureninuria, 236800* *Vertebral, cardiac, renal, and limb defects syndrome 2, 617661*
L1CAM	*100*	*98,8*	*100*	*100*	*MASA syndrome, 303350* *Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000* *Corpus callosum, partial agenesis of, 304100* *CRASH syndrome, 303350* *Hydrocephalus with Hirschsprung disease, 307000* *Hydrocephalus due to aqueductal stenosis, 307000*
L2HGDH	*98,9*	*96,4*	*100*	*100*	*L-2-hydroxyglutaric aciduria, 236792*
LACC1	*99,7*	*98,4*	*100*	*100*	*Juvenile arthritis, 618795*
LACTB	*99,4*	*95,1*	*100*	*99,9*	*No OMIM disease ID*
LAGE3	*96,1*	*84,3*	*100*	*100*	*Galloway-Mowat syndrome 2, X-linked, 301006*
LAMA1	*99,9*	*99,3*	*100*	*100*	*Poretti-Boltshauser syndrome, 615960*
LAMA2	*99,9*	*99,1*	*100*	*100*	*Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138* *Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855*
LAMA3	*99,9*	*99,3*	*100*	*100*	*Laryngoonychocutaneous syndrome, 245660* *Epidermolysis bullosa, junctional, Herlitz type, 226700* *Epidermolysis bullosa, generalized atrophic benign, 226650*
LAMA4	*100*	*99,7*	*100*	*100*	*Cardiomyopathy, dilated, 1JJ, 615235*

LAMA5	*98,5*	*96,3*	*100*	*99,9*	*No OMIM disease ID*
LAMB1	*100*	*99,6*	*100*	*100*	*Lissencephaly 5, 615191*
LAMB2	*99,9*	*99,3*	*100*	*100*	*Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199* *Pierson syndrome, 609049*
LAMB3	*99,9*	*98,8*	*100*	*100*	*Epidermolysis bullosa, junctional, non-Herlitz type, 226650* *Epidermolysis bullosa, junctional, Herlitz type, 226700* *Amelogenesis imperfecta, type IA, 104530*
LAMC2	*99,4*	*96,8*	*100*	*100*	*Epidermolysis bullosa, junctional, non-Herlitz type, 226650* *Epidermolysis bullosa, junctional, Herlitz type, 226700*
LAMC3	*98,9*	*97,5*	*100*	*99,8*	*Cortical malformations, occipital, 614115*
LAMP2	*99,3*	*96*	*100*	*99,7*	*Danon disease, 300257*
LAMTOR2	*100*	*100*	*100*	*100*	*Immunodeficiency due to defect in MAPBP-interacting protein, 610798*
LAPTM5	*96,9*	*91,6*	*100*	*100*	*No OMIM disease ID*
LARGE1	*100*	*99,7*	*100*	*100*	*Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840* *Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154*
LARP7	*86,6*	*75,4*	*100*	*99,9*	*Alazami syndrome, 615071*
LARS1	*99,4*	*97,2*	*100*	*99,9*	*?Infantile liver failure syndrome 1, 615438*
LARS2	*100*	*100*	*100*	*100*	*Perrault syndrome 4, 615300* *Hydrops, lactic acidosis, and sideroblastic anemia, 617021*
LAS1L	*99,5*	*95,7*	*100*	*100*	*Wilson-Turner syndrome, 309585*
LAT	*100*	*99,4*	*100*	*100*	*Immunodeficiency 52, 617514*
LBR	*97,9*	*91*	*100*	*100*	*Pelger-Huet anomaly, 169400* *Pelger-Huet anomaly with mild skeletal anomalies, 618019* *?Reynolds syndrome, 613471* *Greenberg skeletal dysplasia, 215140*
LBX1	*100*	*100*	*100*	*100*	*No OMIM disease ID*
LCA5	*99,6*	*97,9*	*100*	*100*	*Leber congenital amaurosis 5, 604537*
LCAT	*98,8*	*93,3*	*100*	*100*	*Fish-eye disease, 136120* *Norum disease, 245900*
LCK	*98,2*	*96,1*	*100*	*100*	*?Immunodeficiency 22, 615758*
LCP2	*99,6*	*95,4*	*100*	*100*	*?Immunodeficiency 81, 619374*
LCT	*99,6*	*97,4*	*100*	*100*	*Lactase deficiency, congenital, 223000*

LDB3	*95,4*	*94,8*	*100*	*100*	*Left ventricular noncompaction 3, 601493* *Cardiomyopathy, hypertrophic, 24, 601493* *Myopathy, myofibrillar, 4, 609452* *Cardiomyopathy, dilated, 1C, with or without LVNC, 601493*
LDHA	*94,4*	*89,3*	*100*	*100*	*Glycogen storage disease XI, 612933*
LDHB	*90,5*	*77,9*	*100*	*100*	*No OMIM disease ID*
LDHD	*100*	*99,6*	*100*	*100*	*D-lactic aciduria with susceptibility to gout, 245450*
LDLR	*99,8*	*98*	*100*	*100*	*LDL cholesterol level QTL2, 143890* *Hypercholesterolemia, familial, 1, 143890*
LDLRAP1	*98,9*	*94*	*100*	*100*	*Hypercholesterolemia, familial, 4, 603813*
LEF1	*100*	*99,9*	*100*	*100*	*Sebaceous tumors, somatic,*
LEFTY2	*94,3*	*84,3*	*100*	*100*	*No OMIM disease ID*
LEMD2	*99,9*	*96,1*	*100*	*100*	*Marbach-Rustad progeroid syndrome, 619322* *Cataract 46, juvenile-onset, 212500*
LEMD3	*99,5*	*97,8*	*100*	*100*	*Buschke-Ollendorff syndrome, 166700* *Osteopoikilosis with or without melorheostosis, 166700*
LEP	*100*	*99,6*	*100*	*100*	*Obesity, morbid, due to leptin deficiency, 614962*
LEPR	*94,1*	*92,3*	*94,6*	*94,5*	*Obesity, morbid, due to leptin receptor deficiency, 614963*
LFNG	*88,6*	*86,5*	*92*	*87,3*	*Spondylocostal dysostosis 3, autosomal recessive, 609813*
LGI1	*98,3*	*98,1*	*100*	*100*	*Epilepsy, familial temporal lobe, 1, 600512*
LGI4	*99,7*	*97,9*	*100*	*100*	*Arthrogryposis multiplex congenita 1, neurogenic, with myelin defect, 617468*
LHB	*91,7*	*42,8*	*100*	*100*	*Hypogonadotropic hypogonadism 23 with or without anosmia, 228300*
LHCGR	*96,6*	*92,4*	*100*	*100*	*Leydig cell adenoma, somatic, with precocious puberty, 176410* *Leydig cell hypoplasia with pseudohermaphroditism, 238320* *Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320* *Luteinizing hormone resistance, female, 238320* *Precocious puberty, male, 176410*
LHFPL5	*100*	*100*	*100*	*100*	*Deafness, autosomal recessive 67, 610265*
LHX1	*100*	*99,8*	*100*	*100*	*No OMIM disease ID*
LHX3	*96,6*	*96,2*	*100*	*100*	*Pituitary hormone deficiency, combined, 3, 221750*
LHX4	*100*	*100*	*100*	*100*	*Pituitary hormone deficiency, combined, 4, 262700*
LIAS	*99,8*	*98,9*	*100*	*100*	*Hyperglycinemia, lactic acidosis, and seizures, 614462*

LIFR	*99,3*	*97,8*	*100*	*99,9*	*Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559*
LIG1	*100*	*99,7*	*100*	*100*	*No OMIM disease ID*
LIG4	*99,8*	*99,3*	*100*	*100*	*LIG4 syndrome, 606593*
LIM2	*100*	*99,8*	*100*	*100*	*Cataract 19, multiple types, 615277*
LIMS2	*94,1*	*92,7*	*100*	*99,7*	*?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827*
LINGO1	*100*	*100*	*100*	*100*	*Mental retardation, autosomal recessive 64, 618103*
LINS1	*99,8*	*98,8*	*100*	*100*	*Mental retardation, autosomal recessive 27, 614340*
LIPA	*96,9*	*94,6*	*95,2*	*95,2*	*Wolman disease, 278000* *Cholesteryl ester storage disease, 278000*
LIPC	*100*	*99,4*	*100*	*100*	*Hepatic lipase deficiency, 614025*
LIPE	*100*	*99,2*	*100*	*100*	*Lipodystrophy, familial partial, type 6, 615980*
LIPH	*100*	*99*	*100*	*100*	*Hypotrichosis 7, 604379* *Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379*
LIPN	*99,6*	*99,4*	*100*	*100*	*Ichthyosis, congenital, autosomal recessive 8, 613943*
LIPT1	*99,7*	*99,5*	*100*	*100*	*Lipoyltransferase 1 deficiency, 616299*
LIPT2	*98,4*	*82,4*	*100*	*100*	*Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668*
LITAF	*97,1*	*91,9*	*100*	*100*	*Charcot-Marie-Tooth disease, type 1C, 601098*
LMAN1	*99,8*	*98,5*	*100*	*100*	*Combined factor V and VIII deficiency, 227300*
LMAN2L	*100*	*99,5*	*100*	*100*	*?Mental retardation, autosomal recessive, 52, 616887*
LMBR1	*98,1*	*94,7*	*98,7*	*98,5*	*Triphalangeal thumb, type I, 174500* *Syndactyly, type IV, 186200* *Laurin-Sandrow syndrome, 135750* *Hypoplastic or aplastic tibia with polydactyly, 188740* *Polydactyly, preaxial type II, 174500* *Acheiropody, 200500* *Triphalangeal thumb-polysyndactyly syndrome, 174500*
LMBRD1	*94,1*	*89,1*	*96,1*	*95,7*	*Methylmalonic aciduria and homocystinuria, cblF type, 277380*
LMBRD2	*99,1*	*95,6*	*100*	*100*	*No OMIM disease ID*
LMF1	*100*	*99,7*	*100*	*100*	*Lipase deficiency, combined, 246650*
LMNA	*96,1*	*90,6*	*100*	*100*	*Mandibuloacral dysplasia, 248370* *Heart-hand syndrome, Slovenian type, 610140* *Cardiomyopathy, dilated, 1A, 115200*

					Restrictive dermopathy, lethal, 275210 *Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516* *Charcot-Marie-Tooth disease, type 2B1, 605588* *Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350* *Hutchinson-Gilford progeria, 176670* *Lipodystrophy, familial partial, type 2, 151660* *Muscular dystrophy, congenital, 613205* *Malouf syndrome, 212112*
LMNB1	*99,9*	*99,2*	*100*	*100*	*Leukodystrophy, adult-onset, autosomal dominant, 169500* *Microcephaly 26, primary, autosomal dominant, 619179*
LMNB2	*98,3*	*95,5*	*97,9*	*96,7*	*Microcephaly 27, primary, autosomal dominant, 619180* *?Epilepsy, progressive myoclonic, 9, 616540*
LMOD1	*100*	*99,8*	*100*	*100*	*?Megacystis-microcolon-intestinal hypoperistalsis syndrome 3, 619362*
LMOD3	*99,8*	*99*	*100*	*100*	*Nemaline myopathy 10, 616165*
LMX1A	*100*	*99,9*	*100*	*100*	*Deafness, autosomal dominant 7, 601412*
LMX1B	*99,3*	*96,8*	*100*	*100*	*Focal segmental glomerulosclerosis 10, 256020* *Nail-patella syndrome, 161200*
LNPK	*97,2*	*92,1*	*93,3*	*93,2*	*Neurodevelopmental disorder with epilepsy and hypoplasia of the corpus callosum, 618090*
LONP1	*100*	*99,9*	*100*	*100*	*CODAS syndrome, 600373*
LORICRIN	*99,5*	*81,1*	*100*	*100*	*Vohwinkel syndrome with ichthyosis, 604117*
LOX	*99,8*	*99,6*	*100*	*100*	*Aortic aneurysm, familial thoracic 10, 617168*
LOXHD1	*99,9*	*98,7*	*100*	*100*	*Deafness, autosomal recessive 77, 613079*
LOXL3	*100*	*99,8*	*100*	*100*	*No OMIM disease ID*
LPAR6	*99,3*	*96,8*	*100*	*100*	*Hypotrichosis 8, 278150* *Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150*
LPIN1	*99,4*	*97,2*	*100*	*100*	*Myoglobinuria, acute recurrent, autosomal recessive, 268200*
LPIN2	*99,9*	*99,7*	*100*	*100*	*Majeed syndrome, 609628*
LPL	*100*	*100*	*100*	*100*	*Lipoprotein lipase deficiency, 238600* *Combined hyperlipidemia, familial, 144250*
LPP	*100*	*99,9*	*100*	*100*	*Leukemia, acute myeloid, 601626* *Lipoma,*
LRAT	*100*	*100*	*100*	*100*	*Leber congenital amaurosis 14, 613341* *Retinal dystrophy, early-onset severe, 613341* *Retinitis pigmentosa, juvenile, 613341*

LRBA	*99,9*	*99,7*	*100*	*100*	*Immunodeficiency, common variable, 8, with autoimmunity, 614700*
LRIF1	*100*	*99,9*	*100*	*100*	*?Facioscapulohumeral muscular dystrophy 3, digenic, 619477*
LRIG2	*99,8*	*99,2*	*100*	*100*	*Urofacial syndrome 2, 615112*
LRIG3	*100*	*99,4*	*100*	*99,7*	*No OMIM disease ID*
LRIT3	*94,1*	*92,2*	*100*	*100*	*Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058*
LRMDA	*97,4*	*95,5*	*99,6*	*99,6*	*Albinism, oculocutaneous, type VII, 615179*
LRP1	*99,8*	*99,1*	*100*	*100*	*?Keratosis pilaris atrophicans, 604093*
LRP12	*100*	*99,9*	*100*	*100*	*Oculopharyngodistal myopathy 1, 164310*
LRP2	*100*	*99,8*	*100*	*100*	*Donnai-Barrow syndrome, 222448*
LRP4	*99,1*	*98,4*	*100*	*100*	*?Myasthenic syndrome, congenital, 17, 616304* *Sclerosteosis 2, 614305* *Cenani-Lenz syndactyly syndrome, 212780*
LRP5	*99,2*	*98,2*	*99,8*	*99,2*	*Osteopetrosis, autosomal dominant 1, 607634* *Hyperostosis, endosteal, 144750* *Osteosclerosis, 144750* *Polycystic liver disease 4 with or without kidney cysts, 617875* *Osteoporosis-pseudoglioma syndrome, 259770* *Exudative vitreoretinopathy 4, 601813* *van Buchem disease, type 2, 607636*
LRP6	*100*	*99,2*	*100*	*100*	*Tooth agenesis, selective, 7, 616724*
LRPAP1	*100*	*99,9*	*100*	*100*	*Myopia 23, autosomal recessive, 615431*
LRPPRC	*99,7*	*99,3*	*100*	*99,9*	*Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111*
LRRC10	*100*	*100*	*100*	*100*	*No OMIM disease ID*
LRRC32	*100*	*100*	*100*	*100*	*Cleft palate, proliferative retinopathy, and developmental delay, 619074*
LRRC56	*99,8*	*99*	*100*	*100*	*Ciliary dyskinesia, primary, 39, 618254*
LRRC8A	*100*	*99,7*	*100*	*100*	*?Agammaglobulinemia 5, 613506*
LRRK1	*98,7*	*97,5*	*100*	*100*	*No OMIM disease ID*
LRRK2	*99,5*	*97*	*100*	*100*	*No OMIM disease ID*
LRSAM1	*100*	*100*	*100*	*100*	*Charcot-Marie-Tooth disease, axonal, type 2P, 614436*
LRTOMT	*100*	*99,2*	*100*	*100*	*Deafness, autosomal recessive 63, 611451*
LSM11	*99,9*	*97,6*	*100*	*98,9*	*?Aicardi-Goutieres syndrome 8, 619486*

LSS	*100*	*99,4*	*100*	*100*	*Hypotrichosis 14, 618275* *Cataract 44, 616509* *Alopecia-mental retardation syndrome 4, 618840*
LTBP1	*99,2*	*97,4*	*100*	*99,9*	*Cutis laxa, autosomal recessive, type IIE, 619451*
LTBP2	*99,8*	*98,9*	*100*	*100*	*Glaucoma 3, primary congenital, D, 613086* *Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750* *?Weill-Marchesani syndrome 3, recessive, 614819*
LTBP3	*99,8*	*98,6*	*100*	*99,9*	*Dental anomalies and short stature, 601216* *Geleophysic dysplasia 3, 617809*
LTBP4	*99,9*	*98,1*	*100*	*100*	*Cutis laxa, autosomal recessive, type IC, 613177*
LTC4S	*76,4*	*69*	*100*	*100*	*No OMIM disease ID*
LYRM4	*66,7*	*65,6*	*66,3*	*66,3*	*?Combined oxidative phosphorylation deficiency 19, 615595*
LYRM7	*95,6*	*86,5*	*100*	*99,2*	*Mitochondrial complex III deficiency, nuclear type 8, 615838*
LYST	*99,4*	*97,8*	*100*	*100*	*Chediak-Higashi syndrome, 214500*
LYZ	*100*	*99,9*	*100*	*100*	*Amyloidosis, renal, 105200*
LZTFL1	*99,7*	*99,4*	*100*	*99,9*	*Bardet-Biedl syndrome 17, 615994*
LZTR1	*100*	*99,9*	*100*	*100*	*Noonan syndrome 2, 605275* *Noonan syndrome 10, 616564*
LZTS1	*100*	*99,6*	*100*	*100*	*Esophageal squamous cell carcinoma, somatic, 133239*
M1AP	*100*	*99,8*	*100*	*100*	*Spermatogenic failure 48, 619108*
MAB21L1	*100*	*100*	*100*	*100*	*Cerebellar, ocular, craniofacial, and genital syndrome, 618479*
MAB21L2	*100*	*100*	*100*	*100*	*Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877*
MACF1	*99,6*	*98,7*	*100*	*100*	*Lissencephaly 9 with complex brainstem malformation, 618325*
MAD1L1	*99,9*	*98,4*	*100*	*100*	*Prostate cancer, somatic, 176807* *Lymphoma, somatic,*
MAD2L2	*100*	*99,9*	*100*	*100*	*?Fanconi anemia, complementation group V, 617243*
MADD	*100*	*99,2*	*100*	*100*	*Neurodevelopmental disorder with dysmorphic facies, impaired speech and hypotonia, 619005* *DEEAH syndrome, 619004*
MAF	*84,3*	*78,9*	*88,6*	*82,2*	*Cataract 21, multiple types, 610202* *Ayme-Gripp syndrome, 601088*
MAFA	*93,5*	*72,7*	*98,6*	*94*	*Insulinomatosis and diabetes mellitus, 147630*
MAFB	*100*	*99,8*	*100*	*100*	*Duane retraction syndrome 3, 617041* *Multicentric carpotarsal osteolysis syndrome, 166300*

MAG	*100*	*100*	*100*	*100*	*Spastic paraplegia 75, autosomal recessive, 616680*
MAGED2	*99,5*	*97,6*	*100*	*99,8*	*Bartter syndrome, type 5, antenatal, transient, 300971*
MAGEL2	*94,1*	*89,1*	*100*	*100*	*Schaaf-Yang syndrome, 615547*
MAGI2	*94,2*	*91*	*94,7*	*93,3*	*Nephrotic syndrome, type 15, 617609*
MAGT1	*98*	*94,5*	*98,7*	*98,4*	*Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853* *Congenital disorder of glycosylation, type Icc, 301031*
MAK	*99,7*	*97,9*	*100*	*100*	*Retinitis pigmentosa 62, 614181*
MAL2	*100*	*99*	*100*	*99,7*	*No OMIM disease ID*
MALT1	*90,9*	*87,7*	*100*	*99,9*	*Immunodeficiency 12, 615468*
MAML2	*100*	*99,8*	*100*	*100*	*Mucoepidermoid salivary gland carcinoma,*
MAMLD1	*99,7*	*97,8*	*100*	*100*	*Hypospadias 2, X-linked, 300758*
MAN1B1	*100*	*99,7*	*100*	*100*	*Rafiq syndrome, 614202*
MAN2B1	*99,6*	*97,4*	*100*	*100*	*Mannosidosis, alpha-, types I and II, 248500*
MAN2B2	*99,9*	*99,2*	*100*	*100*	*No OMIM disease ID*
MANBA	*87,1*	*84,9*	*100*	*99,9*	*Mannosidosis, beta, 248510*
MAOA	*100*	*99,8*	*99,4*	*97,9*	*Brunner syndrome, 300615*
MAP1B	*98,9*	*97,1*	*100*	*100*	*Periventricular nodular heterotopia 9, 618918*
MAP1LC3B2	*100*	*100*	*100*	*100*	*No OMIM disease ID*
MAP2K1	*99,6*	*96,1*	*100*	*100*	*Cardiofaciocutaneous syndrome 3, 615279* *Melorheostosis, isolated, somatic mosaic, 155950*
MAP2K2	*98,5*	*95,3*	*100*	*100*	*Cardiofaciocutaneous syndrome 4, 615280*
MAP3K1	*97*	*93*	*100*	*99,9*	*46XY sex reversal 6, 613762*
MAP3K14	*100*	*99,8*	*100*	*100*	*No OMIM disease ID*
MAP3K20	*99,9*	*99,2*	*100*	*99,9*	*Centronuclear myopathy 6 with fiber-type disproportion, 617760* *Split-foot malformation with mesoaxial polydactyly, 616890*
MAP3K7	*99,8*	*99,6*	*100*	*100*	*Frontometaphyseal dysplasia 2, 617137* *Cardiospondylocarpofacial syndrome, 157800*
MAP3K8	*100*	*99,9*	*100*	*99,9*	*Lung cancer, somatic, 211980*
MAP4K4	*100*	*98,6*	*100*	*100*	*No OMIM disease ID*
MAPK1	*100*	*99,9*	*100*	*99,6*	*Noonan syndrome 13, 619087*
MAPK8	*99,9*	*99,7*	*100*	*100*	*No OMIM disease ID*

MAPK8IP3	*99,4*	*99*	*100*	*100*	*Neurodevelopmental disorder with or without variable brain abnormalities, 618443*
MAPKAPK3	*100*	*98,6*	*100*	*100*	*?Macular dystrophy, patterned, 3, 617111*
MAPKAPK5	*92,2*	*92,2*	*100*	*100*	*No OMIM disease ID*
MAPKBP1	*100*	*100*	*100*	*100*	*Nephronophthisis 20, 617271*
MAPRE2	*100*	*98,5*	*100*	*100*	*Symmetric circumferential skin creases, congenital, 2, 616734*
MAPT	*99,9*	*98,9*	*100*	*100*	*Supranuclear palsy, progressive, 601104* *Supranuclear palsy, progressive atypical, 260540* *Dementia, frontotemporal, with or without parkinsonism, 600274* *Pick disease, 172700*
MARCHF6	*99,3*	*98*	*100*	*99,9*	*Epilepsy, familial adult myoclonic, 3, 613608*
MARK3	*99,6*	*97,6*	*100*	*99,9*	*?Visual impairment and progressive phthisis bulbi, 618283*
MARS1	*99*	*96,1*	*100*	*100*	*Interstitial lung and liver disease, 615486* *Charcot-Marie-Tooth disease, axonal, type 2U, 616280*
MARS2	*100*	*100*	*100*	*100*	*?Combined oxidative phosphorylation deficiency 25, 616430* *Spastic ataxia 3, autosomal recessive, 611390*
MARVELD2	*98,5*	*94,9*	*100*	*100*	*Deafness, autosomal recessive 49, 610153*
MASP1	*100*	*99,6*	*100*	*100*	*3MC syndrome 1, 257920*
MASP2	*100*	*99,3*	*100*	*100*	*MASP2 deficiency, 613791*
MAST1	*99,8*	*99,3*	*100*	*100*	*Mega-corpus-callosum syndrome with cerebellar hypoplasia and cortical malformations, 618273*
MASTL	*99,9*	*99,7*	*100*	*100*	*No OMIM disease ID*
MAT1A	*99,9*	*98,5*	*100*	*100*	*Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850* *Methionine adenosyltransferase deficiency, autosomal recessive, 250850*
MAT2A	*99,2*	*93,8*	*100*	*100*	*No OMIM disease ID*
MATN3	*84,8*	*84,1*	*100*	*100*	*Spondyloepimetaphyseal dysplasia, Borochowitz-Cormier-Daire type, 608728* *Epiphyseal dysplasia, multiple, 5, 607078*
MATR3	*96,5*	*90,6*	*100*	*100*	*Amyotrophic lateral sclerosis 21, 606070*
MAX	*99,8*	*97,7*	*100*	*100*	*No OMIM disease ID*
MBD5	*99,9*	*99,8*	*100*	*100*	*Mental retardation, autosomal dominant 1, 156200*
MBL2	*99,9*	*99,4*	*100*	*100*	*No OMIM disease ID*
MBOAT7	*100*	*99,3*	*100*	*100*	*Mental retardation, autosomal recessive 57, 617188*
MBTPS1	*99,4*	*97,3*	*100*	*100*	*?Spondyloepiphyseal dysplasia, Kondo-Fu type, 618392*

MBTPS2	*99,9*	*98,5*	*100*	*100*	*Keratosis follicularis spinulosa decalvans, X-linked, 308800* *Osteogenesis imperfecta, type XIX, 301014* *IFAP syndrome with or without BRESHECK syndrome, 308205* *?Olmsted syndrome, X-linked, 300918*
MC1R	*100*	*100*	*100*	*100*	*No OMIM disease ID*
MC2R	*99,7*	*97,4*	*100*	*100*	*Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200*
MC4R	*100*	*100*	*100*	*100*	*Obesity (BMIQ20), 618406*
MCC	*100*	*99,7*	*100*	*100*	*Colorectal cancer, somatic, 114500*
MCCC1	*99,9*	*98,7*	*100*	*100*	*3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200*
MCCC2	*99,9*	*99,1*	*100*	*100*	*3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210*
MCEE	*100*	*100*	*100*	*100*	*Methylmalonyl-CoA epimerase deficiency, 251120*
MCFD2	*99,4*	*94,9*	*100*	*100*	*Factor V and factor VIII, combined deficiency of, 613625*
MCIDAS	*98,4*	*95,2*	*100*	*100*	*Ciliary dyskinesia, primary, 42, 618695*
MCM10	*100*	*99,5*	*100*	*100*	*Immunodeficiency 80 with or without cardiomyopathy, 619313*
MCM2	*100*	*99,9*	*100*	*100*	*?Deafness, autosomal dominant 70, 616968*
MCM3AP	*99,9*	*99,1*	*100*	*100*	*Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124*
MCM4	*95,3*	*95*	*95,5*	*95,5*	*Immunodeficiency 54, 609981*
MCM5	*100*	*99,7*	*100*	*100*	*?Meier-Gorlin syndrome 8, 617564*
MCM6	*100*	*100*	*100*	*100*	*Lactase persistence/nonpersistence, 223100*
MCM8	*99,9*	*98,8*	*94,4*	*94,3*	*?Premature ovarian failure 10, 612885*
MCM9	*99,9*	*99*	*100*	*100*	*Ovarian dysgenesis 4, 616185*
MCOLN1	*99,8*	*98,8*	*100*	*100*	*Mucolipidosis IV, 252650*
MCPH1	*99,8*	*98,6*	*100*	*100*	*Microcephaly 1, primary, autosomal recessive, 251200*
MCTP2	*99,4*	*97,7*	*100*	*100*	*No OMIM disease ID*
MCUR1	*99,1*	*91,3*	*100*	*99,9*	*No OMIM disease ID*
MDH1	*99,7*	*99,1*	*100*	*100*	*?Developmental and epileptic encephalopathy 88, 618959*
MDH2	*98*	*98*	*100*	*100*	*Developmental and epileptic encephalopathy 51, 617339*
MDM2	*92,1*	*88,2*	*92,6*	*92,6*	*?Lessel-Kubisch syndrome, 618681*
MDM4	*100*	*98,8*	*100*	*100*	*?Bone marrow failure syndrome 6, 618849*
MECOM	*100*	*99,6*	*100*	*100*	*Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738*

MECP2	*99,8*	*97,5*	*100*	*99,7*	*Intellectual developmental disorder, X-linked, syndromic 13, 300055* *Rett syndrome, atypical, 312750* *Encephalopathy, neonatal severe, 300673* *Intellectual developmental disorder, X-linked syndromic, Lubs type, 300260* *Rett syndrome, 312750* *Rett syndrome, preserved speech variant, 312750*
MECR	*100*	*98,7*	*100*	*100*	*Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282*
MED12	*99,3*	*94,1*	*100*	*100*	*Lujan-Fryns syndrome, 309520* *Ohdo syndrome, X-linked, 300895* *Opitz-Kaveggia syndrome, 305450*
MED12L	*100*	*99,9*	*100*	*100*	*Nizon-Isidor syndrome, 618872*
MED13	*99,9*	*99,6*	*100*	*100*	*Intellectual developmental disorder 61, 618009*
MED13L	*100*	*99,5*	*100*	*100*	*Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789* *Transposition of the great arteries, dextro-looped 1, 608808*
MED17	*95,8*	*92,4*	*100*	*100*	*Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668*
MED23	*99,9*	*99*	*100*	*100*	*Mental retardation, autosomal recessive 18, 614249*
MED25	*100*	*99,9*	*100*	*99,9*	*Basel-Vanagait-Smirin-Yosef syndrome, 616449*
MED27	*79*	*65,9*	*84,7*	*84,7*	*Neurodevelopmental disorder with spasticity, cataracts, and cerebellar hypoplasia, 619286*
MEF2C	*99,7*	*95,7*	*100*	*100*	*Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443*
MEFV	*99,6*	*97,6*	*96,4*	*96,4*	*Neutrophilic dermatosis, acute febrile, 608068* *Familial Mediterranean fever, AR, 249100* *Familial Mediterranean fever, AD, 134610*
MEGF10	*100*	*99,9*	*100*	*100*	*Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399* *Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399*
MEGF8	*100*	*99,2*	*100*	*100*	*Carpenter syndrome 2, 614976*
MEI1	*99,9*	*98,7*	*100*	*100*	*Hydatidiform mole, recurrent, 3, 618431*
MEIOB	*99,3*	*98,6*	*100*	*99,9*	*?Spermatogenic failure 22, 617706*
MEIS2	*100*	*99,7*	*100*	*100*	*Cleft palate, cardiac defects, and mental retardation, 600987*
MEN1	*96,2*	*94,1*	*100*	*100*	*Multiple endocrine neoplasia 1, 131100* *Lipoma, somatic,* *Angiofibroma, somatic,* *Carcinoid tumor of lung,* *Adrenal adenoma, somatic,* *Parathyroid adenoma, somatic,*

MEOX1	*100*	*98*	*100*	*100*	*Klippel-Feil syndrome 2, 214300*
MERTK	*99,4*	*98,6*	*99,1*	*99,1*	*Retinitis pigmentosa 38, 613862*
MESD	*100*	*98,8*	*100*	*100*	*Osteogenesis imperfecta, type XX, 618644*
MESP2	*95,7*	*90,6*	*97,5*	*97,5*	*Spondylocostal dysostosis 2, autosomal recessive, 608681*
MET	*100*	*99,4*	*100*	*100*	*Renal cell carcinoma, papillary, 1, familial and somatic, 605074* *Hepatocellular carcinoma, childhood type, somatic, 114550* *?Deafness, autosomal recessive 97, 616705*
EEF1AKNMT	*99,4*	*97,8*	*100*	*100*	*No OMIM disease ID*
METTL23	*100*	*100*	*100*	*100*	*Mental retardation, autosomal recessive 44, 615942*
METTL5	*98,9*	*97,4*	*99,8*	*97,6*	*Intellectual developmental disorder, autosomal recessive 72, 618665*
MFAP5	*100*	*98,7*	*100*	*100*	*Aortic aneurysm, familial thoracic 9, 616166*
MFF	*93,9*	*89,4*	*100*	*100*	*Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086*
MFN2	*100*	*99,8*	*100*	*100*	*Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260* *Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087* *Hereditary motor and sensory neuropathy VIA, 601152*
MFRP	*100*	*100*	*100*	*100*	*Microphthalmia, isolated 5, 611040* *Nanophthalmos 2, 609549*
MFSD2A	*99,5*	*97,3*	*100*	*100*	*Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities, 616486*
MFSD8	*99,6*	*99,4*	*100*	*100*	*Macular dystrophy with central cone involvement, 616170* *Ceroid lipofuscinosis, neuronal, 7, 610951*
MGAT2	*100*	*99,9*	*100*	*100*	*Congenital disorder of glycosylation, type IIa, 212066*
MGME1	*100*	*99,9*	*100*	*100*	*Mitochondrial DNA depletion syndrome 11, 615084*
MGP	*98,7*	*93,6*	*100*	*100*	*Keutel syndrome, 245150*
MIA3	*99,8*	*99,3*	*100*	*100*	*?Ondontochondrodysplasia 2 with hearing loss and diabetes, 619269*
MIB1	*100*	*99,6*	*100*	*99,9*	*Left ventricular noncompaction 7, 615092*
MICOS13	*100*	*98,9*	*100*	*99,9*	*Combined oxidative phosphorylation deficiency 37, 618329*
MICU1	*97,3*	*92,2*	*100*	*100*	*Myopathy with extrapyramidal signs, 615673*
MICU2	*96,7*	*92,5*	*100*	*99,9*	*No OMIM disease ID*
MID1	*99,6*	*97,7*	*100*	*100*	*Opitz GBBB syndrome, type I, 300000*
MID2	*99,8*	*98,6*	*99,9*	*99,8*	*?Intellectual developmental disorder, X-linked 101, 300928*
MIEF2	*100*	*99,1*	*100*	*100*	*?Combined oxidative phosphorylation deficiency 49, 619024*

MINPP1	*99,7*	*99,3*	*100*	*99,9*	*No OMIM disease ID*
MIP	*99,7*	*94,1*	*100*	*100*	*Cataract 15, multiple types, 615274*
MIPEP	*99,5*	*97,1*	*100*	*100*	*Combined oxidative phosphorylation deficiency 31, 617228*
MIR140	*NC*	*NC*	*NC*	*NC*	*Spondyloepiphyseal dysplasia, Nishimura type, 618618*
MIR17HG	*NC*	*NC*	*NC*	*NC*	*No OMIM disease ID*
MIR184	*NC*	*NC*	*NC*	*NC*	*EDICT syndrome, 614303*
MIR204	*NC*	*NC*	*NC*	*NC*	*?Retinal dystrophy and iris coloboma with or without cataract, 616722*
MIR96	*NC*	*NC*	*NC*	*NC*	*Deafness, autosomal dominant 50, 613074*
MITF	*100*	*99,9*	*100*	*100*	*Waardenburg syndrome, type 2A, 193510* *Tietz albinism-deafness syndrome, 103500* *Waardenburg syndrome/ocular albinism, digenic, 103470* *COMMAD syndrome, 617306*
MKKS	*100*	*100*	*100*	*100*	*McKusick-Kaufman syndrome, 236700* *Bardet-Biedl syndrome 6, 605231*
MKRN3	*96*	*96*	*96*	*96*	*Precocious puberty, central, 2, 615346*
MKS1	*99,4*	*96,3*	*100*	*100*	*Bardet-Biedl syndrome 13, 615990* *Meckel syndrome 1, 249000* *Joubert syndrome 28, 617121*
MLC1	*100*	*98,8*	*100*	*100*	*Megalencephalic leukoencephalopathy with subcortical cysts, 604004*
MLH1	*100*	*99,9*	*100*	*100*	*Colorectal cancer, hereditary nonpolyposis, type 2, 609310* *Muir-Torre syndrome, 158320* *Mismatch repair cancer syndrome 1, 276300*
MLH3	*100*	*99,9*	*100*	*100*	*Colorectal cancer, somatic, 114500* *Colorectal cancer, hereditary nonpolyposis, type 7, 614385*
MLIP	*99,9*	*98,8*	*100*	*100*	*No OMIM disease ID*
MLLT10	*96,8*	*95,4*	*97,1*	*97,1*	*Leukemia, acute myeloid, 601626*
MLLT6	*99,4*	*95,7*	*100*	*100*	*No OMIM disease ID*
MLPH	*100*	*98,5*	*100*	*100*	*Griscelli syndrome, type 3, 609227*
MLYCD	*96,8*	*92,5*	*100*	*99,4*	*Malonyl-CoA decarboxylase deficiency, 248360*
MMAA	*100*	*100*	*100*	*100*	*Methylmalonic aciduria, vitamin B12-responsive, cblA type, 251100*
MMAB	*100*	*99,9*	*100*	*100*	*Methylmalonic aciduria, vitamin B12-responsive, cblB type, 251110*
MMACHC	*100*	*100*	*100*	*100*	*Methylmalonic aciduria and homocystinuria, cblC type, 277400*

MMADHC	*91,6*	*81,3*	*89,7*	*89,7*	*Methylmalonic aciduria, cblD type, variant 2, 277410* *Methylmalonic aciduria and homocystinuria, cblD type, 277410* *Homocystinuria, cblD type, variant 1, 277410*
MME	*99,7*	*98,6*	*98*	*97,9*	*?Spinocerebellar ataxia 43, 617018* *Charcot-Marie-Tooth disease, axonal, type 2T, 617017*
MMGT1	*99,2*	*98,4*	*100*	*99,8*	*No OMIM disease ID*
MMP1	*99,8*	*98,8*	*100*	*100*	*COPD, rate of decline of lung function in, 606963*
MMP13	*93,6*	*92,2*	*92,4*	*92,3*	*?Spondyloepimetaphyseal dysplasia, Missouri type, 602111* *Metaphyseal anadysplasia 1, 602111* *Metaphyseal dysplasia, Spahr type, 250400*
MMP14	*100*	*99,4*	*100*	*100*	*?Winchester syndrome, 277950*
MMP19	*100*	*98,9*	*100*	*100*	*Cavitary optic disc anomalies, 611543*
MMP2	*100*	*100*	*100*	*100*	*Multicentric osteolysis, nodulosis, and arthropathy, 259600*
MMP20	*99,9*	*99,3*	*100*	*100*	*Amelogenesis imperfecta, type IIA2, 612529*
MMP21	*99,8*	*99,2*	*100*	*100*	*Heterotaxy, visceral, 7, autosomal, 616749*
MMP9	*99,6*	*96,8*	*100*	*100*	*Metaphyseal anadysplasia 2, 613073*
MMUT	*99,7*	*98,2*	*100*	*100*	*Methylmalonic aciduria, mut(0) type, 251000*
MN1	*100*	*99,7*	*100*	*100*	*CEBALID syndrome, 618774* *Meningioma, 607174*
MNS1	*99,2*	*96*	*100*	*99,9*	*Heterotaxy, visceral, 9, autosomal, with male infertility, 618948*
MNX1	*70,8*	*59,2*	*85,7*	*79*	*Currarino syndrome, 176450*
MOCOS	*99,9*	*97,8*	*100*	*100*	*Xanthinuria, type II, 603592*
MOCS1	*98,9*	*95,5*	*100*	*100*	*Molybdenum cofactor deficiency A, 252150*
MOCS2	*99,4*	*99,4*	*100*	*100*	*Molybdenum cofactor deficiency B, 252160*
MOG	*99,9*	*99,5*	*100*	*100*	*?Narcolepsy 7, 614250*
MOGS	*100*	*99,9*	*100*	*100*	*Congenital disorder of glycosylation, type IIb, 606056*
MORC2	*100*	*99,5*	*100*	*100*	*Charcot-Marie-Tooth disease, axonal, type 2Z, 616688* *Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy, 619090*
MPC1	*100*	*99,6*	*100*	*100*	*Mitochondrial pyruvate carrier deficiency, 614741*
MPDU1	*100*	*99,2*	*100*	*100*	*Congenital disorder of glycosylation, type If, 609180*
MPDZ	*99,8*	*98,5*	*100*	*100*	*Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219*
MPEG1	*100*	*100*	*100*	*100*	*Immunodeficiency 77, 619223*

