PRECONCEPTION SCREENING GENE PANEL DG 2.16 (2183 genes)

Releasedate: 07-06-2019

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM disease ID*
AAAS	102,3	100.0%	99.6%	Achalasia-addisonianism-alacrimia syndrome, 231550
AARS	103,7	100.0%	99.5%	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 Epileptic encephalopathy, early infantile, 29, 616339
AARS2	122,7	100.0%	99.8%	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889
AASS	131,5	99.9%	99.4%	Hyperlysinemia, 238700 Saccharopinuria, 268700
ABAT	83,2	99.9%	98.3%	GABA-transaminase deficiency, 613163
ABCA1	97	99.9%	98.4%	HDL deficiency, type 2, 604091 Tangier disease, 205400 {Coronary artery disease in familial hypercholesterolemia, protection against}, 143890
ABCA12	129,3	99.6%	98.4%	Ichthyosis, congenital, autosomal recessive 4A, 601277 Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500
ABCA3	119,4	100.0%	99.5%	Surfactant metabolism dysfunction, pulmonary, 3, 610921
ABCA4	104,5	99.9%	98.9%	Cone-rod dystrophy 3, 604116 Fundus flavimaculatus, 248200 Retinal dystrophy, early-onset severe, 248200 Retinitis pigmentosa 19, 601718 Stargardt disease 1, 248200 {Macular degeneration, age-related, 2}, 153800
ABCB11	134,9	100.0%	99.2%	Cholestasis, benign recurrent intrahepatic, 2, 605479 Cholestasis, progressive familial intrahepatic 2, 601847
ABCB4	123,3	99.9%	99.2%	Cholestasis, intrahepatic, of pregnancy, 3, 614972 Cholestasis, progressive familial intrahepatic 3, 602347 Gallbladder disease 1, 600803
ABCC2	110,7	100.0%	99.8%	Dubin-Johnson syndrome, 237500
ABCC6	109,1	93.6%	92.8%	Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850
ABCC8	125,8	100.0%	99.9%	Diabetes mellitus, noninsulin-dependent, 125853 Diabetes mellitus, permanent neonatal, 606176

				Diabetes mellitus, transient neonatal 2, 610374 Hyperinsulinemic hypoglycemia, familial, 1, 256450 Hypoglycemia of infancy, leucine-sensitive, 240800
ABCD3	108,1	99.5%	97.3%	?Bile acid synthesis defect, congenital, 5, 616278
ABCD4	129	99.8%	98.4%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	139,5	100.0%	99.9%	Sitosterolemia, 210250
ABCG8	133,9	99.7%	98.2%	Sitosterolemia, 210250 {Gallbladder disease 4}, 611465
ABHD12	93,1	100.0%	98.9%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ABHD5	180,9	100.0%	100.0%	Chanarin-Dorfman syndrome, 275630
ACACA	109,9	98.3%	97.5%	Acetyl-CoA carboxylase deficiency, 613933
ACAD8	122,1	100.0%	99.9%	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	124,3	99.9%	99.1%	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADM	129,5	99.8%	99.2%	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	148,2	100.0%	100.0%	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	113,1	99.9%	97.3%	2-methylbutyrylglycinuria, 610006
ACADVL	115,8	99.8%	98.0%	VLCAD deficiency, 201475
ACAN	121,3	94.6%	89.1%	?Spondyloepiphyseal dysplasia, Kimberley type, 608361 Short stature and advanced bone age, with or without early-onset osteoarthritis and/or osteochondritis dissecans, 165800 Spondyloepimetaphyseal dysplasia, aggrecan type, 612813
ACAT1	110,2	99.7%	98.3%	Alpha-methylacetoacetic aciduria, 203750
ACD	159,6	100.0%	100.0%	?Dyskeratosis congenita, autosomal dominant 6, 616553 ?Dyskeratosis congenita, autosomal recessive 7, 616553
ACE	120,1	99.9%	99.5%	Renal tubular dysgenesis, 267430 [Angiotensin I-converting enzyme, benign serum increase], 0 {Microvascular complications of diabetes 3}, 612624 {Myocardial infarction, susceptibility to}, 0 {SARS, progression of}, 0 {Stroke, hemorrhagic}, 614519
ACER3	115,7	99.6%	98.9%	?Leukodystrophy, progressive, early childhood-onset, 617762
ACO2	115,3	95.8%	89.5%	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559
ACOX1	123,7	100.0%	100.0%	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACOX2	111	100.0%	99.3%	Bile acid synthesis defect, congenital, 6, 617308
ACP5	172,6	100.0%	99.6%	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACSF3	145,8	99.9%	99.1%	Combined malonic and methylmalonic aciduria, 614265

ACTA1	95,3	99.8%	97.9%	?Myopathy, scapulohumeroperoneal, 616852 Myopathy, actin, congenital, with cores, 161800 Myopathy, actin, congenital, with excess of thin myofilaments, 161800 Myopathy, congenital, with fiber-type disproportion 1, 255310 Nemaline myopathy 3, autosomal dominant or recessive, 161800
ACY1	118,5	100.0%	98.6%	Aminoacylase 1 deficiency, 609924
ADA	104,6	100.0%	99.6%	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADA2	83,5	99.9%	97.6%	?Sneddon syndrome, 182410 Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688
ADAM17	119	99.8%	98.6%	?Inflammatory skin and bowel disease, neonatal, 1, 614328
ADAM22	133,9	99.9%	99.4%	?Epileptic encephalopathy, early infantile, 61, 617933
ADAM9	141,3	99.9%	99.0%	Cone-rod dystrophy 9, 612775
ADAMTS10	122,8	100.0%	99.8%	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS13	103,8	98.1%	95.2%	Thrombotic thrombocytopenic purpura, familial, 274150
ADAMTS17	109,2	97.6%	92.3%	Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTS18	129,7	100.0%	99.9%	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
ADAMTS2	126,3	100.0%	99.6%	Ehlers-Danlos syndrome, dermatosparaxis type, 225410
ADAMTSL2	115,9	99.0%	96.3%	Geleophysic dysplasia 1, 231050
ADAMTSL4	122,7	100.0%	99.6%	Ectopia lentis et pupillae, 225200 Ectopia lentis, isolated, autosomal recessive, 225100
ADAR	109,2	99.9%	99.3%	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
ADAT3	128	100.0%	99.9%	Mental retardation, autosomal recessive 36, 615286
ADCY1	134,5	96.8%	95.4%	?Deafness, autosomal recessive 44, 610154
ADCY6	162,8	100.0%	100.0%	?Lethal congenital contracture syndrome 8, 616287
ADD3	143,5	99.9%	99.6%	Cerebral palsy, spastic quadriplegic, 3, 617008
ADGRG1	147,2	100.0%	100.0%	Polymicrogyria, bilateral frontoparietal, 606854 Polymicrogyria, bilateral perisylvian, 615752
ADGRG6	135,7	99.8%	98.7%	Lethal congenital contracture syndrome 9, 616503
ADGRV1	126	99.7%	98.4%	?Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472
ADK	102,4	99.8%	98.0%	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADSL	138,6	99.2%	98.6%	Adenylosuccinase deficiency, 103050
ADSSL1	110,7	95.5%	87.9%	Myopathy, distal, 5, 617030
AEBP1	147,7	100.0%	100.0%	Ehlers-Danlos syndrome, classic-like, 2, 618000

AFG3L2	98,3	95.9%	86.1%	Spastic ataxia 5, autosomal recessive, 614487 Spinocerebellar ataxia 28, 610246
AGA	142,7	100.0%	100.0%	Aspartylglucosaminuria, 208400
AGBL5	104,9	100.0%	99.4%	Retinitis pigmentosa 75, 617023
AGK	108,5	99.5%	95.7%	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AGL	146,9	100.0%	99.4%	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGPAT2	162,6	99.1%	94.8%	Lipodystrophy, congenital generalized, type 1, 608594
AGPS	75,4	99.5%	97.8%	Rhizomelic chondrodysplasia punctata, type 3, 600121
AGRN	151,6	98.4%	94.5%	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120
AGT	192,1	100.0%	100.0%	Renal tubular dysgenesis, 267430 {Hypertension, essential, susceptibility to}, 145500 {Preeclampsia, susceptibility to}, 0
AGTR1	143,6	91.9%	91.6%	Renal tubular dysgenesis, 267430 {Hypertension, essential}, 145500
AGXT	160,8	100.0%	100.0%	Hyperoxaluria, primary, type 1, 259900
AHCY	111,6	99.9%	97.7%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHI1	129,8	99.7%	98.3%	Joubert syndrome 3, 608629
AHSG	160,8	100.0%	99.8%	?Alopecia-mental retardation syndrome 1, 203650
AICDA	128,4	100.0%	99.5%	Immunodeficiency with hyper-IgM, type 2, 605258
AIMP1	79,4	99.1%	92.4%	Leukodystrophy, hypomyelinating, 3, 260600
AIMP2	117,6	96.1%	89.3%	Leukodystrophy, hypomyelinating, 17, 618006
AIPL1	113	100.0%	99.9%	Cone-rod dystrophy, 604393 Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393
AIRE	102,3	100.0%	99.9%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AK1	136,9	100.0%	99.9%	Hemolytic anemia due to adenylate kinase deficiency, 612631
AK2	96,1	98.7%	94.4%	Reticular dysgenesis, 267500
AKR1C2	135,8	94.9%	87.9%	46XY sex reversal 8, 614279
AKR1D1	91,5	99.6%	95.7%	Bile acid synthesis defect, congenital, 2, 235555
ALAD	94,9	99.5%	94.7%	Porphyria, acute hepatic, 612740 {Lead poisoning, susceptibility to}, 612740
ALB	156,5	100.0%	99.2%	Analbuminemia, 616000 [Dysalbuminemic hyperthyroxinemia], 615999
ALDH18A1	113,7	100.0%	99.8%	Cutis laxa, autosomal dominant 3, 616603 Cutis laxa, autosomal recessive, type IIIA, 219150

				Spastic paraplegia 9A, autosomal dominant, 601162 Spastic paraplegia 9B, autosomal recessive, 616586
ALDH1A3	102,4	99.7%	97.0%	Microphthalmia, isolated 8, 615113
ALDH3A2	113,5	95.3%	94.3%	Sjogren-Larsson syndrome, 270200
ALDH4A1	123,9	100.0%	99.8%	Hyperprolinemia, type II, 239510
ALDH5A1	91	99.3%	93.2%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	104,9	100.0%	99.6%	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	66,7	93.5%	86.1%	Epilepsy, pyridoxine-dependent, 266100
ALDOA	120,4	76.5%	74.5%	Glycogen storage disease XII, 611881
ALDOB	135,3	100.0%	99.3%	Fructose intolerance, hereditary, 229600
ALG1	46,5	53.2%	50.2%	Congenital disorder of glycosylation, type Ik, 608540
ALG11	129,3	96.8%	96.3%	Congenital disorder of glycosylation, type Ip, 613661
ALG12	155,7	100.0%	99.9%	Congenital disorder of glycosylation, type Ig, 607143
ALG14	199,4	100.0%	100.0%	?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227
ALG2	103,2	100.0%	100.0%	?Congenital disorder of glycosylation, type Ii, 607906 Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228
ALG3	106,5	100.0%	99.9%	Congenital disorder of glycosylation, type Id, 601110
ALG6	101,6	99.1%	95.6%	Congenital disorder of glycosylation, type Ic, 603147
ALG8	118,5	96.6%	96.2%	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	113	100.0%	99.6%	Congenital disorder of glycosylation, type Il, 608776 Gillessen-Kaesbach-Nishimura syndrome, 263210
ALMS1	172,8	100.0%	99.7%	Alstrom syndrome, 203800
ALOX12B	125,6	100.0%	99.8%	Ichthyosis, congenital, autosomal recessive 2, 242100
ALOXE3	124,7	100.0%	99.4%	Ichthyosis, congenital, autosomal recessive 3, 606545
ALPK3	113,9	99.4%	97.2%	Cardiomyopathy, familial hypertrophic 27, 618052
ALPL	154,8	100.0%	99.7%	Hypophosphatasia, adult, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500 Odontohypophosphatasia, 146300
ALS2	145,1	100.0%	99.8%	Amyotrophic lateral sclerosis 2, juvenile, 205100 Primary lateral sclerosis, juvenile, 606353 Spastic paralysis, infantile onset ascending, 607225
ALX1	134,2	99.9%	98.5%	?Frontonasal dysplasia 3, 613456
ALX3	134,6	91.1%	79.0%	Frontonasal dysplasia 1, 136760
ALX4	157,1	100.0%	100.0%	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597

				{Craniosynostosis 5, susceptibility to}, 615529
AMACR	157,7	100.0%	100.0%	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
AMBN	175,1	99.3%	97.1%	Amelogenesis imperfecta, type IF, 616270
AMN	101,5	98.1%	90.6%	Megaloblastic anemia-1, Norwegian type, 261100
AMPD1	115,8	99.9%	98.6%	Myopathy due to myoadenylate deaminase deficiency, 615511
AMPD2	132,3	100.0%	99.9%	?Spastic paraplegia 63, 615686 Pontocerebellar hypoplasia, type 9, 615809
AMT	142,7	100.0%	100.0%	Glycine encephalopathy, 605899
ANGPTL3	94	99.2%	95.9%	Hypobetalipoproteinemia, familial, 2, 605019
ANK3	139,9	99.4%	99.0%	?Mental retardation, autosomal recessive, 37, 615493
ANKH	111,6	100.0%	99.9%	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000
ANKLE2	144,7	100.0%	99.8%	?Microcephaly 16, primary, autosomal recessive, 616681
ANKS6	94,2	98.3%	94.4%	Nephronophthisis 16, 615382
ANO10	106	98.9%	96.3%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANO5	131	99.6%	97.3%	Gnathodiaphyseal dysplasia, 166260 Miyoshi muscular dystrophy 3, 613319 Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307
ANO6	133,3	99.8%	98.0%	Scott syndrome, 262890
ANTXR1	108,3	99.0%	96.9%	GAPO syndrome, 230740 {?Hemangioma, capillary infantile, susceptibility to}, 602089
ANTXR2	119,3	99.9%	98.6%	Hyaline fibromatosis syndrome, 228600
AP1S1	101	100.0%	99.8%	MEDNIK syndrome, 609313
AP3B1	112,1	99.5%	96.5%	Hermansky-Pudlak syndrome 2, 608233
AP3B2	125,6	99.4%	97.6%	Epileptic encephalopathy, early infantile, 48, 617276
AP3D1	125,2	98.4%	97.9%	?Hermansky-Pudlak syndrome 10, 617050
AP4B1	121	99.9%	98.4%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	106,6	99.8%	98.8%	Spastic paraplegia 51, autosomal recessive, 613744 Stuttering, familial persistent, 1, 184450
AP4M1	129,3	99.7%	98.1%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	66,2	78.5%	71.3%	Spastic paraplegia 52, autosomal recessive, 614067
AP5Z1	121,3	100.0%	100.0%	Spastic paraplegia 48, autosomal recessive, 613647
APC2	122,4	99.9%	98.7%	?Sotos syndrome 3, 617169
APOB	154,7	100.0%	99.7%	Hypercholesterolemia, due to ligand-defective apo B, 144010 Hypobetalipoproteinemia, 615558
APOC2	103,7	100.0%	100.0%	Hyperlipoproteinemia, type Ib, 207750

APOE	83,3	100.0%	99.9%	Alzheimer disease-2, 104310 Hyperlipoproteinemia, type III, 617347 Lipoprotein glomerulopathy, 611771 Sea-blue histiocyte disease, 269600 {?Macular degeneration, age-related}, 603075 {Coronary artery disease, severe, susceptibility to}, 617347
APOPT1	NC	NC	NC	Mitochondrial complex IV deficiency, 220110
APRT	93,2	100.0%	100.0%	Adenine phosphoribosyltransferase deficiency, 614723
APTX	96,3	94.1%	91.3%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
AQP2	126,1	100.0%	99.9%	Diabetes insipidus, nephrogenic, 125800
ARFGEF2	125,2	99.7%	98.7%	Periventricular heterotopia with microcephaly, 608097
ARG1	159,1	100.0%	100.0%	Argininemia, 207800
ARHGDIA	202,4	100.0%	100.0%	Nephrotic syndrome, type 8, 615244
ARHGEF18	140,4	99.5%	97.3%	Retinitis pigmentosa 78, 617433
ARHGEF2	113,5	100.0%	99.8%	?Neurodevelopmental disorder with midbrain and hindbrain malformations, 617523
ARL13B	102,2	100.0%	99.4%	Joubert syndrome 8, 612291
ARL2BP	63,8	92.4%	83.1%	Retinitis pigmentosa with or without situs inversus, 615434
ARL6	100,3	99.8%	98.2%	?Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151 {Bardet-Biedl syndrome 1, modifier of}, 209900
ARL6IP1	62,7	94.8%	76.6%	?Spastic paraplegia 61, autosomal recessive, 615685
ARMC4	107,2	94.4%	93.5%	Ciliary dyskinesia, primary, 23, 615451
ARMC9	124,8	100.0%	99.3%	Joubert syndrome 30, 617622
ARNT2	120,9	100.0%	100.0%	?Webb-Dattani syndrome, 615926
ARSA	138,5	100.0%	100.0%	Metachromatic leukodystrophy, 250100
ARSB	109,4	99.9%	98.9%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARV1	108,9	100.0%	99.2%	Epileptic encephalopathy, early infantile, 38, 617020
ASAH1	125,7	99.3%	97.2%	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASCC1	125,4	95.7%	92.0%	?Spinal muscular atrophy with congenital bone fractures 2, 616867 Barrett esophagus/esophageal adenocarcinoma, 614266
ASL	123,6	100.0%	98.5%	Argininosuccinic aciduria, 207900
ASNS	82,8	98.6%	92.2%	Asparagine synthetase deficiency, 615574
ASPA	116,1	99.7%	96.9%	Canavan disease, 271900
ASPH	111,3	99.9%	98.8%	Traboulsi syndrome, 601552
ASPM	111,6	99.7%	98.0%	Microcephaly 5, primary, autosomal recessive, 608716
ASS1	97,4	95.0%	87.1%	Citrullinemia, 215700

ATAD1	65,2	99.3%	89.8%	Hyperekplexia 4, 618011
ATCAY	152,7	100.0%	99.7%	Ataxia, cerebellar, Cayman type, 601238
ATF6	125,2	100.0%	99.3%	Achromatopsia 7, 616517
ATG5	126,9	99.1%	97.0%	?Spinocerebellar ataxia, autosomal recessive 25, 617584
ATIC	113,9	100.0%	99.7%	AICA-ribosiduria due to ATIC deficiency, 608688
ATM	110,9	99.6%	97.2%	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic, 0 Lymphoma, mantle cell, somatic, 0 T-cell prolymphocytic leukemia, somatic, 0 {Breast cancer, susceptibility to}, 114480
ATOH7	176,2	98.6%	97.0%	Persistent hyperplastic primary vitreous, autosomal recessive, 221900
ATP13A2	134,1	99.9%	99.7%	Kufor-Rakeb syndrome, 606693 Spastic paraplegia 78, autosomal recessive, 617225
ATP2A1	146,4	100.0%	100.0%	Brody myopathy, 601003
ATP5A1	NC	NC	NC	?Combined oxidative phosphorylation deficiency 22, 616045 ?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4, 615228
ATP5D	NC	NC	NC	Mitochondrial complex V (ATP synthase) deficiency, 618120
ATP5E	NC	NC	NC	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053
ATP6V0A2	117,4	99.9%	99.0%	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250
ATP6V0A4	105,4	100.0%	99.2%	Renal tubular acidosis, distal, autosomal recessive, 602722
ATP6V1A	133,4	99.6%	97.3%	Cutis laxa, autosomal recessive, type IID, 617403 Epileptic encephalopathy, infantile or early childhood, 3, 618012
ATP6V1B1	172,2	100.0%	100.0%	Renal tubular acidosis with deafness, 267300
ATP6V1E1	66,5	92.3%	85.9%	Cutis laxa, autosomal recessive, type IIC, 617402
ATP7B	128,7	99.9%	99.1%	Wilson disease, 277900
ATP8A2	115,2	99.9%	99.5%	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268
ATP8B1	114	97.5%	94.6%	Cholestasis, benign recurrent intrahepatic, 243300 Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, progressive familial intrahepatic 1, 211600
ATPAF2	103,5	100.0%	100.0%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
ATR	144,6	99.8%	98.6%	?Cutaneous telangiectasia and cancer syndrome, familial, 614564 Seckel syndrome 1, 210600
AUH	127	100.0%	99.7%	3-methylglutaconic aciduria, type I, 250950
AURKC	69,1	99.7%	94.3%	Spermatogenic failure 5, 243060
B2M	198,9	100.0%	99.8%	?Amyloidosis, familial visceral, 105200 Immunodeficiency 43, 241600

B3GALNT2	93,9	92.9%	91.0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B3GALT6	81,7	82.6%	77.6%	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B3GAT3	121	99.6%	96.5%	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B3GLCT	96,6	99.7%	99.1%	Peters-plus syndrome, 261540
B4GALNT1	151,2	99.8%	97.9%	Spastic paraplegia 26, autosomal recessive, 609195
B4GALT1	112,2	99.8%	97.7%	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	123,9	99.8%	98.1%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
B4GAT1	136,9	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
B9D1	103,7	92.2%	92.1%	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120
B9D2	105,8	100.0%	100.0%	?Meckel syndrome 10, 614175 Joubert syndrome 34, 614175
BAAT	103	99.8%	97.3%	Hypercholanemia, familial, 607748
BANF1	51,1	96.6%	84.1%	Nestor-Guillermo progeria syndrome, 614008
BBIP1	119,7	97.3%	90.3%	?Bardet-Biedl syndrome 18, 615995
BBS1	146,4	100.0%	100.0%	Bardet-Biedl syndrome 1, 209900
BBS10	158,1	100.0%	99.9%	Bardet-Biedl syndrome 10, 615987
BBS12	187,1	100.0%	100.0%	Bardet-Biedl syndrome 12, 615989
BBS2	150,7	99.9%	99.6%	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	110,2	99.9%	99.2%	Bardet-Biedl syndrome 4, 615982
BBS5	98,5	98.0%	93.3%	Bardet-Biedl syndrome 5, 615983
BBS7	142,9	99.1%	96.5%	Bardet-Biedl syndrome 7, 615984
BBS9	113,6	98.6%	94.4%	Bardet-Biedl syndrome 9, 615986
BCKDHA	176,9	100.0%	99.8%	Maple syrup urine disease, type Ia, 248600
BCKDHB	123,3	98.6%	92.8%	Maple syrup urine disease, type Ib, 248600
BCKDK	203,5	100.0%	100.0%	Branched-chain ketoacid dehydrogenase kinase deficiency, 614923
BCL10	126,9	100.0%	100.0%	?Immunodeficiency 37, 616098 Lymphoma, MALT, somatic, 137245 {Lymphoma, follicular, somatic}, 605027 {Male germ cell tumor, somatic}, 273300 {Mesothelioma, somatic}, 156240 {Sezary syndrome, somatic}, 0
BCS1L	147,9	100.0%	100.0%	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000

				Mitochondrial complex III deficiency, nuclear type 1, 124000
BFSP1	100,1	100.0%	99.1%	Cataract 33, multiple types, 611391
BFSP2	99,3	99.9%	98.2%	Cataract 12, multiple types, 611597
BHLHA9	33,5	94.5%	73.0%	?Camptosynpolydactyly, complex, 607539 Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432
BIN1	113,4	99.9%	98.4%	Centronuclear myopathy 2, 255200
BLM	111	99.6%	98.0%	Bloom syndrome, 210900
BLNK	91,4	97.0%	93.1%	?Agammaglobulinemia 4, 613502
BLOC1S3	67,4	100.0%	99.9%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	101,1	99.2%	95.1%	?Hermansky-pudlak syndrome 9, 614171
BLVRA	111,9	100.0%	99.9%	Hyperbiliverdinemia, 614156
BMP1	152,7	100.0%	100.0%	Osteogenesis imperfecta, type XIII, 614856
BMPER	127,1	100.0%	99.5%	Diaphanospondylodysostosis, 608022
BMPR1B	139,4	100.0%	100.0%	Acromesomelic dysplasia, Demirhan type, 609441 Brachydactyly, type A1, D, 616849 Brachydactyly, type A2, 112600
BOLA3	48,1	99.9%	92.5%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BPGM	101,3	100.0%	100.0%	Erythrocytosis, familial, 8, 222800
BRAT1	142	100.0%	99.3%	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056 Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BRCA1	161,4	99.1%	98.1%	Fanconi anemia, complementation group S, 617883 {Breast-ovarian cancer, familial, 1}, 604370 {Pancreatic cancer, susceptibility to, 4}, 614320
BRCA2	106,2	99.6%	98.7%	Fanconi anemia, complementation group D1, 605724 Wilms tumor, 194070 {Breast cancer, male, susceptibility to}, 114480 {Breast-ovarian cancer, familial, 2}, 612555 {Glioblastoma 3}, 613029 {Medulloblastoma}, 155255 {Pancreatic cancer 2}, 613347 {Prostate cancer}, 176807
BRF1	109	99.8%	98.1%	Cerebellofaciodental syndrome, 616202
BSCL2	105,2	100.0%	100.0%	Encephalopathy, progressive, with or without lipodystrophy, 615924 Lipodystrophy, congenital generalized, type 2, 269700 Neuropathy, distal hereditary motor, type VA, 600794 Silver spastic paraplegia syndrome, 270685
BSND	139,4	100.0%	99.9%	Bartter syndrome, type 4a, 602522 Sensorineural deafness with mild renal dysfunction, 602522

BTD	126,6	99.9%	99.7%	Biotinidase deficiency, 253260
BUB1B	122	99.8%	98.7%	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430
BVES	112,6	99.7%	98.5%	Muscular dystrophy, limb-girdle, autosomal recessive 25, 616812
C12orf57	145,6	100.0%	100.0%	Temtamy syndrome, 218340
C12orf65	110,4	100.0%	99.6%	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035
C15orf41	122	100.0%	99.6%	Dyserythropoietic anemia, congenital, type Ib, 615631
C19orf12	104,2	100.0%	99.8%	?Spastic paraplegia 43, autosomal recessive, 615043 Neurodegeneration with brain iron accumulation 4, 614298
C1QA	196,1	100.0%	100.0%	C1q deficiency, 613652
C1QB	161,5	100.0%	100.0%	C1q deficiency, 613652
C1QBP	66,7	91.9%	79.5%	Combined oxidative phosphorylation deficiency 33, 617713
C1QC	187	100.0%	99.6%	C1q deficiency, 613652
C1S	96,8	99.8%	97.8%	C1s deficiency, 613783 Ehlers-Danlos syndrome, periodontal type, 2, 617174
C2	126,3	100.0%	100.0%	C2 deficiency, 217000 {Macular degeneration, age-related, 14, reduced risk of}, 615489
C21orf2	NC	NC	NC	Retinal dystrophy with macular staphyloma, 617547 Spondylometaphyseal dysplasia, axial, 602271
C21orf59	NC	NC	NC	Ciliary dyskinesia, primary, 26, 615500
C2CD3	116,9	95.8%	95.2%	Orofaciodigital syndrome XIV, 615948
C2orf71	NC	NC	NC	Retinitis pigmentosa 54, 613428
C3	141,6	100.0%	99.4%	C3 deficiency, 613779 {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 {Macular degeneration, age-related, 9}, 611378
C4A	86,4	98.5%	96.1%	C4a deficiency, 614380 [Blood group, Rodgers], 614374
C4B	83	98.7%	96.6%	C4B deficiency, 614379
C4orf26	NC	NC	NC	Amelogenesis imperfecta, type IIA4, 614832
C5	120,9	99.5%	97.7%	C5 deficiency, 609536 [Eculizumab, poor response to], 615749
C5orf42	NC	NC	NC	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
C8A	105,6	100.0%	99.4%	C8 deficiency, type I, 613790
C8B	105,6	99.9%	98.7%	C8 deficiency, type II, 613789