MPI	*100*	*99,5*	*100*	*100*	*Congenital disorder of glycosylation, type Ib, 602579*
MPIG6B	*100*	*99,9*	*100*	*100*	*?Thrombocytopenia, anemia, and myelofibrosis, 617441*
MPL	*100*	*99,8*	*100*	*100*	*Myelofibrosis with myeloid metaplasia, somatic, 254450* *Thrombocythemia 2, 601977* *Thrombocytopenia, congenital amegakaryocytic, 604498*
MPLKIP	*100*	*99,4*	*100*	*100*	*Trichothiodystrophy 4, nonphotosensitive, 234050*
MPO	*99,9*	*98,7*	*100*	*100*	*Myeloperoxidase deficiency, 254600*
MPV17	*100*	*98,7*	*100*	*100*	*Charcot-Marie-Tooth disease, axonal, type 2EE, 618400* *Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810*
MPZ	*85,6*	*81,9*	*100*	*100*	*Charcot-Marie-Tooth disease, type 2I, 607677* *Dejerine-Sottas disease, 145900* *Charcot-Marie-Tooth disease, type 1B, 118200* *Roussy-Levy syndrome, 180800* *Charcot-Marie-Tooth disease, dominant intermediate D, 607791* *Hypomyelinating neuropathy, congenital, 2, 618184* *Charcot-Marie-Tooth disease, type 2J, 607736*
MPZL2	*100*	*99,9*	*100*	*100*	*Deafness, autosomal recessive 111, 618145*
MRAP	*100*	*100*	*100*	*100*	*Glucocorticoid deficiency 2, 607398*
MRAS	*100*	*99,3*	*100*	*100*	*Noonan syndrome 11, 618499*
MRE11	*98,2*	*88,6*	*100*	*100*	*Ataxia-telangiectasia-like disorder 1, 604391*
MRM2	*100*	*98,9*	*98,9*	*98,9*	*?Mitochondrial DNA depletion syndrome 17, 618567*
MRPL12	*100*	*99,1*	*100*	*100*	*?Combined oxidative phosphorylation deficiency 45, 618951*
MRPL24	*100*	*100*	*100*	*100*	*No OMIM disease ID*
MRPL3	*91,7*	*82,1*	*100*	*100*	*Combined oxidative phosphorylation deficiency 9, 614582*
MRPL40	*99,5*	*91,6*	*100*	*100*	*No OMIM disease ID*
MRPL44	*99,5*	*97,4*	*100*	*100*	*?Combined oxidative phosphorylation deficiency 16, 615395*
MRPL57	*100*	*100*	*100*	*100*	*No OMIM disease ID*
MRPS14	*100*	*100*	*100*	*100*	*?Combined oxidative phosphorylation deficiency 38, 618378*
MRPS16	*100*	*98,8*	*100*	*100*	*Combined oxidative phosphorylation deficiency 2, 610498*
MRPS2	*99,6*	*97*	*100*	*100*	*Combined oxidative phosphorylation deficiency 36, 617950*
MRPS22	*99,7*	*98,3*	*100*	*100*	*Ovarian dysgenesis 7, 618117* *Combined oxidative phosphorylation deficiency 5, 611719*

MRPS23	*99,7*	*98,8*	*100*	*100*	*?Combined oxidative phosphorylation defiency 46, 618952*
MRPS25	*100*	*99,8*	*82,7*	*82,7*	*?Combined oxidative phosphorylation deficiency 50, 619025*
MRPS28	*87,6*	*86,6*	*86,6*	*86,6*	*?Combined oxidative phosphorylation deficiency 47, 618958*
MRPS34	*98,6*	*93,3*	*100*	*100*	*Combined oxidative phosphorylation deficiency 32, 617664*
MRPS36	*94*	*75,2*	*100*	*100*	*No OMIM disease ID*
MRPS7	*100*	*100*	*100*	*100*	*?Combined oxidative phosphorylation deficiency 34, 617872*
MRRF	*100*	*100*	*100*	*100*	*No OMIM disease ID*
MRTFA	*91*	*89*	*92,8*	*92,8*	*?Immunodeficiency 66, 618847*
MS4A1	*99,5*	*96,5*	*100*	*99,9*	*?Immunodeficiency, common variable, 5, 613495*
MSH2	*98,5*	*94,5*	*100*	*100*	*Muir-Torre syndrome, 158320* *Colorectal cancer, hereditary nonpolyposis, type 1, 120435* *Mismatch repair cancer syndrome 2, 619096*
MSH3	*97,8*	*97,1*	*100*	*100*	*Familial adenomatous polyposis 4, 617100* *Endometrial carcinoma, somatic, 608089*
MSH4	*98,8*	*96,5*	*100*	*99,5*	*No OMIM disease ID*
MSH5	*100*	*99,5*	*100*	*100*	*?Premature ovarian failure 13, 617442*
MSH6	*100*	*99,3*	*100*	*100*	*Colorectal cancer, hereditary nonpolyposis, type 5, 614350* *Mismatch repair cancer syndrome 3, 619097*
MSL2	*100*	*100*	*100*	*100*	*No OMIM disease ID*
MSL3	*83,4*	*75,3*	*97,8*	*96,6*	*Basilicata-Akhtar syndrome, 301032*
MSMO1	*93,1*	*86,8*	*100*	*100*	*Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834*
MSN	*98,2*	*92*	*100*	*100*	*Immunodeficiency 50, 300988*
MSR1	*99,7*	*99,6*	*100*	*100*	*Barrett esophagus/esophageal adenocarcinoma, 614266*
MSRB3	*99,8*	*99,8*	*100*	*100*	*Deafness, autosomal recessive 74, 613718*
MSTN	*100*	*99,9*	*100*	*100*	*?Muscle hypertrophy, 614160*
MSTO1	*99*	*96,3*	*100*	*100*	*Myopathy, mitochondrial, and ataxia, 617675*
MSX1	*97,7*	*92,6*	*100*	*100*	*Tooth agenesis, selective, 1, with or without orofacial cleft, 106600* *Ectodermal dysplasia 3, Witkop type, 189500* *Orofacial cleft 5, 608874*
MSX2	*100*	*98*	*100*	*100*	*Parietal foramina with cleidocranial dysplasia, 168550* *Craniosynostosis 2, 604757* *Parietal foramina 1, 168500*

MTAP	*98,3*	*91,8*	*100*	*100*	*Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250*
MTFMT	*99,9*	*99,5*	*100*	*100*	*Combined oxidative phosphorylation deficiency 15, 614947* *Mitochondrial complex I deficiency, nuclear type 27, 618248*
MTHFD1	*99,9*	*98,4*	*100*	*100*	*Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780*
MTHFR	*97,3*	*95,9*	*100*	*100*	*Homocystinuria due to MTHFR deficiency, 236250*
MTHFS	*75*	*75*	*100*	*100*	*Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367*
MTM1	*98,7*	*92*	*100*	*99,7*	*Myotubular myopathy, X-linked, 310400*
MTMR2	*99,5*	*98,4*	*100*	*100*	*Charcot-Marie-Tooth disease, type 4B1, 601382*
MTO1	*90,9*	*88,8*	*92,8*	*91,4*	*Combined oxidative phosphorylation deficiency 10, 614702*
MTOR	*99,9*	*98,9*	*100*	*100*	*Focal cortical dysplasia, type II, somatic, 607341* *Smith-Kingsmore syndrome, 616638*
MTPAP	*99,1*	*94,1*	*100*	*100*	*?Spastic ataxia 4, autosomal recessive, 613672*
MTR	*100*	*99,9*	*100*	*100*	*Homocystinuria-megaloblastic anemia, cblG complementation type, 250940*
C12orf65	*99*	*94,5*	*100*	*100*	*Spastic paraplegia 55, autosomal recessive, 615035* *Combined oxidative phosphorylation deficiency 7, 613559*
MTRR	*99,8*	*98,4*	*100*	*100*	*Homocystinuria-megaloblastic anemia, cbl E type, 236270*
MTTP	*99,9*	*99,2*	*100*	*100*	*Abetalipoproteinemia, 200100*
MTX2	*98,2*	*88,5*	*100*	*100*	*Mandibuloacral dysplasia progeroid syndrome, 619127*
MUC1	*91*	*81,4*	*100*	*100*	*Tubulointerstitial kidney disease, autosomal dominant, 2, 174000*
MUSK	*100*	*99,9*	*100*	*100*	*Fetal akinesia deformation sequence 1, 208150* *Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325*
MUTYH	*100*	*100*	*100*	*100*	*Adenomas, multiple colorectal, 608456* *Gastric cancer, somatic, 613659*
MVD	*99,7*	*97,5*	*100*	*100*	*Porokeratosis 7, multiple types, 614714*
MVK	*91,4*	*90,5*	*90,5*	*90,5*	*Hyper-IgD syndrome, 260920* *Porokeratosis 3, multiple types, 175900* *Mevalonic aciduria, 610377*
MXI1	*99,3*	*95,4*	*98,4*	*94,2*	*Prostate cancer, somatic, 176807* *Neurofibrosarcoma, somatic,*
MYBPC1	*99,8*	*99,1*	*100*	*99,9*	*Myopathy, congenital, with tremor, 618524* *Lethal congenital contracture syndrome 4, 614915* *Arthrogryposis, distal, type 1B, 614335*

MYBPC3	*99,8*	*97,6*	*100*	*100*	*Cardiomyopathy, hypertrophic, 4, 115197* *Cardiomyopathy, dilated, 1MM, 615396* *Left ventricular noncompaction 10, 615396*
MYBPHL	*99,5*	*96,6*	*100*	*100*	*No OMIM disease ID*
MYC	*65,4*	*63,7*	*100*	*100*	*Burkitt lymphoma, somatic, 113970*
MYCN	*100*	*100*	*99,2*	*96,2*	*Feingold syndrome 1, 164280*
MYD88	*100*	*99,5*	*100*	*100*	*Macroglobulinemia, Waldenstrom, somatic, 153600* *Immunodeficiency 68, 612260*
MYF5	*100*	*100*	*100*	*100*	*Ophthalmoplegia, external, with rib and vertebral anomalies, 618155*
MYH11	*100*	*99,7*	*100*	*100*	*Megacystis-microcolon-intestinal hypoperistalsis syndrome 2, 619351* *Aortic aneurysm, familial thoracic 4, 132900* *Visceral myopathy 2, 619350*
MYH14	*98,2*	*93,3*	*100*	*100*	*?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369* *Deafness, autosomal dominant 4A, 600652*
MYH2	*99,9*	*99,1*	*100*	*100*	*Proximal myopathy and ophthalmoplegia, 605637*
MYH3	*99,9*	*98,4*	*100*	*100*	*Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A, 178110* *Contractures, pterygia, and spondylocarpotarsal fusion syndrome 1B, 618469* *Arthrogryposis, distal, type 2B3 (Sheldon-Hall), 618436* *Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700*
MYH6	*99,2*	*96,1*	*100*	*100*	*Atrial septal defect 3, 614089* *Cardiomyopathy, dilated, 1EE, 613252* *Cardiomyopathy, hypertrophic, 14, 613251*
MYH7	*99,1*	*96,7*	*100*	*100*	*Laing distal myopathy, 160500* *Cardiomyopathy, hypertrophic, 1, 192600* *Left ventricular noncompaction 5, 613426* *Cardiomyopathy, dilated, 1S, 613426* *Scapuloperoneal syndrome, myopathic type, 181430* *Myopathy, myosin storage, autosomal dominant, 608358* *Myopathy, myosin storage, autosomal recessive, 255160*
MYH7B	*98,3*	*94,8*	*100*	*100*	*No OMIM disease ID*
MYH8	*100*	*99,2*	*100*	*100*	*Carney complex variant, 608837* *Trismus-pseudocamptodactyly syndrome, 158300*
MYH9	*99,9*	*98,9*	*100*	*100*	*Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100* *Deafness, autosomal dominant 17, 603622*
MYL1	*99,8*	*99,1*	*100*	*100*	*Myopathy, congenital, with fast-twitch (type II) fiber atrophy, 618414*

MYL2	*94,8*	*81,1*	*99,6*	*97,3*	*Cardiomyopathy, hypertrophic, 10, 608758* *Myopathy, myofibrillar, 12, infantile-onset, with cardiomyopathy, 619424*
MYL3	*100*	*100*	*100*	*100*	*Cardiomyopathy, hypertrophic, 8, 608751*
MYL4	*100*	*100*	*100*	*100*	*?Atrial fibrillation, familial, 18, 617280*
MYL7	*100*	*99,6*	*100*	*100*	*No OMIM disease ID*
MYL9	*100*	*100*	*100*	*100*	*?Megacystis-microcolon-intestinal hypoperistalsis syndrome 4, 619365*
MYLK	*100*	*99,6*	*100*	*100*	*Megacystis-microcolon-intestinal hypoperistalsis syndrome 1, 249210* *Aortic aneurysm, familial thoracic 7, 613780*
MYLK2	*100*	*100*	*100*	*100*	*Cardiomyopathy, hypertrophic, 1, digenic, 192600*
MYLK3	*99,7*	*98,1*	*100*	*100*	*No OMIM disease ID*
MYLPF	*100*	*100*	*100*	*100*	*Arthrogryposis, distal, type 1C, 619110*
MYMK	*100*	*100*	*100*	*100*	*Carey-Fineman-Ziter syndrome, 254940*
MYO15A	*99,1*	*97,6*	*100*	*99,8*	*Deafness, autosomal recessive 3, 600316*
MYO18B	*100*	*99,3*	*100*	*100*	*Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549*
MYO1A	*99,9*	*99,2*	*100*	*100*	*No OMIM disease ID*
MYO1E	*99,9*	*98,6*	*100*	*100*	*Glomerulosclerosis, focal segmental, 6, 614131*
MYO1H	*99,6*	*99,4*	*100*	*100*	*?Central hypoventilation syndrome, congenital, 2, and autonomic dysfunction, 619482*
MYO3A	*99,1*	*95,4*	*100*	*99,9*	*Deafness, autosomal recessive 30, 607101*
MYO5A	*99,6*	*98,3*	*100*	*100*	*Griscelli syndrome, type 1, 214450*
MYO5B	*98,5*	*94,8*	*100*	*100*	*Diarrhea 2, with microvillus atrophy, 251850*
MYO6	*99,1*	*96,3*	*100*	*100*	*Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346* *Deafness, autosomal dominant 22, 606346* *Deafness, autosomal recessive 37, 607821*
MYO7A	*99,7*	*98,3*	*100*	*100*	*Deafness, autosomal recessive 2, 600060* *Usher syndrome, type 1B, 276900* *Deafness, autosomal dominant 11, 601317*
MYO9A	*99,8*	*98,9*	*100*	*100*	*Myasthenic syndrome, congenital, 24, presynaptic, 618198*
MYOC	*99,9*	*98*	*100*	*100*	*Glaucoma 1A, primary open angle, 137750*
MYOCD	*100*	*100*	*100*	*100*	*Megabladder, congenital, 618719*
MYOD1	*100*	*100*	*100*	*100*	*Myopathy, congenital, with diaphragmatic defects, respiratory insufficiency, and dysmorphic facies, 618975*
MYOF	*99,4*	*98,8*	*100*	*100*	*?Angioedema, hereditary, 7, 619366*

MYOM1	*99,7*	*98*	*100*	*100*	*No OMIM disease ID*
MYORG	*100*	*100*	*100*	*100*	*Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317*
MYOT	*100*	*99,2*	*100*	*100*	*Myopathy, myofibrillar, 3, 609200* *Myopathy, spheroid body, 182920*
MYOZ2	*100*	*100*	*100*	*100*	*Cardiomyopathy, hypertrophic, 16, 613838*
MYPN	*100*	*99,5*	*100*	*100*	*Cardiomyopathy, hypertrophic, 22, 615248* *Cardiomyopathy, familial restrictive, 4, 615248* *Cardiomyopathy, dilated, 1KK, 615248* *Nemaline myopathy 11, autosomal recessive, 617336*
MYRF	*99*	*97,8*	*100*	*100*	*Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113* *Cardiac-urogenital syndrome, 618280*
MYSM1	*96,1*	*95,4*	*96,4*	*96,3*	*Bone marrow failure syndrome 4, 618116*
MYT1L	*87*	*86,3*	*90,2*	*90,1*	*Mental retardation, autosomal dominant 39, 616521*
NAA10	*99,8*	*97,9*	*99,9*	*99,9*	*Microphthalmia, syndromic 1, 309800* *Ogden syndrome, 300855*
NAA15	*94,8*	*91,2*	*96,8*	*96,7*	*Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787*
NAA20	*99,9*	*99,3*	*100*	*100*	*No OMIM disease ID*
NACC1	*100*	*100*	*100*	*100*	*Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393*
NADK2	*99,7*	*99,3*	*99,5*	*96,8*	*2,4-dienoyl-CoA reductase deficiency, 616034*
NADSYN1	*100*	*100*	*100*	*100*	*Vertebral, cardiac, renal, and limb defects syndrome 3, 618845*
NAGA	*100*	*100*	*100*	*100*	*Schindler disease, type I, 609241* *Kanzaki disease, 609242* *Schindler disease, type III, 609241*
NAGLU	*93,8*	*91,7*	*99,9*	*98,7*	*?Charcot-Marie-Tooth disease, axonal, type 2V, 616491* *Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920*
NAGS	*99,9*	*97,9*	*100*	*100*	*N-acetylglutamate synthase deficiency, 237310*
NALCN	*99,7*	*98,9*	*99,8*	*99,7*	*Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266* *Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419*
NANOS1	*100*	*98,3*	*96,4*	*89,3*	*Spermatogenic failure 12, 615413*
NANS	*100*	*99,9*	*100*	*100*	*Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442*
NARS1	*100*	*100*	*100*	*100*	*Neurodevelopmental disorder with microcephaly, impaired language, epilepsy, and gait abnormalities, autosomal dominant, 619092*

					Neurodevelopmental disorder with microcephaly, impaired language, and gait abnormalities, autosomal recessive, 619091
NARS2	*97,9*	*97,1*	*100*	*99,9*	*Combined oxidative phosphorylation deficiency 24, 616239* *?Deafness, autosomal recessive 94, 618434*
NAT8L	*100*	*98*	*92,9*	*87,1*	*?N-acetylaspartate deficiency, 614063*
NAXD	*100*	*99,9*	*100*	*100*	*Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321*
NAXE	*100*	*98,6*	*100*	*100*	*Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186*
NBAS	*99,9*	*99,3*	*100*	*100*	*Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800* *Infantile liver failure syndrome 2, 616483*
NBEA	*91,8*	*90,3*	*100*	*100*	*Neurodevelopmental disorder with or without early-onset generalized epilepsy, 619157*
NBEAL2	*99,5*	*99,3*	*100*	*100*	*Gray platelet syndrome, 139090*
NBN	*99,2*	*97,8*	*100*	*99,9*	*Leukemia, acute lymphoblastic, 613065* *Aplastic anemia, 609135* *Nijmegen breakage syndrome, 251260*
NCAPD2	*99,9*	*99*	*100*	*100*	*?Microcephaly 21, primary, autosomal recessive, 617983*
NCAPD3	*99,7*	*98*	*100*	*100*	*Microcephaly 22, primary, autosomal recessive, 617984*
NCAPG2	*99,8*	*99*	*100*	*100*	*Khan-Khan-Katsanis syndrome, 618460*
NCAPH	*100*	*100*	*100*	*100*	*?Microcephaly 23, primary, autosomal recessive, 617985*
NCDN	*100*	*99,9*	*100*	*100*	*Neurodevelopmental disorder with infantile epileptic spasms, 619373*
NCF1	*26*	*25,7*	*100*	*99,9*	*Chronic granulomatous disease 1, autosomal recessive, 233700*
NCF2	*99,8*	*97*	*100*	*100*	*Chronic granulomatous disease 2, autosomal recessive, 233710*
NCF4	*100*	*100*	*100*	*100*	*Chronic granulomatous disease 3, autosomal recessive, 613960*
NCKAP1	*98,5*	*96,1*	*100*	*100*	*No OMIM disease ID*
NCKAP1L	*100*	*99,9*	*100*	*99,9*	*Immunodeficiency 72 with autoinflammation, 618982*
NCOA3	*99*	*95,9*	*100*	*100*	*No OMIM disease ID*
NCOA4	*93,9*	*90,4*	*100*	*100*	*No OMIM disease ID*
NCSTN	*100*	*99,7*	*100*	*100*	*Acne inversa, familial, 1, 142690*
NDE1	*100*	*99,4*	*100*	*100*	*Lissencephaly 4 (with microcephaly), 614019* *?Microhydranencephaly, 605013*
NDN	*98,5*	*90,7*	*100*	*100*	*Prader-Willi syndrome, 176270*
NDNF	*100*	*100*	*100*	*100*	*Hypogonadotropic hypogonadism 25 with anosmia, 618841*

NDP	*100*	*99,7*	*100*	*100*	*Exudative vitreoretinopathy 2, X-linked, 305390* *Norrie disease, 310600*
NDRG1	*100*	*99,9*	*100*	*100*	*Charcot-Marie-Tooth disease, type 4D, 601455*
NDST1	*100*	*100*	*100*	*100*	*Mental retardation, autosomal recessive 46, 616116*
NDUFA1	*99,8*	*99,3*	*100*	*100*	*Mitochondrial complex I deficiency, nuclear type 12, 301020*
NDUFA10	*99,9*	*98,6*	*100*	*100*	*Mitochondrial complex I deficiency, nuclear type 22, 618243*
NDUFA11	*100*	*99,8*	*100*	*99,9*	*Mitochondrial complex I deficiency, nuclear type 14, 618236*
NDUFA12	*99,6*	*99,6*	*100*	*100*	*Mitochondrial complex I deficiency, nuclear type 23, 618244*
NDUFA13	*92,2*	*90*	*100*	*100*	*?Mitochondrial complex I deficiency, nuclear type 28, 618249*
NDUFA2	*100*	*100*	*100*	*100*	*Mitochondrial complex I deficiency, nuclear type 13, 618235*
NDUFA3	*88,7*	*88*	*92,2*	*88,4*	*No OMIM disease ID*
NDUFA4	*99,1*	*96,5*	*100*	*100*	*?Mitochondrial complex IV deficiency, nuclear type 21, 619065*
NDUFA5	*92,3*	*75,3*	*100*	*99,7*	*No OMIM disease ID*
NDUFA6	*100*	*100*	*100*	*100*	*Mitochondrial complex I deficiency, nuclear type 33, 618253*
NDUFA7	*100*	*99,7*	*100*	*100*	*No OMIM disease ID*
NDUFA8	*100*	*97,3*	*100*	*100*	*Mitochondrial complex I deficiency, nuclear type 37, 619272*
NDUFA9	*99,3*	*95,2*	*100*	*100*	*Mitochondrial complex I deficiency, nuclear type 26, 618247*
NDUFAB1	*98,9*	*91,9*	*100*	*100*	*No OMIM disease ID*
NDUFAF1	*100*	*100*	*100*	*100*	*Mitochondrial complex I deficiency, nuclear type 11, 618234*
NDUFAF2	*91*	*77,5*	*100*	*99,6*	*Mitochondrial complex I deficiency, nuclear type 10, 618233*
NDUFAF3	*100*	*99,9*	*100*	*100*	*Mitochondrial complex I deficiency, nuclear type 18, 618240*
NDUFAF4	*99,6*	*96,9*	*100*	*99,8*	*Mitochondrial complex I deficiency, nuclear type 15, 618237*
NDUFAF5	*99,7*	*99,1*	*100*	*100*	*Mitochondrial complex I deficiency, nuclear type 16, 618238*
NDUFAF6	*99,3*	*96,9*	*100*	*99,9*	*Mitochondrial complex I deficiency, nuclear type 17, 618239* *Fanconi renotubular syndrome 5, 618913*
NDUFAF7	*99,8*	*99,3*	*100*	*100*	*No OMIM disease ID*
NDUFAF8	*62,6*	*61,7*	*100*	*100*	*Mitochondrial complex I deficiency, nuclear type 34, 618776*
NDUFB1	*67,8*	*54,3*	*100*	*100*	*No OMIM disease ID*
NDUFB10	*100*	*100*	*100*	*100*	*?Mitochondrial complex I deficiency, nuclear type 35, 619003*

NDUFB11	*99,1*	*94,8*	*99,9*	*99,1*	*Linear skin defects with multiple congenital anomalies 3, 300952* *?Mitochondrial complex I deficiency, nuclear type 30, 301021*
NDUFB2	*100*	*99,4*	*100*	*100*	*No OMIM disease ID*
NDUFB3	*88,6*	*71*	*100*	*100*	*Mitochondrial complex I deficiency, nuclear type 25, 618246*
NDUFB4	*87,2*	*84,9*	*100*	*100*	*No OMIM disease ID*
NDUFB5	*100*	*100*	*100*	*100*	*No OMIM disease ID*
NDUFB6	*97,2*	*84,7*	*100*	*100*	*No OMIM disease ID*
NDUFB7	*99,9*	*97,4*	*100*	*100*	*No OMIM disease ID*
NDUFB8	*100*	*99,5*	*100*	*100*	*Mitochondrial complex I deficiency, nuclear type 32, 618252*
NDUFB9	*96,1*	*91,5*	*98,7*	*98,7*	*?Mitochondrial complex I deficiency, nuclear type 24, 618245*
NDUFC1	*99,5*	*99,3*	*100*	*100*	*No OMIM disease ID*
NDUFC2	*99,1*	*91,9*	*100*	*100*	*Mitochondrial complex I deficiency, nuclear type 36, 619170*
NDUFS1	*99,9*	*99,1*	*100*	*99,9*	*Mitochondrial complex I deficiency, nuclear type 5, 618226*
NDUFS2	*100*	*100*	*100*	*100*	*Mitochondrial complex I deficiency, nuclear type 6, 618228*
NDUFS3	*90,7*	*90,6*	*92,8*	*90,7*	*Mitochondrial complex I deficiency, nuclear type 8, 618230*
NDUFS4	*99,7*	*99,7*	*100*	*100*	*Mitochondrial complex I deficiency, nuclear type 1, 252010*
NDUFS5	*100*	*100*	*100*	*99,7*	*No OMIM disease ID*
NDUFS6	*100*	*99,8*	*100*	*100*	*Mitochondrial complex I deficiency, nuclear type 9, 618232*
NDUFS7	*100*	*99,7*	*100*	*100*	*Mitochondrial complex I deficiency, nuclear type 3, 618224*
NDUFS8	*100*	*99,1*	*100*	*100*	*Mitochondrial complex I deficiency, nuclear type 2, 618222*
NDUFV1	*99*	*97*	*100*	*100*	*Mitochondrial complex I deficiency, nuclear type 4, 618225*
NDUFV2	*85,8*	*78,7*	*100*	*100*	*Mitochondrial complex I deficiency, nuclear type 7, 618229*
NDUFV3	*99,9*	*98,4*	*100*	*100*	*No OMIM disease ID*
NEB	*82,9*	*82,5*	*99,9*	*99,8*	*Nemaline myopathy 2, autosomal recessive, 256030* *Arthrogryposis multiplex congenita 6, 619334*
NEBL	*99,2*	*97,1*	*100*	*100*	*No OMIM disease ID*
NECAP1	*100*	*100*	*100*	*100*	*Developmental and epileptic encephalopathy 21, 615833*
NECTIN1	*100*	*99,7*	*100*	*100*	*Cleft lip/palate-ectodermal dysplasia syndrome, 225060* *Orofacial cleft 7, 225060*
NECTIN4	*100*	*99,9*	*100*	*100*	*Ectodermal dysplasia-syndactyly syndrome 1, 613573*

NEDD4L	*71,9*	*71,7*	*100*	*99,9*	*Periventricular nodular heterotopia 7, 617201*
NEFH	*96,3*	*88,2*	*100*	*100*	*Charcot-Marie-Tooth disease, axonal, type 2CC, 616924*
NEFL	*99,4*	*96,8*	*100*	*100*	*Charcot-Marie-Tooth disease, type 1F, 607734* *Charcot-Marie-Tooth disease, dominant intermediate G, 617882* *Charcot-Marie-Tooth disease, type 2E, 607684*
NEK1	*99,5*	*98,2*	*100*	*99,9*	*Short-rib thoracic dysplasia 6 with or without polydactyly, 263520*
NEK10	*99,2*	*95,4*	*100*	*100*	*Ciliary dyskinesia, primary, 44, 618781*
NEK11	*99,8*	*98,9*	*100*	*99,9*	*No OMIM disease ID*
NEK2	*99,3*	*93,4*	*96,1*	*96,1*	*?Retinitis pigmentosa 67, 615565*
NEK4	*96,5*	*94,6*	*94,6*	*94,6*	*No OMIM disease ID*
NEK8	*100*	*99,8*	*100*	*100*	*Renal-hepatic-pancreatic dysplasia 2, 615415* *?Nephronophthisis 9, 613824*
NEK9	*99,9*	*99*	*100*	*100*	*?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262* *Nevus comedonicus, somatic, 617025* *Lethal congenital contracture syndrome 10, 617022*
NEMF	*99,6*	*98,3*	*100*	*99,9*	*Intellectual developmental disorder with speech delay and axonal peripheral neuropathy, 619099*
NEPRO	*99,8*	*99,5*	*100*	*100*	*Anauxetic dysplasia 3, 618853*
NEU1	*99,3*	*96,1*	*100*	*100*	*Sialidosis, type II, 256550* *Sialidosis, type I, 256550*
NEUROD1	*100*	*99,4*	*100*	*100*	*Maturity-onset diabetes of the young 6, 606394*
NEUROD2	*100*	*100*	*100*	*100*	*Developmental and epileptic encephalopathy 72, 618374*
NEUROG3	*100*	*100*	*100*	*100*	*Diarrhea 4, malabsorptive, congenital, 610370*
NEXMIF	*99,9*	*99*	*100*	*100*	*Intellectual developmental disorder, X-linked 98, 300912*
NEXN	*87,9*	*71,5*	*100*	*99,8*	*Cardiomyopathy, dilated, 1CC, 613122* *Cardiomyopathy, hypertrophic, 20, 613876*
NF1	*91,8*	*89,3*	*100*	*100*	*Watson syndrome, 193520* *Leukemia, juvenile myelomonocytic, 607785* *Neurofibromatosis, familial spinal, 162210* *Neurofibromatosis, type 1, 162200* *Neurofibromatosis-Noonan syndrome, 601321*
NF2	*100*	*99,6*	*100*	*100*	*Neurofibromatosis, type 2, 101000* *Meningioma, NF2-related, somatic, 607174* *Schwannomatosis, somatic, 162091*
NFASC	*100*	*99,5*	*100*	*100*	*Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356*

NFAT5	*99,9*	*98,7*	*100*	*100*	*No OMIM disease ID*
NFATC1	*100*	*100*	*100*	*99,9*	*No OMIM disease ID*
NFE2	*100*	*100*	*100*	*100*	*No OMIM disease ID*
NFE2L2	*100*	*99,9*	*100*	*100*	*Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744*
NFIA	*99,2*	*98,4*	*99,2*	*99,2*	*Brain malformations with or without urinary tract defects, 613735*
NFIB	*97,4*	*96*	*100*	*99,9*	*Macrocephaly, acquired, with impaired intellectual development, 618286*
NFIX	*100*	*99,3*	*99,4*	*98,6*	*Marshall-Smith syndrome, 602535* *Sotos syndrome 2, 614753*
NFKB1	*99,5*	*97,7*	*100*	*100*	*Immunodeficiency, common variable, 12, 616576*
NFKB2	*97,7*	*94,9*	*100*	*100*	*Immunodeficiency, common variable, 10, 615577*
NFKBIA	*92,4*	*83*	*100*	*100*	*Ectodermal dysplasia and immunodeficiency 2, 612132*
NFS1	*89,2*	*83,6*	*89,5*	*89,5*	*Combined oxidative phosphorylation deficiency 52, 619386*
NFU1	*98,7*	*87,7*	*100*	*100*	*Multiple mitochondrial dysfunctions syndrome 1, 605711*
NGF	*100*	*100*	*100*	*100*	*Neuropathy, hereditary sensory and autonomic, type V, 608654*
NGLY1	*99,8*	*99,7*	*100*	*100*	*Congenital disorder of deglycosylation, 615273*
NHEJ1	*99,8*	*97,2*	*100*	*100*	*Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291*
NHLRC1	*100*	*99,8*	*100*	*100*	*Epilepsy, progressive myoclonic 2B (Lafora), 254780*
NHLRC2	*99,7*	*97,8*	*100*	*99,9*	*FINCA syndrome, 618278*
NHP2	*100*	*100*	*100*	*100*	*Dyskeratosis congenita, autosomal recessive 2, 613987*
NHS	*96,1*	*94,1*	*100*	*100*	*Cataract 40, X-linked, 302200* *Nance-Horan syndrome, 302350*
NIN	*99,9*	*99,5*	*99,1*	*99,1*	*?Seckel syndrome 7, 614851*
NIPA1	*100*	*100*	*99,7*	*98,1*	*Spastic paraplegia 6, autosomal dominant, 600363*
NIPAL4	*100*	*98,9*	*100*	*100*	*Ichthyosis, congenital, autosomal recessive 6, 612281*
NIPBL	*98,4*	*96,3*	*100*	*99,9*	*Cornelia de Lange syndrome 1, 122470*
NKAP	*99*	*94,1*	*99,9*	*99,4*	*Intellectual developmental disorder, X-linked, syndromic, Hackman-Di Donato type, 301039*
NKX2-1	*99,3*	*89,3*	*100*	*100*	*Chorea, hereditary benign, 118700* *Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978*
NKX2-5	*100*	*100*	*100*	*100*	*Hypoplastic left heart syndrome 2, 614435* *Tetralogy of Fallot, 187500*

					Hypothyroidism, congenital nongoitrous, 5, 225250 *Conotruncal heart malformations, variable, 217095* *Ventricular septal defect 3, 614432* *Atrial septal defect 7, with or without AV conduction defects, 108900*
NKX2-6	*100*	*100*	*100*	*100*	*Persistent truncus arteriosus, 217095* *Conotruncal heart malformations, 217095*
NKX3-2	*100*	*99,3*	*100*	*100*	*Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330*
NKX6-2	*88,2*	*81,9*	*100*	*100*	*Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560*
NLGN2	*93,7*	*88,6*	*100*	*100*	*No OMIM disease ID*
NLGN3	*99,7*	*98,2*	*100*	*100*	*No OMIM disease ID*
NLGN4X	*99,8*	*98,2*	*100*	*100*	*Intellectual developmental disorder, X-linked, 300495*
NLRC4	*100*	*99,9*	*100*	*100*	*?Familial cold autoinflammatory syndrome 4, 616115* *Autoinflammation with infantile enterocolitis, 616050*
NLRP1	*99,3*	*97,2*	*100*	*100*	*?Respiratory papillomatosis, juvenile recurrent, congenital, 618803* *Autoinflammation with arthritis and dyskeratosis, 617388* *Palmoplantar carcinoma, multiple self-healing, 615225*
NLRP12	*100*	*100*	*100*	*100*	*Familial cold autoinflammatory syndrome 2, 611762*
NLRP3	*100*	*99,9*	*100*	*100*	*CINCA syndrome, 607115* *Familial cold inflammatory syndrome 1, 120100* *Keratoendothelitis fugax hereditaria, 148200* *Deafness, autosomal dominant 34, with or without inflammation, 617772* *Muckle-Wells syndrome, 191900*
NLRP6	*99,2*	*97,6*	*100*	*100*	*No OMIM disease ID*
NLRP7	*99,9*	*99*	*100*	*100*	*Hydatidiform mole, recurrent, 1, 231090*
NME1	*100*	*100*	*100*	*100*	*No OMIM disease ID*
NME3	*96,1*	*91*	*100*	*100*	*No OMIM disease ID*
NME5	*99,7*	*99,7*	*100*	*100*	*No OMIM disease ID*
NME8	*98,9*	*94,2*	*100*	*100*	*Ciliary dyskinesia, primary, 6, 610852*
NMNAT1	*100*	*99,2*	*99,4*	*96,7*	*Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis, 619260* *Leber congenital amaurosis 9, 608553*
NMNAT2	*100*	*98,8*	*100*	*100*	*No OMIM disease ID*
NNT	*96,4*	*96*	*96,4*	*96,4*	*Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736*

NOBOX	*99,3*	*96,6*	*100*	*100*	*Premature ovarian failure 5, 611548*
NOD2	*100*	*99,9*	*100*	*99,9*	*Blau syndrome, 186580*
NODAL	*100*	*100*	*100*	*100*	*Heterotaxy, visceral, 5, 270100*
NOG	*100*	*100*	*100*	*100*	*Symphalangism, proximal, 1A, 185800* *Brachydactyly, type B2, 611377* *Stapes ankylosis with broad thumbs and toes, 184460* *Tarsal-carpal coalition syndrome, 186570* *Multiple synostoses syndrome 1, 186500*
NOL3	*95,1*	*87*	*100*	*100*	*?Myoclonus, familial, 1, 614937*
NONO	*99,8*	*97,2*	*100*	*100*	*Intellectual developmental disorder, X-linked syndromic 34, 300967*
NOP10	*100*	*99,2*	*100*	*100*	*Dyskeratosis congenita, autosomal recessive 1, 224230*
NOP56	*99,8*	*98,5*	*100*	*100*	*Spinocerebellar ataxia 36, 614153*
NOS1AP	*100*	*100*	*100*	*100*	*Nephrotic syndrome, type 22, 619155*
NOS2	*96,6*	*92,9*	*100*	*100*	*No OMIM disease ID*
NOTCH1	*99,3*	*97,9*	*100*	*100*	*Adams-Oliver syndrome 5, 616028* *Aortic valve disease 1, 109730*
NOTCH2	*100*	*99,2*	*100*	*100*	*Alagille syndrome 2, 610205* *Hajdu-Cheney syndrome, 102500*
NOTCH2NLC	*100*	*99,8*	*100*	*100*	*Tremor, hereditary essential, 6, 618866* *Oculopharyngodistal myopathy 3, 619473* *Neuronal intranuclear inclusion disease, 603472*
NOTCH3	*94,5*	*90,7*	*100*	*99,8*	*Lateral meningocele syndrome, 130720* *?Myofibromatosis, infantile 2, 615293* *Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310*
NOVA2	*99*	*92,8*	*96,8*	*92,8*	*Neurodevelopmental disorder with or without autistic features and/or structural brain abnormalities, 618859*
NPAT	*99,7*	*98,8*	*100*	*100*	*No OMIM disease ID*
NPC1	*99,9*	*99*	*100*	*100*	*Niemann-Pick disease, type C1, 257220* *Niemann-Pick disease, type D, 257220*
NPC2	*100*	*99,2*	*100*	*100*	*Niemann-pick disease, type C2, 607625*
NPHP1	*99,8*	*99,1*	*100*	*100*	*Joubert syndrome 4, 609583* *Nephronophthisis 1, juvenile, 256100* *Senior-Loken syndrome-1, 266900*

NPHP3	*99,6*	*98,5*	*100*	*99,9*	*Nephronophthisis 3, 604387* *Renal-hepatic-pancreatic dysplasia 1, 208540* *Meckel syndrome 7, 267010*
NPHP4	*100*	*99,8*	*100*	*100*	*Senior-Loken syndrome 4, 606996* *Nephronophthisis 4, 606966*
NPHS1	*99,7*	*99*	*100*	*100*	*Nephrotic syndrome, type 1, 256300*
NPHS2	*100*	*99,6*	*100*	*99,9*	*Nephrotic syndrome, type 2, 600995*
NPL	*100*	*99,3*	*100*	*100*	*No OMIM disease ID*
NPM1	*95,3*	*84,9*	*100*	*100*	*Leukemia, acute myeloid, somatic, 601626*
NPPA	*100*	*100*	*100*	*100*	*Atrial standstill 2, 615745* *Atrial fibrillation, familial, 6, 612201*
NPPB	*100*	*100*	*100*	*100*	*No OMIM disease ID*
NPPC	*100*	*99,3*	*100*	*100*	*No OMIM disease ID*
NPR2	*100*	*99,2*	*100*	*100*	*Acromesomelic dysplasia, Maroteaux type, 602875* *Epiphyseal chondrodysplasia, Miura type, 615923* *Short stature with nonspecific skeletal abnormalities, 616255*
NPR3	*100*	*100*	*100*	*100*	*No OMIM disease ID*
NPRL2	*100*	*100*	*100*	*100*	*Epilepsy, familial focal, with variable foci 2, 617116*
NPRL3	*100*	*99,9*	*100*	*100*	*Epilepsy, familial focal, with variable foci 3, 617118*
NR0B1	*99,9*	*99,2*	*100*	*100*	*Adrenal hypoplasia, congenital, 300200* *46XY sex reversal 2, dosage-sensitive, 300018*
NR0B2	*100*	*99,6*	*100*	*100*	*Obesity, mild, early-onset, 601665*
NR1H4	*99,6*	*98,6*	*100*	*100*	*Cholestasis, progressive familial intrahepatic, 5, 617049*
NR2E3	*100*	*99,6*	*100*	*100*	*Retinitis pigmentosa 37, 611131* *Enhanced S-cone syndrome, 268100*
NR2F1	*100*	*100*	*98,2*	*93*	*Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722*
NR2F2	*100*	*99,1*	*100*	*100*	*46,XX sex reversal 5, 618901* *Congenital heart defects, multiple types, 4, 615779*
NR3C1	*100*	*99,9*	*100*	*100*	*Glucocorticoid resistance, 615962*
NR3C2	*99,9*	*99,8*	*100*	*100*	*Pseudohypoaldosteronism type I, autosomal dominant, 177735* *Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115*
NR4A2	*100*	*100*	*100*	*100*	*No OMIM disease ID*
NR4A3	*100*	*99,9*	*100*	*100*	*Chondrosarcoma, extraskeletal myxoid, 612237*

NR5A1	*100*	*100*	*100*	*100*	*46, XX sex reversal 4, 617480* *Premature ovarian failure 7, 612964* *46XY sex reversal 3, 612965* *Adrenocortical insufficiency, 612964* *Spermatogenic failure 8, 613957*
NRAS	*100*	*100*	*100*	*100*	*Noonan syndrome 6, 613224* *?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470* *Melanocytic nevus syndrome, congenital, somatic, 137550* *Epidermal nevus, somatic, 162900* *Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200* *Thyroid carcinoma, follicular, somatic, 188470* *Neurocutaneous melanosis, somatic, 249400* *Colorectal cancer, somatic, 114500*
NRIP1	*100*	*100*	*100*	*100*	*?Congenital anomalies of kidney and urinary tract 3, 618270*
NRL	*99,6*	*96,4*	*100*	*100*	*Retinitis pigmentosa 27, 613750* *Retinal degeneration, autosomal recessive, clumped pigment type,*
NRROS	*100*	*100*	*100*	*100*	*Seizures, early-onset, with neurodegeneration and brain calcification, 618875*
NRXN1	*97,5*	*96,9*	*99,9*	*99,7*	*Pitt-Hopkins-like syndrome 2, 614325*
NSD1	*100*	*99,8*	*100*	*100*	*Sotos syndrome 1, 117550*
NSD2	*99,9*	*98,3*	*100*	*100*	*No OMIM disease ID*
NSDHL	*99,8*	*96,3*	*100*	*100*	*CK syndrome, 300831* *CHILD syndrome, 308050*
NSF	*99,5*	*99,3*	*100*	*100*	*Developmental and epileptic encephalopathy 96, 619340*
NSMCE2	*99,5*	*98,7*	*100*	*100*	*Seckel syndrome 10, 617253*
NSMCE3	*100*	*100*	*100*	*100*	*Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241*
NSMF	*96,9*	*95,5*	*100*	*100*	*Hypogonadotropic hypogonadism 9 with or without anosmia, 614838*
NSUN2	*95,5*	*92,9*	*100*	*100*	*Mental retardation, autosomal recessive 5, 611091*
NSUN3	*100*	*100*	*100*	*100*	*Combined oxidative phosphorylation deficiency 48, 619012*
NT5C2	*97,7*	*94,6*	*100*	*100*	*Spastic paraplegia 45, autosomal recessive, 613162*
NT5C3A	*94,6*	*82,2*	*100*	*100*	*Anemia, hemolytic, due to UMPH1 deficiency, 266120*
NT5E	*100*	*100*	*100*	*100*	*Calcification of joints and arteries, 211800*
NTF4	*98,9*	*92,2*	*100*	*100*	*Glaucoma 1, open angle, 1O, 613100*
NTHL1	*100*	*99,9*	*100*	*100*	*Familial adenomatous polyposis 3, 616415*

NTN1	*100*	*100*	*100*	*99,9*	*Mirror movements 4, 618264*
NTNG2	*99,1*	*97,3*	*100*	*99,6*	*Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718*
NTRK1	*99,9*	*98,5*	*100*	*100*	*Insensitivity to pain, congenital, with anhidrosis, 256800*
NTRK2	*100*	*99,9*	*100*	*100*	*Developmental and epileptic encephalopathy 58, 617830* *Obesity, hyperphagia, and developmental delay, 613886*
NUAK2	*100*	*99,8*	*100*	*100*	*?Anencephaly 2, 619452*
NUBPL	*99,5*	*96,9*	*100*	*100*	*Mitochondrial complex I deficiency, nuclear type 21, 618242*
NUDT2	*100*	*100*	*100*	*100*	*No OMIM disease ID*
NUMA1	*100*	*99,5*	*100*	*100*	*Leukemia, acute promyelocytic, somatic, 612376*
NUP107	*99,7*	*98,4*	*100*	*99,9*	*?Ovarian dysgenesis 6, 618078* *Galloway-Mowat syndrome 7, 618348* *Nephrotic syndrome, type 11, 616730*
NUP133	*99,4*	*97,3*	*100*	*100*	*?Galloway-Mowat syndrome 8, 618349* *Nephrotic syndrome, type 18, 618177*
NUP155	*98,7*	*96,2*	*100*	*99,9*	*?Atrial fibrillation 15, 615770*
NUP160	*100*	*99,8*	*100*	*100*	*?Nephrotic syndrome, type 19, 618178*
NUP188	*99,9*	*99,1*	*100*	*100*	*Sandestig-Stefanova syndrome, 618804*
NUP205	*99,9*	*99,3*	*100*	*99,9*	*?Nephrotic syndrome, type 13, 616893*
NUP214	*99,8*	*99*	*100*	*100*	*Leukemia, T-cell acute lymphoblastic, somatic, 613065* *Leukemia, acute myeloid, somatic, 601626*
NUP37	*100*	*100*	*100*	*100*	*?Microcephaly 24, primary, autosomal recessive, 618179*
NUP62	*100*	*100*	*100*	*100*	*Striatonigral degeneration, infantile, 271930*
NUP85	*100*	*100*	*100*	*100*	*Nephrotic syndrome, type 17, 618176*
NUP88	*99,8*	*99,8*	*100*	*100*	*Fetal akinesia deformation sequence 4, 618393*
NUP93	*96,7*	*92,7*	*95,5*	*95,5*	*Nephrotic syndrome, type 12, 616892*
NUS1	*56,5*	*42*	*100*	*99,9*	*Mental retardation, autosomal dominant 55, with seizures, 617831* *?Congenital disorder of glycosylation, type 1aa, 617082*
NUTM2B-AS1	*NC*	*NC*	*NC*	*NC*	*?Oculopharyngeal myopathy with leukoencephalopathy 1, 618637*
NXF5	*58,3*	*57*	*99,9*	*99,9*	*No OMIM disease ID*
NXN	*100*	*100*	*100*	*99,7*	*Robinow syndrome, autosomal recessive 2, 618529*
NYX	*97,4*	*96,1*	*100*	*99,6*	*Night blindness, congenital stationary (complete), 1A, X-linked, 310500*

OAS1	*100*	*100*	*100*	*100*	*No OMIM disease ID*
OAT	*82*	*73*	*100*	*100*	*Gyrate atrophy of choroid and retina with or without ornithinemia, 258870*
OBSCN	*99,4*	*98,5*	*100*	*100*	*No OMIM disease ID*
OBSL1	*100*	*99,8*	*100*	*100*	*3-M syndrome 2, 612921*
OCA2	*99,9*	*98,3*	*100*	*100*	*Albinism, brown oculocutaneous, 203200* *Albinism, oculocutaneous, type II, 203200*
OCLN	*100*	*99,9*	*100*	*100*	*Pseudo-TORCH syndrome 1, 251290*
OCRL	*99,4*	*97,6*	*100*	*99,9*	*Dent disease 2, 300555* *Lowe syndrome, 309000*
CCDC114	*100*	*99,8*	*100*	*100*	*Ciliary dyskinesia, primary, 20, 615067*
ARMC4	*92,4*	*89,9*	*96,3*	*96,2*	*Ciliary dyskinesia, primary, 23, 615451*
CCDC151	*100*	*99,7*	*100*	*100*	*Ciliary dyskinesia, primary, 30, 616037*
TTC25	*100*	*99,7*	*100*	*100*	*Ciliary dyskinesia, primary, 35, 617092*
ODAM	*99,8*	*99,2*	*100*	*100*	*No OMIM disease ID*
ODAPH	*100*	*100*	*100*	*100*	*Amelogenesis imperfecta, type IIA4, 614832*
ODC1	*100*	*99,3*	*100*	*100*	*Bachmann-Bupp syndrome, 619075*
OFD1	*87,1*	*71,3*	*100*	*99,8*	*Simpson-Golabi-Behmel syndrome, type 2, 300209* *?Retinitis pigmentosa 23, 300424* *Orofaciodigital syndrome I, 311200* *Joubert syndrome 10, 300804*
OGDH	*100*	*99,8*	*100*	*100*	*No OMIM disease ID*
OGG1	*100*	*99,6*	*100*	*100*	*Renal cell carcinoma, clear cell, somatic, 144700*
OGT	*99,7*	*98,3*	*100*	*100*	*Intellectual developmental disorder, X-linked 106, 300997*
OPA1	*99,5*	*96,7*	*100*	*99,9*	*Optic atrophy plus syndrome, 125250* *Optic atrophy 1, 165500* *Behr syndrome, 210000* *?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896*
OPA3	*100*	*99,5*	*100*	*100*	*3-methylglutaconic aciduria, type III, 258501* *Optic atrophy 3 with cataract, 165300*
OPCML	*99,6*	*99,6*	*100*	*100*	*Ovarian cancer, somatic, 167000*
OPHN1	*99,3*	*96,3*	*99,5*	*98,4*	*Intellectual developmental disorder, X-linked syndromic, Billuart type, 300486*
OPLAH	*100*	*99,8*	*100*	*100*	*5-oxoprolinase deficiency, 260005*

OPN1LW	*66,3*	*59,8*	*97,6*	*97,2*	*Blue cone monochromacy, 303700* *Colorblindness, protan, 303900*
OPN1MW	*69,8*	*62,9*	*99*	*97,7*	*Colorblindness, deutan, 303800* *Blue cone monochromacy, 303700*
OPN1SW	*100*	*100*	*100*	*100*	*Colorblindness, tritan, 190900*
OPTN	*99,9*	*99,9*	*100*	*100*	*Glaucoma 1, open angle, E, 137760* *Amyotrophic lateral sclerosis 12 with or without frontotemporal dementia, 613435*
ORAI1	*99,3*	*97,1*	*99,4*	*96,7*	*Immunodeficiency 9, 612782* *Myopathy, tubular aggregate, 2, 615883*
ORC1	*99,9*	*97,9*	*100*	*100*	*Meier-Gorlin syndrome 1, 224690*
ORC4	*96,8*	*90,6*	*100*	*100*	*Meier-Gorlin syndrome 2, 613800*
ORC6	*100*	*99,8*	*100*	*100*	*Meier-Gorlin syndrome 3, 613803*
OSBPL2	*100*	*100*	*100*	*100*	*Deafness, autosomal dominant 67, 616340*
OSGEP	*99,8*	*95,7*	*100*	*100*	*Galloway-Mowat syndrome 3, 617729*
OSMR	*100*	*99,6*	*100*	*100*	*Amyloidosis, primary localized cutaneous, 1, 105250*
OSTM1	*98,7*	*92,9*	*100*	*100*	*Osteopetrosis, autosomal recessive 5, 259720*
OTC	*100*	*99,9*	*100*	*99,7*	*Ornithine transcarbamylase deficiency, 311250*
OTOA	*99,7*	*98,2*	*100*	*100*	*Deafness, autosomal recessive 22, 607039*
OTOF	*100*	*99,8*	*100*	*100*	*Auditory neuropathy, autosomal recessive, 1, 601071* *Deafness, autosomal recessive 9, 601071*
OTOG	*99,3*	*98,5*	*100*	*100*	*Deafness, autosomal recessive 18B, 614945*
OTOGL	*99,3*	*97,1*	*100*	*99,9*	*Deafness, autosomal recessive 84B, 614944*
OTUD5	*89*	*76,4*	*98,4*	*95,2*	*Multiple congenital anomalies-neurodevelopmental syndrome, X-linked, 301056*
OTUD6B	*99,7*	*98,6*	*100*	*99,8*	*Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452*
OTULIN	*92,9*	*87*	*98,8*	*94,1*	*Autoinflammation, panniculitis, and dermatosis syndrome, 617099*
OTX2	*100*	*99*	*100*	*100*	*Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125* *Pituitary hormone deficiency, combined, 6, 613986* *Microphthalmia, syndromic 5, 610125*
OVOL2	*97,8*	*91,8*	*100*	*100*	*Corneal dystrophy, posterior polymorphous, 1, 122000*
OXA1L	*100*	*99,4*	*100*	*100*	*No OMIM disease ID*
OXCT1	*99,4*	*97,6*	*100*	*100*	*Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050*
OXR1	*99,2*	*96,3*	*100*	*99,9*	*Cerebellar hypoplasia/atrophy, epilepsy, and global developmental delay, 213000*

P2RX2	*100*	*100*	*100*	*100*	*Deafness, autosomal dominant 41, 608224*
P2RY12	*100*	*100*	*100*	*100*	*Bleeding disorder, platelet-type, 8, 609821*
P3H1	*100*	*100*	*100*	*100*	*Osteogenesis imperfecta, type VIII, 610915*
P3H2	*99,9*	*98*	*100*	*100*	*Myopia, high, with cataract and vitreoretinal degeneration, 614292*
P4HA2	*99,9*	*98,7*	*100*	*100*	*Myopia 25, autosomal dominant, 617238*
P4HB	*94,6*	*94*	*100*	*100*	*Cole-Carpenter syndrome 1, 112240*
P4HTM	*99,3*	*97,6*	*100*	*99,6*	*Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493*
PABPN1	*64,8*	*54,7*	*100*	*99,2*	*Oculopharyngeal muscular dystrophy, 164300*
PACS1	*99,8*	*99*	*100*	*100*	*Schuurs-Hoeijmakers syndrome, 615009*
PACS2	*99,8*	*97,1*	*99,9*	*99,6*	*Developmental and epileptic encephalopathy 66, 618067*
PADI3	*100*	*100*	*100*	*100*	*Uncombable hair syndrome, 191480*
PADI6	*99,9*	*98,7*	*100*	*100*	*Preimplantation embryonic lethality 2, 617234*
PAFAH1B1	*93,5*	*84,9*	*100*	*100*	*Subcortical laminar heterotopia, 607432* *Lissencephaly 1, 607432*
PAH	*100*	*100*	*100*	*100*	*Phenylketonuria, 261600*
PAK1	*100*	*99,4*	*100*	*100*	*Intellectual developmental disorder with macrocephaly, seizures, and speech delay, 618158*
PAK3	*98,9*	*93,6*	*100*	*99,5*	*Intellectual developmental disorder, X-linked 30, 300558*
PALB2	*100*	*99,9*	*100*	*100*	*Fanconi anemia, complementation group N, 610832*
MPP5	*99,8*	*99,1*	*100*	*100*	*No OMIM disease ID*
PAM16	*65,3*	*65,2*	*82,9*	*82,9*	*Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320*
PANK2	*100*	*99,7*	*100*	*100*	*HARP syndrome, 607236* *Neurodegeneration with brain iron accumulation 1, 234200*
PANX1	*100*	*100*	*100*	*100*	*Oocyte maturation defect 7, 618550*
PAPPA2	*100*	*99,6*	*100*	*100*	*Short stature, Dauber-Argente type, 619489*
PAPSS2	*99,8*	*98*	*100*	*100*	*Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847*
PARK7	*100*	*99,8*	*100*	*100*	*Parkinson disease 7, autosomal recessive early-onset, 606324*
PARN	*81,1*	*80,4*	*88,3*	*87,6*	*Dyskeratosis congenita, autosomal recessive 6, 616353* *Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371*
PARP1	*99,9*	*98,8*	*100*	*100*	*No OMIM disease ID*
PARS2	*100*	*100*	*100*	*100*	*Developmental and epileptic encephalopathy 75, 618437*

PATL2	*99,9*	*95*	*100*	*100*	*Oocyte maturation defect 4, 617743*
PAX1	*92,6*	*87,5*	*100*	*99,7*	*Otofaciocervical syndrome 2, 615560*
PAX2	*100*	*100*	*100*	*100*	*Glomerulosclerosis, focal segmental, 7, 616002* *Papillorenal syndrome, 120330*
PAX3	*100*	*99,8*	*100*	*100*	*Craniofacial-deafness-hand syndrome, 122880* *Waardenburg syndrome, type 3, 148820* *Waardenburg syndrome, type 1, 193500* *Rhabdomyosarcoma 2, alveolar, 268220*
PAX4	*100*	*99,5*	*100*	*100*	*Maturity-onset diabetes of the young, type IX, 612225* *Diabetes mellitus, type 2, 125853*
PAX5	*98,8*	*95,4*	*100*	*100*	*No OMIM disease ID*
PAX6	*100*	*99,9*	*100*	*100*	*Optic nerve hypoplasia, 165550* *Cataract with late-onset corneal dystrophy, 106210* *?Coloboma, ocular, 120200* *?Coloboma of optic nerve, 120430* *Aniridia, 106210* *Anterior segment dysgenesis 5, multiple subtypes, 604229* *?Morning glory disc anomaly, 120430* *Foveal hypoplasia 1, 136520* *Keratitis, 148190*
PAX7	*100*	*100*	*100*	*100*	*Rhabdomyosarcoma 2, alveolar, 268220* *Myopathy, congenital, progressive, with scoliosis, 618578*
PAX8	*100*	*99,6*	*100*	*100*	*Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700*
PAX9	*99,7*	*99,6*	*100*	*100*	*Tooth agenesis, selective, 3, 604625*
PBRM1	*99,9*	*99,3*	*100*	*100*	*?Renal cell carcinoma, clear cell, 144700*
PBX1	*100*	*99,1*	*100*	*100*	*Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641*
PC	*99,7*	*98*	*100*	*100*	*Pyruvate carboxylase deficiency, 266150*
PCARE	*99,6*	*98,1*	*100*	*100*	*Retinitis pigmentosa 54, 613428*
PCBD1	*100*	*99,8*	*100*	*100*	*Hyperphenylalaninemia, BH4-deficient, D, 264070*
PCCA	*98,9*	*93,4*	*100*	*100*	*Propionicacidemia, 606054*
PCCB	*96,7*	*95,4*	*99*	*96,2*	*Propionicacidemia, 606054*
PCDH12	*100*	*100*	*100*	*100*	*Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280*

PCDH15	*97,9*	*96,8*	*100*	*100*	*Usher syndrome, type 1D/F digenic, 601067* *Deafness, autosomal recessive 23, 609533* *Usher syndrome, type 1F, 602083*
PCDH19	*99,7*	*97,7*	*100*	*100*	*Developmental and epileptic encephalopathy 9, 300088*
PCDHGC4	*100*	*100*	*100*	*100*	*No OMIM disease ID*
PCGF2	*99,6*	*93,3*	*100*	*100*	*Turnpenny-Fry syndrome, 618371*
PCK1	*100*	*100*	*100*	*100*	*?Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680*
PCK2	*100*	*100*	*100*	*100*	*No OMIM disease ID*
PCLO	*99,4*	*98,3*	*100*	*100*	*?Pontocerebellar hypoplasia, type 3, 608027*
PCNA	*99,8*	*98,3*	*100*	*100*	*?Ataxia-telangiectasia-like disorder 2, 615919*
PCNT	*99,3*	*96,5*	*100*	*100*	*Microcephalic osteodysplastic primordial dwarfism, type II, 210720*
PCSK1	*99,9*	*99,4*	*100*	*100*	*Obesity with impaired prohormone processing, 600955*
PCSK9	*93,9*	*92,6*	*100*	*100*	*Hypercholesterolemia, familial, 3, 603776*
PCYT1A	*99,2*	*95,7*	*100*	*100*	*Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940*
PCYT2	*100*	*98,3*	*99,6*	*97,8*	*Spastic paraplegia 82, autosomal recessive, 618770*
PDCD1	*100*	*100*	*100*	*100*	*No OMIM disease ID*
PDCD10	*99,7*	*99,5*	*100*	*99,9*	*Cerebral cavernous malformations 3, 603285*
PDE10A	*65,7*	*64,3*	*86,5*	*83,2*	*Striatal degeneration, autosomal dominant, 616922* *Dyskinesia, limb and orofacial, infantile-onset, 616921*
PDE11A	*99,9*	*99,7*	*100*	*100*	*Pigmented nodular adrenocortical disease, primary, 2, 610475*
PDE1C	*99,9*	*99,5*	*100*	*100*	*?Deafness, autosomal dominant 74, 618140*
PDE2A	*100*	*99,5*	*100*	*100*	*Intellectual developmental disorder with paroxysmal dyskinesia or seizures, 619150*
PDE3A	*99,8*	*99,1*	*100*	*100*	*Hypertension and brachydactyly syndrome, 112410*
PDE4D	*95,7*	*93,1*	*100*	*99,6*	*Acrodysostosis 2, with or without hormone resistance, 614613*
PDE6A	*100*	*99,5*	*100*	*100*	*Retinitis pigmentosa 43, 613810*
PDE6B	*100*	*99,8*	*100*	*100*	*Retinitis pigmentosa-40, 613801* *Night blindness, congenital stationary, autosomal dominant 2, 163500*
PDE6C	*99,7*	*97*	*100*	*100*	*Cone dystrophy 4, 613093*
PDE6D	*100*	*99,9*	*100*	*100*	*Joubert syndrome 22, 615665*
PDE6G	*100*	*100*	*100*	*100*	*Retinitis pigmentosa 57, 613582*

PDE6H	*99,8*	*86,1*	*100*	*100*	*Retinal cone dystrophy 3, 610024* *Achromatopsia 6, 610024*
PDE8B	*100*	*99,4*	*100*	*100*	*Pigmented nodular adrenocortical disease, primary, 3, 614190* *Striatal degeneration, autosomal dominant, 609161*
PDGFB	*100*	*100*	*100*	*100*	*Meningioma, SIS-related, 607174* *Basal ganglia calcification, idiopathic, 5, 615483* *Dermatofibrosarcoma protuberans, 607907*
PDGFRA	*100*	*100*	*100*	*100*	*Gastrointestinal stromal tumor/GIST-plus syndrome, somatic or familial, 175510* *Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685*
PDGFRB	*99,2*	*97,3*	*100*	*100*	*Premature aging syndrome, Penttinen type, 601812* *Kosaki overgrowth syndrome, 616592* *Myofibromatosis, infantile, 1, 228550* *Basal ganglia calcification, idiopathic, 4, 615007*
PDGFRL	*100*	*99,8*	*100*	*100*	*Hepatocellular cancer, somatic, 114550* *Colorectal cancer, somatic, 114500*
PDHA1	*98,8*	*95,9*	*100*	*100*	*Pyruvate dehydrogenase E1-alpha deficiency, 312170*
PDHB	*99,2*	*96,8*	*100*	*100*	*Pyruvate dehydrogenase E1-beta deficiency, 614111*
PDHX	*99,8*	*99,6*	*100*	*100*	*Lacticacidemia due to PDX1 deficiency, 245349*
PDK1	*99,7*	*99,2*	*100*	*99,9*	*No OMIM disease ID*
PDK2	*100*	*100*	*100*	*100*	*No OMIM disease ID*
PDK3	*98,8*	*95,5*	*100*	*99,8*	*?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905*
PDK4	*99,7*	*99,5*	*100*	*99,9*	*No OMIM disease ID*
PDLIM3	*100*	*99,7*	*100*	*100*	*No OMIM disease ID*
PDLIM5	*92,9*	*90,1*	*96,7*	*94,4*	*No OMIM disease ID*
PDP1	*100*	*100*	*100*	*100*	*Pyruvate dehydrogenase phosphatase deficiency, 608782*
PDSS1	*95,2*	*87,8*	*97,4*	*97,4*	*Coenzyme Q10 deficiency, primary, 2, 614651*
PDSS2	*98,4*	*94,3*	*100*	*100*	*Coenzyme Q10 deficiency, primary, 3, 614652*
PDX1	*94,8*	*82,2*	*100*	*99,9*	*Pancreatic agenesis 1, 260370* *MODY, type IV, 606392*
PDXK	*79,4*	*77,1*	*99,9*	*99,1*	*Neuropathy, hereditary motor and sensory, type VIC, with optic atrophy, 618511*
PDYN	*100*	*100*	*100*	*100*	*Spinocerebellar ataxia 23, 610245*
PDZD7	*96*	*91,5*	*100*	*99,7*	*Deafness, autosomal recessive 57, 618003* *Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472*

PEPD	*100*	*99,4*	*100*	*100*	*Prolidase deficiency, 170100*
PER2	*100*	*99*	*100*	*100*	*?Advanced sleep phase syndrome, familial, 1, 604348*
PER3	*99,9*	*98,6*	*100*	*100*	*?Advanced sleep phase syndrome, familial, 3, 616882*
PERCC1	*3,4*	*0*	*100*	*100*	*Diarrhea 11, malabsorptive, congenital, 618662*
PERP	*100*	*100*	*100*	*100*	*Erythrokeratodermia variabilis et progressiva 7, 619209* *Olmsted syndrome 2, 619208*
PET100	*100*	*99,2*	*100*	*100*	*Mitochondrial complex IV deficiency, nuclear type 12, 619055*
PET117	*100*	*100*	*100*	*100*	*?Mitochondrial complex IV deficiency, nuclear type 19, 619063*
PEX1	*99,8*	*99,4*	*100*	*100*	*Heimler syndrome 1, 234580* *Peroxisome biogenesis disorder 1B (NALD/IRD), 601539* *Peroxisome biogenesis disorder 1A (Zellweger), 214100*
PEX10	*98,8*	*90,6*	*100*	*100*	*Peroxisome biogenesis disorder 6A (Zellweger), 614870* *Peroxisome biogenesis disorder 6B, 614871*
PEX11B	*100*	*98,3*	*100*	*100*	*Peroxisome biogenesis disorder 14B, 614920*
PEX12	*100*	*100*	*100*	*100*	*Peroxisome biogenesis disorder 3B, 266510* *Peroxisome biogenesis disorder 3A (Zellweger), 614859*
PEX13	*100*	*100*	*100*	*100*	*Peroxisome biogenesis disorder 11A (Zellweger), 614883* *Peroxisome biogenesis disorder 11B, 614885*
PEX14	*95,8*	*89,4*	*100*	*100*	*Peroxisome biogenesis disorder 13A (Zellweger), 614887*
PEX16	*97,1*	*93,9*	*100*	*100*	*Peroxisome biogenesis disorder 8B, 614877* *Peroxisome biogenesis disorder 8A (Zellweger), 614876*
PEX19	*99*	*94,4*	*100*	*100*	*Peroxisome biogenesis disorder 12A (Zellweger), 614886*
PEX2	*100*	*100*	*100*	*100*	*Peroxisome biogenesis disorder 5A (Zellweger), 614866* *Peroxisome biogenesis disorder 5B, 614867*
PEX26	*100*	*99,8*	*100*	*100*	*Peroxisome biogenesis disorder 7B, 614873* *Peroxisome biogenesis disorder 7A (Zellweger), 614872*
PEX3	*99,4*	*99,2*	*100*	*100*	*Peroxisome biogenesis disorder 10A (Zellweger), 614882* *?Peroxisome biogenesis disorder 10B, 617370*
PEX5	*99,9*	*98,8*	*100*	*100*	*Peroxisome biogenesis disorder 2B, 202370* *Peroxisome biogenesis disorder 2A (Zellweger), 214110* *Rhizomelic chondrodysplasia punctata, type 5, 616716*
PEX6	*96,4*	*88*	*100*	*100*	*Peroxisome biogenesis disorder 4B, 614863* *Peroxisome biogenesis disorder 4A (Zellweger), 614862* *Heimler syndrome 2, 616617*

PEX7	*88*	*81*	*91,3*	*91,2*	*Rhizomelic chondrodysplasia punctata, type 1, 215100* *Peroxisome biogenesis disorder 9B, 614879*
PFKM	*100*	*99,7*	*100*	*100*	*Glycogen storage disease VII, 232800*
PFN1	*100*	*100*	*100*	*100*	*Amyotrophic lateral sclerosis 18, 614808*
PGAM2	*100*	*100*	*100*	*100*	*Glycogen storage disease X, 261670*
PGAP1	*98,7*	*94,6*	*100*	*99,8*	*Neurodevelopmental disorder with dysmorphic features, spasticity, and brain abnormalities, 615802*
PGAP2	*100*	*99,9*	*100*	*100*	*Hyperphosphatasia with mental retardation syndrome 3, 614207*
PGAP3	*62,6*	*58,1*	*100*	*100*	*Hyperphosphatasia with mental retardation syndrome 4, 615716*
PGK1	*90,3*	*73,2*	*100*	*100*	*Phosphoglycerate kinase 1 deficiency, 300653*
PGM1	*94,2*	*94,1*	*94,2*	*94,2*	*Congenital disorder of glycosylation, type It, 614921*
PGM2L1	*99,8*	*97,7*	*100*	*100*	*No OMIM disease ID*
PGM3	*99,9*	*99,7*	*91,7*	*91,7*	*Immunodeficiency 23, 615816*
PHACTR1	*100*	*99,6*	*100*	*99,9*	*Developmental and epileptic encephalopathy 70, 618298*
PHC1	*100*	*99,1*	*100*	*100*	*?Microcephaly 11, primary, autosomal recessive, 615414*
PHEX	*99,9*	*98,9*	*100*	*99,2*	*Hypophosphatemic rickets, X-linked dominant, 307800*
PHF21A	*100*	*99,7*	*100*	*100*	*Intellectual developmental disorder with behavioral abnormalities and craniofacial dysmorphism with or without seizures, 618725*
PHF6	*96,2*	*84,7*	*100*	*99,2*	*Borjeson-Forssman-Lehmann syndrome, 301900*
PHF8	*98,9*	*94,4*	*100*	*100*	*Intellectual developmental disorder, X-linked, syndromic, Siderius type, 300263*
PHGDH	*99,9*	*98,2*	*100*	*100*	*Neu-Laxova syndrome 1, 256520* *Phosphoglycerate dehydrogenase deficiency, 601815*
PHIP	*98,2*	*95,7*	*99,9*	*99,6*	*Chung-Jansen syndrome, 617991*
PHKA1	*97,8*	*93,4*	*100*	*99,6*	*Muscle glycogenosis, 300559*
PHKA2	*100*	*99,1*	*100*	*99,4*	*Glycogen storage disease, type IXa2, 306000* *Glycogen storage disease, type IXa1, 306000*
PHKB	*99,7*	*99,1*	*100*	*100*	*Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750*
PHKG1	*99,8*	*97,5*	*100*	*100*	*No OMIM disease ID*
PHKG2	*100*	*99,9*	*100*	*100*	*Glycogen storage disease IXc, 613027*
PHOX2A	*92,8*	*74,4*	*100*	*100*	*Fibrosis of extraocular muscles, congenital, 2, 602078*
PHOX2B	*100*	*100*	*99,8*	*98,5*	*Neuroblastoma with Hirschsprung disease, 613013* *Central hypoventilation syndrome, congenital, 1, with or without Hirschsprung disease, 209880*

PHYH	*100*	*98,9*	*100*	*100*	*Refsum disease, 266500*
PI4K2A	*93,4*	*87,6*	*100*	*100*	*No OMIM disease ID*
PI4KA	*92,6*	*88,7*	*100*	*99,9*	*Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531*
PI4KB	*99,9*	*98,6*	*100*	*100*	*No OMIM disease ID*
PIBF1	*99,1*	*95*	*100*	*99,9*	*Joubert syndrome 33, 617767*
PICALM	*98,9*	*95,2*	*100*	*100*	*Leukemia, acute myeloid, somatic, 601626*
PIDD1	*100*	*99,5*	*100*	*100*	*No OMIM disease ID*
PIEZO1	*99,9*	*98,9*	*100*	*100*	*Lymphatic malformation 6, 616843* *Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380*
PIEZO2	*99,8*	*99,2*	*100*	*100*	*Arthrogryposis, distal, type 5, 108145* *Arthrogryposis, distal, with impaired proprioception and touch, 617146* *Arthrogryposis, distal, type 3, 114300* *?Marden-Walker syndrome, 248700*
PIGA	*91,6*	*82,5*	*100*	*99,8*	*Paroxysmal nocturnal hemoglobinuria, somatic, 300818* *Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868*
PIGB	*99,5*	*97,3*	*100*	*100*	*Developmental and epileptic encephalopathy 80, 618580*
PIGC	*96*	*86,2*	*100*	*100*	*Glycosylphosphatidylinositol biosynthesis defect 16, 617816*
PIGF	*70,7*	*65,3*	*100*	*100*	*Onychodystrophy, osteodystrophy, impaired intellectual development, and seizures syndrome, 619356*
PIGG	*100*	*99,6*	*100*	*100*	*Mental retardation, autosomal recessive 53, 616917*
PIGH	*81,9*	*64,4*	*75,9*	*74,4*	*Glycosylphosphatidylinositol biosynthesis defect 17, 618010*
PIGK	*98,8*	*94,2*	*100*	*100*	*Neurodevelopmental disorder with hypotonia and cerebellar atrophy, with or without seizures, 618879*
PIGL	*100*	*99,6*	*100*	*100*	*CHIME syndrome, 280000*
PIGM	*100*	*100*	*100*	*100*	*Glycosylphosphatidylinositol deficiency, 610293*
PIGN	*93,1*	*89,6*	*98,8*	*98,6*	*Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080*
PIGO	*100*	*99,8*	*100*	*100*	*Hyperphosphatasia with mental retardation syndrome 2, 614749*
PIGP	*95,6*	*85,5*	*100*	*99,9*	*Developmental and epileptic encephalopathy 55, 617599*
PIGQ	*93,4*	*91,6*	*100*	*100*	*Developmental and epileptic encephalopathy 77, 618548*
PIGS	*100*	*99,6*	*100*	*100*	*Developmental and epileptic encephalopathy 95, 618143*
PIGT	*98,1*	*98*	*100*	*100*	*?Paroxysmal nocturnal hemoglobinuria 2, 615399* *Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398*
PIGU	*100*	*99,5*	*100*	*98,9*	*Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis, 618590*

PIGV	*100*	*100*	*100*	*100*	*Hyperphosphatasia with mental retardation syndrome 1, 239300*
PIGW	*100*	*99,7*	*100*	*100*	*Glycosylphosphatidylinositol biosynthesis defect 11, 616025*
PIGY	*100*	*100*	*100*	*100*	*Hyperphosphatasia with mental retardation syndrome 6, 616809*
PIK3C2A	*99*	*95,9*	*100*	*100*	*Oculoskeletodental syndrome, 618440*
PIK3CA	*97,7*	*97,3*	*100*	*100*	*CLOVE syndrome, somatic, 612918* *Hepatocellular carcinoma, somatic, 114550* *Breast cancer, somatic, 114480* *Ovarian cancer, somatic, 167000* *Colorectal cancer, somatic, 114500* *Macrodactyly, somatic, 155500* *CLAPO syndrome, somatic, 613089* *Keratosis, seborrheic, somatic, 182000* *Nevus, epidermal, somatic, 162900* *Gastric cancer, somatic, 613659* *Nonsmall cell lung cancer, somatic, 211980* *Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501* *Cowden syndrome 5, 615108*
PIK3CD	*99,3*	*97,6*	*100*	*100*	*Immunodeficiency 14A, autosomal dominant, 615513* *Immunodeficiency 14B, autosomal recessive, 619281* *?Roifman-Chitayat syndrome, digenic, 613328*
PIK3CG	*100*	*100*	*100*	*100*	*No OMIM disease ID*
PIK3R1	*99,7*	*98,4*	*100*	*100*	*Immunodeficiency 36, 616005* *?Agammaglobulinemia 7, autosomal recessive, 615214* *SHORT syndrome, 269880*
PIK3R2	*90,9*	*89,1*	*99,7*	*98*	*Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387*
PIK3R5	*100*	*99,9*	*100*	*100*	*Ataxia-oculomotor apraxia 3, 615217*
PIKFYVE	*99,9*	*99,3*	*100*	*100*	*Corneal fleck dystrophy, 121850*
PINK1	*91*	*85,8*	*100*	*99,5*	*Parkinson disease 6, early onset, 605909*
PIP5K1C	*99,2*	*96,7*	*99,9*	*99,2*	*Lethal congenital contractural syndrome 3, 611369*
PISD	*100*	*99,7*	*100*	*100*	*Liberfarb syndrome, 618889*
PITPNM3	*99,7*	*98,6*	*100*	*100*	*Cone-rod dystrophy 5, 600977*
PITRM1	*98,2*	*96,2*	*100*	*100*	*Spinocerebellar ataxia, autosomal recessive 30, 619405*
PITX1	*96,4*	*91,5*	*100*	*100*	*Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800*

PITX2	*99,8*	*97,2*	*100*	*100*	*Ring dermoid of cornea, 180550* *Axenfeld-Rieger syndrome, type 1, 180500* *Anterior segment dysgenesis 4, 137600*
PITX3	*100*	*98*	*100*	*100*	*Cataract 11, multiple types, 610623* *Anterior segment dysgenesis 1, multiple subtypes, 107250* *Cataract 11, syndromic, autosomal recessive, 610623*
PJA1	*100*	*100*	*100*	*100*	*No OMIM disease ID*
PJVK	*100*	*99,7*	*100*	*99,9*	*Deafness, autosomal recessive 59, 610220*
PKD1	*40,6*	*32,8*	*99,3*	*99*	*Polycystic kidney disease 1, 173900*
PKD1L1	*100*	*99,3*	*100*	*100*	*Heterotaxy, visceral, 8, autosomal, 617205*
PKD2	*96*	*93,3*	*99,6*	*97,9*	*Polycystic kidney disease 2, 613095*
PKDCC	*91,7*	*84,5*	*98*	*94,6*	*Rhizomelic limb shortening with dysmorphic features, 618821*
PKHD1	*100*	*99,6*	*100*	*100*	*Polycystic kidney disease 4, with or without hepatic disease, 263200*
PKLR	*99,9*	*98*	*100*	*100*	*Adenosine triphosphate, elevated, of erythrocytes, 102900* *Pyruvate kinase deficiency, 266200*
PKP1	*99,9*	*98,6*	*100*	*100*	*Ectodermal dysplasia/skin fragility syndrome, 604536*
PKP2	*94,3*	*86,9*	*95*	*95*	*Arrhythmogenic right ventricular dysplasia 9, 609040*
PKP4	*99,2*	*96,1*	*100*	*100*	*No OMIM disease ID*
PLA2G4A	*99,5*	*99,1*	*100*	*99,9*	*Gastrointestinal ulceration, recurrent, with dysfunctional platelets, 618372*
PLA2G5	*100*	*100*	*100*	*100*	*No OMIM disease ID*
PLA2G6	*92,1*	*90,7*	*92,3*	*92,3*	*Parkinson disease 14, autosomal recessive, 612953* *Neurodegeneration with brain iron accumulation 2B, 610217* *Infantile neuroaxonal dystrophy 1, 256600*
PLA2G7	*99,8*	*99,3*	*100*	*100*	*Platelet-activating factor acetylhydrolase deficiency, 614278*
PLAA	*99,6*	*98,4*	*100*	*100*	*Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527*
PLAG1	*100*	*100*	*100*	*100*	*Adenomas, salivary gland pleomorphic, somatic, 181030* *Silver-Russell syndrome 4, 618907*
PLAT	*100*	*98,5*	*100*	*100*	*No OMIM disease ID*
PLAU	*100*	*99,6*	*100*	*100*	*Quebec platelet disorder, 601709*
PLCB1	*99,9*	*99,4*	*100*	*100*	*Developmental and epileptic encephalopathy 12, 613722*
PLCB3	*100*	*99,3*	*100*	*100*	*Spondylometaphyseal dysplasia with corneal dystrophy, 618961*
PLCB4	*99,8*	*98,7*	*100*	*100*	*Auriculocondylar syndrome 2, 614669*