C8orf37	144,1	99.8%	99.4%	Bardet-Biedl syndrome 21, 617406 Cone-rod dystrophy 16, 614500 Retinitis pigmentosa 64, 614500
C9	121	99.9%	99.3%	C9 deficiency, 613825 {Macular degeneration, age-related, 15, susceptibility to}, 615591
CA12	100,7	100.0%	100.0%	Hyperchlorhidrosis, isolated, 143860
CA2	137,4	100.0%	99.9%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA5A	93,2	99.6%	95.7%	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CA8	107,5	99.7%	97.6%	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CABP2	76,6	78.5%	71.0%	Deafness, autosomal recessive 93, 614899
CABP4	148,3	100.0%	100.0%	Cone-rod synaptic disorder, congenital nonprogressive, 610427
CACNA1D	127,4	98.0%	97.7%	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896
CACNA2D4	95,9	99.0%	97.1%	Retinal cone dystrophy 4, 610478
CAD	136,7	99.9%	99.2%	Epileptic encephalopathy, early infantile, 50, 616457
CANT1	144,9	100.0%	100.0%	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719
CAPN1	162,5	100.0%	100.0%	Spastic paraplegia 76, autosomal recessive, 616907
CAPN10	116,5	100.0%	99.8%	{Diabetes mellitus, noninsulin-dependent 1}, 601283
CAPN3	98,3	99.2%	97.0%	Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129 Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600
CARD11	138,2	100.0%	99.6%	B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11A, 615206 Immunodeficiency 11B with atopic dermatitis, 617638
CARD9	133,2	100.0%	99.5%	Candidiasis, familial, 2, autosomal recessive, 212050
CARS2	128,2	100.0%	100.0%	Combined oxidative phosphorylation deficiency 27, 616672
CASP14	85,8	100.0%	99.9%	Ichthyosis, congenital, autosomal recessive 12, 617320
CASP8	128,1	95.6%	95.2%	?Autoimmune lymphoproliferative syndrome, type IIB, 607271 Hepatocellular carcinoma, somatic, 114550 {Breast cancer, protection against}, 114480 {Lung cancer, protection against}, 211980
CASQ2	113,5	100.0%	99.1%	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CASR	154,3	100.0%	99.7%	Hyperparathyroidism, neonatal, 239200 Hypocalcemia, autosomal dominant, 601198 Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 Hypocalciuric hypercalcemia, type I, 145980 {Epilepsy idiopathic generalized, susceptibility to, 8}, 612899

CAST	112,4	99.8%	97.1%	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295
CAT	136,9	100.0%	100.0%	Acatalasemia, 614097
CATSPER1	115,6	100.0%	99.6%	Spermatogenic failure 7, 612997
CAV1	189,3	100.0%	100.0%	?Lipodystrophy, congenital generalized, type 3, 612526 Lipodystrophy, familial partial, type 7, 606721 Pulmonary hypertension, primary, 3, 615343
CAVIN1	174,1	100.0%	100.0%	Lipodystrophy, congenital generalized, type 4, 613327
CBS	123,3	99.9%	99.0%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CBX2	149,7	100.0%	100.0%	?46XY sex reversal 5, 613080
CC2D1A	135	100.0%	99.5%	Mental retardation, autosomal recessive 3, 608443
CC2D2A	111,7	99.0%	97.1%	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284
CCBE1	75,3	99.8%	99.1%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CCDC103	116,2	100.0%	99.8%	Ciliary dyskinesia, primary, 17, 614679
CCDC114	134,1	100.0%	99.8%	Ciliary dyskinesia, primary, 20, 615067
CCDC115	77,9	88.9%	87.1%	Congenital disorder of glycosylation, type IIo, 616828
CCDC151	127,2	100.0%	100.0%	Ciliary dyskinesia, primary, 30, 616037
CCDC174	120,5	99.7%	97.5%	Hypotonia, infantile, with psychomotor retardation, 616816
CCDC39	86,7	99.4%	96.8%	Ciliary dyskinesia, primary, 14, 613807
CCDC40	112	99.4%	98.4%	Ciliary dyskinesia, primary, 15, 613808
CCDC65	80,3	99.6%	97.1%	Ciliary dyskinesia, primary, 27, 615504
CCDC8	186,9	100.0%	100.0%	3-M syndrome 3, 614205
CCDC88A	92,8	99.3%	96.7%	?PEHO syndrome-like, 617507
CCDC88C	108,2	100.0%	99.4%	?Spinocerebellar ataxia 40, 616053 Hydrocephalus, congenital, 1, 236600
CCNO	130,9	100.0%	99.8%	Ciliary dyskinesia, primary, 29, 615872
CCT5	117,9	99.9%	98.9%	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CD151	122,2	100.0%	100.0%	Nephropathy with pretibial epidermolysis bullosa and deafness, 609057 [Blood group, Raph], 179620
CD19	108,8	100.0%	99.9%	Immunodeficiency, common variable, 3, 613493
CD247	95,5	100.0%	99.5%	?Immunodeficiency 25, 610163
CD27	105,3	100.0%	100.0%	Lymphoproliferative syndrome 2, 615122
CD2AP	121,7	99.7%	98.3%	Glomerulosclerosis, focal segmental, 3, 607832
CD320	113,9	100.0%	99.9%	Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646
CD3D	138,9	100.0%	99.9%	Immunodeficiency 19, 615617

CD3E	125,9	100.0%	98.9%	Immunodeficiency 18, 615615 Immunodeficiency 18, SCID variant, 615615
CD3G	136,6	100.0%	100.0%	Immunodeficiency 17, CD3 gamma deficient, 615607
CD40	147,8	100.0%	99.9%	Immunodeficiency with hyper-IgM, type 3, 606843
CD59	149,6	93.5%	85.8%	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD79A	133,5	100.0%	99.3%	Agammaglobulinemia 3, 613501
CD79B	194,3	100.0%	100.0%	Agammaglobulinemia 6, 612692
CD81	158,3	100.0%	100.0%	Immunodeficiency, common variable, 6, 613496
CD8A	150,8	100.0%	99.9%	CD8 deficiency, familial, 608957
CDAN1	112,4	100.0%	99.6%	Dyserythropoietic anemia, congenital, type Ia, 224120
CDC14A	150,1	99.6%	97.4%	Deafness, autosomal recessive 32, with or without immotile sperm, 608653
CDC45	138,9	99.6%	98.1%	Meier-Gorlin syndrome 7, 617063
CDC6	139,4	99.9%	99.8%	?Meier-Gorlin syndrome 5, 613805
CDCA7	112	100.0%	99.5%	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910
CDH11	126,5	100.0%	100.0%	Elsahy-Waters syndrome, 211380
CDH23	172,7	100.0%	100.0%	Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D, 601067 Usher syndrome, type 1D/F digenic, 601067 {Pituitary adenoma 5, multiple types}, 617540
CDH3	140,5	100.0%	99.8%	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 Hypotrichosis, congenital, with juvenile macular dystrophy, 601553
CDHR1	143,6	99.9%	99.0%	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660
CDK10	131,3	100.0%	100.0%	Al Kaissi syndrome, 617694
CDK5	107,2	100.0%	99.8%	?Lissencephaly 7 with cerebellar hypoplasia, 616342
CDK5RAP2	107	99.8%	98.8%	Microcephaly 3, primary, autosomal recessive, 604804
CDK6	110	99.6%	97.4%	?Microcephaly 12, primary, autosomal recessive, 616080
CDSN	131	100.0%	100.0%	Hypotrichosis 2, 146520 Peeling skin syndrome 1, 270300
CDT1	130,9	100.0%	99.9%	Meier-Gorlin syndrome 4, 613804
CEBPE	99,1	100.0%	99.9%	Specific granule deficiency, 245480
CENPE	76,6	98.5%	93.2%	?Microcephaly 13, primary, autosomal recessive, 616051
CENPF	139,9	99.8%	98.7%	Stromme syndrome, 243605
CENPJ	136	99.9%	99.2%	?Seckel syndrome 4, 613676 Microcephaly 6, primary, autosomal recessive, 608393
CEP104	104	99.3%	97.5%	Joubert syndrome 25, 616781
CEP120	131,7	100.0%	99.4%	Joubert syndrome 31, 617761

				Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP135	89,9	98.8%	92.6%	Microcephaly 8, primary, autosomal recessive, 614673
CEP152	144,4	99.7%	98.2%	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823
CEP164	89,3	99.8%	98.0%	Nephronophthisis 15, 614845
CEP19	181,7	100.0%	100.0%	Morbid obesity and spermatogenic failure, 615703
CEP290	82,6	97.3%	91.7%	?Bardet-Biedl syndrome 14, 615991 Joubert syndrome 5, 610188 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Senior-Loken syndrome 6, 610189
CEP41	78,2	98.9%	94.4%	Joubert syndrome 15, 614464
CEP55	124,5	100.0%	100.0%	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500
CEP57	85,1	98.7%	91.6%	Mosaic variegated aneuploidy syndrome 2, 614114
CEP63	120,8	98.2%	94.7%	?Seckel syndrome 6, 614728
CEP78	120,1	99.7%	97.6%	Cone-rod dystrophy and hearing loss, 617236
CEP83	108,8	99.4%	96.6%	Nephronophthisis 18, 615862
CEP89	127,5	97.4%	94.7%	No OMIM phenotype Complex IV deficiency,isolated (van Bon (2013) Hum Mol Genet 22,3138) ?Intellectual disability (Vulto-van Silfhout (2013) Hum Mutat 34,1679)
CERKL	114,1	99.4%	97.2%	Retinitis pigmentosa 26, 608380
CERS1	70	92.6%	81.4%	?Epilepsy, progressive myoclonic, 8, 616230
CERS3	95,2	99.8%	98.2%	Ichthyosis, congenital, autosomal recessive 9, 615023
CFAP53	131,8	99.1%	97.0%	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFD	113,8	96.9%	89.7%	Complement factor D deficiency, 613912
CFH	155,4	99.4%	97.9%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814 {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 {Macular degeneration, age-related, 4}, 610698
CFI	139	99.5%	97.0%	Complement factor I deficiency, 610984 {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 {Macular degeneration, age-related, 13, susceptibility to}, 615439
CFL2	119,2	100.0%	99.2%	Nemaline myopathy 7, autosomal recessive, 610687
CFTR	113,5	99.4%	97.4%	Congenital bilateral absence of vas deferens, 277180 Cystic fibrosis, 219700 Sweat chloride elevation without CF, 0 {Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400

				{Hypertrypsinemia, neonatal}, 0 {Pancreatitis, hereditary}, 167800
CHAT	117,1	95.4%	86.9%	Myasthenic syndrome, congenital, 6, presynaptic, 254210
CHKB	115,4	100.0%	100.0%	Muscular dystrophy, congenital, megaconial type, 602541
CHMP1A	123,2	100.0%	99.8%	Pontocerebellar hypoplasia, type 8, 614961
CHRM3	131	100.0%	100.0%	?Prune belly syndrome, 100100
CHRNA1	92,6	94.6%	93.3%	Multiple pterygium syndrome, lethal type, 253290 Myasthenic syndrome, congenital, 1A, slow-channel, 601462 Myasthenic syndrome, congenital, 1B, fast-channel, 608930
CHRNB1	128,2	100.0%	99.7%	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314 Myasthenic syndrome, congenital, 2A, slow-channel, 616313
CHRND	140,4	99.8%	98.0%	?Myasthenic syndrome, congenital, 3A, slow-channel, 616321 ?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323 Multiple pterygium syndrome, lethal type, 253290 Myasthenic syndrome, congenital, 3B, fast-channel, 616322
CHRNE	167,8	100.0%	100.0%	Myasthenic syndrome, congenital, 4A, slow-channel, 605809 Myasthenic syndrome, congenital, 4B, fast-channel, 616324 Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931
CHRNG	141,3	100.0%	100.0%	Escobar syndrome, 265000 Multiple pterygium syndrome, lethal type, 253290
CHST14	160,6	99.9%	98.9%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	133,8	100.0%	100.0%	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHST6	279,8	100.0%	100.0%	Macular corneal dystrophy, 217800
CHST8	257,9	100.0%	100.0%	?Peeling skin syndrome 3, 616265
CHSY1	125,9	99.3%	97.9%	Temtamy preaxial brachydactyly syndrome, 605282
CHUK	126,5	99.8%	99.2%	Cocoon syndrome, 613630
CIB2	198	99.9%	99.4%	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CIDEC	83,6	99.9%	96.4%	?Lipodystrophy, familial partial, type 5, 615238
CIITA	148,8	100.0%	99.9%	Bare lymphocyte syndrome, type II, complementation group A, 209920 {Rheumatoid arthritis, susceptibility to}, 180300
CISD2	113,6	83.4%	83.3%	Wolfram syndrome 2, 604928
CIT	101,4	99.9%	98.5%	Microcephaly 17, primary, autosomal recessive, 617090
CKAP2L	156	99.9%	98.9%	Filippi syndrome, 272440
CLCF1	87,2	100.0%	99.6%	Cold-induced sweating syndrome 2, 610313
CLCN1	125,1	100.0%	99.8%	Myotonia congenita, dominant, 160800 Myotonia congenita, recessive, 255700

				Myotonia levior, recessive, 0
CLCN2	115,1	100.0%	99.7%	Hyperaldosteronism, familial, type II, 605635 Leukoencephalopathy with ataxia, 615651 {Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628 {Epilepsy, juvenile absence, susceptibility to, 2}, 607628 {Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628
CLCN7	146,7	99.8%	98.7%	Osteopetrosis, autosomal dominant 2, 166600 Osteopetrosis, autosomal recessive 4, 611490
CLCNKB	99,4	99.7%	97.1%	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090
CLDN1	122,5	100.0%	100.0%	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626
CLDN10	138,2	100.0%	100.0%	HELIX syndrome, 617671
CLDN14	111,7	100.0%	99.9%	Deafness, autosomal recessive 29, 614035
CLDN16	126,6	100.0%	100.0%	Hypomagnesemia 3, renal, 248250
CLDN19	125,4	99.1%	95.1%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLIC5	97,6	100.0%	99.9%	?Deafness, autosomal recessive 103, 616042
CLIP1	118,1	99.9%	98.8%	No OMIM phenotype Intellectual disability, autosomal recessive (Larti (2015) Eur J Hum Genet 23,331)
CLMP	86,2	100.0%	99.6%	Congenital short bowel syndrome, 615237
CLN3	114,7	92.6%	91.9%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	138,7	99.9%	98.8%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	132,3	100.0%	99.9%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	144,5	83.5%	83.5%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLP1	135,7	100.0%	100.0%	Pontocerebellar hypoplasia, type 10, 615803
CLPB	125,6	99.8%	97.9%	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CLPP	139,3	100.0%	99.2%	Perrault syndrome 3, 614129
CLRN1	135,3	100.0%	99.4%	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902
CNGA1	110,4	92.5%	86.5%	Retinitis pigmentosa 49, 613756
CNGA3	149,6	100.0%	99.7%	Achromatopsia 2, 216900
CNGB1	107,4	99.5%	98.0%	Retinitis pigmentosa 45, 613767
CNGB3	101,5	98.6%	93.9%	Achromatopsia 3, 262300 Macular degeneration, juvenile, 248200
CNNM2	199,8	100.0%	100.0%	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418

CNNM4	161,6	100.0%	99.5%	Jalili syndrome, 217080
CNPY3	78,4	100.0%	100.0%	Epileptic encephalopathy, early infantile, 60, 617929
CNTN1	128	99.7%	98.7%	?Myopathy, congenital, Compton-North, 612540
CNTN2	123,7	92.7%	92.7%	?Epilepsy, myoclonic, familial adult, 5, 615400
CNTNAP1	157,2	99.9%	99.1%	Hypomyelinating neuropathy, congenital, 3, 618186 Lethal congenital contracture syndrome 7, 616286
CNTNAP2	127,1	100.0%	99.8%	Cortical dysplasia-focal epilepsy syndrome, 610042 Pitt-Hopkins like syndrome 1, 610042 {Autism susceptibility 15}, 612100
COA5	74,9	86.6%	83.5%	?Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 3, 616500
COA6	112,5	99.4%	97.0%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 4, 616501
COA7	122	100.0%	100.0%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387
COASY	172,7	100.0%	100.0%	Neurodegeneration with brain iron accumulation 6, 615643 Pontocerebellar hypoplasia, type 12, 618266
COG1	108,4	100.0%	99.9%	Congenital disorder of glycosylation, type IIg, 611209
COG2	124,6	99.7%	98.1%	?Congenital disorder of glycosylation, type IIq, 617395
COG4	94,5	100.0%	99.6%	Congenital disorder of glycosylation, type IIj, 613489 Saul-Wilson syndrome, 618150
COG5	126,3	99.9%	98.4%	Congenital disorder of glycosylation, type IIi, 613612
COG6	90,4	99.1%	96.0%	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328
COG7	106,1	100.0%	99.7%	Congenital disorder of glycosylation, type IIe, 608779
COG8	145	100.0%	98.5%	Congenital disorder of glycosylation, type IIh, 611182
COL11A1	96,6	97.9%	94.0%	Fibrochondrogenesis 1, 228520 Marshall syndrome, 154780 Stickler syndrome, type II, 604841 {Lumbar disc herniation, susceptibility to}, 603932
COL11A2	111,6	100.0%	99.4%	Deafness, autosomal dominant 13, 601868 Deafness, autosomal recessive 53, 609706 Fibrochondrogenesis 2, 614524 Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840 Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150
COL12A1	124,2	99.8%	99.1%	?Ullrich congenital muscular dystrophy 2, 616470 Bethlem myopathy 2, 616471
COL13A1	89,8	100.0%	99.3%	Myasthenic syndrome, congenital, 19, 616720
COL17A1	104,5	99.3%	96.9%	Epidermolysis bullosa, junctional, localisata variant, 226650 Epidermolysis bullosa, junctional, non-Herlitz type, 226650

				Epithelial recurrent erosion dystrophy, 122400
COL18A1	133,5	99.5%	96.9%	Knobloch syndrome, type 1, 267750
COL1A2	93,3	98.5%	94.6%	Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821 Ehlers-Danlos syndrome, cardiac valvular type, 225320 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type III, 259420 Osteogenesis imperfecta, type IV, 166220 {Osteoporosis, postmenopausal}, 166710
COL25A1	131,9	99.5%	98.9%	Fibrosis of extraocular muscles, congenital, 5, 616219
COL27A1	142,2	99.9%	99.2%	Steel syndrome, 615155
COL4A3	90,5	99.6%	97.7%	Alport syndrome 2, autosomal recessive, 203780 Alport syndrome 3, autosomal dominant, 104200 Hematuria, benign familial, 141200
COL4A4	92,4	99.8%	97.4%	Alport syndrome 2, autosomal recessive, 203780 Hematuria, familial benign, 141200
COL6A1	158,8	100.0%	99.8%	Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090
COL6A2	175,3	100.0%	99.8%	?Myosclerosis, congenital, 255600 Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090
COL6A3	154	100.0%	99.8%	Bethlem myopathy 1, 158810 Dystonia 27, 616411 Ullrich congenital muscular dystrophy 1, 254090
COL7A1	139,7	99.8%	98.9%	EBD inversa, 226600 EBD, Bart type, 132000 EBD, localisata variant, 0 Epidermolysis bullosa dystrophica, AD, 131750 Epidermolysis bullosa dystrophica, AR, 226600 Epidermolysis bullosa pruriginosa, 604129 Epidermolysis bullosa, pretibial, 131850 Toenail dystrophy, isolated, 607523 Transient bullous of the newborn, 131705
COL9A1	132,3	100.0%	99.7%	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	95,2	99.9%	98.8%	?Stickler syndrome, type V, 614284 Epiphyseal dysplasia, multiple, 2, 600204
COLEC10	120,1	100.0%	99.9%	3MC syndrome 3, 248340
COLEC11	180,6	100.0%	100.0%	3MC syndrome 2, 265050

COLQ	100,9	99.8%	97.5%	Myasthenic syndrome, congenital, 5, 603034
COPB2	139	99.8%	99.0%	?Microcephaly 19, primary, autosomal recessive, 617800
COQ2	103,5	97.6%	97.1%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ4	105	91.3%	90.2%	Coenzyme Q10 deficiency, primary, 7, 616276
COQ6	127,5	99.9%	98.6%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	138,3	99.9%	99.6%	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ8A	161,8	100.0%	99.9%	Coenzyme Q10 deficiency, primary, 4, 612016
COQ8B	99,5	100.0%	99.8%	Nephrotic syndrome, type 9, 615573
COQ9	73,8	100.0%	98.1%	Coenzyme Q10 deficiency, primary, 5, 614654
CORO1A	150,8	99.9%	98.5%	Immunodeficiency 8, 615401
COX10	220,4	100.0%	99.9%	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 Mitochondrial complex IV deficiency, 220110
COX14	95,2	100.0%	100.0%	?Mitochondrial complex IV deficiency, 220110
COX15	87,7	99.9%	98.3%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000
COX20	66,2	96.4%	85.3%	Mitochondrial complex IV deficiency, 220110
COX4I2	116,5	100.0%	99.6%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX6A1	148,3	100.0%	99.9%	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
COX6B1	139,1	100.0%	100.0%	Mitochondrial complex IV deficiency, 220110
COX8A	109,4	100.0%	100.0%	?Mitochondrial complex IV deficiency, 220110
CP	100,6	93.1%	87.4%	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 [Hypoceruloplasminemia, hereditary], 604290
CPA6	109,1	99.4%	96.9%	Epilepsy, familial temporal lobe, 5, 614417 Febrile seizures, familial, 11, 614418
CPLX1	102,3	100.0%	100.0%	Epileptic encephalopathy, early infantile, 63, 617976
CPN1	97,9	99.8%	97.8%	Carboxypeptidase N deficiency, 212070
CPOX	134,1	99.5%	97.2%	Coproporphyria, 121300 Harderoporphyria, 121300
CPS1	133,8	100.0%	99.9%	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}, 0
CPT1A	131,5	99.9%	98.4%	CPT deficiency, hepatic, type IA, 255120
CPT2	139,2	98.3%	98.2%	CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 CPT II deficiency, myopathic, stress-induced, 255110

				{Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212
CR2	131,7	100.0%	99.9%	Immunodeficiency, common variable, 7, 614699 {Systemic lupus erythematosus, susceptibility to, 9}, 610927
CRADD	113,2	100.0%	98.2%	Mental retardation, autosomal recessive 34, with variant lissencephaly, 614499
CRAT	115,5	100.0%	99.9%	?Neurodegeneration with brain iron accumulation 8, 617917
CRB1	155	100.0%	100.0%	Leber congenital amaurosis 8, 613835 Pigmented paravenous chorioretinal atrophy, 172870 Retinitis pigmentosa-12, 600105
CRB2	116	99.8%	98.5%	Focal segmental glomerulosclerosis 9, 616220 Ventriculomegaly with cystic kidney disease, 219730
CRBN	125,2	88.1%	87.3%	Mental retardation, autosomal recessive 2, 607417
CREB3L1	135,8	100.0%	99.8%	Osteogenesis imperfecta, type XVI, 616229
CRIPT	42,6	98.7%	91.2%	Short stature with microcephaly and distinctive facies, 615789
CRLF1	126,4	93.2%	90.7%	Cold-induced sweating syndrome 1, 272430
CRTAP	120,2	100.0%	99.1%	Osteogenesis imperfecta, type VII, 610682
CRYAA	131	95.8%	90.7%	Cataract 9, multiple types, 604219
CRYAB	94	99.7%	96.8%	Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, 2, 608810 Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869
CRYBB1	125,2	100.0%	99.8%	Cataract 17, multiple types, 611544
CRYBB3	139,5	100.0%	100.0%	Cataract 22, 609741
CSF2RB	123,2	99.8%	98.4%	Surfactant metabolism dysfunction, pulmonary, 5, 614370
CSPP1	119	100.0%	99.1%	Joubert syndrome 21, 615636
CSTA	110,9	99.9%	99.1%	Peeling skin syndrome 4, 607936
CSTB	70	99.3%	90.9%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTC1	105,5	100.0%	99.3%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTDP1	128,1	95.1%	88.0%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTNS	112,6	100.0%	99.5%	Cystinosis, atypical nephropathic, 219800 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750
CTPS1	108,8	100.0%	99.9%	Immunodeficiency 24, 615897
CTSA	132,9	100.0%	99.9%	Galactosialidosis, 256540
CTSC	116,2	100.0%	100.0%	Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000 Periodontitis 1, juvenile, 170650

CTSD	171	99.8%	97.8%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	107	91.3%	81.8%	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362
CTSK	86,3	100.0%	99.8%	Pycnodysostosis, 265800
CUBN	103,2	99.6%	97.6%	Megaloblastic anemia-1, Finnish type, 261100
CUL7	129,2	100.0%	99.8%	3-M syndrome 1, 273750
CWC27	84,5	99.8%	97.5%	Retinitis pigmentosa with or without skeletal anomalies, 250410
CWF19L1	103,2	99.9%	99.3%	Spinocerebellar ataxia, autosomal recessive 17, 616127
CYB5A	132,5	100.0%	100.0%	Methemoglobinemia and ambiguous genitalia, 250790
CYB5R3	152,1	99.2%	98.3%	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYBA	110,5	96.7%	86.9%	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690
CYC1	152,2	99.3%	95.6%	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYP11A1	121,2	99.2%	95.0%	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	155,9	100.0%	100.0%	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 Aldosteronism, glucocorticoid-remediable, 103900
CYP11B2	156	100.0%	100.0%	Aldosterone to renin ratio raised, 0 Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 {Low renin hypertension, susceptibility to}, 0
CYP17A1	108,5	100.0%	99.6%	17,20-lyase deficiency, isolated, 202110 17-alpha-hydroxylase/17,20-lyase deficiency, 202110
CYP19A1	125,7	99.4%	97.3%	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300
CYP1B1	134,4	100.0%	100.0%	Anterior segment dysgenesis 6, multiple subtypes, 617315 Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300
CYP24A1	169,1	100.0%	100.0%	Hypercalcemia, infantile, 1, 143880
CYP26B1	168,7	100.0%	100.0%	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
CYP26C1	133	100.0%	99.9%	Focal facial dermal dysplasia 4, 614974
CYP27A1	173	100.0%	99.7%	Cerebrotendinous xanthomatosis, 213700
CYP27B1	147,1	100.0%	99.7%	Vitamin D-dependent rickets, type I, 264700
CYP2C8	90,3	98.7%	95.3%	{Drug metabolism, altered, CYP2C8-related}, 618018
CYP2R1	130,7	99.7%	96.8%	Rickets due to defect in vitamin D 25-hydroxylation, 600081
CYP2U1	134,3	98.4%	95.5%	Spastic paraplegia 56, autosomal recessive, 615030
CYP4F22	115,3	100.0%	98.8%	Ichthyosis, congenital, autosomal recessive 5, 604777
CYP4V2	137,2	99.9%	98.3%	Bietti crystalline corneoretinal dystrophy, 210370
CYP7B1	103,8	99.6%	96.6%	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, autosomal recessive, 270800

D2HGDH	142	100.0%	99.4%	D-2-hydroxyglutaric aciduria, 600721
DAG1	189	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818
DARS	125,4	99.9%	99.0%	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2	126,8	100.0%	98.6%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBH	145,6	100.0%	99.9%	Orthostatic hypotension 1, due to DBH deficiency, 223360
DBT	109,9	99.6%	96.9%	Maple syrup urine disease, type II, 248600
DCAF17	90,4	99.9%	97.9%	Woodhouse-Sakati syndrome, 241080
DCC	118,6	100.0%	99.8%	Colorectal cancer, somatic, 114500 Esophageal carcinoma, somatic, 133239 Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 Mirror movements 1 and/or agenesis of the corpus callosum, 157600
DCDC2	150	99.9%	99.8%	?Deafness, autosomal recessive 66, 610212 Nephronophthisis 19, 616217 Sclerosing cholangitis, neonatal, 617394
DCHS1	149,4	100.0%	99.9%	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390
DCLRE1C	138,9	99.9%	97.2%	Omenn syndrome, 603554 Severe combined immunodeficiency, Athabascan type, 602450
DCPS	128	100.0%	99.6%	Al-Raqad syndrome, 616459
DDB2	147,3	99.8%	98.4%	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DDC	97,9	99.5%	95.0%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	161,6	99.9%	98.4%	Spastic paraplegia 28, autosomal recessive, 609340
DDHD2	129,6	100.0%	99.5%	Spastic paraplegia 54, autosomal recessive, 615033
DDOST	114	100.0%	99.8%	?Congenital disorder of glycosylation, type Ir, 614507
DDR2	115,8	100.0%	99.3%	Spondylometaepiphyseal dysplasia, short limb-hand type, 271665 Warburg-Cinotti syndrome, 618175
DDRGK1	100	100.0%	99.8%	Spondyloepimetaphyseal dysplasia, Shohat type, 602557
DDX11	100,7	86.7%	81.2%	Warsaw breakage syndrome, 613398
DDX59	141,5	100.0%	99.8%	Orofaciodigital syndrome V, 174300
DEAF1	113,4	99.8%	97.6%	?Dyskinesia, seizures, and intellectual developmental disorder, 617171 Mental retardation, autosomal dominant 24, 615828
DENND5A	99,2	99.8%	98.9%	Epileptic encephalopathy, early infantile, 49, 617281
DES	125	100.0%	100.0%	Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400
DFNB59	NC	NC	NC	Deafness, autosomal recessive 59, 610220