PLCD1	*99,9*	*97,3*	*100*	*100*	*Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600*
PLCE1	*99,8*	*98,9*	*100*	*100*	*Nephrotic syndrome, type 3, 610725*
PLCG2	*100*	*99,3*	*100*	*100*	*Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878* *Familial cold autoinflammatory syndrome 3, 614468*
PLCZ1	*99*	*96,8*	*100*	*99,9*	*Spermatogenic failure 17, 617214*
PLD1	*99,8*	*98,7*	*100*	*100*	*Cardiac valvular defect, developmental, 212093*
PLD3	*100*	*99,2*	*100*	*100*	*?Spinocerebellar ataxia 46, 617770*
PLEC	*100*	*99,9*	*100*	*100*	*?Epidermolysis bullosa simplex with nail dystrophy, 616487* *Epidermolysis bullosa simplex, Ogna type, 131950* *Epidermolysis bullosa simplex with pyloric atresia, 612138* *Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723* *Epidermolysis bullosa simplex with muscular dystrophy, 226670*
PLEKHG2	*99,8*	*98,1*	*100*	*100*	*Leukodystrophy and acquired microcephaly with or without dystonia, 616763*
PLEKHG5	*96,1*	*93,4*	*96,3*	*96,2*	*Spinal muscular atrophy, distal, autosomal recessive, 4, 611067* *Charcot-Marie-Tooth disease, recessive intermediate C, 615376*
PLEKHM1	*100*	*99,9*	*100*	*100*	*?Osteopetrosis, autosomal recessive 6, 611497* *Osteopetrosis, autosomal dominant 3, 618107*
PLEKHM2	*100*	*99,9*	*100*	*100*	*No OMIM disease ID*
PLG	*87,8*	*87,6*	*100*	*100*	*Dysplasminogenemia, 217090* *Angioedema, hereditary, 4, 619360* *Plasminogen deficiency, type I, 217090*
PLIN1	*99,6*	*95,2*	*100*	*99,8*	*Lipodystrophy, familial partial, type 4, 613877*
PLK1	*100*	*99*	*100*	*100*	*No OMIM disease ID*
PLK4	*99,4*	*98,4*	*100*	*100*	*Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171*
PLN	*100*	*100*	*100*	*100*	*Cardiomyopathy, dilated, 1P, 609909* *Cardiomyopathy, hypertrophic, 18, 613874*
PLOD1	*100*	*98,2*	*100*	*100*	*Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400*
PLOD2	*99,2*	*98,1*	*100*	*100*	*Bruck syndrome 2, 609220*
PLOD3	*100*	*98,7*	*100*	*100*	*Lysyl hydroxylase 3 deficiency, 612394*
PLP1	*99,9*	*97,7*	*100*	*100*	*Pelizaeus-Merzbacher disease, 312080* *Spastic paraplegia 2, X-linked, 312920*
PLPBP	*95,1*	*88,9*	*100*	*99,9*	*Epilepsy, early-onset, vitamin B6-dependent, 617290*
PLPP6	*99,3*	*93,5*	*100*	*100*	*No OMIM disease ID*

PLS1	*99,7*	*98,8*	*100*	*99,9*	*Deafness, autosomal dominant 76, 618787*
PLS3	*97,3*	*96,1*	*97,2*	*97,1*	*Bone mineral density QTL18, osteoporosis, 300910*
PLVAP	*100*	*100*	*100*	*100*	*Diarrhea 10, protein-losing enteropathy type, 618183*
PLXNA1	*100*	*99,9*	*100*	*100*	*No OMIM disease ID*
PLXND1	*98,9*	*96,1*	*99,8*	*99,5*	*No OMIM disease ID*
PMEPA1	*100*	*98,8*	*100*	*99,2*	*No OMIM disease ID*
PMFBP1	*99,8*	*98,5*	*100*	*100*	*Spermatogenic failure 31, 618112*
PML	*100*	*100*	*100*	*100*	*Leukemia, acute promyelocytic, PML/RARA type,*
PMM2	*99,8*	*99,8*	*100*	*100*	*Congenital disorder of glycosylation, type Ia, 212065*
PMP2	*99,7*	*99,7*	*100*	*100*	*Charcot-Marie-Tooth disease, demyelinating, type 1G, 618279*
PMP22	*100*	*100*	*100*	*100*	*Charcot-Marie-Tooth disease, type 1A, 118220* *Roussy-Levy syndrome, 180800* *Charcot-Marie-Tooth disease, type 1E, 118300* *?Neuropathy, inflammatory demyelinating, 139393* *Neuropathy, recurrent, with pressure palsies, 162500* *Dejerine-Sottas disease, 145900*
PMPCA	*97,6*	*93,5*	*100*	*100*	*Spinocerebellar ataxia, autosomal recessive 2, 213200*
PMPCB	*99,9*	*99,2*	*100*	*100*	*Multiple mitochondrial dysfunctions syndrome 6, 617954*
PMS2	*83,9*	*81,6*	*100*	*100*	*Colorectal cancer, hereditary nonpolyposis, type 4, 614337* *Mismatch repair cancer syndrome 4, 619101*
PMS2CL	*NC*	*NC*	*NC*	*NC*	*No OMIM disease ID*
PMVK	*100*	*100*	*100*	*100*	*Porokeratosis 1, multiple types, 175800*
PNKD	*100*	*99,9*	*100*	*100*	*Paroxysmal nonkinesigenic dyskinesia 1, 118800*
PNKP	*100*	*100*	*100*	*100*	*?Charcot-Marie-Tooth disease, type 2B2, 605589* *Ataxia-oculomotor apraxia 4, 616267* *Microcephaly, seizures, and developmental delay, 613402*
PNLDC1	*100*	*100*	*100*	*100*	*No OMIM disease ID*
PNLIP	*99,9*	*99,6*	*100*	*100*	*?Pancreatic lipase deficiency, 614338*
PNMT	*99,7*	*96,8*	*100*	*100*	*No OMIM disease ID*
PNP	*99,8*	*98,7*	*100*	*100*	*Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179*
PNPLA1	*100*	*100*	*100*	*100*	*Ichthyosis, congenital, autosomal recessive 10, 615024*

PNPLA2	*99,8*	*96,1*	*100*	*100*	*Neutral lipid storage disease with myopathy, 610717*
PNPLA6	*100*	*99,8*	*100*	*100*	*Spastic paraplegia 39, autosomal recessive, 612020* *Oliver-McFarlane syndrome, 275400* *?Laurence-Moon syndrome, 245800* *Boucher-Neuhauser syndrome, 215470*
PNPLA8	*99,7*	*99,5*	*100*	*99,9*	*?Mitochondrial myopathy with lactic acidosis, 251950*
PNPO	*99,9*	*97,1*	*100*	*100*	*Pyridoxamine 5'-phosphate oxidase deficiency, 610090*
PNPT1	*96,9*	*86,1*	*100*	*99,9*	*Deafness, autosomal recessive 70, 614934* *Combined oxidative phosphorylation deficiency 13, 614932*
POC1A	*100*	*100*	*100*	*100*	*Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813*
POC1B	*99,5*	*97,3*	*100*	*100*	*Cone-rod dystrophy 20, 615973*
POC5	*99,4*	*97,1*	*100*	*100*	*No OMIM disease ID*
PODXL	*94,3*	*94*	*94,4*	*94,3*	*No OMIM disease ID*
POF1B	*94,5*	*83,3*	*100*	*99,4*	*?Premature ovarian failure 2B, 300604*
POFUT1	*100*	*98,9*	*100*	*100*	*Dowling-Degos disease 2, 615327*
POGLUT1	*99,8*	*95,9*	*100*	*100*	*Dowling-Degos disease 4, 615696* *?Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232*
POGZ	*99,4*	*98,8*	*100*	*100*	*White-Sutton syndrome, 616364*
POLA1	*99*	*93,6*	*100*	*99,7*	*Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220* *Van Esch-O'Driscoll syndrome, 301030*
POLD1	*98,4*	*95,1*	*100*	*100*	*Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381*
POLE	*100*	*99,5*	*100*	*100*	*FILS syndrome, 615139* *IMAGE-I syndrome, 618336*
POLE2	*96,8*	*86,4*	*100*	*99,8*	*No OMIM disease ID*
POLG	*99,9*	*98,8*	*100*	*100*	*Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459* *Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662* *Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700* *Progressive external ophthalmoplegia, autosomal dominant 1, 157640* *Progressive external ophthalmoplegia, autosomal recessive 1, 258450*
POLG2	*99,3*	*97,2*	*100*	*99,8*	*Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131* *?Mitochondrial DNA depletion syndrome 16 (hepatic type), 618528* *?Mitochondrial DNA depletion syndrome 16B (neuroophthalmic type), 619425*
POLH	*100*	*99,1*	*100*	*100*	*Xeroderma pigmentosum, variant type, 278750*
POLL	*99,8*	*97,4*	*100*	*100*	*No OMIM disease ID*

POLR1A	*99,9*	*98,8*	*100*	*100*	*Acrofacial dysostosis, Cincinnati type, 616462*
POLR1B	*100*	*99,9*	*100*	*100*	*Treacher-Collins syndrome 4, 618939*
POLR1C	*89,6*	*84,8*	*82,8*	*82,8*	*Leukodystrophy, hypomyelinating, 11, 616494* *Treacher Collins syndrome 3, 248390*
POLR1D	*91,6*	*91,6*	*100*	*99,8*	*Treacher Collins syndrome 2, 613717*
POLR2A	*100*	*100*	*100*	*100*	*Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603*
POLR3A	*99,9*	*99*	*100*	*100*	*Wiedemann-Rautenstrauch syndrome, 264090* *Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694*
POLR3B	*99,7*	*97,6*	*100*	*100*	*Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381*
POLR3GL	*99,8*	*95,8*	*100*	*100*	*Short stature, oligodontia, dysmorphic facies, and motor delay, 619234*
POLR3K	*100*	*100*	*100*	*100*	*Leukodystrophy, hypomyelinating, 21, 619310*
POLRMT	*85,5*	*65,9*	*100*	*100*	*No OMIM disease ID*
POMC	*100*	*100*	*100*	*100*	*Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734*
POMGNT1	*100*	*99,8*	*100*	*100*	*Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151* *Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157* *Retinitis pigmentosa 76, 617123* *Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280*
POMGNT2	*100*	*100*	*100*	*100*	*Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135* *Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830*
POMK	*100*	*100*	*100*	*100*	*?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094* *Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249*
POMP	*99,7*	*98,8*	*100*	*100*	*Proteasome-associated autoinflammatory syndrome 2, 618048* *Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952*
POMT1	*99,5*	*97,3*	*100*	*100*	*Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670* *Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308* *Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155*
POMT2	*99,8*	*97,3*	*100*	*100*	*Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158* *Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156* *Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150*
POP1	*100*	*99,3*	*100*	*100*	*Anauxetic dysplasia 2, 617396*
POPDC3	*100*	*100*	*100*	*100*	*Muscular dystrophy, limb-girdle, autosomal recessive 26, 618848*
POR	*99,5*	*98*	*100*	*100*	*Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750* *Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571*

PORCN	*100*	*99,1*	*100*	*100*	*Focal dermal hypoplasia, 305600*
POT1	*99,5*	*98,5*	*100*	*100*	*No OMIM disease ID*
POU1F1	*99,9*	*98,2*	*100*	*100*	*Pituitary hormone deficiency, combined, 1, 613038*
POU2AF1	*99,9*	*97,4*	*100*	*100*	*No OMIM disease ID*
POU3F3	*76,5*	*63,8*	*90,8*	*79,1*	*Snijders Blok-Fisher syndrome, 618604*
POU3F4	*100*	*100*	*100*	*100*	*Deafness, X-linked 2, 304400*
POU4F1	*78,2*	*72,5*	*86,7*	*79*	*Ataxia, intention tremor, and hypotonia syndrome, childhood-onset, 619352*
POU4F3	*100*	*100*	*100*	*100*	*Deafness, autosomal dominant 15, 602459*
POU6F2	*95,2*	*95,1*	*100*	*100*	*No OMIM disease ID*
PPA2	*97,3*	*88,6*	*100*	*100*	*?Sudden cardiac failure, alcohol-induced, 617223* *Sudden cardiac failure, infantile, 617222*
PPARG	*100*	*100*	*98,3*	*98,3*	*Insulin resistance, severe, digenic, 604367* *Lipodystrophy, familial partial, type 3, 604367* *Obesity, severe, 601665* *Carotid intimal medial thickness 1, 609338*
PPCS	*100*	*99,1*	*100*	*100*	*Cardiomyopathy, dilated, 2C, 618189*
PPIB	*100*	*99,9*	*100*	*100*	*Osteogenesis imperfecta, type IX, 259440*
PPIL1	*100*	*100*	*100*	*100*	*Pontocerebellar hypoplasia, type 14, 619301*
PPIP5K2	*98,2*	*94,8*	*100*	*99,9*	*Deafness, autosomal recessive 100, 618422*
PPM1D	*100*	*99,9*	*100*	*100*	*Breast cancer, somatic, 114480* *Jansen de Vries syndrome, 617450*
PPM1K	*100*	*100*	*100*	*100*	*?Maple syrup urine disease, mild variant, 615135*
PPOX	*99,3*	*95,7*	*100*	*100*	*Porphyria variegata, 176200*
PPP1CB	*99,8*	*98,7*	*100*	*100*	*Noonan syndrome-like disorder with loose anagen hair 2, 617506*
PPP1R12A	*97,8*	*95,9*	*100*	*99,8*	*Genitourinary and/or/brain malformation syndrome, 618820*
PPP1R15B	*100*	*99,6*	*100*	*100*	*Microcephaly, short stature, and impaired glucose metabolism 2, 616817*
PPP1R21	*99,3*	*95,5*	*100*	*100*	*Neurodevelopmental disorder with hypotonia, facial dysmorphism, and brain abnormalities, 619383*
PPP1R3A	*99,7*	*99,3*	*100*	*100*	*Insulin resistance, severe, digenic, 125853*
PPP2CA	*100*	*100*	*100*	*100*	*Neurodevelopmental disorder and language delay with or without structural brain abnormalities, 618354*
PPP2R1A	*91,6*	*91,6*	*93,6*	*93,6*	*Mental retardation, autosomal dominant 36, 616362*
PPP2R1B	*99,9*	*99,5*	*100*	*100*	*Lung cancer, somatic, 211980*

PPP2R2B	*99,8*	*97,6*	*100*	*100*	*Spinocerebellar ataxia 12, 604326*
PPP2R3C	*98,3*	*89,4*	*100*	*99,8*	*Gonadal dysgenesis, dysmorphic facies, retinal dystrophy, and myopathy, 618419* *Spermatogenic failure 36, 618420*
PPP2R5B	*100*	*100*	*100*	*100*	*No OMIM disease ID*
PPP2R5C	*96,2*	*90,5*	*100*	*99,9*	*No OMIM disease ID*
PPP2R5D	*100*	*99,8*	*100*	*100*	*Mental retardation, autosomal dominant 35, 616355*
PPP3CA	*99,7*	*96,7*	*100*	*100*	*Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265* *Developmental and epileptic encephalopathy 91, 617711*
PPT1	*90,3*	*89,9*	*82,5*	*82,5*	*Ceroid lipofuscinosis, neuronal, 1, 256730*
PQBP1	*100*	*99,3*	*100*	*100*	*Renpenning syndrome, 309500*
PRCC	*99,9*	*98,8*	*100*	*100*	*Renal cell carcinoma, papillary, 605074*
PRCD	*100*	*100*	*100*	*100*	*Retinitis pigmentosa 36, 610599*
PRDM12	*91,7*	*89,6*	*92,8*	*91*	*Neuropathy, hereditary sensory and autonomic, type VIII, 616488*
PRDM13	*98,5*	*93,5*	*100*	*100*	*No OMIM disease ID*
PRDM15	*99,6*	*96,9*	*99,7*	*98,8*	*No OMIM disease ID*
PRDM16	*99,9*	*99,2*	*100*	*100*	*Left ventricular noncompaction 8, 615373* *Cardiomyopathy, dilated, 1LL, 615373*
PRDM5	*99,8*	*98,4*	*100*	*100*	*Brittle cornea syndrome 2, 614170*
PRDM6	*95,9*	*88,9*	*100*	*100*	*Patent ductus arteriosus 3, 617039*
PRDM8	*93,5*	*89,2*	*100*	*100*	*?Epilepsy, progressive myoclonic, 10, 616640*
PRDX1	*100*	*99,9*	*100*	*100*	*Methylmalonic aciduria and homocystinuria, cblC type, digenic, 277400*
PREPL	*98,9*	*96,6*	*100*	*100*	*Myasthenic syndrome, congenital, 22, 616224*
PRF1	*91,2*	*90,1*	*100*	*100*	*Hemophagocytic lymphohistiocytosis, familial, 2, 603553* *Aplastic anemia, 609135* *Lymphoma, non-Hodgkin, 605027*
PRG4	*92,7*	*83,2*	*100*	*100*	*Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250*
PRICKLE1	*100*	*99,9*	*100*	*100*	*Epilepsy, progressive myoclonic 1B, 612437*
PRIMPOL	*97,5*	*94,6*	*100*	*99,8*	*Myopia 22, autosomal dominant, 615420*
PRKAA1	*99,8*	*99,6*	*100*	*100*	*No OMIM disease ID*
PRKACA	*80,1*	*79,6*	*100*	*100*	*Cushing syndrome, ACTH-independent adrenal, somatic, 615830* *Cardioacrofacial dysplasia 1, 619142*

PRKACB	*98,5*	*96,1*	*100*	*99,9*	*Cardioacrofacial dysplasia 2, 619143*
PRKACG	*100*	*99,4*	*100*	*100*	*?Bleeding disorder, platelet-type, 19, 616176*
PRKAG2	*99,4*	*96,1*	*99,9*	*99,3*	*Glycogen storage disease of heart, lethal congenital, 261740* *Wolff-Parkinson-White syndrome, 194200* *Cardiomyopathy, hypertrophic 6, 600858*
PRKAR1A	*97*	*89,1*	*100*	*100*	*Pigmented nodular adrenocortical disease, primary, 1, 610489* *Acrodysostosis 1, with or without hormone resistance, 101800* *Carney complex, type 1, 160980* *Myxoma, intracardiac, 255960* *Adrenocortical tumor, somatic,*
PRKAR1B	*100*	*100*	*100*	*100*	*No OMIM disease ID*
PRKCA	*100*	*100*	*100*	*100*	*Pituitary tumor, invasive,*
PRKCB	*100*	*99,8*	*100*	*100*	*No OMIM disease ID*
PRKCD	*100*	*99,9*	*100*	*100*	*Autoimmune lymphoproliferative syndrome, type III, 615559*
PRKCG	*99,9*	*98,3*	*100*	*100*	*Spinocerebellar ataxia 14, 605361*
PRKCSH	*99,5*	*94,1*	*100*	*100*	*Polycystic liver disease 1, 174050*
PRKD1	*99,6*	*98,9*	*100*	*99,9*	*Congenital heart defects and ectodermal dysplasia, 617364*
PRKDC	*99,2*	*96,9*	*100*	*100*	*Immunodeficiency 26, with or without neurologic abnormalities, 615966*
PRKG1	*92,4*	*91*	*92,7*	*92,7*	*Aortic aneurysm, familial thoracic 8, 615436*
PRKG2	*97,6*	*96,7*	*100*	*100*	*No OMIM disease ID*
PRKN	*66,9*	*65,8*	*75,4*	*75,3*	*Adenocarcinoma of lung, somatic, 211980* *Parkinson disease, juvenile, type 2, 600116* *Ovarian cancer, somatic, 167000*
PRKRA	*99,8*	*99,5*	*100*	*100*	*Dystonia 16, 612067*
PRLR	*100*	*99,6*	*100*	*100*	*Multiple fibroadenomas of the breast, 615554* *Hyperprolactinemia, 615555*
PRMT7	*100*	*99,9*	*100*	*100*	*Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157*
PRNP	*100*	*99,6*	*100*	*100*	*Spongiform encephalopathy with neuropsychiatric features, 606688* *Gerstmann-Straussler disease, 137440* *Huntington disease-like 1, 603218* *Insomnia, fatal familial, 600072* *Cerebral amyloid angiopathy, PRNP-related, 137440* *Creutzfeldt-Jakob disease, 123400*

PROC	*100*	*100*	*100*	*100*	*Thrombophilia due to protein C deficiency, autosomal recessive, 612304* *Thrombophilia due to protein C deficiency, autosomal dominant, 176860*
PRODH	*84*	*80,2*	*100*	*100*	*Hyperprolinemia, type I, 239500*
PROK2	*99,9*	*98,9*	*100*	*100*	*Hypogonadotropic hypogonadism 4 with or without anosmia, 610628*
PROKR2	*100*	*100*	*100*	*100*	*Hypogonadotropic hypogonadism 3 with or without anosmia, 244200*
PROM1	*97,2*	*96,5*	*100*	*99,9*	*Macular dystrophy, retinal, 2, 608051* *Retinitis pigmentosa 41, 612095* *Stargardt disease 4, 603786* *Cone-rod dystrophy 12, 612657*
PROP1	*91*	*80,2*	*100*	*100*	*Pituitary hormone deficiency, combined, 2, 262600*
PRORP	*99,7*	*97,9*	*100*	*100*	*No OMIM disease ID*
PROS1	*96,4*	*89,4*	*98,4*	*98,4*	*Thrombophilia due to protein S deficiency, autosomal dominant, 612336* *Thrombophilia due to protein S deficiency, autosomal recessive, 614514*
PROZ	*100*	*99,1*	*100*	*100*	*No OMIM disease ID*
PRPF3	*97,8*	*92,9*	*100*	*99,9*	*Retinitis pigmentosa 18, 601414*
PRPF31	*99,3*	*95,1*	*100*	*100*	*Retinitis pigmentosa 11, 600138*
PRPF4	*99,9*	*99,8*	*100*	*100*	*Retinitis pigmentosa 70, 615922*
PRPF6	*100*	*99,7*	*100*	*100*	*Retinitis pigmentosa 60, 613983*
PRPF8	*99,9*	*98,6*	*100*	*100*	*Retinitis pigmentosa 13, 600059*
PRPH2	*100*	*100*	*100*	*100*	*Macular dystrophy, patterned, 1, 169150* *Choroidal dystrophy, central areolar 2, 613105* *Retinitis punctata albescens, 136880* *Leber congenital amaurosis 18, 608133* *Macular dystrophy, vitelliform, 3, 608161* *Retinitis pigmentosa 7 and digenic form, 608133*
PRPS1	*86,4*	*86,3*	*100*	*99,7*	*Arts syndrome, 301835* *Phosphoribosylpyrophosphate synthetase superactivity, 300661* *Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070* *Deafness, X-linked 1, 304500* *Gout, PRPS-related, 300661*
PRR11	*100*	*99,9*	*100*	*100*	*No OMIM disease ID*
PRR12	*98,4*	*96,7*	*100*	*100*	*No OMIM disease ID*

PRRT2	*100*	*98,8*	*100*	*100*	*Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066* *Seizures, benign familial infantile, 2, 605751* *Episodic kinesigenic dyskinesia 1, 128200*
PRRX1	*100*	*99,4*	*100*	*100*	*Agnathia-otocephaly complex, 202650*
PRSS1	*100*	*100*	*100*	*100*	*Pancreatitis, hereditary, 167800*
PRSS12	*100*	*99,9*	*100*	*100*	*Mental retardation, autosomal recessive 1, 249500*
PRSS56	*100*	*98*	*100*	*100*	*Microphthalmia, isolated 6, 613517*
PRUNE1	*93,6*	*93,1*	*93,6*	*93,6*	*Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481*
PRX	*96,1*	*95,8*	*97,5*	*96,4*	*Charcot-Marie-Tooth disease, type 4F, 614895* *Dejerine-Sottas disease, 145900*
PSAP	*100*	*99,6*	*100*	*100*	*Combined SAP deficiency, 611721* *Krabbe disease, atypical, 611722* *Metachromatic leukodystrophy due to SAP-b deficiency, 249900* *Gaucher disease, atypical, 610539*
PSAT1	*92*	*75,1*	*100*	*100*	*Neu-Laxova syndrome 2, 616038* *?Phosphoserine aminotransferase deficiency, 610992*
PSEN1	*99,9*	*99,9*	*100*	*100*	*Pick disease, 172700* *Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822* *Dementia, frontotemporal, 600274* *?Acne inversa, familial, 3, 613737* *Cardiomyopathy, dilated, 1U, 613694* *Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822* *Alzheimer disease, type 3, 607822*
PSEN2	*100*	*99,9*	*100*	*100*	*Alzheimer disease-4, 606889* *Cardiomyopathy, dilated, 1V, 613697*
PSENEN	*100*	*99,9*	*100*	*100*	*Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736*
PSIP1	*97,4*	*88*	*100*	*99,9*	*No OMIM disease ID*
PSMA3	*98,7*	*92,4*	*100*	*100*	*No OMIM disease ID*
PSMB1	*100*	*99,8*	*100*	*100*	*No OMIM disease ID*
PSMB10	*100*	*99,7*	*100*	*100*	*Proteasome-associated autoinflammatory syndrome 5, 619175*
PSMB4	*100*	*100*	*100*	*100*	*?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591*
PSMB8	*99,8*	*97,5*	*100*	*100*	*Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040*
PSMB9	*99,5*	*95,4*	*100*	*100*	*?Proteasome-associated autoinflammatory syndrome 3, digenic, 617591*
PSMC3	*100*	*100*	*100*	*100*	*?Deafness, cataract, impaired intellectual development, and polyneuropathy, 619354*

PSMC3IP	*100*	*100*	*100*	*100*	*Ovarian dysgenesis 3, 614324*
PSMC5	*100*	*100*	*100*	*100*	*No OMIM disease ID*
PSMD12	*96,7*	*89,6*	*100*	*100*	*Stankiewicz-Isidor syndrome, 617516*
PSMG2	*99,8*	*98,2*	*100*	*100*	*?Proteasome-associated autoinflammatory syndrome 4, 619183*
PSPH	*100*	*100*	*100*	*100*	*Phosphoserine phosphatase deficiency, 614023*
PSTPIP1	*100*	*99,2*	*100*	*100*	*Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416*
PTCD3	*99,2*	*96,9*	*100*	*100*	*?Combined oxidative phosphorylation deficiency 51, 619057*
PTCH1	*99,3*	*96,6*	*100*	*99,9*	*Basal cell carcinoma, somatic, 605462* *Holoprosencephaly 7, 610828* *Basal cell nevus syndrome, 109400*
PTCH2	*99,9*	*98,4*	*100*	*100*	*Medulloblastoma, somatic, 155255* *Basal cell nevus syndrome, 109400* *Basal cell carcinoma, somatic, 605462*
PTCHD1	*100*	*99,9*	*100*	*100*	*No OMIM disease ID*
PTDSS1	*100*	*100*	*100*	*100*	*Lenz-Majewski hyperostotic dwarfism, 151050*
PTEN	*99,5*	*97,2*	*100*	*100*	*Lhermitte-Duclos syndrome, 158350* *Cowden syndrome 1, 158350* *Prostate cancer, somatic, 176807* *Macrocephaly/autism syndrome, 605309*
PTF1A	*98,8*	*91,1*	*98,7*	*92,9*	*Pancreatic and cerebellar agenesis, 609069* *Pancreatic agenesis 2, 615935*
PTGIS	*99*	*95,1*	*100*	*100*	*Hypertension, essential, 145500*
PTGS1	*100*	*99,9*	*100*	*100*	*No OMIM disease ID*
PTH	*99,3*	*93,8*	*100*	*100*	*Hypoparathyroidism, familial isolated 1, 146200*
PTH1R	*99,6*	*95,9*	*100*	*100*	*Metaphyseal chondrodysplasia, Murk Jansen type, 156400* *Eiken syndrome, 600002* *Failure of tooth eruption, primary, 125350* *Chondrodysplasia, Blomstrand type, 215045*
PTHLH	*99,8*	*98,3*	*100*	*100*	*Brachydactyly, type E2, 613382*
PTPN11	*97,7*	*87,6*	*100*	*100*	*Noonan syndrome 1, 163950* *LEOPARD syndrome 1, 151100* *Metachondromatosis, 156250* *Leukemia, juvenile myelomonocytic, somatic, 607785*
PTPN12	*99,3*	*97,3*	*100*	*100*	*Colon cancer, somatic, 114500*

PTPN14	*99,4*	*96,9*	*100*	*100*	*Choanal atresia and lymphedema, 613611*
PTPN22	*99,6*	*95,2*	*100*	*100*	*No OMIM disease ID*
PTPN23	*100*	*100*	*100*	*100*	*Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity, 618890*
PTPRC	*98,8*	*93,9*	*100*	*99,9*	*Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971*
PTPRF	*100*	*99,8*	*100*	*100*	*?Breasts and/or nipples, aplasia or hypoplasia of, 2, 616001*
PTPRJ	*97,6*	*96,4*	*100*	*99,6*	*Colon cancer, somatic, 114500*
PTPRO	*99,7*	*98,9*	*100*	*100*	*Nephrotic syndrome, type 6, 614196*
PTPRQ	*94,4*	*92,5*	*92,8*	*92,2*	*Deafness, autosomal dominant 73, 617663* *Deafness, autosomal recessive 84A, 613391*
PTRH2	*100*	*100*	*100*	*100*	*Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263*
PTRHD1	*100*	*100*	*100*	*100*	*No OMIM disease ID*
PTS	*99,5*	*99*	*100*	*99,9*	*Hyperphenylalaninemia, BH4-deficient, A, 261640*
PUF60	*99,9*	*99,2*	*100*	*100*	*Verheij syndrome, 615583*
PUM1	*100*	*99,4*	*100*	*99,9*	*Spinocerebellar ataxia 47, 617931*
PURA	*98,6*	*94,6*	*100*	*100*	*Mental retardation, autosomal dominant 31, 616158*
PUS1	*99,9*	*98*	*99,9*	*98,2*	*Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462*
PUS3	*100*	*100*	*100*	*100*	*Neurodevelopmental disorder with microcephaly and gray sclerae, 617051*
PUS7	*99,8*	*99,7*	*100*	*100*	*Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342*
PXDN	*99,9*	*99,6*	*100*	*100*	*Anterior segment dysgenesis 7, with sclerocornea, 269400*
PYCR1	*100*	*98,2*	*100*	*100*	*Cutis laxa, autosomal recessive, type IIIB, 614438* *Cutis laxa, autosomal recessive, type IIB, 612940*
PYCR2	*100*	*99,3*	*100*	*100*	*Leukodystrophy, hypomyelinating, 10, 616420*
PYGL	*100*	*100*	*100*	*100*	*Glycogen storage disease VI, 232700*
PYGM	*100*	*100*	*100*	*100*	*McArdle disease, 232600*
PYROXD1	*92,1*	*78,7*	*100*	*100*	*Myopathy, myofibrillar, 8, 617258*
QARS1	*100*	*100*	*100*	*100*	*Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760*
QDPR	*100*	*98,9*	*100*	*100*	*Hyperphenylalaninemia, BH4-deficient, C, 261630*
QRICH1	*100*	*99*	*100*	*100*	*Ververi-Brady syndrome, 617982*
QRICH2	*94,4*	*93,6*	*100*	*100*	*Spermatogenic failure 35, 618341*
QRSL1	*98,6*	*92,8*	*100*	*99,9*	*Combined oxidative phosphorylation deficiency 40, 618835*

RAB11B	*100*	*100*	*100*	*100*	*Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807*
RAB14	*98,8*	*94,4*	*100*	*99,9*	*No OMIM disease ID*
RAB18	*98,9*	*94,5*	*99,9*	*99,8*	*Warburg micro syndrome 3, 614222*
RAB23	*99,7*	*99,7*	*100*	*100*	*Carpenter syndrome, 201000*
RAB27A	*99,5*	*99,5*	*100*	*99,9*	*Griscelli syndrome, type 2, 607624*
RAB28	*99,2*	*97,2*	*100*	*99,9*	*Cone-rod dystrophy 18, 615374*
RAB33B	*85*	*85*	*100*	*100*	*Smith-McCort dysplasia 2, 615222*
RAB39B	*100*	*99,8*	*100*	*100*	*Intellectual developmental disorder, X-linked 72, 300271* *Waisman syndrome, 311510*
RAB3GAP1	*99,2*	*98,7*	*99,4*	*99,3*	*Martsolf syndrome 2, 619420* *Warburg micro syndrome 1, 600118*
RAB3GAP2	*99,1*	*96,3*	*100*	*99,9*	*Martsolf syndrome 1, 212720* *Warburg micro syndrome 2, 614225*
RAB7A	*100*	*100*	*100*	*100*	*Charcot-Marie-Tooth disease, type 2B, 600882*
RAC1	*99,6*	*96,3*	*100*	*100*	*Mental retardation, autosomal dominant 48, 617751*
RAC2	*99,8*	*95,4*	*100*	*100*	*Immunodeficiency 73A with defective neutrophil chemotaxix and leukocytosis, 608203* *?Immunodeficiency 73C with defective neutrophil chemotaxis and hypogammaglobulinemia, 618987* *Immunodeficiency 73B with defective neutrophil chemotaxis and lymphopenia, 618986*
RAC3	*98*	*94,3*	*99,4*	*97,3*	*Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577*
RAD21	*99,2*	*95,9*	*100*	*100*	*Cornelia de Lange syndrome 4, 614701* *?Mungan syndrome, 611376*
RAD50	*96,6*	*89,7*	*100*	*100*	*Nijmegen breakage syndrome-like disorder, 613078*
RAD51	*89,4*	*89,4*	*89,4*	*89,4*	*Mirror movements 2, 614508* *Fanconi anemia, complementation group R, 617244*
RAD51B	*98,9*	*95*	*95,4*	*92,7*	*No OMIM disease ID*
RAD51C	*99,8*	*99,4*	*100*	*100*	*Fanconi anemia, complementation group O, 613390*
RAD51D	*100*	*99,6*	*100*	*100*	*No OMIM disease ID*
RAD54B	*99,4*	*96,9*	*100*	*100*	*Colon cancer, somatic, 114500* *Lymphoma, non-Hodgkin, somatic, 605027*
RAD54L	*100*	*99*	*100*	*100*	*Lymphoma, non-Hodgkin, somatic, 605027* *Adenocarcinoma, colonic, somatic,*

RAF1	*99,9*	*99,2*	*100*	*100*	*Cardiomyopathy, dilated, 1NN, 615916* *Noonan syndrome 5, 611553* *LEOPARD syndrome 2, 611554*
RAG1	*100*	*100*	*100*	*100*	*Omenn syndrome, 603554* *Severe combined immunodeficiency, B cell-negative, 601457* *Combined cellular and humoral immune defects with granulomas, 233650* *Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889*
RAG2	*100*	*100*	*100*	*100*	*Severe combined immunodeficiency, B cell-negative, 601457* *Combined cellular and humoral immune defects with granulomas, 233650* *Omenn syndrome, 603554*
RAI1	*100*	*100*	*100*	*100*	*Smith-Magenis syndrome, 182290*
RALA	*89,1*	*82,1*	*100*	*100*	*Hiatt-Neu-Cooper neurodevelopmental syndrome, 619311*
RALGAPA1	*73,6*	*61,2*	*100*	*99,9*	*Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation, 618797*
RANBP2	*49,5*	*49,1*	*100*	*100*	*No OMIM disease ID*
RANGRF	*100*	*99,2*	*100*	*100*	*No OMIM disease ID*
RAP1GDS1	*99,4*	*96*	*100*	*100*	*Lymphocytic leukemia, acute T-cell,*
RAPGEF2	*99,5*	*98,7*	*100*	*100*	*?Epilepsy, familial adult myoclonic, 7, 618075*
RAPSN	*100*	*99,6*	*100*	*100*	*Fetal akinesia deformation sequence 2, 618388* *Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326*
RARB	*100*	*99,8*	*100*	*100*	*Microphthalmia, syndromic 12, 615524*
RARS1	*94,1*	*91,8*	*94,4*	*94,2*	*Leukodystrophy, hypomyelinating, 9, 616140*
RARS2	*99,7*	*98,6*	*100*	*100*	*Pontocerebellar hypoplasia, type 6, 611523*
RASA1	*98,3*	*96*	*100*	*99,9*	*Capillary malformation-arteriovenous malformation 1, 608354* *Basal cell carcinoma, somatic, 605462*
RASEF	*99,9*	*99,5*	*100*	*100*	*No OMIM disease ID*
RASGRP1	*100*	*99,6*	*100*	*100*	*Immunodeficiency 64, 618534*
RASGRP2	*100*	*98,3*	*100*	*100*	*?Bleeding disorder, platelet-type, 18, 615888*
RAX	*98,1*	*89,3*	*99,9*	*97,7*	*Microphthalmia, isolated 3, 611038*
RAX2	*100*	*95,5*	*100*	*100*	*Cone-rod dystrophy 11, 610381* *?Macular degeneration, age-related, 6, 613757*
RB1	*96,3*	*93,2*	*100*	*99,9*	*Small cell cancer of the lung, somatic, 182280* *Bladder cancer, somatic, 109800*

					Retinoblastoma, trilateral, 180200 *Osteosarcoma, somatic, 259500* *Retinoblastoma, 180200*
RB1CC1	*99,1*	*95,5*	*100*	*100*	*Breast cancer, somatic, 114480*
RBBP6	*97,9*	*95,2*	*100*	*100*	*No OMIM disease ID*
RBBP8	*99,7*	*99,4*	*100*	*99,9*	*Seckel syndrome 2, 606744* *Jawad syndrome, 251255* *Pancreatic carcinoma, somatic,*
RBCK1	*99,9*	*98,3*	*100*	*100*	*Polyglucosan body myopathy 1 with or without immunodeficiency, 615895*
RBFOX1	*89,2*	*88,6*	*99,8*	*98,2*	*No OMIM disease ID*
RBM10	*99,8*	*97,3*	*100*	*100*	*TARP syndrome, 311900*
RBM20	*100*	*99,4*	*100*	*100*	*Cardiomyopathy, dilated, 1DD, 613172*
RBM28	*100*	*100*	*100*	*100*	*?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079*
RBM8A	*99,6*	*95,3*	*100*	*100*	*Thrombocytopenia-absent radius syndrome, 274000*
RBMX	*91,8*	*79*	*100*	*99,8*	*?Intellectual developmental disorder, syndromic 11, Shashi type, 300238*
RBP3	*100*	*100*	*100*	*100*	*?Retinitis pigmentosa 66, 615233*
RBP4	*99,6*	*96,3*	*100*	*100*	*Microphthalmia, isolated, with coloboma 10, 616428* *Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147*
RBPJ	*97,4*	*89*	*100*	*100*	*Adams-Oliver syndrome 3, 614814*
RC3H1	*100*	*99,4*	*100*	*100*	*?Immune dysregulation and systemic hyperinflammation syndrome, 618998*
RCBTB1	*99,7*	*98,6*	*100*	*100*	*Retinal dystrophy with or without extraocular anomalies, 617175*
RD3	*100*	*100*	*100*	*100*	*Leber congenital amaurosis 12, 610612*
RDH11	*99,6*	*96,9*	*100*	*100*	*?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108*
RDH12	*99,3*	*95,4*	*100*	*100*	*Leber congenital amaurosis 13, 612712*
RDH5	*100*	*100*	*100*	*100*	*Fundus albipunctatus, 136880*
RDX	*87,2*	*69,3*	*100*	*99,9*	*Deafness, autosomal recessive 24, 611022*
REC114	*99,6*	*96,9*	*100*	*100*	*Oocyte maturation defect 10, 619176*
RECQL4	*99,9*	*98,6*	*100*	*100*	*Baller-Gerold syndrome, 218600* *Rothmund-Thomson syndrome, type 2, 268400* *RAPADILINO syndrome, 266280*
REEP1	*78,6*	*76,4*	*100*	*100*	*?Neuronopathy, distal hereditary motor, type VB, 614751* *Spastic paraplegia 31, autosomal dominant, 610250*

REEP2	*99,8*	*98*	*100*	*100*	*?Spastic paraplegia 72, autosomal dominant, 615625* *?Spastic paraplegia 72, autosomal recessive, 615625*
REEP6	*100*	*99,9*	*97,9*	*90,8*	*Retinitis pigmentosa 77, 617304*
RELA	*99,6*	*98*	*100*	*100*	*?Mucocutaneous ulceration, chronic, 618287*
RELB	*99*	*91,5*	*100*	*100*	*?Immunodeficiency 53, 617585*
RELN	*100*	*99,6*	*100*	*100*	*Lissencephaly 2 (Norman-Roberts type), 257320*
RELT	*100*	*99,9*	*100*	*100*	*Amelogenesis imperfecta, type IIIC, 618386*
REN	*100*	*100*	*100*	*100*	*Renal tubular dysgenesis, 267430* *Tubulointerstitial kidney disease, autosomal dominant, 4, 613092*
REPS1	*98,5*	*96,1*	*100*	*99,9*	*?Neurodegeneration with brain iron accumulation 7, 617916*
RERE	*93,3*	*85,2*	*99,9*	*99,9*	*Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975*
REST	*98,4*	*97,4*	*98,6*	*98,6*	*?Deafness, autosomal dominant 27, 612431* *Fibromatosis, gingival, 5, 617626*
RET	*100*	*98,7*	*100*	*100*	*Multiple endocrine neoplasia IIA, 171400* *Medullary thyroid carcinoma, 155240* *Pheochromocytoma, 171300* *Multiple endocrine neoplasia IIB, 162300*
RETREG1	*99,1*	*96,1*	*100*	*100*	*Neuropathy, hereditary sensory and autonomic, type IIB, 613115*
REV3L	*97,4*	*97*	*97,6*	*97,6*	*No OMIM disease ID*
RFC1	*99,5*	*97,4*	*100*	*99,9*	*Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome, 614575*
RFT1	*99,7*	*98,4*	*100*	*100*	*Congenital disorder of glycosylation, type In, 612015*
RFWD3	*100*	*99,4*	*100*	*100*	*?Fanconi anemia, complementation group W, 617784*
RFX3	*100*	*100*	*100*	*100*	*No OMIM disease ID*
RFX5	*99,7*	*98,1*	*100*	*100*	*Bare lymphocyte syndrome, type II, complementation group C, 209920* *Bare lymphocyte syndrome, type II, complementation group E, 209920*
RFX6	*99,9*	*99,5*	*100*	*100*	*Mitchell-Riley syndrome, 615710*
RFXANK	*100*	*99,7*	*100*	*100*	*MHC class II deficiency, complementation group B, 209920*
RFXAP	*99,9*	*98,6*	*100*	*100*	*Bare lymphocyte syndrome, type II, complementation group D, 209920*
RGR	*99*	*97,6*	*99*	*99*	*Retinitis pigmentosa 44, 613769*
RGS10	*95,9*	*91,7*	*100*	*100*	*No OMIM disease ID*
RGS9	*97,9*	*96,5*	*100*	*100*	*Bradyopsia, 608415*
RGS9BP	*100*	*99,4*	*100*	*100*	*Bradyopsia, 608415*

RHAG	*100*	*99,1*	*100*	*100*	*Overhydrated hereditary stomatocytosis, 185000* *Anemia, hemolytic, Rh-null, regulator type, 268150*
RHBDF2	*99,8*	*98,4*	*100*	*100*	*Tylosis with esophageal cancer, 148500*
RHCE	*97,5*	*97*	*96,5*	*96,5*	*Rh-null disease, amorph type, 617970*
RHEB	*86,5*	*70,4*	*100*	*100*	*No OMIM disease ID*
RHO	*100*	*100*	*100*	*100*	*Night blindness, congenital stationary, autosomal dominant 1, 610445* *Retinitis pigmentosa 4, autosomal dominant or recessive, 613731* *Retinitis punctata albescens, 136880*
RHOA	*81,2*	*80,7*	*80,7*	*80,7*	*Ectodermal dysplasia with facial dysmorphism and acral, ocular, and brain anomalies, somatic mosaic, 618727*
RHOBTB2	*100*	*100*	*100*	*100*	*Developmental and epileptic encephalopathy 64, 618004*
RHOG	*100*	*100*	*100*	*100*	*No OMIM disease ID*
RHOH	*100*	*100*	*100*	*100*	*No OMIM disease ID*
RIC1	*99,9*	*99,8*	*100*	*100*	*CATIFA syndrome, 618761*
RIMS1	*99,7*	*97,3*	*100*	*100*	*Cone-rod dystrophy 7, 603649*
RIMS2	*96,6*	*94,5*	*97,8*	*97,7*	*Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970*
RIN2	*100*	*99,7*	*100*	*100*	*Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075*
RINT1	*99,6*	*97,6*	*100*	*99,9*	*Infantile liver failure syndrome 3, 618641*
RIPK1	*99,8*	*98,5*	*100*	*100*	*Immunodeficiency 57 with autoinflammation, 618108* *Autoinflammation with episodic fever and lymphadenopathy, 618852*
RIPK4	*100*	*99,9*	*100*	*100*	*CHAND syndrome, 214350* *Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650*
RIPOR2	*99,9*	*99,3*	*100*	*100*	*?Deafness, autosomal recessive 104, 616515*
RIPPLY2	*99*	*94,7*	*100*	*99,9*	*?Spondylocostal dysostosis 6, 616566*
RIT1	*100*	*100*	*100*	*100*	*Noonan syndrome 8, 615355*
RLBP1	*100*	*99,8*	*100*	*100*	*Bothnia retinal dystrophy, 607475* *Newfoundland rod-cone dystrophy, 607476* *Retinitis punctata albescens, 136880* *Fundus albipunctatus, 136880*
RLIM	*99,8*	*98*	*100*	*100*	*Tonne-Kalscheuer syndrome, 300978*
RMND1	*99,7*	*97,2*	*100*	*99,9*	*Combined oxidative phosphorylation deficiency 11, 614922*
RMRP	*NC*	*NC*	*NC*	*NC*	*Anauxetic dysplasia 1, 607095* *Metaphyseal dysplasia without hypotrichosis, 250460* *Cartilage-hair hypoplasia, 250250*

RNASEH1	*98,7*	*95,8*	*100*	*100*	*Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479*
RNASEH2A	*100*	*99,7*	*100*	*100*	*Aicardi-Goutieres syndrome 4, 610333*
RNASEH2B	*81*	*78,2*	*91*	*90,9*	*Aicardi-Goutieres syndrome 2, 610181*
RNASEH2C	*100*	*100*	*100*	*100*	*Aicardi-Goutieres syndrome 3, 610329*
RNASEL	*100*	*99,6*	*100*	*100*	*Prostate cancer 1, 601518*
RNASET2	*95,7*	*91*	*100*	*100*	*Leukoencephalopathy, cystic, without megalencephaly, 612951*
RNF113A	*100*	*100*	*100*	*100*	*Trichothiodystrophy 5, nonphotosensitive, 300953*
RNF125	*100*	*98,3*	*100*	*100*	*Tenorio syndrome, 616260*
RNF13	*94,8*	*83,2*	*100*	*99,7*	*Developmental and epileptic encephalopathy 73, 618379*
RNF139	*100*	*100*	*100*	*100*	*Renal cell carcinoma, 144700*
RNF168	*99,9*	*99,6*	*100*	*100*	*RIDDLE syndrome, 611943*
RNF170	*98,2*	*94,2*	*100*	*100*	*Ataxia, sensory, 1, autosomal dominant, 608984*
RNF2	*99,7*	*98,2*	*100*	*100*	*Luo-Schoch-Yamamoto syndrome, 619460*
RNF212	*99,9*	*99,2*	*100*	*100*	*Recombination rate QTL 1, 612042*
RNF216	*99,6*	*98,2*	*100*	*100*	*Cerebellar ataxia and hypogonadotropic hypogonadism, 212840*
RNF31	*100*	*98,6*	*100*	*100*	*No OMIM disease ID*
RNF43	*99,8*	*98,3*	*100*	*100*	*Sessile serrated polyposis cancer syndrome, 617108*
RNF6	*100*	*99,8*	*100*	*100*	*Esophageal carcinoma, somatic, 133239*
RNPC3	*94*	*75,1*	*100*	*100*	*?Growth hormone deficiency, isolated, type V, 618160*
RNU4ATAC	*NC*	*NC*	*NC*	*NC*	*Roifman syndrome, 616651* *Lowry-Wood syndrome, 226960* *Microcephalic osteodysplastic primordial dwarfism, type I, 210710*
RNU7-1	*NC*	*NC*	*NC*	*NC*	*Aicardi-Goutieres syndrome 9, 619487*
ROBO1	*100*	*99,5*	*100*	*100*	*No OMIM disease ID*
ROBO2	*99,1*	*97,3*	*100*	*100*	*Vesicoureteral reflux 2, 610878*
ROBO3	*99*	*95,9*	*100*	*100*	*Gaze palsy, familial horizontal, with progressive scoliosis, 1, 607313*
ROBO4	*100*	*99*	*100*	*100*	*Aortic valve disease 3, 618496*
ROGDI	*98,6*	*95,2*	*99,9*	*98,1*	*Kohlschutter-Tonz syndrome, 226750*
ROM1	*100*	*99,9*	*100*	*100*	*Retinitis pigmentosa 7, digenic form, 608133*
ROR1	*97,2*	*96,8*	*100*	*99,4*	*?Deafness, autosomal recessive 108, 617654*

ROR2	*100*	*99,4*	*97*	*97*	*Brachydactyly, type B1, 113000* *Robinow syndrome, autosomal recessive, 268310*
RORA	*96,2*	*89,4*	*100*	*100*	*Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060*
RORC	*100*	*100*	*100*	*100*	*Immunodeficiency 42, 616622*
RP1	*91,2*	*90,7*	*100*	*100*	*Retinitis pigmentosa 1, 180100*
RP1L1	*100*	*100*	*100*	*100*	*Occult macular dystrophy, 613587* *Retinitis pigmentosa 88, 618826*
RP2	*99,8*	*99,8*	*100*	*100*	*Retinitis pigmentosa 2, 312600*
RP9	*82,5*	*72,7*	*100*	*99,7*	*?Retinitis pigmentosa 9, 180104*
RPE65	*99,9*	*98,7*	*100*	*100*	*Retinitis pigmentosa 20, 613794* *Retinitis pigmentosa 87 with choroidal involvement, 618697* *Leber congenital amaurosis 2, 204100*
RPGR	*76,4*	*70,8*	*100*	*99,2*	*Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455* *Cone-rod dystrophy, X-linked, 1, 304020* *Retinitis pigmentosa 3, 300029* *Macular degeneration, X-linked atrophic, 300834*
RPGRIP1	*100*	*99,7*	*100*	*100*	*Cone-rod dystrophy 13, 608194* *Leber congenital amaurosis 6, 613826*
RPGRIP1L	*96,5*	*95,3*	*100*	*99,4*	*Joubert syndrome 7, 611560* *Meckel syndrome 5, 611561* *?COACH syndrome 3, 619113*
RPIA	*99,1*	*96,1*	*100*	*100*	*Ribose 5-phosphate isomerase deficiency, 608611*
RPL10	*96,7*	*87,5*	*100*	*100*	*Intellectual developmental disorder, X-linked, syndromic, 35, 300998*
RPL11	*99,9*	*97,9*	*100*	*100*	*Diamond-Blackfan anemia 7, 612562*
RPL13	*95,6*	*84,7*	*100*	*100*	*Spondyloepimetaphyseal dysplasia, Isidor-Toutain type, 618728*
RPL15	*84,9*	*70,4*	*100*	*99,5*	*?Diamond-Blackfan anemia 12, 615550*
RPL18	*100*	*99,9*	*100*	*100*	*?Diamond-Blackfan anemia 18, 618310*
RPL21	*81,1*	*62*	*100*	*100*	*Hypotrichosis 12, 615885*
RPL26	*94,2*	*75,5*	*100*	*100*	*?Diamond-Blackfan anemia 11, 614900*
RPL27	*68*	*56,6*	*100*	*100*	*?Diamond-Blackfan anemia 16, 617408*
RPL31	*97,6*	*87,4*	*100*	*100*	*No OMIM disease ID*
RPL35	*90,1*	*79,1*	*100*	*100*	*?Diamond-Blackfan anemia 19, 618312*
RPL35A	*94,7*	*84,9*	*100*	*100*	*Diamond-Blackfan anemia 5, 612528*

RPL3L	*100*	*99,3*	*100*	*100*	*Cardiomyopathy, dilated, 2D, 619371*
RPL4	*83,3*	*73,3*	*100*	*100*	*No OMIM disease ID*
RPL5	*81,9*	*59,7*	*100*	*100*	*Diamond-Blackfan anemia 6, 612561*
RPL9	*98,6*	*88*	*100*	*100*	*No OMIM disease ID*
RPN2	*100*	*100*	*100*	*100*	*No OMIM disease ID*
RPS10	*96,6*	*87,6*	*100*	*100*	*Diamond-Blackfan anemia 9, 613308*
RPS14	*96,7*	*92,6*	*100*	*100*	*Macrocytic anemia, refractory, due to 5q deletion, somatic, 153550*
RPS15A	*95,3*	*84,1*	*80,4*	*80,4*	*?Diamond-Blackfan anemia 20, 618313*
RPS17	*85*	*67,8*	*100*	*100*	*Diamond-Blackfan anemia 4, 612527*
RPS19	*100*	*99,9*	*100*	*100*	*Diamond-Blackfan anemia 1, 105650*
RPS20	*96,6*	*87,5*	*100*	*100*	*No OMIM disease ID*
RPS23	*85,4*	*75,7*	*100*	*100*	*Brachycephaly, trichomegaly, and developmental delay, 617412*
RPS24	*96,2*	*90,3*	*100*	*100*	*Diamond-blackfan anemia 3, 610629*
RPS26	*93,2*	*81,2*	*100*	*100*	*Diamond-Blackfan anemia 10, 613309*
RPS27	*95,5*	*70*	*100*	*100*	*?Diamond-Blackfan anemia 17, 617409*
RPS28	*99,7*	*86,3*	*100*	*100*	*Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164*
RPS29	*78*	*70,5*	*100*	*100*	*Diamond-Blackfan anemia 13, 615909*
RPS6KA3	*98,4*	*91,4*	*99,9*	*98,3*	*Intellectual developmental disorder, X-linked 19, 300844* *Coffin-Lowry syndrome, 303600*
RPS7	*81,7*	*66,9*	*100*	*100*	*Diamond-Blackfan anemia 8, 612563*
RPSA	*100*	*99,9*	*100*	*100*	*Asplenia, isolated congenital, 271400*
RRAD	*88*	*82,4*	*99,6*	*96,7*	*No OMIM disease ID*
RRAGC	*99,9*	*99,4*	*100*	*100*	*No OMIM disease ID*
RRAS	*99,7*	*95,8*	*100*	*99,9*	*No OMIM disease ID*
RRAS2	*95,9*	*87,4*	*100*	*100*	*Noonan syndrome 12, 618624* *Ovarian carcinoma,*
RREB1	*99,8*	*99,2*	*100*	*100*	*No OMIM disease ID*
RRM1	*99,9*	*99,5*	*100*	*100*	*No OMIM disease ID*

RRM2B	*100*	*99,8*	*100*	*99,9*	*Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075* *Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075* *Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077*
RRP7A	*99,9*	*97*	*100*	*99,8*	*?Microcephaly 28, primary, autosomal recessive, 619453*
RS1	*98,4*	*89,6*	*100*	*100*	*Retinoschisis, 312700*
RSPH1	*99,9*	*99,9*	*100*	*100*	*Ciliary dyskinesia, primary, 24, 615481*
RSPH3	*99,6*	*98,4*	*100*	*99,9*	*Ciliary dyskinesia, primary, 32, 616481*
RSPH4A	*98,2*	*95,4*	*100*	*100*	*Ciliary dyskinesia, primary, 11, 612649*
RSPH9	*99,7*	*96,3*	*100*	*100*	*Ciliary dyskinesia, primary, 12, 612650*
RSPO1	*100*	*99,9*	*100*	*100*	*Palmoplantar hyperkeratosis and true hermaphroditism, 610644* *Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644*
RSPO2	*94,8*	*88,3*	*100*	*100*	*?Humerofemoral hypoplasia with radiotibial ray deficiency, 618022* *Tetraamelia syndrome 2, 618021*
RSPO4	*100*	*99,2*	*100*	*100*	*Anonychia congenita, 206800*
RSPRY1	*99,9*	*99,9*	*100*	*100*	*Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723*
RSRC1	*99*	*94,6*	*100*	*100*	*Intellectual developmental disorder, autosomal recessive 70, 618402*
RTEL1	*99,7*	*97,2*	*100*	*100*	*Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373* *Dyskeratosis congenita, autosomal dominant 4, 615190* *Dyskeratosis congenita, autosomal recessive 5, 615190*
RTN2	*99,9*	*99,1*	*100*	*100*	*Spastic paraplegia 12, autosomal dominant, 604805*
RTN4IP1	*99,6*	*97,3*	*100*	*100*	*Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732*
RTTN	*98,6*	*97,6*	*100*	*100*	*Microcephaly, short stature, and polymicrogyria with seizures, 614833*
RUBCN	*99,7*	*97,9*	*100*	*100*	*Spinocerebellar ataxia, autosomal recessive 15, 615705*
RUNX1	*98,6*	*93*	*100*	*100*	*Platelet disorder, familial, with associated myeloid malignancy, 601399* *Leukemia, acute myeloid, 601626*
RUNX2	*72,2*	*72,2*	*100*	*100*	*Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510* *Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600* *Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600* *Cleidocranial dysplasia, 119600*
RUSC2	*100*	*100*	*100*	*100*	*Mental retardation, autosomal recessive 61, 617773*
RXYLT1	*99,2*	*95,9*	*100*	*99,9*	*Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041*
RYR1	*97,1*	*94*	*99,4*	*99*	*Neuromuscular disease, congenital, with uniform type 1 fiber, 117000* *Central core disease, 117000*

					King-Denborough syndrome, 145600 *Minicore myopathy with external ophthalmoplegia, 255320*
RYR2	*99,8*	*98,8*	*100*	*100*	*Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772* *Ventricular arrythmias due to cardiac ryanodine receptor calcium release deficiency syndrome, 115000* *Arrhythmogenic right ventricular dysplasia 2, 600996*
S1PR2	*99*	*96,4*	*100*	*100*	*Deafness, autosomal recessive 68, 610419*
SACS	*99,9*	*99,9*	*100*	*100*	*Spastic ataxia, Charlevoix-Saguenay type, 270550*
SAG	*100*	*100*	*100*	*100*	*Retinitis pigmentosa 47, 613758* *Oguchi disease-1, 258100*
SALL1	*99,7*	*97,5*	*100*	*100*	*Townes-Brocks syndrome 1, 107480* *Townes-Brocks branchiootorenal-like syndrome, 107480*
SALL2	*100*	*100*	*100*	*100*	*?Coloboma, ocular, autosomal recessive, 216820*
SALL4	*99,1*	*96,4*	*100*	*100*	*?IVIC syndrome, 147750* *Duane-radial ray syndrome, 607323*
SAMD11	*92,2*	*85*	*100*	*100*	*No OMIM disease ID*
SAMD12	*100*	*100*	*100*	*100*	*Epilepsy, familial adult myoclonic, 1, 601068*
SAMD9	*99,9*	*99,8*	*100*	*100*	*Tumoral calcinosis, familial, normophosphatemic, 610455* *Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041* *MIRAGE syndrome, 617053*
SAMD9L	*100*	*99,9*	*100*	*100*	*Ataxia-pancytopenia syndrome, 159550* *Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270*
SAMHD1	*98,5*	*97,9*	*100*	*100*	*?Chilblain lupus 2, 614415* *Aicardi-Goutieres syndrome 5, 612952*
SAR1B	*94,8*	*88,6*	*100*	*100*	*Chylomicron retention disease, 246700*
SARDH	*93,4*	*91,6*	*91,4*	*91,4*	*No OMIM disease ID*
SARS1	*100*	*99,3*	*100*	*100*	*?Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709*
SARS2	*95,7*	*94,5*	*100*	*100*	*Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845*
SART3	*99,7*	*98,7*	*100*	*100*	*No OMIM disease ID*
SASH1	*99,8*	*98,3*	*100*	*100*	*Dyschromatosis universalis hereditaria 1, 127500* *?Cancer, alopecia, pigment dyscrasia, onychodystrophy, and keratoderma, 618373*
SASH3	*99,8*	*94,9*	*100*	*100*	*No OMIM disease ID*
SASS6	*99,1*	*98,1*	*100*	*99,7*	*?Microcephaly 14, primary, autosomal recessive, 616402*
SAT1	*99,7*	*96,5*	*100*	*100*	*No OMIM disease ID*

SATB1	*96,4*	*96*	*100*	*99,7*	*Kohlschutter-Tonz syndrome-like, 619229* *Developmental delay with dysmorphic facies and dental anomalies, 619228*
SATB2	*99,5*	*96,5*	*100*	*100*	*Glass syndrome, 612313*
SBDS	*100*	*99,9*	*100*	*100*	*Shwachman-Diamond syndrome, 260400*
SBF1	*99,1*	*98*	*100*	*100*	*Charcot-Marie-Tooth disease, type 4B3, 615284*
SBF2	*99,7*	*98,6*	*100*	*100*	*Charcot-Marie-Tooth disease, type 4B2, 604563*
SC5D	*99,9*	*99,1*	*100*	*100*	*Lathosterolosis, 607330*
SCAF4	*99,3*	*97,1*	*100*	*100*	*No OMIM disease ID*
SCAMP5	*100*	*100*	*100*	*100*	*No OMIM disease ID*
SCAPER	*99,5*	*97,1*	*100*	*99,9*	*Intellectual developmental disorder and retinitis pigmentosa, 618195*
SCARB2	*99,9*	*99,4*	*100*	*100*	*Epilepsy, progressive myoclonic 4, with or without renal failure, 254900*
SCARF2	*97,4*	*88,9*	*99,8*	*99,2*	*Van den Ende-Gupta syndrome, 600920*
SCD5	*100*	*99,1*	*100*	*100*	*?Deafness, autosomal dominant 79, 619086*
SCIMP	*80,4*	*79,9*	*97,8*	*89,1*	*No OMIM disease ID*
SCLT1	*95,4*	*89,6*	*95,1*	*95*	*No OMIM disease ID*
SCN10A	*99,9*	*98,5*	*100*	*100*	*Episodic pain syndrome, familial, 2, 615551*
SCN11A	*99,3*	*97,5*	*100*	*100*	*Episodic pain syndrome, familial, 3, 615552* *Neuropathy, hereditary sensory and autonomic, type VII, 615548*
SCN1A	*99,7*	*99,1*	*100*	*100*	*Developmental and epileptic encephalopathy 6B, non-Dravet, 619317* *Migraine, familial hemiplegic, 3, 609634* *Dravet syndrome, 607208* *Febrile seizures, familial, 3A, 604403* *Generalized epilepsy with febrile seizures plus, type 2, 604403*
SCN1B	*98,2*	*96,3*	*99,7*	*98,9*	*Generalized epilepsy with febrile seizures plus, type 1, 604233* *Developmental and epileptic encephalopathy 52, 617350* *Cardiac conduction defect, nonspecific, 612838* *Atrial fibrillation, familial, 13, 615377* *Brugada syndrome 5, 612838*
SCN2A	*99,4*	*97,4*	*100*	*99,9*	*Seizures, benign familial infantile, 3, 607745* *Developmental and epileptic encephalopathy 11, 613721* *Episodic ataxia, type 9, 618924*
SCN2B	*100*	*100*	*100*	*100*	*Atrial fibrillation, familial, 14, 615378*

SCN3A	*99,8*	*99,1*	*100*	*100*	*Epilepsy, familial focal, with variable foci 4, 617935* *Developmental and epileptic encephalopathy 62, 617938*
SCN3B	*100*	*100*	*100*	*100*	*Atrial fibrillation, familial, 16, 613120* *Brugada syndrome 7, 613120*
SCN4A	*99,9*	*99,4*	*100*	*100*	*Paramyotonia congenita, 168300* *Hypokalemic periodic paralysis, type 2, 613345* *Myotonia congenita, atypical, acetazolamide-responsive, 608390* *Myasthenic syndrome, congenital, 16, 614198* *Hyperkalemic periodic paralysis, type 2, 170500*
SCN4B	*99,9*	*97,1*	*100*	*100*	*Atrial fibrillation, familial, 17, 611819* *Long QT syndrome 10, 611819*
SCN5A	*99*	*98,7*	*100*	*100*	*Ventricular fibrillation, familial, 1, 603829* *Heart block, progressive, type IA, 113900* *Cardiomyopathy, dilated, 1E, 601154* *Heart block, nonprogressive, 113900* *Long QT syndrome 3, 603830* *Sick sinus syndrome 1, 608567* *Brugada syndrome 1, 601144* *Atrial fibrillation, familial, 10, 614022*
SCN7A	*97,7*	*91,2*	*100*	*99,9*	*No OMIM disease ID*
SCN8A	*100*	*99,5*	*100*	*100*	*?Myoclonus, familial, 2, 618364* *Seizures, benign familial infantile, 5, 617080* *Cognitive impairment with or without cerebellar ataxia, 614306* *Developmental and epileptic encephalopathy 13, 614558*
SCN9A	*99,1*	*97*	*100*	*100*	*Erythermalgia, primary, 133020* *Insensitivity to pain, congenital, 243000* *Small fiber neuropathy, 133020* *Paroxysmal extreme pain disorder, 167400* *Neuropathy, hereditary sensory and autonomic, type IID, 243000*
SCNN1A	*99,7*	*97,5*	*100*	*100*	*Pseudohypoaldosteronism, type I, 264350* *?Liddle syndrome 3, 618126* *Bronchiectasis with or without elevated sweat chloride 2, 613021*
SCNN1B	*100*	*99,8*	*100*	*100*	*Bronchiectasis with or without elevated sweat chloride 1, 211400* *Pseudohypoaldosteronism, type I, 264350* *Liddle syndrome 1, 177200*
SCNN1G	*99,8*	*98,4*	*100*	*100*	*Bronchiectasis with or without elevated sweat chloride 3, 613071* *Pseudohypoaldosteronism, type I, 264350* *Liddle syndrome 2, 618114*

SCO1	*97,6*	*94,4*	*100*	*100*	*Mitochondrial complex IV deficiency, nuclear type 4, 619048*
SCO2	*100*	*100*	*100*	*100*	*Myopia 6, 608908* *Mitochondrial complex IV deficiency, nuclear type 2, 604377*
SCP2	*99,9*	*97,9*	*100*	*100*	*?Leukoencephalopathy with dystonia and motor neuropathy, 613724*
SCUBE3	*100*	*99,8*	*100*	*100*	*Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 619184*
SCYL1	*100*	*99,9*	*100*	*100*	*Spinocerebellar ataxia, autosomal recessive 21, 616719*
SCYL2	*96,2*	*88*	*100*	*100*	*Arthrogryposis multiplex congenita 4, neurogenic, with agenesis of the corpus callosum, 618766*
SDCCAG8	*99,8*	*99,8*	*100*	*100*	*Senior-Loken syndrome 7, 613615* *Bardet-Biedl syndrome 16, 615993*
SDHA	*84,5*	*77,9*	*100*	*100*	*Cardiomyopathy, dilated, 1GG, 613642* *Mitochondrial complex II deficiency, nuclear type 1, 252011* *Neurodegeneration with ataxia and late-onset optic atrophy, 619259* *Paragangliomas 5, 614165*
SDHAF1	*100*	*98,4*	*100*	*100*	*Mitochondrial complex II deficiency, nuclear type 2, 619166*
SDHAF2	*94,6*	*93,5*	*99,4*	*96,4*	*Paragangliomas 2, 601650*
SDHB	*100*	*100*	*100*	*100*	*Paragangliomas 4, 115310* *Mitochondrial complex II deficiency, nuclear type 4, 619224* *Gastrointestinal stromal tumor, 606764* *Pheochromocytoma, 171300* *Paraganglioma and gastric stromal sarcoma, 606864*
SDHC	*100*	*98,9*	*100*	*100*	*Paragangliomas 3, 605373* *Paraganglioma and gastric stromal sarcoma, 606864* *Gastrointestinal stromal tumor, 606764*
SDHD	*53,8*	*49*	*80,1*	*80,1*	*Paragangliomas 1, with or without deafness, 168000* *Paraganglioma and gastric stromal sarcoma, 606864* *Mitochondrial complex II deficiency, nuclear type 3, 619167* *Pheochromocytoma, 171300*
SDR9C7	*100*	*100*	*100*	*100*	*Ichthyosis, congenital, autosomal recessive 13, 617574*
SEC23A	*99,7*	*97*	*100*	*100*	*Craniolenticulosutural dysplasia, 607812*
SEC23B	*99,9*	*99,1*	*100*	*100*	*?Cowden syndrome 7, 616858* *Dyserythropoietic anemia, congenital, type II, 224100*
SEC24D	*99,9*	*99,3*	*100*	*100*	*Cole-Carpenter syndrome 2, 616294*
SEC31A	*99*	*96,2*	*100*	*100*	*?Neurodevelopmental disorder with spastic quadriplegia, optic atrophy, seizures, and structural brain anomalies, 618651*

SEC61A1	*100*	*100*	*100*	*100*	*Tubulointerstitial kidney disease, autosomal dominant, 5, 617056*
SEC61B	*97,4*	*89,1*	*100*	*100*	*No OMIM disease ID*
SEC63	*86,2*	*77,3*	*100*	*100*	*Polycystic liver disease 2, 617004*
SECISBP2	*99,5*	*95,9*	*100*	*100*	*Thyroid hormone metabolism, abnormal, 609698*
SELENBP1	*100*	*99,7*	*100*	*100*	*Extraoral halitosis due to MTO deficiency, 618148*
SELENOI	*99,9*	*99,8*	*100*	*99,9*	*Spastic paraplegia 81, autosomal recessive, 618768*
SELENON	*84,3*	*84*	*87,8*	*85,1*	*Myopathy, congenital, with fiber-type disproportion, 255310* *Muscular dystrophy, rigid spine, 1, 602771*
SEMA3A	*100*	*99,7*	*100*	*100*	*No OMIM disease ID*
SEMA3E	*99,1*	*98,9*	*100*	*100*	*?CHARGE syndrome, 214800*
SEMA4A	*100*	*99,4*	*100*	*100*	*Retinitis pigmentosa 35, 610282* *Cone-rod dystrophy 10, 610283*
SEMA6B	*82,4*	*75,5*	*100*	*100*	*Epilepsy, progressive myoclonic, 11, 618876*
SEPSECS	*99,9*	*99,6*	*100*	*100*	*Pontocerebellar hypoplasia type 2D, 613811*
SEPTIN12	*100*	*99,1*	*100*	*100*	*Spermatogenic failure 10, 614822*
SEPTIN9	*100*	*99,5*	*100*	*100*	*Amyotrophy, hereditary neuralgic, 162100*
SERAC1	*99,6*	*99,5*	*100*	*99,9*	*3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739*
SERPINA1	*100*	*100*	*100*	*100*	*Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490* *Emphysema due to AAT deficiency, 613490* *Emphysema-cirrhosis, due to AAT deficiency, 613490*
SERPINA12	*100*	*100*	*100*	*100*	*No OMIM disease ID*
SERPINA3	*100*	*100*	*100*	*100*	*Alpha-1-antichymotrypsin deficiency,* *Cerebrovascular disease, occlusive,*
SERPINA6	*100*	*100*	*100*	*100*	*Corticosteroid-binding globulin deficiency, 611489*
SERPINB6	*93,4*	*93,4*	*100*	*100*	*?Deafness, autosomal recessive 91, 613453*
SERPINB7	*100*	*99,8*	*100*	*99,6*	*Palmoplantar keratoderma, Nagashima type, 615598*
SERPINB8	*95*	*95*	*100*	*100*	*Peeling skin syndrome 5, 617115*
SERPINC1	*100*	*100*	*100*	*100*	*Thrombophilia due to antithrombin III deficiency, 613118*
SERPIND1	*100*	*100*	*100*	*100*	*Thrombophilia due to heparin cofactor II deficiency, 612356*
SERPINE1	*100*	*100*	*100*	*100*	*Plasminogen activator inhibitor-1 deficiency, 613329*
SERPINF1	*100*	*99,9*	*100*	*100*	*Osteogenesis imperfecta, type VI, 613982*

SERPINF2	*100*	*99,9*	*100*	*100*	*Alpha-2-plasmin inhibitor deficiency, 262850*
SERPING1	*99,6*	*96,4*	*100*	*100*	*Angioedema, hereditary, 1 and 2, 106100* *Complement component 4, partial deficiency of, 120790*
SERPINH1	*99,8*	*98*	*100*	*100*	*Osteogenesis imperfecta, type X, 613848*
SERPINI1	*99,8*	*98,9*	*100*	*99,9*	*Encephalopathy, familial, with neuroserpin inclusion bodies, 604218*
SET	*96,7*	*87,4*	*99,3*	*97,3*	*Mental retardation, autosomal dominant 58, 618106*
SETBP1	*99,5*	*98,3*	*100*	*100*	*Schinzel-Giedion midface retraction syndrome, 269150* *Mental retardation, autosomal dominant 29, 616078*
SETD1A	*100*	*99,7*	*100*	*100*	*Epilepsy, early-onset, with or without developmental delay, 618832* *Neurodevelopmental disorder with speech impairment and dysmorphic facies, 619056*
SETD1B	*98,3*	*97,6*	*100*	*100*	*Intellectual developmental disorder with seizures and language delay, 619000*
SETD2	*99,9*	*99,6*	*100*	*100*	*Luscan-Lumish syndrome, 616831*
SETD5	*100*	*99,7*	*98*	*98*	*Mental retardation, autosomal dominant 23, 615761*
SETX	*99,8*	*99,6*	*100*	*100*	*Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002* *Amyotrophic lateral sclerosis 4, juvenile, 602433*
SEZ6	*100*	*98,3*	*100*	*100*	*No OMIM disease ID*
SF3B1	*99,5*	*98,2*	*100*	*100*	*Myelodysplastic syndrome, somatic, 614286*
SF3B4	*99,8*	*94,1*	*100*	*100*	*Acrofacial dysostosis 1, Nager type, 154400*
SFRP4	*99,8*	*99,1*	*100*	*100*	*Pyle disease, 265900*
SFTPA1	*100*	*100*	*100*	*100*	*No OMIM disease ID*
SFTPA2	*100*	*100*	*100*	*100*	*Pulmonary fibrosis, idiopathic, 178500*
SFTPB	*100*	*99,2*	*100*	*100*	*Surfactant metabolism dysfunction, pulmonary, 1, 265120*
SFTPC	*99,5*	*95,7*	*100*	*100*	*Surfactant metabolism dysfunction, pulmonary, 2, 610913*
SFXN4	*99,6*	*97,4*	*100*	*100*	*Combined oxidative phosphorylation deficiency 18, 615578*
SGCA	*100*	*99,6*	*100*	*100*	*Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099*
SGCB	*97,8*	*96,5*	*100*	*99,9*	*Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286*
SGCD	*99,6*	*96,5*	*100*	*100*	*Cardiomyopathy, dilated, 1L, 606685* *Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287*
SGCE	*88*	*83,7*	*91,2*	*91,2*	*Dystonia-11, myoclonic, 159900*
SGCG	*100*	*99,4*	*100*	*100*	*Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700*
SGMS2	*100*	*100*	*100*	*100*	*Calvarial doughnut lesions with bone fragility with or without spondylometaphyseal dysplasia, 126550*

SGO1	*99,5*	*99*	*100*	*100*	*Chronic atrial and intestinal dysrhythmia, 616201*
SGPL1	*100*	*100*	*100*	*100*	*Nephrotic syndrome, type 14, 617575*
SGSH	*94,8*	*94,1*	*100*	*100*	*Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900*
SH2B3	*99*	*94,7*	*100*	*100*	*Thrombocythemia, somatic, 187950* *Myelofibrosis, somatic, 254450* *Erythrocytosis, somatic, 133100*
SH2D1A	*97,8*	*92,9*	*100*	*100*	*Lymphoproliferative syndrome, X-linked, 1, 308240*
SH3BP2	*91,4*	*91,3*	*97,2*	*95,9*	*Cherubism, 118400*
SH3KBP1	*98,9*	*93,5*	*100*	*99,9*	*?Immunodeficiency 61, 300310*
SH3PXD2B	*100*	*99,9*	*100*	*100*	*Frank-ter Haar syndrome, 249420*
SH3TC2	*100*	*99,4*	*100*	*100*	*Charcot-Marie-Tooth disease, type 4C, 601596* *Mononeuropathy of the median nerve, mild, 613353*
SHANK2	*97,7*	*97,5*	*98,9*	*98,9*	*No OMIM disease ID*
SHANK3	*92,4*	*84,9*	*96*	*91,6*	*Phelan-McDermid syndrome, 606232*
SHH	*100*	*100*	*100*	*100*	*Microphthalmia with coloboma 5, 611638* *Schizencephaly, 269160* *Single median maxillary central incisor, 147250* *Holoprosencephaly 3, 142945*
SHMT2	*100*	*100*	*100*	*100*	*Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121*
SHOC2	*99,8*	*99,6*	*100*	*99,9*	*Noonan syndrome-like with loose anagen hair 1, 607721*
SHOX	*71,5*	*60,7*	*95,1*	*95,1*	*Short stature, idiopathic familial, 300582* *Leri-Weill dyschondrosteosis, 127300* *Langer mesomelic dysplasia, 249700* *Short stature, idiopathic familial, 300582* *Langer mesomelic dysplasia, 249700* *Leri-Weill dyschondrosteosis, 127300*
SHROOM3	*98,5*	*97,5*	*100*	*100*	*No OMIM disease ID*
SHROOM4	*99,8*	*98,1*	*100*	*100*	*Intellectual developmental disorder, X-linked syndromic, Stocco dos Santos type, 300434*
SI	*99*	*95,9*	*100*	*99,9*	*Sucrase-isomaltase deficiency, congenital, 222900*
SIAH1	*100*	*99,9*	*100*	*100*	*Buratti-Harel syndrome, 619314*
SIGLEC7	*100*	*99,7*	*100*	*100*	*No OMIM disease ID*
SIGMAR1	*100*	*100*	*100*	*100*	*?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726* *?Amyotrophic lateral sclerosis 16, juvenile, 614373*

SIK1	*98,2*	*93,9*	*100*	*100*	*Developmental and epileptic encephalopathy 30, 616341*
SIK3	*99,7*	*98*	*99,3*	*98*	*?Spondyloepimetaphyseal dysplasia, Krakow type, 618162*
SIL1	*98,7*	*96*	*100*	*100*	*Marinesco-Sjogren syndrome, 248800*
SIN3A	*99,8*	*98,2*	*100*	*100*	*Witteveen-Kolk syndrome, 613406*
SIN3B	*96,6*	*96*	*100*	*100*	*No OMIM disease ID*
SIPA1L3	*100*	*99,3*	*100*	*100*	*?Cataract 45, 616851*
SIX1	*100*	*99,7*	*100*	*100*	*Deafness, autosomal dominant 23, 605192* *Branchiootic syndrome 3, 608389*
SIX3	*99,3*	*96,9*	*100*	*99,8*	*Schizencephaly, 269160* *Holoprosencephaly 2, 157170*
SIX5	*96,9*	*90,1*	*100*	*100*	*Branchiootorenal syndrome 2, 610896*
SIX6	*100*	*100*	*100*	*100*	*Optic disc anomalies with retinal and/or macular dystrophy, 212550*
SKI	*99,7*	*97,1*	*100*	*99,7*	*Shprintzen-Goldberg syndrome, 182212*
SKIV2L	*100*	*99,5*	*100*	*100*	*Trichohepatoenteric syndrome 2, 614602*
SLC10A1	*99,8*	*97,4*	*100*	*100*	*Hypercholanemia, familial 2, 619256*
SLC10A2	*100*	*100*	*100*	*100*	*?Bile acid malabsorption, primary, 1, 613291*
SLC10A7	*99,5*	*98,1*	*100*	*100*	*Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363*
SLC11A2	*98,1*	*97,4*	*100*	*100*	*Anemia, hypochromic microcytic, with iron overload 1, 206100*
SLC12A1	*96,2*	*96*	*96,2*	*96,2*	*Bartter syndrome, type 1, 601678*
SLC12A2	*94,4*	*92,4*	*100*	*100*	*Kilquist syndrome, 619080* *Delpire-McNeill syndrome, 619083* *Deafness, autosomal dominant 78, 619081*
SLC12A3	*100*	*100*	*100*	*100*	*Gitelman syndrome, 263800*
SLC12A5	*83,9*	*83,8*	*97,4*	*97,4*	*Developmental and epileptic encephalopathy 34, 616645*
SLC12A6	*100*	*100*	*100*	*100*	*Agenesis of the corpus callosum with peripheral neuropathy, 218000*
SLC13A3	*99,8*	*97,9*	*100*	*100*	*Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate, 618384*
SLC13A5	*100*	*100*	*100*	*100*	*Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905*
SLC16A1	*100*	*98,6*	*100*	*100*	*Hyperinsulinemic hypoglycemia, familial, 7, 610021* *Erythrocyte lactate transporter defect, 245340* *Monocarboxylate transporter 1 deficiency, 616095*
SLC16A12	*100*	*99,9*	*100*	*100*	*Cataract 47, juvenile, with microcornea, 612018*