DGKE	127,8	99.8%	98.3%	Nephrotic syndrome, type 7, 615008 {Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008
DGUOK	119,4	99.9%	97.9%	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880 Portal hypertension, noncirrhotic, 617068 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070
DHCR24	155,8	100.0%	99.9%	Desmosterolosis, 602398
DHCR7	144,9	100.0%	100.0%	Smith-Lemli-Opitz syndrome, 270400
DHDDS	81	97.1%	93.8%	?Congenital disorder of glycosylation, type 1bb, 613861 Developmental delay and seizures with or without movement abnormalities, 617836 Retinitis pigmentosa 59, 613861
DHFR	50	94.1%	83.1%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHH	164,9	100.0%	100.0%	46XY partial gonadal dysgenesis, with minifascicular neuropathy, 607080 46XY sex reversal 7, 233420
DHODH	98,8	100.0%	100.0%	Miller syndrome, 263750
DHTKD1	122,4	99.9%	98.8%	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DIAPH1	101,7	99.9%	99.6%	Deafness, autosomal dominant 1, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632
DIS3L2	143,3	100.0%	99.8%	Perlman syndrome, 267000
DLAT	100,2	99.8%	99.2%	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	119,2	99.9%	99.7%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLL3	108,5	96.7%	92.5%	Spondylocostal dysostosis 1, autosomal recessive, 277300
DLX5	145,3	100.0%	99.8%	?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600
DMGDH	134,7	100.0%	99.8%	Dimethylglycine dehydrogenase deficiency, 605850
DMP1	133	100.0%	99.9%	Hypophosphatemic rickets, AR, 241520
DMXL2	154	99.8%	98.9%	?Deafness, autosomal dominant 71, 617605 ?Polyendocrine-polyneuropathy syndrome, 616113
DNA2	124,3	99.7%	97.3%	?Seckel syndrome 8, 615807 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156
DNAAF1	112,8	100.0%	99.5%	Ciliary dyskinesia, primary, 13, 613193
DNAAF2	150,1	99.9%	98.7%	Ciliary dyskinesia, primary, 10, 612518
DNAAF3	115	99.9%	98.5%	Ciliary dyskinesia, primary, 2, 606763
DNAAF4	94,1	99.9%	98.2%	Ciliary dyskinesia, primary, 25, 615482 {Dyslexia, susceptibility to, 1}, 127700
DNAAF5	107,6	95.4%	85.2%	Ciliary dyskinesia, primary, 18, 614874
DNAH11	131,3	99.8%	98.7%	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH5	114,1	99.9%	99.1%	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644

DNAI1	115,4	100.0%	100.0%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	151,7	99.6%	96.6%	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJB2	118,4	100.0%	100.0%	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881
DNAJC12	140,7	87.4%	87.4%	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC19	93,8	98.4%	92.3%	3-methylglutaconic aciduria, type V, 610198
DNAJC21	128,1	99.9%	99.5%	Bone marrow failure syndrome 3, 617052
DNAJC3	137,4	100.0%	99.7%	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192
DNAJC6	126,5	99.9%	99.0%	Parkinson disease 19a, juvenile-onset, 615528 Parkinson disease 19b, early-onset, 615528
DNAL1	104	99.3%	96.2%	Ciliary dyskinesia, primary, 16, 614017
DNASE1L3	113	100.0%	99.7%	Systemic lupus erythematosus 16, 614420
DNM1L	119,5	99.9%	98.7%	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 Optic atrophy 5, 610708
DNM2	123,9	99.7%	96.7%	Centronuclear myopathy 1, 160150 Charcot-Marie-Tooth disease, axonal type 2M, 606482 Charcot-Marie-Tooth disease, dominant intermediate B, 606482 Lethal congenital contracture syndrome 5, 615368
DNMT3B	116,4	100.0%	99.9%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK2	119,5	100.0%	99.5%	Immunodeficiency 40, 616433
DOCK6	121,5	99.6%	98.6%	Adams-Oliver syndrome 2, 614219
DOCK7	120,7	99.6%	97.8%	Epileptic encephalopathy, early infantile, 23, 615859
DOCK8	112,1	100.0%	99.6%	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
DOK7	135,1	94.0%	93.3%	?Fetal akinesia deformation sequence 3, 618389 Myasthenic syndrome, congenital, 10, 254300
DOLK	157,2	100.0%	100.0%	Congenital disorder of glycosylation, type Im, 610768
DONSON	90,2	99.0%	92.4%	Microcephaly, short stature, and limb abnormalities, 617604 Microcephaly-micromelia syndrome, 251230
DPAGT1	87,5	100.0%	99.9%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPH1	161,6	100.0%	100.0%	Developmental delay with short stature, dysmorphic features, and sparse hair, 616901
DPM1	134,7	95.2%	88.2%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	88,5	99.8%	97.6%	Congenital disorder of glycosylation, type Iu, 615042
DPM3	200,5	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937
DPY19L2	85,9	74.4%	70.2%	Spermatogenic failure 9, 613958
DPYD	141,6	99.5%	96.4%	5-fluorouracil toxicity, 274270 Dihydropyrimidine dehydrogenase deficiency, 274270
DPYS	117,5	100.0%	99.8%	Dihydropyrimidinuria, 222748

DRAM2	130,4	100.0%	100.0%	Cone-rod dystrophy 21, 616502
DRC1	91,2	100.0%	99.3%	Ciliary dyskinesia, primary, 21, 615294
DSC2	123,7	99.6%	97.2%	Arrhythmogenic right ventricular dysplasia 11, 610476 Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476
DSC3	95,6	99.3%	97.3%	?Hypotrichosis and recurrent skin vesicles, 613102
DSE	89,7	99.7%	97.2%	Ehlers-Danlos syndrome, musculocontractural type 2, 615539
DSG4	158,3	99.8%	99.0%	Hypotrichosis 6, 607903
DSP	140,6	100.0%	99.6%	Arrhythmogenic right ventricular dysplasia 8, 607450 Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821 Epidermolysis bullosa, lethal acantholytic, 609638 Keratosis palmoplantaris striata II, 612908 Skin fragility-woolly hair syndrome, 607655
DST	144,8	99.9%	99.2%	?Neuropathy, hereditary sensory and autonomic, type VI, 614653 Epidermolysis bullosa simplex, autosomal recessive 2, 615425
DSTYK	121,7	99.6%	97.7%	Congenital anomalies of kidney and urinary tract 1, 610805 Spastic paraplegia 23, 270750
DTNBP1	113,4	99.8%	97.9%	Hermansky-Pudlak syndrome 7, 614076
DUOX2	128,8	99.5%	96.4%	Thyroid dyshormonogenesis 6, 607200
DUOXA2	140,5	100.0%	100.0%	Thyroid dyshormonogenesis 5, 274900
DYM	103,3	97.4%	95.5%	Dyggve-Melchior-Clausen disease, 223800 Smith-McCort dysplasia, 607326
DYNC2H1	102,2	98.8%	95.5%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYNC2LI1	95,4	99.7%	97.0%	Short-rib thoracic dysplasia 15 with polydactyly, 617088
DYSF	133,6	100.0%	99.9%	Miyoshi muscular dystrophy 1, 254130 Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601 Myopathy, distal, with anterior tibial onset, 606768
DZIP1L	96,7	99.7%	98.0%	Polycystic kidney disease 5, 617610
EARS2	99	99.7%	97.8%	Combined oxidative phosphorylation deficiency 12, 614924
ECEL1	107,1	100.0%	97.4%	Arthrogryposis, distal, type 5D, 615065
ECHS1	103,8	100.0%	99.7%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ECM1	158,6	99.9%	99.0%	Urbach-Wiethe disease, 247100
EDAR	126,6	100.0%	100.0%	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490 Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900 [Hair morphology 1, hair thickness], 612630
EDARADD	89,8	99.7%	98.3%	Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940 Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941

EDC3	113,7	100.0%	99.0%	?Mental retardation, autosomal recessive 50, 616460
EDN1	156,5	100.0%	99.9%	Auriculocondylar syndrome 3, 615706 Question mark ears, isolated, 612798 {High density lipoprotein cholesterol level QTL 7}, 0
EDN3	135,2	100.0%	100.0%	Central hypoventilation syndrome, congenital, 209880 Waardenburg syndrome, type 4B, 613265 {Hirschsprung disease, susceptibility to, 4}, 613712
EDNRB	120,9	96.9%	92.5%	ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580 {Hirschsprung disease, susceptibility to, 2}, 600155
EFEMP2	129,4	100.0%	100.0%	Cutis laxa, autosomal recessive, type IB, 614437
EFL1	150,5	99.5%	98.1%	Shwachman-Diamond syndrome 2, 617941
EGF	110,5	100.0%	99.7%	Hypomagnesemia 4, renal, 611718
EGFR	135,2	100.0%	100.0%	?Inflammatory skin and bowel disease, neonatal, 2, 616069 Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980 Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980 {Nonsmall cell lung cancer, susceptibility to}, 211980
EGR2	130	100.0%	100.0%	Charcot-Marie-Tooth disease, type 1D, 607678 Dejerine-Sottas disease, 145900 Hypomyelinating neuropathy, congenital, 1, 605253
EIF2AK3	134,2	99.5%	96.3%	Wolcott-Rallison syndrome, 226980
EIF2AK4	129,8	99.7%	98.6%	Pulmonary venoocclusive disease 2, 234810
EIF2B1	121,7	100.0%	99.9%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B2	109,7	99.4%	92.4%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B3	134,7	100.0%	100.0%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B4	122	100.0%	99.6%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B5	103,1	100.0%	99.6%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF4A3	87,5	100.0%	99.4%	Robin sequence with cleft mandible and limb anomalies, 268305
ELAC2	109,4	99.9%	99.0%	Combined oxidative phosphorylation deficiency 17, 615440 {Prostate cancer, hereditary, 2, susceptibility to}, 614731
ELMOD3	135,8	100.0%	99.9%	?Deafness, autosomal recessive 88, 615429
ELOVL4	104,4	99.9%	99.1%	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110
ELP1	118,7	99.7%	98.4%	Dysautonomia, familial, 223900

ELP2	120,6	99.8%	98.0%	Mental retardation, autosomal recessive 58, 617270
EMC1	105,7	100.0%	98.9%	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EMG1	123,2	100.0%	99.9%	Bowen-Conradi syndrome, 211180
EML1	129,9	100.0%	99.6%	Band heterotopia, 600348
EMP2	78,4	99.5%	96.1%	Nephrotic syndrome, type 10, 615861
ENAM	139,5	100.0%	100.0%	Amelogenesis imperfecta, type IB, 104500 Amelogenesis imperfecta, type IC, 204650
ENO3	173,2	100.0%	100.0%	?Glycogen storage disease XIII, 612932
ENPP1	129,2	97.5%	93.3%	Arterial calcification, generalized, of infancy, 1, 208000 Cole disease, 615522 Hypophosphatemic rickets, autosomal recessive, 2, 613312 {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853 {Obesity, susceptibility to}, 601665
ENTPD1	125	100.0%	99.9%	Spastic paraplegia 64, autosomal recessive, 615683
EOGT	102,7	79.3%	78.1%	Adams-Oliver syndrome 4, 615297
EPB41	119,2	99.6%	97.4%	Elliptocytosis-1, 611804
EPB42	128	100.0%	99.3%	Spherocytosis, type 5, 612690
EPCAM	76,5	99.7%	95.7%	Colorectal cancer, hereditary nonpolyposis, type 8, 613244 Diarrhea 5, with tufting enteropathy, congenital, 613217
EPG5	110,3	99.3%	97.9%	Vici syndrome, 242840
EPHX1	116	99.2%	96.0%	?Hypercholanemia, familial, 607748
EPM2A	116,5	90.9%	88.8%	Epilepsy, progressive myoclonic 2A (Lafora), 254780
EPO	98,6	100.0%	99.4%	?Diamond-Blackfan anemia-like, 617911 Erythrocytosis, familial, 5, 617907 {Microvascular complications of diabetes 2}, 612623
EPRS	139,4	99.9%	99.2%	Leukodystrophy, hypomyelinating, 15, 617951
EPS8	114,6	99.8%	96.6%	?Deafness, autosomal recessive 102, 615974
EPS8L2	159,7	99.0%	96.3%	Deafness autosomal recessive 106, 617637
ERAL1	156,5	100.0%	100.0%	Perrault syndrome 6, 617565
ERBB3	113,3	99.9%	99.2%	?Lethal congenital contractural syndrome 2, 607598 {?Erythroleukemia, familial, susceptibility to}, 133180
ERCC1	85,7	100.0%	98.1%	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	128	100.0%	99.8%	?Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy 1, photosensitive, 601675 Xeroderma pigmentosum, group D, 278730
ERCC3	92	99.9%	98.4%	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651

ERCC4	132	100.0%	99.8%	Fanconi anemia, complementation group Q, 615272 Xeroderma pigmentosum, group F, 278760 Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 XFE progeroid syndrome, 610965
ERCC5	126,3	99.9%	99.5%	Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	158,2	100.0%	99.9%	Cerebrooculofacioskeletal syndrome 1, 214150 Cockayne syndrome, type B, 133540 De Sanctis-Cacchione syndrome, 278800 Premature ovarian failure 11, 616946 UV-sensitive syndrome 1, 600630 {Lung cancer, susceptibility to}, 211980 {Macular degeneration, age-related, susceptibility to, 5}, 613761
ERCC6L2	121,7	99.9%	99.0%	Bone marrow failure syndrome 2, 615715
ERCC8	82,8	98.9%	90.0%	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621
ERGIC1	178	95.3%	94.5%	?Arthrogryposis multiplex congenita, neurogenic type, 208100
ERLIN1	141,4	100.0%	100.0%	Spastic paraplegia 62, 615681
ERLIN2	115,3	100.0%	99.2%	Spastic paraplegia 18, autosomal recessive, 611225
ESCO2	115,6	99.4%	97.3%	Roberts syndrome, 268300 SC phocomelia syndrome, 269000
ESPN	30,8	53.5%	42.4%	Deafness, autosomal recessive 36, 609006 Deafness, neurosensory, without vestibular involvement, autosomal dominant, 0
ESR1	137	100.0%	100.0%	Breast cancer, somatic, 114480 Estrogen resistance, 615363 {Atherosclerosis, susceptibility to}, 0 {HDL response to hormone replacement, augmented}, 0 {Migraine, susceptibility to}, 157300 {Myocardial infarction, susceptibility to}, 608446
ESRP1	95,3	99.8%	98.4%	?Deafness, autosomal recessive 109, 618013
ESRRB	121,7	99.9%	99.1%	Deafness, autosomal recessive 35, 608565
ETFA	132,7	100.0%	99.8%	Glutaric acidemia IIA, 231680
ETFB	116,8	100.0%	100.0%	Glutaric acidemia IIB, 231680
ETFDH	114,4	100.0%	99.3%	Glutaric acidemia IIC, 231680
ETHE1	97,3	99.9%	97.8%	Ethylmalonic encephalopathy, 602473
EVC	106,3	95.9%	92.4%	?Weyers acrofacial dysostosis, 193530 Ellis-van Creveld syndrome, 225500

EVC2	110,2	99.4%	96.3%	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530
EXOSC2	110,3	100.0%	99.9%	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EXOSC3	125,1	96.4%	87.8%	Pontocerebellar hypoplasia, type 1B, 614678
EXOSC8	89	98.2%	90.3%	Pontocerebellar hypoplasia, type 1C, 616081
EXOSC9	132,5	99.6%	95.1%	Pontocerebellar hypoplasia, type 1D, 618065
EXPH5	163,6	100.0%	99.9%	Epidermolysis bullosa, nonspecific, autosomal recessive, 615028
EXT2	118	99.9%	99.1%	?Seizures, scoliosis, and macrocephaly syndrome, 616682 Exostoses, multiple, type 2, 133701
EXTL3	184,1	100.0%	100.0%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
EYS	132,5	99.7%	97.5%	Retinitis pigmentosa 25, 602772
F10	173,6	99.8%	99.1%	Factor X deficiency, 227600
F11	126,7	100.0%	99.9%	Factor XI deficiency, autosomal dominant, 612416 Factor XI deficiency, autosomal recessive, 612416
F12	151,3	99.9%	99.5%	Angioedema, hereditary, type III, 610618 Factor XII deficiency, 234000
F13A1	112,9	100.0%	99.6%	Factor XIIIA deficiency, 613225 {Myocardial infarction, protection against}, 608446 {Venous thrombosis, protection against}, 188050
F13B	105,8	98.6%	92.9%	Factor XIIIB deficiency, 613235
F2	128,1	99.8%	97.1%	Dysprothrombinemia, 613679 Hypoprothrombinemia, 613679 Thrombophilia due to thrombin defect, 188050 {Pregnancy loss, recurrent, susceptibility to, 2}, 614390 {Stroke, ischemic, susceptibility to}, 601367
F5	145,5	99.5%	97.7%	Factor V deficiency, 227400 Thrombophilia due to activated protein C resistance, 188055 {Budd-Chiari syndrome}, 600880 {Pregnancy loss, recurrent, susceptibility to, 1}, 614389 {Stroke, ischemic, susceptibility to}, 601367 {Thrombophilia, susceptibility to, due to factor V Leiden}, 188055
F7	162	100.0%	100.0%	Factor VII deficiency, 227500 {Myocardial infarction, decreased susceptibility to}, 608446
FA2H	92,7	98.8%	92.5%	Spastic paraplegia 35, autosomal recessive, 612319
FADD	181,7	100.0%	100.0%	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations, 613759
FAH	128,4	100.0%	99.8%	Tyrosinemia, type I, 276700
FAM126A	125,4	100.0%	99.4%	Leukodystrophy, hypomyelinating, 5, 610532

FAM161A	136,9	99.9%	99.1%	Retinitis pigmentosa 28, 606068
FAM20A	111,1	100.0%	99.4%	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM20C	145,2	100.0%	100.0%	Raine syndrome, 259775
FAN1	132,2	100.0%	99.9%	Interstitial nephritis, karyomegalic, 614817
FANCA	112,4	99.9%	98.9%	Fanconi anemia, complementation group A, 227650
FANCB	76,4	98.6%	93.2%	Fanconi anemia, complementation group B, 300514
FANCC	100,8	99.7%	99.2%	Fanconi anemia, complementation group C, 227645
FANCD2	115,6	99.1%	96.6%	Fanconi anemia, complementation group D2, 227646
FANCE	118,2	96.6%	89.9%	Fanconi anemia, complementation group E, 600901
FANCF	244,4	100.0%	100.0%	Fanconi anemia, complementation group F, 603467
FANCG	140,7	100.0%	99.8%	Fanconi anemia, complementation group G, 614082
FANCI	136,2	99.9%	98.9%	Fanconi anemia, complementation group I, 609053
FANCL	105,8	99.7%	98.0%	Fanconi anemia, complementation group L, 614083
FAR1	73,7	97.2%	91.8%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154
FARS2	161,9	100.0%	100.0%	Combined oxidative phosphorylation deficiency 14, 614946 Spastic paraplegia 77, autosomal recessive, 617046
FARSB	78	97.4%	92.7%	Rajab interstitial lung disease with brain calcifications, 613658
FASTKD2	115,5	99.6%	97.9%	?Mitochondrial complex IV deficiency, 220110
FAT4	190,3	100.0%	99.9%	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 Van Maldergem syndrome 2, 615546
FBLN5	96,6	91.8%	91.5%	Cutis laxa, autosomal dominant 2, 614434 Cutis laxa, autosomal recessive, type IA, 219100 Macular degeneration, age-related, 3, 608895 Neuropathy, hereditary, with or without age-related macular degeneration, 608895
FBP1	105,5	100.0%	99.3%	Fructose-1,6-bisphosphatase deficiency, 229700
FBXL4	168,9	100.0%	100.0%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FBXO31	111,5	99.6%	97.3%	?Mental retardation, autosomal recessive 45, 615979
FBXO7	152,8	99.9%	99.6%	Parkinson disease 15, autosomal recessive, 260300
FCGR3A	163,2	99.6%	96.7%	Immunodeficiency 20, 615707
FCN3	124,7	100.0%	100.0%	Immunodeficiency due to ficolin 3 deficiency, 613860
FDXR	122,3	99.9%	99.1%	Auditory neuropathy and optic atrophy, 617717
FECH	104	100.0%	99.7%	Protoporphyria, erythropoietic, 1, 177000
FERMT1	90,8	99.6%	96.6%	Kindler syndrome, 173650
FERMT3	144,9	100.0%	99.9%	Leukocyte adhesion deficiency, type III, 612840
FEZF1	179,7	100.0%	100.0%	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FGA	137	99.3%	97.3%	Afibrinogenemia, congenital, 202400 Amyloidosis, familial visceral, 105200

				Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, congenital, 616004
FGB	136,7	99.7%	98.2%	Afibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Hypofibrinogenemia, congenital, 202400
FGD4	104,3	99.7%	97.8%	Charcot-Marie-Tooth disease, type 4H, 609311
FGF20	125,3	100.0%	98.3%	?Renal hypodysplasia/aplasia 2, 615721
FGF23	122,3	99.7%	97.7%	Hypophosphatemic rickets, autosomal dominant, 193100 Osteomalacia, tumor-induced, 0 Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993
FGF3	139,5	100.0%	100.0%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGG	126,4	99.8%	98.0%	Afibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, 616004 Hypofibrinogenemia, congenital, 202400
FH	128	95.0%	88.5%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FIBP	124,7	100.0%	99.8%	Thauvin-Robinet-Faivre syndrome, 617107
FIG4	157,5	100.0%	99.6%	?Polymicrogyria, bilateral temporooccipital, 612691 Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228 Yunis-Varon syndrome, 216340
FKBP10	157,5	99.5%	97.3%	Bruck syndrome 1, 259450 Osteogenesis imperfecta, type XI, 610968
FKBP14	80,8	99.8%	97.9%	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557
FKRP	153,3	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155
FKTN	107,5	99.7%	96.1%	Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588
FLAD1	170,7	100.0%	99.6%	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100
FLG	147,1	100.0%	99.9%	Ichthyosis vulgaris, 146700 {Dermatitis, atopic, susceptibility to, 2}, 605803
FLNB	123,6	99.7%	98.7%	Atelosteogenesis, type I, 108720 Atelosteogenesis, type III, 108721 Boomerang dysplasia, 112310

				Larsen syndrome, 150250 Spondylocarpotarsal synostosis syndrome, 272460
FLVCR1	146,1	99.9%	99.2%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FLVCR2	124,8	100.0%	100.0%	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790
FMN2	109,6	86.6%	84.2%	Mental retardation, autosomal recessive 47, 616193
FMO3	131,9	100.0%	99.3%	Trimethylaminuria, 602079
FOLR1	107,4	100.0%	99.9%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXE1	87,5	100.0%	99.7%	Bamforth-Lazarus syndrome, 241850 {Thyroid cancer, nonmedullary, 4}, 616534
FOXE3	88,3	89.7%	82.5%	Anterior segment dysgenesis 2, multiple subtypes, 610256 Cataract 34, multiple types, 612968 {Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349
FOXI1	194,5	100.0%	100.0%	Enlarged vestibular aqueduct, 600791
FOXN1	133	100.0%	99.5%	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXRED1	121	99.8%	98.2%	Mitochondrial complex I deficiency, nuclear type 19, 618241
FRAS1	119,2	99.9%	99.2%	Fraser syndrome 1, 219000
FREM1	110,7	99.8%	98.4%	Bifid nose with or without anorectal and renal anomalies, 608980 Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485
FREM2	152,4	100.0%	99.5%	Cryptophthalmos, unilateral or bilateral, isolated, 123570 Fraser syndrome 2, 617666
FRMD4A	115,2	91.4%	90.5%	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819
FRRS1L	99,1	85.5%	79.1%	Epileptic encephalopathy, early infantile, 37, 616981
FSHB	112	100.0%	100.0%	Hypogonadotropic hypogonadism 24 without anosmia, 229070
FSHR	96,6	99.9%	97.4%	Ovarian dysgenesis 1, 233300 Ovarian hyperstimulation syndrome, 608115 Ovarian response to FSH stimulation, 276400
FTCD	116,8	98.7%	95.2%	Glutamate formiminotransferase deficiency, 229100
FTO	95,8	83.8%	83.6%	Growth retardation, developmental delay, facial dysmorphism, 612938 {Obesity, susceptibility to, BMIQ14}, 612460
FUCA1	125,9	100.0%	99.9%	Fucosidosis, 230000
FUT8	130,4	99.9%	99.2%	Congenital disorder of glycosylation with defective fucosylation 1, 618005
FXN	64,9	99.7%	96.8%	Friedreich ataxia, 229300 Friedreich ataxia with retained reflexes, 229300
FYCO1	123,2	100.0%	100.0%	Cataract 18, autosomal recessive, 610019
FZD6	186,4	100.0%	100.0%	Nail disorder, nonsyndromic congenital, 10, (claw-shaped nails), 614157
G6PC	146,8	100.0%	99.9%	Glycogen storage disease Ia, 232200

G6PC3	114,6	100.0%	100.0%	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
GAA	160,8	100.0%	99.9%	Glycogen storage disease II, 232300
GAB1	155,1	100.0%	99.5%	?Deafness, autosomal recessive 26, 605428
GAD1	112,7	99.9%	99.7%	?Cerebral palsy, spastic quadriplegic, 1, 603513
GALC	102,9	99.8%	98.8%	Krabbe disease, 245200
GALE	140	100.0%	100.0%	Galactose epimerase deficiency, 230350
GALK1	165,2	100.0%	99.9%	Galactokinase deficiency with cataracts, 230200
GALNS	108,3	100.0%	99.3%	Mucopolysaccharidosis IVA, 253000
GALNT3	125,8	99.9%	98.7%	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GALT	152,6	100.0%	100.0%	Galactosemia, 230400
GAMT	112,5	98.3%	91.5%	Cerebral creatine deficiency syndrome 2, 612736
GAN	142,2	99.9%	99.4%	Giant axonal neuropathy-1, 256850
GAS8	127,3	100.0%	99.4%	Ciliary dyskinesia, primary, 33, 616726
GATM	137,3	100.0%	100.0%	Cerebral creatine deficiency syndrome 3, 612718
GBA	169,8	100.0%	100.0%	Gaucher disease, perinatal lethal, 608013 Gaucher disease, type I, 230800 Gaucher disease, type II, 230900 Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 {Lewy body dementia, susceptibility to}, 127750 {Parkinson disease, late-onset, susceptibility to}, 168600
GBA2	141,5	100.0%	99.6%	Spastic paraplegia 46, autosomal recessive, 614409
GBE1	157,4	99.9%	99.7%	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GCDH	145,9	100.0%	99.1%	Glutaricaciduria, type I, 231670
GCH1	84,8	100.0%	99.5%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GCK	138,6	100.0%	100.0%	Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, permanent neonatal, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485 MODY, type II, 125851
GCLC	143,7	99.3%	95.8%	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 {Myocardial infarction, susceptibility to}, 608446
GCNT2	151,2	99.5%	99.5%	Adult i phenotype without cataract, 110800 Cataract 13 with adult i phenotype, 116700 [Blood group, Ii], 110800