SLC16A2	*97,6*	*88,4*	*100*	*100*	*Allan-Herndon-Dudley syndrome, 300523*
SLC17A5	*99,6*	*96,2*	*100*	*100*	*Salla disease, 604369* *Sialic acid storage disorder, infantile, 269920*
SLC17A8	*100*	*99,9*	*100*	*100*	*Deafness, autosomal dominant 25, 605583*
SLC17A9	*96,7*	*95,7*	*100*	*100*	*Porokeratosis 8, disseminated superficial actinic type, 616063*
SLC18A2	*99,9*	*99,6*	*100*	*100*	*?Parkinsonism-dystonia, infantile, 2, 618049*
SLC18A3	*100*	*100*	*100*	*100*	*Myasthenic syndrome, congenital, 21, presynaptic, 617239*
SLC19A1	*99,2*	*96*	*100*	*100*	*?Megaloblastic anemia, folate-responsive, 601775*
SLC19A2	*100*	*98,5*	*100*	*100*	*Thiamine-responsive megaloblastic anemia syndrome, 249270*
SLC19A3	*97,8*	*97*	*98,7*	*98,7*	*Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483*
SLC1A1	*100*	*99,5*	*100*	*100*	*Dicarboxylic aminoaciduria, 222730*
SLC1A2	*96,1*	*94,8*	*100*	*100*	*Developmental and epileptic encephalopathy 41, 617105*
SLC1A3	*100*	*100*	*100*	*100*	*Episodic ataxia, type 6, 612656*
SLC1A4	*99,6*	*97*	*100*	*100*	*Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657*
SLC20A2	*99,9*	*97,6*	*100*	*100*	*Basal ganglia calcification, idiopathic, 1, 213600*
SLC22A12	*100*	*99,8*	*100*	*100*	*Hypouricemia, renal, 220150*
SLC22A18	*100*	*98,7*	*100*	*100*	*Breast cancer, somatic, 114480* *Lung cancer, somatic, 211980* *Rhabdomyosarcoma, somatic, 268210*
SLC22A4	*100*	*99,4*	*100*	*100*	*No OMIM disease ID*
SLC22A5	*100*	*99,6*	*100*	*100*	*Carnitine deficiency, systemic primary, 212140*
SLC24A1	*100*	*99,9*	*100*	*100*	*Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830*
SLC24A4	*100*	*99,5*	*100*	*100*	*Amelogenesis imperfecta, type IIA5, 615887*
SLC24A5	*99,6*	*98,5*	*100*	*100*	*Albinism, oculocutaneous, type VI, 113750*
SLC25A1	*96,9*	*89,8*	*99,7*	*98,2*	*Combined D-2- and L-2-hydroxyglutaric aciduria, 615182* *Myasthenic syndrome, congenital, 23, presynaptic, 618197*
SLC25A10	*76,4*	*70,3*	*100*	*100*	*?Mitochondrial DNA depletion syndrome 19, 618972*
SLC25A11	*100*	*100*	*100*	*100*	*Paragangliomas 6, 618464*
SLC25A12	*100*	*99,2*	*100*	*100*	*Developmental and epileptic encephalopathy 39, 612949*
SLC25A13	*100*	*99,4*	*100*	*100*	*Citrullinemia, type II, neonatal-onset, 605814* *Citrullinemia, adult-onset type II, 603471*

SLC25A15	*99,3*	*96,6*	*100*	*100*	*Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970*
SLC25A19	*99,9*	*98*	*100*	*100*	*Microcephaly, Amish type, 607196* *Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710*
SLC25A20	*100*	*98,9*	*100*	*100*	*Carnitine-acylcarnitine translocase deficiency, 212138*
SLC25A21	*100*	*99,6*	*100*	*100*	*?Mitochondrial DNA depletion syndrome 18, 618811*
SLC25A22	*99,2*	*96,5*	*100*	*100*	*Developmental and epileptic encephalopathy 3, 609304*
SLC25A24	*99,3*	*98,8*	*99,7*	*99,7*	*Fontaine progeroid syndrome, 612289*
SLC25A26	*99,8*	*98,1*	*100*	*100*	*Combined oxidative phosphorylation deficiency 28, 616794*
SLC25A3	*99,7*	*96,9*	*100*	*100*	*Mitochondrial phosphate carrier deficiency, 610773*
SLC25A32	*100*	*100*	*100*	*99,9*	*?Exercise intolerance, riboflavin-responsive, 616839*
SLC25A37	*100*	*100*	*100*	*100*	*No OMIM disease ID*
SLC25A38	*97,4*	*93,3*	*100*	*100*	*Anemia, sideroblastic, 2, pyridoxine-refractory, 205950*
SLC25A4	*100*	*99,8*	*100*	*100*	*Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418* *Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283* *Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184*
SLC25A42	*97,1*	*94,3*	*100*	*100*	*Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416*
SLC25A46	*99,7*	*98,6*	*100*	*100*	*Neuropathy, hereditary motor and sensory, type VIB, 616505* *Pontocerebellar hypoplasia, type 1E, 619303*
SLC26A1	*100*	*99,7*	*100*	*100*	*?Nephrolithiasis, calcium oxalate, 167030*
SLC26A2	*100*	*100*	*100*	*100*	*Epiphyseal dysplasia, multiple, 4, 226900* *De la Chapelle dysplasia, 256050* *Diastrophic dysplasia, 222600* *Diastrophic dysplasia, broad bone-platyspondylic variant, 222600* *Achondrogenesis Ib, 600972* *Atelosteogenesis, type II, 256050*
SLC26A3	*100*	*99,5*	*100*	*100*	*Diarrhea 1, secretory chloride, congenital, 214700*
SLC26A4	*99,9*	*99,7*	*100*	*100*	*Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791* *Pendred syndrome, 274600*
SLC26A5	*98,7*	*95,9*	*100*	*100*	*?Deafness, autosomal recessive 61, 613865*
SLC26A8	*99,9*	*99,3*	*100*	*100*	*Spermatogenic failure 3, 606766*
SLC27A4	*100*	*99,9*	*100*	*100*	*Ichthyosis prematurity syndrome, 608649*
SLC28A1	*100*	*98,3*	*100*	*100*	*No OMIM disease ID*

SLC29A3	*100*	*99,5*	*100*	*100*	*Histiocytosis-lymphadenopathy plus syndrome, 602782*
SLC2A1	*92,8*	*92,7*	*100*	*100*	*Dystonia 9, 601042* *GLUT1 deficiency syndrome 1, infantile onset, severe, 606777* *Stomatin-deficient cryohydrocytosis with neurologic defects, 608885* *GLUT1 deficiency syndrome 2, childhood onset, 612126*
SLC2A10	*97,7*	*97,7*	*100*	*100*	*Arterial tortuosity syndrome, 208050*
SLC2A2	*100*	*99,8*	*100*	*100*	*Fanconi-Bickel syndrome, 227810*
SLC2A9	*99,3*	*95*	*100*	*100*	*Hypouricemia, renal, 2, 612076*
SLC30A10	*100*	*100*	*100*	*100*	*Hypermanganesemia with dystonia 1, 613280*
SLC30A2	*100*	*99*	*100*	*100*	*Zinc deficiency, transient neonatal, 608118*
SLC30A5	*99,4*	*96,8*	*100*	*99,9*	*No OMIM disease ID*
SLC30A9	*98,7*	*94,3*	*100*	*99,9*	*?Birk-Landau-Perez syndrome, 617595*
SLC33A1	*99,8*	*98,5*	*100*	*99,8*	*Spastic paraplegia 42, autosomal dominant, 612539* *Congenital cataracts, hearing loss, and neurodegeneration, 614482*
SLC34A1	*100*	*99,7*	*100*	*100*	*?Fanconi renotubular syndrome 2, 613388* *Hypercalcemia, infantile, 2, 616963* *Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286*
SLC34A2	*100*	*99,6*	*100*	*100*	*Pulmonary alveolar microlithiasis, 265100*
SLC34A3	*100*	*99,3*	*100*	*100*	*Hypophosphatemic rickets with hypercalciuria, 241530*
SLC35A1	*99,7*	*99,3*	*100*	*100*	*Congenital disorder of glycosylation, type IIf, 603585*
SLC35A2	*99,6*	*97,7*	*100*	*100*	*Congenital disorder of glycosylation, type IIm, 300896*
SLC35A3	*80,4*	*78,8*	*81*	*80,9*	*?Arthrogryposis, mental retardation, and seizures, 615553*
SLC35C1	*100*	*99,4*	*100*	*100*	*Congenital disorder of glycosylation, type IIc, 266265*
SLC35D1	*99,6*	*97,6*	*100*	*99,2*	*Schneckenbecken dysplasia, 269250*
SLC36A2	*100*	*99,8*	*99,9*	*99,8*	*Iminoglycinuria, digenic, 242600* *Hyperglycinuria, 138500*
SLC37A4	*99,8*	*97,6*	*100*	*100*	*Glycogen storage disease Ib, 232220* *Congenital disorder of glycosylation, type IIw, 619525* *Glycogen storage disease Ic, 232240*
SLC38A8	*99,9*	*97,9*	*100*	*100*	*Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218*
SLC39A13	*99,9*	*97,9*	*100*	*100*	*Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350*
SLC39A14	*100*	*99*	*93,5*	*93,5*	*?Hyperostosis cranalis interna, 144755* *Hypermanganesemia with dystonia 2, 617013*

SLC39A4	*99,3*	*96,3*	*100*	*100*	*Acrodermatitis enteropathica, 201100*
SLC39A5	*99,9*	*99*	*100*	*100*	*Myopia 24, autosomal dominant, 615946*
SLC39A7	*100*	*99,8*	*100*	*100*	*No OMIM disease ID*
SLC39A8	*100*	*99,7*	*100*	*100*	*Congenital disorder of glycosylation, type IIn, 616721*
SLC3A1	*100*	*99,7*	*96,6*	*96,6*	*Cystinuria, 220100*
SLC40A1	*99,9*	*98,6*	*100*	*99,9*	*Hemochromatosis, type 4, 606069*
SLC41A1	*100*	*99,9*	*100*	*100*	*?Nephronophthisis-like nephropathy 2, 619468*
SLC44A1	*98,2*	*98,1*	*100*	*99,9*	*Neurodegeneration, childhood-onset, with ataxia, tremor, optic atrophy, and cognitive decline, 618868*
SLC44A4	*100*	*99,1*	*100*	*100*	*?Deafness, autosomal dominant 72, 617606*
SLC45A1	*100*	*100*	*100*	*100*	*Intellectual developmental disorder with neuropsychiatric features, 617532*
SLC45A2	*100*	*99,9*	*100*	*100*	*Albinism, oculocutaneous, type IV, 606574*
SLC46A1	*100*	*98,5*	*100*	*100*	*Folate malabsorption, hereditary, 229050*
SLC4A1	*100*	*99,9*	*96,1*	*96,1*	*Distal renal tubular acidosis 1, 179800* *Spherocytosis, type 4, 612653* *Distal renal tubular acidosis 4 with hemolytic anemia, 611590* *Cryohydrocytosis, 185020* *Ovalocytosis, SA type, 166900*
SLC4A11	*100*	*99,9*	*100*	*100*	*Corneal endothelial dystrophy, autosomal recessive, 217700* *Corneal dystrophy, Fuchs endothelial, 4, 613268* *Corneal endothelial dystrophy and perceptive deafness, 217400*
SLC4A4	*99,9*	*99,4*	*100*	*100*	*Renal tubular acidosis, proximal, with ocular abnormalities, 604278*
SLC51A	*100*	*99,8*	*100*	*100*	*?Cholestasis, progressive familial intrahepatic, 6, 619484*
SLC51B	*100*	*99,9*	*100*	*100*	*?Bile acid malabsorption, primary, 2, 619481*
SLC52A1	*100*	*100*	*100*	*100*	*Riboflavin deficiency, 615026*
SLC52A2	*100*	*100*	*100*	*100*	*Brown-Vialetto-Van Laere syndrome 2, 614707*
SLC52A3	*100*	*100*	*100*	*100*	*?Fazio-Londe disease, 211500* *Brown-Vialetto-Van Laere syndrome 1, 211530*
SLC5A1	*100*	*99,8*	*100*	*100*	*Glucose/galactose malabsorption, 606824*
SLC5A2	*100*	*100*	*100*	*100*	*Renal glucosuria, 233100*
SLC5A5	*100*	*99,9*	*100*	*100*	*Thyroid dyshormonogenesis 1, 274400*
SLC5A6	*100*	*100*	*100*	*100*	*Neurodegeneration, infantile-onset, biotin-responsive, 618973*

SLC5A7	*100*	*100*	*100*	*100*	*Neuronopathy, distal hereditary motor, type VIIA, 158580* *Myasthenic syndrome, congenital, 20, presynaptic, 617143*
SLC6A1	*96,7*	*96,6*	*100*	*100*	*Myoclonic-atonic epilepsy, 616421*
SLC6A17	*100*	*100*	*100*	*100*	*Mental retardation, autosomal recessive 48, 616269*
SLC6A19	*100*	*100*	*100*	*100*	*Iminoglycinuria, digenic, 242600* *Hartnup disorder, 234500* *Hyperglycinuria, 138500*
SLC6A2	*100*	*99,6*	*100*	*100*	*?Orthostatic intolerance, 604715*
SLC6A20	*100*	*99,8*	*100*	*100*	*Iminoglycinuria, digenic, 242600* *Hyperglycinuria, 138500*
SLC6A3	*100*	*99,9*	*100*	*100*	*Parkinsonism-dystonia, infantile, 1, 613135*
SLC6A5	*100*	*99,9*	*100*	*100*	*Hyperekplexia 3, 614618*
SLC6A6	*85,5*	*83,3*	*100*	*100*	*Hypotaurinemic retinal degeneration and cardiomyopathy, 145350*
SLC6A8	*94,8*	*83*	*99,9*	*99,5*	*Cerebral creatine deficiency syndrome 1, 300352*
SLC6A9	*100*	*99,6*	*100*	*100*	*Glycine encephalopathy with normal serum glycine, 617301*
SLC7A14	*100*	*100*	*100*	*100*	*Retinitis pigmentosa 68, 615725*
SLC7A6OS	*99,9*	*98,4*	*100*	*100*	*Epilepsy, progressive myoclonic, 12, 619191*
SLC7A7	*100*	*99,9*	*100*	*100*	*Lysinuric protein intolerance, 222700*
SLC7A9	*100*	*99,4*	*100*	*100*	*Cystinuria, 220100*
SLC9A1	*100*	*100*	*100*	*100*	*Lichtenstein-Knorr syndrome, 616291*
SLC9A3	*90,5*	*86*	*96*	*93,6*	*Diarrhea 8, secretory sodium, congenital, 616868*
SLC9A3R1	*99,9*	*98,2*	*100*	*100*	*Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287*
SLC9A6	*94,7*	*90,2*	*99,7*	*97*	*Intellectual developmental disorder, X-linked syndromic, Christianson type, 300243*
SLC9A7	*97,4*	*89,8*	*100*	*99,6*	*Intellectual developmental disorder, X-linked 108, 301024*
SLCO1B1	*98,3*	*92*	*100*	*99,5*	*Hyperbilirubinemia, Rotor type, digenic, 237450*
SLCO1B3	*98,1*	*88,4*	*100*	*99,8*	*Hyperbilirubinemia, Rotor type, digenic, 237450*
SLCO2A1	*99,9*	*98*	*100*	*100*	*Hypertrophic osteoarthropathy, primary, autosomal dominant, 167100* *Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441*
SLCO5A1	*99,3*	*98,2*	*100*	*100*	*No OMIM disease ID*
SLFN14	*100*	*100*	*100*	*100*	*Bleeding disorder, platelet-type, 20, 616913*
SLIT3	*97,9*	*95,6*	*100*	*100*	*No OMIM disease ID*

SLITRK1	*100*	*99,9*	*100*	*100*	*Tourette syndrome, 137580* *?Trichotillomania, 613229*
SLITRK6	*100*	*99,9*	*100*	*100*	*Deafness and myopia, 221200*
SLMAP	*98,4*	*92,5*	*100*	*99,9*	*No OMIM disease ID*
SLURP1	*100*	*99,4*	*100*	*100*	*Meleda disease, 248300*
SLX4	*100*	*99,9*	*100*	*100*	*Fanconi anemia, complementation group P, 613951*
SMAD1	*99,8*	*98,2*	*100*	*100*	*No OMIM disease ID*
SMAD2	*100*	*99,8*	*100*	*100*	*No OMIM disease ID*
SMAD3	*99,9*	*98,4*	*100*	*100*	*Loeys-Dietz syndrome 3, 613795*
SMAD4	*99,9*	*99,9*	*100*	*100*	*Pancreatic cancer, somatic, 260350* *Myhre syndrome, 139210* *Polyposis, juvenile intestinal, 174900* *Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050*
SMAD6	*90,7*	*79,3*	*100*	*99,9*	*Aortic valve disease 2, 614823*
SMAD9	*100*	*99,3*	*100*	*100*	*Pulmonary hypertension, primary, 2, 615342*
SMARCA1	*99,1*	*96,3*	*99,9*	*98,8*	*No OMIM disease ID*
SMARCA2	*96,7*	*96,3*	*97,8*	*96,9*	*Nicolaides-Baraitser syndrome, 601358* *Blepharophimosis-impaired intellectual development syndrome, 619293*
SMARCA4	*99,9*	*99,2*	*100*	*100*	*Coffin-Siris syndrome 4, 614609*
SMARCA5	*99,3*	*96,9*	*100*	*99,9*	*No OMIM disease ID*
SMARCAD1	*98,8*	*95,4*	*100*	*100*	*Basan syndrome, 129200* *Huriez syndrome, 181600* *Adermatoglyphia, 136000*
SMARCAL1	*100*	*99,8*	*100*	*100*	*Schimke immunoosseous dysplasia, 242900*
SMARCB1	*100*	*99,9*	*100*	*100*	*Rhabdoid tumors, somatic, 609322* *Coffin-Siris syndrome 3, 614608*
SMARCC2	*98,8*	*95,7*	*100*	*100*	*Coffin-Siris syndrome 8, 618362*
SMARCD1	*94,9*	*89,8*	*100*	*99,9*	*Coffin-Siris syndrome 11, 618779*
SMARCD2	*87*	*85,8*	*99,9*	*98,6*	*Specific granule deficiency 2, 617475*
SMARCE1	*93,7*	*85,9*	*100*	*100*	*Coffin-Siris syndrome 5, 616938*
SMC1A	*99,6*	*97,1*	*100*	*99,9*	*Cornelia de Lange syndrome 2, 300590* *Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044*

SMC3	*94,5*	*89*	*100*	*99,9*	*Cornelia de Lange syndrome 3, 610759*
SMCHD1	*99,3*	*96,4*	*100*	*99,9*	*Bosma arhinia microphthalmia syndrome, 603457* *Fascioscapulohumeral muscular dystrophy 2, digenic, 158901*
SMDT1	*100*	*100*	*100*	*100*	*No OMIM disease ID*
SMG8	*100*	*100*	*100*	*100*	*Alzahrani-Kuwahara syndrome, 619268*
SMG9	*100*	*100*	*100*	*100*	*Heart and brain malformation syndrome, 616920*
SMN1	*99,7*	*96,1*	*94,6*	*94,6*	*Spinal muscular atrophy-2, 253550* *Spinal muscular atrophy-4, 271150* *Spinal muscular atrophy-3, 253400* *Spinal muscular atrophy-1, 253300*
SMO	*98,9*	*94,7*	*100*	*100*	*Pallister-Hall-like syndrome, 241800* *Basal cell carcinoma, somatic, 605462* *Curry-Jones syndrome, somatic mosaic, 601707*
SMOC1	*99,8*	*98,2*	*100*	*100*	*Microphthalmia with limb anomalies, 206920*
SMOC2	*76,7*	*74,9*	*100*	*100*	*Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400*
SMPD1	*100*	*99,9*	*100*	*100*	*Niemann-Pick disease, type B, 607616* *Niemann-Pick disease, type A, 257200*
SMPD4	*99,6*	*95*	*100*	*100*	*Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622*
SMPX	*99,7*	*94,5*	*100*	*100*	*Deafness, X-linked 4, 300066*
SMS	*87,9*	*72,1*	*100*	*99,5*	*Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type, 309583*
SNAI2	*99,9*	*98*	*100*	*100*	*Waardenburg syndrome, type 2D, 608890* *Piebaldism, 172800*
SNAP25	*100*	*99,8*	*100*	*100*	*?Myasthenic syndrome, congenital, 18, 616330*
SNAP29	*100*	*100*	*100*	*100*	*Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528*
SNCA	*79,1*	*79,1*	*79,1*	*79,1*	*Dementia, Lewy body, 127750* *Parkinson disease 1, 168601* *Parkinson disease 4, 605543*
SNCB	*100*	*98,8*	*100*	*100*	*Dementia, Lewy body, 127750*
SNIP1	*99,2*	*97,3*	*100*	*100*	*Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501*
SNORA31	*NC*	*NC*	*NC*	*NC*	*No OMIM disease ID*
SNORD118	*NC*	*NC*	*NC*	*NC*	*Leukoencephalopathy, brain calcifications, and cysts, 614561*
SNRNP200	*99,8*	*98,3*	*100*	*100*	*Retinitis pigmentosa 33, 610359*
SNRPB	*100*	*98,6*	*100*	*100*	*Cerebrocostomandibular syndrome, 117650*

SNRPE	*98,7*	*90,9*	*100*	*100*	*Hypotrichosis 11, 615059*
SNRPN	*99,8*	*96,5*	*100*	*100*	*Prader-Willi syndrome, 176270*
SNTA1	*92,6*	*80,2*	*99,9*	*98,6*	*Long QT syndrome 12, 612955*
SNX10	*96,2*	*95,9*	*99,9*	*99,3*	*Osteopetrosis, autosomal recessive 8, 615085*
SNX14	*98,9*	*93,6*	*100*	*100*	*Spinocerebellar ataxia, autosomal recessive 20, 616354*
SNX27	*100*	*99,1*	*100*	*100*	*No OMIM disease ID*
SOBP	*98,5*	*95,9*	*97,4*	*95,5*	*Mental retardation, anterior maxillary protrusion, and strabismus, 613671*
SOCS1	*100*	*100*	*100*	*100*	*Autoinflammatory syndrome, familial, with or without immunodeficiency, 619375*
SOCS4	*99,9*	*99,3*	*100*	*99,9*	*No OMIM disease ID*
SOD1	*100*	*100*	*100*	*100*	*Spastic tetraplegia and axial hypotonia, progressive, 618598* *Amyotrophic lateral sclerosis 1, 105400*
SOD2	*100*	*100*	*100*	*100*	*No OMIM disease ID*
SOHLH1	*99,7*	*96,8*	*100*	*100*	*Ovarian dysgenesis 5, 617690* *Spermatogenic failure 32, 618115*
SON	*97,6*	*92,6*	*100*	*100*	*ZTTK syndrome, 617140*
SORD	*90,6*	*89,4*	*98,4*	*95,1*	*Sorbitol dehydrogenase deficiency with peripheral neuropathy, 618912*
SOS1	*99,6*	*97,9*	*100*	*99,9*	*Noonan syndrome 4, 610733* *?Fibromatosis, gingival, 1, 135300*
SOS2	*99,6*	*98,7*	*100*	*99,9*	*Noonan syndrome 9, 616559*
SOST	*100*	*99,6*	*100*	*100*	*Sclerosteosis 1, 269500* *Craniodiaphyseal dysplasia, autosomal dominant, 122860*
SOX10	*99,9*	*97,2*	*100*	*100*	*Waardenburg syndrome, type 4C, 613266* *PCWH syndrome, 609136* *Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584*
SOX11	*100*	*100*	*100*	*100*	*Coffin-Siris syndrome 9, 615866*
SOX17	*100*	*99,9*	*100*	*100*	*Vesicoureteral reflux 3, 613674*
SOX18	*75,2*	*55,5*	*95,7*	*91,7*	*Hypotrichosis-lymphedema-telangiectasia syndrome, 607823* *Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940*
SOX2	*100*	*99,8*	*100*	*100*	*Optic nerve hypoplasia and abnormalities of the central nervous system, 206900* *Microphthalmia, syndromic 3, 206900*
SOX3	*94,9*	*81,2*	*100*	*99,6*	*Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency, 300123* *Panhypopituitarism, X-linked, 312000*
SOX4	*97,7*	*90,6*	*99,8*	*97,9*	*Coffin-Siris syndrome 10, 618506*

SOX5	*99,8*	*97,6*	*100*	*100*	*Lamb-Shaffer syndrome, 616803*
SOX6	*99,9*	*98,9*	*100*	*99,9*	*Tolchin-Le Caignec syndrome, 618971*
SOX9	*100*	*99,9*	*100*	*100*	*Campomelic dysplasia with autosomal sex reversal, 114290* *Acampomelic campomelic dysplasia, 114290* *Campomelic dysplasia, 114290*
SP110	*100*	*100*	*100*	*100*	*Hepatic venoocclusive disease with immunodeficiency, 235550*
SP7	*99,9*	*99,2*	*100*	*100*	*Osteogenesis imperfecta, type XII, 613849*
SPAG1	*98,7*	*93,9*	*99,6*	*97,9*	*Ciliary dyskinesia, primary, 28, 615505*
SPAG17	*99,9*	*99,3*	*100*	*100*	*?Spermatogenic failure 55, 619380*
SPAG6	*99,9*	*99,5*	*100*	*100*	*No OMIM disease ID*
SPARC	*100*	*100*	*100*	*100*	*Osteogenesis imperfecta, type XVII, 616507*
SPART	*99,7*	*96,4*	*100*	*100*	*Troyer syndrome, 275900*
SPAST	*99,4*	*98,1*	*100*	*100*	*Spastic paraplegia 4, autosomal dominant, 182601*
SPATA16	*99,9*	*99,1*	*100*	*100*	*?Spermatogenic failure 6, 102530*
SPATA5	*99,8*	*99,5*	*100*	*100*	*Epilepsy, hearing loss, and mental retardation syndrome, 616577*
SPATA7	*99,6*	*98,2*	*100*	*100*	*Retinitis pigmentosa, juvenile, autosomal recessive, 604232* *Leber congenital amaurosis 3, 604232*
SPECC1L	*96*	*95*	*97,1*	*96,1*	*Opitz GBBB syndrome, type II, 145410* *Teebi hypertelorism syndrome, 145420* *?Facial clefting, oblique, 1, 600251*
SPEF2	*98*	*95,5*	*100*	*100*	*Spermatogenic failure 43, 618751*
SPEG	*97,2*	*91,1*	*99,7*	*99,7*	*Centronuclear myopathy 5, 615959*
SPEN	*100*	*99,8*	*100*	*100*	*Radio-Tartaglia syndrome, 619312*
SPG11	*99,8*	*99*	*100*	*100*	*Amyotrophic lateral sclerosis 5, juvenile, 602099* *Charcot-Marie-Tooth disease, axonal, type 2X, 616668* *Spastic paraplegia 11, autosomal recessive, 604360*
SPG21	*98,9*	*94,7*	*100*	*100*	*Mast syndrome, 248900*
SPG7	*90,4*	*86,7*	*100*	*100*	*Spastic paraplegia 7, autosomal recessive, 607259*
SPINK1	*99,9*	*99*	*100*	*100*	*Tropical calcific pancreatitis, 608189* *Pancreatitis, hereditary, 167800*
SPINK2	*99,3*	*98,2*	*99,3*	*99,3*	*?Spermatogenic failure 29, 618091*
SPINK5	*99,8*	*99,6*	*100*	*99,9*	*Netherton syndrome, 256500*

SPINT2	*97,5*	*78,8*	*100*	*100*	*Diarrhea 3, secretory sodium, congenital, syndromic, 270420*
SPNS2	*92,3*	*89,6*	*96,7*	*95*	*?Deafness, autosomal recessive 115, 618457*
SPOCK1	*100*	*99,7*	*100*	*100*	*No OMIM disease ID*
SPOP	*100*	*100*	*100*	*100*	*Nabais Sa-de Vries syndrome, type 1, 618828* *Nabais Sa-de Vries syndrome, type 2, 618829*
SPP2	*99,8*	*99,8*	*100*	*100*	*No OMIM disease ID*
SPPL2A	*84,3*	*70,5*	*100*	*99,7*	*No OMIM disease ID*
SPR	*100*	*99,4*	*100*	*100*	*Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716*
SPRED1	*99,8*	*98,2*	*100*	*100*	*Legius syndrome, 611431*
SPRTN	*100*	*100*	*100*	*100*	*Ruijs-Aalfs syndrome, 616200*
SPRY4	*100*	*100*	*100*	*100*	*Hypogonadotropic hypogonadism 17 with or without anosmia, 615266*
SPTA1	*99,9*	*98,8*	*100*	*100*	*Spherocytosis, type 3, 270970* *Elliptocytosis-2, 130600* *Pyropoikilocytosis, 266140*
SPTAN1	*99,1*	*97,9*	*100*	*100*	*Developmental and epileptic encephalopathy 5, 613477*
SPTB	*100*	*99,9*	*100*	*100*	*Anemia, neonatal hemolytic, fatal or near-fatal, 617948* *Elliptocytosis-3, 617948* *Spherocytosis, type 2, 616649*
SPTBN1	*99,9*	*99,4*	*100*	*100*	*Developmental delay, impaired speech, and behavioral abnormalities, 619475*
SPTBN2	*100*	*99,4*	*100*	*99,9*	*Spinocerebellar ataxia 5, 600224* *Spinocerebellar ataxia, autosomal recessive 14, 615386*
SPTBN4	*98,1*	*92,1*	*100*	*100*	*Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519*
SPTLC1	*98,7*	*93,7*	*100*	*100*	*Neuropathy, hereditary sensory and autonomic, type IA, 162400*
SPTLC2	*100*	*100*	*100*	*99,9*	*Neuropathy, hereditary sensory and autonomic, type IC, 613640*
SPTLC3	*100*	*99,8*	*100*	*100*	*No OMIM disease ID*
SQOR	*100*	*98*	*100*	*100*	*Sulfide:quinone oxidoreductase deficiency, 619221*
SQSTM1	*99,8*	*97,8*	*100*	*100*	*Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145* *Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437* *Myopathy, distal, with rimmed vacuoles, 617158* *Paget disease of bone 3, 167250*
SRC	*100*	*99,6*	*100*	*100*	*?Thrombocytopenia 6, 616937* *Colon cancer, advanced, somatic, 114500*
SRCAP	*99,7*	*98,9*	*100*	*100*	*Floating-Harbor syndrome, 136140*

SRD5A2	*100*	*98,8*	*100*	*100*	*Pseudovaginal perineoscrotal hypospadias, 264600*
SRD5A3	*100*	*99,1*	*100*	*100*	*Kahrizi syndrome, 612713* *Congenital disorder of glycosylation, type Iq, 612379*
SREBF1	*99,3*	*97,1*	*96,9*	*96,9*	*Ichthyosis, follicular, with atrichia and photophobia syndrome 2, 619016* *Mucoepithelial dysplasia, hereditary, 158310*
SRI	*99,8*	*96,4*	*100*	*100*	*No OMIM disease ID*
SRP54	*98*	*93,4*	*100*	*100*	*Neutropenia, severe congenital, 8, autosomal dominant, 618752*
SRP72	*98*	*90,1*	*100*	*100*	*Bone marrow failure syndrome 1, 614675*
SRPK3	*99*	*95,7*	*100*	*100*	*No OMIM disease ID*
SRPX2	*99,3*	*93,6*	*100*	*100*	*?Rolandic epilepsy, impaired intellectual development, and speech dyspraxia, 300643*
SRRM2	*100*	*99,8*	*100*	*100*	*No OMIM disease ID*
SRY	*50*	*49,9*	*50*	*50*	*46XY sex reversal 1, 400044*
SSBP1	*99,1*	*94,1*	*100*	*100*	*Optic atrophy 13 with retinal and foveal abnormalities, 165510*
SSR4	*100*	*99,5*	*100*	*100*	*Congenital disorder of glycosylation, type Iy, 300934*
SSTR5	*100*	*100*	*100*	*100*	*No OMIM disease ID*
SSX1	*82*	*81,3*	*100*	*100*	*?Sarcoma, synovial, 300813*
SSX2	*64,5*	*60,9*	*100*	*100*	*?Sarcoma, synovial, 300813*
ST14	*99,9*	*98,9*	*100*	*100*	*Ichthyosis, congenital, autosomal recessive 11, 602400*
ST3GAL3	*68,8*	*68,2*	*95,3*	*95,2*	*Developmental and epileptic encephalopathy 15, 615006* *Intellectual developmental disorder, autosomal recessive 12, 611090*
ST3GAL5	*85,9*	*84*	*98,7*	*98,6*	*Salt and pepper developmental regression syndrome, 609056*
STAB2	*100*	*99,8*	*100*	*100*	*No OMIM disease ID*
STAC3	*100*	*100*	*100*	*100*	*Myopathy, congenital, Baily-Bloch, 255995*
STAG1	*99,4*	*96,2*	*100*	*100*	*Mental retardation, autosomal dominant 47, 617635*
STAG2	*97*	*86,9*	*100*	*99,3*	*Holoprosencephaly 13, X-linked, 301043* *Mullegama-Klein-Martinez syndrome, 301022*
STAG3	*93,5*	*92,8*	*100*	*100*	*Premature ovarian failure 8, 615723*
STAMBP	*99,4*	*96,4*	*100*	*100*	*Microcephaly-capillary malformation syndrome, 614261*
STAR	*100*	*99,9*	*100*	*100*	*Lipoid adrenal hyperplasia, 201710*
STARD7	*98,9*	*93,2*	*100*	*100*	*Epilepsy, familial adult myoclonic, 2, 607876*

STAT1	*93,1*	*90,2*	*95,7*	*95*	*Immunodeficiency 31C, chronic mucocutaneous candidiasis, autosomal dominant, 614162* *Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892* *Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796*
STAT2	*100*	*99,4*	*100*	*100*	*Pseudo-TORCH syndrome 3, 618886* *Immunodeficiency 44, 616636*
STAT3	*99,9*	*99*	*100*	*100*	*Hyper-IgE recurrent infection syndrome, 147060* *Autoimmune disease, multisystem, infantile-onset, 1, 615952*
STAT4	*99,8*	*99,4*	*100*	*100*	*No OMIM disease ID*
STAT5B	*99,9*	*98,1*	*100*	*100*	*Growth hormone insensitivity with immune dysregulation 1, autosomal recessive, 245590* *Growth hormone insensitivity with immune dysregulation 2, autosomal dominant, 618985* *Leukemia, acute promyelocytic, somatic, 102578*
STAT6	*100*	*99,3*	*100*	*100*	*No OMIM disease ID*
STEAP3	*100*	*99,7*	*100*	*100*	*?Anemia, hypochromic microcytic, with iron overload 2, 615234*
CXorf56	*99,4*	*92,9*	*100*	*99,8*	*?Intellectual developmental disorder, X-linked 107, 301013*
STIL	*99,9*	*99,7*	*100*	*100*	*Microcephaly 7, primary, autosomal recessive, 612703*
STIM1	*99,9*	*97,5*	*100*	*100*	*Myopathy, tubular aggregate, 1, 160565* *Stormorken syndrome, 185070* *Immunodeficiency 10, 612783*
STING1	*99,7*	*96,3*	*100*	*100*	*STING-associated vasculopathy, infantile-onset, 615934*
STK11	*92,4*	*91,9*	*100*	*100*	*Melanoma, malignant, somatic, 155600* *Pancreatic cancer, somatic, 260350* *Peutz-Jeghers syndrome, 175200* *Testicular tumor, somatic, 273300*
STK36	*100*	*99,1*	*100*	*100*	*?Ciliary dyskinesia, primary, 46, 619436*
STK4	*99,9*	*99,7*	*100*	*100*	*T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868*
STN1	*99,9*	*99,8*	*100*	*100*	*Cerebroretinal microangiopathy with calcifications and cysts 2, 617341*
STOX1	*80,5*	*80,5*	*94,2*	*89*	*Preeclampsia/eclampsia 4, 609404*
STRA6	*100*	*99,9*	*100*	*100*	*Microphthalmia, syndromic 9, 601186* *Microphthalmia, isolated, with coloboma 8, 601186*
STRADA	*100*	*99*	*100*	*100*	*Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087*
STRC	*99,9*	*98,3*	*100*	*100*	*Deafness, autosomal recessive 16, 603720*
STS	*96,8*	*93,8*	*97,4*	*97,2*	*Ichthyosis, X-linked, 308100*
STT3A	*100*	*100*	*100*	*100*	*Congenital disorder of glycosylation, type Iw, 615596*

STT3B	*99,7*	*99,4*	*100*	*100*	*?Congenital disorder of glycosylation, type Ix, 615597*
STUB1	*100*	*98,2*	*100*	*100*	*Spinocerebellar ataxia 48, 618093* *Spinocerebellar ataxia, autosomal recessive 16, 615768*
STX11	*100*	*100*	*100*	*100*	*Hemophagocytic lymphohistiocytosis, familial, 4, 603552*
STX16	*100*	*99*	*100*	*100*	*Pseudohypoparathyroidism, type IB, 603233*
STX1B	*100*	*100*	*100*	*100*	*Generalized epilepsy with febrile seizures plus, type 9, 616172*
STX3	*93*	*92,5*	*100*	*100*	*Retinal dystrophy and microvillus inclusion disease, 619446* *Diarrhea 12, with microvillus atrophy, 619445*
STX5	*95*	*89,3*	*100*	*100*	*No OMIM disease ID*
STXBP1	*96,8*	*96,2*	*100*	*100*	*Developmental and epileptic encephalopathy 4, 612164*
STXBP2	*82,4*	*79,9*	*99,7*	*98*	*Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease, 613101*
SUCLA2	*88,8*	*79,4*	*99,9*	*99,8*	*Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073*
SUCLG1	*100*	*99,7*	*100*	*99,8*	*Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400*
SUCLG2	*91,7*	*79,1*	*100*	*100*	*No OMIM disease ID*
SUFU	*100*	*100*	*100*	*100*	*Joubert syndrome 32, 617757* *Medulloblastoma, desmoplastic, 155255* *Basal cell nevus syndrome, 109400*
SUGCT	*99,6*	*97,6*	*100*	*99,9*	*Glutaric aciduria III, 231690*
SULF1	*99,9*	*99,2*	*100*	*100*	*No OMIM disease ID*
SULT2B1	*100*	*100*	*100*	*100*	*Ichthyosis, congenital, autosomal recessive 14, 617571*
SUMF1	*98,3*	*92,5*	*100*	*100*	*Multiple sulfatase deficiency, 272200*
SUMO1	*60,5*	*45,3*	*69,4*	*69,4*	*?Orofacial cleft 10, 613705*
SUN5	*100*	*99,7*	*100*	*100*	*Spermatogenic failure 16, 617187*
SUOX	*100*	*100*	*100*	*100*	*Sulfite oxidase deficiency, 272300*
SUPT16H	*97*	*89,3*	*100*	*100*	*Neurodevelopmental disorder with dysmorphic facies and thin corpus callosum, 619480*
SURF1	*89,5*	*88,1*	*100*	*100*	*Charcot-Marie-Tooth disease, type 4K, 616684* *Mitochondrial complex IV deficiency, nuclear type 1, 220110*
SUZ12	*90,7*	*86,2*	*100*	*99,9*	*Imagawa-Matsumoto syndrome, 618786*
SVBP	*100*	*100*	*100*	*100*	*Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569*
SVIL	*99,9*	*98,7*	*100*	*100*	*Myofibrillar myopathy 10, 619040*