GCSH	32,1	88.4%	69.8%	?Glycine encephalopathy, 605899
GDAP1	145,6	99.8%	99.0%	Charcot-Marie-Tooth disease, axonal, type 2K, 607831 Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706 Charcot-Marie-Tooth disease, recessive intermediate, A, 608340 Charcot-Marie-Tooth disease, type 4A, 214400
GDF1	50,7	97.8%	84.7%	Congenital heart defects, multiple types, 6, 613854 Right atrial isomerism (Ivemark), 208530
GDF5	169,6	100.0%	100.0%	?Acromesomelic dysplasia, Hunter-Thompson type, 201250 Brachydactyly, type A1, C, 615072 Brachydactyly, type A2, 112600 Brachydactyly, type C, 113100 Chondrodysplasia, Grebe type, 200700 Du Pan syndrome, 228900 Multiple synostoses syndrome 2, 610017 Symphalangism, proximal, 1B, 615298 {Osteoarthritis-5}, 612400
GEMIN4	148,4	100.0%	99.7%	Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities, 617913
GFER	90,6	100.0%	99.6%	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076
GFM1	104,2	99.9%	99.0%	Combined oxidative phosphorylation deficiency 1, 609060
GFPT1	146	99.9%	99.1%	Myasthenia, congenital, 12, with tubular aggregates, 610542
GGCX	101,2	100.0%	99.4%	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842 Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450
GGT1	12,1	20.3%	18.2%	?Glutathioninuria, 231950
GH1	159,5	100.0%	100.0%	Growth hormone deficiency, isolated, type IA, 262400 Growth hormone deficiency, isolated, type IB, 612781 Growth hormone deficiency, isolated, type II, 173100 Kowarski syndrome, 262650
GHR	150,6	99.5%	99.4%	Growth hormone insensitivity, partial, 604271 Increased responsiveness to growth hormone, 604271 Laron dwarfism, 262500 {Hypercholesterolemia, familial, modifier of}, 143890
GHRHR	108,1	96.0%	95.2%	Growth hormone deficiency, isolated, type IV, 618157
GHSR	173,6	98.7%	95.2%	Growth hormone deficiency, isolated partial, 615925
GIF	NC	NC	NC	Intrinsic factor deficiency, 261000
GINS1	122,7	98.1%	90.6%	Immunodeficiency 55, 617827
GIPC3	127,8	98.9%	95.2%	Deafness, autosomal recessive 15, 601869
GJA1	156,2	100.0%	100.0%	Atrioventricular septal defect 3, 600309 Craniometaphyseal dysplasia, autosomal recessive, 218400

				Erythrokeratodermia variabilis et progressiva 3, 617525 Hypoplastic left heart syndrome 1, 241550 Oculodentodigital dysplasia, 164200 Oculodentodigital dysplasia, autosomal recessive, 257850 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100
GJB2	141,4	100.0%	100.0%	Bart-Pumphrey syndrome, 149200 Deafness, autosomal dominant 3A, 601544 Deafness, autosomal recessive 1A, 220290 Hystrix-like ichthyosis with deafness, 602540 Keratitis-ichthyosis-deafness syndrome, 148210 Keratoderma, palmoplantar, with deafness, 148350 Vohwinkel syndrome, 124500
GJB6	140,9	100.0%	100.0%	Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290 Ectodermal dysplasia 2, Clouston type, 129500
GJC2	45,3	92.6%	75.4%	Leukodystrophy, hypomyelinating, 2, 608804 Lymphatic malformation 3, 613480 Spastic paraplegia 44, autosomal recessive, 613206
GLB1	82,6	99.7%	95.4%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLDC	59,2	90.6%	79.2%	Glycine encephalopathy, 605899
GLDN	106,6	99.8%	96.9%	Lethal congenital contracture syndrome 11, 617194
GLE1	97,1	100.0%	99.9%	Congenital arthrogryposis with anterior horn cell disease, 611890 Lethal congenital contracture syndrome 1, 253310
GLIS2	129,6	100.0%	100.0%	Nephronophthisis 7, 611498
GLIS3	123,7	100.0%	99.5%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GLRA1	96,8	100.0%	99.7%	Hyperekplexia 1, 149400
GLRB	104,6	99.5%	94.5%	Hyperekplexia 2, 614619
GLRX5	137,6	99.6%	96.1%	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GLUL	77	99.7%	96.5%	Glutamine deficiency, congenital, 610015
GLYCTK	161,3	100.0%	99.5%	D-glyceric aciduria, 220120
GM2A	122	100.0%	100.0%	GM2-gangliosidosis, AB variant, 272750
GMPPA	147,2	100.0%	99.8%	Alacrima, achalasia, and mental retardation syndrome, 615510

GMPPB	211,8	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352
GNAT2	100,7	99.8%	98.2%	Achromatopsia 4, 613856
GNB3	146	100.0%	99.9%	Night blindness, congenital stationary, type 1H, 617024 {Hypertension, essential, susceptibility to}, 145500
GNB5	113	99.9%	97.5%	Intellectual developmental disorder with cardiac arrhythmia, 617173 Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182
GNE	113,8	100.0%	99.3%	Nonaka myopathy, 605820 Sialuria, 269921
GNMT	126,9	99.9%	98.7%	Glycine N-methyltransferase deficiency, 606664
GNPAT	127,2	99.5%	96.8%	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	148	100.0%	99.3%	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GNPTG	177,6	100.0%	98.5%	Mucolipidosis III gamma, 252605
GNRHR	145,4	100.0%	100.0%	Hypogonadotropic hypogonadism 7 without anosmia, 146110
GNS	94,5	99.6%	95.2%	Mucopolysaccharidosis type IIID, 252940
GORAB	165,7	100.0%	98.9%	Geroderma osteodysplasticum, 231070
GOSR2	102,6	95.8%	93.7%	Epilepsy, progressive myoclonic 6, 614018
GP1BA	136,8	98.7%	95.7%	Bernard-Soulier syndrome, type A1 (recessive), 231200 Bernard-Soulier syndrome, type A2 (dominant), 153670 von Willebrand disease, platelet-type, 177820 {Nonarteritic anterior ischemic optic neuropathy, susceptibility to}, 258660
GP1BB	68,6	94.5%	83.1%	Bernard-Soulier syndrome, type B, 231200 Giant platelet disorder, isolated, 231200
GP6	123,2	100.0%	99.7%	Bleeding disorder, platelet-type, 11, 614201
GP9	134,3	99.9%	98.3%	Bernard-Soulier syndrome, type C, 231200
GPAA1	123,1	100.0%	98.3%	Glycosylphosphatidylinositol biosynthesis defect 15, 617810
GPC6	126,6	100.0%	100.0%	Omodysplasia 1, 258315
GPD1	87,7	100.0%	99.4%	Hypertriglyceridemia, transient infantile, 614480
GPHN	144,7	99.9%	98.8%	Molybdenum cofactor deficiency C, 615501
GPI	141,4	100.0%	99.8%	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470
GPIHBP1	149,5	100.0%	100.0%	Hyperlipoproteinemia, type 1D, 615947
GPNMB	150,5	100.0%	100.0%	Amyloidosis, primary localized cutaneous, 3, 617920
GPR179	146,8	100.0%	100.0%	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
GPR88	146,7	100.0%	99.6%	?Chorea, childhood-onset, with psychomotor retardation, 616939
GPSM2	119,6	99.8%	99.0%	Chudley-McCullough syndrome, 604213

GPT2	121,7	100.0%	99.4%	Mental retardation, autosomal recessive 49, 616281
GPX4	165,9	94.4%	90.7%	Spondylometaphyseal dysplasia, Sedaghatian type, 250220
GRHPR	99,6	84.2%	81.7%	Hyperoxaluria, primary, type II, 260000
GRID2	146,8	99.9%	99.4%	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRIK2	121,7	96.2%	95.4%	Mental retardation, autosomal recessive, 6, 611092
GRIN1	166,1	100.0%	99.9%	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254 Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820
GRIP1	111,1	100.0%	99.3%	Fraser syndrome 3, 617667
GRK1	135,7	100.0%	100.0%	Oguchi disease-2, 613411
GRM1	156,6	100.0%	99.9%	Spinocerebellar ataxia 44, 617691 Spinocerebellar ataxia, autosomal recessive 13, 614831
GRM6	138,3	98.0%	92.4%	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270
GRN	174,1	100.0%	100.0%	Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
GRXCR1	154,5	100.0%	99.7%	Deafness, autosomal recessive 25, 613285
GRXCR2	112,4	100.0%	99.9%	?Deafness, autosomal recessive 101, 615837
GSC	133,8	100.0%	98.9%	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471
GSS	93,3	100.0%	99.2%	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900
GTF2E2	85,2	100.0%	98.2%	Trichothiodystrophy 6, nonphotosensitive, 616943
GTF2H5	81,8	99.9%	95.9%	Trichothiodystrophy 3, photosensitive, 616395
GTPBP2	128,9	99.7%	98.6%	Jaberi-Elahi syndrome, 617988
GTPBP3	164,7	100.0%	100.0%	Combined oxidative phosphorylation deficiency 23, 616198
GUCY1A3	NC	NC	NC	Moyamoya 6 with achalasia, 615750
GUCY2C	117,5	100.0%	99.4%	Diarrhea 6, 614616 Meconium ileus, 614665
GUCY2D	108,9	100.0%	99.9%	?Choroidal dystrophy, central areolar 1, 215500 Cone-rod dystrophy 6, 601777 Leber congenital amaurosis 1, 204000
GUF1	95,1	99.9%	98.4%	?Epileptic encephalopathy, early infantile, 40, 617065
GUSB	99,5	92.5%	90.5%	Mucopolysaccharidosis VII, 253220
GYG1	125,6	100.0%	99.4%	?Glycogen storage disease XV, 613507 Polyglucosan body myopathy 2, 616199
GYS1	109,6	100.0%	98.6%	Glycogen storage disease 0, muscle, 611556
GYS2	120,1	99.7%	98.0%	Glycogen storage disease 0, liver, 240600
GZF1	187,8	100.0%	99.7%	Joint laxity, short stature, and myopia, 617662

H6PD	193,5	99.0%	99.0%	Cortisone reductase deficiency 1, 604931
HAAO	104,6	100.0%	99.8%	Vertebral, cardiac, renal, and limb defects syndrome 1, 617660
HACE1	136,2	99.9%	99.1%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADH	111,1	99.3%	98.8%	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HADHA	72,9	96.3%	89.3%	Fatty liver, acute, of pregnancy, 609016 HELLP syndrome, maternal, of pregnancy, 609016 LCHAD deficiency, 609016 Trifunctional protein deficiency, 609015
HADHB	77,6	96.7%	83.8%	Trifunctional protein deficiency, 609015
HAMP	169	100.0%	100.0%	Hemochromatosis, type 2B, 613313
HARS	134,8	100.0%	100.0%	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504
HARS2	136,6	100.0%	99.9%	?Perrault syndrome 2, 614926
HAX1	137,4	100.0%	100.0%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HBB	131,9	100.0%	100.0%	Delta-beta thalassemia, 141749 Erythrocytosis 6, 617980 Heinz body anemia, 140700 Hereditary persistence of fetal hemoglobin, 141749 Methmoglobinemia, beta type, 617971 Sickle cell anemia, 603903 Thalassemia, beta, 613985 Thalassemia-beta, dominant inclusion-body, 603902 {Malaria, resistance to}, 611162
HELLS	102,8	98.8%	92.7%	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911
HEPACAM	118,3	95.5%	88.7%	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926
HERC1	142,1	100.0%	99.7%	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
HERC2	95,1	80.0%	76.1%	Mental retardation, autosomal recessive 38, 615516 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220
HES7	53,9	90.1%	72.7%	Spondylocostal dysostosis 4, autosomal recessive, 613686
HESX1	66,2	100.0%	98.7%	Growth hormone deficiency with pituitary anomalies, 182230 Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230
HEXA	106,3	93.7%	92.4%	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800

				[Hex A pseudodeficiency], 272800
HEXB	163	99.7%	98.5%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HFE	108	100.0%	98.9%	Hemochromatosis, 235200 [Transferrin serum level QTL2], 614193 {Alzheimer disease, susceptibility to}, 104300 {Microvascular complications of diabetes 7}, 612635 {Porphyria cutanea tarda, susceptibility to}, 176100 {Porphyria variegata, susceptibility to}, 176200
HFE2	NC	NC	NC	Hemochromatosis, type 2A, 602390
HFM1	54,6	96.7%	90.6%	Premature ovarian failure 9, 615724
HGD	98,2	100.0%	99.8%	Alkaptonuria, 203500
HGF	138	99.9%	99.2%	Deafness, autosomal recessive 39, 608265
HGSNAT	98,3	87.2%	86.2%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544
HIBCH	69,9	96.3%	79.8%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HIKESHI	54,5	96.8%	84.3%	Leukodystrophy, hypomyelinating, 13, 616881
HINT1	63,3	91.2%	79.1%	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200
HK1	116,5	100.0%	99.7%	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285 Retinitis pigmentosa 79, 617460
HLCS	142,3	100.0%	100.0%	Holocarboxylase synthetase deficiency, 253270
HMGCL	119,4	99.9%	98.7%	HMG-CoA lyase deficiency, 246450
HMGCS2	102,4	100.0%	99.4%	HMG-CoA synthase-2 deficiency, 605911
HMOX1	137,4	96.5%	90.7%	Heme oxygenase-1 deficiency, 614034 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963
HMX1	39,8	85.4%	63.9%	Oculoauricular syndrome, 612109
HNMT	133,1	100.0%	99.9%	Mental retardation, autosomal recessive 51, 616739 {Asthma, susceptibility to}, 600807
HOGA1	149,6	100.0%	99.1%	Hyperoxaluria, primary, type III, 613616
HOXA1	164	100.0%	100.0%	Athabaskan brainstem dysgenesis syndrome, 601536 Bosley-Salih-Alorainy syndrome, 601536
HOXA2	83	100.0%	99.9%	?Microtia, hearing impairment, and cleft palate (AR), 612290 Microtia with or without hearing impairment (AD), 612290
HOXB1	147,8	100.0%	100.0%	Facial paresis, hereditary congenital, 3, 614744
HOXC13	172,8	100.0%	100.0%	Ectodermal dysplasia 9, hair/nail type, 614931
HPCA	253,9	100.0%	100.0%	Dystonia 2, torsion, autosomal recessive, 224500
HPD	148,1	100.0%	99.7%	Hawkinsinuria, 140350

				Tyrosinemia, type III, 276710
HPGD	90,6	99.9%	98.9%	Cranioosteoarthropathy, 259100 Digital clubbing, isolated congenital, 119900 Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100
HPS1	115,8	100.0%	99.9%	Hermansky-Pudlak syndrome 1, 203300
HPS3	132,7	99.9%	98.8%	Hermansky-Pudlak syndrome 3, 614072
HPS4	128,1	100.0%	99.9%	Hermansky-Pudlak syndrome 4, 614073
HPS5	122,8	99.9%	98.7%	Hermansky-Pudlak syndrome 5, 614074
HPS6	164,6	99.9%	97.8%	Hermansky-Pudlak syndrome 6, 614075
HPSE2	100,6	100.0%	99.7%	Urofacial syndrome 1, 236730
HR	117,4	99.6%	97.3%	Alopecia universalis, 203655 Atrichia with papular lesions, 209500 Hypotrichosis 4, 146550
HSD11B2	165,6	94.3%	87.3%	Apparent mineralocorticoid excess, 218030
HSD17B3	116,4	100.0%	99.9%	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	109,4	96.3%	93.6%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B2	131,8	100.0%	99.9%	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810
HSD3B7	143,7	99.5%	96.2%	Bile acid synthesis defect, congenital, 1, 607765
HSPA9	82,6	89.5%	84.2%	Anemia, sideroblastic, 4, 182170 Even-plus syndrome, 616854
HSPD1	74,3	98.1%	92.5%	Leukodystrophy, hypomyelinating, 4, 612233 Spastic paraplegia 13, autosomal dominant, 605280
HSPG2	119,8	99.5%	98.8%	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
HTRA1	89	95.2%	87.0%	CARASIL syndrome, 600142 Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779 {Macular degeneration, age-related, 7}, 610149 {Macular degeneration, age-related, neovascular type}, 610149
HTRA2	132,6	100.0%	99.6%	3-methylglutaconic aciduria, type VIII, 617248 {Parkinson disease 13}, 610297
HYAL1	110,7	100.0%	100.0%	?Mucopolysaccharidosis type IX, 601492
HYDIN	106,8	99.8%	98.9%	Ciliary dyskinesia, primary, 5, 608647
HYLS1	156,6	100.0%	100.0%	Hydrolethalus syndrome, 236680
IARS	125,4	99.9%	99.0%	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093
IARS2	142,9	100.0%	99.9%	?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007

IBA57	137,4	99.3%	95.9%	?Spastic paraplegia 74, autosomal recessive, 616451 Multiple mitochondrial dysfunctions syndrome 3, 615330
ICK	110	99.8%	99.3%	Endocrine-cerebroosteodysplasia, 612651 {Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924
ICOS	156,4	100.0%	99.9%	Immunodeficiency, common variable, 1, 607594
IDH3B	128,2	95.5%	95.4%	Retinitis pigmentosa 46, 612572
IDUA	148,1	98.9%	94.6%	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016
IER3IP1	106,3	94.3%	82.8%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFNAR2	129,5	99.5%	96.9%	?Immunodeficiency 45, 616669 {Hepatitis B virus, susceptibility to}, 610424
IFNGR1	145	99.9%	99.3%	Immunodeficiency 27A, mycobacteriosis, AR, 209950 Immunodeficiency 27B, mycobacteriosis, AD, 615978 {H. pylori infection, susceptibility to}, 600263 {Hepatitis B virus infection, susceptibility to}, 610424 {Tuberculosis infection, protection against}, 607948 {Tuberculosis, susceptibility to}, 607948
IFNGR2	125,2	97.0%	93.5%	Immunodeficiency 28, mycobacteriosis, 614889
IFT122	120,5	99.9%	99.0%	Cranioectodermal dysplasia 1, 218330
IFT140	117,6	99.9%	99.2%	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	94,5	100.0%	99.4%	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	115,8	100.0%	100.0%	?Bardet-Biedl syndrome 19, 615996
IFT43	112,4	100.0%	100.0%	?Cranioectodermal dysplasia 3, 614099 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866
IFT52	121	100.0%	99.7%	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102
IFT57	120,5	99.9%	99.3%	?Orofaciodigital syndrome XVIII, 617927
IFT74	84,7	99.1%	95.8%	?Bardet-Biedl syndrome 20, 617119
IFT80	64,9	96.7%	84.7%	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IFT81	92,3	93.6%	89.0%	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IGF1	98	100.0%	99.8%	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IGF1R	114,9	100.0%	99.6%	Insulin-like growth factor I, resistance to, 270450
IGFALS	108,2	100.0%	99.9%	Acid-labile subunit, deficiency of, 615961
IGFBP7	82,7	99.4%	95.3%	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis, 614224

IGHM	170	100.0%	100.0%	Agammaglobulinemia 1, 601495
IGHMBP2	108,3	99.6%	97.4%	Charcot-Marie-Tooth disease, axonal, type 2S, 616155 Neuronopathy, distal hereditary motor, type VI, 604320
IGKC	113,2	100.0%	100.0%	Kappa light chain deficiency, 614102
IGLL1	92,2	100.0%	99.6%	Agammaglobulinemia 2, 613500
IHH	171,9	100.0%	100.0%	Acrocapitofemoral dysplasia, 607778 Brachydactyly, type A1, 112500
IKBKB	110	99.3%	96.4%	Immunodeficiency 15A, 618204 Immunodeficiency 15B, 615592
IL10RA	144,4	100.0%	100.0%	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	130,1	100.0%	99.3%	Inflammatory bowel disease 25, early onset, autosomal recessive, 612567 {Hepatitis B virus, susceptibility to}, 610424
IL11RA	131,5	100.0%	99.6%	Craniosynostosis and dental anomalies, 614188
IL12B	94,8	99.9%	97.0%	Immunodeficiency 29, mycobacteriosis, 614890
IL12RB1	112,9	98.0%	95.6%	Immunodeficiency 30, 614891
IL17RA	149,1	100.0%	100.0%	Immunodeficiency 51, 613953
IL17RC	123,6	100.0%	100.0%	Candidiasis, familial, 9, 616445
IL1RN	139,3	100.0%	99.7%	Interleukin 1 receptor antagonist deficiency, 612852 {Gastric cancer risk after H. pylori infection}, 137215 {Microvascular complications of diabetes 4}, 612628
IL21R	145	100.0%	100.0%	Immunodeficiency 56, 615207 [IgE, elevated level of], 147050
IL2RA	100,6	99.9%	98.7%	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367 {Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942
IL36RN	92,8	100.0%	100.0%	Psoriasis 14, pustular, 614204
IL7R	114,1	100.0%	99.9%	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
ILDR1	111,5	99.9%	98.7%	Deafness, autosomal recessive 42, 609646
IMPA1	72,1	96.7%	86.7%	Mental retardation, autosomal recessive 59, 617323
IMPAD1	170,4	100.0%	99.9%	Chondrodysplasia with joint dislocations, GPAPP type, 614078
IMPG2	125,6	99.4%	97.6%	Macular dystrophy, vitelliform, 5, 616152 Retinitis pigmentosa 56, 613581
INPP5E	116,8	100.0%	98.6%	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INPP5K	88,8	100.0%	99.3%	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404
INPPL1	127,9	99.8%	98.0%	Opsismodysplasia, 258480
INSR	116,4	99.0%	95.1%	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968

				Leprechaunism, 246200 Rabson-Mendenhall syndrome, 262190
INTU	115,4	99.8%	98.7%	?Orofaciodigital syndrome XVII, 617926 ?Short-rib thoracic dysplasia 20 with polydactyly, 617925
INVS	143,7	100.0%	99.9%	Nephronophthisis 2, infantile, 602088
IQCB1	93,3	91.6%	80.0%	Senior-Loken syndrome 5, 609254
IRAK4	103,9	99.7%	96.5%	Invasive pneumococcal disease, recurrent isolated, 1, 610799 IRAK4 deficiency, 607676
IRF7	152,6	100.0%	99.8%	?Immunodeficiency 39, 616345
IRF8	111,8	99.9%	98.6%	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893 Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990
IRX5	134,9	100.0%	99.8%	Hamamy syndrome, 611174
ISCA1	67,2	93.5%	84.0%	Multiple mitochondrial dysfunctions syndrome 5, 617613
ISCA2	105,1	99.8%	95.8%	Multiple mitochondrial dysfunctions syndrome 4, 616370
ISCU	117,2	100.0%	99.9%	Myopathy with lactic acidosis, hereditary, 255125
ISG15	184,2	100.0%	100.0%	Immunodeficiency 38, 616126
ISPD	NC	NC	NC	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052
ITCH	116,3	95.4%	94.6%	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITGA2B	124,4	99.9%	98.9%	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Glanzmann thrombasthenia, 273800 Thrombocytopenia, neonatal alloimmune, BAK antigen related, 0
ITGA3	150,1	99.7%	98.0%	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748
ITGA6	138,9	99.9%	99.0%	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730
ITGA7	129,2	99.7%	98.2%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
ITGA8	115,5	100.0%	99.5%	Renal hypodysplasia/aplasia 1, 191830
ITGB2	156,8	100.0%	100.0%	Leukocyte adhesion deficiency, 116920
ITGB3	112,8	100.0%	99.8%	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Glanzmann thrombasthenia, 273800 Purpura, posttransfusion, 0 Thrombocytopenia, neonatal alloimmune, 0 {Myocardial infarction, susceptibility to}, 608446
ITGB4	152,1	99.2%	97.4%	Epidermolysis bullosa of hands and feet, 131800 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa, junctional, with pyloric atresia, 226730
ITGB6	127,5	96.7%	95.0%	Amelogenesis imperfecta, type IH, 616221
ITK	103,1	99.9%	99.1%	Lymphoproliferative syndrome 1, 613011

ITPA	130,2	100.0%	100.0%	Epileptic encephalopathy, early infantile, 35, 616647 [Inosine triphosphatase deficiency], 613850
IVD	100	100.0%	99.9%	Isovaleric acidemia, 243500
IYD	105,5	99.3%	94.7%	Thyroid dyshormonogenesis 4, 274800
JAGN1	118,5	100.0%	100.0%	Neutropenia, severe congenital, 6, autosomal recessive, 616022
JAK3	122,1	98.8%	97.2%	SCID, autosomal recessive, T-negative/B-positive type, 600802
JAM3	126,9	100.0%	99.9%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
JPH1	168,8	100.0%	99.9%	?Charcot-Marie-Tooth disease, axonal, autosomal dominant, type 2K, 607831
JUP	124,5	100.0%	99.8%	Arrhythmogenic right ventricular dysplasia 12, 611528 Naxos disease, 601214
KALRN	124,4	99.9%	99.3%	{Coronary heart disease, susceptibility to, 5}, 608901
KANK2	163,4	100.0%	99.9%	Nephrotic syndrome, type 16, 617783 Palmoplantar keratoderma and woolly hair, 616099
KARS	104,1	100.0%	98.8%	?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness, autosomal recessive 89, 613916
KATNB1	154,2	100.0%	100.0%	Lissencephaly 6, with microcephaly, 616212
KCNE1	369,2	100.0%	100.0%	Jervell and Lange-Nielsen syndrome 2, 612347 Long QT syndrome 5, 613695
KCNJ1	157,5	100.0%	100.0%	Bartter syndrome, type 2, 241200
KCNJ10	148,6	89.2%	88.1%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ11	199,7	100.0%	100.0%	Diabetes mellitus, transient neonatal, 3, 610582 Diabetes, permanent neonatal, with or without neurologic features, 606176 Hyperinsulinemic hypoglycemia, familial, 2, 601820 Maturity-onset diabetes of the young, type 13, 616329 {Diabetes mellitus, type 2, susceptibility to}, 125853
KCNJ13	138,9	100.0%	100.0%	Leber congenital amaurosis 16, 614186 Snowflake vitreoretinal degeneration, 193230
KCNQ1	135,8	97.9%	95.3%	Atrial fibrillation, familial, 3, 607554 Jervell and Lange-Nielsen syndrome, 220400 Long QT syndrome 1, 192500 Short QT syndrome 2, 609621 {Long QT syndrome 1, acquired, susceptibility to}, 192500
KCNV2	138,9	100.0%	100.0%	Retinal cone dystrophy 3B, 610356
KCTD7	154,9	95.0%	95.0%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KERA	175,3	100.0%	100.0%	Cornea plana 2, autosomal recessive, 217300
KHDC3L	160	100.0%	99.7%	Hydatidiform mole, recurrent, 2, 614293

KIAA0586	117,7	97.0%	93.0%	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546
KIAA0753	113,2	99.9%	98.7%	?Orofaciodigital syndrome XV, 617127
KIAA1109	140,8	99.9%	98.9%	Alkuraya-Kucinskas syndrome, 617822
KIF14	116,6	99.6%	97.9%	?Meckel syndrome 12, 616258 Microcephaly 20, primary, autosomal recessive, 617914
KIF1A	115	99.7%	97.6%	Mental retardation, autosomal dominant 9, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal recessive, 610357
KIF1BP	161,1	96.1%	96.0%	Goldberg-Shprintzen megacolon syndrome, 609460
KIF1C	147,7	100.0%	99.4%	Spastic ataxia 2, autosomal recessive, 611302
KIF7	105,2	98.2%	93.5%	?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990
KISS1R	156,4	100.0%	100.0%	?Precocious puberty, central, 1, 176400 Hypogonadotropic hypogonadism 8 with or without anosmia, 614837
KIZ	145	99.9%	98.1%	Retinitis pigmentosa 69, 615780
KL	171,8	99.2%	98.1%	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994
KLC2	127	99.9%	99.2%	Spastic paraplegia, optic atrophy, and neuropathy, 609541
KLHL3	105,6	99.9%	97.7%	Pseudohypoaldosteronism, type IID, 614495
KLHL40	130,6	100.0%	100.0%	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	172,8	100.0%	99.8%	Nemaline myopathy 9, 615731
KLK4	164,3	100.0%	100.0%	Amelogenesis imperfecta, type IIA1, 204700
KLKB1	132,3	100.0%	99.5%	Fletcher factor (prekallikrein) deficiency, 612423
KMT2B	141,1	96.9%	93.5%	Dystonia 28, childhood-onset, 617284
KNL1	105,5	99.0%	97.2%	Microcephaly 4, primary, autosomal recessive, 604321
KPTN	145,7	100.0%	100.0%	Mental retardation, autosomal recessive 41, 615637
KRT10	128,8	99.9%	98.6%	Epidermolytic hyperkeratosis, 113800 Ichthyosis with confetti, 609165 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602
KRT14	42,6	89.0%	80.0%	Dermatopathia pigmentosa reticularis, 125595 Epidermolysis bullosa simplex, Dowling-Meara type, 131760 Epidermolysis bullosa simplex, Koebner type, 131900 Epidermolysis bullosa simplex, recessive 1, 601001 Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 Naegeli-Franceschetti-Jadassohn syndrome, 161000

KRT18	34,2	89.8%	71.1%	Cirrhosis, cryptogenic, 215600 {Cirrhosis, noncryptogenic, susceptibility to}, 215600
KRT5	110,6	100.0%	100.0%	Dowling-Degos disease 1, 179850 Epidermolysis bullosa simplex, Dowling-Meara type, 131760 Epidermolysis bullosa simplex, Koebner type, 131900 Epidermolysis bullosa simplex, recessive 1, 601001 Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 Epidermolysis bullosa simplex-MCR, 609352 Epidermolysis bullosa simplex-MP, 131960
KRT8	37	89.5%	67.8%	Cirrhosis, cryptogenic, 215600 {Cirrhosis, noncryptogenic, susceptibility to}, 215600
KRT85	101,3	99.0%	95.2%	Ectodermal dysplasia 4, hair/nail type, 602032
KY	112,6	100.0%	99.8%	Myopathy, myofibrillar, 7, 617114
KYNU	104,7	98.8%	93.8%	?Hydroxykynureninuria, 236800 Vertebral, cardiac, renal, and limb defects syndrome 2, 617661
L2HGDH	124	99.0%	96.7%	L-2-hydroxyglutaric aciduria, 236792
LAMA1	116	100.0%	99.5%	Poretti-Boltshauser syndrome, 615960
LAMA2	130,6	100.0%	99.5%	Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855 Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138
LAMA3	125,2	99.9%	99.6%	Epidermolysis bullosa, generalized atrophic benign, 226650 Epidermolysis bullosa, junctional, Herlitz type, 226700 Laryngoonychocutaneous syndrome, 245660
LAMB1	142,9	100.0%	99.7%	Lissencephaly 5, 615191
LAMB2	166,5	100.0%	99.6%	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 Pierson syndrome, 609049
LAMB3	116,9	100.0%	99.4%	Amelogenesis imperfecta, type IA, 104530 Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LAMC2	100,5	99.7%	98.3%	Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LAMC3	148,1	99.7%	98.6%	Cortical malformations, occipital, 614115
LAMTOR2	172,2	100.0%	100.0%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LARGE1	115,2	100.0%	99.7%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840
LARP7	75,5	88.8%	75.2%	Alazami syndrome, 615071
LARS	131,5	99.7%	98.0%	?Infantile liver failure syndrome 1, 615438
LARS2	122,8	100.0%	100.0%	?Hydrops, lactic acidosis, and sideroblastic anemia, 617021 Perrault syndrome 4, 615300

LAT	114,4	100.0%	99.3%	Immunodeficiency 52, 617514
LBR	103	98.3%	91.5%	?Reynolds syndrome, 613471 Greenberg skeletal dysplasia, 215140 Pelger-Huet anomaly, 169400 Pelger-Huet anomaly with mild skeletal anomalies, 618019
LCA5	139,9	99.8%	98.9%	Leber congenital amaurosis 5, 604537
LCAT	140,7	99.4%	95.1%	Fish-eye disease, 136120 Norum disease, 245900
LCK	148,6	99.3%	97.3%	?Immunodeficiency 22, 615758
LCT	118,8	99.8%	97.9%	Lactase deficiency, congenital, 223000
LDHA	55,6	96.6%	88.0%	Glycogen storage disease XI, 612933
LDLRAP1	149	99.9%	99.1%	Hypercholesterolemia, familial, autosomal recessive, 603813
LEMD2	103,2	100.0%	99.6%	Cataract 46, juvenile-onset, 212500
LEP	174,7	100.0%	99.9%	Obesity, morbid, due to leptin deficiency, 614962
LEPR	106,5	94.2%	91.4%	Obesity, morbid, due to leptin receptor deficiency, 614963
LFNG	117,6	92.8%	87.7%	Spondylocostal dysostosis 3, autosomal recessive, 609813
LGI4	99,9	99.4%	96.7%	Arthrogryposis multiplex congenita, neurogenic, with myelin defect, 617468
LHB	23,5	92.3%	52.6%	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300
LHCGR	137	97.9%	94.1%	Leydig cell adenoma, somatic, with precocious puberty, 176410 Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320 Leydig cell hypoplasia with pseudohermaphroditism, 238320 Luteinizing hormone resistance, female, 238320 Precocious puberty, male, 176410
LHFPL5	207,7	100.0%	100.0%	Deafness, autosomal recessive 67, 610265
LHX3	116,2	96.6%	96.4%	Pituitary hormone deficiency, combined, 3, 221750
LIAS	125,3	99.9%	98.7%	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIFR	110,3	99.7%	97.4%	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LIG4	173,4	100.0%	99.8%	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500
LIM2	112,9	100.0%	99.5%	Cataract 19, multiple types, 615277
LIMS2	118,5	95.7%	93.2%	?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827
LINS1	133,6	100.0%	99.3%	Mental retardation, autosomal recessive 27, 614340
LIPA	105	96.5%	94.4%	Cholesteryl ester storage disease, 278000 Wolman disease, 278000
LIPC	98,5	100.0%	99.1%	Hepatic lipase deficiency, 614025 [High density lipoprotein cholesterol level QTL 12], 612797 {Diabetes mellitus, noninsulin-dependent}, 125853

LIPE	121,8	100.0%	99.5%	Lipodystrophy, familial partial, type 6, 615980
LIPH	120,2	100.0%	99.7%	Hypotrichosis 7, 604379 Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379
LIPN	113,9	100.0%	99.1%	Ichthyosis, congenital, autosomal recessive 8, 613943
LIPT1	203,2	100.0%	99.9%	Lipoyltransferase 1 deficiency, 616299
LIPT2	91,2	99.9%	99.3%	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668
LMAN1	144,3	99.8%	99.4%	Combined factor V and VIII deficiency, 227300
LMAN2L	109,3	100.0%	99.2%	?Mental retardation, autosomal recessive, 52, 616887
LMBR1	122,9	98.4%	96.3%	Acheiropody, 200500 Hypoplastic or aplastic tibia with polydactyly, 188740 Laurin-Sandrow syndrome, 135750 Polydactyly, preaxial type II, 174500 Syndactyly, type IV, 186200 Triphalangeal thumb, type I, 174500 Triphalangeal thumb-polysyndactyly syndrome, 174500
LMBRD1	100,1	98.9%	94.1%	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LMF1	136,8	100.0%	99.9%	Lipase deficiency, combined, 246650
LMNA	104,7	97.7%	91.9%	Cardiomyopathy, dilated, 1A, 115200 Charcot-Marie-Tooth disease, type 2B1, 605588 Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 Heart-hand syndrome, Slovenian type, 610140 Hutchinson-Gilford progeria, 176670 Lipodystrophy, familial partial, type 2, 151660 Malouf syndrome, 212112 Mandibuloacral dysplasia, 248370 Muscular dystrophy, congenital, 613205 Restrictive dermopathy, lethal, 275210
LMNB2	140,4	99.2%	97.4%	?Epilepsy, progressive myoclonic, 9, 616540 {Lipodystrophy, partial, acquired, susceptibility to}, 608709
LMOD3	128,6	100.0%	99.8%	Nemaline myopathy 10, 616165
LONP1	148	100.0%	100.0%	CODAS syndrome, 600373
LOXHD1	113,3	99.9%	99.5%	Deafness, autosomal recessive 77, 613079
LPAR6	100,3	99.8%	98.4%	Hypotrichosis 8, 278150 Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150
LPIN1	123,4	99.1%	96.4%	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	97,8	100.0%	99.6%	Majeed syndrome, 609628
LPL	128,2	100.0%	99.9%	Combined hyperlipidemia, familial, 144250

				Lipoprotein lipase deficiency, 238600 [High density lipoprotein cholesterol level QTL 11], 0
LRAT	240,7	100.0%	100.0%	Leber congenital amaurosis 14, 613341 Retinal dystrophy, early-onset severe, 613341 Retinitis pigmentosa, juvenile, 613341
LRBA	129,8	100.0%	99.6%	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRIG2	126,8	99.8%	99.1%	Urofacial syndrome 2, 615112
LRIT3	108	94.3%	93.1%	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRMDA	114,9	99.4%	97.6%	Albinism, oculocutaneous, type VII, 615179
LRP1	172,3	99.8%	99.3%	?Keratosis pilaris atrophicans, 604093
LRP2	139,2	100.0%	99.9%	Donnai-Barrow syndrome, 222448
LRP4	128	99.7%	99.0%	?Myasthenic syndrome, congenital, 17, 616304 Cenani-Lenz syndactyly syndrome, 212780 Sclerosteosis 2, 614305
LRP5	168,1	99.8%	98.7%	Exudative vitreoretinopathy 4, 601813 Hyperostosis, endosteal, 144750 Osteopetrosis, autosomal dominant 1, 607634 Osteoporosis-pseudoglioma syndrome, 259770 Osteosclerosis, 144750 Polycystic liver disease 4 with or without kidney cysts, 617875 van Buchem disease, type 2, 607636 [Bone mineral density variability 1], 601884 {Osteoporosis}, 166710
LRPAP1	141	100.0%	99.6%	Myopia 23, autosomal recessive, 615431
LRPPRC	129,3	100.0%	99.6%	Leigh syndrome, French-Canadian type, 220111
LRRC6	139,3	99.8%	97.3%	Ciliary dyskinesia, primary, 19, 614935
LRSAM1	135,4	100.0%	99.9%	Charcot-Marie-Tooth disease, axonal, type 2P, 614436
LRTOMT	114,4	99.1%	94.4%	Deafness, autosomal recessive 63, 611451
LSS	127,6	100.0%	99.7%	Cataract 44, 616509 Hypotrichosis 14, 618275
LTBP2	112,9	99.9%	99.3%	?Weill-Marchesani syndrome 3, recessive, 614819 Glaucoma 3, primary congenital, D, 613086 Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750
LTBP3	147,5	100.0%	99.6%	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
LTBP4	148	100.0%	99.4%	Cutis laxa, autosomal recessive, type IC, 613177
LTC4S	83,3	94.2%	79.5%	Leukotriene C4 synthase deficiency, 614037
LYRM4	77,5	67.4%	62.4%	?Combined oxidative phosphorylation deficiency 19, 615595

LYRM7	61,9	98.7%	91.3%	Mitochondrial complex III deficiency, nuclear type 8, 615838
LYST	136,3	99.4%	97.8%	Chediak-Higashi syndrome, 214500
LZTFL1	117	99.8%	99.2%	Bardet-Biedl syndrome 17, 615994
MAB21L2	237,8	100.0%	100.0%	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877
MAD2L2	139,1	100.0%	99.8%	?Fanconi anemia, complementation group V, 617243
MAG	160,7	100.0%	100.0%	Spastic paraplegia 75, autosomal recessive, 616680
MAGI2	90,4	94.6%	91.7%	Nephrotic syndrome, type 15, 617609
MAK	131,8	99.5%	97.4%	Retinitis pigmentosa 62, 614181
MALT1	129,4	93.0%	89.4%	Immunodeficiency 12, 615468
MAN1B1	125,5	100.0%	99.8%	Mental retardation, autosomal recessive 15, 614202
MAN2B1	128,6	99.9%	98.6%	Mannosidosis, alpha-, types I and II, 248500
MANBA	118,3	99.5%	97.5%	Mannosidosis, beta, 248510
MAP3K20	109,8	99.9%	98.9%	Centronuclear myopathy 6 with fiber-type disproportion, 617760 Split-foot malformation with mesoaxial polydactyly, 616890
MAPKBP1	132,5	100.0%	100.0%	Nephronophthisis 20, 617271
MAPT	151,6	99.9%	99.6%	Dementia, frontotemporal, with or without parkinsonism, 600274 Pick disease, 172700 Supranuclear palsy, progressive, 601104 Supranuclear palsy, progressive atypical, 260540 {Parkinson disease, susceptibility to}, 168600
MARS	100,4	99.9%	98.0%	Charcot-Marie-Tooth disease, axonal, type 2U, 616280 Interstitial lung and liver disease, 615486
MARS2	178,4	100.0%	100.0%	?Combined oxidative phosphorylation deficiency 25, 616430 Spastic ataxia 3, autosomal recessive, 611390
MARVELD2	139	98.8%	95.9%	Deafness, autosomal recessive 49, 610153
MASP1	131,1	100.0%	99.3%	3MC syndrome 1, 257920
MAT1A	144,1	99.8%	98.4%	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MATN3	99,6	86.5%	84.5%	?Spondyloepimetaphyseal dysplasia, 608728 Epiphyseal dysplasia, multiple, 5, 607078 {Osteoarthritis susceptibility 2}, 140600
MBOAT7	110	100.0%	99.7%	Mental retardation, autosomal recessive 57, 617188
MC2R	148,3	100.0%	99.2%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MCCC1	137,6	100.0%	99.4%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	119	100.0%	99.7%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	121,1	100.0%	99.9%	Methylmalonyl-CoA epimerase deficiency, 251120

MCFD2	91,4	97.6%	91.0%	Factor V and factor VIII, combined deficiency of, 613625
MCM4	133,1	100.0%	99.5%	Immunodeficiency 54, 609981
MCM5	122,5	100.0%	100.0%	?Meier-Gorlin syndrome 8, 617564
MCM9	128,7	99.9%	99.7%	Ovarian dysgenesis 4, 616185
MCOLN1	157,1	99.9%	99.0%	Mucolipidosis IV, 252650
MCPH1	133,1	99.9%	98.5%	Microcephaly 1, primary, autosomal recessive, 251200
MDH2	109,4	98.0%	97.9%	Epileptic encephalopathy, early infantile, 51, 617339
MECR	108,2	100.0%	99.7%	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MED17	132,4	97.5%	94.7%	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	133,6	99.8%	98.4%	Mental retardation, autosomal recessive 18, 614249
MED25	132,7	100.0%	99.7%	?Charcot-Marie-Tooth disease, type 2B2, 605589 Basel-Vanagait-Smirin-Yosef syndrome, 616449
MEFV	126,8	98.6%	96.5%	Familial Mediterranean fever, AD, 134610 Familial Mediterranean fever, AR, 249100
MEGF10	125,9	100.0%	99.8%	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399
MEGF8	144	100.0%	99.5%	Carpenter syndrome 2, 614976
MEOX1	105	99.9%	97.4%	Klippel-Feil syndrome 2, 214300
MERTK	128,2	99.4%	98.7%	Retinitis pigmentosa 38, 613862
MESP2	128	97.0%	94.9%	Spondylocostal dysostosis 2, autosomal recessive, 608681
MET	151,3	99.9%	99.3%	?Deafness, autosomal recessive 97, 616705 Hepatocellular carcinoma, childhood type, somatic, 114550 Renal cell carcinoma, papillary, 1, familial and somatic, 605074 {Osteofibrous dysplasia, susceptibility to}, 607278
METTL23	116,9	100.0%	100.0%	Mental retardation, autosomal recessive 44, 615942
MFF	86,2	93.7%	89.6%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFN2	122,8	100.0%	99.9%	Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260 Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 Hereditary motor and sensory neuropathy VIA, 601152
MFRP	128,1	100.0%	100.0%	Microphthalmia, isolated 5, 611040 Nanophthalmos 2, 609549
MFSD2A	114,3	100.0%	99.3%	Microcephaly 15, primary, autosomal recessive, 616486
MFSD8	121,3	100.0%	99.6%	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170
MGAT2	145,4	100.0%	100.0%	Congenital disorder of glycosylation, type IIa, 212066
MGME1	142,4	100.0%	99.9%	Mitochondrial DNA depletion syndrome 11, 615084
MGP	134,2	98.7%	94.6%	Keutel syndrome, 245150

MICU1	103,3	98.8%	96.5%	Myopathy with extrapyramidal signs, 615673
MIPEP	99,3	99.4%	96.6%	Combined oxidative phosphorylation deficiency 31, 617228
MITF	141,1	100.0%	99.8%	COMMAD syndrome, 617306 Tietz albinism-deafness syndrome, 103500 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456
MKKS	155,7	83.2%	83.2%	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKS1	92,4	99.6%	97.8%	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000
MLC1	96,7	100.0%	99.9%	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MLH1	139,2	99.9%	99.3%	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MLPH	97,4	99.7%	97.2%	Griscelli syndrome, type 3, 609227
MLYCD	95,7	99.4%	96.5%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	168,5	100.0%	100.0%	Methylmalonic aciduria, vitamin B12-responsive, 251100
MMAB	94,6	100.0%	99.7%	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110
MMACHC	196	100.0%	100.0%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	81,2	92.7%	79.5%	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410
MME	112,7	99.7%	98.1%	?Spinocerebellar ataxia 43, 617018 Charcot-Marie-Tooth disease, axonal, type 2T, 617017
MMP13	112,5	93.4%	92.1%	Metaphyseal anadysplasia 1, 602111 Metaphyseal dysplasia, Spahr type, 250400 Spondyloepimetaphyseal dysplasia, Missouri type, 602111
MMP14	148	100.0%	99.7%	?Winchester syndrome, 277950
MMP2	154,2	100.0%	100.0%	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP20	90,8	99.8%	97.6%	Amelogenesis imperfecta, type IIA2, 612529
MMP21	94,9	100.0%	98.0%	Heterotaxy, visceral, 7, autosomal, 616749
MMP9	143,9	100.0%	99.1%	Metaphyseal anadysplasia 2, 613073
MOCOS	147,2	99.9%	99.1%	Xanthinuria, type II, 603592
MOCS1	91,2	98.8%	95.7%	Molybdenum cofactor deficiency A, 252150

MOCS2	137,7	99.6%	99.5%	Molybdenum cofactor deficiency B, 252160
MOGS	141	100.0%	100.0%	Congenital disorder of glycosylation, type IIb, 606056
MPC1	153,1	100.0%	99.6%	Mitochondrial pyruvate carrier deficiency, 614741
MPDU1	102,4	100.0%	99.6%	Congenital disorder of glycosylation, type If, 609180
MPDZ	128,3	99.6%	98.2%	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219
MPI	110,1	100.0%	99.9%	Congenital disorder of glycosylation, type Ib, 602579
MPL	125,8	100.0%	99.8%	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498
MPLKIP	104,3	100.0%	99.9%	Trichothiodystrophy 4, nonphotosensitive, 234050
MPO	153,5	99.9%	99.4%	Myeloperoxidase deficiency, 254600 {Alzheimer disease, susceptibility to}, 104300 {Lung cancer, protection against, in smokers}, 0
MPV17	88,6	100.0%	98.9%	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MPZ	125	100.0%	98.9%	Charcot-Marie-Tooth disease, dominant intermediate D, 607791 Charcot-Marie-Tooth disease, type 1B, 118200 Charcot-Marie-Tooth disease, type 2I, 607677 Charcot-Marie-Tooth disease, type 2J, 607736 Dejerine-Sottas disease, 145900 Hypomyelinating neuropathy, congenital, 2, 618184 Roussy-Levy syndrome, 180800
MRAP	163,4	100.0%	100.0%	Glucocorticoid deficiency 2, 607398
MRE11	49,7	97.3%	86.0%	Ataxia-telangiectasia-like disorder 1, 604391
MRPL3	63,4	93.2%	81.4%	Combined oxidative phosphorylation deficiency 9, 614582
MRPL44	126,3	100.0%	99.7%	?Combined oxidative phosphorylation deficiency 16, 615395
MRPS16	127	99.9%	98.5%	Combined oxidative phosphorylation deficiency 2, 610498
MRPS2	158,6	100.0%	99.9%	Combined oxidative phosphorylation deficiency 36, 617950
MRPS22	134,6	99.9%	98.7%	Combined oxidative phosphorylation deficiency 5, 611719 Ovarian dysgenesis 7, 618117
MRPS34	169	100.0%	99.9%	Combined oxidative phosphorylation deficiency 32, 617664
MRPS7	153,2	100.0%	100.0%	?Combined oxidative phosphorylation deficiency 34, 617872
MS4A1	127,6	99.9%	98.8%	Immunodeficiency, common variable, 5, 613495
MSH2	111,7	99.4%	96.4%	Colorectal cancer, hereditary nonpolyposis, type 1, 120435 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MSH3	139,6	99.8%	99.2%	Endometrial carcinoma, somatic, 608089

				Familial adenomatous polyposis 4, 617100
MSH6	165,1	100.0%	100.0%	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 Mismatch repair cancer syndrome, 276300 {Endometrial cancer, familial}, 608089
MSMO1	51,6	95.8%	88.5%	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MSRB3	131,9	99.9%	99.6%	Deafness, autosomal recessive 74, 613718
MSTO1	101,4	99.3%	96.1%	Myopathy, mitochondrial, and ataxia, 617675
MTFMT	132,5	100.0%	99.8%	Combined oxidative phosphorylation deficiency 15, 614947 Mitochondrial complex I deficiency, nuclear type 27, 618248
MTHFD1	115,4	99.8%	97.4%	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTHFR	114,9	98.2%	96.4%	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}, 0
MTMR2	99,1	99.9%	98.5%	Charcot-Marie-Tooth disease, type 4B1, 601382
MTO1	143,7	91.4%	89.6%	Combined oxidative phosphorylation deficiency 10, 614702
MTPAP	122,3	99.0%	93.2%	?Spastic ataxia 4, autosomal recessive, 613672
MTR	131,4	99.9%	99.4%	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTRR	131,1	100.0%	99.0%	Homocystinuria-megaloblastic anemia, cbl E type, 236270 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTTP	114,7	99.9%	99.4%	Abetalipoproteinemia, 200100 {Metabolic syndrome, protection against}, 605552
MUSK	131,5	100.0%	100.0%	Fetal akinesia deformation sequence 1, 208150 Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325
MUT	NC	NC	NC	Methylmalonic aciduria, mut(0) type, 251000
MUTYH	152	100.0%	100.0%	Adenomas, multiple colorectal, 608456 Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600 Gastric cancer, somatic, 613659
MVK	121,4	91.0%	90.5%	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900
MYBPC1	127,8	99.9%	99.2%	Arthrogryposis, distal, type 1B, 614335 Lethal congenital contracture syndrome 4, 614915
MYD88	194,7	100.0%	99.8%	Macroglobulinemia, Waldenstrom, somatic, 153600 Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260

MYH2	108,2	100.0%	99.5%	Proximal myopathy and ophthalmoplegia, 605637
MYL3	98	100.0%	100.0%	Cardiomyopathy, hypertrophic, 8, 608751
MYMK	140,5	100.0%	100.0%	Carey-Fineman-Ziter syndrome, 254940
MYO15A	143,7	99.8%	98.8%	Deafness, autosomal recessive 3, 600316
MYO18B	122,1	99.9%	99.2%	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549
MYO1E	115,4	99.8%	98.7%	Glomerulosclerosis, focal segmental, 6, 614131
MYO3A	113	99.2%	95.3%	Deafness, autosomal recessive 30, 607101
MYO5A	109	99.7%	98.6%	Griscelli syndrome, type 1, 214450
MYO5B	108,3	97.9%	94.7%	Microvillus inclusion disease, 251850
MYO6	101,5	99.5%	96.4%	Deafness, autosomal dominant 22, 606346 Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346 Deafness, autosomal recessive 37, 607821
MYO7A	125,2	99.8%	98.5%	Deafness, autosomal dominant 11, 601317 Deafness, autosomal recessive 2, 600060 Usher syndrome, type 1B, 276900
MYPN	124,8	99.9%	99.0%	Cardiomyopathy, dilated, 1KK, 615248 Cardiomyopathy, familial restrictive, 4, 615248 Cardiomyopathy, hypertrophic, 22, 615248 Nemaline myopathy 11, autosomal recessive, 617336
NADK2	163,3	99.9%	99.0%	?2,4-dienoyl-CoA reductase deficiency, 616034
NAGA	121,7	100.0%	100.0%	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241
NAGLU	117,7	97.1%	94.1%	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NAGS	104,3	100.0%	99.9%	N-acetylglutamate synthase deficiency, 237310
NALCN	117	99.7%	98.7%	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419
NANS	97,2	99.9%	98.4%	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NARS2	120,7	97.6%	97.2%	Combined oxidative phosphorylation deficiency 24, 616239
NAT8L	91,4	98.8%	94.6%	?N-acetylaspartate deficiency, 614063
NAXE	81,4	99.7%	97.0%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NBAS	138,5	99.9%	99.1%	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NBEAL2	166	100.0%	99.5%	Gray platelet syndrome, 139090
NBN	93,8	99.8%	98.4%	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065

				Nijmegen breakage syndrome, 251260
NCAPD2	118,1	100.0%	99.4%	?Microcephaly 21, primary, autosomal recessive, 617983
NCAPD3	100,1	99.8%	98.2%	Microcephaly 22, primary, autosomal recessive, 617984
NCAPH	119,6	100.0%	99.9%	?Microcephaly 23, primary, autosomal recessive, 617985
NCF1	23,1	27.8%	22.5%	Chronic granulomatous disease due to deficiency of NCF-1, 233700
NCF2	109,2	99.8%	98.2%	Chronic granulomatous disease due to deficiency of NCF-2, 233710
NCF4	148,6	100.0%	100.0%	?Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III, 613960
NDE1	89,3	100.0%	99.6%	?Microhydranencephaly, 605013 Lissencephaly 4 (with microcephaly), 614019
NDRG1	114	100.0%	100.0%	Charcot-Marie-Tooth disease, type 4D, 601455
NDST1	188,4	100.0%	100.0%	Mental retardation, autosomal recessive 46, 616116
NDUFA10	114,6	99.9%	98.9%	Mitochondrial complex I deficiency, nuclear type 22, 618243
NDUFA11	116	99.8%	97.4%	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFA12	160,8	100.0%	100.0%	?Mitochondrial complex I deficiency, nuclear type 23, 618244
NDUFA13	121,3	92.3%	91.7%	?Mitochondrial complex I deficiency, nuclear type 28, 618249 {Thyroid carcinoma, Hurthle cell}, 607464
NDUFA2	162,6	100.0%	99.6%	?Mitochondrial complex I deficiency, nuclear type 13, 618235
NDUFA9	101,6	99.7%	96.5%	Mitochondrial complex I deficiency, nuclear type 26, 618247
NDUFAF1	98,5	100.0%	99.9%	Mitochondrial complex I deficiency, nuclear type 11, 618234
NDUFAF2	54,1	94.3%	82.0%	Mitochondrial complex I deficiency, nuclear type 10, 618233
NDUFAF3	141	100.0%	99.9%	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF4	98,3	99.2%	94.5%	Mitochondrial complex I deficiency, nuclear type 15, 618237
NDUFAF5	124,9	99.9%	99.1%	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFAF6	91,9	99.8%	98.5%	Mitochondrial complex I deficiency, nuclear type 17, 618239
NDUFB11	103,3	98.6%	95.0%	?Mitochondrial complex I deficiency, nuclear type 30, 301021 Linear skin defects with multiple congenital anomalies 3, 300952
NDUFB3	23,3	89.7%	62.5%	Mitochondrial complex I deficiency, nuclear type 25, 618246
NDUFB8	105,3	100.0%	99.8%	Mitochondrial complex I deficiency, nuclear type 32, 618252
NDUFB9	105,2	97.8%	93.3%	?Mitochondrial complex I deficiency, nuclear type 24, 618245
NDUFS1	143,5	99.9%	99.8%	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	100,1	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	124,8	90.7%	90.5%	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	144,5	100.0%	99.7%	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS6	111,9	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFS7	140,5	100.0%	99.9%	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFS8	156,8	100.0%	99.7%	Mitochondrial complex I deficiency, nuclear type 2, 618222
NDUFV1	141,7	99.9%	98.8%	Mitochondrial complex I deficiency, nuclear type 4, 618225