SYCE1	*99,9*	*98,8*	*100*	*100*	*?Spermatogenic failure 15, 616950* *?Premature ovarian failure 12, 616947*
SYCP2	*96,8*	*88,9*	*100*	*99,8*	*Spermatogenic failure 1, 258150*
SYCP3	*99*	*98,3*	*100*	*100*	*Pregnancy loss, recurrent, 4, 270960* *Spermatogenic failure 4, 270960*
SYK	*100*	*100*	*100*	*100*	*Immunodeficiency 82 with systemic inflammation, 619381*
SYN1	*82*	*71,6*	*100*	*99,9*	*Intellectual developmental disorder, X-linked 50, 300115* *Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491*
SYNCRIP	*97*	*82*	*100*	*100*	*No OMIM disease ID*
SYNE1	*98,1*	*97,5*	*98,8*	*98,8*	*Arthrogryposis multiplex congenita 3, myogenic type, 618484* *Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998* *Spinocerebellar ataxia, autosomal recessive 8, 610743*
SYNE2	*99,4*	*97,2*	*100*	*99,9*	*Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999*
SYNE4	*99,9*	*97,5*	*100*	*100*	*Deafness, autosomal recessive 76, 615540*
SYNGAP1	*98,8*	*97,4*	*100*	*100*	*Mental retardation, autosomal dominant 5, 612621*
SYNJ1	*99,7*	*98,1*	*100*	*100*	*Parkinson disease 20, early-onset, 615530* *Developmental and epileptic encephalopathy 53, 617389*
SYP	*99,9*	*96,2*	*100*	*100*	*Intellectual developmental disorder, X-linked 96, 300802*
SYT1	*99,5*	*97,5*	*100*	*100*	*Baker-Gordon syndrome, 618218*
SYT14	*61*	*60,4*	*100*	*100*	*?Spinocerebellar ataxia, autosomal recessive 11, 614229*
SYT2	*100*	*98,8*	*100*	*100*	*Myasthenic syndrome, congenital, 7A, presynaptic, and distal motor neuropathy, autosomal dominant, 616040* *Myasthenic syndrome, congenital, 7B, presynaptic, autosomal recessive, 619461*
SZT2	*99,6*	*99,3*	*100*	*99,9*	*Developmental and epileptic encephalopathy 18, 615476*
TAB2	*99,8*	*99,2*	*100*	*100*	*Congenital heart defects, nonsyndromic, 2, 614980*
TAC3	*99,9*	*93,6*	*100*	*100*	*Hypogonadotropic hypogonadism 10 with or without anosmia, 614839*
TACO1	*98,9*	*93,7*	*100*	*100*	*Mitochondrial complex IV deficiency, nuclear type 8, 619052*
TACR3	*100*	*100*	*100*	*100*	*Hypogonadotropic hypogonadism 11 with or without anosmia, 614840*
TACSTD2	*99,1*	*96,6*	*100*	*100*	*Corneal dystrophy, gelatinous drop-like, 204870*
TAF1	*99,2*	*95,7*	*100*	*100*	*Intellectual developmental disorder, X-linked syndromic 33, 300966* *Dystonia-Parkinsonism, X-linked, 314250*
TAF13	*99,6*	*99,1*	*100*	*99,9*	*Mental retardation, autosomal recessive 60, 617432*
TAF1C	*100*	*100*	*100*	*100*	*No OMIM disease ID*

TAF2	*99,5*	*98,6*	*100*	*99,9*	*Mental retardation, autosomal recessive 40, 615599*
TAF4B	*97,5*	*93,7*	*100*	*100*	*?Spermatogenic failure 13, 615841*
TAF6	*99,6*	*98,1*	*100*	*100*	*Alazami-Yuan syndrome, 617126*
TAZ	*99,3*	*93,7*	*100*	*100*	*Barth syndrome, 302060*
TAL1	*89,2*	*77,2*	*100*	*100*	*Leukemia, T-cell acute lymphocytic, somatic, 613065*
TAL2	*100*	*100*	*100*	*100*	*Leukemia, T-cell acute lymphocytic, somatic, 613065*
TALDO1	*100*	*98*	*100*	*100*	*Transaldolase deficiency, 606003*
TANC2	*99,8*	*98,9*	*100*	*100*	*Intellectual developmental disorder with autistic features and language delay, with or without seizures, 618906*
TANGO2	*100*	*99,3*	*100*	*100*	*Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878*
TAOK1	*99,6*	*97,5*	*100*	*100*	*No OMIM disease ID*
TAP1	*100*	*97,6*	*100*	*100*	*Bare lymphocyte syndrome, type I, 604571*
TAP2	*99,9*	*98,6*	*100*	*100*	*Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571*
TAPBP	*96,5*	*95,1*	*96,6*	*96,6*	*Bare lymphocyte syndrome, type I, 604571*
TAPT1	*93*	*87,1*	*98,5*	*94,1*	*Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897*
TARDBP	*100*	*99,9*	*100*	*100*	*Frontotemporal lobar degeneration, TARDBP-related, 612069* *Amyotrophic lateral sclerosis 10, with or without FTD, 612069*
TARS1	*99,8*	*97,4*	*100*	*100*	*Trichothiodystrophy 7, nonphotosensitive, 618546*
TARS2	*99,9*	*98,8*	*100*	*100*	*?Combined oxidative phosphorylation deficiency 21, 615918*
TASP1	*99,8*	*98,7*	*100*	*100*	*Suleiman-El-Hattab syndrome, 618950*
TAT	*100*	*100*	*100*	*100*	*Tyrosinemia, type II, 276600*
TAX1BP3	*100*	*100*	*100*	*100*	*No OMIM disease ID*
TBC1D20	*94,3*	*93,9*	*100*	*99,7*	*Warburg micro syndrome 4, 615663*
TBC1D23	*98,7*	*94,5*	*100*	*99,7*	*Pontocerebellar hypoplasia, type 11, 617695*
TBC1D24	*100*	*100*	*100*	*100*	*Deafness, autosomal recessive 86, 614617* *Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp, 608105* *Myoclonic epilepsy, infantile, familial, 605021* *Deafness, autosomal dominant 65, 616044* *Developmental and epileptic encephalopathy 16, 615338* *DOORS syndrome, 220500*
TBC1D2B	*99*	*97,1*	*98,5*	*97,9*	*Neurodevelopmental disorder with seizures and gingival overgrowth, 619323*

TBC1D32	*98,7*	*96,4*	*100*	*99,9*	*No OMIM disease ID*
TBC1D7	*99,7*	*99,3*	*100*	*100*	*Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000*
TBC1D8B	*98*	*92,1*	*100*	*99,7*	*Nephrotic syndrome, type 20, 301028*
TBCD	*95,5*	*93,3*	*100*	*100*	*Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193*
TBCE	*99,7*	*96,6*	*100*	*100*	*Kenny-Caffey syndrome, type 1, 244460* *Hypoparathyroidism-retardation-dysmorphism syndrome, 241410* *Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207*
TBCK	*99,4*	*95,8*	*100*	*99,9*	*Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900*
TBK1	*99,3*	*97,5*	*100*	*99,8*	*Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439*
TBL1X	*94,1*	*88,1*	*100*	*99,9*	*Hypothyroidism, congenital, nongoitrous, 8, 301033*
TBL1XR1	*93,4*	*80,7*	*100*	*100*	*Pierpont syndrome, 602342* *Mental retardation, autosomal dominant 41, 616944*
TBL1Y	*48,8*	*43,3*	*50*	*49,9*	*?Deafness, Y-linked 2, 400047*
TBP	*99,9*	*99,2*	*100*	*99,9*	*Spinocerebellar ataxia 17, 607136*
TBR1	*100*	*99,6*	*100*	*100*	*Intellectual developmental disorder with autism and speech delay, 606053*
TBX1	*87,4*	*77,6*	*93,7*	*90,2*	*Tetralogy of Fallot, 187500* *DiGeorge syndrome, 188400* *Conotruncal anomaly face syndrome, 217095* *Velocardiofacial syndrome, 192430*
TBX15	*100*	*99,7*	*100*	*100*	*Cousin syndrome, 260660*
TBX18	*99,5*	*97,1*	*100*	*100*	*Congenital anomalies of kidney and urinary tract 2, 143400*
TBX19	*100*	*100*	*100*	*100*	*Adrenocorticotropic hormone deficiency, 201400*
TBX2	*99,9*	*97,8*	*98,4*	*95,7*	*Vertebral anomalies and variable endocrine and T-cell dysfunction, 618223*
TBX20	*100*	*99,8*	*100*	*100*	*Atrial septal defect 4, 611363*
TBX21	*97,8*	*88*	*100*	*100*	*Asthma and nasal polyps, 208550*
TBX22	*98,4*	*93,8*	*100*	*99,9*	*Cleft palate with ankyloglossia, 303400* *?Abruzzo-Erickson syndrome, 302905*
TBX3	*99,4*	*97,3*	*100*	*100*	*Ulnar-mammary syndrome, 181450*
TBX4	*98,1*	*95,4*	*100*	*99,9*	*Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension, 147891* *Amelia, posterior, with pelvic and pulmonary hypoplasia syndrome, 601360*
TBX5	*100*	*100*	*100*	*100*	*Holt-Oram syndrome, 142900*
TBX6	*99,2*	*94,8*	*100*	*100*	*Spondylocostal dysostosis 5, 122600*

TBXA2R	*97,4*	*93,9*	*99,8*	*98,7*	*No OMIM disease ID*
TBXAS1	*100*	*100*	*100*	*100*	*Ghosal hematodiaphyseal syndrome, 231095*
TBXT	*99,3*	*94,9*	*100*	*100*	*Sacral agenesis with vertebral anomalies, 615709*
TCAP	*100*	*100*	*100*	*100*	*Cardiomyopathy, hypertrophic, 25, 607487* *Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954*
TCF12	*99,9*	*99,7*	*100*	*100*	*Craniosynostosis 3, 615314*
TCF20	*100*	*100*	*100*	*100*	*Developmental delay with variable intellectual impairment and behavioral abnormalities, 618430*
TCF3	*98*	*93,8*	*100*	*100*	*Agammaglobulinemia 8, autosomal dominant, 616941*
TCF4	*100*	*99,9*	*100*	*100*	*Pitt-Hopkins syndrome, 610954* *Corneal dystrophy, Fuchs endothelial, 3, 613267*
TCF7L2	*99,3*	*97*	*100*	*100*	*No OMIM disease ID*
TCHH	*100*	*99,9*	*100*	*100*	*?Uncombable hair syndrome 3, 617252*
TCIRG1	*98,5*	*93,4*	*100*	*100*	*Osteopetrosis, autosomal recessive 1, 259700*
TCN2	*100*	*100*	*100*	*100*	*Transcobalamin II deficiency, 275350*
TCOF1	*99,7*	*98,7*	*100*	*100*	*Treacher Collins syndrome 1, 154500*
TCTN1	*96,8*	*92,8*	*94,7*	*94,7*	*Joubert syndrome 13, 614173*
TCTN2	*99,9*	*99,1*	*100*	*100*	*Joubert syndrome 24, 616654* *?Meckel syndrome 8, 613885*
TCTN3	*100*	*100*	*100*	*100*	*Joubert syndrome 18, 614815* *Orofaciodigital syndrome IV, 258860*
TDGF1	*98,8*	*91,8*	*100*	*100*	*Forebrain defects,*
TDP1	*99,9*	*99,4*	*100*	*100*	*?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250*
TDP2	*99,6*	*99,5*	*100*	*99,9*	*Spinocerebellar ataxia, autosomal recessive 23, 616949*
TDRD7	*99,9*	*99,3*	*100*	*100*	*Cataract 36, 613887*
TDRD9	*99,3*	*97,7*	*100*	*100*	*?Spermatogenic failure 30, 618110*
TDRKH	*94,7*	*94,6*	*100*	*100*	*No OMIM disease ID*
TEAD1	*100*	*99,7*	*100*	*100*	*Sveinsson chorioretinal atrophy, 108985*
TECPR2	*100*	*100*	*100*	*100*	*Spastic paraplegia 49, autosomal recessive, 615031*
TECR	*100*	*98,5*	*100*	*100*	*Mental retardation, autosomal recessive 14, 614020*
TECRL	*97,5*	*91,7*	*100*	*99,4*	*Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021*

TECTA	*100*	*99,8*	*100*	*100*	*Deafness, autosomal dominant 8/12, 601543* *Deafness, autosomal recessive 21, 603629*
TEK	*100*	*99,9*	*100*	*100*	*Venous malformations, multiple cutaneous and mucosal, 600195* *Glaucoma 3, primary congenital, E, 617272*
TELO2	*99,9*	*98*	*100*	*100*	*You-Hoover-Fong syndrome, 616954*
TENM3	*100*	*99,7*	*100*	*100*	*Microphthalmia, syndromic 15, 615145* *?Microphthalmia, isolated, with coloboma 9, 615145*
TENM4	*100*	*99,3*	*100*	*100*	*Essential tremor, hereditary, 5, 616736*
TENT5A	*100*	*99,5*	*100*	*100*	*Osteogenesis imperfecta, type XVIII, 617952*
TERB1	*99,5*	*98,6*	*100*	*100*	*No OMIM disease ID*
TERB2	*97,4*	*90,9*	*100*	*99,7*	*No OMIM disease ID*
TERC	*NC*	*NC*	*NC*	*NC*	*Dyskeratosis congenita, autosomal dominant 1, 127550*
TERF1	*67,1*	*44,2*	*100*	*99,9*	*No OMIM disease ID*
TERF2	*99,9*	*98*	*100*	*99,8*	*No OMIM disease ID*
TERF2IP	*100*	*99,9*	*83,7*	*83,7*	*No OMIM disease ID*
TERT	*97*	*94,8*	*100*	*100*	*No OMIM disease ID*
TET2	*100*	*100*	*100*	*100*	*Myelodysplastic syndrome, somatic, 614286* *Immunodeficiency 75, 619126*
TET3	*94,4*	*94,4*	*100*	*100*	*Beck-Fahrner syndrome, 618798*
TEX11	*92,7*	*84,5*	*97,1*	*96,4*	*Spermatogenic failure, X-linked, 2, 309120*
TEX14	*99,9*	*98,6*	*100*	*100*	*Spermatogenic failure 23, 617707*
TEX15	*99,6*	*99,2*	*100*	*100*	*Spermatogenic failure 25, 617960*
TF	*100*	*99,9*	*100*	*100*	*Atransferrinemia, 209300*
TFAM	*98*	*78,5*	*100*	*100*	*?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156*
TFAP2A	*98,1*	*92,1*	*100*	*100*	*Branchiooculofacial syndrome, 113620*
TFAP2B	*98,8*	*96,4*	*100*	*100*	*Patent ductus arteriosus 2, 617035* *Char syndrome, 169100*
TFB2M	*100*	*98,5*	*100*	*99,9*	*No OMIM disease ID*
TFE3	*98,1*	*91*	*100*	*100*	*Renal cell carcinoma, papillary, 1, 300854*
TFG	*97*	*96*	*100*	*99,9*	*?Spastic paraplegia 57, autosomal recessive, 615658* *Hereditary motor and sensory neuropathy, Okinawa type, 604484*
TFR2	*99,3*	*96,9*	*100*	*100*	*Hemochromatosis, type 3, 604250*

TFRC	*99,9*	*99,6*	*100*	*100*	*Immunodeficiency 46, 616740*
TG	*99,9*	*98,5*	*100*	*100*	*Thyroid dyshormonogenesis 3, 274700*
TGDS	*99,4*	*95,9*	*100*	*99,9*	*Catel-Manzke syndrome, 616145*
TGFB1	*100*	*98,8*	*100*	*100*	*Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213* *Camurati-Engelmann disease, 131300*
TGFB2	*100*	*100*	*100*	*100*	*Loeys-Dietz syndrome 4, 614816*
TGFB3	*100*	*100*	*100*	*100*	*Arrhythmogenic right ventricular dysplasia 1, 107970* *Loeys-Dietz syndrome 5, 615582*
TGFBI	*99,9*	*96,3*	*100*	*100*	*Corneal dystrophy, Avellino type, 607541* *Corneal dystrophy, Reis-Bucklers type, 608470* *Corneal dystrophy, Thiel-Behnke type, 602082* *Corneal dystrophy, Groenouw type I, 121900* *Corneal dystrophy, epithelial basement membrane, 121820* *Corneal dystrophy, lattice type I, 122200* *Corneal dystrophy, lattice type IIIA, 608471*
TGFBR1	*93,6*	*93,6*	*98,8*	*97,6*	*Loeys-Dietz syndrome 1, 609192*
TGFBR2	*100*	*99,9*	*100*	*100*	*Loeys-Dietz syndrome 2, 610168* *Colorectal cancer, hereditary nonpolyposis, type 6, 614331* *Esophageal cancer, somatic, 133239*
TGIF1	*100*	*100*	*100*	*100*	*Holoprosencephaly 4, 142946*
TGM1	*100*	*99,5*	*100*	*100*	*Ichthyosis, congenital, autosomal recessive 1, 242300*
TGM3	*100*	*98,7*	*100*	*100*	*?Uncombable hair syndrome 2, 617251*
TGM5	*100*	*99,4*	*100*	*100*	*Peeling skin syndrome 2, 609796*
TGM6	*99,8*	*98,1*	*100*	*100*	*Spinocerebellar ataxia 35, 613908*
TH	*99,8*	*98*	*100*	*100*	*Segawa syndrome, recessive, 605407*
THAP1	*100*	*100*	*100*	*100*	*Dystonia 6, torsion, 602629*
THBD	*100*	*99,9*	*100*	*100*	*Thrombophilia due to thrombomodulin defect, 614486*
THBS4	*100*	*99,4*	*100*	*100*	*No OMIM disease ID*
THG1L	*100*	*100*	*100*	*100*	*Spinocerebellar ataxia, autosomal recessive 28, 618800*
THOC1	*99,5*	*97,6*	*100*	*100*	*No OMIM disease ID*
THOC2	*98,1*	*91,2*	*100*	*99,5*	*Intellectual developmental disorder, X-linked 12, 300957*
THOC6	*100*	*100*	*100*	*100*	*Beaulieu-Boycott-Innes syndrome, 613680*

THPO	*81,4*	*78,7*	*100*	*100*	*Thrombocythemia 1, 187950*
THRA	*100*	*99,8*	*100*	*100*	*Hypothyroidism, congenital, nongoitrous, 6, 614450*
THRB	*100*	*99,6*	*100*	*100*	*Thyroid hormone resistance, autosomal recessive, 274300* *Thyroid hormone resistance, 188570* *Thyroid hormone resistance, selective pituitary, 145650*
THSD1	*100*	*100*	*100*	*100*	*?Aneurysm, intracranial berry, 12, 618734*
TIA1	*99,4*	*95,3*	*100*	*100*	*Welander distal myopathy, 604454* *Amyotrophic lateral sclerosis 26 with or without frontotemporal dementia, 619133*
TICAM1	*100*	*98,8*	*100*	*100*	*No OMIM disease ID*
TIE1	*100*	*99,3*	*100*	*100*	*Lymphatic malformation 11, 619401*
TIMM22	*100*	*99,2*	*100*	*100*	*?Combined oxidative phosphorylation deficiency 43, 618851*
TIMM44	*100*	*99,9*	*100*	*100*	*No OMIM disease ID*
TIMM50	*98,4*	*95*	*100*	*100*	*3-methylglutaconic aciduria, type IX, 617698*
TIMM8A	*96,2*	*83,1*	*100*	*100*	*Mohr-Tranebjaerg syndrome, 304700*
TIMMDC1	*99,9*	*99,8*	*100*	*100*	*Mitochondrial complex I deficiency, nuclear type 31, 618251*
TIMP3	*100*	*100*	*100*	*100*	*Sorsby fundus dystrophy, 136900*
TINF2	*100*	*100*	*100*	*100*	*Dyskeratosis congenita, autosomal dominant 3, 613990* *Revesz syndrome, 268130*
TIRAP	*100*	*100*	*100*	*100*	*No OMIM disease ID*
TJP1	*100*	*99,7*	*100*	*100*	*No OMIM disease ID*
TJP2	*92,8*	*92,3*	*98,8*	*98,8*	*Hypercholanemia, familial 1, 607748* *Cholestasis, progressive familial intrahepatic 4, 615878*
TK2	*99*	*96*	*100*	*100*	*Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560* *?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069*
TKFC	*100*	*99,8*	*100*	*100*	*Triokinase and FMN cyclase deficiency syndrome, 618805*
TKT	*98,6*	*96,8*	*98,7*	*98,7*	*Short stature, developmental delay, and congenital heart defects, 617044*
TLE6	*99,9*	*97,8*	*100*	*100*	*Preimplantation embryonic lethality, 616814*
TLK2	*98,5*	*93,2*	*100*	*100*	*Mental retardation, autosomal dominant 57, 618050*
TLL1	*99,9*	*99,7*	*100*	*100*	*Atrial septal defect 6, 613087*
TLN1	*99,9*	*98,3*	*100*	*100*	*No OMIM disease ID*
TLR3	*99,8*	*99*	*100*	*100*	*No OMIM disease ID*

TLR4	*100*	*99*	*100*	*100*	*No OMIM disease ID*
TLR5	*100*	*100*	*100*	*100*	*No OMIM disease ID*
TLR7	*100*	*99,7*	*100*	*100*	*Immunodeficiency 74, COVID19-related, X-linked, 301051*
TLR8	*99,9*	*99,8*	*100*	*100*	*No OMIM disease ID*
TMC1	*99,8*	*96,6*	*100*	*99,9*	*Deafness, autosomal dominant 36, 606705* *Deafness, autosomal recessive 7, 600974*
TMC6	*100*	*99,6*	*100*	*100*	*Epidermodysplasia verruciformis, 226400*
TMC8	*99,9*	*98,9*	*100*	*100*	*Epidermodysplasia verruciformis 2, 618231*
TMCO1	*87,8*	*87*	*88*	*87,9*	*Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980*
TMCO3	*99,9*	*98,6*	*100*	*100*	*No OMIM disease ID*
TMEM106B	*99,3*	*98,3*	*100*	*99,9*	*Leukodystrophy, hypomyelinating, 16, 617964*
TMEM107	*100*	*100*	*100*	*100*	*Orofaciodigital syndrome XVI, 617563* *Meckel syndrome 13, 617562* *?Joubert syndrome 29, 617562*
TMEM126A	*95,4*	*80*	*100*	*100*	*Optic atrophy 7, 612989*
TMEM126B	*99,6*	*97,2*	*100*	*100*	*Mitochondrial complex I deficiency, nuclear type 29, 618250*
TMEM127	*99,9*	*97,7*	*100*	*100*	*No OMIM disease ID*
TMEM132E	*97,5*	*94,8*	*100*	*100*	*Deafness, autosomal recessive 99, 618481*
TMEM138	*99,8*	*93,1*	*100*	*100*	*Joubert syndrome 16, 614465*
TMEM14C	*100*	*99,7*	*100*	*100*	*No OMIM disease ID*
TMEM165	*100*	*100*	*100*	*100*	*Congenital disorder of glycosylation, type IIk, 614727*
TMEM186	*100*	*100*	*100*	*100*	*No OMIM disease ID*
TMEM199	*100*	*100*	*100*	*100*	*Congenital disorder of glycosylation, type IIp, 616829*
TMEM216	*98,5*	*92,8*	*100*	*100*	*Joubert syndrome 2, 608091* *Meckel syndrome 2, 603194*
TMEM218	*100*	*99,8*	*100*	*100*	*No OMIM disease ID*
TMEM222	*100*	*99,5*	*100*	*100*	*Neurodevelopmental disorder with motor and speech delay and behavioral abnormalities, 619470*
TMEM231	*100*	*99,3*	*100*	*100*	*Joubert syndrome 20, 614970* *Meckel syndrome 11, 615397*
TMEM237	*99,8*	*99,3*	*100*	*100*	*Joubert syndrome 14, 614424*
TMEM240	*100*	*100*	*100*	*100*	*Spinocerebellar ataxia 21, 607454*

TMEM251	*100*	*98,8*	*100*	*100*	*Dysostosis multiplex, Ain-Naz type, 619345*
TMEM260	*98,7*	*95,4*	*100*	*100*	*Structural heart defects and renal anomalies syndrome, 617478*
TMEM38B	*99,8*	*99,8*	*100*	*100*	*Osteogenesis imperfecta, type XIV, 615066*
TMEM43	*99,9*	*98,4*	*100*	*100*	*Arrhythmogenic right ventricular dysplasia 5, 604400* *Emery-Dreifuss muscular dystrophy 7, AD, 614302*
TMEM63A	*100*	*99,2*	*100*	*100*	*Leukodystrophy, hypomyelinating, 19, transient infantile, 618688*
TMEM65	*89,3*	*83,2*	*95,1*	*87*	*No OMIM disease ID*
TMEM67	*98,6*	*93,5*	*100*	*99,6*	*Nephronophthisis 11, 613550* *Joubert syndrome 6, 610688* *Meckel syndrome 3, 607361* *?RHYNS syndrome, 602152* *COACH syndrome 1, 216360*
TMEM70	*98,4*	*94,6*	*100*	*100*	*Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052*
TMEM94	*100*	*100*	*100*	*100*	*Intellectual developmental disorder with cardiac defects and dysmorphic facies, 618316*
TMEM98	*99,2*	*95,8*	*100*	*100*	*Nanophthalmos 4, 615972*
TMIE	*99,9*	*97,5*	*100*	*100*	*Deafness, autosomal recessive 6, 600971*
TMLHE	*98,6*	*94,1*	*100*	*99,7*	*No OMIM disease ID*
TMPO	*98*	*93,8*	*100*	*100*	*No OMIM disease ID*
TMPRSS15	*98,2*	*95,7*	*100*	*99,9*	*Enterokinase deficiency, 226200*
TMPRSS3	*100*	*99,3*	*100*	*100*	*Deafness, autosomal recessive 8/10, 601072*
TMPRSS6	*100*	*99,3*	*100*	*100*	*Iron-refractory iron deficiency anemia, 206200*
TMTC2	*97,5*	*97,5*	*97,5*	*97,5*	*No OMIM disease ID*
TMTC3	*98,7*	*95,8*	*100*	*100*	*Lissencephaly 8, 617255*
TMX2	*100*	*99,2*	*100*	*100*	*Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730*
TNC	*100*	*99,8*	*100*	*100*	*Deafness, autosomal dominant 56, 615629*
TNFAIP3	*100*	*99,9*	*100*	*100*	*Autoinflammatory syndrome, familial, Behcet-like, 616744*
TNFRSF10B	*100*	*100*	*100*	*100*	*Squamous cell carcinoma, head and neck, 275355*
TNFRSF11A	*94,9*	*93,8*	*99,1*	*97,7*	*Osteopetrosis, autosomal recessive 7, 612301* *Osteolysis, familial expansile, 174810*
TNFRSF11B	*100*	*100*	*100*	*100*	*Paget disease of bone 5, juvenile-onset, 239000*

TNFRSF13B	*100*	*99,9*	*100*	*100*	*Immunodeficiency, common variable, 2, 240500* *Immunoglobulin A deficiency 2, 609529*
TNFRSF13C	*85*	*75,6*	*100*	*100*	*Immunodeficiency, common variable, 4, 613494*
TNFRSF1A	*89,3*	*86,5*	*92,8*	*92,8*	*Periodic fever, familial, 142680*
TNFRSF4	*97,7*	*89*	*100*	*100*	*?Immunodeficiency 16, 615593*
TNFRSF9	*100*	*100*	*100*	*100*	*No OMIM disease ID*
TNFSF11	*100*	*100*	*100*	*100*	*Osteopetrosis, autosomal recessive 2, 259710*
TNFSF12	*99,5*	*95,3*	*100*	*100*	*No OMIM disease ID*
TNFSF13	*99*	*94,2*	*100*	*100*	*No OMIM disease ID*
TNIK	*99,8*	*98,5*	*100*	*100*	*Mental retardation, autosomal recessive 54, 617028*
TNNC1	*100*	*100*	*100*	*100*	*Cardiomyopathy, dilated, 1Z, 611879* *Cardiomyopathy, hypertrophic, 13, 613243*
TNNI2	*100*	*99,9*	*100*	*100*	*Arthrogryposis, distal, type 2B1, 601680*
TNNI3	*99,6*	*95,5*	*100*	*100*	*?Cardiomyopathy, dilated, 2A, 611880* *Cardiomyopathy, hypertrophic, 7, 613690* *Cardiomyopathy, familial restrictive, 1, 115210* *Cardiomyopathy, dilated, 1FF, 613286*
TNNI3K	*99,8*	*99,6*	*100*	*100*	*Cardiac conduction disease with or without dilated cardiomyopathy, 616117*
TNNT1	*99,6*	*97,1*	*100*	*100*	*Nemaline myopathy 5, Amish type, 605355*
TNNT2	*94,6*	*90,7*	*100*	*99,7*	*Cardiomyopathy, dilated, 1D, 601494* *Cardiomyopathy, hypertrophic, 2, 115195* *Cardiomyopathy, familial restrictive, 3, 612422* *Left ventricular noncompaction 6, 601494*
TNNT3	*100*	*99,6*	*100*	*100*	*Arthrogryposis, distal, type 2B2, 618435*
TNPO3	*100*	*99,8*	*100*	*100*	*Muscular dystrophy, limb-girdle, autosomal dominant 2, 608423*
TNR	*100*	*99,6*	*100*	*100*	*No OMIM disease ID*
TNRC6A	*99,7*	*98,8*	*100*	*100*	*?Epilepsy, familial adult myoclonic, 6, 618074*
TNRC6B	*99,9*	*99,2*	*100*	*100*	*Global developmental delay with speech and behavioral abnormalities, 619243*
TNS2	*100*	*100*	*100*	*100*	*No OMIM disease ID*
TNXB	*98,7*	*93,9*	*100*	*100*	*Ehlers-Danlos syndrome, classic-like, 1, 606408* *Vesicoureteral reflux 8, 615963*
TOE1	*100*	*100*	*100*	*100*	*Pontocerebellar hypoplasia, type 7, 614969*

TOGARAM1	*99,6*	*97,5*	*100*	*99,9*	*Joubert syndrome 37, 619185*
TOMM70	*99,9*	*99,3*	*100*	*100*	*No OMIM disease ID*
TONSL	*99,9*	*98,4*	*100*	*100*	*Spondyloepimetaphyseal dysplasia, sponastrime type, 271510*
TOP1	*99,9*	*97,5*	*100*	*100*	*DNA topoisomerase I, camptothecin-resistant,*
TOP2A	*99,4*	*98*	*100*	*100*	*DNA topoisomerase II, resistance to inhibition of, by amsacrine,*
TOP2B	*98,9*	*95,7*	*100*	*99,9*	*No OMIM disease ID*
TOP3A	*99,6*	*96,5*	*100*	*100*	*?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098* *Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097*
TOPORS	*100*	*100*	*100*	*100*	*Retinitis pigmentosa 31, 609923*
TOR1A	*91,3*	*91,3*	*91,7*	*91,3*	*Arthrogryposis multiplex congenita 5, 618947* *Dystonia-1, torsion, 128100*
TOR1AIP1	*99,2*	*96,1*	*100*	*100*	*?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072*
TP53	*99*	*95,2*	*91,7*	*91,7*	*Hepatocellular carcinoma, somatic, 114550* *Breast cancer, somatic, 114480* *Li-Fraumeni syndrome, 151623* *Pancreatic cancer, somatic, 260350* *Nasopharyngeal carcinoma, somatic, 607107* *Bone marrow failure syndrome 5, 618165*
TP53RK	*95,5*	*84,9*	*100*	*100*	*Galloway-Mowat syndrome 4, 617730*
TP63	*100*	*100*	*100*	*100*	*Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292* *Hay-Wells syndrome, 106260* *Split-hand/foot malformation 4, 605289* *Orofacial cleft 8, 618149* *Rapp-Hodgkin syndrome, 129400* *ADULT syndrome, 103285* *Limb-mammary syndrome, 603543*
TP73	*100*	*100*	*100*	*100*	*Ciliary dyskinesia, primary, 47, and lissencephaly, 619466*
TPCN2	*95,5*	*92,8*	*100*	*100*	*No OMIM disease ID*
TPI1	*99,8*	*98*	*100*	*100*	*Hemolytic anemia due to triosephosphate isomerase deficiency, 615512*
TPK1	*99,5*	*97,2*	*100*	*100*	*Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458*
TPM1	*100*	*99,3*	*100*	*99,8*	*Left ventricular noncompaction 9, 611878* *Cardiomyopathy, hypertrophic, 3, 115196* *Cardiomyopathy, dilated, 1Y, 611878*

TPM2	*100*	*99,8*	*100*	*99,9*	*Arthrogryposis, distal, type 2B4, 108120* *Arthrogryposis, distal, type 1A, 108120* *Nemaline myopathy 4, autosomal dominant, 609285* *CAP myopathy 2, 609285*
TPM3	*87,7*	*84,3*	*100*	*100*	*CAP myopathy 1, 609284* *Myopathy, congenital, with fiber-type disproportion, 255310* *Nemaline myopathy 1, autosomal dominant or recessive, 609284*
TPM4	*81,5*	*64,6*	*100*	*100*	*No OMIM disease ID*
TPMT	*98,4*	*82*	*100*	*100*	*No OMIM disease ID*
TPO	*100*	*99,2*	*100*	*100*	*Thyroid dyshormonogenesis 2A, 274500*
TPP1	*100*	*100*	*100*	*100*	*Ceroid lipofuscinosis, neuronal, 2, 204500* *Spinocerebellar ataxia, autosomal recessive 7, 609270*
TPP2	*99,2*	*95,1*	*100*	*100*	*Immunodeficiency 78 with autoimmunity and developmental delay, 619220*
TPRKB	*80,2*	*75,2*	*81,9*	*81,7*	*Galloway-Mowat syndrome 5, 617731*
TPRN	*89,7*	*83,4*	*94,5*	*88,3*	*Deafness, autosomal recessive 79, 613307*
TRAC	*100*	*100*	*100*	*100*	*Immunodeficiency 7, TCR-alpha/beta deficient, 615387*
TRAF3	*100*	*99,2*	*100*	*100*	*No OMIM disease ID*
TRAF3IP1	*98,7*	*95,4*	*100*	*100*	*Senior-Loken syndrome 9, 616629*
TRAF3IP2	*99,7*	*97,8*	*100*	*100*	*?Candidiasis, familial, 8, 615527*
TRAF6	*96,2*	*86,1*	*100*	*100*	*No OMIM disease ID*
TRAF7	*100*	*100*	*100*	*100*	*Cardiac, facial, and digital anomalies with developmental delay, 618164*
TRAIP	*100*	*100*	*100*	*99,9*	*Seckel syndrome 9, 616777*
TRAK1	*93,3*	*93,1*	*100*	*99,9*	*Developmental and epileptic encephalopathy 68, 618201*
TRAPPC11	*99,7*	*98,7*	*100*	*99,9*	*Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356*
TRAPPC12	*100*	*99,9*	*100*	*100*	*Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669*
MAP11	*100*	*98,8*	*100*	*100*	*?Microcephaly 25, primary, autosomal recessive, 618351*
TRAPPC2	*91,6*	*73,2*	*100*	*99,9*	*Spondyloepiphyseal dysplasia tarda, 313400*
TRAPPC2L	*100*	*100*	*100*	*100*	*Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331*
TRAPPC4	*100*	*100*	*100*	*100*	*Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy, 618741*
TRAPPC6B	*99,2*	*96,4*	*100*	*100*	*Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862*
TRAPPC9	*100*	*99,7*	*100*	*100*	*Mental retardation, autosomal recessive 13, 613192*

TRDN	*97,7*	*89,1*	*100*	*99,5*	*Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441*
TREH	*97,7*	*93*	*100*	*100*	*Trehalase deficiency, 612119*
TREM2	*100*	*99,3*	*100*	*100*	*Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193*
TREX1	*100*	*100*	*100*	*100*	*Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315* *Aicardi-Goutieres syndrome 1, dominant and recessive, 225750* *Chilblain lupus, 610448*
TRH	*99,7*	*97,5*	*100*	*100*	*No OMIM disease ID*
TRHR	*99,9*	*98,5*	*100*	*100*	*Hypothyroidism, congenital, nongoitrous, 7, 618573*
TRIM2	*93,8*	*93,5*	*93,9*	*93,9*	*Charcot-Marie-Tooth disease, type 2R, 615490*
TRIM22	*100*	*99,9*	*100*	*100*	*No OMIM disease ID*
TRIM28	*97,7*	*96,3*	*99,8*	*99,2*	*No OMIM disease ID*
TRIM32	*100*	*99,9*	*100*	*100*	*?Bardet-Biedl syndrome 11, 615988* *Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110*
TRIM36	*99,8*	*98,7*	*100*	*100*	*?Anencephaly 1, 206500*
TRIM37	*98,3*	*97,1*	*98,7*	*98,6*	*Mulibrey nanism, 253250*
TRIM44	*100*	*99*	*100*	*100*	*?Aniridia 3, 617142*
TRIM63	*100*	*99,9*	*100*	*100*	*No OMIM disease ID*
TRIM71	*100*	*100*	*100*	*99,6*	*Hydrocephalus, congenital communicating, 1, 618667*
TRIM8	*98,9*	*96,2*	*100*	*100*	*Focal segmental glomerulosclerosis and neurodevelopmental syndrome, 619428*
TRIO	*99,3*	*97,5*	*99,2*	*98,2*	*Intellectual developmental disorder, autosomal dominant 44, with microcephaly, 617061* *Intellectual developmental disorder, autosomal dominant 63, with macrocephaly, 618825*
TRIOBP	*98,5*	*96,6*	*99,9*	*99,2*	*Deafness, autosomal recessive 28, 609823*
TRIP11	*97,2*	*92,6*	*100*	*99,9*	*Odontochondrodysplasia 1, 184260* *Achondrogenesis, type IA, 200600*
TRIP12	*99,7*	*98,6*	*100*	*100*	*Mental retardation, autosomal dominant 49, 617752*
TRIP13	*100*	*99,9*	*100*	*100*	*Oocyte maturation defect 9, 619011* *Mosaic variegated aneuploidy syndrome 3, 617598*
TRIP4	*99,8*	*99*	*100*	*100*	*?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066* *Spinal muscular atrophy with congenital bone fractures 1, 616866*
TRIT1	*100*	*100*	*100*	*100*	*Combined oxidative phosphorylation deficiency 35, 617873*
TRMT1	*99,5*	*96,2*	*100*	*100*	*Mental retardation, autosomal recessive 68, 618302*
TRMT10A	*99,7*	*99,5*	*100*	*100*	*Microcephaly, short stature, and impaired glucose metabolism 1, 616033*

TRMT10C	*100*	*99,9*	*100*	*99,9*	*Combined oxidative phosphorylation deficiency 30, 616974*
TRMT5	*99,8*	*99,1*	*100*	*100*	*Combined oxidative phosphorylation deficiency 26, 616539*
TRMU	*99,9*	*99,6*	*100*	*99,9*	*Liver failure, transient infantile, 613070*
TRNT1	*99,7*	*97,4*	*100*	*99,9*	*Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084* *Retinitis pigmentosa and erythrocytic microcytosis, 616959*
TRPA1	*96,4*	*89,9*	*100*	*99,9*	*?Episodic pain syndrome, familial, 1, 615040*
TRPC3	*99,8*	*97,9*	*100*	*100*	*?Spinocerebellar ataxia 41, 616410*
TRPC6	*97,1*	*94,5*	*100*	*100*	*Glomerulosclerosis, focal segmental, 2, 603965*
TRPM1	*100*	*99,4*	*100*	*100*	*Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216*
TRPM3	*99,9*	*99,4*	*100*	*100*	*No OMIM disease ID*
TRPM4	*100*	*99,8*	*100*	*100*	*Progressive familial heart block, type IB, 604559* *Erythrokeratodermia veriabilis et progressiva 6, 618531*
TRPM6	*99,9*	*99,1*	*100*	*100*	*Hypomagnesemia 1, intestinal, 602014*
TRPM8	*99,8*	*98,5*	*100*	*100*	*No OMIM disease ID*
TRPS1	*100*	*99,9*	*100*	*100*	*Trichorhinophalangeal syndrome, type III, 190351* *Trichorhinophalangeal syndrome, type I, 190350*
TRPV1	*99,9*	*99*	*100*	*100*	*No OMIM disease ID*
TRPV3	*99,8*	*98,6*	*97,1*	*97,1*	*?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400* *Olmsted syndrome 1, 614594*
TRPV4	*100*	*99,9*	*100*	*100*	*Spondylometaphyseal dysplasia, Kozlowski type, 184252* *Digital arthropathy-brachydactyly, familial, 606835* *SED, Maroteaux type, 184095* *Metatropic dysplasia, 156530* *Scapuloperoneal spinal muscular atrophy, 181405* *Hereditary motor and sensory neuropathy, type IIc, 606071* *?Avascular necrosis of femoral head, primary, 2, 617383* *Neuronopathy, distal hereditary motor, type VIII, 600175* *Parastremmatic dwarfism, 168400* *Brachyolmia type 3, 113500*
TRPV6	*100*	*99,9*	*99,9*	*99,2*	*Hyperparathyroidism, transient neonatal, 618188*
TRRAP	*99,9*	*99,1*	*100*	*100*	*?Deafness, autosomal dominant 75, 618778* *Developmental delay with or without dysmorphic facies and autism, 618454*