NDUFV2	74,2	92.4%	77.3%	Mitochondrial complex I deficiency, nuclear type 7, 618229
NEB	100,1	83.0%	82.4%	Nemaline myopathy 2, autosomal recessive, 256030
NECAP1	102,3	100.0%	100.0%	?Epileptic encephalopathy, early infantile, 21, 615833
NECTIN1	134	100.0%	99.9%	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060
NECTIN4	121,6	100.0%	99.9%	Ectodermal dysplasia-syndactyly syndrome 1, 613573
NEK1	115,9	99.7%	98.1%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520 {Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892
NEK2	86,4	98.6%	92.4%	?Retinitis pigmentosa 67, 615565
NEK8	141,3	100.0%	99.9%	?Nephronophthisis 9, 613824 Renal-hepatic-pancreatic dysplasia 2, 615415
NEK9	118,9	99.8%	98.2%	?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262 Lethal congenital contracture syndrome 10, 617022 Nevus comedonicus, somatic, 617025
NEU1	141,3	99.3%	96.4%	Sialidosis, type I, 256550 Sialidosis, type II, 256550
NEUROG3	172,9	100.0%	100.0%	Diarrhea 4, malabsorptive, congenital, 610370
NFU1	61,8	97.4%	82.1%	Multiple mitochondrial dysfunctions syndrome 1, 605711
NGF	199	100.0%	100.0%	Neuropathy, hereditary sensory and autonomic, type V, 608654
NGLY1	135,4	100.0%	99.7%	Congenital disorder of deglycosylation, 615273
NHEJ1	58,5	99.7%	92.8%	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
NHLRC1	169,7	100.0%	100.0%	Epilepsy, progressive myoclonic 2B (Lafora), 254780
NHP2	121,9	100.0%	99.2%	Dyskeratosis congenita, autosomal recessive 2, 613987
NIN	127	99.9%	99.4%	?Seckel syndrome 7, 614851
NIPAL4	126,7	100.0%	99.3%	Ichthyosis, congenital, autosomal recessive 6, 612281
NKX2-6	139,9	100.0%	100.0%	Conotruncal heart malformations, 217095 Persistent truncus arteriosus, 217095
NKX3-2	138,4	100.0%	99.8%	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330
NKX6-2	122,9	98.1%	91.2%	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560
NLRP1	117,7	99.5%	97.6%	Autoinflammation with arthritis and dyskeratosis, 617388 Palmoplantar carcinoma, multiple self-healing, 615225 {Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579
NLRP7	124,5	99.9%	98.8%	Hydatidiform mole, recurrent, 1, 231090
NME8	104,7	98.6%	93.8%	Ciliary dyskinesia, primary, 6, 610852
NMNAT1	113,5	100.0%	98.5%	Leber congenital amaurosis 9, 608553
NNT	124,6	100.0%	98.5%	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736

NOP10	120,5	100.0%	100.0%	Dyskeratosis congenita, autosomal recessive 1, 224230
NPC1	117,8	100.0%	99.2%	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220
NPC2	124,7	100.0%	99.9%	Niemann-pick disease, type C2, 607625
NPHP1	121,2	99.8%	98.5%	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NPHP3	121,4	99.8%	98.5%	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540
NPHP4	125,6	100.0%	99.7%	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
NPHS1	106,3	100.0%	99.5%	Nephrotic syndrome, type 1, 256300
NPHS2	114,5	100.0%	99.5%	Nephrotic syndrome, type 2, 600995
NPPA	158,5	100.0%	100.0%	Atrial fibrillation, familial, 6, 612201 Atrial standstill 2, 615745
NPR2	144,1	100.0%	99.4%	Acromesomelic dysplasia, Maroteaux type, 602875 Epiphyseal chondrodysplasia, Miura type, 615923 Short stature with nonspecific skeletal abnormalities, 616255
NR0B2	102,8	100.0%	99.7%	Obesity, mild, early-onset, 601665
NR1H4	122,3	99.8%	98.3%	Cholestasis, progressive familial intrahepatic, 5, 617049
NR2E3	105,1	99.9%	98.6%	Enhanced S-cone syndrome, 268100 Retinitis pigmentosa 37, 611131
NRXN1	141,6	97.6%	97.3%	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332
NSMCE2	81,4	99.9%	98.6%	Seckel syndrome 10, 617253
NSMCE3	194	100.0%	100.0%	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241
NSUN2	94,7	97.6%	93.5%	Mental retardation, autosomal recessive 5, 611091
NT5C2	121,2	97.9%	96.3%	Spastic paraplegia 45, autosomal recessive, 613162
NT5C3A	64,1	97.2%	85.7%	Anemia, hemolytic, due to UMPH1 deficiency, 266120
NT5E	151,4	100.0%	99.9%	Calcification of joints and arteries, 211800
NTHL1	121,6	100.0%	100.0%	Familial adenomatous polyposis 3, 616415
NTRK1	133	100.0%	99.3%	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma, familial, 155240
NUBPL	102	98.9%	95.5%	Mitochondrial complex I deficiency, nuclear type 21, 618242
NUP107	126,6	99.8%	98.6%	?Ovarian dysgenesis 6, 618078 Galloway-Mowat syndrome 7, 618348

				Nephrotic syndrome, type 11, 616730
NUP205	133,5	99.6%	98.7%	?Nephrotic syndrome, type 13, 616893
NUP62	111,8	100.0%	100.0%	Striatonigral degeneration, infantile, 271930
NUP93	117,8	96.9%	93.8%	Nephrotic syndrome, type 12, 616892
NUS1	53,3	71.5%	44.1%	?Congenital disorder of glycosylation, type 1aa, 617082 Mental retardation, autosomal dominant 55, with seizures, 617831
OAT	68,2	81.7%	70.1%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OBSL1	147,2	100.0%	99.8%	3-M syndrome 2, 612921
OCA2	116,8	99.7%	97.7%	Albinism, brown oculocutaneous, 203200 Albinism, oculocutaneous, type II, 203200 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220
OCLN	173,9	100.0%	100.0%	Pseudo-TORCH syndrome 1, 251290
OGDH	172,3	99.9%	99.4%	Alpha-ketoglutarate dehydrogenase deficiency, 203740
OPA1	124,7	99.7%	97.4%	?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896 Behr syndrome, 210000 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250 {Glaucoma, normal tension, susceptibility to}, 606657
OPA3	156,6	100.0%	99.2%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPLAH	141,5	100.0%	99.9%	5-oxoprolinase deficiency, 260005
ORAI1	198,9	99.8%	98.2%	Immunodeficiency 9, 612782 Myopathy, tubular aggregate, 2, 615883
ORC1	90,3	99.9%	98.7%	Meier-Gorlin syndrome 1, 224690
ORC4	73,6	98.1%	92.0%	Meier-Gorlin syndrome 2, 613800
ORC6	127,6	100.0%	99.9%	Meier-Gorlin syndrome 3, 613803
OSGEP	98,1	100.0%	97.3%	Galloway-Mowat syndrome 3, 617729
OSTM1	109,3	98.2%	92.5%	Osteopetrosis, autosomal recessive 5, 259720
OTOA	97,2	99.7%	98.3%	Deafness, autosomal recessive 22, 607039
OTOF	135,6	100.0%	99.8%	Auditory neuropathy, autosomal recessive, 1, 601071 Deafness, autosomal recessive 9, 601071
OTOG	134,6	99.6%	98.8%	Deafness, autosomal recessive 18B, 614945
OTOGL	104,6	99.4%	97.0%	Deafness, autosomal recessive 84B, 614944
OTUD6B	117,7	99.8%	99.3%	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452
OTULIN	132,6	98.7%	95.2%	Autoinflammation, panniculitis, and dermatosis syndrome, 617099
OXCT1	125,5	99.7%	98.2%	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050

P2RY12	186,2	100.0%	100.0%	Bleeding disorder, platelet-type, 8, 609821
P3H1	129,3	100.0%	100.0%	Osteogenesis imperfecta, type VIII, 610915
P3H2	98,8	99.9%	99.4%	Myopia, high, with cataract and vitreoretinal degeneration, 614292
PAH	126,4	100.0%	100.0%	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600
PAM16	64,5	66.4%	65.3%	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320
PANK2	154,1	100.0%	100.0%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PAPSS2	103,8	99.7%	97.7%	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847
PARK7	83,5	100.0%	99.8%	Parkinson disease 7, autosomal recessive early-onset, 606324
PARN	127,3	99.9%	99.5%	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
PATL2	93,7	99.8%	96.2%	Oocyte maturation defect 4, 617743
PAX1	189,5	97.0%	92.1%	?Otofaciocervical syndrome 2, 615560
PAX3	106,9	100.0%	99.7%	Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220 Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820
PC	155,4	99.9%	98.7%	Pyruvate carboxylase deficiency, 266150
PCBD1	103,9	100.0%	99.7%	Hyperphenylalaninemia, BH4-deficient, D, 264070
PCCA	99,2	99.3%	95.5%	Propionicacidemia, 606054
PCCB	111,8	99.3%	96.9%	Propionicacidemia, 606054
PCDH12	182,1	100.0%	100.0%	Microcephaly, seizures, spasticity, and brain calcification, 251280
PCDH15	140,3	99.2%	99.0%	Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1F, 602083
PCK1	119,4	100.0%	99.9%	?Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680
PCK2	164,9	100.0%	100.0%	PEPCK deficiency, mitochondrial, 261650
PCLO	142,1	99.8%	99.0%	?Pontocerebellar hypoplasia, type 3, 608027
PCNA	92	100.0%	98.2%	?Ataxia-telangiectasia-like disorder 2, 615919
PCNT	115,4	99.7%	97.7%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PCSK1	141,9	100.0%	99.2%	Obesity with impaired prohormone processing, 600955 {Obesity, susceptibility to, BMIQ12}, 612362
PCYT1A	95,6	97.9%	94.4%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDE10A	107,6	81.4%	80.4%	Dyskinesia, limb and orofacial, infantile-onset, 616921 Striatal degeneration, autosomal dominant, 616922
PDE6A	102,6	100.0%	99.2%	Retinitis pigmentosa 43, 613810

PDE6B	157,3	100.0%	100.0%	Night blindness, congenital stationary, autosomal dominant 2, 163500 Retinitis pigmentosa-40, 613801
PDE6C	116,2	99.5%	97.2%	Cone dystrophy 4, 613093
PDE6D	114,7	100.0%	99.9%	?Joubert syndrome 22, 615665
PDE6G	125,8	100.0%	99.5%	Retinitis pigmentosa 57, 613582
PDE6H	58,2	98.5%	76.0%	Achromatopsia 6, 610024 Retinal cone dystrophy 3, 610024
PDHB	111,4	99.2%	97.2%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	129	99.9%	99.5%	Lacticacidemia due to PDX1 deficiency, 245349
PDP1	129,1	100.0%	100.0%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	104,8	96.7%	87.7%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	112,9	99.6%	96.1%	Coenzyme Q10 deficiency, primary, 3, 614652
PDX1	72,9	99.1%	95.2%	MODY, type IV, 606392 Pancreatic agenesis 1, 260370 {Diabetes mellitus, type II, susceptibility to}, 125853
PDZD7	93,9	99.6%	97.8%	Deafness, autosomal recessive 57, 618003 Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472 {Retinal disease in Usher syndrome type IIA, modifier of}, 276901
PEPD	117,4	100.0%	99.6%	Prolidase deficiency, 170100
PET100	87,9	98.0%	87.6%	Mitochondrial complex IV deficiency, 220110
PEX1	127,9	99.9%	99.3%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	113,3	99.9%	97.4%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX11B	87,9	100.0%	99.4%	?Peroxisome biogenesis disorder 14B, 614920
PEX12	120,6	100.0%	100.0%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	179,6	100.0%	100.0%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	130,5	99.8%	97.8%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	140,8	98.6%	94.8%	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	84,9	100.0%	98.9%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	134,9	100.0%	100.0%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	94,3	100.0%	99.6%	Peroxisome biogenesis disorder 7A (Zellweger), 614872

				Peroxisome biogenesis disorder 7B, 614873
PEX3	113,9	99.9%	99.2%	?Peroxisome biogenesis disorder 10B, 617370 Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	107,9	100.0%	99.2%	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	106,5	98.5%	92.0%	Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863
PEX7	111	91.2%	89.3%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PFKM	113,7	100.0%	99.2%	Glycogen storage disease VII, 232800
PGAM2	163,6	100.0%	100.0%	Glycogen storage disease X, 261670
PGAP1	110,9	99.1%	95.8%	Mental retardation, autosomal recessive 42, 615802
PGAP2	134,7	100.0%	99.5%	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGAP3	70,3	63.5%	59.9%	Hyperphosphatasia with mental retardation syndrome 4, 615716
PGM1	128,8	100.0%	99.8%	Congenital disorder of glycosylation, type It, 614921
PGM3	149,3	99.9%	99.6%	Immunodeficiency 23, 615816
PHGDH	106,6	100.0%	99.3%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHKB	125,3	99.9%	99.1%	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750
PHKG2	155,6	100.0%	100.0%	Cirrhosis due to liver phosphorylase kinase deficiency, 0 Glycogen storage disease IXc, 613027
PHOX2A	56	98.6%	88.9%	Fibrosis of extraocular muscles, congenital, 2, 602078
PHYH	74	99.9%	96.9%	Refsum disease, 266500
PI4KA	91,8	93.7%	89.7%	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531
PIBF1	74,3	99.2%	94.4%	Joubert syndrome 33, 617767
PIEZO2	104,2	99.9%	99.2%	?Marden-Walker syndrome, 248700 Arthrogryposis, distal, type 3, 114300 Arthrogryposis, distal, type 5, 108145 Arthrogryposis, distal, with impaired proprioception and touch, 617146
PIGC	85,9	99.3%	92.2%	Glycosylphosphatidylinositol biosynthesis defect 16, 617816
PIGG	143,4	100.0%	99.5%	Mental retardation, autosomal recessive 53, 616917
PIGH	93,8	78.8%	67.3%	Glycosylphosphatidylinositol biosynthesis defect 17, 618010
PIGL	122,1	99.7%	99.6%	CHIME syndrome, 280000
PIGM	148,9	100.0%	100.0%	Glycosylphosphatidylinositol deficiency, 610293
PIGN	106,3	93.6%	91.1%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080

PIGO	144,5	100.0%	99.9%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGP	89,3	94.8%	86.0%	?Epileptic encephalopathy, early infantile, 55, 617599
PIGT	159,3	98.1%	98.1%	?Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGV	124,4	100.0%	100.0%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIGW	145	100.0%	99.8%	Glycosylphosphatidylinositol biosynthesis defect 11, 616025
PIGY	90,3	100.0%	99.9%	Hyperphosphatasia with mental retardation syndrome 6, 616809
PIK3R1	124,3	99.9%	98.9%	?Agammaglobulinemia 7, autosomal recessive, 615214 Immunodeficiency 36, 616005 SHORT syndrome, 269880
PIK3R5	120,7	100.0%	99.9%	Ataxia-oculomotor apraxia 3, 615217
PINK1	87,3	96.4%	90.7%	Parkinson disease 6, early onset, 605909
PIP5K1C	136,6	99.8%	97.6%	Lethal congenital contractural syndrome 3, 611369
PKD1L1	108,7	100.0%	99.3%	Heterotaxy, visceral, 8, autosomal, 617205
PKHD1	130,4	99.9%	99.4%	Polycystic kidney disease 4, with or without hepatic disease, 263200
PKLR	169,2	100.0%	99.7%	Adenosine triphosphate, elevated, of erythrocytes, 102900 Pyruvate kinase deficiency, 266200
PKP1	120,8	99.8%	98.4%	Ectodermal dysplasia/skin fragility syndrome, 604536
PLA2G6	111,9	99.8%	98.2%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953
PLA2G7	120,9	99.8%	98.6%	Platelet-activating factor acetylhydrolase deficiency, 614278 {Asthma, susceptibility to}, 600807 {Atopy, susceptibility to}, 147050
PLAA	163	99.7%	98.6%	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527
PLCB1	134,9	100.0%	99.7%	Epileptic encephalopathy, early infantile, 12, 613722
PLCD1	116,1	100.0%	99.3%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCE1	125	99.8%	99.0%	Nephrotic syndrome, type 3, 610725
PLD1	116,4	99.9%	99.3%	Cardiac valvular defect, developmental, 212093
PLEC	144,1	100.0%	100.0%	?Epidermolysis bullosa simplex with nail dystrophy, 616487 Epidermolysis bullosa simplex with muscular dystrophy, 226670 Epidermolysis bullosa simplex with pyloric atresia, 612138 Epidermolysis bullosa simplex, Ogna type, 131950 Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723
PLEKHG2	152,2	100.0%	99.0%	Leukodystrophy and acquired microcephaly with or without dystonia, 616763
PLEKHG5	101,6	99.8%	97.7%	Charcot-Marie-Tooth disease, recessive intermediate C, 615376 Spinal muscular atrophy, distal, autosomal recessive, 4, 611067

PLEKHM1	127,8	100.0%	99.9%	?Osteopetrosis, autosomal recessive 6, 611497 Osteopetrosis, autosomal dominant 3, 618107
PLG	93,4	87.8%	86.8%	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PLK4	149,7	99.8%	98.2%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLOD1	131,9	99.8%	97.3%	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD2	121,3	99.6%	97.3%	Bruck syndrome 2, 609220
PLOD3	109,7	100.0%	99.9%	Lysyl hydroxylase 3 deficiency, 612394
PLPBP	95,3	99.6%	95.3%	Epilepsy, early-onset, vitamin B6-dependent, 617290
PMM2	127,7	100.0%	99.7%	Congenital disorder of glycosylation, type Ia, 212065
PMPCA	108,1	99.1%	95.9%	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMPCB	121,6	100.0%	99.2%	Multiple mitochondrial dysfunctions syndrome 6, 617954
PMS2	94,7	83.4%	81.0%	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 Mismatch repair cancer syndrome, 276300
PNKP	109	100.0%	99.9%	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNLIP	133,9	100.0%	97.7%	?Pancreatic lipase deficiency, 614338
PNP	108,6	100.0%	99.5%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA1	164,8	100.0%	100.0%	Ichthyosis, congenital, autosomal recessive 10, 615024
PNPLA2	142,7	100.0%	99.8%	Neutral lipid storage disease with myopathy, 610717
PNPLA6	137,9	99.9%	99.5%	?Laurence-Moon syndrome, 245800 Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 Spastic paraplegia 39, autosomal recessive, 612020
PNPLA8	121,2	100.0%	99.7%	?Mitochondrial myopathy with lactic acidosis, 251950
PNPO	74,4	100.0%	99.3%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
PNPT1	56,1	96.2%	84.3%	Combined oxidative phosphorylation deficiency 13, 614932 Deafness, autosomal recessive 70, 614934
POC1A	112,9	100.0%	100.0%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POC1B	79,6	99.7%	97.9%	Cone-rod dystrophy 20, 615973
POGLUT1	101,2	100.0%	98.7%	?Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232 Dowling-Degos disease 4, 615696
POLG	113,9	100.0%	99.6%	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Progressive external ophthalmoplegia, autosomal recessive 1, 258450

POLH	116,2	99.9%	98.6%	Xeroderma pigmentosum, variant type, 278750
POLR1C	98,3	98.9%	94.9%	Leukodystrophy, hypomyelinating, 11, 616494 Treacher Collins syndrome 3, 248390
POLR1D	183,1	91.6%	91.6%	Treacher Collins syndrome 2, 613717
POLR3A	116,8	100.0%	99.9%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 Wiedemann-Rautenstrauch syndrome, 264090
POLR3B	129,8	99.7%	98.2%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMC	148,2	100.0%	100.0%	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734 {Obesity, early-onset, susceptibility to}, 601665
POMGNT1	115,5	100.0%	99.6%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 Retinitis pigmentosa 76, 617123
POMGNT2	201,7	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830 Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135
POMK	138,7	100.0%	100.0%	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249
POMP	124,6	99.9%	97.6%	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952 Proteasome-associated autoinflammatory syndrome 2, 618048
POMT1	130,6	99.7%	97.8%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308
POMT2	103,3	100.0%	98.4%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158
POP1	120,1	100.0%	99.4%	Anauxetic dysplasia 2, 617396
POR	175,5	99.2%	97.1%	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571
POU1F1	109,1	99.9%	98.2%	Pituitary hormone deficiency, combined, 1, 613038
PPA2	92,3	98.8%	91.7%	?Sudden cardiac failure, alcohol-induced, 617223 Sudden cardiac failure, infantile, 617222
PPIB	106,9	100.0%	100.0%	Osteogenesis imperfecta, type IX, 259440
PPP1R15B	124	100.0%	99.9%	Microcephaly, short stature, and impaired glucose metabolism 2, 616817
PPT1	136,6	90.2%	89.2%	Ceroid lipofuscinosis, neuronal, 1, 256730
PRCD	95,6	100.0%	100.0%	Retinitis pigmentosa 36, 610599
PRDM12	138,9	92.6%	90.4%	Neuropathy, hereditary sensory and autonomic, type VIII, 616488
PRDM5	137,8	99.6%	98.7%	Brittle cornea syndrome 2, 614170

PRDM8	116,4	99.7%	95.9%	?Epilepsy, progressive myoclonic, 10, 616640
PRDX1	95	100.0%	99.7%	Methylmalonic aciduria and homocystinuria, cblC type, digenic, 277400
PREPL	103,9	99.7%	97.7%	Myasthenic syndrome, congenital, 22, 616224
PRF1	138,1	91.2%	90.6%	Aplastic anemia, 609135 Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027
PRG4	131,1	99.2%	92.4%	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250
PRICKLE1	100	100.0%	99.8%	Epilepsy, progressive myoclonic 1B, 612437
PRKCD	164,7	100.0%	100.0%	Autoimmune lymphoproliferative syndrome, type III, 615559
PRKDC	97,9	99.3%	96.5%	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PRKN	82,1	79.9%	78.1%	Adenocarcinoma of lung, somatic, 211980 Ovarian cancer, somatic, 167000 Parkinson disease, juvenile, type 2, 600116
PRKRA	190,7	100.0%	100.0%	Dystonia 16, 612067
PRMT7	119,1	100.0%	99.9%	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157
PROC	142,6	100.0%	100.0%	Thrombophilia due to protein C deficiency, autosomal dominant, 176860 Thrombophilia due to protein C deficiency, autosomal recessive, 612304
PRODH	81,8	89.0%	81.7%	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850
PROM1	107,4	97.8%	95.8%	Cone-rod dystrophy 12, 612657 Macular dystrophy, retinal, 2, 608051 Retinitis pigmentosa 41, 612095 Stargardt disease 4, 603786
PROP1	96,9	92.5%	83.7%	Pituitary hormone deficiency, combined, 2, 262600
PROS1	95,8	97.7%	92.7%	Thrombophilia due to protein S deficiency, autosomal dominant, 612336 Thrombophilia due to protein S deficiency, autosomal recessive, 614514
PRSS12	137,8	100.0%	99.6%	Mental retardation, autosomal recessive 1, 249500
PRSS56	95,4	100.0%	99.2%	Microphthalmia, isolated 6, 613517
PRUNE1	118	100.0%	99.3%	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481
PRX	156,5	100.0%	99.9%	Charcot-Marie-Tooth disease, type 4F, 614895 Dejerine-Sottas disease, 145900
PSAP	98,1	100.0%	99.3%	Combined SAP deficiency, 611721 Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PSAT1	42,8	90.3%	72.5%	?Phosphoserine aminotransferase deficiency, 610992 Neu-Laxova syndrome 2, 616038

PSMB4	119,2	100.0%	99.9%	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591
PSMB8	113,5	100.0%	98.8%	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040
PSMB9	81,5	99.9%	97.7%	?Proteasome-associated autoinflammatory syndrome 3, digenic, 617591
PSMC3IP	104,9	100.0%	100.0%	Ovarian dysgenesis 3, 614324
PSPH	126,6	100.0%	99.8%	Phosphoserine phosphatase deficiency, 614023
PTF1A	120,6	99.9%	98.2%	Pancreatic agenesis 2, 615935 Pancreatic and cerebellar agenesis, 609069
PTH1R	106,6	100.0%	99.1%	Chondrodysplasia, Blomstrand type, 215045 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Metaphyseal chondrodysplasia, Murk Jansen type, 156400
PTPN14	159,2	99.3%	96.8%	Choanal atresia and lymphedema, 613611
PTPRC	100,6	98.7%	93.9%	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971 {Hepatitis C virus, susceptibility to}, 609532
PTPRO	128,1	99.9%	99.4%	Nephrotic syndrome, type 6, 614196
PTPRQ	102,5	94.5%	92.3%	Deafness, autosomal dominant 73, 617663 Deafness, autosomal recessive 84A, 613391
PTRH2	200,8	100.0%	100.0%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PTS	101,5	99.8%	98.4%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUS1	113,3	99.8%	97.5%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PUS3	158,3	100.0%	100.0%	Mental retardation, autosomal recessive 55, 617051
PXDN	138,4	100.0%	99.6%	Anterior segment dysgenesis 7, with sclerocornea, 269400
PYCR1	96	99.7%	97.4%	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
PYCR2	116,5	99.7%	96.9%	Leukodystrophy, hypomyelinating, 10, 616420
PYGL	141	100.0%	100.0%	Glycogen storage disease VI, 232700
PYGM	121,1	100.0%	99.9%	McArdle disease, 232600
PYROXD1	48,6	93.0%	78.5%	Myopathy, myofibrillar, 8, 617258
QARS	129,2	100.0%	99.8%	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	97,9	100.0%	99.2%	Hyperphenylalaninemia, BH4-deficient, C, 261630
RAB18	83,4	99.7%	97.2%	Warburg micro syndrome 3, 614222
RAB23	107,4	100.0%	99.2%	Carpenter syndrome, 201000
RAB27A	126,1	100.0%	99.8%	Griscelli syndrome, type 2, 607624
RAB28	66,6	98.9%	92.3%	Cone-rod dystrophy 18, 615374
RAB33B	191,3	100.0%	100.0%	Smith-McCort dysplasia 2, 615222
RAB3GAP1	121,7	99.4%	98.9%	Warburg micro syndrome 1, 600118
RAB3GAP2	91,6	99.7%	96.9%	Martsolf syndrome, 212720

				Warburg micro syndrome 2, 614225
RAD50	102	97.5%	91.1%	Nijmegen breakage syndrome-like disorder, 613078
RAD51C	140,6	99.9%	99.5%	Fanconi anemia, complementation group O, 613390 {Breast-ovarian cancer, familial, susceptibility to, 3}, 613399
RAG1	150,9	100.0%	100.0%	Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889 Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457
RAG2	186,2	100.0%	100.0%	Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457
RAPSN	149	99.8%	97.7%	Fetal akinesia deformation sequence 2, 618388 Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326
RARB	93,2	100.0%	100.0%	Microphthalmia, syndromic 12, 615524
RARS	93,4	93.6%	90.0%	Leukodystrophy, hypomyelinating, 9, 616140
RARS2	104	100.0%	99.4%	Pontocerebellar hypoplasia, type 6, 611523
RAX	135,9	99.9%	98.5%	Microphthalmia, isolated 3, 611038
RBBP8	120,6	99.9%	99.3%	Jawad syndrome, 251255 Pancreatic carcinoma, somatic, 0 Seckel syndrome 2, 606744
RBCK1	107,9	100.0%	99.2%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RBM28	130,1	100.0%	99.9%	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RBM8A	87,4	99.8%	97.4%	Thrombocytopenia-absent radius syndrome, 274000
RBP3	150	100.0%	100.0%	?Retinitis pigmentosa 66, 615233
RBP4	137,9	99.2%	95.8%	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RCBTB1	95,9	99.8%	99.3%	Retinal dystrophy with or without extraocular anomalies, 617175
RD3	165,7	100.0%	100.0%	Leber congenital amaurosis 12, 610612
RDH11	93	99.9%	98.5%	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108
RDH12	80,7	99.8%	97.2%	Leber congenital amaurosis 13, 612712
RDH5	167,7	100.0%	100.0%	Fundus albipunctatus, 136880
RDX	38,4	88.0%	69.1%	Deafness, autosomal recessive 24, 611022
RECQL4	159,9	100.0%	99.8%	Baller-Gerold syndrome, 218600 RAPADILINO syndrome, 266280 Rothmund-Thomson syndrome, 268400
REEP2	173,9	100.0%	99.3%	?Spastic paraplegia 72, autosomal dominant, 615625