TSC1	*99,5*	*98,2*	*100*	*100*	*Focal cortical dysplasia, type II, somatic, 607341* *Tuberous sclerosis-1, 191100* *Lymphangioleiomyomatosis, 606690*
TSC2	*100*	*99,8*	*100*	*100*	*Lymphangioleiomyomatosis, somatic, 606690* *?Focal cortical dysplasia, type II, somatic, 607341* *Tuberous sclerosis-2, 613254*
TSEN15	*78,9*	*77*	*100*	*100*	*Pontocerebellar hypoplasia, type 2F, 617026*
TSEN2	*99,9*	*99,2*	*100*	*100*	*Pontocerebellar hypoplasia type 2B, 612389*
TSEN34	*92,1*	*85,6*	*100*	*100*	*?Pontocerebellar hypoplasia type 2C, 612390*
TSEN54	*96,7*	*94,8*	*99,9*	*99,2*	*Pontocerebellar hypoplasia type 2A, 277470* *Pontocerebellar hypoplasia type 4, 225753* *?Pontocerebellar hypoplasia type 5, 610204*
TSFM	*100*	*99,3*	*94,9*	*94,9*	*Combined oxidative phosphorylation deficiency 3, 610505*
TSGA10	*89,3*	*88,5*	*100*	*99,7*	*?Spermatogenic failure 26, 617961*
TSHB	*100*	*100*	*100*	*100*	*Hypothyroidism, congenital, nongoitrous 4, 275100*
TSHR	*95,9*	*95,1*	*100*	*100*	*Hyperthyroidism, familial gestational, 603373* *Hyperthyroidism, nonautoimmune, 609152* *Hypothyroidism, congenital, nongoitrous, 1, 275200* *Thyroid adenoma, hyperfunctioning, somatic,* *Thyroid carcinoma with thyrotoxicosis,*
TSHZ1	*98,8*	*98,8*	*100*	*100*	*Aural atresia, congenital, 607842*
TSPAN12	*100*	*99,9*	*100*	*100*	*Exudative vitreoretinopathy 5, 613310*
TSPAN7	*100*	*99,8*	*100*	*100*	*Intellectual developmental disorder, X-linked 58, 300210*
TSPEAR	*100*	*99,7*	*100*	*100*	*?Deafness, autosomal recessive 98, 614861* *Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180*
TSPYL1	*100*	*100*	*100*	*100*	*Sudden infant death with dysgenesis of the testes syndrome, 608800*
TSR2	*99,9*	*98,3*	*100*	*100*	*?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946*
TTBK2	*99,9*	*98,3*	*100*	*100*	*Spinocerebellar ataxia 11, 604432*
TTC12	*99,9*	*98,9*	*100*	*100*	*Ciliary dyskinesia, primary, 45, 618801*
TTC19	*83,8*	*74,1*	*100*	*99,8*	*Mitochondrial complex III deficiency, nuclear type 2, 615157*
TTC21A	*100*	*99,9*	*100*	*100*	*Spermatogenic failure 37, 618429*
TTC21B	*99,7*	*99,1*	*100*	*99,9*	*Short-rib thoracic dysplasia 4 with or without polydactyly, 613819* *Nephronophthisis 12, 613820*

TTC26	*99,8*	*97,9*	*100*	*100*	*No OMIM disease ID*
TTC29	*98,9*	*94,6*	*100*	*99,9*	*Spermatogenic failure 42, 618745*
TTC37	*99,7*	*98,8*	*100*	*100*	*Trichohepatoenteric syndrome 1, 222470*
TTC5	*99,9*	*99*	*100*	*100*	*Neurodevelopmental disorder with cerebral atrophy and variable facial dysmorphism, 619244*
TTC7A	*99,6*	*97,1*	*100*	*100*	*Gastrointestinal defects and immunodeficiency syndrome, 243150*
TTC8	*99,5*	*98*	*100*	*100*	*Bardet-Biedl syndrome 8, 615985* *?Retinitis pigmentosa 51, 613464*
TTI2	*100*	*99,9*	*100*	*100*	*Mental retardation, autosomal recessive 39, 615541*
TTLL5	*99,9*	*98,1*	*100*	*100*	*Cone-rod dystrophy 19, 615860*
TTN	*98,5*	*98*	*100*	*100*	*Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807* *Cardiomyopathy, familial hypertrophic, 9, 613765* *Tibial muscular dystrophy, tardive, 600334* *Salih myopathy, 611705* *Cardiomyopathy, dilated, 1G, 604145* *Myopathy, myofibrillar, 9, with early respiratory failure, 603689*
TTPA	*96,2*	*89,6*	*100*	*100*	*Ataxia with isolated vitamin E deficiency, 277460*
TTR	*94,6*	*94,6*	*94,6*	*94,6*	*Amyloidosis, hereditary, transthyretin-related, 105210* *Carpal tunnel syndrome, familial, 115430*
TUB	*99,8*	*97,1*	*100*	*100*	*?Retinal dystrophy and obesity, 616188*
TUBA1A	*99,5*	*93,2*	*100*	*100*	*Lissencephaly 3, 611603*
TUBA3D	*99,9*	*96,5*	*100*	*100*	*Keratoconus 9, 617928*
TUBA4A	*100*	*100*	*100*	*100*	*Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia, 616208*
TUBA8	*99,9*	*99,2*	*100*	*100*	*No OMIM disease ID*
TUBB	*96,8*	*93,7*	*99,9*	*99,8*	*Symmetric circumferential skin creases, congenital, 1, 156610* *Cortical dysplasia, complex, with other brain malformations 6, 615771*
TUBB1	*100*	*100*	*100*	*100*	*Macrothrombocytopenia, autosomal dominant, TUBB1-related, 613112*
TUBB2A	*96,9*	*95,7*	*100*	*100*	*Cortical dysplasia, complex, with other brain malformations 5, 615763*
TUBB2B	*100*	*99,7*	*100*	*100*	*Cortical dysplasia, complex, with other brain malformations 7, 610031*
TUBB3	*98,5*	*96,8*	*100*	*100*	*Fibrosis of extraocular muscles, congenital, 3A, 600638* *Cortical dysplasia, complex, with other brain malformations 1, 614039*
TUBB4A	*96*	*95,6*	*99*	*96,9*	*Dystonia 4, torsion, autosomal dominant, 128101* *Leukodystrophy, hypomyelinating, 6, 612438*
TUBB4B	*100*	*98,7*	*100*	*100*	*Leber congenital amaurosis with early-onset deafness, 617879*

TUBB6	*90,6*	*90,2*	*100*	*99,9*	*?Facial palsy, congenitla, with ptosis and velopharyngeal dysfunction, 617732*
TUBB8	*83,1*	*51,3*	*100*	*100*	*Oocyte maturation defect 2, 616780*
TUBG1	*100*	*100*	*100*	*100*	*Cortical dysplasia, complex, with other brain malformations 4, 615412*
TUBGCP2	*99,1*	*95,5*	*97*	*97*	*Pachygyria, microcephaly, developmental delay, and dysmorphic facies, with or without seizures, 618737*
TUBGCP4	*98,9*	*94,7*	*100*	*99,8*	*Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335*
TUBGCP6	*100*	*99,4*	*100*	*100*	*Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270*
TUFM	*99,9*	*97,6*	*100*	*100*	*Combined oxidative phosphorylation deficiency 4, 610678*
TULP1	*99,8*	*98,2*	*100*	*100*	*Leber congenital amaurosis 15, 613843* *Retinitis pigmentosa 14, 600132*
TUSC3	*100*	*99,7*	*100*	*100*	*Mental retardation, autosomal recessive 7, 611093*
TWIST1	*100*	*99,4*	*96,7*	*90,6*	*Craniosynostosis 1, 123100* *Robinow-Sorauf syndrome, 180750* *Sweeney-Cox syndrome, 617746* *Saethre-Chotzen syndrome with or without eyelid anomalies, 101400*
TWIST2	*100*	*99,9*	*100*	*100*	*Ablepharon-macrostomia syndrome, 200110* *Barber-Say syndrome, 209885* *Focal facial dermal dysplasia 3, Setleis type, 227260*
TWNK	*100*	*99,9*	*100*	*100*	*Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245* *Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286* *Perrault syndrome 5, 616138*
TXN2	*100*	*98,5*	*100*	*100*	*?Combined oxidative phosphorylation deficiency 29, 616811*
TXNL4A	*99,3*	*99,1*	*100*	*100*	*Burn-McKeown syndrome, 608572*
TXNRD2	*96,8*	*95,9*	*100*	*100*	*?Glucocorticoid deficiency 5, 617825*
TYK2	*100*	*99,3*	*100*	*100*	*Immunodeficiency 35, 611521*
TYMP	*100*	*99,4*	*100*	*100*	*Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041*
TYMS	*100*	*99,7*	*100*	*100*	*No OMIM disease ID*
TYR	*100*	*100*	*100*	*100*	*Albinism, oculocutaneous, type IB, 606952* *Waardenburg syndrome/albinism, digenic, 103470* *Albinism, oculocutaneous, type IA, 203100*
TYROBP	*100*	*100*	*100*	*100*	*Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770*
TYRP1	*100*	*99,9*	*100*	*100*	*Albinism, oculocutaneous, type III, 203290*
U2AF2	*99,8*	*97,7*	*100*	*100*	*No OMIM disease ID*

UBA1	*99,2*	*97,3*	*99,9*	*99,3*	*Spinal muscular atrophy, X-linked 2, infantile, 301830* *VEXAS syndrome, somatic, 301054*
UBA5	*97,4*	*86,6*	*100*	*100*	*?Spinocerebellar ataxia, autosomal recessive 24, 617133* *Developmental and epileptic encephalopathy 44, 617132*
UBAP1	*98,1*	*91,8*	*100*	*100*	*Spastic paraplegia 80, autosomal dominant, 618418*
UBB	*100*	*99,5*	*100*	*100*	*No OMIM disease ID*
UBE2A	*99,5*	*97,4*	*100*	*100*	*Intellectual developmental disorder, X-linked syndromic, Nascimento type, 300860*
UBE2T	*99,9*	*99,3*	*100*	*100*	*Fanconi anemia, complementation group T, 616435*
UBE3A	*98,9*	*94,1*	*100*	*100*	*Angelman syndrome, 105830*
UBE3B	*100*	*99,7*	*100*	*100*	*Kaufman oculocerebrofacial syndrome, 244450*
UBE4A	*99,7*	*98,7*	*100*	*100*	*No OMIM disease ID*
UBIAD1	*99,5*	*96,6*	*100*	*100*	*Corneal dystrophy, Schnyder type, 121800*
UBQLN2	*99,8*	*98,7*	*100*	*100*	*Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857*
UBR1	*99,6*	*99,1*	*98*	*97,9*	*Johanson-Blizzard syndrome, 243800*
UBR2	*99*	*98,1*	*100*	*99,7*	*No OMIM disease ID*
UBR7	*99,9*	*99,9*	*100*	*100*	*Li-Campeau syndrome, 619189*
UBTF	*99,9*	*99,1*	*100*	*100*	*Neurodegeneration, childhood-onset, with brain atrophy, 617672*
UCHL1	*99,3*	*90,5*	*100*	*100*	*Spastic paraplegia 79, autosomal recessive, 615491*
UFC1	*100*	*100*	*100*	*100*	*Neurodevelopmental disorder with spasticity and poor growth, 618076*
UFM1	*72,4*	*69,1*	*100*	*100*	*Leukodystrophy, hypomyelinating, 14, 617899*
UFSP2	*99,7*	*98,9*	*100*	*99,9*	*?Hip dysplasia, Beukes type, 142669* *?Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974*
UGDH	*99,7*	*99,1*	*100*	*100*	*Developmental and epileptic encephalopathy 84, 618792*
UGP2	*98,7*	*98,2*	*96,4*	*96,3*	*Developmental and epileptic encephalopathy 83, 618744*
UGT1A1	*100*	*100*	*100*	*100*	*Crigler-Najjar syndrome, type I, 218800* *Hyperbilirubinemia, familial transient neonatal, 237900* *Crigler-Najjar syndrome, type II, 606785*
UMOD	*97,5*	*95,9*	*100*	*100*	*Tubulointerstitial kidney disease, autosomal dominant, 1, 162000*
UMPS	*99,9*	*98,7*	*97*	*97*	*Orotic aciduria, 258900*
UNC119	*100*	*98,8*	*100*	*100*	*?Immunodeficiency 13, 615518* *?Cone-rod dystrophy,*

UNC13A	*99,4*	*97,8*	*100*	*99,9*	*No OMIM disease ID*
UNC13D	*99,3*	*97,4*	*100*	*100*	*Hemophagocytic lymphohistiocytosis, familial, 3, 608898*
UNC45A	*84,6*	*81,6*	*100*	*100*	*Osteootohepatoenteric syndrome, 619377*
UNC45B	*99,4*	*98*	*100*	*100*	*?Cataract 43, 616279* *Myofibrillar myopathy 11, 619178*
UNC80	*97,9*	*97,1*	*100*	*100*	*Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801*
UNC93B1	*60,6*	*60*	*100*	*100*	*No OMIM disease ID*
UNG	*99,9*	*97,9*	*100*	*100*	*Immunodeficiency with hyper IgM, type 5, 608106*
UPB1	*100*	*100*	*100*	*100*	*Beta-ureidopropionase deficiency, 613161*
UPF1	*99,8*	*99,6*	*98,3*	*97,6*	*No OMIM disease ID*
UPF3B	*90,8*	*80,3*	*100*	*99,4*	*Intellectual developmental disorder, X-linked syndromic 14, 300676*
UPK3A	*100*	*99,7*	*100*	*100*	*No OMIM disease ID*
UQCC1	*100*	*100*	*100*	*99,9*	*No OMIM disease ID*
UQCC2	*99,9*	*98,5*	*100*	*100*	*Mitochondrial complex III deficiency, nuclear type 7, 615824*
UQCC3	*100*	*97,5*	*100*	*100*	*?Mitochondrial complex III deficiency, nuclear type 9, 616111*
UQCR10	*100*	*100*	*100*	*100*	*No OMIM disease ID*
UQCR11	*100*	*100*	*100*	*100*	*No OMIM disease ID*
UQCRB	*97,7*	*92,1*	*100*	*100*	*Mitochondrial complex III deficiency, nuclear type 3, 615158*
UQCRC1	*99,8*	*98,3*	*100*	*100*	*Parkinsonism with polyneuropathy, 619279*
UQCRC2	*100*	*98,8*	*100*	*100*	*Mitochondrial complex III deficiency, nuclear type 5, 615160*
UQCRFS1	*94,1*	*88,8*	*100*	*100*	*Mitochondrial complex III deficiency, nuclear type 10, 618775*
UQCRH	*100*	*97,3*	*100*	*100*	*No OMIM disease ID*
UQCRQ	*100*	*100*	*100*	*100*	*Mitochondrial complex III deficiency, nuclear type 4, 615159*
UROC1	*100*	*99,9*	*100*	*100*	*?Urocanase deficiency, 276880*
UROD	*98,5*	*95,5*	*100*	*100*	*Porphyria, hepatoerythropoietic, 176100* *Porphyria cutanea tarda, 176100*
UROS	*100*	*99,9*	*100*	*100*	*Porphyria, congenital erythropoietic, 263700*
USB1	*100*	*98,8*	*100*	*100*	*Poikiloderma with neutropenia, 604173*
USH1C	*99,9*	*99,2*	*100*	*100*	*Usher syndrome, type 1C, 276904* *Deafness, autosomal recessive 18A, 602092*

USH1G	*99,7*	*96,6*	*100*	*100*	*Usher syndrome, type 1G, 606943*
USH2A	*100*	*99,7*	*99,5*	*99,5*	*Usher syndrome, type 2A, 276901* *Retinitis pigmentosa 39, 613809*
USP18	*95,9*	*95,9*	*100*	*100*	*Pseudo-TORCH syndrome 2, 617397*
USP26	*100*	*99,5*	*100*	*100*	*No OMIM disease ID*
USP27X	*100*	*99,9*	*100*	*100*	*Intellectual developmental disorder, X-linked 105, 300984*
USP45	*99,4*	*97,6*	*100*	*99,9*	*?Leber congenital amaurosis 19, 618513*
USP48	*99,9*	*98,9*	*100*	*100*	*No OMIM disease ID*
USP7	*90,8*	*85,2*	*94,8*	*94,7*	*Hao-Fountain syndrome, 616863*
USP8	*96,4*	*85,9*	*100*	*99,9*	*Pituitary adenoma 4, ACTH-secreting, somatic, 219090*
USP9X	*98,1*	*91,7*	*100*	*99,8*	*Intellectual developmental disorder, X-linked 99, 300919* *Intellectual developmental disorder, X-linked 99, syndromic, female-restricted, 300968*
UST	*99,9*	*99,3*	*100*	*100*	*No OMIM disease ID*
UVSSA	*100*	*100*	*100*	*100*	*UV-sensitive syndrome 3, 614640*
VAC14	*99,8*	*98,5*	*100*	*100*	*Striatonigral degeneration, childhood-onset, 617054*
VAMP1	*100*	*99,8*	*100*	*100*	*Myasthenic syndrome, congenital, 25, 618323* *Spastic ataxia 1, autosomal dominant, 108600*
VAMP2	*99,1*	*97*	*100*	*100*	*Neurodevelopmental disorder with hypotonia and autistic features with or without hyperkinetic movements, 618760*
VANGL1	*100*	*100*	*100*	*100*	*Caudal regression syndrome, 600145*
VANGL2	*100*	*99,3*	*100*	*100*	*Neural tube defects, 182940*
VAPB	*99,8*	*99,4*	*100*	*99,9*	*Spinal muscular atrophy, late-onset, Finkel type, 182980* *Amyotrophic lateral sclerosis 8, 608627*
VARS1	*100*	*99,7*	*100*	*100*	*Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802*
VARS2	*100*	*99*	*100*	*100*	*Combined oxidative phosphorylation deficiency 20, 615917*
VAV1	*98,2*	*95,1*	*97,1*	*97,1*	*No OMIM disease ID*
VAX1	*98,9*	*93,9*	*96*	*92,3*	*?Microphthalmia, syndromic 11, 614402*
VCAN	*100*	*99,9*	*100*	*100*	*Wagner syndrome 1, 143200*
VCL	*99,9*	*98,5*	*100*	*100*	*Cardiomyopathy, dilated, 1W, 611407* *Cardiomyopathy, hypertrophic, 15, 613255*

VCP	*100*	*99,1*	*100*	*100*	*Frontotemporal dementia and/or amyotrophic lateral sclerosis 6, 613954* *Charcot-Marie-Tooth disease, type 2Y, 616687* *Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320*
VDR	*96,7*	*94,4*	*99,5*	*97,7*	*Rickets, vitamin D-resistant, type IIA, 277440*
VEGFC	*100*	*100*	*100*	*100*	*Lymphatic malformation 4, 615907*
VHL	*95,5*	*90,6*	*100*	*100*	*Erythrocytosis, familial, 2, 263400* *von Hippel-Lindau syndrome, 193300* *Renal cell carcinoma, somatic, 144700* *Pheochromocytoma, 171300* *Hemangioblastoma, cerebellar, somatic,*
VIM	*98,8*	*96,8*	*100*	*100*	*Cataract 30, pulverulent, 116300*
VIPAS39	*100*	*100*	*100*	*100*	*Arthrogryposis, renal dysfunction, and cholestasis 2, 613404*
VKORC1	*100*	*99,8*	*93*	*93*	*Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473* *Warfarin resistance, 122700*
VLDLR	*100*	*100*	*100*	*100*	*Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050*
VMA21	*99*	*92,6*	*100*	*99,8*	*Myopathy, X-linked, with excessive autophagy, 310440*
VPS11	*94,6*	*92,2*	*100*	*100*	*Leukodystrophy, hypomyelinating, 12, 616683*
VPS13A	*98,4*	*94,6*	*100*	*99,9*	*Choreoacanthocytosis, 200150*
VPS13B	*99,4*	*97,8*	*99,4*	*99,3*	*Cohen syndrome, 216550*
VPS13C	*99*	*95,8*	*100*	*99,9*	*Parkinson disease 23, autosomal recessive, early onset, 616840*
VPS13D	*100*	*99,4*	*100*	*100*	*Spinocerebellar ataxia, autosomal recessive 4, 607317*
VPS16	*100*	*100*	*100*	*100*	*Dystonia 30, 619291*
VPS33A	*91,9*	*89,9*	*89,9*	*89,9*	*Mucopolysaccharidosis-plus syndrome, 617303*
VPS33B	*100*	*99,9*	*100*	*100*	*Arthrogryposis, renal dysfunction, and cholestasis 1, 208085*
VPS35	*96,1*	*87,4*	*100*	*100*	*No OMIM disease ID*
VPS35L	*100*	*99,8*	*100*	*100*	*Ritscher-Schinzel syndrome 3, 619135*
VPS37A	*91,3*	*76*	*100*	*100*	*Spastic paraplegia 53, autosomal recessive, 614898*
VPS41	*99,8*	*98,8*	*100*	*100*	*Spinocerebellar ataxia, autosomal recessive 29, 619389*
VPS45	*97,8*	*95,1*	*95,3*	*95,3*	*Neutropenia, severe congenital, 5, autosomal recessive, 615285*
VPS4A	*100*	*99,9*	*100*	*100*	*CIMDAG syndrome, 619273*
VPS51	*96,5*	*84,1*	*100*	*100*	*Pontocerebellar hypoplasia, type 13, 618606*

VPS53	*91,1*	*89,9*	*100*	*99,4*	*Pontocerebellar hypoplasia, type 2E, 615851*
VRK1	*99,4*	*97,8*	*100*	*100*	*Pontocerebellar hypoplasia type 1A, 607596*
VSX1	*85,4*	*81,3*	*100*	*100*	*?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195* *Keratoconus 1, 148300*
VSX2	*100*	*99,2*	*100*	*100*	*Microphthalmia, isolated 2, 610093* *Microphthalmia with coloboma 3, 610092*
VWA1	*85,4*	*77,7*	*99,6*	*96,1*	*Neuropathy, hereditary motor, with myopathic features, 619216*
VWA3B	*99,9*	*98,9*	*100*	*100*	*?Spinocerebellar ataxia, autosomal recessive 22, 616948*
VWF	*99,9*	*99*	*100*	*100*	*von Willebrand disease, type 1, 193400* *von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554* *von Willebrand disease, type 3, 277480*
WAC	*99,8*	*99,6*	*100*	*100*	*Desanto-Shinawi syndrome, 616708*
WARS1	*99,6*	*97,4*	*100*	*100*	*Neuronopathy, distal hereditary motor, type IX, 617721*
WARS2	*100*	*99,8*	*100*	*99,8*	*Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710*
WAS	*94,1*	*83,7*	*100*	*100*	*Wiskott-Aldrich syndrome, 301000* *Neutropenia, severe congenital, X-linked, 300299* *Thrombocytopenia, X-linked, intermittent, 313900* *Thrombocytopenia, X-linked, 313900*
WASF1	*99,2*	*94,1*	*100*	*100*	*Neurodevelopmental disorder with absent language and variable seizures, 618707*
WASHC4	*98,9*	*96*	*100*	*99,8*	*?Mental retardation, autosomal recessive 43, 615817*
WASHC5	*99,8*	*99,7*	*100*	*100*	*Ritscher-Schinzel syndrome 1, 220210* *Spastic paraplegia 8, autosomal dominant, 603563*
WBP11	*96,3*	*83,3*	*100*	*99,9*	*Vertebral, cardiac, tracheoesophageal, renal, and limb defects, 619227*
WBP2	*100*	*99,2*	*100*	*100*	*Deafness, autosomal recessive 107, 617639*
WDFY3	*99,8*	*99,1*	*100*	*100*	*?Microcephaly 18, primary, autosomal dominant, 617520*
WDPCP	*98*	*94,1*	*98,1*	*98*	*?Bardet-Biedl syndrome 15, 615992* *Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085*
WDR1	*99,9*	*98,7*	*100*	*100*	*Periodic fever, immunodeficiency, and thrombocytopenia syndrome, 150550*
WDR11	*98,2*	*96,5*	*100*	*100*	*Hypogonadotropic hypogonadism 14 with or without anosmia, 614858*
WDR13	*99,8*	*98,7*	*100*	*100*	*No OMIM disease ID*
WDR19	*99,8*	*98,6*	*100*	*99,9*	*Nephronophthisis 13, 614377* *Senior-Loken syndrome 8, 616307*

					?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 *?Cranioectodermal dysplasia 4, 614378*
WDR26	*89,2*	*84,2*	*95,9*	*92,9*	*Skraban-Deardorff syndrome, 617616*
WDR35	*99,6*	*98,4*	*100*	*100*	*Short-rib thoracic dysplasia 7 with or without polydactyly, 614091* *Cranioectodermal dysplasia 2, 613610*
WDR36	*99,7*	*97,8*	*100*	*100*	*Glaucoma 1, open angle, G, 609887*
WDR37	*86,5*	*86,3*	*86,5*	*86,5*	*Neurooculocardiogenitourinary syndrome, 618652*
WDR4	*100*	*100*	*100*	*100*	*Galloway-Mowat syndrome 6, 618347* *Microcephaly, growth deficiency, seizures, and brain malformations, 618346*
WDR45	*98,2*	*92*	*100*	*100*	*Neurodegeneration with brain iron accumulation 5, 300894*
WDR45B	*94,8*	*80,3*	*100*	*100*	*Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977*
WDR62	*100*	*99,9*	*100*	*100*	*Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317*
WDR72	*96,5*	*95,6*	*96,9*	*96,9*	*Amelogenesis imperfecta, type IIA3, 613211*
WDR73	*100*	*100*	*100*	*100*	*Galloway-Mowat syndrome 1, 251300*
WDR81	*100*	*100*	*100*	*100*	*Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185* *Hydrocephalus, congenital, 3, with brain anomalies, 617967*
WEE2	*99,9*	*99,6*	*100*	*100*	*Oocyte maturation defect 5, 617996*
WFS1	*100*	*99,8*	*100*	*100*	*Deafness, autosomal dominant 6/14/38, 600965* *?Cataract 41, 116400* *Wolfram-like syndrome, autosomal dominant, 614296* *Wolfram syndrome 1, 222300*
WHRN	*99,6*	*97,6*	*100*	*100*	*Deafness, autosomal recessive 31, 607084* *Usher syndrome, type 2D, 611383*
WIPF1	*99,9*	*98,5*	*100*	*100*	*Wiskott-Aldrich syndrome 2, 614493*
WIPI2	*99,7*	*98,1*	*100*	*100*	*?Intellectual developmental disorder with short stature and variable skeletal anomalies, 618453*
WNK1	*99,8*	*99,3*	*100*	*100*	*Neuropathy, hereditary sensory and autonomic, type II, 201300* *Pseudohypoaldosteronism, type IIC, 614492*
WNK4	*99,9*	*99*	*100*	*100*	*Pseudohypoaldosteronism, type IIB, 614491*
WNT1	*99,9*	*96,6*	*100*	*100*	*Osteogenesis imperfecta, type XV, 615220*
WNT10A	*100*	*98,9*	*100*	*100*	*Schopf-Schulz-Passarge syndrome, 224750* *Tooth agenesis, selective, 4, 150400* *Odontoonychodermal dysplasia, 257980*
WNT10B	*100*	*99,6*	*100*	*100*	*Tooth agenesis, selective, 8, 617073* *Split-hand/foot malformation 6, 225300*

WNT2B	*97,2*	*88,6*	*100*	*100*	*Diarrhea 9, 618168*
WNT3	*100*	*99,9*	*100*	*100*	*?Tetra-amelia syndrome 1, 273395*
WNT4	*97,8*	*93,6*	*99,3*	*96,5*	*?SERKAL syndrome, 611812* *Mullerian aplasia and hyperandrogenism, 158330*
WNT5A	*100*	*100*	*100*	*100*	*Robinow syndrome, autosomal dominant 1, 180700*
WNT6	*100*	*99,3*	*100*	*100*	*No OMIM disease ID*
WNT7A	*100*	*100*	*100*	*100*	*Fuhrmann syndrome, 228930* *Ulna and fibula, absence of, with severe limb deficiency, 276820*
WRAP53	*100*	*100*	*100*	*99,9*	*Dyskeratosis congenita, autosomal recessive 3, 613988*
WRN	*99,3*	*98,2*	*100*	*99,9*	*Werner syndrome, 277700*
WT1	*97,6*	*96,1*	*97,7*	*97,7*	*Mesothelioma, somatic, 156240* *Meacham syndrome, 608978* *Frasier syndrome, 136680* *Nephrotic syndrome, type 4, 256370* *Denys-Drash syndrome, 194080* *Wilms tumor, type 1, 194070*
WWOX	*100*	*99,9*	*100*	*100*	*Esophageal squamous cell carcinoma, somatic, 133239* *Developmental and epileptic encephalopathy 28, 616211* *Spinocerebellar ataxia, autosomal recessive 12, 614322*
XDH	*100*	*99,8*	*100*	*100*	*Xanthinuria, type I, 278300*
XIAP	*93,1*	*88,3*	*99,9*	*99,6*	*Lymphoproliferative syndrome, X-linked, 2, 300635*
XIRP2	*99,9*	*99,7*	*100*	*99,9*	*No OMIM disease ID*
XIST	*NC*	*NC*	*NC*	*NC*	*X-inactivation, familial skewed, 300087*
XK	*99,7*	*97,6*	*100*	*100*	*McLeod syndrome with or without chronic granulomatous disease, 300842*
XPA	*99,2*	*97,3*	*100*	*100*	*Xeroderma pigmentosum, group A, 278700*
XPC	*100*	*99,9*	*100*	*100*	*Xeroderma pigmentosum, group C, 278720*
XPNPEP3	*100*	*100*	*100*	*100*	*Nephronophthisis-like nephropathy 1, 613159*
XPO5	*100*	*99,5*	*99,9*	*99,7*	*No OMIM disease ID*
XPR1	*99,9*	*99,8*	*100*	*100*	*Basal ganglia calcification, idiopathic, 6, 616413*
XRCC1	*99,6*	*97,2*	*100*	*100*	*?Spinocerebellar ataxia, autosomal recessive 26, 617633*
XRCC2	*99,6*	*95,7*	*100*	*100*	*Spermatogenic failure, 619145* *?Premature ovarian failure 17, 619146* *?Fanconi anemia, complementation group U, 617247*

XRCC4	*99,7*	*98,4*	*100*	*100*	*Short stature, microcephaly, and endocrine dysfunction, 616541*
XYLT1	*97,8*	*91,1*	*97,7*	*94,1*	*Desbuquois dysplasia 2, 615777*
XYLT2	*99,9*	*97,1*	*96,7*	*96,7*	*Spondyloocular syndrome, 605822*
YAP1	*96,6*	*90,3*	*100*	*100*	*Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433*
YARS1	*100*	*99,8*	*100*	*100*	*Infantile-onset multisystem neurologic, endocrine, and pancreatic disease 2, 619418* *Charcot-Marie-Tooth disease, dominant intermediate C, 608323*
YARS2	*99,9*	*99,4*	*100*	*100*	*Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561*
YEATS2	*99,7*	*98*	*100*	*100*	*?Epilepsy, myoclonic, familial adult, 4, 615127*
YIF1B	*99,9*	*99,2*	*90,1*	*90,1*	*Kaya-Barakat-Masson syndrome, 619125*
YIPF5	*100*	*100*	*100*	*100*	*Microcephaly, epilepsy, and diabetes syndrome 2, 619278*
YME1L1	*98,9*	*93,7*	*100*	*100*	*?Optic atrophy 11, 617302*
YPEL2	*96,6*	*90,8*	*100*	*99,5*	*No OMIM disease ID*
YWHAE	*100*	*100*	*100*	*100*	*No OMIM disease ID*
YWHAG	*100*	*99,9*	*100*	*100*	*Developmental and epileptic encephalopathy 56, 617665*
YWHAZ	*76,7*	*66,8*	*100*	*100*	*No OMIM disease ID*
YY1	*99,9*	*99,3*	*100*	*100*	*Gabriele-de Vries syndrome, 617557*
YY1AP1	*98,5*	*97*	*100*	*100*	*Grange syndrome, 602531*
ZAP70	*100*	*99,7*	*100*	*100*	*Immunodeficiency 48, 269840* *Autoimmune disease, multisystem, infantile-onset, 2, 617006*
ZBTB11	*99,9*	*99,3*	*100*	*100*	*Intellectual developmental disorder, autosomal recessive 69, 618383*
ZBTB16	*100*	*100*	*100*	*100*	*Skeletal defects, genital hypoplasia, and mental retardation, 612447* *Leukemia, acute promyelocytic, PL2F/RARA type,*
ZBTB17	*100*	*100*	*100*	*100*	*No OMIM disease ID*
ZBTB18	*100*	*99,8*	*100*	*99,8*	*Mental retardation, autosomal dominant 22, 612337*
ZBTB20	*100*	*100*	*100*	*100*	*Primrose syndrome, 259050*
ZBTB24	*100*	*100*	*100*	*100*	*Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069*
ZBTB42	*100*	*100*	*100*	*100*	*?Lethal congenital contracture syndrome 6, 616248*
ZC3H14	*99,7*	*98,2*	*100*	*99,9*	*Mental retardation, autosomal recessive 56, 617125*
ZC4H2	*100*	*98,1*	*100*	*100*	*Wieacker-Wolff syndrome, 314580* *Wieacker-Wolff syndrome, female-restricted, 301041*
ZCCHC8	*99,7*	*98*	*100*	*100*	*?Pulmonary fibrosis and/or bone marrow failure, telomere-related, 5, 618674*

ZDHHC9	*97,3*	*84,5*	*100*	*100*	*Mental retardation, X-linked syndromic, Raymond type, 300799*
ZEB1	*100*	*99,5*	*100*	*100*	*Corneal dystrophy, posterior polymorphous, 3, 609141* *Corneal dystrophy, Fuchs endothelial, 6, 613270*
ZEB2	*99,7*	*98,5*	*97,4*	*97,4*	*Mowat-Wilson syndrome, 235730*
ZFHX2	*99,8*	*99,1*	*100*	*100*	*?Marsili syndrome, 147430*
ZFHX3	*100*	*99,4*	*100*	*100*	*Prostate cancer, somatic, 176807*
ZFHX4	*100*	*99,7*	*100*	*100*	*No OMIM disease ID*
ZFP57	*100*	*99,6*	*100*	*100*	*Diabetes mellitus, transient neonatal 1, 601410*
ZFPM2	*100*	*99,9*	*100*	*100*	*Diaphragmatic hernia 3, 610187* *46XY sex reversal 9, 616067* *Tetralogy of Fallot, 187500*
ZFYVE26	*99,7*	*97,8*	*100*	*100*	*Spastic paraplegia 15, autosomal recessive, 270700*
ZFYVE27	*100*	*99,8*	*100*	*100*	*Spastic paraplegia 33, autosomal dominant, 610244*
ZIC1	*100*	*100*	*100*	*100*	*?Craniosynostosis 6, 616602* *Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736*
ZIC2	*100*	*99,3*	*97,7*	*94,2*	*Holoprosencephaly 5, 609637*
ZIC3	*100*	*99,9*	*100*	*100*	*Congenital heart defects, nonsyndromic, 1, X-linked, 306955* *Heterotaxy, visceral, 1, X-linked, 306955* *VACTERL association, X-linked, 314390*
ZMIZ1	*99,8*	*99*	*100*	*100*	*Neurodevelopmental disorder with dysmorphic facies and distal skeletal anomalies, 618659*
ZMPSTE24	*99,6*	*99,4*	*100*	*99,9*	*Mandibuloacral dysplasia with type B lipodystrophy, 608612* *Restrictive dermopathy, lethal, 275210*
ZMYM2	*99,7*	*97,8*	*100*	*100*	*No OMIM disease ID*
ZMYND10	*100*	*100*	*100*	*100*	*Ciliary dyskinesia, primary, 22, 615444*
ZMYND11	*99,9*	*99,7*	*100*	*100*	*Mental retardation, autosomal dominant 30, 616083*
ZMYND15	*99,9*	*99,1*	*100*	*100*	*?Spermatogenic failure 14, 615842*
ZNF141	*99,9*	*99,6*	*100*	*100*	*?Polydactyly, postaxial, type A6, 615226*
ZNF142	*100*	*99,7*	*100*	*100*	*Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425*
ZNF148	*99,9*	*99,8*	*100*	*100*	*Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260*
ZNF292	*99,5*	*98,1*	*99,6*	*99,6*	*Intellectual developmental disorder, autosomal dominant 64, 619188*
ZNF335	*100*	*99,7*	*100*	*100*	*Microcephaly 10, primary, autosomal recessive, 615095*
ZNF341	*97,3*	*95,9*	*100*	*100*	*Hyper-IgE recurrent infection syndrome 3, autosomal recessive, 618282*

ZNF407	*99,9*	*99,1*	*100*	*100*	*No OMIM disease ID*
ZNF408	*100*	*100*	*100*	*100*	*Retinitis pigmentosa 72, 616469* *?Exudative vitreoretinopathy 6, 616468*
ZNF41	*100*	*99,7*	*100*	*100*	*No OMIM disease ID*
ZNF423	*100*	*100*	*100*	*100*	*Nephronophthisis 14, 614844* *Joubert syndrome 19, 614844*
ZNF462	*100*	*99,8*	*100*	*100*	*Weiss-Kruszka syndrome, 618619*
ZNF469	*100*	*100*	*100*	*100*	*Brittle cornea syndrome 1, 229200*
ZNF513	*100*	*100*	*100*	*100*	*?Retinitis pigmentosa 58, 613617*
ZNF526	*100*	*100*	*100*	*100*	*No OMIM disease ID*
ZNF592	*100*	*99,8*	*100*	*100*	*No OMIM disease ID*
ZNF644	*100*	*99,8*	*100*	*100*	*Myopia 21, autosomal dominant, 614167*
ZNF687	*100*	*100*	*100*	*100*	*Paget disease of bone 6, 616833*
ZNF699	*99,9*	*99,2*	*100*	*100*	*DEGCAGS syndrome, 619488*
ZNF711	*99,4*	*96,5*	*100*	*99,8*	*Intellectual developmental disorder, X-linked 97, 300803*
ZNF750	*100*	*99,8*	*100*	*100*	*Seborrhea-like dermatitis with psoriasiform elements, 610227*
ZNFX1	*100*	*99,7*	*100*	*100*	*No OMIM disease ID*
ZNHIT3	*74,4*	*74,4*	*75,7*	*74,4*	*PEHO syndrome, 260565*
ZP1	*100*	*100*	*100*	*100*	*Oocyte maturation defect 1, 615774*
ZP2	*99,8*	*98,4*	*100*	*100*	*Oocyte maturation defect 6, 618353*
ZP3	*100*	*100*	*100*	*100*	*Oocyte maturation defect 3, 617712*
ZPR1	*99,3*	*96,7*	*100*	*100*	*?Growth restriction, hypoplastic kidneys, alopecia, and distinctive facies, 619321*
ZSWIM6	*95,1*	*91,6*	*94,3*	*91*	*Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865* *Acromelic frontonasal dysostosis, 603671*

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.
Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.
Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.
TWIST is the chemistry used for (in-house) TURBO/RAPID WES analysis.
Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.

Genes with coverage denoting NC are non-protein coding genes.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : September 16th , 2021.
This list is accurate for panel version DG 3.2.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions betw een {} signify risk factors