				?Spastic paraplegia 72, autosomal recessive, 615625
REEP6	206,2	100.0%	99.7%	Retinitis pigmentosa 77, 617304
RELB	108	99.2%	92.7%	?Immunodeficiency 53, 617585
RELN	130,1	100.0%	99.6%	Lissencephaly 2 (Norman-Roberts type), 257320 {Epilepsy, familial temporal lobe, 7}, 616436
REN	127,5	100.0%	100.0%	Hyperuricemic nephropathy, familial juvenile 2, 613092 Renal tubular dysgenesis, 267430 [Hyperproreninemia], 0
REPS1	121,5	99.0%	96.4%	?Neurodegeneration with brain iron accumulation 7, 617916
RETREG1	126,9	99.7%	98.8%	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
RFT1	105,7	100.0%	99.2%	Congenital disorder of glycosylation, type In, 612015
RFWD3	103,7	100.0%	99.5%	?Fanconi anemia, complementation group W, 617784
RFX5	109	99.8%	97.5%	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFX6	147	99.9%	99.7%	Mitchell-Riley syndrome, 615710
RFXANK	123,1	100.0%	99.3%	MHC class II deficiency, complementation group B, 209920
RFXAP	116,5	100.0%	99.3%	Bare lymphocyte syndrome, type II, complementation group D, 209920
RGR	120,8	100.0%	99.1%	Retinitis pigmentosa 44, 613769
RHO	165,3	100.0%	100.0%	Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 Retinitis punctata albescens, 136880
RIN2	119,5	100.0%	99.6%	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075
RIPK4	167,5	100.0%	100.0%	CHAND syndrome, 214350 Popliteal pterygium syndrome, Bartsocas-Papas type, 263650
RIPOR2	111,5	100.0%	99.8%	?Deafness, autosomal recessive 104, 616515
RIPPLY2	78,7	100.0%	98.7%	?Spondylocostal dysostosis 6, 616566
RLBP1	120,4	100.0%	99.7%	Bothnia retinal dystrophy, 607475 Fundus albipunctatus, 136880 Newfoundland rod-cone dystrophy, 607476 Retinitis punctata albescens, 136880
RMND1	132,6	100.0%	99.0%	Combined oxidative phosphorylation deficiency 11, 614922
RMRP	NC	NC	NC	Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RNASEH1	101,1	97.6%	92.5%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479
RNASEH2A	129,8	100.0%	99.7%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	100,8	98.9%	95.2%	Aicardi-Goutieres syndrome 2, 610181

RNASEH2C	281,7	100.0%	100.0%	Aicardi-Goutieres syndrome 3, 610329
RNASET2	102,2	95.4%	90.2%	Leukoencephalopathy, cystic, without megalencephaly, 612951
RNF168	182	100.0%	99.6%	RIDDLE syndrome, 611943
RNF216	125	99.9%	98.1%	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840
ROBO3	104,3	99.4%	96.9%	Gaze palsy, familial horizontal, with progressive scoliosis, 1, 607313
ROGDI	127,6	100.0%	99.4%	Kohlschutter-Tonz syndrome, 226750
ROR1	149,1	99.1%	97.2%	?Deafness, autosomal recessive 108, 617654
ROR2	160,6	100.0%	99.7%	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RORC	118,6	100.0%	100.0%	Immunodeficiency 42, 616622
RP1	109,4	91.4%	90.8%	Retinitis pigmentosa 1, 180100
RPE65	131,9	100.0%	99.8%	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794
RPGRIP1	128	100.0%	99.7%	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826
RPGRIP1L	123,4	96.7%	95.4%	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561
RPIA	113	100.0%	98.8%	?Ribose 5-phosphate isomerase deficiency, 608611
RRM2B	143,9	99.9%	99.4%	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077
RSPH1	122,6	100.0%	99.9%	Ciliary dyskinesia, primary, 24, 615481
RSPH3	139,5	99.9%	99.3%	Ciliary dyskinesia, primary, 32, 616481
RSPH4A	146,9	98.1%	95.3%	Ciliary dyskinesia, primary, 11, 612649
RSPH9	131,2	99.7%	97.1%	Ciliary dyskinesia, primary, 12, 612650
RSPO1	103,8	100.0%	99.9%	Palmoplantar hyperkeratosis and true hermaphroditism, 610644 Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644
RSPO2	136,4	96.6%	90.1%	?Humerofemoral hypoplasia with radiotibial ray deficiency, 618022 Tetraamelia syndrome 2, 618021
RSPO4	144,4	100.0%	100.0%	Anonychia congenita, 206800
RSPRY1	142	100.0%	99.9%	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723
RTEL1	131,1	99.7%	97.7%	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373
RTN4IP1	79,6	100.0%	98.0%	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
RTTN	117,9	98.8%	97.4%	Microcephaly, short stature, and polymicrogyria with seizures, 614833

RUBCN	99,3	99.9%	99.1%	?Spinocerebellar ataxia, autosomal recessive 15, 615705
RUSC2	188,1	100.0%	100.0%	Mental retardation, autosomal recessive 61, 617773
RYR1	117,1	98.7%	95.7%	Central core disease, 117000 King-Denborough syndrome, 145600 Minicore myopathy with external ophthalmoplegia, 255320 Neuromuscular disease, congenital, with uniform type 1 fiber, 117000 {Malignant hyperthermia susceptibility 1}, 145600
S1PR2	200,8	99.5%	96.8%	Deafness, autosomal recessive 68, 610419
SACS	150,4	100.0%	99.9%	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAG	127	100.0%	99.9%	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758
SALL2	128,4	100.0%	100.0%	?Coloboma, ocular, autosomal recessive, 216820
SAMD9	163,9	100.0%	99.9%	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455
SAMHD1	133,4	99.8%	98.5%	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SAR1B	120,9	97.2%	90.5%	Chylomicron retention disease, 246700
SARS	107,6	99.9%	98.8%	?Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709
SARS2	117,9	95.1%	93.2%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SASS6	77,6	99.6%	97.6%	?Microcephaly 14, primary, autosomal recessive, 616402
SBDS	166,2	100.0%	100.0%	Shwachman-Diamond syndrome, 260400 {Aplastic anemia, susceptibility to}, 609135
SBF1	122	99.4%	97.9%	Charcot-Marie-Tooth disease, type 4B3, 615284
SBF2	107,7	99.9%	99.0%	Charcot-Marie-Tooth disease, type 4B2, 604563
SC5D	153,6	99.8%	99.3%	Lathosterolosis, 607330
SCAPER	138,7	98.2%	96.4%	Intellectual developmental disorder and retinitis pigmentosa, 618195
SCARB2	105,8	99.8%	99.1%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCARF2	104,8	99.5%	95.7%	Van den Ende-Gupta syndrome, 600920
SCN1B	169,7	99.9%	98.1%	Atrial fibrillation, familial, 13, 615377 Brugada syndrome 5, 612838 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233 Epileptic encephalopathy, early infantile, 52, 617350
SCN4A	167,9	99.8%	99.3%	Hyperkalemic periodic paralysis, type 2, 170500 Hypokalemic periodic paralysis, type 2, 613345 Myasthenic syndrome, congenital, 16, 614198 Myotonia congenita, atypical, acetazolamide-responsive, 608390

				Paramyotonia congenita, 168300
SCN9A	128,4	99.1%	97.7%	Epilepsy, generalized, with febrile seizures plus, type 7, 613863 Erythermalgia, primary, 133020 Febrile seizures, familial, 3B, 613863 HSAN2D, autosomal recessive, 243000 Insensitivity to pain, congenital, 243000 Paroxysmal extreme pain disorder, 167400 Small fiber neuropathy, 133020 {Dravet syndrome, modifier of}, 607208
SCNN1A	127	99.8%	98.3%	?Liddle syndrome 3, 618126 Bronchiectasis with or without elevated sweat chloride 2, 613021 Pseudohypoaldosteronism, type I, 264350
SCNN1B	130,6	100.0%	100.0%	Bronchiectasis with or without elevated sweat chloride 1, 211400 Liddle syndrome 1, 177200 Pseudohypoaldosteronism, type I, 264350
SCNN1G	142,2	99.5%	97.2%	Bronchiectasis with or without elevated sweat chloride 3, 613071 Liddle syndrome 2, 618114 Pseudohypoaldosteronism, type I, 264350
SCO1	100,1	99.8%	98.1%	Mitochondrial complex IV deficiency, 220110
SCO2	115,7	100.0%	99.9%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908
SCP2	107,8	99.7%	96.4%	?Leukoencephalopathy with dystonia and motor neuropathy, 613724
SCYL1	146,2	100.0%	100.0%	Spinocerebellar ataxia, autosomal recessive 21, 616719
SDCCAG8	124,1	100.0%	99.7%	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SDHA	88,9	85.1%	77.7%	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165
SDHAF1	83	100.0%	100.0%	Mitochondrial complex II deficiency, 252011
SDHD	43,7	52.7%	50.6%	Mitochondrial complex II deficiency, 252011 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 1, with or without deafness, 168000 Pheochromocytoma, 171300
SDR9C7	168	100.0%	99.9%	Ichthyosis, congenital, autosomal recessive 13, 617574
SEC23A	122,6	99.7%	97.9%	Craniolenticulosutural dysplasia, 607812
SEC23B	131	99.8%	99.0%	?Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100

SEC24D	126,3	99.9%	99.5%	Cole-Carpenter syndrome 2, 616294
SECISBP2	108	99.7%	97.1%	Thyroid hormone metabolism, abnormal, 609698
SELENON	131	84.9%	83.9%	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310
SEMA4A	124,3	100.0%	99.3%	Cone-rod dystrophy 10, 610283 Retinitis pigmentosa 35, 610282
SEPSECS	159,6	100.0%	99.6%	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	111	99.7%	99.0%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPINA1	104,9	100.0%	99.7%	Emphysema due to AAT deficiency, 613490 Emphysema-cirrhosis, due to AAT deficiency, 613490 Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963
SERPINA6	138,3	100.0%	100.0%	Corticosteroid-binding globulin deficiency, 611489
SERPINB6	138,9	95.9%	95.9%	?Deafness, autosomal recessive 91, 613453
SERPINB7	124,3	100.0%	99.6%	Palmoplantar keratoderma, Nagashima type, 615598
SERPINB8	125,8	95.0%	95.0%	Peeling skin syndrome 5, 617115
SERPINC1	117,9	100.0%	100.0%	Thrombophilia due to antithrombin III deficiency, 613118
SERPINE1	134	100.0%	100.0%	Plasminogen activator inhibitor-1 deficiency, 613329 {Transcription of plasminogen activator inhibitor, modulator of}, 0
SERPINF1	104	100.0%	99.9%	Osteogenesis imperfecta, type VI, 613982
SERPINF2	151,6	100.0%	99.9%	Alpha-2-plasmin inhibitor deficiency, 262850
SERPING1	96,7	99.5%	96.7%	Angioedema, hereditary, types I and II, 106100 Complement component 4, partial deficiency of, 120790
SERPINH1	195,8	100.0%	99.6%	Osteogenesis imperfecta, type X, 613848 {Preterm premature rupture of the membranes, susceptibility to}, 610504
SETX	151,6	100.0%	99.6%	Amyotrophic lateral sclerosis 4, juvenile, 602433 Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002
SFRP4	125,4	99.9%	99.0%	Pyle disease, 265900
SFTPB	94,1	100.0%	99.5%	Surfactant metabolism dysfunction, pulmonary, 1, 265120
SFXN4	124	100.0%	99.7%	Combined oxidative phosphorylation deficiency 18, 615578
SGCA	158,4	100.0%	100.0%	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099
SGCB	140,1	99.3%	96.7%	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286
SGCD	78	99.8%	97.2%	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287
SGCG	114,5	100.0%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700
SGO1	107,8	99.9%	98.9%	Chronic atrial and intestinal dysrhythmia, 616201
SGPL1	132,3	100.0%	100.0%	Nephrotic syndrome, type 14, 617575

SGSH	140,2	97.6%	94.7%	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SH3PXD2B	161,4	100.0%	99.9%	Frank-ter Haar syndrome, 249420
SH3TC2	102,1	100.0%	99.4%	Charcot-Marie-Tooth disease, type 4C, 601596 Mononeuropathy of the median nerve, mild, 613353
SI	118,7	99.4%	95.9%	Sucrase-isomaltase deficiency, congenital, 222900
SIGMAR1	146,6	100.0%	100.0%	?Amyotrophic lateral sclerosis 16, juvenile, 614373 ?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726
SIL1	129,5	98.9%	96.2%	Marinesco-Sjogren syndrome, 248800
SIX6	278,5	100.0%	100.0%	Optic disc anomalies with retinal and/or macular dystrophy, 212550
SKIV2L	138,4	100.0%	99.7%	Trichohepatoenteric syndrome 2, 614602
SLC10A2	122,1	100.0%	100.0%	Bile acid malabsorption, primary, 613291
SLC11A2	96,2	99.9%	98.7%	Anemia, hypochromic microcytic, with iron overload 1, 206100
SLC12A1	144,2	100.0%	99.8%	Bartter syndrome, type 1, 601678
SLC12A3	140	100.0%	100.0%	Gitelman syndrome, 263800
SLC12A5	111,9	86.1%	84.1%	Epileptic encephalopathy, early infantile, 34, 616645 {Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685
SLC12A6	118,9	100.0%	99.9%	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC13A5	141,9	100.0%	99.9%	Epileptic encephalopathy, early infantile, 25, 615905
SLC17A5	137,7	99.8%	96.1%	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC18A2	106,8	100.0%	99.8%	?Parkinsonism-dystonia, infantile, 2, 618049
SLC18A3	256,7	100.0%	100.0%	Myasthenic syndrome, congenital, 21, presynaptic, 617239
SLC19A2	101,3	100.0%	99.6%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	134,6	100.0%	99.9%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A1	145,6	100.0%	99.6%	Dicarboxylic aminoaciduria, 222730 {?Schizophrenia susceptibility 18}, 615232
SLC1A4	146,4	100.0%	99.5%	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC22A12	117,7	100.0%	99.8%	Hypouricemia, renal, 220150
SLC22A5	129,7	100.0%	100.0%	Carnitine deficiency, systemic primary, 212140
SLC24A1	167	100.0%	100.0%	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
SLC24A4	103,5	100.0%	99.8%	Amelogenesis imperfecta, type IIA5, 615887 [Skin/hair/eye pigmentation 6, blond/brown hair], 210750 [Skin/hair/eye pigmentation 6, blue/green eyes], 210750
SLC24A5	104,1	99.9%	99.3%	Albinism, oculocutaneous, type VI, 113750 [Skin/hair/eye pigmentation 4, fair/dark skin], 113750
SLC25A1	103,2	99.3%	95.1%	?Myasthenic syndrome, congenital, 23, presynaptic, 618197 Combined D-2- and L-2-hydroxyglutaric aciduria, 615182

SLC25A12	150,9	99.9%	99.9%	Epileptic encephalopathy, early infantile, 39, 612949
SLC25A13	120,1	99.8%	98.9%	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A15	146,8	97.9%	93.6%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A19	77,4	99.9%	97.8%	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A20	90,7	100.0%	100.0%	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A22	123,8	100.0%	99.1%	Epileptic encephalopathy, early infantile, 3, 609304
SLC25A26	98,4	99.9%	99.3%	Combined oxidative phosphorylation deficiency 28, 616794
SLC25A3	129,8	99.5%	96.9%	Mitochondrial phosphate carrier deficiency, 610773
SLC25A38	94,5	99.1%	95.2%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC25A4	130,9	100.0%	99.9%	Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184 Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283
SLC25A46	173	99.8%	98.3%	Neuropathy, hereditary motor and sensory, type VIB, 616505
SLC26A1	149,3	100.0%	100.0%	?Nephrolithiasis, calcium oxalate, 167030
SLC26A2	205,1	100.0%	99.9%	Achondrogenesis Ib, 600972 Atelosteogenesis, type II, 256050 De la Chapelle dysplasia, 256050 Diastrophic dysplasia, 222600 Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 Epiphyseal dysplasia, multiple, 4, 226900
SLC26A3	132,5	99.9%	99.2%	Diarrhea 1, secretory chloride, congenital, 214700
SLC26A4	113	100.0%	99.5%	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791 Pendred syndrome, 274600
SLC26A5	130,3	98.3%	95.4%	?Deafness, autosomal recessive 61, 613865
SLC27A4	150,9	100.0%	100.0%	Ichthyosis prematurity syndrome, 608649
SLC29A3	173,3	100.0%	99.5%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC2A1	148,9	92.8%	92.8%	Dystonia 9, 601042 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 GLUT1 deficiency syndrome 2, childhood onset, 612126 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847
SLC2A10	152,6	98.0%	97.5%	Arterial tortuosity syndrome, 208050
SLC2A2	158,1	100.0%	99.9%	Fanconi-Bickel syndrome, 227810 {Diabetes mellitus, noninsulin-dependent}, 125853
SLC2A9	104,8	100.0%	98.7%	Hypouricemia, renal, 2, 612076

				{Uric acid concentration, serum, QTL 2}, 612076
SLC30A10	176,1	100.0%	100.0%	Hypermanganesemia with dystonia 1, 613280
SLC30A9	88,6	98.7%	93.1%	?Birk-Landau-Perez syndrome, 617595
SLC33A1	132	99.7%	97.7%	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539
SLC34A1	149,8	100.0%	99.9%	?Fanconi renotubular syndrome 2, 613388 Hypercalcemia, infantile, 2, 616963 Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286
SLC34A2	140,3	100.0%	99.7%	Pulmonary alveolar microlithiasis, 265100
SLC34A3	141,1	99.9%	99.0%	Hypophosphatemic rickets with hypercalciuria, 241530
SLC35A1	127,1	100.0%	99.4%	Congenital disorder of glycosylation, type IIf, 603585
SLC35A3	66,6	80.6%	78.3%	?Arthrogryposis, mental retardation, and seizures, 615553
SLC35C1	187,8	100.0%	99.8%	Congenital disorder of glycosylation, type IIc, 266265
SLC35D1	125	99.5%	97.2%	Schneckenbecken dysplasia, 269250
SLC37A4	114,3	100.0%	99.6%	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240
SLC38A8	71,5	99.4%	95.5%	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218
SLC39A13	145,1	100.0%	99.9%	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350
SLC39A14	95,4	99.9%	97.9%	?Hyperostosis cranalis interna, 144755 Hypermanganesemia with dystonia 2, 617013
SLC39A4	114,2	100.0%	99.0%	Acrodermatitis enteropathica, 201100
SLC39A8	140,9	100.0%	99.8%	Congenital disorder of glycosylation, type IIn, 616721
SLC3A1	144,5	100.0%	99.4%	Cystinuria, 220100
SLC45A1	141	100.0%	100.0%	Intellectual developmental disorder with neuropsychiatric features, 617532
SLC45A2	115,2	100.0%	99.8%	Albinism, oculocutaneous, type IV, 606574 [Skin/hair/eye pigmentation 5, black/nonblack hair], 227240 [Skin/hair/eye pigmentation 5, dark/fair skin], 227240 [Skin/hair/eye pigmentation 5, dark/light eyes], 227240
SLC46A1	111,1	99.9%	98.4%	Folate malabsorption, hereditary, 229050
SLC4A1	139,2	100.0%	99.8%	Cryohydrocytosis, 185020 Ovalocytosis, SA type, 166900 Renal tubular acidosis, distal, AD, 179800 Renal tubular acidosis, distal, AR, 611590 Spherocytosis, type 4, 612653 [Blood group, Diego], 110500 [Blood group, Froese], 601551 [Blood group, Swann], 601550

				[Blood group, Waldner], 112010 [Blood group, Wright], 112050 [Malaria, resistance to], 611162
SLC4A11	157,4	100.0%	100.0%	Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy and perceptive deafness, 217400 Corneal endothelial dystrophy, autosomal recessive, 217700
SLC4A4	113,9	99.8%	98.3%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC52A2	185,4	100.0%	100.0%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	118,8	100.0%	99.8%	?Fazio-Londe disease, 211500 Brown-Vialetto-Van Laere syndrome 1, 211530
SLC5A1	110,7	100.0%	99.3%	Glucose/galactose malabsorption, 606824
SLC5A2	135	100.0%	100.0%	Renal glucosuria, 233100
SLC5A5	105,7	100.0%	99.9%	Thyroid dyshormonogenesis 1, 274400
SLC5A7	100,2	100.0%	99.9%	Myasthenic syndrome, congenital, 20, presynaptic, 617143 Neuronopathy, distal hereditary motor, type VIIA, 158580
SLC6A17	149,8	100.0%	100.0%	Mental retardation, autosomal recessive 48, 616269
SLC6A19	129,3	100.0%	100.0%	Hartnup disorder, 234500 Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC6A3	133	100.0%	99.9%	Parkinsonism-dystonia, infantile, 1, 613135 {Nicotine dependence, protection against}, 188890
SLC6A5	128,7	100.0%	100.0%	Hyperekplexia 3, 614618
SLC6A9	148,8	100.0%	100.0%	Glycine encephalopathy with normal serum glycine, 617301
SLC7A14	145,3	100.0%	100.0%	Retinitis pigmentosa 68, 615725
SLC7A7	105,5	100.0%	99.6%	Lysinuric protein intolerance, 222700
SLC7A9	119,8	100.0%	98.8%	Cystinuria, 220100
SLC9A1	142,4	100.0%	100.0%	?Lichtenstein-Knorr syndrome, 616291
SLC9A3	161,7	100.0%	99.8%	Diarrhea 8, secretory sodium, congenital, 616868
SLCO2A1	97,7	99.9%	98.2%	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
SLITRK6	169,5	100.0%	100.0%	Deafness and myopia, 221200
SLURP1	100,1	100.0%	99.4%	Meleda disease, 248300
SLX4	124,2	100.0%	99.7%	Fanconi anemia, complementation group P, 613951
SMARCAL1	113,2	100.0%	99.6%	Schimke immunoosseous dysplasia, 242900
SMARCD2	92,3	87.3%	85.8%	Specific granule deficiency 2, 617475
SMG9	94,5	100.0%	100.0%	Heart and brain malformation syndrome, 616920
SMN1	97,6	99.8%	96.9%	Spinal muscular atrophy-1, 253300 Spinal muscular atrophy-2, 253550

				Spinal muscular atrophy-3, 253400 Spinal muscular atrophy-4, 271150
SMOC1	115,1	99.8%	98.2%	Microphthalmia with limb anomalies, 206920
SMOC2	88,7	77.0%	75.7%	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SMPD1	146,4	100.0%	99.2%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SNAI2	102,7	99.9%	99.1%	Piebaldism, 172800 Waardenburg syndrome, type 2D, 608890
SNAP29	168,4	100.0%	100.0%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNIP1	131	100.0%	99.2%	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501
SNORD118	NC	NC	NC	Leukoencephalopathy, brain calcifications, and cysts, 614561
SNX10	131,4	96.2%	95.7%	Osteopetrosis, autosomal recessive 8, 615085
SNX14	84,1	99.0%	95.4%	Spinocerebellar ataxia, autosomal recessive 20, 616354
SOBP	182,7	98.8%	97.8%	Mental retardation, anterior maxillary protrusion, and strabismus, 613671
SOD1	123,6	100.0%	100.0%	Amyotrophic lateral sclerosis 1, 105400
SOST	182,9	100.0%	99.6%	Craniodiaphyseal dysplasia, autosomal dominant, 122860 Sclerosteosis 1, 269500 Van Buchem disease, 239100
SOX18	50	91.5%	76.2%	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823 Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940
SP110	109,3	100.0%	99.8%	Hepatic venoocclusive disease with immunodeficiency, 235550 {Mycobacterium tuberculosis, susceptibility to}, 607948
SP7	148,4	100.0%	99.3%	Osteogenesis imperfecta, type XII, 613849
SPAG1	101,6	99.1%	95.3%	Ciliary dyskinesia, primary, 28, 615505
SPARC	134,3	100.0%	100.0%	Osteogenesis imperfecta, type XVII, 616507
SPART	132,6	99.8%	98.2%	Troyer syndrome, 275900
SPATA5	139,5	100.0%	99.8%	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPATA7	122,7	99.4%	97.4%	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, autosomal recessive, 604232
SPEG	128,6	99.2%	97.1%	Centronuclear myopathy 5, 615959
SPG11	116,1	99.7%	98.4%	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360
SPG21	120,6	99.7%	96.8%	Mast syndrome, 248900
SPG7	115,2	99.3%	96.4%	Spastic paraplegia 7, autosomal recessive, 607259
SPINK5	128	99.9%	99.5%	Netherton syndrome, 256500
SPINT2	68,8	99.7%	90.0%	Diarrhea 3, secretory sodium, congenital, syndromic, 270420

SPR	145,7	100.0%	99.8%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRTN	160,6	100.0%	100.0%	Ruijs-Aalfs syndrome, 616200
SPTA1	105,2	99.9%	98.6%	Elliptocytosis-2, 130600 Pyropoikilocytosis, 266140 Spherocytosis, type 3, 270970
SPTB	142,9	100.0%	100.0%	Anemia, neonatal hemolytic, fatal or near-fatal, 617948 Elliptocytosis-3, 617948 Spherocytosis, type 2, 616649
SPTBN2	126,2	100.0%	99.7%	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia, autosomal recessive 14, 615386
SPTBN4	103,8	99.8%	98.1%	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519
SQSTM1	117,8	99.9%	99.2%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 Myopathy, distal, with rimmed vacuoles, 617158 Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 Paget disease of bone 3, 167250
SRD5A2	85,6	100.0%	98.1%	Pseudovaginal perineoscrotal hypospadias, 264600
SRD5A3	139,9	99.8%	98.3%	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713
ST14	154,8	100.0%	99.9%	Ichthyosis, congenital, autosomal recessive 11, 602400
ST3GAL3	134,7	100.0%	99.5%	?Epileptic encephalopathy, early infantile, 15, 615006 Mental retardation, autosomal recessive 12, 611090
ST3GAL5	101,8	89.0%	84.9%	Salt and pepper developmental regression syndrome, 609056
STAC3	114,7	100.0%	100.0%	Myopathy, congenital, Baily-Bloch, 255995
STAMBP	93,7	99.8%	97.9%	Microcephaly-capillary malformation syndrome, 614261
STAR	135	100.0%	100.0%	Lipoid adrenal hyperplasia, 201710
STAT1	117,8	99.6%	97.7%	Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796 Immunodeficiency 31C, autosomal dominant, 614162
STAT2	110	100.0%	99.8%	Immunodeficiency 44, 616636
STAT5B	114,1	99.8%	97.8%	Growth hormone insensitivity with immunodeficiency, 245590 Leukemia, acute promyelocytic, somatic, 102578
STIL	154,1	100.0%	99.8%	Microcephaly 7, primary, autosomal recessive, 612703
STIM1	120,7	99.8%	96.8%	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070
STK4	122,5	100.0%	99.7%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STN1	82,2	100.0%	99.6%	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341

STRA6	117,6	100.0%	99.8%	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186
STRADA	108,6	100.0%	98.8%	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087
STRC	99	99.9%	98.1%	Deafness, autosomal recessive 16, 603720
STT3A	123,1	100.0%	99.9%	?Congenital disorder of glycosylation, type Iw, 615596
STT3B	127,4	99.9%	99.6%	?Congenital disorder of glycosylation, type Ix, 615597
STUB1	173,9	100.0%	99.5%	?Spinocerebellar ataxia 48, 618093 Spinocerebellar ataxia, autosomal recessive 16, 615768
STX11	298,3	100.0%	100.0%	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STXBP2	100,2	83.7%	80.4%	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
SUCLA2	58,8	91.7%	82.6%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	102,9	99.9%	99.6%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUFU	132,8	100.0%	99.9%	Basal cell nevus syndrome, 109400 Joubert syndrome 32, 617757 Medulloblastoma, desmoplastic, 155255 {Meningioma, familial, susceptibility to}, 607174
SULT2B1	124,7	100.0%	100.0%	Ichthyosis, congenital, autosomal recessive 14, 617571
SUMF1	89,7	99.7%	96.8%	Multiple sulfatase deficiency, 272200
SUOX	167,2	100.0%	100.0%	Sulfite oxidase deficiency, 272300
SURF1	84,8	91.3%	88.4%	Charcot-Marie-Tooth disease, type 4K, 616684 Leigh syndrome, due to COX IV deficiency, 256000
SYNE1	121,6	98.3%	97.8%	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 Spinocerebellar ataxia, autosomal recessive 8, 610743
SYNE4	83,7	99.9%	98.5%	Deafness, autosomal recessive 76, 615540
SYNJ1	126,6	99.9%	98.5%	Epileptic encephalopathy, early infantile, 53, 617389 Parkinson disease 20, early-onset, 615530
SYT14	104,1	60.3%	58.0%	?Spinocerebellar ataxia, autosomal recessive 11, 614229
SZT2	135,6	99.6%	99.4%	Epileptic encephalopathy, early infantile, 18, 615476
T	NC	NC	NC	Sacral agenesis with vertebral anomalies, 615709 {Neural tube defects, susceptibility to}, 182940
TAC3	61,8	99.9%	95.4%	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACO1	93,9	99.5%	95.3%	Mitochondrial complex IV deficiency, 220110
TACR3	146,1	100.0%	99.7%	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
TACSTD2	268,7	100.0%	99.9%	Corneal dystrophy, gelatinous drop-like, 204870
TAF13	100,1	100.0%	99.9%	Mental retardation, autosomal recessive 60, 617432
TAF2	113,9	99.8%	98.5%	Mental retardation, autosomal recessive 40, 615599
TAF6	127,3	100.0%	99.3%	Alazami-Yuan syndrome, 617126

TALDO1	148,2	100.0%	99.6%	Transaldolase deficiency, 606003
TANGO2	127,3	100.0%	100.0%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TAP1	117,7	99.9%	97.3%	Bare lymphocyte syndrome, type I, 604571
TAP2	93	99.6%	98.4%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	116,6	96.6%	96.5%	Bare lymphocyte syndrome, type I, 604571
TAPT1	89,2	97.9%	92.2%	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897
TARS2	89	99.7%	96.4%	?Combined oxidative phosphorylation deficiency 21, 615918
TAT	115	100.0%	100.0%	Tyrosinemia, type II, 276600
TBC1D20	115,7	96.3%	93.8%	Warburg micro syndrome 4, 615663
TBC1D23	92,7	99.2%	95.4%	Pontocerebellar hypoplasia, type 11, 617695
TBC1D24	177,7	100.0%	100.0%	Deafness , autosomal recessive 86, 614617 Deafness, autosomal dominant 65, 616044 DOORS syndrome, 220500 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021
TBC1D7	99,6	99.8%	99.3%	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000
TBCD	136,2	98.2%	94.3%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TBCE	116,4	98.7%	94.7%	Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Kenny-Caffey syndrome, type 1, 244460
TBCK	101,6	99.5%	96.1%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900
TBX15	106,3	100.0%	99.7%	Cousin syndrome, 260660
TBX19	156,9	100.0%	99.9%	Adrenocorticotropic hormone deficiency, 201400
TBX6	124,4	99.7%	96.5%	Spondylocostal dysostosis 5, 122600
TBXAS1	128,8	100.0%	100.0%	?Thromboxane synthase deficiency, 614158 Ghosal hematodiaphyseal syndrome, 231095
TCAP	100,1	100.0%	100.0%	Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954
TCIRG1	131,4	99.2%	96.6%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	148,5	100.0%	100.0%	Transcobalamin II deficiency, 275350
TCTEX1D2	123,6	100.0%	99.4%	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405
TCTN1	94,8	95.6%	92.3%	Joubert syndrome 13, 614173
TCTN2	122,4	99.9%	99.0%	?Meckel syndrome 8, 613885 Joubert syndrome 24, 616654
TCTN3	116,3	100.0%	99.9%	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860

TDP1	103,9	99.9%	99.5%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250
TDP2	173	99.9%	99.4%	Spinocerebellar ataxia, autosomal recessive 23, 616949
TDRD7	136,7	99.8%	98.7%	Cataract 36, 613887
TECPR2	137,2	100.0%	100.0%	Spastic paraplegia 49, autosomal recessive, 615031
TECR	124,9	100.0%	99.6%	Mental retardation, autosomal recessive 14, 614020
TECRL	75	97.1%	90.3%	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021
TECTA	164,9	100.0%	99.9%	Deafness, autosomal dominant 8/12, 601543 Deafness, autosomal recessive 21, 603629
TELO2	122,6	99.8%	97.5%	You-Hoover-Fong syndrome, 616954
TENM3	148,6	99.8%	99.3%	Microphthalmia, isolated, with coloboma 9, 615145
TF	101,6	100.0%	99.7%	Atransferrinemia, 209300
TFAM	67,1	94.2%	74.5%	?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156
TFG	106,2	96.8%	95.3%	?Spastic paraplegia 57, autosomal recessive, 615658 Hereditary motor and sensory neuropathy, Okinawa type, 604484
TFR2	124	99.6%	98.3%	Hemochromatosis, type 3, 604250
TFRC	132	99.9%	99.0%	Immunodeficiency 46, 616740
TG	117	99.9%	99.1%	Thyroid dyshormonogenesis 3, 274700 {Autoimmune thyroid disease, susceptibility to, 3}, 608175
TGDS	88,3	99.4%	96.6%	Catel-Manzke syndrome, 616145
TGM1	141	100.0%	99.9%	Ichthyosis, congenital, autosomal recessive 1, 242300
TGM5	144,8	100.0%	99.9%	Peeling skin syndrome 2, 609796
TH	96,3	100.0%	98.2%	Segawa syndrome, recessive, 605407
THOC6	228,9	100.0%	100.0%	Beaulieu-Boycott-Innes syndrome, 613680
THRB	141,2	99.9%	99.3%	Thyroid hormone resistance, 188570 Thyroid hormone resistance, autosomal recessive, 274300 Thyroid hormone resistance, selective pituitary, 145650
TIMM50	122,9	99.9%	98.7%	3-methylglutaconic aciduria, type IX, 617698
TIMMDC1	161,4	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 31, 618251
TJP2	109,3	94.0%	93.4%	Cholestasis, progressive familial intrahepatic 4, 615878 Hypercholanemia, familial, 607748
TK2	103,8	100.0%	99.2%	?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069 Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560
TKT	115,6	98.7%	98.1%	Short stature, developmental delay, and congenital heart defects, 617044
TLE6	116,7	99.9%	98.6%	Preimplantation embryonic lethality, 616814
TMC1	110,1	99.8%	97.5%	Deafness, autosomal dominant 36, 606705 Deafness, autosomal recessive 7, 600974
TMC6	91,1	100.0%	99.7%	Epidermodysplasia verruciformis, 226400

TMC8	133	100.0%	99.7%	Epidermodysplasia verruciformis 2, 618231
TMCO1	81,7	88.0%	87.5%	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980
TMEM107	148,7	100.0%	100.0%	?Joubert syndrome 29, 617562 Meckel syndrome 13, 617562 Orofaciodigital syndrome XVI, 617563
TMEM126A	104,8	96.2%	82.8%	Optic atrophy 7, 612989
TMEM126B	87,8	99.5%	95.9%	Mitochondrial complex I deficiency, nuclear type 29, 618250
TMEM138	82,7	100.0%	99.2%	Joubert syndrome 16, 614465
TMEM165	148,2	100.0%	99.8%	Congenital disorder of glycosylation, type IIk, 614727
TMEM199	118,4	100.0%	99.8%	Congenital disorder of glycosylation, type IIp, 616829
TMEM216	88	99.7%	95.7%	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	101,1	100.0%	99.3%	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	117,7	100.0%	99.2%	Joubert syndrome 14, 614424
TMEM260	117,5	99.6%	97.6%	Structural heart defects and renal anomalies syndrome, 617478
TMEM38B	107,9	100.0%	99.0%	Osteogenesis imperfecta, type XIV, 615066
TMEM5	NC	NC	NC	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
TMEM67	83,1	99.1%	94.6%	?RHYNS syndrome, 602152 COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991
TMEM70	117,9	99.8%	97.6%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMIE	104,5	100.0%	99.7%	Deafness, autosomal recessive 6, 600971
TMPRSS15	106,6	98.5%	95.1%	Enterokinase deficiency, 226200
TMPRSS3	98,5	100.0%	99.4%	Deafness, autosomal recessive 8/10, 601072
TMPRSS6	107	100.0%	99.4%	Iron-refractory iron deficiency anemia, 206200
TMTC3	92,8	99.5%	97.6%	Lissencephaly 8, 617255
TNFRSF11B	172,4	100.0%	100.0%	Paget disease of bone 5, juvenile-onset, 239000
TNFRSF13B	100,8	100.0%	99.7%	Immunodeficiency, common variable, 2, 240500 Immunoglobulin A deficiency 2, 609529
TNFRSF13C	98,4	96.2%	82.4%	Immunodeficiency, common variable, 4, 613494
TNFRSF4	79,8	99.5%	96.8%	?Immunodeficiency 16, 615593
TNFSF11	129,8	100.0%	100.0%	Osteopetrosis, autosomal recessive 2, 259710
TNIK	106,3	99.9%	98.7%	Mental retardation, autosomal recessive 54, 617028

TNNI3	107,3	99.6%	94.9%	?Cardiomyopathy, dilated, 2A, 611880 Cardiomyopathy, dilated, 1FF, 613286 Cardiomyopathy, familial restrictive, 1, 115210 Cardiomyopathy, hypertrophic, 7, 613690
TNNT1	104,4	99.9%	98.5%	Nemaline myopathy 5, Amish type, 605355
TNXB	105,6	99.5%	95.8%	Ehlers-Danlos syndrome, classic-like, 1, 606408 Vesicoureteral reflux 8, 615963
TOE1	141,1	100.0%	99.8%	Pontocerebellar hypoplasia, type 7, 614969
TOR1AIP1	133,3	99.5%	97.2%	?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072
TP53RK	81,3	99.7%	96.1%	Galloway-Mowat syndrome 4, 617730
TPI1	112,1	99.9%	96.4%	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512
TPK1	94	100.0%	98.7%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TPO	137,6	100.0%	99.6%	Thyroid dyshormonogenesis 2A, 274500
TPP1	123,7	100.0%	99.9%	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TPRKB	59,4	80.3%	73.9%	Galloway-Mowat syndrome 5, 617731
TPRN	98,7	91.5%	86.7%	Deafness, autosomal recessive 79, 613307
TRAC	127,5	100.0%	100.0%	Immunodeficiency 7, TCR-alpha/beta deficient, 615387
TRAF3IP1	84,2	99.4%	97.1%	Senior-Loken syndrome 9, 616629
TRAIP	123,4	100.0%	100.0%	Seckel syndrome 9, 616777
TRAK1	149,3	100.0%	99.6%	Epileptic encephalopathy, early infantile, 68, 618201
TRAPPC11	125,6	99.9%	99.0%	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356
TRAPPC12	162,3	100.0%	100.0%	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669
TRAPPC6B	79	99.9%	98.2%	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862
TRAPPC9	125,2	100.0%	99.7%	Mental retardation, autosomal recessive 13, 613192
TRDN	82,7	97.5%	88.4%	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441
TREM2	127	100.0%	99.9%	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193
TREX1	233,4	100.0%	100.0%	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700
TRH	112,3	99.8%	97.3%	Thyrotropin-releasing hormone deficiency, 275120
TRIM2	136,6	93.9%	93.6%	Charcot-Marie-Tooth disease, type 2R, 615490
TRIM32	123	100.0%	100.0%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRIM36	136	99.9%	99.0%	?Anencephaly, 206500
TRIM37	113,8	98.5%	97.4%	Mulibrey nanism, 253250

TRIOBP	156,9	99.1%	97.5%	Deafness, autosomal recessive 28, 609823
TRIP11	90,9	97.5%	92.6%	Achondrogenesis, type IA, 200600 Osteochondrodysplasia, 184260
TRIP4	103,3	99.8%	98.5%	?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066 Spinal muscular atrophy with congenital bone fractures 1, 616866
TRIT1	107,9	100.0%	99.9%	Combined oxidative phosphorylation deficiency 35, 617873
TRMT10A	119,5	99.9%	99.2%	Microcephaly, short stature, and impaired glucose metabolism 1, 616033
TRMT10C	138,5	100.0%	100.0%	Combined oxidative phosphorylation deficiency 30, 616974
TRMT5	175,7	99.8%	98.7%	Combined oxidative phosphorylation deficiency 26, 616539
TRMU	100	100.0%	99.4%	Liver failure, transient infantile, 613070 {Deafness, mitochondrial, modifier of}, 580000
TRNT1	101,5	99.2%	96.5%	Retinitis pigmentosa and erythrocytic microcytosis, 616959 Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084
TRPM1	128,8	100.0%	99.0%	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TRPM6	126,6	99.9%	99.1%	Hypomagnesemia 1, intestinal, 602014
TSEN15	89,8	99.7%	96.4%	Pontocerebellar hypoplasia, type 2F, 617026
TSEN2	95,6	99.9%	98.9%	Pontocerebellar hypoplasia type 2B, 612389
TSEN34	77,6	98.7%	93.6%	?Pontocerebellar hypoplasia type 2C, 612390
TSEN54	114,4	99.4%	96.8%	?Pontocerebellar hypoplasia type 5, 610204 Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753
TSFM	120	100.0%	99.2%	Combined oxidative phosphorylation deficiency 3, 610505
TSHB	229,7	100.0%	100.0%	Hypothyroidism, congenital, nongoitrous 4, 275100
TSHR	153	100.0%	99.0%	Hyperthyroidism, familial gestational, 603373 Hyperthyroidism, nonautoimmune, 609152 Hypothyroidism, congenital, nongoitrous, 1, 275200 Thyroid adenoma, hyperfunctioning, somatic, 0 Thyroid carcinoma with thyrotoxicosis, 0
TSPAN12	135,3	100.0%	99.4%	Exudative vitreoretinopathy 5, 613310
TSPEAR	139,3	100.0%	99.8%	?Deafness, autosomal recessive 98, 614861 Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180
TSPYL1	144,9	100.0%	100.0%	Sudden infant death with dysgenesis of the testes syndrome, 608800
TTC19	83,4	97.0%	82.6%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTC21B	119,5	99.7%	98.8%	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
TTC25	93,7	100.0%	99.6%	Ciliary dyskinesia, primary, 35, 617092
TTC37	135,1	99.9%	99.2%	Trichohepatoenteric syndrome 1, 222470

TTC7A	115	99.8%	98.0%	Gastrointestinal defects and immunodeficiency syndrome, 243150
TTC8	115,2	99.8%	98.8%	?Retinitis pigmentosa 51, 613464 Bardet-Biedl syndrome 8, 615985
TTI2	96,2	100.0%	99.9%	Mental retardation, autosomal recessive 39, 615541
TTLL5	136,1	100.0%	98.7%	Cone-rod dystrophy 19, 615860
TTPA	109,2	97.6%	92.5%	Ataxia with isolated vitamin E deficiency, 277460
TUBA8	126,1	100.0%	99.5%	Cortical dysplasia, complex, with other brain malformations 8, 613180
TUBGCP4	104,6	98.0%	94.7%	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TUBGCP6	155,1	100.0%	99.5%	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
TUFM	130,6	100.0%	99.2%	Combined oxidative phosphorylation deficiency 4, 610678
TULP1	117,9	100.0%	99.6%	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132
TUSC3	155,1	99.9%	99.5%	Mental retardation, autosomal recessive 7, 611093
TWIST2	132,6	100.0%	100.0%	Ablepharon-macrostomia syndrome, 200110 Barber-Say syndrome, 209885 Focal facial dermal dysplasia 3, Setleis type, 227260
TWNK	159,6	100.0%	100.0%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Perrault syndrome 5, 616138 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286
TXN2	61,4	100.0%	99.8%	?Combined oxidative phosphorylation deficiency 29, 616811
TXNL4A	122,6	99.5%	98.3%	Burn-McKeown syndrome, 608572
TYK2	129,9	100.0%	99.5%	Immunodeficiency 35, 611521
TYMP	120,9	100.0%	100.0%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYR	147,9	100.0%	99.9%	Albinism, oculocutaneous, type IA, 203100 Albinism, oculocutaneous, type IB, 606952 Waardenburg syndrome/albinism, digenic, 103470 [Skin/hair/eye pigmentation 3, blue/green eyes], 601800 [Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 {Melanoma, cutaneous malignant, susceptibility to, 8}, 601800
TYROBP	83,3	100.0%	100.0%	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770
TYRP1	152,3	100.0%	100.0%	Albinism, oculocutaneous, type III, 203290 [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271
UBA5	79,9	97.7%	86.6%	?Spinocerebellar ataxia, autosomal recessive 24, 617133 Epileptic encephalopathy, early infantile, 44, 617132
UBE2T	91,1	100.0%	99.3%	Fanconi anemia, complementation group T, 616435
UBE3B	113,5	100.0%	99.7%	Kaufman oculocerebrofacial syndrome, 244450
UBR1	119,9	99.8%	99.0%	Johanson-Blizzard syndrome, 243800

UCHL1	98,7	99.5%	95.4%	Spastic paraplegia 79, autosomal recessive, 615491 {?Parkinson disease 5, susceptibility to}, 613643
UFC1	121,3	100.0%	100.0%	Neurodevelopmental disorder with spasticity and poor growth, 618076
UFM1	104,9	73.2%	69.6%	Leukodystrophy, hypomyelinating, 14, 617899
UGT1A1	184,1	100.0%	100.0%	Crigler-Najjar syndrome, type I, 218800 Crigler-Najjar syndrome, type II, 606785 Hyperbilirubinemia, familial transient neonatal, 237900 [Bilirubin, serum level of, QTL1], 601816 [Gilbert syndrome], 143500
UMPS	149,3	100.0%	98.8%	Orotic aciduria, 258900
UNC13D	108,2	99.8%	98.7%	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC80	111,2	100.0%	99.5%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801
UNG	116,8	99.5%	95.6%	Immunodeficiency with hyper IgM, type 5, 608106
UPB1	143	100.0%	100.0%	Beta-ureidopropionase deficiency, 613161
UQCC2	132,2	100.0%	98.1%	Mitochondrial complex III deficiency, nuclear type 7, 615824
UQCC3	123	100.0%	99.9%	?Mitochondrial complex III deficiency, nuclear type 9, 616111
UQCRB	106	99.2%	95.4%	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC2	105,3	99.6%	97.2%	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRQ	158,9	100.0%	100.0%	Mitochondrial complex III deficiency, nuclear type 4, 615159
UROC1	132,8	100.0%	99.9%	?Urocanase deficiency, 276880
UROD	130,8	98.9%	95.6%	Porphyria cutanea tarda, 176100 Porphyria, hepatoerythropoietic, 176100
UROS	103,8	100.0%	99.7%	Porphyria, congenital erythropoietic, 263700
USB1	118,2	99.8%	97.2%	Poikiloderma with neutropenia, 604173
USH1C	92,5	99.9%	99.2%	Deafness, autosomal recessive 18A, 602092 Usher syndrome, type 1C, 276904
USH1G	191,3	99.9%	98.8%	Usher syndrome, type 1G, 606943
USH2A	129,3	100.0%	99.7%	Retinitis pigmentosa 39, 613809 Usher syndrome, type 2A, 276901
USP18	144,4	95.9%	95.9%	Pseudo-TORCH syndrome 2, 617397
UVSSA	122,6	99.2%	98.9%	UV-sensitive syndrome 3, 614640
VAC14	98,9	99.8%	98.6%	Striatonigral degeneration, childhood-onset, 617054
VARS	129,8	100.0%	99.9%	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802
VARS2	120,1	100.0%	99.8%	Combined oxidative phosphorylation deficiency 20, 615917
VAX1	95,1	99.5%	95.7%	?Microphthalmia, syndromic 11, 614402
VDR	108,8	99.1%	96.0%	?Osteoporosis, involutional, 166710 Rickets, vitamin D-resistant, type IIA, 277440

VHL	169,6	100.0%	98.3%	Erythrocytosis, familial, 2, 263400 Hemangioblastoma, cerebellar, somatic, 0 Pheochromocytoma, 171300 Renal cell carcinoma, somatic, 144700 von Hippel-Lindau syndrome, 193300
VIPAS39	114,7	100.0%	99.9%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VKORC1	146,5	100.0%	99.9%	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473 Warfarin resistance, 122700
VLDLR	141,4	100.0%	99.9%	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VPS11	119,6	95.4%	93.1%	Leukodystrophy, hypomyelinating, 12, 616683
VPS13A	78,2	99.2%	95.3%	Choreoacanthocytosis, 200150
VPS13B	134,5	99.3%	98.0%	Cohen syndrome, 216550
VPS13C	110,2	99.5%	97.0%	Parkinson disease 23, autosomal recessive, early onset, 616840
VPS33A	103,9	96.2%	94.4%	Mucopolysaccharidosis-plus syndrome, 617303
VPS33B	107,2	100.0%	99.9%	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
VPS37A	64,3	91.3%	79.3%	Spastic paraplegia 53, autosomal recessive, 614898
VPS45	126,5	97.3%	94.4%	Neutropenia, severe congenital, 5, autosomal recessive, 615285
VPS53	111,3	91.1%	89.6%	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	129,6	99.8%	98.7%	Pontocerebellar hypoplasia type 1A, 607596
VSX2	120,2	100.0%	99.8%	Microphthalmia with coloboma 3, 610092 Microphthalmia, isolated 2, 610093
VWA3B	124,9	99.9%	98.9%	?Spinocerebellar ataxia, autosomal recessive 22, 616948
VWF	98,2	99.9%	99.1%	von Willebrand disease, type 1, 193400 von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 von Willibrand disease, type 3, 277480
WARS2	132,3	99.9%	99.1%	Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710
WASHC4	108,9	99.3%	95.9%	?Mental retardation, autosomal recessive 43, 615817
WASHC5	134,2	99.9%	99.5%	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, autosomal dominant, 603563
WBP2	94,3	100.0%	99.9%	Deafness, autosomal recessive 107, 617639
WDPCP	106,7	97.8%	94.9%	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR19	126,8	100.0%	99.2%	?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307

WDR34	116,1	100.0%	100.0%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
WDR35	141,8	99.7%	98.4%	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091
WDR45B	72,7	97.5%	90.1%	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977
WDR60	108,1	99.7%	98.1%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WDR62	152,6	100.0%	99.8%	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
WDR72	123,8	96.8%	96.1%	Amelogenesis imperfecta, type IIA3, 613211
WDR73	153,2	100.0%	99.9%	Galloway-Mowat syndrome 1, 251300
WDR81	184,8	100.0%	100.0%	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185 Hydrocephalus, congenital, 3, with brain anomalies, 617967
WEE2	97,9	99.9%	98.7%	Oocyte maturation defect 5, 617996
WFS1	189,9	100.0%	99.9%	?Cataract 41, 116400 Deafness, autosomal dominant 6/14/38, 600965 Wolfram syndrome 1, 222300 Wolfram-like syndrome, autosomal dominant, 614296 {Diabetes mellitus, noninsulin-dependent, association with}, 125853
WHRN	132,8	99.9%	99.0%	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
WIPF1	89,1	100.0%	99.1%	?Wiskott-Aldrich syndrome 2, 614493
WISP3	NC	NC	NC	Arthropathy, progressive pseudorheumatoid, of childhood, 208230 Spondyloepiphyseal dysplasia tarda with progressive arthropathy, 208230
WNK1	134,4	100.0%	99.5%	Neuropathy, hereditary sensory and autonomic, type II, 201300 Pseudohypoaldosteronism, type IIC, 614492
WNT1	255,8	100.0%	99.8%	Osteogenesis imperfecta, type XV, 615220 {Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221
WNT10A	141,8	100.0%	99.9%	Odontoonychodermal dysplasia, 257980 Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400
WNT10B	157	100.0%	100.0%	Split-hand/foot malformation 6, 225300 Tooth agenesis, selective, 8, 617073
WNT3	169,3	100.0%	99.8%	?Tetra-amelia syndrome 1, 273395
WNT4	226,5	99.5%	97.3%	?SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330
WNT7A	195,8	100.0%	100.0%	Fuhrmann syndrome, 228930 Ulna and fibula, absence of, with severe limb deficiency, 276820
WRAP53	162,8	100.0%	100.0%	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	124,8	99.7%	98.8%	Werner syndrome, 277700

WWOX	116,1	100.0%	99.9%	Epileptic encephalopathy, early infantile, 28, 616211 Esophageal squamous cell carcinoma, somatic, 133239 Spinocerebellar ataxia, autosomal recessive 12, 614322
XDH	93,8	100.0%	99.7%	Xanthinuria, type I, 278300
XPA	74,7	99.7%	98.2%	Xeroderma pigmentosum, group A, 278700
XPC	143,5	100.0%	99.8%	Xeroderma pigmentosum, group C, 278720
XPNPEP3	99,9	100.0%	99.4%	Nephronophthisis-like nephropathy 1, 613159
XRCC1	111,4	99.9%	99.1%	?Spinocerebellar ataxia, autosomal recessive 26, 617633
XRCC2	171,8	99.8%	96.5%	?Fanconi anemia, complementation group U, 617247
XRCC4	143	99.9%	99.0%	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	128,1	99.9%	98.2%	Desbuquois dysplasia 2, 615777 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
XYLT2	147,5	99.7%	98.1%	Spondyloocular syndrome, 605822 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
YARS2	175,2	99.9%	99.6%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
YME1L1	103,9	98.2%	93.5%	?Optic atrophy 11, 617302
YY1AP1	143,6	98.6%	97.1%	Grange syndrome, 602531
ZAP70	186,1	100.0%	99.9%	Autoimmune disease, multisystem, infantile-onset, 2, 617006 Immunodeficiency 48, 269840
ZBTB16	148,4	100.0%	100.0%	Leukemia, acute promyelocytic, PL2F/RARA type, 0 Skeletal defects, genital hypoplasia, and mental retardation, 612447
ZBTB24	155,5	100.0%	100.0%	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069
ZBTB42	131,4	100.0%	100.0%	?Lethal congenital contracture syndrome 6, 616248
ZC3H14	152,9	99.8%	98.5%	Mental retardation, autosomal recessive 56, 617125
ZFYVE26	104,8	99.9%	98.7%	Spastic paraplegia 15, autosomal recessive, 270700
ZMPSTE24	128,7	100.0%	99.6%	Mandibuloacral dysplasia with type B lipodystrophy, 608612 Restrictive dermopathy, lethal, 275210
ZMYND10	123	100.0%	100.0%	Ciliary dyskinesia, primary, 22, 615444
ZNF335	134,2	100.0%	99.8%	Microcephaly 10, primary, autosomal recessive, 615095
ZNF408	144,5	100.0%	100.0%	?Exudative vitreoretinopathy 6, 616468 Retinitis pigmentosa 72, 616469
ZNF423	192,9	100.0%	100.0%	Joubert syndrome 19, 614844 Nephronophthisis 14, 614844
ZNF469	157,6	100.0%	100.0%	Brittle cornea syndrome 1, 229200
ZNF513	135,3	100.0%	100.0%	?Retinitis pigmentosa 58, 613617
ZNHIT3	136	74.4%	74.4%	PEHO syndrome, 260565
ZP1	178,3	100.0%	100.0%	Oocyte maturation defect 1, 615774

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan;43(Database issue):D1079-85.
Median Coverage describes the average number of reads seen across 50 exomes.
% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : May 8th, 2019.
This list is accurate for panel version DG 2.16
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors