MULTIPLE CONGENITAL ANOMALIES GENE PANEL DG 2.7/DG 2.8

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
A2M	109.0	99%	99%	Alpha-2-macroglobulin deficiency, 614036 {Alzheimer disease, susceptibility to}, 104300
A4GALT	123.7	100%	100%	NOR polyagglutination syndrome, 111400 [Blood group, P1Pk system, P phenotype], 111400 [Blood group, P1Pk system, p phenotype], 111400
AAAS	77.2	100%	97%	Achalasia-addisonianism-alacrimia syndrome, 231550
AAGAB	133.1	99%	92%	Keratoderma, palmoplantar, punctate type IA, 148600
AARS	112.5	99%	96%	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 Epileptic encephalopathy, early infantile, 29, 616339
AARS2	109.2	99%	97%	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889
AASS	120.8	100%	96%	Hyperlysinemia, 238700 Saccharopinuria, 268700
ABAT	79.3	100%	100%	GABA-transaminase deficiency, 613163
ABCA1	112.7	99%	98%	HDL deficiency, type 2, 604091 Tangier disease, 205400 {Coronary artery disease in familial hypercholesterolemia, protection against}, 143890
ABCA12	131.5	99%	96%	Ichthyosis, autosomal recessive 4B (harlequin), 242500 Ichthyosis, congenital, autosomal recessive 4A, 601277
ABCA3	109.2	99%	98%	Surfactant metabolism dysfunction, pulmonary, 3, 610921
ABCA4	106.1	100%	97%	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	150.3	100%	98%	Cholestasis, benign recurrent intrahepatic, 2, 605479 Cholestasis, progressive familial intrahepatic 2, 601847

ABCB4	110.5	98%	94%	Cholestasis, intrahepatic, of pregnancy, 3, 614972 Cholestasis, progressive familial intrahepatic 3, 602347 Gallbladder disease 1, 600803
ABCB6	106.3	99%	98%	Dyschromatosis universalis hereditaria 3, 615402 Microphthalmia, isolated, with coloboma 7, 614497 Pseudohyperkalemia, familial, 2, due to red cell leak, 609153 [Blood group, Langereis system], 111600
ABCB7	170.5	100%	97%	Anemia, sideroblastic, with ataxia, 301310
ABCC2	132.7	100%	100%	Dubin-Johnson syndrome, 237500
ABCC6	86.5	92%	90%	Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850
ABCC8	128.1	100%	99%	Diabetes mellitus, noninsulin-dependent, 125853 Diabetes mellitus, permanent neonatal, 606176 Diabetes mellitus, transient neonatal 2, 610374 Hyperinsulinemic hypoglycemia, familial, 1, 256450 Hypoglycemia of infancy, leucine-sensitive, 240800
ABCC9	141.6	100%	98%	Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia, 239850
ABCD1	95.4	73%	68%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABCD4	123.5	100%	98%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	129.0	100%	100%	Sitosterolemia, 210250
ABCG8	140.8	100%	96%	Sitosterolemia, 210250 {Gallbladder disease 4}, 611465
ABHD12	89.3	100%	88%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ABHD5	206.1	100%	100%	Chanarin-Dorfman syndrome, 275630
ABL1	121.5	100%	99%	Leukemia, Philadelphia chromosome-positive, resistant to imatinib
ACAD8	111.4	100%	100%	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	113.4	98%	94%	Mitochondrial complex I deficiency due to ACAD9 deficiency, 611126
ACADM	94.6	98%	93%	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	113.7	99%	96%	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	111.4	96%	87%	2-methylbutyrylglycinuria, 610006

ACADVL	103.2	100%	95%	VLCAD deficiency, 201475
ACAN	105.3	91%	82%	Osteochondritis dissecans, short stature, and early-onset osteoarthritis, 165800 Spondyloepimetaphyseal dysplasia, aggrecan type, 612813 Spondyloepiphyseal dysplasia, Kimberley type, 608361
ACAT1	106.6	96%	84%	Alpha-methylacetoacetic aciduria, 203750
ACE	98.6	92%	87%	Renal tubular dysgenesis, 267430 [Angiotensin I-converting enzyme, benign serum increase] {Microvascular complications of diabetes 3}, 612624 {Myocardial infarction, susceptibility to} {SARS, progression of} {Stroke, hemorrhagic}, 614519
ACO2	106.8	94%	88%	Infantile cerebellar-retinal degeneration, 614559 ?Optic atrophy 9, 616289
ACOX1	158.8	100%	100%	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACP5	179.2	100%	97%	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACSF3	123.6	100%	98%	Combined malonic and methylmalonic aciduria, 614265
ACSL4	136.5	98%	89%	Mental retardation, X-linked 63, 300387
ACSL6	109.9	100%	98%	Myelodysplastic syndrome Myelogenous leukemia, acute
ACTA1	82.9	100%	95%	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 ?Myopathy, scapulohumeroperoneal, 616852
ACTA2	126.5	100%	98%	Aortic aneurysm, familial thoracic 6, 611788 Moyamoya disease 5, 614042 Multisystemic smooth muscle dysfunction syndrome, 613834
ACTB	90.3	100%	90%	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ACTC1	143.2	100%	99%	Atrial septal defect 5, 612794 Cardiomyopathy, dilated, 1R, 613424 Cardiomyopathy, hypertrophic, 11, 612098 Left ventricular noncompaction 4, 613424
ACTG1	101.9	100%	100%	Baraitser-Winter syndrome 2, 614583 Deafness, autosomal dominant 20/26, 604717

ACTG2	118.9	99%	96%	Visceral myopathy, 155310
ACTN1	128.7	100%	99%	Bleeding disorder, platelet-type, 15, 615193
ACTN2	132.6	100%	100%	Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158 Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158
ACTN4	113.0	100%	100%	Glomerulosclerosis, focal segmental, 1, 603278
ACVR1	142.8	100%	98%	Fibrodysplasia ossificans progressiva, 135100
ACVR1B	149.7	99%	94%	Pancreatic cancer, somatic
ACVR2B	125.8	95%	90%	Heterotaxy, visceral, 4, autosomal, 613751
ACVRL1	106.2	99%	97%	Telangiectasia, hereditary hemorrhagic, type 2, 600376
ACY1	121.8	99%	95%	Aminoacylase 1 deficiency, 609924
ADA	96.3	98%	95%	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADAM10	131.1	99%	98%	Reticulate acropigmentation of Kitamura, 615537 {Alzheimer disease 18, susceptibility to}, 615590
ADAM9	134.7	100%	94%	Cone-rod dystrophy 9, 612775
ADAMTS10	93.0	99%	96%	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS13	93.0	95%	91%	Thrombotic thrombocytopenic purpura, familial, 274150
ADAMTS17	105.9	88%	87%	Weill-Marchesani-like syndrome, 613195
ADAMTS18	140.7	100%	98%	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
ADAMTS2	108.0	100%	96%	Ehlers-Danlos syndrome, type VIIC, 225410
ADAMTSL2	98.4	87%	82%	Geleophysic dysplasia 1, 231050
ADAMTSL4	77.7	99%	98%	Ectopia lentis et pupillae, 225200 Ectopia lentis, isolated, autosomal recessive, 225100
ADAR	106.4	100%	98%	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
ADAT3	66.4	100%	97%	Mental retardation, autosomal recessive 36, 615286
ADCK3	113.7	100%	99%	Coenzyme Q10 deficiency, primary, 4, 612016
ADCK4	84.7	100%	99%	Nephrotic syndrome, type 9, 615573
ADCY5	113.8	92%	89%	Dyskinesia, familial, with facial myokymia, 606703
ADD3	157.7	99%	98%	Cerebral palsy,spastic quadriplegic,3, 617008
ADIPOQ	113.0	100%	100%	Adiponectin deficiency, 612556
ADK	84.1	99%	93%	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADNP	210.8	100%	99%	Helsmoortel-van der Aa syndrome, 615873
ADRA2B	172.1	100%	100%	Epilepsy, myoclonic, familial adult, 2, 607876

ADSL	152.2	100%	100%	Adenylosuccinase deficiency, 103050
ADSSL1	109.3	98%	90%	Myopathy, distal, 5, 617030
AFF2	152.0	100%	100%	Mental retardation, X-linked, FRAXE type, 309548
AFF4	101.4	96%	95%	CHOPS syndrome, 616368
AFG3L2	96.0	89%	78%	Ataxia, spastic, 5, autosomal recessive, 614487 Spinocerebellar ataxia 28, 610246
AGA	130.4	100%	100%	Aspartylglucosaminuria, 208400
AGBL1	120.0	99%	99%	Corneal dystrophy, Fuchs endothelial, 8, 615523
AGBL5	96.7	99%	97%	Retinitis pigmentosa 75,617023
AGK	113.9	98%	94%	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AGL	133.0	100%	97%	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGPAT2	103.0	99%	94%	Lipodystrophy, congenital generalized, type 1, 608594
AGPS	49.4	94%	78%	Rhizomelic chondrodysplasia punctata, type 3, 600121
AGRN	103.1	97%	90%	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120
AGT	157.6	100%	100%	Renal tubular dysgenesis, 267430 {Hypertension, essential, susceptibility to}, 145500 {Preeclampsia, susceptibility to}
AGTR1	132.6	100%	100%	Renal tubular dysgenesis, 267430 {Hypertension, essential}, 145500
AGXT	125.0	100%	100%	Hyperoxaluria, primary, type 1, 259900
AHCY	106.4	100%	98%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHDC1	110.1	100%	96%	Xia-Gibbs syndrome, 615829
AHI1	123.6	99%	91%	Joubert syndrome-3, 608629
AICDA	115.0	99%	97%	Immunodeficiency with hyper-IgM, type 2, 605258
AIFM1	143.5	100%	100%	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Deafness, X-linked 5, 300614
AIMP1	84.3	97%	91%	Leukodystrophy, hypomyelinating, 3, 260600
AIP	135.3	98%	95%	Pituitary adenoma, ACTH-secreting, 219090 Pituitary adenoma, growth hormone-secreting, 102200 Pituitary adenoma, prolactin-secreting, 600634

AIPL1	107.4	100%	100%	Cone-rod dystrophy, 604393 Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393
AIRE	69.3	98%	91%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AK1	95.6	100%	100%	Hemolytic anemia due to adenylate kinase deficiency, 612631
AK2	91.7	97%	88%	Reticular dysgenesis, 267500
AKR1C2	139.6	95%	84%	46XY sex reversal 8, 614279 Obesity, hyperphagia, and developmental delay
AKR1D1	89.3	93%	87%	Bile acid synthesis defect, congenital, 2, 235555
AKT1	148.4	100%	98%	Breast cancer, somatic, 114480 Colorectal cancer, somatic, 114500 Cowden syndrome 6, 615109 Ovarian cancer, somatic, 167000 Proteus syndrome, somatic, 176920 {Schizophrenia, susceptibility to}, 181500
AKT2	125.5	99%	98%	Diabetes mellitus, type II, 125853 Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900
AKT3	69.2	99%	89%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937
ALAD	99.9	100%	100%	Porphyria, acute hepatic, 612740 {Lead poisoning, susceptibility to}, 612740
ALAS2	110.7	100%	96%	Anemia, sideroblastic, 1, 300751 Protoporphyria, erythropoietic, X-linked, 300752
ALB	141.4	100%	97%	Analbuminemia, 616000 [Dysalbuminemic hyperthyroxinemia], 615999
ALDH18A1	115.4	100%	99%	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	101.2	94%	91%	Microphthalmia, isolated 8, 615113
ALDH2	108.7	100%	98%	Alcohol sensitivity, acute, 610251 {Esophageal cancer, alcohol-related, susceptibility to} {Hangover, susceptibility to}, 610251 {Sublingual nitroglycerin, susceptibility to poor response to}
ALDH3A2	122.0	100%	100%	Sjogren-Larsson syndrome, 270200

ALDH4A1	104.2	98%	97%	Hyperprolinemia, type II, 239510
ALDH5A1	71.5	90%	80%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	113.1	100%	100%	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	62.7	91%	83%	Epilepsy, pyridoxine-dependent, 266100
ALDOA	148.8	100%	96%	Glycogen storage disease XII, 611881
ALDOB	136.6	100%	98%	Fructose intolerance, 229600
ALG1	49.1	52%	47%	Congenital disorder of glycosylation, type Ik, 608540
ALG11	144.7	100%	100%	Congenital disorder of glycosylation, type Ip, 613661
ALG12	132.7	100%	100%	Congenital disorder of glycosylation, type Ig, 607143
ALG13	105.4	100%	96%	Epileptic encephalopathy, early infantile, 36, 300884
ALG2	95.5	100%	100%	Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 ?Congenital disorder of glycosylation, type Ii, 607906
ALG3	99.2	100%	99%	Congenital disorder of glycosylation, type Id, 601110
ALG6	90.4	95%	94%	Congenital disorder of glycosylation, type Ic, 603147
ALG8	118.3	95%	92%	Congenital disorder of glycosylation, type Ih, 608104
ALG9	112.0	100%	98%	Congenital disorder of glycosylation, type Il, 608776 Gillessen-Kaesbach-Nishimura syndrome, 263210
ALMS1	157.8	99%	99%	Alstrom syndrome, 203800
ALOX12B	118.1	100%	98%	Ichthyosis, congenital, autosomal recessive 2, 242100
ALOXE3	117.1	100%	99%	Ichthyosis, congenital, autosomal recessive 3, 606545
ALPL	135.2	100%	100%	Hypophosphatasia, adult, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500 Odontohypophosphatasia, 146300
ALS2	144.4	100%	99%	Amyotrophic lateral sclerosis 2, juvenile, 205100 Primary lateral sclerosis, juvenile, 606353 Spastic paralysis, infantile onset ascending, 607225
ALX3	102.9	82%	69%	Frontonasal dysplasia 1, 136760
ALX4	102.9	94%	84%	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597 {Craniosynostosis 5, susceptibility to}, 615529
AMACR	130.5	100%	99%	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
AMBN	142.2	94%	88%	Amelogenesis imperfecta, type IF, 616270

AMELX	99.8	100%	99%	Amelogenesis imperfecta, type 1E, 301200
AMER1	108.5	100%	99%	Osteopathia striata with cranial sclerosis, 300373
AMH	32.1	96%	67%	Persistent Mullerian duct syndrome, type I, 261550
AMHR2	115.9	99%	98%	Persistent Mullerian duct syndrome, type II, 261550
AMN	51.5	78%	66%	Megaloblastic anemia-1, Norwegian type, 261100
AMPD1	117.1	100%	100%	Myopathy due to myoadenylate deaminase deficiency, 615511
AMPD2	117.6	100%	96%	Pontocerebellar hypoplasia, type 9, 615809 ?Spastic paraplegia 63, 615686
AMT	134.3	100%	98%	Glycine encephalopathy, 605899
ANG	147.9	100%	96%	Amyotrophic lateral sclerosis 9, 611895
ANGPTL3	82.2	92%	88%	Hypobetalipoproteinemia, familial, 2, 605019
ANK1	118.9	100%	98%	Spherocytosis, type 1, 182900
ANK2	133.8	99%	99%	Cardiac arrhythmia, ankyrin-B-related, 600919 Long QT syndrome 4, 600919
ANKH	97.5	100%	100%	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000
ANKK1	120.0	100%	98%	Dopamine receptor D2, reduced brain density of, 608774
ANKRD11	87.9	94%	90%	KBG syndrome, 148050
ANKRD26	71.0	88%	74%	Thrombocytopenia 2, 188000
ANKS6	79.4	91%	84%	Nephronophthisis 16, 615382
ANLN	129.3	98%	91%	Focal segmental glomerulosclerosis 8, 616032
ANO10	102.8	100%	95%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANO3	141.4	99%	98%	Dystonia 24, 615034
ANO5	128.6	99%	95%	Gnathodiaphyseal dysplasia, 166260 Miyoshi muscular dystrophy 3, 613319 Muscular dystrophy, limb-girdle, type 2L, 611307
ANO6	113.4	97%	92%	Scott syndrome, 262890
ANTXR1	113.9	98%	95%	GAPO syndrome, 230740 {Hemangioma, capillary infantile, susceptibility to}, 602089
ANTXR2	79.9	100%	90%	Hyaline fibromatosis syndrome, 228600
AP1S1	104.8	100%	99%	MEDNIK syndrome, 609313
AP1S2	77.4	74%	70%	Mental retardation, X-linked syndromic 5, 304340
AP2S1	132.4	88%	88%	Hypocalciuric hypercalcemia, familial, type III, 600740
AP3B1	90.0	93%	86%	Hermansky-Pudlak syndrome 2, 608233

AP4B1	131.8	99%	97%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	95.1	100%	98%	Spastic paraplegia 51, autosomal recessive, 613744 Stuttering, familial persistent, 1, 184450
AP4M1	98.9	99%	97%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	58.7	68%	68%	Spastic paraplegia 52, autosomal recessive, 614067
AP5Z1	78.1	100%	99%	Spastic paraplegia 48, autosomal recessive, 613647
APC	143.0	100%	99%	Adenoma, periampullary, somatic Adenomatous polyposis coli, 175100 Brain tumor-polyposis syndrome 2, 175100 Colorectal cancer, somatic, 114500 Desmoid disease, hereditary, 135290 Gardner syndrome, 175100 Gastric cancer, somatic, 613659 Hepatoblastoma, somatic, 114550
APCDD1	132.0	98%	93%	Hypotrichosis 1, 605389
APOA1	81.5	100%	100%	Amyloidosis, 3 or more types, 105200 ApoA-I and apoC-III deficiency, combined Corneal clouding, autosomal recessive Hypoalphalipoproteinemia, 604091
APOA2	103.4	81%	81%	Apolipoprotein A-II deficiency, 107670 {Hypercholesterolemia, familial, modifier of}, 143890
APOA5	121.2	100%	100%	Hyperchylomicronemia, late-onset, 144650 {Hypertriglyceridemia, susceptibility to}, 145750
APOB	166.0	99%	99%	Hypercholesterolemia, due to ligand-defective apo B, 144010 Hypobetalipoproteinemia, 615558
APOC2	83.2	100%	100%	Hyperlipoproteinemia, type Ib, 207750
APOC3	75.8	100%	100%	Apolipoprotein C-III deficiency, 614028
APOE	50.3	96%	85%	Alzheimer disease-2, 104310 Hyperlipoproteinemia, type III Lipoprotein glomerulopathy, 611771 Sea-blue histiocyte disease, 269600 {?Macular degeneration, age-related}, 603075 {Myocardial infarction susceptibility}
APOPT1	69.1	87%	86%	Mitochondrial complex IV deficiency, 220110

APP	133.9	100%	100%	Alzheimer disease 1, familial, 104300 Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants, 605714
APRT	56.4	100%	100%	Adenine phosphoribosyltransferase deficiency, 614723
APTX	108.5	94%	91%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
AQP2	109.4	100%	93%	Diabetes insipidus, nephrogenic, 125800
AQP5	94.5	98%	93%	Palmoplantar keratoderma, Bothnian type, 600231
AR	91.4	94%	89%	Androgen insensitivity, 300068 Androgen insensitivity, partial, with or without breast cancer, 312300 Hypospadias 1, X-linked, 300633 Spinal and bulbar muscular atrophy of Kennedy, 313200 {Prostate cancer, susceptibility to}, 176807
ARFGEF2	140.9	99%	98%	Periventricular heterotopia with microcephaly, 608097
ARG1	131.0	100%	100%	Argininemia, 207800
ARHGAP26	126.8	100%	100%	Leukemia, juvenile myelomonocytic, somatic, 607785
ARHGAP31	98.4	99%	97%	Adams-Oliver syndrome 1, 100300
ARHGDIA	121.4	100%	100%	Nephrotic syndrome, type 8, 615244
ARHGEF6	169.9	95%	93%	Mental retardation, X-linked 46, 300436
ARHGEF9	106.1	100%	99%	Epileptic encephalopathy, early infantile, 8, 300607
ARID1A	112.7	93%	88%	Coffin-Siris syndrome 2, 614607
ARID1B	121.6	92%	87%	Coffin-Siris syndrome 1, 135900
ARL13B	88.6	100%	91%	Joubert syndrome 8, 612291
ARL2BP	63.3	86%	76%	Retinitis pigmentosa with or without situs inversus, 615434
ARL6	85.2	99%	93%	Bardet-Biedl syndrome 3, 600151 ?Retinitis pigmentosa 55, 613575 {Bardet-Biedl syndrome 1, modifier of}, 209900
ARMC4	113.7	90%	89%	Ciliary dyskinesia, primary, 23, 615451
ARMC5	99.8	100%	99%	ACTH-independent macronodular adrenal hyperplasia 2, 615954
ARSA	91.7	100%	100%	Metachromatic leukodystrophy, 250100
ARSB	115.5	98%	94%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARSE	109.5	100%	98%	Chondrodysplasia punctata, X-linked recessive, 302950
ARV1	122.2	100%	98%	Epileptic encephalopathy, early infantile, 38, 617020

ARX	38.5	87%	73%	Epileptic encephalopathy, early infantile, 1, 308350 Hydranencephaly with abnormal genitalia, 300215 Lissencephaly, X-linked 2, 300215 Mental retardation, X-linked 29 and others, 300419 Partington syndrome, 309510 Proud syndrome, 300004
ASAH1	109.1	99%	84%	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASB10	97.0	100%	99%	Glaucoma 1, open angle, F, 603383
ASCC1	141.7	93%	91%	Barrett esophagus/esophageal adenocarcinoma, 614266 ?Spinal muscular atrophy with congenital bone fractures 2,616867
ASCL1	155.3	79%	73%	Central hypoventilation syndrome, congenital, 209880 Haddad syndrome, 209880
ASL	91.7	100%	98%	Argininosuccinic aciduria, 207900
ASNS	85.2	97%	87%	Asparagine synthetase deficiency, 615574
ASPA	112.6	97%	91%	Canavan disease, 271900
ASPH	105.2	97%	91%	Traboulsi syndrome, 601552
ASPM	94.8	97%	91%	Microcephaly 5, primary, autosomal recessive, 608716
ASPSCR1	83.6	100%	96%	Alveolar soft-part sarcoma, 606243
ASS1	81.1	98%	89%	Citrullinemia, 215700
ASXL1	135.3	99%	98%	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ASXL3	139.1	99%	98%	Bainbridge-Ropers syndrome, 615485
ATCAY	136.6	100%	99%	Ataxia, cerebellar, Cayman type, 601238
ATF6	122.4	100%	99%	Achromatopsia 7, 616517
ATIC	104.3	100%	98%	AICA-ribosiduria due to ATIC deficiency, 608688
ATL1	162.1	99%	93%	Neuropathy, hereditary sensory, type ID, 613708 Spastic paraplegia 3A, autosomal dominant, 182600
ATL3	110.4	96%	93%	Neuropathy, hereditary sensory, type IF, 615632
ATM	101.4	98%	92%	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic Lymphoma, mantle cell, somatic T-cell prolymphocytic leukemia, somatic {Breast cancer, susceptibility to}, 114480

ATN1	98.3	98%	94%	Dentatorubro-pallidoluysian atrophy, 125370
ATOH7	82.8	91%	80%	Persistent hyperplastic primary vitreous, autosomal recessive, 221900
ATP13A2	106.7	100%	98%	Kufor-Rakeb syndrome, 606693 ?Ceroid lipofuscinosis, neuronal, 12, 606693
ATP1A2	170.2	100%	100%	Alternating hemiplegia of childhood, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481
ATP1A3	161.8	100%	100%	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235
ATP2A1	136.4	100%	100%	Brody myopathy, 601003
ATP2A2	153.1	100%	98%	Acrokeratosis verruciformis, 101900 Darier disease, 124200
ATP2C1	113.4	100%	97%	Hailey-Hailey disease, 169600
ATP6AP1	128.2	100%	100%	Immunodeficiency 47, 300972
ATP6V0A2	122.9	100%	100%	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250
ATP6V0A4	108.7	99%	97%	Renal tubular acidosis, distal, autosomal recessive, 602722
ATP6V1B1	158.9	100%	100%	Renal tubular acidosis with deafness, 267300
ATP6V1B2	133.9	100%	99%	Deafness, congenital, with onychodystrophy, autosomal dominant, 124480 Zimmermann-Laband syndrome 2, 616455
ATP7A	159.5	99%	98%	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATP7B	134.6	100%	98%	Wilson disease, 277900
ATP8B1	133.1	96%	93%	Cholestasis, benign recurrent intrahepatic, 243300 Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, progressive familial intrahepatic 1, 211600
ATPAF2	90.3	100%	98%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
ATR	129.0	98%	95%	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
ATRX	96.7	99%	97%	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Alpha-thalassemia/mental retardation syndrome, 301040 Mental retardation-hypotonic facies syndrome, X-linked, 309580

ATXN1	125.8	100%	100%	Spinocerebellar ataxia 1, 164400
ATXN10	133.1	95%	92%	Spinocerebellar ataxia 10, 603516
ATXN2	87.7	91%	79%	Spinocerebellar ataxia 2, 183090 {Amyotrophic lateral sclerosis, susceptibility to, 13}, 183090 {Parkinson disease, late-onset, susceptibility to}, 168600
ATXN3	92.8	98%	90%	Machado-Joseph disease, 109150
ATXN7	110.3	95%	91%	Spinocerebellar ataxia 7, 164500
ATXN8OS	NC	NC	NC	Spinocerebellar ataxia 8, 608768
AUH	75.2	100%	95%	3-methylglutaconic aciduria, type I, 250950
AURKC	75.0	99%	94%	Spermatogenic failure 5,243060
AUTS2	89.7	96%	93%	Mental retardation, autosomal dominant 26,615834
AVP	35.3	84%	37%	Diabetes insipidus, neurohypophyseal, 125700
AVPR2	139.2	100%	97%	Diabetes insipidus, nephrogenic, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539
AXIN1	130.6	99%	97%	Hepatocellular carcinoma, somatic, 114550 ?Caudal duplication anomaly, 607864
AXIN2	102.6	100%	98%	Colorectal cancer, somatic, 114500 Oligodontia-colorectal cancer syndrome, 608615
B2M	276.5	100%	100%	Immunodeficiency 43, 241600 ?Amyloidosis, familial visceral, 105200
B3GALNT2	102.7	92%	91%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B3GALT6	47.2	74%	69%	Ehlers-Danlos syndrome, progeroid type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B3GALTL	81.5	92%	86%	Peters-plus syndrome, 261540
B3GAT3	79.7	100%	92%	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B3GNT1	103.0	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
B4GALNT1	122.6	99%	90%	Spastic paraplegia 26, autosomal recessive, 609195
B4GALT1	114.6	100%	98%	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	84.2	99%	94%	Ehlers-Danlos syndrome with short stature and limb anomalies, 130070
B9D2	82.6	100%	100%	Meckel syndrome 10, 614175
BAAT	112.3	97%	93%	Hypercholanemia, familial, 607748
BAG3	110.6	100%	98%	Cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954

BANF1	60.0	99%	95%	Nestor-Guillermo progeria syndrome, 614008
BAP1	118.9	100%	98%	Tumor predisposition syndrome, 614327
BAX	70.4	85%	84%	Colorectal cancer, somatic, 114500 T-cell acute lymphoblastic leukemia, somatic, 613065
BBS1	134.0	100%	100%	Bardet-Biedl syndrome 1, 209900
BBS10	143.8	100%	99%	Bardet-Biedl syndrome 10, 615987
BBS12	175.6	100%	100%	Bardet-Biedl syndrome 12, 615989
BBS2	168.7	100%	100%	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	129.9	100%	98%	Bardet-Biedl syndrome 4, 615982
BBS5	104.0	96%	90%	Bardet-Biedl syndrome 5, 615983
BBS7	113.1	93%	87%	Bardet-Biedl syndrome 7, 615984
BBS9	96.9	95%	92%	Bardet-Biedl syndrome 9, 615986
BCAP31	77.9	99%	92%	Deafness, dystonia, and cerebral hypomyelination, 300475
BCHE	147.0	100%	98%	Apnea, postanesthetic, 177400
BCKDHA	140.4	100%	98%	Maple syrup urine disease, type Ia, 248600
BCKDHB	93.9	93%	90%	Maple syrup urine disease, type Ib, 248600
BCKDK	144.0	100%	100%	Branched-chain ketoacid dehydrogenase kinase deficiency, 614923
BCL10	82.6	100%	98%	Lymphoma, MALT, somatic, 137245 ?Immunodeficiency 37, 616098 {Lymphoma, follicular, somatic}, 605027 {Male germ cell tumor, somatic}, 273300, {Mesothelioma, somatic}, 156240 {Sezary syndrome, somatic},
BCL11A	121.7	98%	97%	Intellectual development disorder with persistence of fetal hemoglobin, 617101
BCL2	109.5	100%	90%	Leukemia/lymphoma, B-cell, 2, 151430
BCL7A	127.0	100%	100%	B-cell non-Hodgkin lymphoma, high-grade
BCMO1	151.4	100%	100%	Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300
BCOR	135.5	100%	98%	Microphthalmia, syndromic 2, 300166
BCR	93.8	88%	82%	Leukemia, acute lymphocytic, somatic, 613065 Leukemia, chronic myeloid, somatic, 608232
BCS1L	145.9	100%	100%	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000

				Mitochondrial complex III deficiency, nuclear type 1, 124000
BDNF	152.9	100%	100%	Central hypoventilation syndrome, congenital, 209880 {Anorexia nervosa, susceptibility to}, 610269 {Bulimia nervosa, age of onset of weight loss in}, 607499 {Memory impairment, susceptibility to} {Obsessive-compulsive disorder, protection against}, 164230
BEAN1	102.6	97%	87%	Spinocerebellar ataxia 31,117210
BEST1	120.9	97%	92%	Bestrophinopathy, autosomal recessive, 611809 Macular dystrophy, vitelliform, 2, 153700 Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma, 193220 Retinitis pigmentosa, concentric, 613194 Retinitis pigmentosa-50, 613194 Vitreoretinochoroidopathy, 193220
BFSP1	90.5	99%	85%	Cataract 33, 611391
BFSP2	78.8	97%	94%	Cataract 12, multiple types, 611597
BGN	136.1	100%	100%	Spondyloepimetaphyseal dysplasia, X-linked, 300106
BHLHA9	12.2	51%	37%	Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432 ?Camptosynpolydactyly, complex, 607539
BICD2	125.6	99%	99%	Spinal muscular atrophy, lower extremity-predominant, 2, AD, 615290
BIN1	98.2	100%	93%	Myopathy, centronuclear, autosomal recessive, 255200
BLK	95.6	100%	100%	Maturity-onset diabetes of the young, type 11, 613375
BLM	118.7	98%	94%	Bloom syndrome, 210900
BLNK	96.2	92%	88%	Agammaglobulinemia 4, 613502
BLOC1S3	32.8	95%	70%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	72.6	98%	84%	Hermansky-pudlak syndrome 9, 614171
BLVRA	112.3	100%	96%	Hyperbiliverdinemia, 614156
BMP1	126.9	100%	99%	Osteogenesis imperfecta, type XIII, 614856
BMP15	129.3	100%	100%	Ovarian dysgenesis 2, 300510 Premature ovarian failure 4, 300510
BMP2	166.6	100%	100%	Brachydactyly, type A2, 112600 {HFE hemochromatosis, modifier of}, 235200

BMP4	127.2	100%	99%	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625
BMPER	137.0	100%	97%	Diaphanospondylodysostosis, 608022
BMPR1A	105.6	100%	96%	Juvenile polyposis syndrome, infantile form, 174900 Polyposis syndrome, hereditary mixed, 2, 610069 Polyposis, juvenile intestinal, 174900
BMPR1B	157.2	100%	100%	Acromesomelic dysplasia, Demirhan type, 609441 Brachydactyly, type A1, D, 616849 Brachydactyly, type A2, 112600
BMPR2	167.3	100%	100%	Pulmonary hypertension, familial primary, 1, with or without HHT, 178600 Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600 Pulmonary venoocclusive disease 1, 265450
BOLA3	44.3	94%	76%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BPGM	98.9	100%	100%	Erythrocytosis due to bisphosphoglycerate mutase deficiency, 222800
BRAF	64.8	85%	76%	Adenocarcinoma of lung, somatic, 211980 Cardiofaciocutaneous syndrome, 115150 Colorectal cancer, somatic LEOPARD syndrome 3, 613707 Melanoma, malignant, somatic Nonsmall cell lung cancer, somatic Noonan syndrome 7, 613706
BRAT1	87.3	100%	96%	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BRCA2	89.3	97%	95%	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	91.2	95%	89%	Cerebellofaciodental syndrome, 616202
BRIP1	114.3	100%	96%	Breast cancer, early-onset, 114480 Fanconi anemia, complementation group J, 609054
BRWD3	126.1	98%	95%	Mental retardation, X-linked 93, 300659

BSCL2	99.3	100%	100%	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	135.5	100%	99%	Bartter syndrome, type 4a, 602522 Sensorineural deafness with mild renal dysfunction, 602522
BTD	125.5	100%	98%	Biotinidase deficiency, 253260
BTK	158.3	100%	100%	Agammaglobulinemia and isolated hormone deficiency, 307200 Agammaglobulinemia, X-linked 1, 300755
BUB1	126.5	99%	94%	Colorectal cancer with chromosomal instability, somatic
BUB1B	144.3	98%	97%	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430
C10orf11	128.5	99%	99%	Albinism, oculocutaneous, type VII, 615179
C10orf2	152.5	100%	100%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Perrault syndrome 5, 616138 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286
C11orf73	52.0	93%	76%	Leukodystrophy,hypomyelinating,13,616881
C12orf57	126.7	100%	100%	Temtamy syndrome, 218340
C12orf65	80.1	97%	91%	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035
C15orf41	117.6	100%	99%	Dyserythropoietic anemia, congenital, type Ib, 615631
C19orf12	92.2	100%	99%	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
C1GALT1C1	146.2	100%	100%	Tn polyagglutination syndrome, somatic, 300622
C1QA	101.1	100%	98%	C1q deficiency, 613652
C1QB	151.4	100%	100%	C1q deficiency, 613652
C1QC	192.1	100%	100%	C1q deficiency, 613652
C1QTNF5	111.5	85%	67%	Retinal degeneration, late-onset, autosomal dominant, 605670
C1S	126.0	100%	100%	C1s deficiency, 613783
C2	13.7	50%	17%	C2 deficiency, 217000 {Macular degeneration, age-related, 14, reduced risk of}, 615489
C21orf59	124.9	100%	99%	Ciliary dyskinesia, primary, 26, 615500
C2orf71	105.9	99%	97%	Retinitis pigmentosa 54, 613428

C3	135.2	100%	99%	C3 deficiency, 613779 {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 {Macular degeneration, age-related, 9}, 611378
C4A	17.6	66%	31%	C4a deficiency, 614380 [Blood group, Rodgers], 614374
C4B	14.0	61%	17%	C4B deficiency, 614379
C4orf26	177.7	100%	100%	Amelogenesis imperfecta, type IIA4, 614832
C5	121.5	96%	91%	C5 deficiency, 609536 [Eculizumab, poor response to], 615749
C5orf42	112.4	98%	94%	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
C6	145.5	100%	99%	C6 deficiency, 612446 Combined C6/C7 deficiency
C7	126.2	100%	90%	C7 deficiency, 610102
C7orf10	112.5	95%	91%	Glutaric aciduria III, 231690
C8A	102.6	100%	99%	C8 deficiency, type I, 613790
C8B	120.4	100%	98%	C8 deficiency, type II, 613789
C8orf37	100.1	100%	99%	Cone-rod dystrophy 16, 614500 Retinitis pigmentosa 64, 614500
C9	126.8	100%	99%	C9 deficiency, 613825 {Macular degeneration, age-related, 15, susceptibility to}, 615591
C9orf72	104.5	100%	95%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1, 105550
CA12	103.4	100%	100%	Hyperchlorhidrosis, isolated, 143860
CA2	126.8	100%	97%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA4	133.0	100%	98%	Retinitis pigmentosa 17, 600852
CA5A	97.7	100%	94%	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CA8	95.6	94%	89%	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CABP2	76.5	99%	95%	Deafness, autosomal recessive 93, 614899
CABP4	93.4	100%	99%	Cone-rod synaptic disorder, congenital nonprogressive, 610427
CACNA1A	82.3	94%	89%	Episodic ataxia, type 2, 108500 Migraine, familial hemiplegic, 1, 141500 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Spinocerebellar ataxia 6, 183086
CACNA1C	134.1	100%	98%	Brugada syndrome 3, 611875

				Timothy syndrome, 601005
CACNA1D	135.8	100%	99%	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896
CACNA1F	115.3	100%	98%	Aland Island eye disease, 300600 Cone-rod dystrophy, X-linked, 3, 300476 Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071
CACNA1G	115.8	99%	97%	Spinocerebellar ataxia 42, 616795
CACNA1H	101.1	98%	95%	Hyperaldosteronism, familial, type IV, 617027 {Epilepsy, childhood absence, susceptibility to, 6}, 611942 {Epilepsy, idiopathic generalized, susceptibility to, 6}, 611942
CACNA1S	120.7	100%	99%	Hypokalemic periodic paralysis, type 1, 170400 {Malignant hyperthermia susceptibility 5}, 601887 {Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580
CACNA2D4	98.1	98%	96%	Retinal cone dystrophy 4, 610478
CACNB2	129.5	99%	95%	Brugada syndrome 4, 611876
CACNB4	105.0	98%	94%	Episodic ataxia, type 5, 613855 {Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 {Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682
CACNG2	130.8	100%	100%	Mental retardation, autosomal dominant 10, 614256
CALM1	124.2	100%	100%	Long QT syndrome 14, 616247 Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916
CALM2	50.0	67%	64%	Long QT syndrome 15, 616249
CALR	90.6	100%	94%	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
CAMTA1	148.8	100%	99%	Cerebellar ataxia, nonprogressive, with mental retardation, 614756
CANT1	125.1	100%	100%	Desbuquois dysplasia 1, 251450
CAPN1	130.2	100%	100%	Spastic paraplegia 76, autosomal recessive, 616907
CAPN3	106.5	99%	94%	Muscular dystrophy, limb-girdle, type 2A, 253600
CAPN5	132.0	100%	93%	Vitreoretinopathy, neovascular inflammatory, 193235
CARD11	135.0	98%	97%	B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11, 615206
CARD14	95.7	99%	95%	Pityriasis rubra pilaris, 173200 Psoriasis 2, 602723
CARD9	101.8	97%	95%	Candidiasis, familial, 2, autosomal recessive, 212050

CARS2	108.9	100%	100%	Combined oxidative phosphorylation deficiency 27, 616672
CASC5	94.5	96%	93%	Microcephaly 4, primary, autosomal recessive, 604321
CASK	123.8	100%	99%	FG syndrome 4, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 Mental retardation, with or without nystagmus, 300422
CASP10	98.1	98%	95%	Autoimmune lymphoproliferative syndrome, type II, 603909 Gastric cancer, somatic, 613659 Lymphoma, non-Hodgkin, somatic, 605027
CASP8	141.5	100%	99%	Hepatocellular carcinoma, somatic, 114550 ?Autoimmune lymphoproliferative syndrome, type IIB, 607271 {Breast cancer, protection against}, 114480 {Lung cancer, protection against}, 211980
CASQ1	102.4	100%	99%	Myopathy, vacuolar, with CASQ1 aggregates, 616231
CASQ2	135.5	100%	98%	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CASR	141.5	99%	99%	Hypercalciuric hypercalcemia Hyperparathyroidism, neonatal, 239200 Hypocalcemia, autosomal dominant, 601198 Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 Hypocalciuric hypercalcemia, type I, 145980 {Calcium, serum level of} {Epilepsy idiopathic generalized, susceptibility to, 8}, 612899
CAST	97.4	94%	91%	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295
CAT	141.3	100%	100%	Acatalasemia, 614097
CATSPER1	146.6	100%	99%	Spermatogenic failure 7, 612997
CAV1	209.7	100%	100%	Pulmonary hypertension, primary, 3, 615343 ?Lipodystrophy, congenital generalized, type 3, 612526 ?Partial lipodystrophy, congenital cataracts, and neurodegeneration syndrome, 606721
CAV3	219.6	100%	100%	Cardiomyopathy, familial hypertrophic, 192600 Creatine phosphokinase, elevated serum, 123320 Long QT syndrome 9, 611818 Muscular dystrophy, limb-girdle, type IC, 607801 Myopathy, distal, Tateyama type, 614321 Rippling muscle disease, 606072
CBL	117.3	99%	98%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia,607785

CBS	97.6	96%	89%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CC2D1A	102.1	100%	98%	Mental retardation, autosomal recessive 3, 608443
CC2D2A	111.9	99%	96%	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284
CCBE1	63.1	95%	84%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CCDC103	88.1	100%	97%	Ciliary dyskinesia, primary, 17, 614679
CCDC11	140.0	97%	94%	Heterotaxy, visceral, 6, autosomal recessive, 614779
CCDC114	106.5	100%	99%	Ciliary dyskinesia, primary, 20, 615067
CCDC115	69.5	89%	88%	Congenital disorder of glycosylation, type IIo, 616828
CCDC151	105.7	100%	98%	Ciliary dyskinesia, primary, 30, 616037
CCDC174	102.3	95%	91%	Hypotonia, infantile, with psychomotor retardation, 616816
CCDC22	114.1	98%	94%	Ritscher-Schinzel syndrome 2, 300963
CCDC39	79.1	95%	90%	Ciliary dyskinesia, primary, 14, 613807
CCDC40	99.4	98%	98%	Ciliary dyskinesia, primary, 15, 613808
CCDC41	95.6	94%	87%	Nephronophthisis 18, 615862
CCDC65	79.0	100%	81%	Ciliary dyskinesia, primary, 27, 615504
CCDC78	97.4	100%	100%	Myopathy, centronuclear, 4, 614807
CCDC8	82.8	100%	100%	3-M syndrome 3, 614205
CCDC88A	71.6	95%	83%	PEHO syndrome, 260565
CCDC88C	95.4	98%	95%	Hydrocephalus, nonsyndromic, autosomal recessive, 236600 ?Spinocerebellar ataxia 40, 616053
CCM2	132.2	97%	97%	Cerebral cavernous malformations-2,603284
CCND2	143.1	100%	100%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938
CCNO	75.2	99%	94%	Ciliary dyskinesia, primary, 29, 615872
CCT5	125.0	100%	99%	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CD151	116.9	100%	100%	Nephropathy with pretibial epidermolysis bullosa and deafness, 609057 [Blood group, Raph], 179620
CD19	77.8	100%	98%	Immunodeficiency, common variable, 3, 613493
CD27	84.2	99%	92%	Lymphoproliferative syndrome 2, 615122
CD2AP	81.8	99%	96%	Glomerulosclerosis, focal segmental, 3, 607832
CD320	77.7	100%	99%	Methylmalonic aciduria due to transcobalamin receptor defect, 613646

CD36	113.1	100%	97%	Platelet glycoprotein IV deficiency, 608404 [Macrothrombocytopenia] {Coronary heart disease, susceptibility to, 7}, 610938 {Malaria, cerebral, reduced risk of}, 611162 {Malaria, cerebral, susceptibility to}, 611162
CD3D	166.1	100%	100%	Immunodeficiency 19, 615617
CD3E	164.4	100%	99%	Immunodeficiency 18, 615615 Immunodeficiency 18, SCID variant, 615615
CD3G	171.8	100%	100%	Immunodeficiency 17, CD3 gamma deficient, 615607
CD4	91.7	100%	100%	OKT4 epitope deficiency, 613949
CD40	160.1	100%	96%	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	174.2	99%	99%	Immunodeficiency, X-linked, with hyper-IgM, 308230
CD59	196.1	90%	84%	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD79A	122.0	100%	97%	Agammaglobulinemia 3, 613501
CD79B	194.1	100%	100%	Agammaglobulinemia 6, 612692
CD81	129.0	100%	99%	Immunodeficiency, common variable, 6, 613496
CD8A	89.8	98%	97%	CD8 deficiency, familial, 608957
CD96	148.5	100%	99%	C syndrome, 211750
CDAN1	89.1	96%	94%	Dyserythropoietic anemia, congenital, type Ia, 224120
CDC14A	133.0	94%	88%	Deafness,autosomal recessive 105,616958
CDC42	80.8	97%	93%	Takenouchi-Kosaki syndrome, 616737
CDC45	144.3	99%	97%	Meier-Gorlin syndrome 7, 617063
CDC6	148.5	100%	99%	Meier-Gorlin syndrome 5, 613805
CDC73	85.8	100%	96%	Hyperparathyroidism, familial primary, 145000 Hyperparathyroidism-jaw tumor syndrome, 145001 Parathyroid adenoma with cystic changes, 145001 Parathyroid carcinoma, 608266
CDCA7	85.3	98%	95%	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910
CDH1	94.4	99%	97%	Endometrial carcinoma, somatic, 608089 Gastric cancer, familial diffuse, with or without cleft lip and/or palate, 137215 Ovarian carcinoma, somatic, 167000 {Breast cancer, lobular}, 114480 {Prostate cancer, susceptibility to}, 176807
CDH15	107.3	99%	95%	Mental retardation, autosomal dominant 3, 612580

CDH23	167.6	100%	99%	Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D, 601067 Usher syndrome, type 1D/F digenic, 601067
CDH3	121.8	99%	95%	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 Hypotrichosis, congenital, with juvenile macular dystrophy, 601553
CDHR1	130.3	100%	98%	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660
CDK5RAP2	116.7	98%	97%	Microcephaly 3, primary, autosomal recessive, 604804
CDKL5	138.6	99%	96%	Epileptic encephalopathy, early infantile, 2, 300672
CDKN1B	62.5	100%	89%	Multiple endocrine neoplasia, type IV, 610755
CDKN1C	32.7	74%	57%	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732
CDKN2A	63.1	92%	90%	Melanoma and neural system tumor syndrome, 155755 Orolaryngeal cancer, multiple, Pancreatic cancer/melanoma syndrome, 606719 {Melanoma, cutaneous malignant, 2}, 155601
CDON	118.9	100%	98%	Holoprosencephaly 11, 614226
CDSN	9.7	37%	11%	Hypotrichosis 2, 146520 Peeling skin syndrome 1, 270300
CDT1	69.2	93%	88%	Meier-Gorlin syndrome 4, 613804
CEACAM16	111.1	100%	98%	Deafness, autosomal dominant 4B, 614614
CEBPA	41.3	86%	61%	Leukemia, acute myeloid, somatic, 601626 ?Leukemia, acute myeloid, 601626
CEBPE	62.7	98%	96%	Specific granule deficiency, 245480
CECR1	85.9	100%	97%	Polyarteritis nodosa, childhood-onset, 615688 ?Sneddon syndrome, 182410
CEL	109.2	84%	81%	Maturity-onset diabetes of the young, type VIII, 609812
CENPF	121.4	99%	97%	Stromme syndrome, 243605
CENPJ	129.4	99%	95%	Microcephaly 6, primary, autosomal recessive, 608393 ?Seckel syndrome 4, 613676
CEP104	106.2	98%	96%	Joubert syndrome 25, 616781
CEP120	117.4	100%	98%	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP152	157.8	97%	94%	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823

CEP164	74.4	98%	93%	Nephronophthisis 15, 614845
CEP19	173.5	100%	100%	Morbid obesity and spermatogenic failure, 615703
CEP290	64.3	85%	74%	Joubert syndrome 5, 610188 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Senior-Loken syndrome 6, 610189 ?Bardet-Biedl syndrome 14, 615991
CEP41	76.4	94%	86%	Joubert syndrome 15, 614464
CEP57	91.9	100%	92%	Mosaic variegated aneuploidy syndrome 2, 614114
CERKL	80.8	97%	88%	Retinitis pigmentosa 26, 608380
CERS3	115.0	100%	100%	Ichthyosis, congenital, autosomal recessive 9, 615023
CES1	129.5	100%	97%	Carboxylesterase 1 deficiency, 114835
CETP	122.3	100%	100%	Hyperalphalipoproteinemia, 143470 [High density lipoprotein cholesterol level QTL 10], 143470
CFC1	46.7	76%	51%	Heterotaxy, visceral, 2, autosomal, 605376
CFD	72.2	85%	81%	Complement factor D deficiency, 613912
CFH	156.9	98%	95%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814 {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 {Macular degeneration, age-related, 4}, 610698
CFHR5	92.7	98%	92%	Nephropathy due to CFHR5 deficiency, 614809
CFI	140.4	97%	96%	Complement factor I deficiency, 610984 {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 {Macular degeneration, age-related, 13, susceptibility to}, 615439
CFL2	118.3	85%	84%	Nemaline myopathy 7, autosomal recessive, 610687
CFP	101.9	100%	96%	Properdin deficiency,X-linked, 312060
CFTR	113.2	98%	92%	Congenital bilateral absence of vas deferens, 277180 Cystic fibrosis, 219700 Sweat chloride elevation without CF {Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400 {Hypertrypsinemia, neonatal} {Pancreatitis, idiopathic}, 167800
CHAMP1	149.4	100%	100%	Mental retardation, autosomal dominant 40, 616579
CHAT	110.9	90%	83%	Myasthenic syndrome, congenital, 6, presynaptic, 254210

CHCHD10	22.1	39%	33%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 Spinal muscular atrophy, Jokela type, 615048 ?Myopathy, isolated mitochondrial, autosomal dominant, 616209
CHCHD2	85.1	99%	87%	Parkinson disease 22, autosomal dominant, 616710
CHD2	118.6	99%	95%	Epileptic encephalopathy, childhood-onset, 615369
CHD7	126.0	99%	98%	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CHEK2	88.1	83%	76%	Li-Fraumeni syndrome, 609265 Osteosarcoma, somatic, 259500 {Breast and colorectal cancer, susceptibility to} {Breast cancer, susceptibility to}, 114480 {Prostate cancer, familial, susceptibility to}, 176807
CHKB	75.2	91%	90%	Muscular dystrophy, congenital, megaconial type, 602541
CHM	132.9	99%	97%	Choroideremia, 303100
CHMP1A	98.2	100%	100%	Pontocerebellar hypoplasia, type 8, 614961
CHMP2B	74.5	94%	70%	Amyotrophic lateral sclerosis 17, 614696 Dementia, familial, nonspecific, 600795
CHMP4B	113.2	100%	98%	Cataract 31, multiple types, 605387
CHN1	131.2	97%	97%	Duane retraction syndrome 2, 604356
CHRDL1	143.2	100%	100%	Megalocornea 1, X-linked 309300
CHRNA1	110.3	100%	100%	Multiple pterygium syndrome, lethal type, 253290 Myasthenic syndrome, congenital, 1A, slow-channel, 601462 Myasthenic syndrome, congenital, 1B, fast-channel, 608930
CHRNA2	193.8	100%	100%	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA4	134.3	96%	95%	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction, susceptibility to}, 188890
CHRNB1	137.3	100%	98%	Myasthenic syndrome, congenital, 2A, slow-channel, 616313 ?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314
CHRNB2	187.5	99%	97%	Epilepsy, nocturnal frontal lobe, 3, 605375
CHRND	143.3	100%	99%	Multiple pterygium syndrome, lethal type, 253290 Myasthenic syndrome, congenital, 3B, fast-channel, 616322 ?Myasthenic syndrome, congenital, 3A, slow-channel, 616321 ?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323

CHRNE	122.7	100%	99%	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	153.5	100%	100%	Escobar syndrome, 265000 Multiple pterygium syndrome, lethal type, 253290
CHST14	145.0	94%	93%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	83.9	100%	95%	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHST6	262.4	100%	100%	Macular corneal dystrophy, 217800
CHSY1	119.9	94%	93%	Temtamy preaxial brachydactyly syndrome, 605282
CHUK	115.6	100%	95%	Cocoon syndrome, 613630
CIB2	201.8	100%	100%	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CIITA	105.4	100%	98%	Bare lymphocyte syndrome, type II, complementation group A, 209920 {Rheumatoid arthritis, susceptibility to}, 180300
CIRH1A	163.0	100%	100%	Cirrhosis, North American Indian childhood type, 604901
CISD2	122.8	83%	83%	Wolfram syndrome 2, 604928
CITED2	99.5	100%	100%	Atrial septal defect 8, 614433 Ventricular septal defect 2, 614431
CKAP2L	162.4	99%	94%	Filippi syndrome, 272440
CLCF1	92.3	100%	97%	Cold-induced sweating syndrome 2, 610313
CLCN1	122.7	99%	98%	Myotonia congenita, dominant, 160800 Myotonia congenita, recessive, 255700 Myotonia levior, recessive
CLCN2	93.2	100%	97%	Leukoencephalopathy with ataxia, 615651 {Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628 {Epilepsy, juvenile absence, susceptibility to, 2}, 607628 {Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628
CLCN4	129.3	100%	100%	Mental retardation, X-linked 49/15, 300114
CLCN5	152.6	99%	99%	Dent disease, 300009 Hypophosphatemic rickets, 300554 Nephrolithiasis, type I, 310468 Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990
CLCN7	118.0	98%	96%	Osteopetrosis, autosomal dominant 2, 166600 Osteopetrosis, autosomal recessive 4, 611490

CLCNKA	89.0	97%	90%	Bartter syndrome, type 4b, digenic, 613090
CLCNKB	88.1	96%	89%	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090
CLDN1	125.4	100%	100%	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626
CLDN14	102.3	100%	100%	Deafness, autosomal recessive 29, 614035
CLDN16	135.0	100%	99%	Hypomagnesemia 3, renal, 248250
CLDN19	111.6	96%	93%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLEC7A	119.3	100%	98%	Candidiasis, familial, 4, autosomal recessive, 613108 {Aspergillosis, susceptibility to}, 614079
CLMP	93.1	100%	98%	Congenital short bowel syndrome, 615237
CLN3	104.9	100%	96%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	136.2	93%	86%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	110.2	95%	92%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	195.1	100%	100%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLP1	147.8	100%	100%	Pontocerebellar hypoplasia, type 10, 615803
CLPB	118.3	96%	96%	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CLPP	94.8	96%	87%	Perrault syndrome 3, 614129
CLRN1	126.0	100%	100%	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902
CNBP	107.6	100%	100%	Myotonic dystrophy 2,602668
CNGA1	102.3	84%	80%	Retinitis pigmentosa 49, 613756
CNGA3	139.4	100%	97%	Achromatopsia-2, 216900
CNGB1	90.5	98%	94%	Retinitis pigmentosa 45, 613767
CNGB3	98.5	99%	87%	Achromatopsia-3, 262300 Macular degeneration, juvenile, 248200
CNNM2	162.3	100%	99%	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNNM4	161.8	98%	97%	Jalili syndrome, 217080
CNTNAP1	137.7	98%	96%	Lethal congenital contracture syndrome 7, 616286
CNTNAP2	121.9	100%	99%	Cortical dysplasia-focal epilepsy syndrome, 610042 Pitt-Hopkins like syndrome 1, 610042 {Autism susceptibility 15}, 612100

COA6	57.9	97%	87%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 4, 616501
COASY	133.9	100%	100%	Neurodegeneration with brain iron accumulation 6, 615643
COCH	191.2	99%	99%	Deafness, autosomal dominant 9, 601369
COG1	109.9	100%	99%	Congenital disorder of glycosylation, type IIg, 611209
COG4	113.3	100%	99%	Congenital disorder of glycosylation, type IIj, 613489
COG5	97.3	93%	92%	Congenital disorder of glycosylation, type IIi, 613612
COG6	76.5	92%	79%	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328
COG7	115.5	100%	100%	Congenital disorder of glycosylation, type IIe, 608779
COG8	103.5	99%	88%	Congenital disorder of glycosylation, type IIh, 611182
COL10A1	81.1	100%	91%	Metaphyseal chondrodysplasia, Schmid type, 156500
COL11A1	79.7	92%	89%	Fibrochondrogenesis 1, 228520 Marshall syndrome, 154780 Stickler syndrome, type II, 604841 {Lumbar disc herniation, susceptibility to}, 603932
COL11A2	10.1	41%	11%	Deafness, autosomal dominant 13, 601868 Deafness, autosomal recessive 53, 609706 Fibrochondrogenesis 2, 614524 Otospondylomegaepiphyseal dysplasia, 215150 Stickler syndrome, type III, 184840 Weissenbacher-Zweymuller syndrome, 277610
COL12A1	122.2	99%	97%	Bethlem myopathy 2, 616471 ?Ullrich congenital muscular dystrophy 2, 616470
COL13A1	75.1	100%	96%	Myasthenic syndrome, congenital, 19, 616720
COL17A1	97.4	97%	93%	Epidermolysis bullosa, junctional, localisata variant, 226650 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epithelial recurrent erosion dystrophy, 122400
COL18A1	75.9	93%	85%	Knobloch syndrome, type 1, 267750
COL1A1	118.0	97%	94%	Caffey disease, 114000 Ehlers-Danlos syndrome, classic, 130000 Ehlers-Danlos syndrome, type VIIA, 130060 Osteogenesis imperfecta, type I, 166200 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type III, 259420

				Osteogenesis imperfecta, type IV, 166220 {Bone mineral density variation QTL, osteoporosis}, 166710
COL1A2	90.7	95%	91%	Ehlers-Danlos syndrome, cardiac valvular form, 225320 Ehlers-Danlos syndrome, type VIIB, 130060 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type III, 259420 Osteogenesis imperfecta, type IV, 166220 {Osteoporosis, postmenopausal}, 166710
COL25A1	109.3	99%	92%	Fibrosis of extraocular muscles, congenital, 5, 616219
COL2A1	92.2	100%	98%	Achondrogenesis, type II or hypochondrogenesis, 200610 Avascular necrosis of the femoral head, 608805 Czech dysplasia, 609162 Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 Kniest dysplasia, 156550 Legg-Calve-Perthes disease, 150600 Osteoarthritis with mild chondrodysplasia, 604864 Otospondylomegaepiphyseal dysplasia, 215150 Platyspondylic skeletal dysplasia, Torrance type, 151210 SED congenita, 183900 SMED Strudwick type, 184250 Spondyloepiphyseal dysplasia, Stanescu type, 616583 Spondyloperipheral dysplasia, 271700 Stickler sydrome, type I, nonsyndromic ocular, 609508 Stickler syndrome, type I, 108300 Vitreoretinopathy with phalangeal epiphyseal dysplasia
COL3A1	94.9	96%	86%	Ehlers-Danlos syndrome, type IV, 130050
COL4A1	79.5	97%	88%	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Brain small vessel disease with or without ocular anomalies, 607595 Porencephaly 1, 175780 ?Retinal arteries, tortuosity of, 180000

				{Hemorrhage, intracerebral, susceptibility to}, 614519
COL4A2	85.6	98%	93%	Porencephaly 2, 614483 {Hemorrhage, intracerebral, susceptibility to}, 614519
COL4A3	77.4	97%	95%	Alport syndrome, autosomal dominant, 104200 Alport syndrome, autosomal recessive, 203780 Hematuria, benign familial, 141200
COL4A3BP	111.2	98%	92%	Mental retardation, autosomal dominant 34, 616351
COL4A4	72.4	97%	91%	Alport syndrome, autosomal recessive, 203780 Hematuria, familial benign
COL4A5	61.5	95%	81%	Alport syndrome, 301050
COL5A1	105.4	99%	95%	Ehlers-Danlos syndrome, classic type, 130000
COL5A2	74.2	99%	95%	Ehlers-Danlos syndrome, classic type, 130000
COL6A1	128.3	99%	97%	Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090
COL6A2	132.4	100%	98%	Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090 ?Myosclerosis, congenital, 255600
COL6A3	145.4	100%	99%	Bethlem myopathy 1, 158810 Dystonia 27, 616411 Ullrich congenital muscular dystrophy 1, 254090
COL7A1	114.8	99%	97%	EBD inversa, 226600 EBD, Bart type, 132000 EBD, localisata variant Epidermolysis bullosa dystrophica, AD, 131750 Epidermolysis bullosa dystrophica, AR, 226600 Epidermolysis bullosa pruriginosa, 604129 Epidermolysis bullosa, pretibial, 131850 Toenail dystrophy, isolated, 607523 Transient bullous of the newborn, 131705
COL8A2	28.7	87%	70%	Corneal dystrophy, Fuchs endothelial, 1, 136800 Corneal dystrophy, posterior polymorphous 2, 609140

COL9A1	100.3	99%	95%	Stickler syndrome, type IV, 614134 /?Epiphyseal dysplasia, multiple, 6, 614135
COL9A2	52.7	96%	88%	Epiphyseal dysplasia, multiple, 2, 600204 ?Stickler syndrome, type V, 614284 {Intervertebral disc disease, susceptibility to}, 603932
COL9A3	62.2	96%	87%	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 {Intervertebral disc disease, susceptibility to}, 603932
COLEC11	168.0	100%	100%	3MC syndrome 2, 265050
COLQ	96.9	100%	94%	Myasthenic syndrome, congenital, 5, 603034
COMP	114.5	93%	92%	Epiphyseal dysplasia, multiple, 1, 132400 Pseudoachondroplasia, 177170
COQ2	68.4	96%	93%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ4	76.0	86%	83%	Coenzyme Q10 deficiency, primary, 7, 616276
COQ6	114.7	99%	96%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ9	81.4	100%	94%	Coenzyme Q10 deficiency, primary, 5, 614654
CORIN	150.7	99%	98%	Preeclampsia/eclampsia 5, 614595
CORO1A	144.5	97%	92%	Immunodeficiency 8, 615401
COX10	194.9	100%	100%	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 Mitochondrial complex IV deficiency, 220110
COX15	86.6	100%	98%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000
COX20	42.6	89%	61%	Mitochondrial complex IV deficiency, 220110
COX4I2	91.9	100%	100%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX6A1	154.4	100%	100%	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
COX6B1	126.5	100%	100%	Mitochondrial complex IV deficiency, 220110
COX7B	52.3	77%	42%	Linear skin defects with multiple congenital anomalies, 300887
CP	115.4	96%	88%	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 [Hypoceruloplasminemia, hereditary], 604290
CPA6	113.8	100%	100%	Epilepsy, familial temporal lobe, 5, 614417 Febrile seizures, familial, 11, 614418
CPN1	109.3	99%	97%	Carboxypeptidase N deficiency, 212070
CPOX	95.3	96%	81%	Coproporphyria, 121300

				Harderoporphyria, 121300
CPS1	133.0	100%	99%	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}
CPT1A	151.0	100%	97%	CPT deficiency, hepatic, type IA, 255120
CPT2	136.9	98%	96%	CPT deficiency, hepatic, type II, 600649 CPT II deficiency, lethal neonatal, 608836 Myopathy due to CPT II deficiency, 255110 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212
CR2	145.2	100%	99%	Immunodeficiency, common variable, 7, 614699 {Systemic lupus erythematosus, susceptibility to, 9}, 610927
CRADD	97.9	98%	95%	Mental retardation, autosomal recessive 34, 614499
CRB1	189.4	100%	100%	Leber congenital amaurosis 8, 613835 Pigmented paravenous chorioretinal atrophy, 172870 Retinitis pigmentosa-12, autosomal recessive, 600105
CRB2	92.0	99%	93%	Focal segmental glomerulosclerosis 9, 616220 Ventriculomegaly with cystic kidney disease, 219730
CRBN	126.2	100%	93%	Mental retardation, autosomal recessive 2, 607417
CREB1	116.1	99%	86%	Histiocytoma, angiomatoid fibrous, somatic, 612160
CREBBP	115.7	99%	95%	Rubinstein-Taybi syndrome, 180849
CRELD1	93.5	100%	100%	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217 {Atrioventricular septal defect, susceptibility to, 2}, 606217
CRIPT	34.9	93%	67%	Short stature with microcephaly and distinctive facies, 615789
CRLF1	94.7	90%	88%	Cold-induced sweating syndrome 1, 272430
CRTAP	88.1	100%	92%	Osteogenesis imperfecta, type VII, 610682
CRTC1	114.8	97%	89%	Mucoepidermoid salivary gland carcinoma
CRX	93.6	99%	97%	Cone-rod retinal dystrophy-2, 120970 Leber congenital amaurosis 7, 613829
CRYAA	101.3	92%	83%	Cataract 9, multiple types, 604219
CRYAB	102.6	98%	93%	Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, 2, 608810 Myopathy, myofibrillar, fatal infantile hypertrophy, alpha-B crystallin-related, 613869
CRYBA1	120.6	100%	99%	Cataract 10, multiple types, 600881

CRYBA4	97.3	100%	100%	Cataract 23, 610425
CRYBB1	100.8	100%	98%	Cataract 17, multiple types, 611544
CRYBB2	146.5	100%	100%	Cataract 3, multiple types, 601547
CRYBB3	113.2	100%	100%	Cataract 22, autosomal recessive, 609741
CRYGB	86.9	100%	98%	Cataract 39, multiple types, autosomal dominant, 615188
CRYGC	109.4	100%	100%	Cataract 2, multiple types, 604307
CRYGD	77.7	100%	100%	Cataract 4, multiple types, 115700
CRYGS	112.0	94%	83%	Cataract 20, multiple types, 116100
CRYM	79.0	100%	98%	Deafness, autosomal dominant 40, 616357
CSF1R	108.8	99%	97%	Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
CSF2RA	52.0	89%	85%	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF2RB	78.2	100%	99%	Surfactant metabolism dysfunction, pulmonary, 5, 614370
CSNK1D	149.8	94%	92%	Advanced sleep-phase syndrome, familial, 2, 615224
CSNK2A1	112.5	96%	84%	Okur-Chung neurodevelopmental syndrome, 617062 Glaucoma, primary congenital (Lee (2011) Mol Vis 17,3583)
CSPP1	97.8	99%	93%	Joubert syndrome 21, 615636
CSRP3	100.0	100%	99%	Cardiomyopathy, hypertrophic, 12, 612124 ?Cardiomyopathy, dilated, 1M, 607482
CST3	98.8	90%	82%	Cerebral amyloid angiopathy, 105150 {Macular degeneration, age-related, 11}, 611953
CSTA	92.5	100%	99%	Peeling skin syndrome 4, 607936
CSTB	90.3	100%	100%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTC1	91.6	100%	99%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTCF	139.6	97%	96%	Mental retardation, autosomal dominant 21, 615502
CTDP1	92.7	87%	83%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTH	156.9	100%	99%	Cystathioninuria, 219500 Homocysteine, total plasma, elevated
CTHRC1	88.4	98%	81%	Barrett esophagus/esophageal adenocarcinoma, 614266
CTLA4	176.4	100%	100%	Autoimmune lymphoproliferative syndrome, type V, 616100 {Celiac disease, susceptibility to, 3}, 609755 {Diabetes mellitus, insulin-dependent, 12}, 601388 {Hashimoto thyroiditis}, 140300 {Systemic lupus erythematosus, susceptibility to}, 152700

CTNNA1	120.4	99%	96%	Macular dystrophy,patterned,608970 Gastric cancer, diffuse (Majewski (2012) J Pathol epub)
CTNNA3	136.8	100%	99%	Arrhythmogenic right ventricular dysplasia, familial, 13, 615616
CTNNB1	142.1	100%	100%	Colorectal cancer, somatic, 114500 Hepatocellular carcinoma, somatic, 114550 Mental retardation, autosomal dominant 19, 615075 Ovarian cancer, somatic, 167000 Pilomatricoma, somatic, 132600
CTNS	113.8	100%	100%	Cystinosis, atypical nephropathic, 219800 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750
CTPS1	121.0	100%	100%	Immunodeficiency 24, 615897
CTSA	113.1	100%	100%	Galactosialidosis, 256540
CTSC	119.9	100%	100%	Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000 Periodontitis 1, juvenile, 170650
CTSD	152.1	100%	97%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	92.5	91%	81%	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362
CTSK	98.4	100%	100%	Pycnodysostosis, 265800
CUBN	113.0	99%	96%	Megaloblastic anemia-1, Finnish type, 261100
CUL3	102.1	95%	90%	Pseudohypoaldosteronism, type IIE, 614496
CUL4B	89.5	98%	96%	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
CUL7	122.3	100%	98%	3-M syndrome 1, 273750
CXCR4	149.4	100%	97%	Myelokathexis, isolated WHIM syndrome, 193670
CYB5A	110.2	100%	100%	Methemoglobinemia, type IV, 250790
CYB5R3	157.7	98%	98%	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYBA	83.7	81%	72%	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690
CYBB	141.6	100%	100%	Chronic granulomatous disease, X-linked, 306400 Immunodeficiency 34, mycobacteriosis, X-linked, 300645
CYC1	159.5	87%	86%	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYCS	66.6	99%	99%	Thrombocytopenia 4, 612004

CYLD	108.2	95%	93%	Brooke-Spiegler syndrome, 605041 Cylindromatosis, familial, 132700 Trichoepithelioma, multiple familial, 1, 601606
CYP11A1	101.7	99%	96%	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	140.1	100%	100%	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 Aldosteronism, glucocorticoid-remediable, 103900
CYP11B2	150.4	100%	100%	Aldosterone to renin ratio raised Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 {Low renin hypertension, susceptibility to}
CYP17A1	109.4	100%	99%	17,20-lyase deficiency, isolated, 202110 17-alpha-hydroxylase/17,20-lyase deficiency, 202110
CYP19A1	163.6	100%	100%	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300
CYP1B1	98.6	100%	94%	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 Peters anomaly, 604229
CYP21A2	11.0	48%	9%	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910
CYP24A1	148.9	100%	99%	Hypercalcemia, infantile, 143880
CYP26B1	146.7	100%	99%	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
CYP26C1	65.5	100%	97%	Focal facial dermal dysplasia 4, 614974
CYP27A1	158.7	94%	93%	Cerebrotendinous xanthomatosis, 213700
CYP27B1	98.6	99%	94%	Vitamin D-dependent rickets, type I, 264700
CYP2A6	119.3	100%	96%	Coumarin resistance, 122700 {Lung cancer, resistance to}, 211980 {Nicotine addiction, protection from}, 188890
CYP2C8	103.4	97%	95%	Rhabdomyolysis, cerivastatin-induced, 601129
CYP2R1	119.6	96%	88%	Rickets due to defect in vitamin D 25-hydroxylation, 600081
CYP2U1	115.4	95%	92%	Spastic paraplegia 56, autosomal recessive, 615030
CYP4F22	103.4	100%	97%	Ichthyosis, congenital, autosomal recessive 5, 604777
CYP4V2	152.0	100%	98%	Bietti crystalline corneoretinal dystrophy, 210370
CYP7B1	88.0	96%	88%	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, autosomal recessive, 270800
D2HGDH	112.5	95%	92%	D-2-hydroxyglutaric aciduria, 600721

DAG1	164.9	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818
DARS	93.3	99%	93%	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2	112.2	100%	98%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBH	124.5	100%	99%	Dopamine beta-hydroxylase deficiency, 223360 [Dopamine-beta-hydroxylase activity levels, plasma]
DBT	98.9	94%	94%	Maple syrup urine disease, type II, 248600
DCAF17	84.2	100%	91%	Woodhouse-Sakati syndrome, 241080
DCC	130.5	100%	100%	Colorectal cancer, somatic, 114500 Esophageal carcinoma, somatic 133239 Mirror movements 1, 157600
DCDC2	125.6	100%	98%	Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212
DCHS1	126.6	98%	97%	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390
DCLRE1C	117.3	96%	90%	Omenn syndrome, 603554 Severe combined immunodeficiency, Athabascan type, 602450
DCN	133.0	95%	94%	Corneal dystrophy, congenital stromal, 610048
DCPS	118.2	100%	99%	Al-Raqad syndrome, 616459
DCTN1	110.2	100%	99%	Neuropathy, distal hereditary motor, type VIIB, 607641 Perry syndrome, 168605 {Amyotrophic lateral sclerosis, susceptibility to}, 105400
DCX	139.7	100%	100%	Lissencephaly, X-linked, 300067 Subcortical laminal heteropia, X-linked, 300067
DDB2	127.9	100%	99%	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DDC	97.6	100%	97%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	132.8	97%	93%	Spastic paraplegia 28, autosomal recessive, 609340
DDHD2	148.2	100%	99%	Spastic paraplegia 54, autosomal recessive, 615033
DDR2	133.7	100%	98%	Spondylometaepiphyseal dysplasia, short limb-hand type, 271665
DDX11	76.1	76%	73%	Warsaw breakage syndrome, 613398
DDX3X	114.8	99%	97%	Mental retardation, X-linked 102, 300958
DDX58	124.3	99%	98%	Singleton-Merten syndrome 2, 616298
DDX59	143.8	99%	98%	Orofaciodigital syndrome V, 174300
DEAF1	126.7	87%	83%	Mental retardation, autosomal dominant 24, 615828

DEPDC5	127.2	99%	99%	Epilepsy, familial focal, with variable foci, 604364
DES	105.1	100%	96%	Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 ?Muscular dystrophy, limb-girdle, type 2R, 615325
DFNA5	96.2	100%	97%	Deafness, autosomal dominant 5, 600994
DFNB59	127.4	100%	100%	Deafness, autosomal recessive 59, 610220
DGKE	126.1	96%	93%	Nephrotic syndrome, type 7, 615008 {Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008
DGUOK	117.3	100%	97%	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHCR24	154.9	100%	100%	Desmosterolosis, 602398
DHCR7	147.7	100%	100%	Smith-Lemli-Opitz syndrome, 270400
DHDDS	89.1	96%	92%	Retinitis pigmentosa 59, 613861
DHFR	44.3	79%	69%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHH	92.5	100%	100%	46XY partial gonadal dysgenesis, with minifascicular neuropathy, 607080 46XY sex reversal 7, 233420
DHODH	91.4	100%	100%	Miller syndrome, 263750
DHTKD1	130.2	99%	98%	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DIABLO	200.7	100%	100%	Deafness, autosomal dominant 64, 614152
DIAPH1	108.4	99%	96%	Deafness, autosomal dominant 1, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632
DIAPH2	71.6	94%	89%	Premature ovarian failure, 300511
DIAPH3	74.5	96%	86%	Auditory neuropathy, autosomal dominant, 1, 609129
DICER1	138.0	99%	96%	Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800 Pleuropulmonary blastoma, 601200 Rhabdomyosarcoma, embryonal, 2, 180295
DIP2B	146.6	99%	97%	Mental retardation, FRA12A type, 136630
DIS3L2	146.8	100%	97%	Perlman syndrome, 267000
DKC1	143.3	100%	100%	Dyskeratosis congenita, X-linked, 305000
DLAT	78.7	100%	91%	Pyruvate dehydrogenase E2 deficiency, 245348
DLC1	167.4	100%	98%	Colorectal cancer, somatic, 114500
DLD	110.9	98%	97%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLG3	114.2	100%	98%	Mental retardation, X-linked 90, 300850

DLL3	58.5	88%	76%	Spondylocostal dysostosis 1, autosomal recessive, 277300
DLL4	170.9	99%	97%	Adams-Oliver syndrome 6, 616589
DLX3	102.9	100%	99%	Amelogenesis imperfecta, type IV, 104510 Trichodontoosseous syndrome, 190320
DMD	152.9	99%	99%	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200
DMGDH	138.4	98%	95%	Dimethylglycine dehydrogenase deficiency, 605850
DMP1	138.4	97%	96%	Hypophosphatemic rickets, AR, 241520
DMPK	98.0	97%	89%	Myotonic dystrophy 1, 160900
DNA2	120.1	99%	95%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156 ?Seckel syndrome 8, 615807
DNAAF1	109.9	100%	96%	Ciliary dyskinesia, primary, 13, 613193
DNAAF2	90.7	100%	94%	Ciliary dyskinesia, primary, 10, 612518
DNAAF3	74.5	99%	90%	Ciliary dyskinesia, primary, 2, 606763
DNAH11	121.4	99%	97%	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH5	114.9	99%	98%	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAI1	102.1	97%	95%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	129.9	95%	93%	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJB2	92.7	100%	99%	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881
DNAJB6	53.2	93%	78%	Muscular dystrophy, limb-girdle, type 1E, 603511
DNAJC19	84.5	99%	98%	3-methylglutaconic aciduria, type V, 610198
DNAJC21	112.7	98%	96%	Bone marrow failure syndrome 3, 617052
DNAJC5	166.2	100%	100%	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350
DNAJC6	147.4	99%	98%	Parkinson disease 19, juvenile-onset, 615528
DNAL1	92.5	99%	73%	Ciliary dyskinesia, primary, 16, 614017
DNASE1L3	121.9	100%	100%	Systemic lupus erythematosus 16, 614420
DNM1	138.3	93%	89%	Epileptic encephalopathy, early infantile, 31, 616346
DNM1L	103.1	99%	91%	Encephalopahty, lethal, due to defective mitochondrial peroxisomal fission, 614388
DNM2	110.9	96%	94%	Charcot-Marie-Tooth disease, axonal, type 2M, 606482 Charcot-Marie-Tooth disease, dominant intermediate B, 606482 Lethal congenital contracture syndrome 5, 615368 Myopathy, centronuclear, 160150

DNMT1	107.0	99%	99%	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 Neuropathy, hereditary sensory, type IE, 614116
DNMT3A	95.6	96%	93%	Tatton-Brown-Rahman syndrome, 615879
DNMT3B	109.3	100%	99%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK2	126.7	100%	100%	Immunodeficiency 40, 616433
DOCK6	105.3	99%	96%	Adams-Oliver syndrome 2, 614219
DOCK7	101.1	96%	93%	Epileptic encephalopathy, early infantile, 23, 615859
DOCK8	122.7	100%	99%	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
DOK7	77.5	96%	91%	Myasthenic syndrome, congenital, 10, 254300 ?Fetal akinesia deformation sequence, 208150
DOLK	153.7	100%	99%	Congenital disorder of glycosylation, type Im, 610768
DPAGT1	96.6	100%	100%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPH1	148.2	100%	99%	Developmental delay with short stature,dysmorphic features and sparse hair,616901
DPM1	105.8	90%	86%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	94.2	100%	99%	Congenital disorder of glycosylation, type Iu, 615042
DPM3	147.7	100%	100%	Congenital disorder of glycosylation, type Io, 612937
DPP6	121.7	99%	95%	Mental retardation, autosomal dominant 33, 616311 {Ventricular fibrillation, paroxysmal familial, 2}, 612956
DPY19L2	64.2	74%	68%	Spermatogenic failure 9, 613958
DPYD	145.1	95%	93%	5-fluorouracil toxicity, 274270 Dihydropyrimidine dehydrogenase deficiency, 274270
DPYS	109.1	100%	99%	Dihydropyrimidinuria, 222748
DRAM2	127.2	100%	100%	Cone-rod dystrophy 21, 616502
DRC1	89.0	100%	99%	Ciliary dyskinesia, primary, 21, 615294
DRD4	73.3	77%	66%	Autonomic nervous system dysfunction [Novelty seeking personality], 601696 {Attention deficit-hyperactivity disorder}, 143465
DRD5	72.7	100%	100%	Dystonia, primary cervical, 126453 {Attention deficit-hyperactivity disorder, susceptibility to}, 143465 {Blepharospasm, primary benign}, 606798
DSC2	124.3	99%	95%	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 Arrhythmogenic right ventricular dysplasia 11, 610476

DSG1	152.5	98%	97%	Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508 Keratosis palmoplantaris striata I, AD, 148700
DSG2	115.3	100%	99%	Arrhythmogenic right ventricular dysplasia 10, 610193 Cardiomyopathy, dilated, 1BB, 612877
DSG4	195.1	98%	94%	Hypotrichosis 6, 607903
DSP	122.6	100%	98%	Arrhythmogenic right ventricular dysplasia 8, 607450 Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821 Epidermolysis bullosa, lethal acantholytic, 609638 Keratosis palmoplantaris striata II, 612908 Skin fragility-woolly hair syndrome, 607655
DSPP	134.7	100%	100%	Deafness, autosomal dominant 39, with dentinogenesis, 605594 Dentin dysplasia, type II, 125420 Dentinogenesis imperfecta, Shields type II, 125490 Dentinogenesis imperfecta, Shields type III, 125500
DST	147.6	99%	97%	Epidermolysis bullosa simplex, autosomal recessive 2, 615425 ?Neuropathy, hereditary sensory and autonomic, type VI, 614653
DTNA	146.8	100%	100%	Left ventricular noncompaction 1, with or without congenital heart defects, 604169
DTNBP1	108.5	100%	96%	Hermansky-Pudlak syndrome 7, 614076
DUOX2	116.1	94%	92%	Thryoid dyshormonogenesis 6, 607200
DUOXA2	112.3	100%	100%	Thyroid dyshormonogenesis 5, 274900
DUSP6	133.4	100%	99%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
DVL1	95.1	98%	91%	Robinow syndrome, autosomal dominant 2, 616331
DVL3	129.7	100%	99%	Robinow syndrome, autosomal dominant 3,616894
DYM	101.5	95%	90%	Dyggve-Melchior-Clausen disease, 223800 Smith-McCort dysplasia, 607326
DYNC1H1	157.8	100%	99%	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600
DYNC2H1	83.0	94%	83%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYNC2LI1	89.2	100%	95%	Short-rib thoracic dysplasia 15 with polydactyly, 617088
DYRK1A	136.7	100%	100%	Mental retardation, autosomal dominant 7, 614104
DYRK1B	86.2	94%	93%	Abdominal obesity-metabolic syndrome 3, 615812

DYSF	114.2	100%	99%	Miyoshi muscular dystrophy 1, 254130 Muscular dystrophy, limb-girdle, type 2B, 253601 Myopathy, distal, with anterior tibial onset, 606768
DYX1C1	65.1	99%	75%	Ciliary dyskinesia, primary, 25, 615482 {Dyslexia, susceptibility to, 1}, 127700
EARS2	78.8	98%	96%	Combined oxidative phosphorylation deficiency 12, 614924
EBP	80.2	100%	96%	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
ECE1	140.1	97%	93%	Hirschsprung disease, cardiac defects, and autonomic dysfunction, 613870 {Hypertension, essential, susceptibility to}, 145500
ECEL1	83.6	87%	83%	Arthrogryposis, distal, type 5D, 615065
ECHS1	108.4	100%	100%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ECM1	141.9	100%	99%	Urbach-Wiethe disease, 247100
EDA	103.5	97%	80%	Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100 Tooth agenesis, selective, X-linked 1, 313500
EDAR	110.2	98%	95%	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	87.4	99%	90%	Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940 Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941
EDN1	122.3	100%	100%	Auriculocondylar syndrome 3, 615706 Question mark ears, isolated, 612798 {High density lipoprotein cholesterol level QTL 7}
EDN3	108.6	100%	93%	Central hypoventilation syndrome, congenital, 209880 Waardenburg syndrome, type 4B, 613265 {Hirschsprung disease, susceptibility to, 4}, 613712
EDNRA	166.5	100%	98%	Mandibulofacial dysostosis with alopecia, 616367 {Migraine, resistance to}, 157300
EDNRB	115.6	93%	93%	ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580 {Hirschsprung disease, susceptibility to, 2}, 600155
EEF1A2	168.9	99%	90%	Epileptic encephalopathy, early infantile, 33, 616409 Mental retardation, autosomal dominant 38, 616393
EFEMP1	166.6	100%	97%	Doyne honeycomb degeneration of retina, 126600

EFEMP2	117.6	100%	100%	Cutis laxa, autosomal recessive, type IB, 614437
EFNB1	147.4	100%	100%	Craniofrontonasal dysplasia, 304110
EFTUD2	100.2	100%	99%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EGF	118.4	100%	99%	Hypomagnesemia 4, renal, 611718
EGFR	137.7	100%	98%	Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980 Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980 ?Inflammatory skin and bowel disease, neonatal, 2, 616069 {Nonsmall cell lung cancer, susceptibility to}, 211980
EGLN1	42.5	79%	60%	Erythrocytosis, familial, 3, 609820 [Hemoglobin, high altitude adaptation], 609070
EGR2	111.3	100%	100%	Charcot-Marie-Tooth disease, type 1D, 607678 Dejerine-Sottas disease, 145900 Neuropathy, congenital hypomyelinating, 1, 605253
EHMT1	134.6	99%	95%	Kleefstra syndrome, 610253
EIF2AK3	132.9	95%	92%	Wolcott-Rallison syndrome, 226980
EIF2AK4	137.4	100%	97%	Pulmonary venoocclusive disease 2, 234810
EIF2B1	118.8	100%	100%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B2	119.2	100%	100%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B3	139.8	100%	100%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B4	114.8	99%	96%	Leukoencephaly with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B5	105.7	100%	98%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF4A3	101.1	100%	99%	Robin sequence with cleft mandible and limb abnormalities,268305
ELAC2	105.4	100%	98%	Combined oxidative phosphorylation deficiency 17, 615440 {Prostate cancer, hereditary, 2, susceptibility to}, 614731
ELANE	90.8	99%	96%	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
ELN	91.1	99%	96%	Cutis laxa, AD, 123700 Supravalvar aortic stenosis, 185500
ELOVL4	74.4	98%	97%	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Stargardt disease 3, 600110 ?Spinocerebellar ataxia 34, 133190

ELOVL5	102.4	100%	99%	Spinocerebellar ataxia 38, 615957
EMC1	109.4	100%	99%	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EMD	128.9	100%	99%	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
EMG1	118.3	100%	100%	Bowen-Conradi syndrome, 211180
EMP2	92.3	100%	99%	Nephrotic syndrome, type 10, 615861
EMR2	142.1	97%	96%	Vibratory urticaria, 125630
EMX2	93.9	100%	99%	Schizencephaly, 269160
ENAM	125.2	100%	100%	Amelogenesis imperfecta, type IB, 104500 Amelogenesis imperfecta, type IC, 204650
ENG	111.5	94%	91%	Telangiectasia, hereditary hemorrhagic, type 1, 187300
ENPP1	129.9	86%	76%	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	139.2	100%	99%	Spastic paraplegia 64, autosomal recessive, 615683
EOGT	130.9	100%	96%	Adams-Oliver syndrome 4, 615297
EP300	161.4	99%	98%	Colorectal cancer, somatic, 114500 Rubinstein-Taybi syndrome 2, 613684
EPAS1	114.8	96%	94%	Erythrocytosis, familial, 4, 611783
EPB41	112.5	99%	96%	Elliptocytosis-1, 611804
EPB42	133.8	100%	99%	Spherocytosis, type 5, 612690
EPCAM	52.9	94%	75%	Colorectal cancer, hereditary nonpolyposis, type 8, 613244 Diarrhea 5, with tufting enteropathy, congenital, 613217
EPG5	112.9	99%	96%	Vici syndrome, 242840
EPHA2	151.5	98%	97%	Cataract 6, multiple types, 116600
EPHX1	108.9	98%	96%	Diphenylhydantoin toxicity Hypercholanemia, familial, 607748 ?Fetal hydantoin syndrome {Preeclampsia, susceptibility to}, 189800
EPM2A	98.4	83%	80%	Epilepsy, progressive myoclonic 2A (Lafora), 254780
ERBB2	115.1	98%	97%	Adenocarcinoma of lung, somatic, 211980 Gastric cancer, somatic, 613659 Glioblastoma, somatic, 137800

				Ovarian cancer, somatic,
ERBB3	114.2	99%	98%	Lethal congenital contractural syndrome 2, 607598
ERBB4	131.0	99%	98%	Amyotrophic lateral sclerosis 19, 615515
ERCC1	74.2	100%	95%	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	116.2	100%	100%	Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy 1, photosensitive, 601675 Xeroderma pigmentosum, group D, 278730
ERCC3	95.9	100%	97%	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651
ERCC4	131.5	99%	97%	Fanconi anemia, complementation group Q, 615272 Xeroderma pigmentosum, group F, 278760 Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 ?XFE progeroid syndrome, 610965
ERCC5	124.3	100%	99%	Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	147.5	100%	100%	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	118.1	99%	97%	Bone marrow failure syndrome 2, 615715
ERCC8	78.9	98%	82%	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621
ERF	90.7	100%	95%	Craniosynostosis 4, 600775
ERLIN1	151.1	100%	100%	Spastic Paraplegia 62, 615681
ERLIN2	132.6	100%	96%	Spastic paraplegia 18, autosomal recessive, 611225
ESCO2	102.3	91%	85%	Roberts syndrome, 268300 SC phocomelia syndrome, 269000

ESPN	36.4	65%	54%	Deafness, autosomal recessive 36, 609006 Deafness, neurosensory, without vestibular involvement, autosomal dominant
ESR1	101.2	98%	97%	Estrogen resistance, 615363 {Atherosclerosis, susceptibility to} {Breast cancer}, 114480 {HDL response to hormone replacement, augmented} {Migraine, susceptibility to}, 157300 {Myocardial infarction, susceptibility to}, 608446
ESRRB	98.4	100%	99%	Deafness, autosomal recessive 35, 608565
ETFA	130.5	100%	100%	Glutaric acidemia IIA, 231680
ETFB	97.9	100%	100%	Glutaric acidemia IIB, 231680
ETFDH	93.8	99%	98%	Glutaric acidemia IIC, 231680
ETHE1	65.8	100%	87%	Ethylmalonic encephalopathy, 602473
ETV6	105.8	100%	100%	Leukemia, acute myeloid, somatic, 601626 Thrombocytopenia 5, 616216
EVC	95.8	90%	85%	Ellis-van Creveld syndrome, 225500 Weyers acrodental dysostosis, 193530
EVC2	95.8	96%	90%	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530
EWSR1	63.7	93%	86%	Ewing sarcoma, 612219 Neuroepithelioma, 612219
EXOSC3	68.1	89%	83%	Pontocerebellar hypoplasia, type 1B, 614678
EXOSC8	69.1	84%	68%	Pontocerebellar hypoplasia, type 1C, 616081
EXPH5	178.8	100%	100%	Epidermolysis bullosa, nonspecific, autosomal recessive, 615028
EXT1	77.9	97%	94%	Chondrosarcoma, 215300 Exostoses, multiple, type 1, 133700
EXT2	162.0	100%	98%	Exostoses, multiple, type 2, 133701 ?Seizures, scoliosis, and macrocephaly syndrome, 616682
EYA1	132.8	100%	99%	Anterior segment anomalies with or without cataract, 113650 Branchiootic syndrome 1, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650 ?Otofaciocervical syndrome, 166780
EYA4	135.5	100%	100%	Cardiomyopathy, dilated, 1J, 605362 Deafness, autosomal dominant 10, 601316

EYS	127.3	98%	93%	Retinitis pigmentosa 25, 602772
EZH2	122.6	99%	97%	Weaver syndrome, 277590
F10	128.4	98%	95%	Factor X deficiency, 227600
F11	136.8	100%	99%	Factor XI deficiency, autosomal dominant, 612416 Factor XI deficiency, autosomal recessive, 612416
F12	98.4	100%	99%	Angioedema, hereditary, type III, 610618 Factor XII deficiency, 234000
F13A1	148.1	100%	98%	Factor XIIIA deficiency, 613225 {Myocardial infarction, protection against}, 608446 {Venous thrombosis, protection against}, 188050
F13B	109.2	94%	84%	Factor XIIIB deficiency, 613235
F2	101.1	100%	97%	Dysprothrombinemia, 613679 Hypoprothrombinemia, 613679 Thrombophilia due to thrombin defect, 188050 {Pregnancy loss, recurrent, susceptibility to, 2}, 614390 {Stroke, ischemic, susceptibility to}, 601367
F5	162.1	98%	97%	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	135.4	100%	100%	Factor VII deficiency, 227500 {Myocardial infarction, decreased susceptibility to}, 608446
F8	151.0	99%	99%	Hemophilia A, 306700
F9	170.7	100%	95%	Hemophilia B, 306900 Thrombophilia, X-linked, due to factor IX defect, 300807 {Deep venous thrombosis, protection against}, 300807 {Warfarin sensitivity}, 122700
FA2H	81.6	94%	79%	Spastic paraplegia 35, autosomal recessive, 612319
FADD	112.5	100%	98%	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovasuclar malformations, 613759
FAH	133.7	100%	99%	Tyrosinemia, type I, 276700
FAM105B	125.2	91%	87%	Autoinflammation,panniculitis, and dermatosis syndrome, 617099

FAM111A	249.9	100%	100%	Gracile bone dysplasia, 602361 Kenny-Caffey syndrome, type 2, 127000
FAM111B	134.3	100%	99%	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704
FAM126A	125.0	95%	95%	Leukodystrophy, hypomyelinating, 5, 610532
FAM134B	98.5	95%	87%	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
FAM161A	97.9	98%	92%	Retinitis pigmentosa 28, 606068
FAM20A	96.9	99%	89%	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM20C	82.4	100%	93%	Raine syndrome, 259775
FAM58A	88.7	80%	78%	STAR syndrome, 300707
FAM83H	70.9	96%	87%	Amelogenesis imperfecta, type III, 130900
FAN1	131.5	100%	100%	Interstitial nephritis, karyomegalic, 614817
FANCA	103.4	100%	98%	Fanconi anemia, complementation group A, 227650
FANCB	87.2	99%	93%	Fanconi anemia, complementation group B, 300514
FANCC	102.9	100%	100%	Fanconi anemia, complementation group C, 227645
FANCD2	116.8	97%	94%	Fanconi anemia, complementation group D2, 227646
FANCE	99.6	84%	84%	Fanconi anemia, complementation group E, 600901
FANCF	113.0	100%	100%	Fanconi anemia, complementation group F, 603467
FANCG	117.3	100%	99%	Fanconi anemia, complementation group G, 614082
FANCI	154.7	99%	96%	Fanconi anemia, complementation group I, 609053
FANCL	67.2	100%	96%	Fanconi anemia, complementation group L, 614083
FAR1	78.2	94%	90%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154
FARS2	183.5	100%	100%	Combined oxidative phosphorylation deficiency 14, 614946 ?Spastic paraplegia 77, autosomal recessive, 617046
FAS	236.0	100%	98%	Autoimmune lymphoproliferative syndrome, type IA, 601859 Squamous cell carcinoma, burn scar-related, somatic {Autoimmune lymphoproliferative syndrome}, 601859
FASLG	78.1	100%	96%	Autoimmune lymphoproliferative syndrome, type IB, 601859 {Lung cancer, susceptibility to}, 211980
FAT4	186.7	100%	99%	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 Van Maldergem syndrome 2, 615546
FBLN5	102.3	91%	91%	Cutis laxa, autosomal dominant 2, 614434 Cutis laxa, autosomal recessive, type IA, 219100 Macular degeneration, age-related, 3, 608895 Neuropathy, hereditary, with or without age-related macular degeneration, 608895

FBN1	141.2	100%	98%	Acromicric dysplasia, 102370 Aortic aneurysm, ascending, and dissection Ectopia lentis, familial, 129600 Geleophysic dysplasia 2, 614185 Marfan lipodystrophy syndrome,616914 Marfan syndrome, 154700 MASS syndrome, 604308 Stiff skin syndrome, 184900 Weill-Marchesani syndrome 2, dominant, 608328
FBN2	145.5	100%	99%	Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118
FBP1	100.7	100%	100%	Fructose-1,6-bisphosphatase deficiency, 229700
FBXL4	180.4	100%	100%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FBXO38	142.4	99%	98%	Neuronopathy, distal hereditary motor, type IID, 615575
FBXO7	192.9	100%	98%	Parkinson disease 15, autosomal recessive, 260300
FCGR3A	156.8	100%	98%	Immunodeficiency 20, 615707
FCGR3B	163.0	99%	97%	Neutropenia, alloimmune neonatal
FCN3	113.1	100%	98%	Immunodeficiency due to ficolin 3 deficiency, 613860
FDPS	60.9	96%	94%	Porokeratosis 9, multiple types, 616631
FECH	114.9	100%	100%	Protoporphyria, erythropoietic, autosomal recessive, 177000
FERMT1	87.6	97%	94%	Kindler syndrome,173650
FERMT3	107.2	100%	100%	Leukocyte adhesion deficiency,type III,612840
FEZF1	125.1	100%	95%	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FGA	142.6	98%	94%	Afibrinogenemia, congenital, 202400 Amyloidosis, familial visceral, 105200 Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, congenital, 616004
FGB	168.0	100%	98%	Afibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Hypofibrinogenemia, congenital, 202400
FGD1	102.8	97%	92%	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400
FGD4	92.4	97%	92%	Charcot-Marie-Tooth disease, type 4H, 609311
FGF10	122.6	100%	100%	Aplasia of lacrimal and salivary glands, 180920

				LADD syndrome, 149730
FGF14	168.9	100%	98%	Spinocerebellar ataxia 27, 609307
FGF16	144.6	100%	99%	Metacarpal 4-5 fusion, 309630
FGF17	129.7	100%	100%	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
FGF23	95.3	98%	95%	Hypophosphatemic rickets, autosomal dominant, 193100 Osteomalacia, tumor-induced Tumoral calcinosis, hyperphosphatemic, familial, 211900
FGF3	68.5	97%	92%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGF5	87.2	100%	99%	Trichomegaly, 190330
FGF8	121.5	82%	75%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGFR1	127.6	97%	96%	Encephalocraniocutaneous lipomatosis,613001 Hartsfield syndrome, 615465 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Jackson-Weiss syndrome, 123150 Osteoglophonic dysplasia, 166250 Pfeiffer syndrome, 101600 Trigonocephaly 1, 190440
FGFR2	121.8	96%	95%	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Apert syndrome, 101200 Beare-Stevenson cutis gyrata syndrome, 123790 Bent bone dysplasia syndrome, 614592 Craniofacial-skeletal-dermatologic dysplasia, 101600 Craniosynostosis, nonspecific Crouzon syndrome, 123500 Gastric cancer, somatic, 613659 Jackson-Weiss syndrome, 123150 LADD syndrome, 149730 Pfeiffer syndrome, 101600 Saethre-Chotzen syndrome, 101400 Scaphocephaly and Axenfeld-Rieger anomaly Scaphocephaly, maxillary retrusion, and mental retardation, 609579

FGFR3	98.9	100%	99%	Achondroplasia, 100800 Bladder cancer, somatic, 109800 CATSHL syndrome, 610474 Cervical cancer, somatic, 603956 Colorectal cancer, somatic, 114500 Crouzon syndrome with acanthosis nigricans, 612247 Hypochondroplasia, 146000 LADD syndrome, 149730 Muenke syndrome, 602849 Nevus, epidermal, somatic, 162900 SADDAN, 616482 Spermatocytic seminoma, somatic, 273300 Thanatophoric dysplasia, type I, 187600 Thanatophoric dysplasia, type II, 187601
FGG	131.4	98%	97%	Afibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, 616004 Hypofibrinogenemia, congenital, 202400
FH	139.2	89%	86%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FHL1	98.8	98%	93%	Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 Hemophagocytic lymphohistiocytosis, familial, 1 Myopathy, X-linked, with postural muscle atrophy, 300696 Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717 Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718 Scapuloperoneal myopathy, X-linked dominant, 300695
FIG4	147.4	100%	96%	Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228 Yunis-Varon syndrome, 216340 ?Polymicrogyria, bilateral temporooccipital, 612691
FIGLA	91.8	90%	85%	Premature ovarian failure 6, 612310
FKBP10	137.9	97%	93%	Bruck syndrome 1, 259450 Osteogenesis imperfecta, type XI, 610968
FKBP14	75.6	99%	93%	Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss, 614557

FKRP	74.3	100%	99%	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	128.4	99%	93%	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	141.7	100%	100%	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100
FLCN	138.8	100%	99%	Birt-Hogg-Dube syndrome, 135150 Colorectal cancer, somatic, 114500 Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700
FLG	170.8	100%	99%	Ichthyosis vulgaris, 146700 {Dermatitis, atopic, susceptibility to, 2}, 605803
FLNA	163.8	100%	100%	Cardiac valvular dysplasia, X-linked, 314400 Congenital short bowel syndrome, 300048 FG syndrome 2, 300321 Frontometaphyseal dysplasia, 305620 Heterotopia, periventricular, 300049 Heterotopia, periventricular, ED variant, 300537 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Terminal osseous dysplasia, 300244
FLNB	129.5	99%	99%	Atelosteogenesis, type I, 108720 Atelosteogenesis, type III, 108721 Boomerang dysplasia, 112310 Larsen syndrome, 150250 Spondylocarpotarsal synostosis syndrome, 272460
FLNC	135.4	100%	99%	Cardiomyopathy, familial hypertrophic, 26 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Myopathy, myofibrillar, 5, 609524

FLRT3	188.9	100%	100%	Hypogonadotropic hypogonadism 21 with anosmia, 615271
FLT3	121.0	98%	97%	Leukemia, acute lymphoblastic, somatic, 613065 Leukemia, acute myeloid, reduced survival in, somatic, 601626 Leukemia, acute myeloid, somatic, 601626
FLT4	143.6	99%	98%	Hemangioma, capillary infantile, somatic, 602089 Lymphedema, hereditary, IA, 153100
FLVCR1	126.5	99%	97%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FLVCR2	152.6	100%	100%	Proliferative vasculopathy and hydraencephaly-hydrocephaly syndrome, 225790
FMN2	76.9	80%	72%	Mental retardation, autosomal recessive 47, 616193
FMO3	138.1	99%	97%	Trimethylaminuria, 602079
FMR1	94.5	96%	87%	Fragile X syndrome, 300624 Fragile X tremor/ataxia syndrome, 300623 Premature ovarian failure 1, 311360
FN1	126.8	100%	98%	Glomerulopathy with fibronectin deposits 2, 601894 Plasma fibronectin deficiency, 614101
FOLR1	107.1	100%	100%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXC1	32.1	97%	83%	Axenfeld-Rieger syndrome, type 3, 602482 Iridogoniodysgenesis, type 1, 601631 Iris hypoplasia and glaucoma, 601631 Rieger or Axenfeld anomalies, 602482
FOXC2	42.2	99%	77%	Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400 Lymphedema-distichiasis syndrome, 153400
FOXE1	24.2	70%	54%	Bamforth-Lazarus syndrome, 241850 {Thyroid cancer, nonmedullary, 4}, 616534
FOXE3	16.1	57%	38%	Anterior segment mesenchymal dysgenesis, 107250 Aphakia, congenital primary, 610256
FOXF1	61.8	96%	87%	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380
FOXG1	93.6	79%	76%	Rett syndrome, congenital variant, 613454
FOXI1	115.2	100%	100%	Enlarged vestibular aqueduct, 600791
FOXL2	25.3	78%	55%	Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 Premature ovarian failure 3, 608996
FOXN1	88.2	99%	96%	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXO1	121.1	94%	91%	Rhabdomyosarcoma,alveolar,268220

FOXP1	116.7	100%	99%	Mental retardation with language impairment and with or without autistic features, 613670
FOXP2	134.9	95%	94%	Speech-language disorder-1, 602081
FOXP3	130.0	100%	90%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790 {Diabetes mellitus, type I, susceptibility to}, 222100
FOXRED1	122.6	100%	99%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
FRAS1	124.7	99%	99%	Fraser syndrome, 219000
FREM1	124.0	99%	97%	Bifid nose with or without anorectal and renal anomalies, 608980 Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485
FREM2	158.3	99%	99%	Fraser syndrome, 219000
FRMD7	151.9	100%	100%	Nystagmus 1, congenital, X-linked, 310700 Nystagmus, infantile periodic alternating, X-linked, 310700
FRRS1L	106.3	62%	62%	Epileptic encephalopathy, early infantile, 37, 616981
FSCN2	130.9	100%	100%	Retinitis pigmentosa 30, 607921
FSHB	118.0	100%	100%	Hypogonadotropic hypogonadism 24 without anosmia, 229070
FSHR	108.0	99%	97%	Ovarian dysgenesis 1, 233300 Ovarian hyperstimulation syndrome, 608115 Ovarian response to FSH stimulation, 276400
FTCD	75.8	92%	85%	Glutamate formiminotransferase deficiency, 229100
FTL	101.3	100%	97%	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159
FTO	116.7	99%	97%	Growth retardation, developmental delay, facial dysmorphism, 612938 {Obesity, susceptibility to, BMIQ14}, 612460
FTSJ1	149.3	100%	98%	Mental retardation, X-linked 9, 309549
FUCA1	121.2	100%	99%	Fucosidosis, 230000
FUS	117.2	100%	93%	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 Tremor, hereditary essential, 4, 614782
FUT6	151.4	100%	100%	Fucosyltransferase 6 deficiency, 613852
FUZ	94.7	100%	100%	Neural tube defects, 182940
FXN	59.8	75%	71%	Friedreich ataxia with retained reflexes, 229300 Friedreich ataxia, 229300
FXYD2	80.6	100%	100%	Hypomagnesemia 2, renal, 154020

FYCO1	106.7	100%	100%	Cataract 18, autosomal recessive, 610019
FZD4	177.8	100%	98%	Exudative vitreoretinopathy 1, 133780 Retinopathy of prematurity, 133780
FZD6	179.4	100%	100%	Nail disorder, nonsyndromic congenital, 10, (claw-shaped nails), 614157
G6PC	166.9	100%	100%	Glycogen storage disease Ia, 232200
G6PC3	115.4	100%	100%	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
G6PD	135.0	100%	99%	Favism, 134700 Hemolytic anemia due to G6PD deficiency, 300908 {Resistance to malaria due to G6PD deficiency}, 611162
GAA	106.3	100%	99%	Glycogen storage disease II, 232300
GABRA1	160.5	100%	100%	Epileptic encephalopathy, early infantile, 19, 615744 {Epilepsy, childhood absence, susceptibility to, 4}, 611136 {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136
GABRG2	134.8	92%	92%	Epilepsy, generalized, with febrile seizures plus, type 3, 611277 Febrile seizures, familial, 8, 611277 {Epilepsy, childhood absence, susceptibility to, 2}, 607681
GALC	94.2	98%	91%	Krabbe disease, 245200
GALE	125.6	100%	100%	Galactose epimerase deficiency, 230350
GALK1	95.7	98%	94%	Galactokinase deficiency with cataracts, 230200
GALNS	84.5	100%	93%	Mucopolysaccharidosis IVA, 253000
GALNT3	118.2	97%	91%	Tumoral calcinosis, hyperphosphatemic, familial, 211900
GALT	137.1	100%	100%	Galactosemia, 230400
GAMT	91.2	98%	88%	Cerebral creatine deficiency syndrome 2, 612736
GAN	177.1	100%	97%	Giant axonal neuropathy-1, 256850
GANAB	103.5	99%	97%	Polycystic kidney disease 3, 600666
GARS	120.6	99%	96%	Charcot-Marie-Tooth disease, type 2D, 601472 Neuropathy, distal hereditary motor, type VA, 600794
GAS8	137.3	100%	99%	Ciliary dyskinesia, primary, 33, 616726
GATA1	92.2	100%	99%	Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835 Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 Thrombocytopenia with beta-thalassemia, X-linked, 314050 Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367

GATA2	106.1	100%	98%	Emberger syndrome, 614038 Immunodeficiency 21, 614172 {Leukemia, acute myeloid, susceptibility to}, 601626 {Myelodysplastic syndrome, susceptibility to}, 614286
GATA3	145.1	100%	99%	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GATA4	65.1	57%	53%	Atrial septal defect 2, 607941 Atrioventricular septal defect 4, 614430 Tetralogy of Fallot, 187500 Ventricular septal defect 1, 614429 ?Testicular anomalies with or without congenital heart disease, 615542
GATA6	47.2	77%	63%	Atrial septal defect 9, 614475 Atrioventricular septal defect 5, 614474 Pancreatic agenesis and congenital heart defects, 600001 Persistent truncus arteriosus, 217095 Tetralogy of Fallot, 187500
GATAD2B	110.1	100%	99%	Mental retardation, autosomal dominant 18, 615074
GATM	140.2	100%	100%	Cerebral creatine deficiency syndrome 3, 612718
GBA	193.2	100%	100%	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	145.8	99%	98%	Spastic paraplegia 46, autosomal recessive, 614409
GBE1	147.0	99%	93%	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GCDH	118.5	93%	91%	Glutaricaciduria, type I, 231670
GCH1	66.9	96%	79%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GCK	126.0	100%	100%	Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, permanent neonatal, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485 MODY, type II, 125851

GCLC	121.1	99%	98%	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 {Myocardial infarction, susceptibility to}, 608446
GCM2	127.0	100%	100%	Hypoparathyroidism, familial isolated, 146200
GCNT2	157.9	100%	100%	Adult i phenotype without cataract, 110800 Cataract 13 with adult i phenotype, 116700 [Blood group, Ii], 110800
GCSH	29.0	60%	37%	Glycine encephalopathy, 605899
GDAP1	153.4	100%	99%	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	27.7	69%	62%	Double-outlet right ventricle, 217095 Right atrial isomerism, 208530 Tetralogy of Fallot, 187500 Transposition of great arteries, dextro-looped 3, 613854
GDF2	128.5	100%	100%	Telangiectasia, hereditary hemorrhagic, type 5, 615506
GDF3	122.6	100%	100%	Klippel-Feil syndrome 3, autosomal dominant, 613702 Microphthalmia with coloboma 6, 613703 Microphthalmia, isolated 7, 613704
GDF5	122.9	100%	100%	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 ?Acromesomelic dysplasia, Hunter-Thompson type, 201250 {Osteoarthritis-5}, 612400
GDF6	70.3	93%	80%	Klippel-Feil syndrome 1, autosomal dominant, 118100 Leber congenital amaurosis 17, 615360 Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094
GDI1	194.0	100%	100%	Mental retardation, X-linked 41, 300849

GDNF	155.8	100%	95%	Central hypoventilation syndrome, 209880 {Hirschsprung disease, susceptibility to, 3}, 613711 {Pheochromocytoma, modifier of}, 171300
GFAP	87.9	100%	100%	Alexander disease, 203450
GFER	75.1	99%	86%	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076
GFI1	81.6	100%	92%	Neutropenia, nonimmune chronic idiopathic, of adults, 607847 Neutropenia, severe congenital 2, autosomal dominant, 613107
GFI1B	132.8	100%	100%	Bleeding disorder, platelet-type, 17, 187900
GFM1	85.8	98%	95%	Combined oxidative phosphorylation deficiency 1, 609060
GFPT1	126.0	99%	96%	Myasthenia, congenital, 12, with tubular aggregates, 610542
GGCX	89.8	99%	96%	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842 Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450
GH1	167.6	100%	100%	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	185.3	100%	100%	Growth hormone insensitivity, partial, 604271 Increased responsiveness to growth hormone Laron dwarfism, 262500 {Hypercholesterolemia, familial, modifier of}, 143890
GHRHR	110.8	100%	100%	Growth hormone deficiency, isolated, type IB, 612781
GHSR	144.6	99%	97%	Growth hormone deficiency, isolated partial, 615925
GIF	135.4	100%	100%	Intrinsic factor deficiency, 261000
GIPC3	100.4	90%	82%	Deafness, autosomal recessive 15, 601869
GJA1	182.8	100%	100%	Atrioventricular septal defect 3, 600309 Craniometaphyseal dysplasia, autosomal recessive, 218400 Erythrokeratodermia variabilis et progressiva, 133200 Hypoplastic left heart syndrome 1, 241550 Oculodentodigital dysplasia, 164200 Oculodentodigital dysplasia, autosomal recessive, 257850 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100
GJA3	120.9	100%	96%	Cataract 14, multiple types, 601885

GJA5	224.4	100%	100%	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic (GJA5/SCN5A), 108770
GJA8	114.0	100%	100%	Cataract 1, multiple types, 116200
GJB1	222.4	100%	100%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GJB2	190.3	100%	100%	Bart-Pumphrey syndrome, 149200 Deafness, autosomal dominant 3A, 601544 Deafness, autosomal recessive 1A, 220290 Hystrix-like ichthyosis with deafness, 602540 Keratitis-ichthyosis-deafness syndrome, 148210 Keratoderma, palmoplantar, with deafness, 148350 Vohwinkel syndrome, 124500
GJB3	283.3	100%	100%	Deafness, autosomal dominant 2B, 612644 Deafness, autosomal dominant, with peripheral neuropathy Deafness, autosomal recessive Deafness, digenic, GJB2/GJB3, 220290 Erythrokeratodermia variabilis et progressiva, 133200
GJB4	330.8	100%	100%	Erythrokeratodermia variabilis with erythema gyratum repens, 133200
GJB6	193.1	100%	100%	Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290 Ectodermal dysplasia 2, Clouston type, 129500
GJC2	38.3	84%	57%	Leukodystrophy, hypomyelinating, 2, 608804 Lymphedema, hereditary, IC, 613480 Spastic paraplegia 44, autosomal recessive, 613206
GK	55.6	83%	67%	Glycerol kinase deficiency, 307030
GLA	89.1	100%	99%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	72.3	99%	95%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLDC	69.0	91%	81%	Glycine encephalopathy, 605899
GLE1	103.5	100%	97%	Arthrogryposis, lethal, with anterior horn cell disease, 611890 Lethal congenital contracture syndrome 1, 253310

GLI2	102.9	98%	96%	Culler-Jones syndrome, 615849 Holoprosencephaly-9, 610829
GLI3	136.7	100%	99%	Greig cephalopolysyndactyly syndrome, 175700 Pallister-Hall syndrome, 146510 Polydactyly, postaxial, types A1 and B, 174200 Polydactyly, preaxial, type IV, 174700 {Hypothalamic hamartomas, somatic}, 241800
GLIS2	92.0	100%	94%	Nephronophthisis 7, 611498
GLIS3	124.3	100%	99%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GLMN	67.1	99%	85%	Glomuvenous malformations,138000
GLRA1	105.1	100%	100%	Hyperekplexia, hereditary 1, autosomal dominant or recessive, 149400
GLRB	88.6	97%	89%	Hyperekplexia 2, autosomal recessive, 614619
GLRX5	79.2	87%	81%	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GLUD1	65.2	89%	80%	Hyperinsulinism-hyperammonemia syndrome, 606762
GLUL	84.2	99%	95%	Glutamine deficiency, congenital, 610015
GLYCTK	186.0	100%	100%	D-glyceric aciduria, 220120
GM2A	116.8	100%	100%	GM2-gangliosidosis, AB variant, 272750
GMNN	93.5	93%	79%	Meier-Gorlin syndrome 6, 616835
GMPPA	125.4	100%	100%	Alacrima, achalasia, and mental retardation syndrome, 615510
GMPPB	207.7	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352
GNA11	136.9	100%	99%	Hypocalcemia, autosomal dominant 2, 615361 Hypocalciuric hypercalcemia, type II, 145981
GNAI2	127.8	100%	100%	Pituitary ACTH-secreting adenoma Ventricular tachycardia, idiopathic, 192605
GNAI3	99.1	100%	95%	Auriculocondylar syndrome 1, 602483
GNAL	118.9	99%	91%	Dystonia 25, 615073
GNAO1	167.2	100%	100%	Epileptic encephalopathy, early infantile, 17, 615473
GNAQ	75.9	89%	72%	Capillary malformations, congenital, 1, somatic, mosaic, 163000 Sturge-Weber syndrome, somatic, mosaic, 185300

GNAS	102.2	97%	95%	Acromegaly, somatic, 102200 ACTH-independent macronodular adrenal hyperplasia, 219080 McCune-Albright syndrome, somatic, mosaic 174800 Osseous heteroplasia, progressive, 166350 Pseudohypoparathyroidism Ia, 103580 Pseudohypoparathyroidism Ib, 603233 Pseudohypoparathyroidism Ic, 612462 Pseudopseudohypoparathyroidism, 612463
GNAS-AS1	NC	NC	NC	Pseudohypoparathyroidism, type IB, 603233
GNAT1	145.8	100%	100%	Night blindness, congenital stationary, autosomal dominant 3, 610444 ?Night blindness, congenital stationary, type 1G, 616389
GNAT2	130.2	100%	100%	Achromatopsia-4, 613856
GNB1	162.4	100%	100%	Leukemia,acute lymphoblastic,somatic, 613065 Mental retardation, autosomal dominant 42, 616973
GNB3	173.2	100%	100%	Night blindness, congenital stationary, type 1H, 617024 {Hypertension, essential, susceptibility to}, 145500
GNB4	133.7	100%	100%	Charcot-Marie-Tooth disease, dominant intermediate F, 615185
GNE	136.1	100%	99%	Nonaka myopathy, 605820 Sialuria, 269921
GNMT	143.4	99%	96%	Glycine N-methyltransferase deficiency, 606664
GNPAT	129.1	99%	95%	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	150.5	97%	97%	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GNPTG	131.6	93%	84%	Mucolipidosis III gamma, 252605
GNRHR	182.5	100%	100%	Hypogonadotropic hypogonadism 7 without anosmia, 146110
GNS	95.0	97%	93%	Mucopolysaccharidosis type IIID, 252940
GORAB	151.7	100%	99%	Geroderma osteodysplasticum, 231070
GOSR2	113.6	97%	96%	Epilepsy, progressive myoclonic 6, 614018
GOT1	110.1	100%	97%	Aspartate aminotransferase, serum level of, QTL1, 614419
GP1BA	135.4	97%	94%	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	30.6	69%	61%	Bernard-Soulier syndrome, type B, 231200

				Giant platelet disorder, isolated, 231200
GP6	110.2	100%	100%	Bleeding disorder, platelet-type, 11, 614201
GP9	60.2	99%	85%	Bernard-Soulier syndrome, type C, 231200
GPC3	119.5	100%	98%	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPC6	129.8	100%	100%	Omodysplasia 1, 258315
GPD1	88.8	99%	92%	Hypertriglyceridemia, transient infantile, 614480
GPD1L	132.0	100%	99%	Brugada syndrome 2, 611777
GPHN	152.5	98%	96%	Molybdenum cofactor deficiency C, 615501
GPI	126.7	100%	100%	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470
GPIHBP1	82.6	100%	99%	Hyperlipoproteinemia, type 1D, 615947
GPR101	152.2	100%	100%	Pituitary adenoma, growth hormone-secreting 2, 300943
GPR126	142.8	99%	98%	Lethal congenital contracture syndrome 9, 616503
GPR143	75.7	91%	87%	Nystagmus 6, congenital, X-linked, 300814 Ocular albinism, type I, Nettleship-Falls type, 300500
GPR179	112.0	100%	99%	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
GPR56	141.3	100%	100%	Polymicrogyria, bilateral frontoparietal, 606854 Polymicrogyria, bilateral perisylvian, 615752
GPR98	129.7	99%	95%	Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472 ?Febrile seizures, familial, 4, 604352
GPSM2	99.3	98%	92%	Chudley-McCullough syndrome, 604213
GPX4	165.8	85%	85%	Spondylometaphyseal dysplasia, Sedaghatian type, 250220
GRHL2	120.8	100%	100%	Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029
GRHL3	131.2	100%	99%	Van der Woude syndrome 2, 606713
GRHPR	96.1	83%	80%	Hyperoxaluria, primary, type II, 260000
GRIA3	115.2	100%	98%	Mental retardation, X-linked 94, 300699
GRID2	158.7	100%	100%	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRIK2	127.9	96%	94%	Mental retardation, autosomal recessive, 6, 611092
GRIN1	136.7	100%	99%	Mental retardation, autosomal dominant 8, 614254
GRIN2A	138.3	100%	99%	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570
GRIN2B	162.5	100%	99%	Epileptic encephalopathy, early infantile, 27, 616139

				Mental retardation, autosomal dominant 6, 613970
GRIP1	123.5	100%	100%	Fraser syndrome, 219000
GRK1	108.4	100%	98%	Oguchi disease-2, 613411
GRM1	163.4	100%	99%	Spinocerebellar ataxia, autosomal recessive 13, 614831
GRM6	130.9	88%	84%	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270
GRN	165.4	100%	100%	Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
GRXCR1	173.3	100%	100%	Deafness, autosomal recessive 25, 613285
GSC	88.5	99%	77%	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471
GSN	104.7	99%	92%	Amyloidosis, Finnish type, 105120
GSS	88.9	100%	99%	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900
GTF2E2	75.1	99%	87%	Trichothiodystrophy 6,nonphotosensitive,616943
GTF2H5	132.3	100%	97%	Trichothiodystrophy 3, photosensitive, 616395
GTPBP3	98.9	100%	99%	Combined oxidative phosphorylation deficiency 23, 616198
GUCA1A	128.2	100%	100%	Cone dystrophy-3, 602093 Cone-rod dystrophy 14, 602093
GUCA1B	132.7	100%	100%	Retinitis pigmentosa 48, 613827
GUCY1A3	151.2	100%	97%	Moyamoya 6 with achalasia, 615750
GUCY2C	127.5	100%	99%	Diarrhea 6, 614616 Meconium ileus, 614665
GUCY2D	80.3	96%	90%	Cone-rod dystrophy 6, 601777 Leber congenital amaurosis 1, 204000
GUSB	96.6	84%	81%	Mucopolysaccharidosis VII, 253220
GYG1	112.9	100%	97%	Polyglucosan body myopathy 2, 616199 ?Glycogen storage disease XV, 613507
GYS1	96.4	100%	98%	Glycogen storage disease 0, muscle, 611556
GYS2	147.2	100%	95%	Glycogen storage disease 0, liver, 240600
H19	NC	NC	NC	Beckwith-Wiedemann syndrome, 130650 Silver-Russell syndrome, 180860 Wilms tumor 2, 194071
H6PD	136.2	99%	98%	Cortisone reductase deficiency 1, 604931

HACE1	123.6	98%	92%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADH	97.1	99%	95%	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HADHA	71.7	95%	90%	Fatty liver, acute, of pregnancy, 609016 HELLP syndrome, maternal, of pregnancy, 609016 LCHAD deficiency, 609016 Trifunctional protein deficiency, 609015
HADHB	83.9	93%	84%	Trifunctional protein deficiency, 609015
HAMP	146.2	100%	100%	Hemochromatosis, type 2B, 613313
HARS	130.7	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504
HAX1	117.0	100%	100%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HBA1	99.9	100%	99%	Erythremias, alpha- Heinz body anemias, alpha-, 140700 Hemoglobin H disease, nondeletional, 613978 Methemoglobinemias, alpha- Thalassemias, alpha-, 604131
HBA2	88.2	93%	85%	Erythrocytosis Heinz body anemia, 140700 Hemoglobin H disease, nondeletional, 613978 Hypochromic microcytic anemia Thalassemia, alpha-, 604131
HBB	151.8	100%	100%	Delta-beta thalassemia, 141749 Erythremias, beta- Heinz body anemias, beta-, 140700 Hereditary persistence of fetal hemoglobin, 141749 Methemoglobinemias, beta- Sickle cell anemia, 603903 Thalassemia-beta, dominant inclusion-body, 603902 Thalassemias, beta-, 613985 {Malaria, resistance to}, 611162
HBD	189.4	100%	100%	Thalassemia due to Hb Lepore Thalassemia, delta-
HBG1	153.0	100%	99%	Fetal hemoglobin quantitative trait locus 1, 141749

HBG2	211.3	100%	100%	Cyanosis, transient neonatal, 613977 Fetal hemoglobin quantitative trait locus 1, 141749
HCCS	116.0	100%	99%	Linear skin defects with multiple congenital anomalies 1, 309801
HCFC1	124.7	99%	98%	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541
HCN1	114.5	100%	95%	Epileptic encephalopathy, early infantile, 24, 615871
HCN4	77.5	98%	92%	Brugada syndrome 8, 613123 Sick sinus syndrome 2, 163800
HDAC8	162.1	100%	100%	Cornelia de Lange syndrome 5, 300882
HEATR2	90.6	87%	81%	Ciliary dyskinesia, primary, 18, 614874
HELLS	85.0	91%	87%	Immunodeficiency-centromeric instability-facial anomalies syndrome 4,616911
HEPACAM	113.8	92%	79%	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926
HERC1	158.5	99%	99%	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
HERC2	92.4	78%	74%	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	34.0	70%	63%	Spondylocostal dysostosis 4, autosomal recessive, 613686
HESX1	62.8	99%	88%	Growth hormone deficiency with pituitary anomalies, 182230 Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230
HEXA	105.8	100%	100%	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800 [Hex A pseudodeficiency], 272800
HEXB	120.7	94%	85%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HFE	119.4	100%	100%	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	106.1	100%	100%	Hemochromatosis type 2A,602390
HFM1	43.9	90%	80%	Premature ovarian failure 9, 615724
HGD	116.4	100%	100%	Alkaptonuria, 203500

HGF	128.7	99%	96%	Deafness, autosomal recessive 39, 608265
HGSNAT	100.1	81%	81%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544
HIBCH	64.0	92%	69%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HINT1	54.0	99%	88%	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200
HIVEP2	168.2	100%	100%	Mental retardation, autosomal dominant 43, 616977
HK1	125.2	99%	98%	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285
HLCS	142.8	100%	100%	Holocarboxylase synthetase deficiency, 253270
HMBS	92.7	99%	95%	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000
HMGCL	117.1	100%	100%	HMG-CoA lyase deficiency, 246450
HMGCS2	122.1	100%	100%	HMG-CoA synthase-2 deficiency, 605911
HMOX1	114.0	93%	88%	Heme oxygenase-1 deficiency, 614034 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963
HMX1	17.6	71%	36%	Oculoauricular syndrome, 612109
HNF1A	123.8	99%	96%	Diabetes mellitus, insulin-dependent, 20, 612520 Hepatic adenoma, somatic, 142330 MODY, type III, 600496 Renal cell carcinoma, 144700 {Diabetes mellitus, insulin-dependent}, 222100 {Diabetes mellitus, noninsulin-dependent, 2}, 125853
HNF1B	111.3	100%	100%	Diabetes mellitus, noninsulin-dependent, 125853 Renal cysts and diabetes syndrome, 137920 {Renal cell carcinoma}, 144700
HNF4A	126.8	98%	97%	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026 MODY, type I, 125850 {Diabetes mellitus, noninsulin-dependent}, 125853
HNMT	121.7	100%	96%	Mental retardation, autosomal recessive 51, 616739 {Asthma, susceptibility to}, 600807
HNRNPA1	65.5	90%	74%	Amyotrophic lateral sclerosis 20, 615426 ?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3, 615424
HNRNPDL	59.3	94%	79%	Muscular dystrophy, limb-girdle, type 1G, 609115
HNRNPK	63.9	83%	74%	Au-Kline syndrome, 616580

HOGA1	115.2	100%	89%	Hyperoxaluria, primary, type III, 613616
HOXA1	127.0	100%	100%	Athabaskan brainstem dysgenesis syndrome, 601536 Bosley-Salih-Alorainy syndrome, 601536
HOXA11	89.9	92%	79%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
HOXA13	41.5	70%	62%	Guttmacher syndrome, 176305 Hand-foot-uterus syndrome, 140000
HOXB1	93.6	100%	100%	Facial paresis, hereditary congenital, 3, 614744
HOXC13	81.4	95%	87%	Ectodermal dysplasia 9, hair/nail type, 614931
HOXD10	114.7	100%	100%	Charcot-Marie-Tooth disease, foot deformity of, 192950 Vertical talus, congenital, 192950
HOXD13	93.2	92%	90%	Brachydactyly, type D, 113200 Brachydactyly, type E, 113300 Syndactyly, type V, 186300 Synpolydactyly 1, 186000 ?Brachydactyly-syndactyly syndrome, 610713
HPCA	223.2	100%	100%	Dystonia 2, torsion, autosomal recessive, 224500
HPD	124.9	100%	100%	Hawkinsinuria, 140350 Tyrosinemia, type III, 276710
HPGD	83.4	100%	97%	Cranioosteoarthropathy, 259100 Digital clubbing, isolated congenital, 119900 Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100
HPRT1	82.3	96%	89%	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322
HPS1	104.9	100%	99%	Hermansky-Pudlak syndrome 1, 203300
HPS3	122.8	100%	98%	Hermansky-Pudlak syndrome 3, 614072
HPS4	124.8	100%	100%	Hermansky-Pudlak syndrome 4, 614073
HPS5	129.0	99%	95%	Hermansky-Pudlak syndrome 5, 614074
HPS6	107.4	98%	85%	Hermansky-Pudlak syndrome 6, 614075
HPSE2	107.3	93%	90%	Urofacial syndrome 1, 236730
HR	86.3	97%	94%	Alopecia universalis, 203655 Atrichia with papular lesions, 209500 Hypotrichosis 4, 146550

HRAS	164.2	100%	100%	Congenital myopathy with excess of muscle spindles, 218040 Costello syndrome, 218040 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 {Bladder cancer, somatic}, 109800 {Nevus sebaceous or woolly hair nevus, somatic}, 162900 {Spitz nevus or nevus spilus, somatic}, 137550 {Thyroid carcinoma, follicular, somatic}, 188470
HRG	150.9	96%	94%	Thrombophilia due to elevated HRG, 613116 Thrombophilia due to HRG deficiency, 613116
HSD11B1	148.7	100%	100%	Cortisone reductase deficiency 2, 614662
HSD11B2	128.2	89%	85%	Apparent mineralocorticoid excess, 218030
HSD17B10	119.0	100%	100%	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 ?Mental retardation, X-linked syndromic 10, 300220
HSD17B3	128.7	100%	100%	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	87.3	92%	89%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B2	139.3	100%	100%	3-beta-hydroxysteroid dehydrogenase, type II, deficiency, 201810
HSD3B7	130.4	100%	93%	Bile acid synthesis defect, congenital, 1, 607765
HSF4	89.9	96%	92%	Cataract 5, multiple types, 116800
HSPA9	79.0	86%	84%	Anemia, sideroblastic, 4, 182170 Even-plus syndrome, 616854
HSPB1	39.0	94%	82%	Charcot-Marie-Tooth disease, axonal, type 2F, 606595 Neuropathy, distal hereditary motor, type IIB, 608634
HSPB8	128.4	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2L, 608673 Neuropathy, distal hereditary motor, type IIA, 158590
HSPD1	74.8	93%	81%	Leukodystrophy, hypomyelinating, 4, 612233 Spastic paraplegia 13, autosomal dominant, 605280
HSPG2	101.2	99%	97%	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
HTR1A	161.3	100%	100%	Periodic fever, menstrual cycle dependent, 614674
HTRA1	83.5	83%	77%	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
HTT	129.6	98%	95%	Huntington disease, 143100
HUWE1	117.8	99%	98%	Mental retardation, X-linked syndromic, Turner type, 300706
HYDIN	114.6	99%	99%	Ciliary dyskinesia, primary, 5, 608647
HYLS1	151.8	100%	100%	Hydrolethalus syndrome, 236680
ICK	125.7	100%	99%	Endocrine-cerebroosteodysplasia, 612651
ICOS	136.6	100%	100%	Immunodeficiency, common variable, 1, 607594
IDH2	81.1	100%	96%	D-2-hydroxyglutaric aciduria 2, 613657
IDH3B	142.9	100%	100%	Retinitis pigmentosa 46, 612572
IDS	112.4	100%	98%	Mucopolysaccharidosis II, 309900
IDUA	85.1	89%	82%	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016
IER3IP1	47.1	87%	77%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	109.1	97%	95%	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFITM5	55.8	93%	85%	Osteogenesis imperfecta, type V, 610967
IFNGR1	125.0	100%	98%	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	120.8	93%	93%	Immunodeficiency 28, mycobacteriosis, 614889
IFT122	130.4	100%	99%	Cranioectodermal dysplasia 1, 218330
IFT140	98.2	99%	97%	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	102.0	99%	97%	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT43	100.8	100%	100%	Cranioectodermal dysplasia 3, 614099
IFT80	48.7	78%	54%	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IGBP1	119.4	99%	95%	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472
IGF1	125.4	100%	100%	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747

IGF1R	121.1	100%	98%	Insulin-like growth factor I, resistance to, 270450
IGF2R	121.1	97%	96%	Hepatocellular carcinoma, somatic, 114550
IGFALS	56.4	100%	94%	Acid-labile subunit, deficiency of, 615961
IGFBP7	60.2	96%	81%	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis, 614224
IGHG2	39.7	82%	60%	IgG2 deficiency, selective
IGHM	176.1	100%	100%	Agammaglobulinemia 1, 601495
IGHMBP2	88.4	96%	89%	Charcot-Marie-Tooth disease, axonal, type 2S, 616155 Neuronopathy, distal hereditary motor, type VI, 604320
IGKC	123.0	100%	100%	Kappa light chain deficiency, 614102
IGLL1	60.7	99%	95%	Agammaglobulinemia 2, 613500
IGSF1	103.0	100%	99%	Hypothyroidism, central, and testicular enlargement, 300888
IHH	107.7	100%	100%	Acrocapitofemoral dysplasia, 607778 Brachydactyly, type A1, 112500
IKBKAP	128.4	99%	98%	Dysautonomia, familial, 223900
IKBKB	107.5	96%	92%	Immunodeficiency 15, 615592
IKBKG	51.0	90%	73%	Ectodermal dysplasia, hypohidrotic, with immune deficiency, 300291 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 Immunodeficiency 33, 300636 Immunodeficiency, isolated, 300584 Incontinentia pigmenti, 308300 Invasive pneumococcal disease, recurrent isolated, 2, 300640
IKZF1	164.1	100%	100%	Immunodeficiency,common variable, 1,616873
IL10RA	125.1	100%	100%	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	166.2	95%	95%	Inflammatory bowel disease 25, early onset, autosomal recessive, 612567 {Hepatitis B virus, susceptibility to}, 610424
IL11RA	122.8	100%	100%	Craniosynostosis and dental anomalies, 614188
IL12B	99.4	100%	97%	Immunodeficiency 29, mycobacteriosis, 614890
IL12RB1	110.8	96%	93%	Immunodeficiency 30, 614891
IL17RC	79.3	99%	96%	Candidiasis, familial, 9, 616445
IL17RD	114.4	100%	96%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
IL1RAPL1	150.3	100%	100%	Mental retardation, X-linked 21/34, 300143
IL1RN	147.6	100%	100%	Interleukin 1 receptor antagonist deficiency, 612852 {Gastric cancer risk after H. pylori infection}, 137215 {Microvascular complications of diabetes 4}, 612628

IL21R	116.3	100%	100%	Immunodeficiency, primary, autosomal recessive, IL21R-related, 615207 [IgE, elevated level of], 147050
IL2RA	104.6	100%	96%	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367 {Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942
IL2RG	83.4	100%	100%	Combined immunodeficiency, X-linked, moderate, 312863 Severe combined immunodeficiency, X-linked, 300400
IL31RA	124.3	100%	100%	Amyloidosis, primary localized cutaneous, 2, 613955
IL36RN	74.6	100%	100%	Psoriasis 14, pustular, 614204
IL7R	120.6	100%	99%	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
ILDR1	94.1	99%	98%	Deafness, autosomal recessive 42, 609646
IMPAD1	126.8	100%	99%	Chondrodysplasia with joint dislocations, GPAPP type, 614078
IMPDH1	45.0	87%	77%	Leber congenital amaurosis 11, 613837 Retinitis pigmentosa 10, 180105
IMPG1	99.8	99%	97%	Macular dystrophy, vitelliform, 4, 616151
IMPG2	151.8	98%	96%	Macular dystrophy, vitelliform, 5, 616152 Retinitis pigmentosa 56, 613581
INF2	73.6	92%	89%	Charcot-Marie-Tooth disease, dominant intermediate E, 614455 Glomerulosclerosis, focal segmental, 5, 613237
ING1	102.6	100%	95%	Squamous cell carcinoma, head and neck, somatic, 275355
INPP5E	86.6	92%	90%	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INPPL1	107.8	97%	90%	Opsismodysplasia, 258480
INS	84.7	100%	100%	Diabetes mellitus, insulin-dependent, 2, 125852 Diabetes mellitus, permanent neonatal, 606176 Hyperproinsulinemia, 616214 Maturity-onset diabetes of the young, type 10, 613370
INSL3	80.9	80%	80%	Cryptorchidism, 219050
INSR	112.2	95%	92%	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968 Leprechaunism, 246200 Rabson-Mendenhall syndrome, 262190
INVS	144.1	99%	99%	Nephronophthisis 2, infantile, 602088
IQCB1	87.6	86%	71%	Senior-Loken syndrome 5, 609254
IQSEC2	72.6	95%	88%	Mental retardation, X-linked 1/78, 309530

IRAK4	87.9	100%	85%	Invasive pneumococcal disease, recurrent isolated, 1, 610799 IRAK4 deficiency, 607676
IRF1	143.6	100%	98%	Gastric cancer, somatic, 613659 Myelodysplastic syndrome, preleukemic Myelogenous leukemia, acute Nonsmall cell lung cancer, somatic, 211980
IRF6	106.9	100%	97%	Popliteal pterygium syndrome 1, 119500 van der Woude syndrome, 119300 {Orofacial cleft 6}, 608864
IRF8	94.4	97%	93%	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893 Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 614894
IRGM	161.8	100%	100%	Inflammatory bowel disease 19, 612278 {Mycobacterium tuberculosis, protection against}, 607948
IRX5	67.0	89%	82%	Hamamy syndrome, 611174
ISCA2	61.6	98%	83%	Multiple mitochondrial dysfunctions syndrome 4, 616370
ISCU	112.0	100%	100%	Myopathy with lactic acidosis, hereditary, 255125
ISG15	121.9	100%	100%	Immunodeficiency 38, 616126
ISPD	97.6	99%	84%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052
ITCH	106.4	95%	95%	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITGA2B	99.8	98%	96%	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Glanzmann thrombasthenia, 273800 Thrombocytopenia, neonatal alloimmune, BAK antigen related
ITGA3	118.9	98%	95%	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748
ITGA6	134.2	99%	98%	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730
ITGA7	108.9	99%	96%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
ITGA8	118.0	99%	97%	Renal hypodysplasia/aplasia 1, 191830
ITGB2	148.7	100%	100%	Leukocyte adhesion deficiency, 116920
ITGB3	125.5	98%	96%	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Glanzmann thrombasthenia, 273800 Purpura, posttransfusion Thrombocytopenia, neonatal alloimmune {Myocardial infarction, susceptibility to}, 608446

ITGB4	123.4	97%	94%	Epidermolysis bullosa of hands and feet, 131800 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa, junctional, with pyloric atresia, 226730
ITGB6	126.9	95%	95%	Amelogenesis imperfecta, type IH, 616221
ITK	110.8	100%	97%	Lymphoproliferative syndrome 1, 613011
ITM2B	101.9	100%	94%	Dementia, familial British, 176500 Dementia, familial Danish, 117300 ?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079
ITPA	112.5	100%	100%	Epileptic encephalopathy, early infantile, 35, 616647 [Inosine triphosphatase deficiency], 613850
ITPR1	135.9	100%	98%	Gillespie syndrome,206700 Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360
IVD	98.7	100%	97%	Isovaleric acidemia, 243500
IYD	101.3	98%	96%	Thyroid dyshormonogenesis 4, 274800
JAG1	127.4	98%	97%	Alagille syndrome, 118450 Tetralogy of Fallot, 187500 ?Deafness, congenital heart defects, and posterior embryotoxon
JAGN1	126.1	100%	100%	Neutropenia, severe congenital, 6, autosomal recessive, 616022
JAK2	83.3	95%	88%	Erythrocytosis, somatic, 133100 Leukemia, acute myeloid, somatic, 601626 Myelofibrosis, somatic, 254450 Polycythemia vera, somatic, 263300 Thrombocythemia 3, 614521 {Budd-Chiari syndrome, somatic}, 600800
JAK3	87.2	96%	93%	SCID, autosomal recessive, T-negative/B-positive type, 600802
JAM3	133.8	100%	97%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
JPH2	86.5	94%	83%	Cardiomyopathy, hypertrophic, 17, 613873
JPH3	117.9	100%	100%	Huntington disease-like 2, 606438
JUP	126.3	99%	98%	Arrhythmogenic right ventricular dysplasia 12, 611528 Naxos disease, 601214
KAL1	100.6	89%	86%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
KANK1	133.8	100%	100%	Cerebral palsy, spastic quadriplegic, 2, 612900
KANK2	130.3	100%	99%	Palmoplantar keratoderma and woolly hair, 616099

KANSL1	63.3	94%	85%	Koolen-De Vries syndrome, 610443
KARS	104.7	100%	99%	Deafness, autosomal recessive 89, 613916 ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641
KAT6A	154.1	100%	99%	Mental retardation, autosomal dominant 32, 616268
KAT6B	150.7	99%	97%	Genitopatellar syndrome, 606170 SBBYSS syndrome, 603736
KATNB1	125.5	100%	100%	Lissencephaly 6, with microcephaly, 616212
KBTBD13	87.8	98%	92%	Nemaline myopathy 6, autosomal dominant, 609273
KCNA1	124.9	99%	98%	Episodic ataxia/myokymia syndrome, 160120
KCNA2	130.8	100%	100%	Epileptic encephalopathy, early infantile, 32, 616366
KCNA5	130.3	100%	93%	Atrial fibrillation, familial, 7, 612240
KCNB1	115.8	100%	100%	Epileptic encephalopathy, early infantile, 26, 616056
KCNC1	155.0	100%	100%	Epilepsy, progressive myoclonic 7, 616187
KCNC3	132.5	66%	57%	Spinocerebellar ataxia 13, 605259
KCND3	168.2	100%	96%	Brugada syndrome 9, 616399 Spinocerebellar ataxia 19, 607346
KCNE1	388.5	100%	100%	Jervell and Lange-Nielsen syndrome 2, 612347 Long QT syndrome 5, 613695
KCNE2	142.0	100%	94%	Atrial fibrillation, familial, 4, 611493 Long QT syndrome 6, 613693
KCNE3	158.2	100%	100%	Brugada syndrome 6, 613119
KCNH1	157.5	100%	99%	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500
KCNH2	90.3	91%	82%	Long QT syndrome 2, 613688 Short QT syndrome 1, 609620 {Long QT syndrome 2, acquired, susceptibility to}, 613688
KCNJ1	205.1	100%	100%	Bartter syndrome, type 2, 241200
KCNJ10	171.3	100%	98%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ11	230.5	100%	100%	Diabetes mellitus, permanent neonatal, with neurologic features, 606176 Diabetes mellitus, transient neonatal, 3, 610582 Diabetes, permanent neonatal, 606176 Hyperinsulinemic hypoglycemia, familial, 2, 601820 Maturity-onset diabetes of the young, type 13, 616329

				{Diabetes mellitus, type 2, susceptibility to}, 125853
KCNJ13	165.1	100%	100%	Leber congenital amaurosis 16, 614186 Snowflake vitreoretinal degeneration, 193230
KCNJ2	172.1	100%	100%	Andersen syndrome, 170390 Atrial fibrillation, familial, 9, 613980 Short QT syndrome 3, 609622
KCNJ5	183.2	99%	96%	Hyperaldosteronism, familial, type III, 613677 Long QT syndrome 13, 613485
KCNJ6	155.0	100%	100%	Keppen-Lubinsky syndrome, 614098
KCNK3	124.7	98%	94%	Pulmonary hypertension, primary, 4, 615344
KCNK9	152.4	100%	100%	Birk-Barel mental retardation dysmorphism syndrome, 612292
KCNMA1	119.7	100%	100%	Generalized epilepsy and paroxysmal dyskinesia, 609446
KCNN4	102.7	100%	100%	Dehydrated hereditary stomatocytosis 2, 616689
KCNQ1	102.6	93%	91%	Atrial fibrillation, familial, 3, 607554 Jervell and Lange-Nielsen syndrome, 220400 Long QT syndrome 1, 192500 Short QT syndrome 2, 609621 {Long QT syndrome 1, acquired, susceptibility to}, 192500
KCNQ1OT1	NC	NC	NC	Beckwith-Wiedemann syndrome, 130650
KCNQ2	84.4	99%	98%	Epileptic encephalopathy, early infantile, 7, 613720 Myokymia, 121200 Seizures, benign neonatal, 1, 121200
KCNQ3	93.9	100%	95%	Seizures, benign neonatal, type 2, 121201
KCNQ4	119.9	94%	85%	Deafness, autosomal dominant 2A, 600101
KCNT1	98.7	95%	89%	Epilepsy, nocturnal frontal lobe, 5, 615005 Epileptic encephalopathy, early infantile, 14, 614959
KCNV2	106.4	100%	100%	Retinal cone dystrophy 3B, 610356
KCTD1	119.4	91%	85%	Scalp-ear-nipple syndrome, 181270
KCTD17	90.2	97%	89%	Dystonia 26, myoclonic, 616398
KCTD7	117.8	94%	92%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726

KDM1A	121.4	97%	93%	Cleft palate, psychomotor retardation, and distinctive facial features, 616728
KDM5C	126.4	97%	96%	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
KDM6A	127.3	94%	89%	Kabuki syndrome 2, 300867
KDR	128.8	100%	99%	Hemangioma, capillary infantile, somatic, 602089 {Hemangioma, capillary infantile, susceptibility to}, 602089
KERA	173.0	100%	100%	Cornea plana congenita, recessive, 217300
KHDC3L	125.7	100%	99%	Hydatidiform mole, recurrent, 2, 614293
KIAA0196	123.8	97%	95%	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, autosomal dominant, 603563
KIAA0586	101.5	99%	89%	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546
KIAA1279	165.1	100%	99%	Goldberg-Shprintzen megacolon syndrome, 609460
KIAA2022	186.8	100%	99%	Mental retardation, X-linked 98, 300912
KIF11	78.5	97%	91%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF1A	112.1	99%	95%	Mental retardation, autosomal dominant 9, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal recessive, 610357
KIF1B	137.7	99%	98%	Pheochromocytoma, 171300 ?Charcot-Marie-Tooth disease, type 2A1, 118210 {Neuroblastoma, susceptibility to, 1}, 256700
KIF1C	93.5	100%	99%	Spastic ataxia 2, autosomal recessive, 611302
KIF21A	111.2	99%	95%	Fibrosis of extraocular muscles, congenital, 1, 135700 Fibrosis of extraocular muscles, congenital, 3B, 135700
KIF22	127.7	99%	98%	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546
KIF2A	93.6	96%	82%	Cortical dysplasia, complex, with other brain malformations 3, 615411
KIF5A	111.5	100%	99%	Spastic paraplegia 10, autosomal dominant, 604187
KIF5C	98.0	99%	97%	Cortical dysplasia, complex, with other brain malformations 2, 615282
KIF7	71.1	96%	84%	Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120
KIRREL3	130.2	100%	98%	Mental retardation, autosomal dominant 4, 612581
KISS1R	82.5	99%	96%	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 ?Precocious puberty, central, 1, 176400

KIT	140.5	100%	99%	Gastrointestinal stromal tumor, familial, 606764 Germ cell tumors, 273300 Leukemia, acute myeloid, 601626 Mast cell disease, 154800 Piebaldism, 172800
KITLG	75.4	92%	85%	Deafness, congenital, unilateral or asymmetric, 616697 Hyperpigmentation with or without hypopigmentation, 145250 [Skin/hair/eye pigmentation 7, blond/brown hair], 611664
KIZ	155.9	100%	95%	Retinitis pigmentosa 69, 615780
KL	150.2	95%	94%	Tumoral calcinosis, hyperphosphatemic, 211900 {Coronary artery disease, susceptibility to}
KLC2	100.9	100%	98%	Spastic paraplegia, optic atrophy, and neuropathy, 609541
KLF1	48.6	90%	83%	Blood group--Lutheran inhibitor, 111150 Dyserythropoietic anemia, congenital, type IV, 613673 [Hereditary persistence of fetal hemoglobin], 613566
KLF11	145.5	100%	100%	Maturity-onset diabetes of the young, type VII, 610508
KLF6	115.1	100%	96%	Gastric cancer, somatic, 613659 Prostate cancer, somatic, 176807
KLHL10	177.6	100%	98%	Spermatogenic failure 11, 615081
KLHL3	122.5	99%	99%	Pseudohypoaldosteronism, type IID, 614495
KLHL40	113.8	100%	100%	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	173.4	100%	99%	Nemaline myopathy 9, 615731
KLHL7	112.9	98%	96%	Cold induced sweating syndrome 3, 617055 Retinitis pigmentosa 42, 612943
KLK4	170.7	100%	98%	Amelogenesis imperfecta, type IIA1, 204700
KLKB1	142.2	93%	92%	Fletcher factor (prekallikrein) deficiency, 612423
KLLN	108.6	100%	100%	Cowden syndrome 4, 615107
KMT2A	141.3	99%	98%	Leukemia, myeloid/lymphoid or mixed-lineage Wiedemann-Steiner syndrome, 605130
KMT2D	126.4	100%	99%	Kabuki syndrome 1, 147920
KPTN	96.1	100%	98%	Mental retardation, autosomal recessive 41, 615637

KRAS	54.9	100%	99%	Bladder cancer, somatic, 109800 Breast cancer, somatic, 114480 Cardiofaciocutaneous syndrome 2, 615278 Gastric cancer, somatic, 137215 Leukemia, acute myeloid, 601626 Lung cancer, somatic, 211980 Noonan syndrome 3, 609942 Pancreatic carcinoma, somatic, 260350 RAS-associated autoimmune leukoproliferative disorder, 614470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200
KRIT1	76.6	98%	90%	Cavernous malformations of CNS and retina, 116860 Cerebral cavernous malformations-1, 116860 Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations, 116860
KRT1	121.0	100%	100%	Epidermolytic hyperkeratosis, 113800 Ichthyosis histrix, Curth-Macklin type, 146590 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602 Keratosis palmoplantaris striata III, 607654 Palmoplantar keratoderma, epidermolytic, 144200 Palmoplantar keratoderma, nonepidermolytic, 600962
KRT10	89.5	99%	90%	Epidermolytic hyperkeratosis, 113800 Ichthyosis with confetti, 609165 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602
KRT12	98.7	97%	93%	Meesmann corneal dystrophy, 122100
KRT13	103.3	100%	99%	White sponge nevus 2, 615785
KRT14	47.9	87%	78%	Dermatopathia pigmentosa reticularis, 125595 Epidermolysis bullosa simplex, Dowling-Meara type, 131760 Epidermolysis bullosa simplex, Koebner type, 131900 Epidermolysis bullosa simplex, recessive 1, 601001 Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 Naegeli-Franceschetti-Jadassohn syndrome, 161000
KRT16	34.1	73%	53%	Pachyonychia congenita 1, 167200 Palmoplantar keratoderma, nonepidermolytic, focal, 613000
KRT17	18.5	50%	34%	Pachyonychia congenita 2, 167210 Steatocystoma multiplex, 184500

KRT18	27.9	64%	60%	Cirrhosis, cryptogenic, 215600 {Cirrhosis, noncryptogenic, susceptibility to}, 215600
KRT2	107.4	98%	97%	Ichthyosis bullosa of Siemens, 146800
KRT25	127.7	100%	100%	Woolly hair, autosomal recessive 3, 616760
KRT3	83.2	100%	99%	Meesmann corneal dystrophy, 122100
KRT4	101.7	100%	100%	White sponge nevus 1, 193900
KRT5	103.1	100%	100%	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-MP, 131960 Epidermylysis bullosa simplex-MCR, 609352
KRT6A	137.9	90%	83%	Pachyonychia congenita 3, 615726
KRT6B	136.6	88%	85%	Pachyonychia congenita 4, 615728
KRT6C	129.4	84%	77%	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735
KRT74	118.7	100%	100%	Woolly hair, autosomal dominant, 194300 ?Ectodermal dysplasia 7, hair/nail type, 614929 ?Hypotrichosis 3, 613981
KRT8	31.5	89%	67%	Cirrhosis, cryptogenic, 215600 {Cirrhosis, noncryptogenic, susceptibility to}, 215600
KRT81	81.1	100%	92%	Monilethrix, 158000
KRT85	90.4	99%	94%	Ectodermal dysplasia 4, hair/nail type, 602032
KRT86	88.2	100%	93%	Monilethrix, 158000
KRT9	67.1	95%	93%	Palmoplantar keratoderma, epidermolytic, 144200
L1CAM	153.4	100%	99%	Corpus callosum, partial agenesis of, 304100 CRASH syndrome, 303350 Hydrocephalus due to aqueductal stenosis, 307000 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 Hydrocephalus with Hirschsprung disease, 307000 MASA syndrome, 303350
L2HGDH	116.2	97%	94%	L-2-hydroxyglutaric aciduria, 236792
LAMA1	124.1	100%	99%	Poretti-Boltshauser syndrome, 615960
LAMA2	134.3	99%	99%	Muscular dystrophy, congenital merosin-deficient, 607855

				Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855
LAMA3	134.4	99%	98%	Epidermolysis bullosa, generalized atrophic benign, 226650 Epidermolysis bullosa, junctional, Herlitz type, 226700 Laryngoonychocutaneous syndrome, 245660
LAMA4	121.3	100%	99%	Cardiomyopathy, dilated, 1JJ, 615235
LAMB1	150.6	100%	98%	Lissencephaly 5, 615191
LAMB2	162.4	99%	98%	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 Pierson syndrome, 609049
LAMB3	105.8	100%	98%	Amelogenesis imperfecta, type IA, 104530 Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LAMC2	105.1	99%	96%	Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LAMC3	101.4	97%	93%	Cortical malformations, occipital, 614115
LAMP2	143.8	92%	92%	Danon disease, 300257
LAMTOR2	149.8	100%	100%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LARGE	113.7	100%	98%	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	46.0	79%	59%	Alazami syndrome, 615071
LARS2	119.8	100%	100%	Perrault syndrome 4, 615300 ?Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LAS1L	110.5	100%	99%	Wilson-Turner syndrome, 309585
LBR	72.7	93%	77%	Greenberg skeletal dysplasia, 215140 Pelger-Huet anomaly, 169400 ?Reynolds syndrome, 613471
LCA5	112.2	99%	95%	Leber congenital amaurosis 5, 604537
LCAT	131.8	100%	88%	Fish-eye disease, 136120 Norum disease, 245900
LCT	111.0	99%	96%	Lactase deficiency, congenital, 223000
LDB3	106.8	94%	93%	Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 Cardiomyopathy, hypertrophic, 24, 601493 Left ventricular noncompaction 3, 601493 Myopathy, myofibrillar, 4, 609452
LDHA	48.5	91%	84%	Glycogen storage disease XI, 612933

LDLR	147.6	97%	95%	Hypercholesterolemia, familial, 143890 LDL cholesterol level QTL2, 143890
LDLRAP1	130.3	100%	90%	Hypercholesterolemia, familial, autosomal recessive, 603813
LEF1	103.9	100%	100%	Sebaceous tumors, somatic, 153245
LEFTY2	40.2	85%	71%	Left-right axis malformations, 601877
LEMD2	60.4	95%	80%	Cataract 46, juvenile-onset, 212500
LEMD3	87.9	95%	90%	Buschke-Ollendorff syndrome, 166700 Melorheostosis with osteopoikilosis, 155950 Osteopoikilosis, 166700
LEP	161.7	100%	96%	Obesity, morbid, due to leptin deficiency, 614962
LEPR	103.7	93%	89%	Obesity, morbid, due to leptin receptor deficiency, 614963
LEPRE1	118.0	100%	100%	Osteogenesis imperfecta, type VIII, 610915
LEPREL1	89.1	99%	91%	Myopia, high, with cataract and vitreoretinal degeneration, 614292
LGI1	177.4	100%	92%	Epilepsy, familial temporal lobe, 1, 600512
LHB	26.2	99%	74%	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300
LHCGR	141.9	92%	92%	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	215.9	100%	100%	Deafness, autosomal recessive 67, 610265
LHX3	88.2	100%	96%	Pituitary hormone deficiency, combined, 3, 221750
LHX4	109.6	100%	100%	Pituitary hormone deficiency, combined, 4, 262700
LIAS	135.1	100%	97%	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIFR	112.9	96%	89%	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LIG1	81.7	100%	97%	DNA ligase I deficiency
LIG4	150.3	100%	100%	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500
LIM2	89.2	100%	98%	Cataract 19, multiple types, 615277
LIMS2	99.5	93%	92%	Muscular dystrophy, limb-girdle, type 2W, 616827
LINS	119.0	100%	97%	Mental retardation, autosomal recessive 27, 614340
LIPA	104.1	95%	93%	Cholesteryl ester storage disease, 278000 Wolman disease, 278000

LIPC	101.3	100%	98%	Hepatic lipase deficiency, 614025 [High density lipoprotein cholesterol level QTL 12], 612797 {Diabetes mellitus, noninsulin-dependent}, 125853
LIPE	93.5	98%	97%	Lipodystrophy, familial partial, type 6, 615980
LIPH	111.6	100%	100%	Hypotrichosis 7, 604379 Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379
LIPN	120.4	100%	92%	Ichthyosis, congenital, autosomal recessive 8, 613943
LIPT1	218.4	100%	100%	Lipoyltransferase 1 deficiency, 616299
LITAF	100.4	97%	90%	Charcot-Marie-Tooth disease, type 1C, 601098
LMAN1	116.1	96%	87%	Combined factor V and VIII deficiency, 227300
LMBR1	90.2	98%	92%	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	65.8	89%	81%	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LMF1	125.2	100%	98%	Lipase deficiency, combined, 246650
LMNA	69.2	91%	86%	Cardiomyopathy, dilated, 1A, 115200 Charcot-Marie-Tooth disease, type 2B1, 605588 Emery-Dreifuss muscular dystrophy 2, AD, 181350 Emery-Dreifuss muscular dystrophy 3, AR, 616516 Heart-hand syndrome, Slovenian type, 610140 Hutchinson-Gilford progeria, 176670 Lipodystrophy, familial partial, 2, 151660 Malouf syndrome, 212112 Mandibuloacral dysplasia, 248370 Muscular dystrophy, congenital, 613205 Muscular dystrophy, limb-girdle, type 1B, 159001 Restrictive dermopathy, lethal, 275210
LMNB1	111.0	100%	97%	Leukodystrophy, adult-onset, autosomal dominant, 169500
LMOD3	130.1	100%	99%	Nemaline myopathy 10, 616165
LMX1B	99.4	100%	88%	Nail-patella syndrome, 161200
LONP1	131.6	96%	90%	CODAS syndrome, 600373

LOR	14.2	80%	25%	Vohwinkel syndrome with ichthyosis, 604117
LOXHD1	114.7	100%	98%	Deafness, autosomal recessive 77, 613079
LPAR6	97.4	96%	93%	Hypotrichosis 8, 278150 Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150
LPIN1	118.6	98%	90%	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	109.0	100%	100%	Majeed syndrome, 609628
LPL	137.8	100%	99%	Combined hyperlipidemia, familial, 144250 Lipoprotein lipase deficiency, 238600 [High density lipoprotein cholesterol level QTL 11]
LPP	112.7	100%	100%	Leukemia, acute myeloid, 601626 Lipoma
LRAT	239.2	100%	100%	Leber congenital amaurosis 14, 613341 Retinal dystrophy, early-onset severe, 613341 Retinitis pigmentosa, juvenile, 613341
LRBA	120.2	98%	95%	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRIG2	138.6	97%	97%	Urofacial syndrome 2, 615112
LRIT3	138.9	93%	92%	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRP2	154.4	100%	99%	Donnai-Barrow syndrome, 222448
LRP4	141.2	98%	98%	Cenani-Lenz syndactyly syndrome, 212780 Sclerosteosis 2, 614305 ?Myasthenic syndrome, congenital, 17, 616304
LRP5	155.0	99%	98%	Exudative vitreoretinopathy 4, 601813 Hyperostosis, endosteal, 144750 Osteopetrosis, autosomal dominant 1, 607634 Osteoporosis-pseudoglioma syndrome, 259770 Osteosclerosis, 144750 van Buchem disease, type 2, 607636 [Bone mineral density variability 1], 601884 {Osteoporosis}, 166710
LRP6	136.3	100%	99%	Tooth agenesis, selective, 7, 616724 {Coronary artery disease, autosomal dominant, 2}, 610947
LRPAP1	121.1	97%	90%	Myopia 23, autosomal recessive, 615431
LRPPRC	113.0	99%	95%	Leigh syndrome, French-Canadian type, 220111
LRRC6	152.6	95%	89%	Ciliary dyskinesia, primary, 19, 614935

LRRC8A	222.9	100%	99%	Agammaglobulinemia 5, 613506
LRSAM1	123.0	99%	98%	Charcot-Marie-Toothe disease, axonal, type 2P, 614436
LRTOMT	108.8	93%	91%	Deafness, autosomal recessive 63, 611451
LSS	101.4	100%	98%	Cataract 44, 616509
LTBP2	89.0	99%	97%	Glaucoma 3, primary congenital, D, 613086 Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750 Weill-Marchesani syndrome 3, recessive, 614819
LTBP3	99.3	97%	95%	Dental anomalies and short stature, 601216
LTBP4	96.2	99%	94%	Cutis laxa, autosomal recessive, type IC, 613177
LYRM7	36.1	82%	57%	Mitochondrial complex III deficiency, nuclear type 8, 615838
LYST	122.8	98%	92%	Chediak-Higashi syndrome, 214500
LYZ	156.8	100%	100%	Amyloidosis, renal, 105200
LZTFL1	111.6	99%	92%	Bardet-Biedl syndrome 17, 615994
LZTR1	124.5	100%	97%	Noonan syndrome 10, 616564 {Schwannomatosis-2, susceptibility to}, 615670
LZTS1	78.8	100%	100%	Esophageal squamous cell carcinoma, 133239
MAB21L2	206.1	100%	100%	Microphthalmia, syndromic 14, 615877
MAD1L1	87.0	100%	93%	Lymphoma, somatic Prostate cancer, somatic, 176807
MAF	48.5	74%	69%	Ayme-Gripp syndrome, 601088 Cataract 21, multiple types, 610202
MAFB	95.6	100%	98%	Duane retraction syndrome 3, 617041 Multicentric carpotarsal osteolysis syndrome, 166300
MAG	121.4	100%	99%	Spastic paraplegia 75, autosomal recessive, 616680
MAGED2	90.0	100%	99%	Bartter syndrome, type 5,antenatal,transient,300971
MAGEL2	115.5	100%	100%	Schaaf-Yang syndrome, 615547
MAGT1	136.6	98%	98%	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853
MAK	134.3	93%	93%	Retinitis pigmentosa 62, 614181
MALT1	125.3	90%	83%	Immunodeficiency 12, 615468
MAML2	110.6	100%	100%	Mucoepidermoid salivary gland carcinoma
MAMLD1	144.7	100%	99%	Hypospadias 2, X-linked, 300758
MAN1B1	128.5	100%	99%	Mental retardation, autosomal recessive 15, 614202
MAN2B1	108.0	98%	94%	Mannosidosis, alpha-, types I and II, 248500

MANBA	112.5	100%	93%	Mannosidosis, beta, 248510
MAOA	148.0	100%	100%	Brunner syndrome, 300615 {Antisocial behavior},300615
MAP2K1	86.9	100%	92%	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	92.2	94%	88%	Cardiofaciocutaneous syndrome 4, 615280
MAP3K1	143.4	92%	88%	46XY sex reversal 6, 613762
MAP3K8	140.2	100%	100%	Lung cancer, somatic, 211980
MAPRE2	175.8	100%	99%	Symmetric circumferential skin creases, congenital, 2, 616734
MAPT	55.9	99%	87%	Dementia, frontotemporal, with or without parkinsonism, 600274 Pick disease, 172700 Supranuclear palsy, progressive atypical, 260540 Supranuclear palsy, progressive, 601104 {Parkinson disease, susceptibility to}, 168600
MARS	112.5	100%	96%	Charcot-Marie-Tooth disease, axonal, type 2U, 616280 Interstitial lung and liver disease, 615486
MARS2	128.6	100%	100%	Spastic ataxia 3, autosomal recessive, 611390 ?Combined oxidative phosphorylation deficiency 25, 616430
MARVELD2	142.2	94%	92%	Deafness, autosomal recessive 49, 610153
MASP1	123.7	100%	99%	3MC syndrome 1, 257920
MASP2	125.0	99%	95%	MASP2 deficiency, 613791
MAT1A	141.9	97%	94%	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MATN3	100.1	84%	84%	Epiphyseal dysplasia, multiple, 5, 607078 Spondyloepimetaphyseal dysplasia, 608728 {Osteoarthritis susceptibility 2}, 140600
MATR3	74.3	95%	86%	Amyotrophic lateral sclerosis 21, 606070
MBD5	160.9	100%	100%	Mental retardation, autosomal dominant 1, 156200
MBTPS2	147.4	100%	98%	IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800 ?Olmsted syndrome, X-linked, 300918
MC2R	165.9	100%	98%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MC4R	263.3	100%	100%	Obesity, autosomal dominant, 601665
MCC	125.4	99%	98%	Colorectal cancer, somatic, 114500

MCCC1	131.2	100%	99%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	116.7	99%	97%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	89.0	100%	100%	Methylmalonyl-CoA epimerase deficiency, 251120
MCFD2	90.9	100%	100%	Factor V and factor VIII, combined deficiency of, 613625
MCM4	145.9	100%	99%	Natural killer cell and glucocorticoid deficiency with DNA repair defect, 609981
MCM6	150.2	100%	100%	Lactase persistence/nonpersistence, 223100
MCM9	128.7	100%	100%	Ovarian dysgenesis 4, 616185
MCOLN1	129.3	99%	96%	Mucolipidosis IV, 252650
MCPH1	123.8	99%	95%	Microcephaly 1, primary, autosomal recessive, 251200
MECOM	133.9	99%	97%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MECP2	100.5	100%	97%	Encephalopathy, neonatal severe, 300673 Mental retardation, X-linked syndromic, Lubs type, 300260 Mental retardation, X-linked, syndromic 13, 300055 Rett syndrome, 312750 Rett syndrome, atypical, 312750 Rett syndrome, preserved speech variant, 312750 {Autism susceptibility, X-linked 3}, 300496
MED12	118.1	100%	97%	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Opitz-Kaveggia syndrome, 305450
MED13L	118.3	99%	97%	Mental retardation and distinctive facial features with or without cardiac defects, 616789 Transposition of the great arteries, dextro-looped 1, 608808
MED17	116.5	98%	93%	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	116.3	99%	97%	Mental retardation, autosomal recessive 18, 614249
MED25	90.7	98%	94%	Basel-Vanagait-Smirin-Yosef syndrome, 616449 ?Charcot-Marie-Tooth disease, type 2B2, 605589
MEF2C	121.4	91%	88%	Chromosome 5q14.3 deletion syndrome, 613443 Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
MEFV	104.9	94%	89%	Familial Mediterranean fever, AD, 134610 Familial Mediterranean fever, AR, 249100
MEGF10	133.4	100%	100%	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399
MEGF8	103.7	99%	96%	Carpenter syndrome 2, 614976

MEN1	103.7	99%	92%	Adrenal adenoma, somatic Angiofibroma, somatic Carcinoid tumor of lung Lipoma, somatic Multiple endocrine neoplasia 1, 131100 Parathyroid adenoma, somatic
MEOX1	82.2	95%	92%	Klippel-Feil syndrome 2, 214300
MERTK	151.4	100%	98%	Retinitis pigmentosa 38, 613862
MESP2	61.2	89%	81%	Spondylocostal dysostosis 2, autosomal recessive, 608681
MET	169.1	100%	99%	Hepatocellular carcinoma, childhood type, somatic, 114550 Renal cell carcinoma, papillary, 1, familial and somatic, 605074 ?Deafness, autosomal recessive 97, 616705 {Osteofibrous dysplasia,susceptibility to},607278
METTL23	119.1	100%	100%	Mental retardation, autosomal recessive 44, 615942
MFAP5	107.5	100%	99%	Aortic aneurysm, familial thoracic 9, 616166
MFN2	120.8	100%	99%	Charcot-Marie-Tooth disease, type 2A2, 609260 Hereditary motor and sensory neuropathy VIA, 601152
MFRP	118.7	100%	100%	Microphthalmia, isolated 5, 611040 Nanophthalmos 2, 609549
MFSD2A	109.3	100%	100%	Microcephaly 15, primary, autosomal recessive, 616486
MFSD8	111.8	100%	99%	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170
MGAT2	117.0	100%	99%	Congenital disorder of glycosylation, type IIa, 212066
MGME1	161.5	100%	99%	Mitochondrial DNA depletion syndrome 11, 615084
MGP	139.7	91%	91%	Keutel syndrome, 245150
MIB1	131.7	100%	98%	Left ventricular noncompaction 7, 615092
MICU1	109.5	96%	92%	Myopathy with extrapyramidal signs, 615673
MID1	182.3	100%	100%	Opitz GBBB syndrome, type I, 300000
MINPP1	137.6	99%	96%	Thyroid carcinoma, follicular, 188470
MIP	99.2	90%	85%	Cataract 15, multiple types, 615274
MIR17HG	NC	NC	NC	Feingold syndrome 2, 614326
MIR184	NC	NC	NC	EDICT syndrome, 614303
MIR96	NC	NC	NC	Deafness, autosomal dominant 50, 613074

MITF	128.3	100%	100%	Tietz albinism-deafness syndrome, 103500 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456
MKKS	185.3	89%	89%	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKL1	97.5	96%	88%	Megakaryoblastic leukemia, acute
MKRN3	96.1	100%	98%	Precocious puberty, central, 2, 615346
MKS1	87.0	99%	97%	Bardet-Biedl syndrome 13, 615990 Meckel syndrome 1, 249000
MLC1	95.3	100%	98%	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MLH1	147.8	100%	99%	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MLH3	143.8	100%	99%	Colorectal cancer, hereditary nonpolyposis, type 7, 614385 Colorectal cancer, somatic, 114500 {Endometrial cancer, susceptibility to}, 608089
MLLT10	121.2	95%	90%	Leukemia,acute myeloid,601626
MLLT11	79.5	100%	97%	Leukemia, acute myelomonocytic, somatic, 607785
MLPH	85.8	100%	98%	Griscelli syndrome, type 3, 609227
MLYCD	68.7	92%	89%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	156.2	100%	100%	Methylmalonic aciduria, vitamin B12-responsive, 251100
MMAB	88.3	100%	97%	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110
MMACHC	159.5	100%	100%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	58.5	82%	64%	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410
MME	88.8	96%	89%	Charcot-Marie-Tooth disease, axonal, type 2T, 617017 ?Spinocerebellar ataxia 43, 617018
MMP1	147.3	100%	97%	COPD, rate of decline of lung function in, 606963 {Epidermolysis bullosa dystrophica, autosomal recessive, modifier of}, 226600
MMP13	114.2	92%	91%	Metaphyseal anadysplasia 1, 602111 Spondyloepimetaphyseal dysplasia, Missouri type, 602111

MMP19	109.2	100%	99%	Cavitary optic disc anomalies, 611543
MMP2	132.0	100%	99%	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP20	90.5	100%	99%	Amelogenesis imperfecta, type IIA2, 612529
MMP21	97.7	89%	83%	Heterotaxy, visceral, 7, autosomal, 616749
MMP9	97.7	100%	98%	Metaphyseal anadysplasia 2, 613073
MN1	85.9	96%	93%	Meningioma, 607174
MNX1	28.9	73%	55%	Currarino syndrome, 176450
MOCOS	152.8	100%	98%	Xanthinuria, type II, 603592
MOCS1	76.9	93%	88%	Molybdenum cofactor deficiency A, 252150
MOCS2	128.2	99%	98%	Molybdenum cofactor deficiency B, 252160
MOGS	94.6	99%	98%	Congenital disorder of glycosylation, type IIb, 606056
MORC2	120.1	100%	99%	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688
MPC1	114.0	100%	98%	Mitochondrial pyruvate carrier deficiency,614741
MPDU1	112.5	100%	98%	Congenital disorder of glycosylation, type If, 609180
MPDZ	141.4	98%	95%	Hydrocephalus, nonsyndromic, autosomal recessive 2, 615219
MPI	108.9	100%	99%	Congenital disorder of glycosylation, type Ib, 602579
MPL	121.9	99%	93%	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498
MPLKIP	73.8	98%	74%	Trichothiodystrophy 4, nonphotosensitive, 234050
MPO	134.2	100%	99%	Myeloperoxidase deficiency, 254600 {Alzheimer disease, susceptibility to}, 104300 {Lung cancer, protection against, in smokers}
MPV17	101.4	100%	99%	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MPZ	92.9	100%	96%	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 Neuropathy, congenital hypomyelinating, 605253 Roussy-Levy syndrome, 180800
MRAP	137.4	100%	100%	Glucocorticoid deficiency 2, 607398
MRE11A	48.8	98%	85%	Ataxia-telangiectasia-like disorder, 604391
MRPL3	54.4	94%	82%	Combined oxidative phosphorylation deficiency 9, 614582

MRPS16	117.4	100%	100%	Combined oxidative phosphorylation deficiency 2, 610498
MRPS22	125.4	98%	89%	Combined oxidative phosphorylation deficiency 5, 611719
MS4A1	106.7	100%	93%	Immunodeficiency, common variable, 5, 613495
MSH2	91.8	95%	87%	Colorectal cancer, hereditary nonpolyposis, type 1, 120435 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MSH3	101.5	99%	94%	Endometrial carcinoma, somatic, 608089 Familial adenomatous polyposis 4,617100
MSH6	142.6	100%	99%	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 Endometrial cancer, familial, 608089 Mismatch repair cancer syndrome, 276300
MSMO1	35.0	85%	70%	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MSR1	173.3	100%	100%	Barrett esophagus/esophageal adenocarcinoma, 614266 Prostate cancer, hereditary, 176807
MSRB3	122.9	100%	95%	Deafness, autosomal recessive 74, 613718
MSTN	176.7	100%	98%	Muscle hypertrophy, 614160
MSX1	56.2	96%	91%	Ectodermal dysplasia 3, Witkop type, 189500 Orofacial cleft 5, 608874 Tooth agenesis, selective, 1, with or without orofacial cleft, 106600
MSX2	81.0	100%	84%	Craniosynostosis, type 2, 604757 Parietal foramina 1, 168500 Parietal foramina with cleidocranial dysplasia, 168550
MTAP	98.3	90%	84%	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250
MTFMT	122.3	98%	94%	Combined oxidative phosphorylation deficiency 15, 614947
MTHFR	117.3	100%	100%	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}
MTM1	115.2	99%	97%	Myotubular myopathy, X-linked, 310400
MTMR2	107.3	100%	100%	Charcot-Marie-Tooth disease, type 4B1, 601382
MTO1	140.9	89%	85%	Combined oxidative phosphorylation deficiency 10, 614702
MTOR	117.2	99%	99%	Smith-Kingsmore syndrome, 616638
MTPAP	108.7	97%	84%	Ataxia, spastic, 4, 613672

MTR	126.4	100%	99%	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTRR	114.9	99%	97%	Homocystinuria-megaloblastic anemia, cbl E type, 236270 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTTP	122.0	100%	96%	Abetalipoproteinemia, 200100 {Metabolic syndrome, protection against}, 605552
MUC1	88.7	97%	92%	Medullary cystic kidney disease 1, 174000
MUSK	146.2	100%	100%	Fetal akinesia deformation sequence, 208150 Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325
MUT	105.1	97%	91%	Methylmalonic aciduria, mut(0) type, 251000
MUTYH	126.5	100%	97%	Adenomas, multiple colorectal, 608456 Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600 Gastric cancer, somatic, 613659
MVD	95.0	100%	100%	Porokeratosis 7, multiple types, 614714
MVK	120.9	100%	100%	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900
MXI1	97.9	88%	83%	Neurofibrosarcoma {Prostate cancer, susceptibility to}, 176807
MYBPC1	145.0	100%	99%	Arthrogryposis, distal, type 1B, 614335 Lethal congenital contracture syndrome 4, 614915
MYBPC3	118.7	99%	94%	Cardiomyopathy, dilated, 1MM, 615396 Cardiomyopathy, hypertrophic, 4, 115197 Left ventricular noncompaction 10, 615396
MYC	151.5	100%	100%	Burkitt lymphoma, 113970
MYCN	80.1	99%	89%	Feingold syndrome, 164280
MYD88	145.1	100%	98%	Macroglobulinemia, Waldenstrom, somatic, 153600 Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260
MYF6	100.2	100%	100%	Myopathy, centronuclear, 3, 614408
MYH11	109.2	100%	98%	Aortic aneurysm, familial thoracic 4, 132900
MYH14	89.2	95%	85%	Deafness, autosomal dominant 4A, 600652 ?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369
MYH2	111.7	98%	96%	Proximal myopathy and ophthalmoplegia, 605637

MYH3	96.3	99%	96%	Arthrogryposis, distal, type 2A, 193700 Arthrogryposis, distal, type 2B, 601680 Arthrogryposis, distal, type 8, 178110
MYH6	99.9	99%	90%	Atrial septal defect 3, 614089 Cardiomyopathy, dilated, 1EE, 613252 Cardiomyopathy, hypertrophic, 14, 613251 {Sick sinus syndrome 3}, 614090
MYH7	98.1	97%	94%	Cardiomyopathy, dilated, 1S, 613426 Cardiomyopathy, hypertrophic, 1, 192600 Left ventricular noncompaction 5, 613426 Liang distal myopathy, 160500 Myopathy, myosin storage, autosomal dominant, 608358 Myopathy, myosin storage, autosomal recessive, 255160 Scapuloperoneal syndrome, myopathic type, 181430
MYH8	115.7	99%	98%	Carney complex variant, 608837 Trismus-pseudocamptodactyly syndrome, 158300
MYH9	115.9	99%	97%	Deafness, autosomal dominant 17, 603622 Epstein syndrome, 153650 Fechtner syndrome, 153640 Macrothrombocytopenia and progressive sensorineural deafness, 600208 May-Hegglin anomaly, 155100 Sebastian syndrome, 605249
MYL2	131.2	99%	94%	Cardiomyopathy, hypertrophic, 10, 608758
MYL3	101.8	100%	100%	Cardiomyopathy, hypertrophic, 8, 608751
MYLK	133.8	99%	99%	Aortic aneurysm, familial thoracic 7, 613780
MYLK2	93.5	100%	98%	Cardiomyopathy, hypertrophic, 1, digenic, 192600
MYO15A	96.8	95%	91%	Deafness, autosomal recessive 3, 600316
MYO18B	110.7	99%	97%	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549
MYO1E	118.8	97%	92%	Glomerulosclerosis, focal segmental, 6, 614131
MYO3A	107.9	96%	91%	Deafness, autosomal recessive 30, 607101
MYO5A	114.3	100%	98%	Griscelli syndrome, type 1, 214450
MYO5B	124.7	97%	96%	Microvillus inclusion disease, 251850
MYO6	78.6	95%	89%	Deafness, autosomal dominant 22, 606346 Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346 Deafness, autosomal recessive 37, 607821

MYO7A	120.1	98%	95%	Deafness, autosomal dominant 11, 601317 Deafness, autosomal recessive 2, 600060 Usher syndrome, type 1B, 276900
MYOC	141.5	100%	99%	Glaucoma 1A, primary open angle, 137750
MYOT	130.7	96%	88%	Muscular dystrophy, limb-girdle, type 1A, 159000 Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
MYOZ2	138.4	100%	100%	Cardiomyopathy, hypertrophic, 16, 613838
MYPN	129.7	99%	98%	Cardiomyopathy, dilated, 1KK, 615248 Cardiomyopathy, familial restrictive, 4, 615248 Cardiomyopathy, hypertrophic, 22, 615248
MYT1L	153.0	99%	99%	Mental retardation, autosomal dominant 39, 616521
NAA10	112.3	100%	97%	Ogden syndrome, 300855 ?Microphthalmia, syndromic 1, 309800
NAGA	118.4	100%	100%	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241
NAGLU	97.6	94%	91%	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491
NAGS	61.6	100%	92%	N-acetylglutamate synthase deficiency, 237310
NALCN	127.0	100%	97%	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419
NANOS1	32.3	93%	74%	Spermatogenic failure 12, 615413
NANS	102.3	100%	100%	Sponyloepimetaphyseal dysplasia, Genevieve type,610442
NARS2	127.1	97%	96%	Combined oxidative phosphorylation deficiency 24, 616239
NBAS	131.9	98%	96%	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NBEAL2	147.3	99%	99%	Gray platelet syndrome, 139090
NBN	74.3	96%	92%	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260
NCF1	14.4	25%	20%	Chronic granulomatous disease due to deficiency of NCF-1, 233700
NCF2	104.8	98%	96%	Chronic granulomatous disease due to deficiency of NCF-2, 233710
NCF4	138.6	100%	100%	Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III, 613960

NCSTN	107.4	100%	100%	Acne inversa, familial, 1, 142690
NDE1	84.3	100%	100%	Lissencephaly 4 (with microcephaly), 614019 ?Microhydranencephaly, 605013
NDN	71.3	81%	70%	Prader-Willi syndrome, 176270
NDP	152.6	100%	100%	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600
NDRG1	114.1	100%	100%	Charcot-Marie-Tooth disease, type 4D, 601455
NDST1	164.7	100%	100%	Mental retardation, autosomal recessive 46, 616116
NDUFA1	215.2	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFA11	76.6	100%	90%	Mitochondrial complex I deficiency, 252010
NDUFA12	129.1	100%	100%	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000
NDUFA2	112.5	100%	100%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFA9	120.0	99%	95%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFAF1	85.6	100%	99%	Mitochondrial complex I deficiency, 252010
NDUFAF2	52.4	82%	70%	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFAF3	96.5	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF4	85.6	99%	96%	Mitochondrial complex I deficiency, 252010
NDUFAF5	96.4	94%	93%	Mitochondrial complex 1 deficiency, 252010
NDUFAF6	73.7	99%	90%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFB11	109.9	94%	87%	Linear skin defects with multiple congenital anomalies 3, 300952
NDUFB3	12.3	62%	28%	Mitochondrial complex I deficiency, 252010
NDUFS1	122.4	100%	99%	Mitochondrial complex I deficiency, 252010
NDUFS2	94.0	99%	98%	Mitochondrial complex I deficiency, 252010
NDUFS3	117.1	90%	90%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
NDUFS4	141.6	100%	99%	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFS6	121.3	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFS7	113.0	100%	98%	Leigh syndrome, 256000
NDUFS8	102.1	100%	100%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFV1	133.9	100%	97%	Mitochondrial complex I deficiency, 252010
NDUFV2	58.2	83%	49%	Mitochondrial complex I deficiency, 252010
NEB	114.4	82%	81%	Nemaline myopathy 2, autosomal recessive, 256030

NEFH	83.7	98%	97%	Charcot-Marie-Tooth disease,axonal,type 2CC,616924 ?{Amyotrophic lateral sclerosis, susceptibility to}, 105400
NEFL	127.7	99%	96%	Charcot-Marie-Tooth disease, type 1F, 607734 Charcot-Marie-Tooth disease, type 2E, 607684
NEK1	99.1	97%	91%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NEK9	123.7	99%	96%	Lethal congenital contracture syndrome 10, 617022 Nevus comedonicus, somatic, 617025 ?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262
NEU1	12.2	61%	15%	Sialidosis, type I, 256550 Sialidosis, type II, 256550
NEUROD1	139.9	100%	100%	Maturity-onset diabetes of the young 6, 606394 {Diabetes mellitus, noninsulin-dependent}, 125853
NEUROG3	85.5	100%	95%	Diarrhea 4, malabsorptive, congenital, 610370
NEXN	65.0	88%	76%	Cardiomyopathy, dilated, 1CC, 613122 Cardiomyopathy, hypertrophic, 20, 613876
NF1	110.4	92%	87%	Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321 Watson syndrome, 193520
NF2	89.6	100%	99%	Meningioma, NF2-related, somatic, 607174 Neurofibromatosis, type 2, 101000 Schwannomatosis, 162091
NFIX	127.6	98%	97%	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753
NFKB1	98.2	99%	90%	Immunodeficiency, common variable, 12, 616576
NFKB2	109.8	96%	91%	Immunodeficiency, common variable, 10, 615577
NFKBIA	94.6	94%	91%	Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency, 612132
NFU1	43.5	98%	81%	Multiple mitochondrial dysfunctions syndrome 1, 605711
NGF	218.4	100%	100%	Neuropathy, hereditary sensory and autonomic, type V, 608654
NGLY1	117.1	100%	99%	Congenital disorder of deglycosylation, 615273
NHEJ1	67.8	100%	100%	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
NHLRC1	126.0	100%	100%	Epilepsy, progressive myoclonic 2B (Lafora), 254780

NHP2	71.0	100%	99%	Dyskeratosis congenita, autosomal recessive 2, 613987
NHS	144.9	95%	93%	Cataract 40, X-linked, 302200 Nance-Horan syndrome, 302350
NIN	139.0	99%	97%	Seckel syndrome 7, 614851
NIPA1	135.0	100%	97%	Spastic paraplegia 6, autosomal dominant, 600363
NIPAL4	129.5	100%	97%	Ichthyosis, congenital, autosomal recessive 6, 612281
NIPBL	99.7	96%	95%	Cornelia de Lange syndrome 1, 122470
NKX2-1	39.2	100%	91%	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 {Thyroid cancer, monmedullary, 1}, 188550
NKX2-5	76.5	99%	99%	Atrial septal defect 7, with or without AV conduction defects, 108900 Conotruncal heart malformations, variable, 217095 Hypoplastic left heart syndrome 2, 614435 Hypothyroidism, congenital nongoitrous, 5, 225250 Tetrology of Fallot, 187500 Ventricular septal defect 3, 614432
NKX2-6	90.1	100%	99%	Conotruncal heart malformations, 217095 Persistent truncus arteriosus, 217095
NKX3-2	42.0	79%	55%	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330
NLGN4X	188.3	100%	97%	Mental retardation, X-linked, 300495 {Asperger syndrome susceptibility, X-linked 2}, 300497 {Autism susceptibility, X-linked 2}, 300495
NLRC4	146.4	100%	99%	Autoinflammation with infantile enterocolitis, 616050 ?Familial cold autoinflammatory syndrome 4, 616115
NLRP12	131.0	99%	99%	Familial cold autoinflammatory syndrome 2, 611762
NLRP3	128.6	100%	99%	CINCA syndrome, 607115 Familial cold-induced inflammatory syndrome 1, 120100 Muckle-Wells syndrome, 191900
NLRP7	120.9	99%	97%	Hydatidiform mole,recurrent, 1,231090
NME1	80.5	100%	96%	Neuroblastoma, 256700
NME8	95.6	96%	90%	Ciliary dyskinesia, primary, 6, 610852
NMNAT1	114.7	100%	96%	Leber congenital amaurosis 9, 608553
NNT	127.7	97%	96%	Glucocorticoid deficiency 4, 614736
NOBOX	67.3	100%	95%	Premature ovarian failure 5, 611548

NOD2	122.6	100%	99%	Blau syndrome, 186580 Sarcoidosis, early-onset, 609464 {Inflammatory bowel disease 1}, 266600 {Psoriatic arthritis, susceptibility to}, 607507
NODAL	131.2	100%	100%	Heterotaxy, visceral, 5, 270100
NOG	148.2	100%	100%	Brachydactyly, type B2, 611377 Multiple synostoses syndrome 1, 186500 Stapes ankylosis with broad thumb and toes, 184460 Symphalangism, proximal, 1A, 185800 Tarsal-carpal coalition syndrome, 186570
NOL3	71.2	91%	80%	Myoclonus, familial cortical, 614937
NONO	110.2	100%	98%	Mental retardation, X-linked, syndromic 34, 300967
NOP10	148.8	100%	100%	Dyskeratosis congenita, autosomal recessive 1, 224230
NOP56	111.5	98%	95%	Spinocerebellar ataxia 36, 614153
NOTCH1	125.3	99%	97%	Adams-Oliver syndrome 5, 616028 Aortic valve disease 1, 109730
NOTCH2	150.5	100%	99%	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NOTCH3	98.3	94%	89%	Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310 Lateral meningocele syndrome, 130720 ?Myofibromatosis, infantile 2, 615293
NPC1	126.2	99%	97%	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220 {Nasopharyngeal carcinoma 1}
NPC2	121.9	100%	100%	Niemann-pick disease, type C2, 607625
NPHP1	118.5	98%	97%	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NPHP3	105.0	97%	91%	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540
NPHP4	122.1	100%	99%	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
NPHS1	90.7	99%	96%	Nephrotic syndrome, type 1, 256300

NPHS2	93.3	100%	98%	Nephrotic syndrome, type 2, 600995
NPM1	63.2	88%	77%	Leukemia, acute myeloid, somatic, 601626
NPPA	89.2	100%	99%	Atrial fibrillation, familial, 6, 612201 Atrial standstill 2, 615745
NPR2	148.3	100%	100%	Acromesomelic dysplasia, Maroteaux type, 602875 Epiphyseal chondrodysplasia, Miura type, 615923 Short stature with nonspecific skeletal abnormalities, 616255
NR0B1	131.4	100%	100%	46XY sex reversal 2, dosage-sensitive, 300018 Adrenal hypoplasia, congenital, with hypogonadotropic hypogonadism, 300200
NR0B2	78.4	100%	100%	Obesity, mild, early-onset, 601665
NR1H4	138.6	96%	89%	Cholestasis, progressive familial intrahepatic 5, 617049
NR2E3	84.7	100%	100%	Enhanced S-cone syndrome, 268100 Retinitis pigmentosa 37, 611131
NR2F1	164.3	100%	100%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NR2F2	170.2	94%	91%	Congenital heart defects, multiple types, 4, 615779
NR3C1	134.1	100%	99%	Glucocorticoid resistance, 615962
NR3C2	149.9	99%	95%	Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115 Pseudohypoaldosteronism type I, autosomal dominant, 177735
NR4A3	88.5	99%	87%	Chondrosarcoma,extraskeletal myxoid,612237
NR5A1	74.8	100%	99%	46XY sex reversal 3, 612965 Adrenocortical insufficiency Premature ovarian failure 7, 612964 Spermatogenic failure 8, 613957
NRAS	175.3	100%	100%	Colorectal cancer, somatic, 114500 Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Thyroid carcinoma, follicular, somatic, 188470 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470
NRL	61.3	99%	85%	Retinal degeneration, autosomal recessive, clumped pigment type Retinitis pigmentosa 27, 613750
NRXN1	142.9	98%	97%	Pitt-Hopkins-like syndrome 2, 614325

				{Schizophrenia, susceptibility to, 17}, 614332
NSD1	139.4	100%	100%	Beckwith-Wiedemann syndrome, 130650 Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550
NSDHL	204.8	100%	100%	CHILD syndrome, 308050 CK syndrome, 300831
NSMF	91.7	96%	94%	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
NSUN2	98.8	94%	87%	Mental retardation, autosomal recessive 5, 611091
NT5C2	126.3	96%	94%	Spastic paraplegia 45, autosomal recessive, 613162
NT5C3A	57.0	84%	66%	Anemia, hemolytic, due to UMPH1 deficiency, 266120
NT5E	150.7	100%	98%	Calcification of joints and arteries, 211800
NTF4	49.9	92%	81%	Glaucoma 1, open angle, 1O, 613100
NTHL1	99.3	96%	93%	Familial adenomatous polyposis 3, 616415
NTRK1	111.1	100%	97%	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma, familial, 155240
NUBPL	81.5	90%	82%	Mitochondrial complex I deficiency, 252010
NUMA1	103.7	100%	99%	Leukemia, acute promyelocytic, somatic, 612376
NUP107	104.8	98%	90%	Nephrotic syndrome, type 11, 616730
NUP214	139.0	100%	99%	Leukemia, acute myeloid, somatic, 601626 Leukemia, T-cell acute lymphoblastic, somatic, 613065
NUP62	109.3	100%	100%	Striatonigral degeneration, infantile, 271930
NUP93	121.0	96%	93%	Nephrotic syndrome, type 12,616892
NYX	86.7	97%	96%	Night blindness, congenital stationary (complete), 1A, X-linked, 310500
OAT	73.4	70%	65%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OBSL1	115.8	98%	95%	3-M syndrome 2, 612921
OCA2	120.2	97%	96%	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	218.4	100%	100%	Band-like calcification with simplified gyration and polymicrogyria, 251290
OCRL	164.3	100%	99%	Dent disease 2, 300555 Lowe syndrome, 309000

OFD1	57.7	88%	78%	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424
OGG1	121.4	100%	100%	Renal cell carcinoma, clear cell, somatic, 144700
OPA1	113.8	97%	92%	Behr syndrome,210000 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type),616896 {Glaucoma, normal tension, susceptibility to}, 606657
OPA3	90.6	97%	94%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPHN1	117.6	100%	99%	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
OPLAH	94.3	100%	97%	5-oxoprolinase deficiency, 260005
OPN1LW	78.3	71%	66%	Blue cone monochromacy, 303700 Colorblindness, protan, 303900
OPN1MW	40.7	66%	62%	Blue cone monochromacy, 303700 Colorblindness, deutan, 303800
OPN1SW	103.4	100%	100%	Colorblindness, tritan, 190900
OPTN	95.5	100%	99%	Amyotrophic lateral sclerosis 12, 613435 Glaucoma 1, open angle, E, 137760 {Glaucoma, normal tension, susceptibility to}, 606657
ORAI1	175.4	93%	89%	Immunodeficiency 9, 612782 Myopathy, tubular aggregate, 2, 615883
ORC1	95.7	98%	93%	Meier-Gorlin syndrome 1, 224690
ORC4	59.1	95%	85%	Meier-Gorlin syndrome 2, 613800
ORC6	122.8	100%	100%	Meier-Gorlin syndrome 3, 613803
OSBPL2	130.3	100%	100%	Deafness, autosomal dominant 67, 616340
OSMR	120.9	100%	99%	Amyloidosis, primary localized cutaneous, 1, 105250
OSTM1	53.7	90%	83%	Osteopetrosis, autosomal recessive 5, 259720
OTC	139.1	100%	100%	Ornithine transcarbamylase deficiency, 311250
OTOA	99.5	98%	95%	Deafness, autosomal recessive 22, 607039
OTOF	116.6	99%	98%	Auditory neuropathy, autosomal recessive, 1, 601071 Deafness, autosomal recessive 9, 601071

OTOG	119.0	99%	97%	Deafness, autosomal recessive 18B, 614945
OTOGL	110.8	98%	93%	Deafness, autosomal recessive 84B, 614944
OTX2	119.3	100%	98%	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125
OVOL2	105.3	100%	97%	Corneal dystrophy, posterior polymorphous, 1, 122000
OXCT1	106.0	100%	96%	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050
P2RX1	104.9	99%	97%	Bleeding disorder due to P2RX1 defect, somatic, 609821
P2RX2	108.6	100%	97%	Deafness, autosomal dominant 41, 608224
P2RY12	187.5	100%	100%	Bleeding disorder, platelet-type, 8, 609821
P4HB	102.1	94%	94%	Cole-Carpenter syndrome 1, 112240
PABPN1	70.4	65%	58%	Oculopharyngeal muscular dystrophy, 164300
PACS1	102.2	97%	95%	Schuss-Hoeijmakers-syndrome, 615009
PAFAH1B1	93.5	88%	78%	Lissencephaly 1, 607432 Subcortical laminar heterotopia, 607432
PAH	153.3	100%	100%	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600
PAK3	104.5	97%	92%	Mental retardation, X-linked 30/47, 300558
PALB2	149.2	100%	100%	Fanconi anemia, complementation group N, 610832 {Breast cancer, susceptibility to}, 114480 {Pancreatic cancer, susceptibility to, 3}, 613348
PAM16	43.8	65%	64%	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320
PANK2	147.7	98%	93%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PAPSS2	86.8	100%	98%	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847
PARK2	109.5	100%	100%	Adenocarcinoma of lung, somatic, 211980 Adenocarcinoma, ovarian, somatic, 167000 Parkinson disease, juvenile, type 2, 600116 {Leprosy, susceptibility to}, 607572
PARK7	74.2	100%	97%	Parkinson disease 7,autosomal recessive early-onset,606324
PARN	114.7	100%	96%	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
PAX2	149.3	100%	100%	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330

PAX3	100.8	100%	99%	Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220 Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820
PAX4	83.3	100%	98%	Diabetes mellitus, type 2, 125853 Maturity-onset diabetes of the young, type IX, 612225 {Diabetes mellitus, ketosis-prone, susceptibility to}, 612227
PAX6	123.5	100%	100%	Aniridia, 106210 Cataract with late-onset corneal dystrophy, 106210 Coloboma of optic nerve, 120430 Coloboma, ocular, 120200 Foveal hypoplasia 1, 136520 Keratitis, 148190 Optic nerve hypoplasia, 165550 Peters anomaly, 604229 ?Morning glory disc anomaly, 120430
PAX7	93.8	100%	99%	Rhabdomyosarcoma 2, alveolar, 268220
PAX8	89.5	100%	99%	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700
PAX9	201.1	99%	95%	Tooth agenesis, selective, 3, 604625
PC	126.0	97%	93%	Pyruvate carboxylase deficiency, 266150
PCBD1	99.4	99%	99%	Hyperphenylalaninemia, BH4-deficient, D, 264070
PCCA	91.1	95%	88%	Propionicacidemia, 606054
PCCB	124.7	96%	94%	Propionicacidemia, 606054
PCDH15	146.8	99%	99%	Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1F, 602083
PCDH19	215.0	100%	100%	Epileptic encephalopathy, early infantile, 9, 300088
PCNT	99.7	97%	94%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PCSK1	130.7	100%	98%	Obesity with impaired prohormone processing, 600955 {Obesity, susceptibility to, BMIQ12}, 612362
PCSK9	87.0	93%	91%	Hypercholesterolemia, familial, 3, 603776 {Low density lipoprotein cholesterol level QTL 1}, 603776
PCYT1A	92.9	99%	95%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDCD10	77.7	98%	74%	Cerebral cavernous malformations 3, 603285

PDE10A	133.5	100%	99%	Dyskinesia,limb and orofacial,infantile-onset,616921 Striatal degeneration,autosomal dominant,616922
PDE11A	138.7	100%	100%	Pigmented nodular adrenocortical disease, primary, 2, 610475
PDE3A	104.0	99%	98%	Hypertension and brachydactyly syndrome, 112410
PDE4D	101.0	98%	94%	Acrodysostosis 2, with or without hormone resistance, 614613 {Stroke, susceptibility to, 1}, 606799
PDE6A	104.9	100%	100%	Retinitis pigmentosa 43, 613810
PDE6B	133.6	100%	100%	Night blindness, congenital stationary, autosomal dominant 2, 163500 Retinitis pigmentosa-40, 613801
PDE6C	124.1	97%	96%	Cone dystrophy 4, 613093
PDE6G	77.3	96%	92%	Retinitis pigmentosa 57, 613582
PDE6H	51.2	74%	48%	Achromatopsia 6, 610024 Retinal cone dystrophy 3, 610024
PDE8B	99.4	100%	99%	Pigmented nodular adrenocortical disease, primary, 3, 614190 Striatal degeneration, autosomal dominant, 609161
PDGFB	90.8	100%	100%	Basal ganglia calcification, idiopathic, 5, 615483 Dermatofibrosarcoma protuberans, 607907 Meningioma, SIS-related, 607174
PDGFRA	135.8	100%	99%	Gastrointestinal stromal tumor, somatic, 606764 Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685
PDGFRB	131.1	98%	95%	Basal ganglia calcification, idiopathic, 4, 615007 Kosaki overgrowth syndrome, 616592 Myeloproliferative disorder with eosinophilia, 131440 Myofibromatosis, infantile, 1, 228550 Premature aging syndrome, Penttinen type, 601812
PDGFRL	122.3	100%	98%	Colorectal cancer, somatic, 114500 Hepatocellular cancer, somatic, 114550
PDHA1	125.5	99%	92%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	118.6	97%	94%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	112.0	97%	95%	Lacticacidemia due to PDX1 deficiency,245349
PDP1	148.8	100%	100%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	104.5	89%	82%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	99.7	94%	94%	Coenzyme Q10 deficiency, primary, 3, 614652

PDX1	20.5	69%	46%	MODY, type IV,606392 Pancreatic agenesis 1, 260370 {Diabetes mellitus, type II, susceptibility to},125853
PDYN	100.0	100%	100%	Spinocerebellar ataxia 23, 610245
PDZD7	84.8	100%	97%	Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472 {Retinal disease in Usher syndrome type IIA, modifier of}, 276901
PEPD	99.1	100%	100%	Prolidase deficiency, 170100
PER2	88.0	100%	99%	Advanced sleep phase syndrome, familial, 1, 604348
PET100	114.0	84%	72%	Mitochondrial complex IV deficiency, 220110
PEX1	101.0	98%	97%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	99.1	96%	90%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX11B	94.0	100%	100%	Peroxisome biogenesis disorder 14B, 614920
PEX12	113.8	99%	98%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	172.4	98%	95%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	116.0	99%	96%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	112.6	99%	93%	Peroxisome biogenesis disorder 8A, (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	98.2	100%	99%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	134.9	100%	100%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	63.5	100%	98%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	83.4	95%	92%	Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	92.3	100%	96%	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	68.5	90%	81%	Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863

PEX7	114.5	89%	84%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PFKM	140.3	100%	99%	Glycogen storage disease VII, 232800
PFN1	139.1	100%	100%	Amyotrophic lateral sclerosis 18, 614808
PGAM2	122.8	100%	100%	Glycogen storage disease X, 261670
PGAP1	90.9	91%	88%	Mental retardation, autosomal recessive 42, 615802
PGAP2	134.1	100%	99%	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGAP3	109.3	96%	92%	Hyperphosphatasia with mental retardation syndrome 4, 615716
PGK1	60.9	90%	84%	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	119.6	100%	100%	Congenital disorder of glycosylation, type It, 614921
PGM3	171.0	100%	100%	Immunodeficiency 23, 615816
PHEX	153.1	98%	96%	Hypophosphatemic rickets, X-linked dominant, 307800
PHF6	79.4	91%	91%	Borjeson-Forssman-Lehmann syndrome, 301900
PHF8	112.2	100%	99%	Mental retardation syndrome, X-linked, Siderius type, 300263
PHGDH	105.9	100%	99%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHKA1	134.7	99%	97%	Muscle glycogenosis, 300559
PHKA2	130.2	100%	100%	Glycogen storage disease, type IXa1, 306000 Glycogen storage disease, type IXa2, 306000
PHKB	126.0	100%	100%	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750
PHKG2	132.3	100%	100%	Cirrhosis due to liver phosphorylase kinase deficiency Glycogen storage disease IXc, 613027
PHOX2A	33.9	49%	22%	Fibrosis of extraocular muscles, congenital, 2, 602078
PHOX2B	94.3	93%	87%	Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, 209880 Neuroblastoma with Hirschsprung disease, 613013 {Neuroblastoma, susceptibility to, 2}, 613013
PHYH	63.6	97%	88%	Refsum disease, 266500
PI4KA	99.2	91%	83%	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531
PICALM	99.7	100%	93%	Leukemia, acute myeloid, somatic, 601626
PIEZO1	128.6	99%	97%	Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380 Lymphedema, hereditary, III, 616843
PIEZO2	112.2	99%	98%	Arthrogryposis, distal, type 3, 114300 Arthrogryposis, distal, type 5, 108145

				?Marden-Walker syndrome, 248700
PIGA	104.0	98%	87%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGG	151.5	100%	96%	Mental retardation,autosomal recessive 53,616917
PIGL	111.8	100%	100%	CHIME syndrome, 280000
PIGM	128.6	100%	100%	Glycosylphosphatidylinositol deficiency, 610293
PIGN	104.4	95%	86%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	103.3	100%	99%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGT	127.6	100%	97%	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 ?Paroxysmal nocturnal hemoglobinuria 2, 615399
PIGV	144.4	100%	100%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIGY	86.2	100%	99%	Hyperphosphatasia with mental retardation syndrome 6, 616809
PIK3CA	109.9	99%	97%	Breast cancer, somatic, 114480 CLOVE syndrome, somatic, 612918 Colorectal cancer, somatic, 114500 Cowden syndrome 5, 615108 Gastric cancer, somatic, 613659 Hepatocellular carcinoma, somatic, 114550 Keratosis, seborrheic, somatic, 182000 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Nevus, epidermal, somatic, 162900 Nonsmall cell lung cancer, somatic, 211980 Ovarian cancer, somatic, 167000
PIK3CD	112.7	98%	95%	Immunodeficiency 14, 615513
PIK3R1	120.6	100%	97%	Immunodeficiency 36, 616005 SHORT syndrome, 269880 ?Agammaglobulinemia 7, autosomal recessive, 615214
PIK3R2	80.1	88%	86%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PIK3R5	99.6	99%	98%	Ataxia-oculomotor apraxia 3, 615217
PIKFYVE	128.3	99%	93%	Corneal fleck dystrophy, 121850
PINK1	86.3	93%	89%	Parkinson disease 6, early onset, 605909
PIP5K1C	106.0	97%	93%	Lethal congenital contractural syndrome 3, 611369
PITPNM3	93.0	98%	95%	Cone-rod dystrophy 5, 600977

PITX1	122.9	92%	88%	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800 Liebenberg syndrome, 186550
PITX2	122.4	98%	95%	Axenfeld-Rieger syndrome, type 1, 180500 Iridogoniodysgenesis, type 2, 137600 Peters anomaly, 604229 Ring dermoid of cornea, 180550
PITX3	51.9	100%	95%	Anterior segment mesenchymal dysgenesis, 107250 Cataract 11, multiple types, 610623 Cataract 11, syndromic, 610623
PKD1	22.7	40%	29%	Polycystic kidney disease, adult type I, 173900
PKD2	96.7	90%	86%	Polycystic kidney disease 2, 613095
PKHD1	138.4	100%	99%	Polycystic kidney and hepatic disease, 263200
PKLR	142.0	100%	99%	Adenosine triphosphate, elevated, of erythrocytes, 102900 Pyruvate kinase deficiency, 266200
PKP1	107.5	100%	98%	Ectodermal dysplasia/skin fragility syndrome, 604536
PKP2	88.1	96%	89%	Arrhythmogenic right ventricular dysplasia 9, 609040
PLA2G4A	125.3	100%	99%	Phospholipase A2, group IV A, deficiency of, 600522
PLA2G6	99.6	99%	97%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953
PLA2G7	121.1	100%	96%	Platelet-activating factor acetylhydrolase deficiency, 614278 {Asthma, susceptibility to}, 600807 {Atopy, susceptibility to}, 147050
PLAG1	194.8	100%	100%	Adenomas, salivary gland pleomorphic, somatic, 181030
PLAU	102.6	100%	95%	Quebec platelet disorder, 601709 {Alzheimer disease, late-onset, susceptibility to}, 104300
PLCB1	138.5	100%	99%	Epileptic encephalopathy, early infantile, 12, 613722
PLCB4	123.4	99%	93%	Auriculocondylar syndrome 2, 614669
PLCD1	103.2	99%	95%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCE1	134.0	99%	98%	Nephrotic syndrome, type 3, 610725
PLCG2	102.0	99%	98%	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468

PLEC	92.8	99%	98%	Epidermolysis bullosa simplex with muscular dystrophy, 226670 Epidermolysis bullosa simplex with pyloric atresia, 612138 Epidermolysis bullosa simplex, Ogna type, 131950 Muscular dystrophy, limb-girdle, type 2Q, 613723 ?Epidermolysis bullosa simplex with nail dystrophy, 616487
PLEKHG2	105.7	100%	99%	Leukodystrophy and acquired microcephaly with or without dystonia, 616763
PLEKHG5	77.0	93%	86%	Charcot-Marie-Tooth disease, recessive intermediate C, 615376 Spinal muscular atrophy, distal, autosomal recessive, 4, 611067
PLEKHM1	51.0	100%	94%	Osteopetrosis, autosomal recessive 6, 611497
PLG	102.8	87%	85%	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PLIN1	74.6	97%	84%	Lipodystrophy, familial partial, type 4, 613877
PLK4	131.0	98%	95%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLN	188.8	100%	100%	Cardiomyopathy, dilated, 1P, 609909 Cardiomyopathy, hypertrophic, 18, 613874
PLOD1	119.0	99%	97%	Ehlers-Danlos syndrome, type VI, 225400
PLOD2	102.5	98%	90%	Bruck syndrome 2, 609220
PLOD3	101.8	100%	99%	Lysyl hydroxylase 3 deficiency, 612394
PLP1	160.9	100%	100%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PLS3	181.2	100%	100%	Bone mineral density QTL18, osteoporosis, 300910
PML	96.4	100%	99%	Leukemia, acute promyelocytic, PML/RARA type
PMM2	153.5	100%	100%	Congenital disorder of glycosylation, type Ia, 212065
PMP22	91.6	90%	88%	Charcot-Marie-Tooth disease, type 1A, 118220 Charcot-Marie-Tooth disease, type 1E, 118300 Dejerine-Sottas disease, 145900 Neuropathy, inflammatory demyelinating, 139393 Neuropathy, recurrent, with pressure palsies, 162500 Roussy-Levy syndrome, 180800
PMPCA	111.3	94%	91%	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMS2	76.8	83%	81%	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 Mismatch repair cancer syndrome, 276300
PMVK	104.6	100%	96%	Porokeratosis 1, multiple types, 175800
PNKD	90.2	100%	96%	Paroxysmal nonkinesigenic dyskinesia,118800

PNKP	81.0	100%	96%	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNP	122.8	100%	100%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA1	146.1	99%	98%	Ichthyosis, congenital, autosomal recessive 10, 615024
PNPLA2	101.3	100%	96%	Neutral lipid storage disease with myopathy, 610717
PNPLA6	112.6	99%	98%	Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 Spastic paraplegia 39, autosomal recessive, 612020 ?Laurence-Moon syndrome, 245800
PNPO	63.0	100%	96%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
PNPT1	46.4	93%	81%	Combined oxidative phosphorylation deficiency 13, 614932 Deafness, autosomal recessive 70, 614934
POC1A	112.9	100%	100%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POC1B	81.2	96%	91%	Cone-rod dystrophy 20, 615973
POF1B	100.9	93%	88%	Premature ovarian failure 2B,300604
POFUT1	118.6	100%	100%	Dowling-Degos disease 2, 615327
POGLUT1	121.5	100%	91%	Dowling-Degos disease 4, 615696
POGZ	141.2	99%	97%	White-Sutton syndrome, 616364
POLA1	147.9	100%	98%	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220
POLD1	88.8	95%	90%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381 {Colorectal cancer, susceptibility to, 10}, 612591
POLE	129.0	99%	98%	FILS syndrome, 615139 {Colorectal cancer, susceptibility to, 12}, 615083
POLG	100.7	100%	100%	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
POLG2	143.0	99%	97%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131
POLH	135.1	100%	98%	Xeroderma pigmentosum, variant type, 278750
POLR1A	108.0	99%	97%	Acrofacial dysostosis, Cincinnati type, 616462
POLR1C	108.2	99%	95%	Leukodystrophy, hypomyelinating, 11, 616494 Treacher Collins syndrome 3, 248390
POLR1D	162.2	100%	100%	Treacher Collins syndrome 2, 613717

POLR3A	126.0	100%	99%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	135.8	100%	97%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMC	84.6	100%	100%	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734 {Obesity, early-onset, susceptibility to}, 601665
POMGNT1	100.3	98%	92%	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
POMGNT2	216.4	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830
POMK	163.3	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249 ?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094
POMP	118.1	86%	85%	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952
POMT1	143.7	98%	95%	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	96.3	97%	95%	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
POR	126.9	100%	100%	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571
PORCN	142.7	100%	100%	Focal dermal hypoplasia, 305600
POU1F1	107.3	99%	90%	Pituitary hormone deficiency, combined, 1, 613038
POU3F4	141.1	100%	100%	Deafness, X-linked 2, 304400
POU4F3	204.9	100%	100%	Deafness, autosomal dominant 15, 602459
PPARG	132.1	100%	100%	Carotid intimal medial thickness 1, 609338 Insulin resistance, severe, digenic, 604367 Lipodystrophy, familial partial, type 3, 604367 Obesity, severe, 601665 [Obesity, resistance to] {Diabetes, type 2}, 125853
PPIB	102.0	100%	100%	Osteogenesis imperfecta, type IX, 259440
PPM1D	133.2	100%	97%	Breast cancer, 114480
PPOX	89.5	98%	96%	Porphyria variegata, 176200

PPP1R15B	120.5	99%	97%	Microcephaly, short stature, and impaired glucose metabolism 2, 616817
PPP1R3A	135.7	99%	97%	Insulin resistance, severe, digenic, 604367
PPP2R1A	119.7	93%	91%	Mental retardation, autosomal dominant 36, 616362
PPP2R1B	152.0	100%	100%	Lung cancer, 211980
PPP2R2B	136.1	100%	99%	Spinocerebellar ataxia 12, 604326
PPP2R5D	129.8	100%	100%	Mental retardation, autosomal dominant 35, 616355
PPT1	153.9	100%	100%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	181.6	97%	97%	Renpenning syndrome, 309500
PRCC	108.6	98%	90%	Renal cell carcinoma, papillary, 605074
PRCD	86.5	100%	100%	Retinitis pigmentosa 36, 610599
PRDM12	92.4	88%	80%	Neuropathy, hereditary sensory and autonomic, type VIII, 616488
PRDM16	136.9	99%	98%	Cardiomyopathy, dilated, 1LL, 615373 Left ventricular noncompaction 8, 615373
PRDM5	113.6	97%	91%	Brittle cornea syndrome 2, 614170
PRDM6	78.4	81%	73%	Patent ductus arteriosus 3, 617039
PRF1	102.5	100%	98%	Aplastic anemia, 609135 Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027
PRG4	123.9	95%	83%	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250
PRICKLE1	109.8	100%	100%	Epilepsy, progressive myoclonic 1B, 612437
PRIMPOL	98.1	94%	90%	Myopia 22,autosomal dominant,615420
PRKAG2	122.6	97%	86%	Cardiomyopathy, hypertrophic 6, 600858 Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200
PRKAR1A	82.3	100%	91%	Acrodysostosis 1, with or without hormone resistance, 101800 Adrenocortical tumor, somatic, Carney complex, type 1, 160980 Myxoma, intracardiac, 255960 Pigmented nodular adrenocortical disease, primary, 1, 610489
PRKCA	153.3	100%	100%	Pituitary tumor, invasive
PRKCD	164.2	100%	100%	Autoimmune lymphoproliferative syndrome, type III, 615559
PRKCG	101.6	97%	92%	Spinocerebellar ataxia 14, 605361
PRKCSH	113.8	99%	87%	Polycystic liver disease, 174050
PRKDC	94.7	97%	92%	Immunodeficiency 26, with or without neurologic abnormalities, 615966

PRKG1	105.8	99%	86%	Aortic aneurysm, familial thoracic 8, 615436
PRKRA	127.6	100%	99%	Dystonia 16, 612067
PRLR	130.5	100%	100%	Multiple fibroadenomas of the breast, 615554 ?Hyperprolactinemia, 615555
PRNP	140.1	100%	100%	Cerebral amyloid angiopathy, PRNP-related, 137440 Creutzfeldt-Jakob disease, 123400 Gerstmann-Straussler disease, 137440 Huntington disease-like 1, 603218 Insomnia, fatal familial, 600072 Prion disease with protracted course, 606688 {Kuru, susceptibility to}, 245300
PROC	111.4	99%	94%	Thrombophilia due to protein C deficiency, autosomal dominant, 176860 Thrombophilia due to protein C deficiency, autosomal recessive, 612304
PRODH	102.3	84%	82%	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850
PROK2	74.9	97%	89%	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	284.6	100%	100%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROM1	111.1	96%	92%	Cone-rod dystrophy 12, 612657 Macular dystrophy, retinal, 2, 608051 Retinitis pigmentosa 41, 612095 Stargardt disease 4, 603786
PROP1	92.5	91%	87%	Pituitary hormone deficiency, combined, 2, 262600
PROS1	68.4	96%	91%	Thrombophilia due to protein S deficiency, autosomal dominant, 612336 Thrombophilia due to protein S deficiency, autosomal recessive, 614514
PRPF3	74.7	97%	92%	Retinitis pigmentosa 18, 601414
PRPF31	99.2	100%	94%	Retinitis pigmentosa 11, 600138
PRPF4	137.2	100%	97%	Retinitis pigmentosa 70, 615922
PRPF6	114.3	100%	100%	Retinitis pigmentosa 60, 613983
PRPF8	121.5	99%	97%	Retinitis pigmentosa 13, 600059
PRPH2	202.7	100%	100%	Choriodal dystrophy, central areolar 2, 613105 Leber congenital amaurosis 18, 608133 Macular dystrophy, patterned, 1, 169150 Macular dystrophy, vitelliform, 3, 608161 Retinitis pigmentosa 7 and digenic, 608133

				Retinitis punctata albescens, 136880
PRPS1	200.6	100%	100%	Arts syndrome, 301835 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Deafness, X-linked 1, 304500 Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661
PRRT2	66.3	97%	96%	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751
PRRX1	75.6	100%	100%	Agnathia-otocephaly complex, 202650
PRSS1	166.1	100%	100%	Pancreatitis, hereditary, 167800 Trypsinogen deficiency, 614044
PRSS12	122.2	99%	96%	Mental retardation, autosomal recessive 1, 249500
PRSS56	45.7	92%	81%	Microphthalmia, isolated 6, 613517
PRX	86.4	100%	99%	Charcot-Marie-Tooth disease, type 4F, 614895 Dejerine-Sottas disease, 145900
PSAP	96.8	99%	95%	Combined SAP deficiency, 611721 Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PSAT1	40.7	80%	70%	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992
PSEN1	129.3	100%	99%	Acne inversa, familial, 3, 613737 Alzheimer disease, type 3, 607822 Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 Cardiomyopathy, dilated, 1U, 613694 Dementia, frontotemporal, 600274 Pick disease, 172700
PSEN2	114.1	100%	100%	Alzheimer disease-4, 606889

				Cardiomyopathy, dilated, 1V, 613697
PSENEN	60.7	100%	100%	Acne inversa, familial, 2, 613736
PSMB8	15.1	45%	32%	Autoinflammation, lipodystrophy, and dermatosis syndrome, 256040
PSMC3IP	98.4	100%	94%	Ovarian dysgenesis 3, 614324
PSPH	109.0	100%	96%	Phosphoserine phosphatase deficiency, 614023
PSTPIP1	78.4	100%	91%	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416
PTCH1	109.5	98%	95%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly-7, 610828
PTCH2	95.5	98%	95%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Medulloblastoma, 155255
PTDSS1	121.7	100%	100%	Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	120.7	100%	99%	Bannayan-Riley-Ruvalcaba syndrome, 153480 Cowden syndrome 1, 158350 Endometrial carcinoma, somatic, 608089 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309 Malignant melanoma, somatic, 155600 PTEN hamartoma tumor syndrome Squamous cell carcinoma, head and neck, somatic, 275355 VATER association with macrocephaly and ventriculomegaly, 276950 {Glioma susceptibility 2}, 613028 {Meningioma}, 607174 {Prostate cancer, somatic}, 176807
PTF1A	67.3	83%	74%	Pancreatic agenesis 2, 615935 Pancreatic and cerebellar agenesis, 609069
PTGIS	104.9	97%	95%	Hypertension, essential, 145500
PTH	105.6	100%	99%	Hypoparathyroidism, autosomal dominant, 146200 Hypoparathyroidism, autosomal recessive, 146200
PTH1R	87.2	99%	98%	Chondrodysplasia, Blomstrand type, 215045 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Metaphyseal chondrodysplasia, Murk Jansen type, 156400

PTHLH	117.9	100%	82%	Brachydactyly, type E2, 613382 Humoral hypercalcemia of malignancy
PTPN11	74.9	96%	86%	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
PTPN12	122.5	95%	90%	Colon cancer, somatic, 114500
PTPN14	158.2	98%	97%	Choanal atresia and lymphedema, 613611
PTPRC	91.6	89%	80%	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971 {Hepatitic C virus, susceptibility to}, 609532
PTPRJ	150.7	96%	95%	Colon cancer, somatic, 114500
PTPRO	137.0	99%	98%	Nephrotic syndrome, type 6, 614196
PTPRQ	98.6	90%	87%	Deafness, autosomal recessive 84A, 613391
PTRF	107.4	100%	98%	Lipodystrophy, congenital generalized, type 4, 613327
PTRH2	237.1	100%	100%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PTS	94.2	94%	83%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUF60	142.8	100%	96%	Verheij syndrome, 615583
PURA	117.2	100%	91%	Mental retardation, autosomal dominant 31, 616158
PUS1	126.0	100%	95%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PVRL1	130.8	100%	100%	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060
PVRL4	118.8	100%	98%	Ectodermal dysplasia-syndactyly syndrome 1, 613573
PXDN	143.0	99%	98%	Corneal opacification and other ocular anomalies, 269400
PYCR1	82.7	97%	95%	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
PYCR2	113.8	100%	95%	Leukodystrophy, hypomyelinating, 10, 616420
PYGL	148.1	100%	100%	Glycogen storage disease VI, 232700
PYGM	123.6	100%	99%	McArdle disease, 232600
QARS	128.6	100%	99%	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	65.9	100%	100%	Hyperphenylalaninemia, BH4-deficient, C, 261630
RAB18	81.2	99%	78%	Warburg micro syndrome 3, 614222
RAB23	105.2	100%	99%	Carpenter syndrome, 201000
RAB27A	137.7	100%	100%	Griscelli syndrome, type 2, 607624
RAB28	49.9	99%	85%	Cone-rod dystrophy 18, 615374

RAB33B	206.0	100%	100%	Smith-McCort dysplasia 2, 615222
RAB39B	144.1	100%	100%	Mental retardation, X-linked 72, 300271 ?Waisman syndrome, 311510
RAB3GAP1	112.3	99%	96%	Warburg micro syndrome 1, 600118
RAB3GAP2	84.5	97%	91%	Martsolf syndrome, 212720 Warburg micro syndrome 2, 614225
RAB7A	135.9	100%	100%	Charcot-Marie-Tooth disease,type 2B, 600882
RAC2	106.4	100%	100%	Neutrophil immunodeficiency syndrome, 608203
RAD21	84.4	99%	94%	Cornelia de Lange syndrome 4, 614701
RAD50	88.5	93%	88%	Nijmegen breakage syndrome-like disorder, 613078
RAD51	122.7	89%	89%	Mirror movements 2, 614508 {Breast cancer, susceptibility to}, 114480
RAD51C	121.9	100%	100%	Fanconi anemia, complementation group O, 613390 {Breast-ovarian cancer, familial, susceptibility to, 3}, 613399
RAD54B	87.9	95%	85%	Colon cancer, somatic, 114500 Lymphoma, non-Hodgkin, somatic, 605027
RAD54L	97.9	99%	97%	Adenocarcinoma, colonic, somatic Lymphoma, non-Hodgkin, somatic, 605027 {Breast cancer, invasive ductal}, 114480
RAF1	105.4	100%	100%	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553
RAG1	183.4	100%	100%	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	214.7	100%	100%	Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457
RAI1	111.0	99%	99%	Smith-Magenis syndrome, 182290
RAP1GDS1	81.5	98%	86%	Lymphocytic leukemia, acute T-cell
RAPSN	105.8	93%	93%	Fetal akinesia deformation sequence, 208150 Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326

RARB	118.7	100%	100%	Microphthalmia, syndromic 12, 615524
RARS	76.7	98%	92%	Leukodystrophy, hypomyelinating, 9, 616140
RARS2	100.2	99%	96%	Pontocerebellar hypoplasia, type 6, 611523
RASA1	88.7	93%	79%	Basal cell carcinoma, somatic, 605462 Capillary malformation-arteriovenous malformation, 608354 Parkes Weber syndrome, 608355
RAX	73.4	91%	81%	Microphthalmia, isolated 3, 611038
RAX2	51.9	99%	78%	Macular degeneration, age-related, 6, 613757 ?Cone-rod dystrophy 11, 610381
RB1	75.2	90%	75%	Bladder cancer, somatic, 109800 Osteosarcoma, somatic, 259500 Retinoblastoma, 180200 Retinoblastoma, trilateral, 180200 Small cell cancer of the lung, somatic, 182280
RB1CC1	104.7	94%	87%	Breast cancer, somatic, 114480
RBBP8	95.8	99%	94%	Jawad syndrome, 251255 Pancreatic carcinoma, somatic Seckel syndrome 2, 606744
RBCK1	92.4	98%	94%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RBM10	122.1	100%	99%	TARP syndrome, 311900
RBM20	154.3	99%	95%	Cardiomyopathy, dilated, 1DD, 613172
RBM8A	100.8	100%	95%	Thrombocytopenia-absent radius syndrome, 274000
RBP4	93.1	93%	89%	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RBPJ	67.2	87%	76%	Adams-Oliver syndrome 3, 614814
RD3	142.0	100%	100%	Leber congenital amaurosis 12, 610612
RDH12	78.5	97%	91%	Leber congenital amaurosis 13, 612712
RDH5	148.7	100%	100%	Fundus albipunctatus, 136880
RDX	35.3	66%	55%	Deafness, autosomal recessive 24, 611022
RECQL4	127.6	97%	96%	Baller-Gerold syndrome, 218600 RAPADILINO syndrome, 266280 Rothmund-Thomson syndrome, 268400
REEP1	91.3	100%	95%	Spastic paraplegia 31, autosomal dominant, 610250 ?Neuronopathy, distal hereditary motor, type VB, 614751

RELN	147.0	100%	99%	Lissencephaly 2 (Norman-Roberts type), 257320 {Epilepsy, familial temporal lobe, 7}, 616436
REN	134.0	100%	100%	Hyperuricemic nephropathy, familial juvenile 2, 613092 Renal tubular dysgenesis, 267430 [Hyperproreninemia]
RERE	67.5	96%	91%	Neurodevelopmental disorder with or without anomalies of the brain,eye or heart, 616975
RET	127.8	98%	96%	Central hypoventilation syndrome, congenital, 209880 Medullary thyroid carcinoma, 155240 Multiple endocrine neoplasia IIA, 171400 Multiple endocrine neoplasia IIB, 162300 Pheochromocytoma, 171300 {Hirschsprung disease, susceptibility to, 1}, 142623
RFT1	97.6	100%	95%	Congenital disorder of glycosylation, type In, 612015
RFX5	93.9	98%	95%	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFX6	135.6	100%	100%	Mitchell-Riley syndrome, 615710
RFXANK	97.7	100%	100%	MHC class II deficiency, complementation group B, 209920
RFXAP	100.9	94%	93%	Bare lymphocyte syndrome, type II, complementation group D, 209920
RGR	111.5	100%	100%	Retinitis pigmentosa 44, 613769
RGS9	91.2	100%	94%	Bradyopsia, 608415
RGS9BP	65.8	100%	99%	Bradyopsia, 608415
RHAG	147.5	100%	96%	Anemia, hemolytic, Rh-null, regulator type, 268150 Overhydrated hereditary stomatocytosis, 185000 Rh-mod syndrome
RHBDF2	92.3	97%	95%	Tylosis with esophageal cancer, 148500
RHCE	153.0	99%	99%	Rh-null disease, amorph type, 111700 [Blood group, Rhesus], 111690
RHO	186.4	100%	100%	Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 Retinitis punctata albescens, 136880
RIMS1	102.9	99%	94%	Cone-rod dystrophy 7, 603649
RIN2	96.2	100%	99%	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075
RIPK4	125.9	100%	99%	Popliteal pterygium syndrome, Bartsocas-Papas type, 263650
RIT1	145.2	100%	100%	Noonan syndrome 8, 615355

RLBP1	109.6	100%	100%	Bothnia retinal dystrophy, 607475 Fundus albipunctatus, 136880 Newfoundland rod-cone dystrophy, 607476 Retinitis punctata albescens, 136880
RLIM	140.4	100%	100%	Mental Retardation, X-linked 61, 300978
RMND1	117.8	99%	96%	Combined oxidative phosphorylation deficiency 11, 614922
RMRP	NC	NC	NC	Anauxetic dysplasia, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RNASEH1	92.0	94%	91%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479
RNASEH2A	111.1	100%	99%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	101.2	90%	87%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	156.2	100%	99%	Aicardi-Goutieres syndrome 3, 610329
RNASEL	121.7	99%	98%	Prostate cancer 1, 601518
RNASET2	73.7	93%	85%	Leukoencephalopathy, cystic, without megalencephaly, 612951
RNF125	148.2	100%	98%	Tenorio syndrome, 616260
RNF135	69.0	93%	87%	Macrocephaly, macrosomia, facial dysmorphism syndrome, 614192
RNF139	199.9	100%	100%	Renal cell carcinoma, 144700
RNF168	202.2	100%	98%	RIDDLE syndrome, 611943
RNF170	112.1	96%	93%	Ataxia, sensory, 1, autosomal dominant, 608984
RNF212	100.9	99%	95%	Recombination rate QTL 1, 612042
RNF216	122.4	99%	95%	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840
RNF6	175.3	100%	100%	Esophageal carcinoma, somatic, 133239
RNU4ATAC	NC	NC	NC	Microcephalic osteodysplastic primordial dwarfism, type I, 210710 Roifman syndrome, 616651
ROBO2	127.4	97%	96%	Vesicoureteral reflux 2, 610878
ROBO3	84.9	98%	93%	Gaze palsy, horizontal, with progressive scoliosis, 607313
ROGDI	106.5	95%	94%	Kohlschutter-Tonz syndrome, 226750
ROM1	100.9	100%	98%	Retinitis pigmentosa 7, digenic, 608133
ROR2	153.6	99%	98%	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RORC	114.0	100%	99%	Immunodeficiency 42, 616622
RP1	112.7	100%	98%	Retinitis pigmentosa 1, 180100
RP1L1	86.7	100%	99%	Occult macular dystrophy, 613587

RP2	213.4	100%	100%	Retinitis pigmentosa 2, 312600
RPE65	122.8	99%	98%	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794
RPGR	108.1	85%	78%	Cone-rod dystrophy, X-linked, 1, 304020 Macular degeneration, X-linked atrophic, 300834 Retinitis pigmentosa 3, 300029 Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455
RPGRIP1	132.6	100%	99%	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826
RPGRIP1L	128.5	96%	93%	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561
RPL11	83.9	100%	99%	Diamond-Blackfan anemia 7, 612562
RPL21	72.1	80%	65%	Hypotrichosis 12, 615885
RPL35A	59.2	93%	81%	Diamond-Blackfan anemia 5, 612528
RPL5	30.2	69%	55%	Diamond-Blackfan anemia 6, 612561
RPS10	97.4	99%	89%	Diamond-Blackfan anemia 9, 613308
RPS14	111.1	96%	92%	Macrocytic anemia, refractory, due to 5q deletion, somatic, 153550
RPS17	40.6	69%	69%	Diamond-Blackfan anemia 4, 612527
RPS19	66.2	100%	99%	Diamond-Blackfan anemia 1, 105650
RPS24	88.7	87%	87%	Diamond-blackfan anemia 3, 610629
RPS26	77.0	99%	91%	Diamond-Blackfan anemia 10, 613309
RPS28	34.8	93%	82%	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164
RPS29	57.5	99%	93%	Diamond-Blackfan anemia 13, 615909
RPS6KA3	98.8	98%	93%	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844
RPS7	89.7	86%	63%	Diamond-Blackfan anemia 8, 612563
RPSA	62.0	100%	100%	Asplenia, isolated congenital, 271400
RRAS2	70.8	81%	75%	Ovarian carcinoma
RRM2B	133.1	99%	97%	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077
RS1	64.1	99%	95%	Retinoschisis, 312700
RSPH1	148.3	100%	100%	Ciliary dyskinesia, primary, 24, 615481

RSPH3	110.8	98%	96%	Ciliary dyskinesia, primary, 32, 616481
RSPH4A	123.7	96%	94%	Ciliary dyskinesia, primary, 11, 612649
RSPH9	130.2	100%	99%	Ciliary dyskinesia, primary, 12, 612650
RSPO1	97.9	100%	100%	Palmoplantar hyperkeratosis and true hermaphroditism, 610644 Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644
RSPO4	103.6	100%	100%	Anonychia congenita, 206800
RSPRY1	141.3	100%	99%	Spondylopeimetaphyseal dsyplasia, Faden-Alkuraya type, 616723
RTEL1	110.0	99%	96%	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373
RTN2	97.1	96%	90%	Spastic paraplegia 12, autosomal dominant, 604805
RTN4IP1	82.5	98%	96%	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
RTTN	121.2	97%	94%	Microcephaly,short stature,and polymicrogyria with seizures,614833
RUNX1	95.2	95%	91%	Leukemia, acute myeloid, 601626 Platelet disorder, familial, with associated myeloid malignancy, 601399
RUNX2	82.1	74%	74%	Cleidocranial dysplasia, 119600 Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600 Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510
RYR1	102.9	96%	92%	Central core disease, 117000 King-Denborough syndrome, 145600 Minicore myopathy with external ophthalmoplegia, 255320 Neuromuscular disease, congenital, with uniform type 1 fiber, 117000 {Malignant hyperthermia susceptibility 1}, 145600
RYR2	124.4	99%	97%	Arrhythmogenic right ventricular dysplasia 2, 600996 Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772
S1PR2	191.7	90%	88%	Deafness, autosomal recessive 68, 610419
SACS	142.5	99%	99%	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAG	119.9	100%	100%	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758
SALL1	115.3	98%	98%	Townes-Brocks branchiootorenal-like syndrome, 107480 Townes-Brocks syndrome, 107480
SALL4	120.3	97%	95%	Duane-radial ray syndrome, 607323 IVIC syndrome, 147750

SAMD9	158.1	100%	99%	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455
SAMD9L	145.2	100%	100%	Ataxia-pancytopenia syndrome, 159550
SAMHD1	120.6	100%	98%	Aicardi-Goutieres syndrome 5, 612952 ?Chilblain lupus 2, 614415
SAR1B	125.6	89%	88%	Chylomicron retention disease, 246700
SARS2	97.2	96%	95%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SATB2	97.5	97%	86%	Glass syndrome, 612313
SBDS	163.6	100%	99%	Shwachman-Diamond syndrome, 260400 {Aplastic anemia, susceptibility to}, 609135
SBF1	85.1	99%	94%	Charcot-Marie-Tooth disease, type 4B3, 615284
SBF2	112.7	98%	96%	Charcot-Marie-Tooth disease, type 4B2, 604563
SC5D	169.7	100%	97%	Lathosterolosis, 607330
SCARB2	113.4	100%	97%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCARF2	62.8	88%	73%	Van den Ende-Gupta syndrome, 600920
SCN10A	155.0	100%	99%	Episodic pain syndrome, familial, 2, 615551
SCN11A	127.1	99%	97%	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN1A	122.5	99%	96%	Dravet syndrome, 607208 Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Febrile seizures, familial, 3A, 604403 Migraine, familial hemiplegic, 3, 609634
SCN1B	146.7	100%	96%	Atrial fibrillation, familial, 13, 615377 Brugada syndrome 5, 612838 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233
SCN2A	132.4	99%	96%	Epileptic encephalopathy, early infantile, 11, 613721 Seizures, benign familial infantile, 3, 607745
SCN2B	170.4	100%	100%	Atrial fibrillation, familial, 14, 615378
SCN3B	132.4	100%	100%	Atrial fibrillation, familial, 16, 613120 Brugada syndrome 7, 613120

SCN4A	188.5	100%	99%	Hyperkalemic periodic paralysis, type 2, 170500 Hypokalemic periodic paralysis, type 2, 613345 Myasthenic syndrome, congenital, 16, 614198 Myotonia congenita, atypical, acetazolamide-responsive, 608390 Paramyotonia congenita, 168300
SCN4B	64.3	100%	97%	Atrial fibrillation, familial, 17, 611819 Long QT syndrome-10, 611819
SCN5A	145.7	100%	100%	Atrial fibrillation, familial, 10, 614022 Brugada syndrome 1, 601144 Cardiomyopathy, dilated, 1E, 601154 Heart block, nonprogressive, 113900 Heart block, progressive, type IA, 113900 Long QT syndrome-3, 603830 Sick sinus syndrome 1, 608567 Ventricular fibrillation, familial, 1, 603829 {Sudden infant death syndrome, susceptibility to}, 272120
SCN8A	180.8	100%	99%	Epileptic encephalopathy, early infantile, 13, 614558 ?Cognitive impairment with or without cerebellar ataxia, 614306
SCN9A	128.5	98%	94%	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	109.4	97%	93%	Bronchiectasis with or without elevated sweat chloride 2, 613021 Pseudohypoaldosteronism, type I, 264350
SCNN1B	138.5	100%	100%	Bronchiectasis with or without elevated sweat chloride 1, 211400 Liddle syndrome, 177200 Pseudohypoaldosteronism, type I, 264350
SCNN1G	120.5	97%	93%	Bronchiectasis with or without elevated sweat chloride 3, 613071 Liddle syndrome, 177200 Pseudohypoaldosteronism, type I, 264350
SCO1	98.1	96%	93%	Mitochondrial complex IV deficiency, 220110

SCO2	87.8	100%	98%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908
SCP2	94.1	100%	97%	Leukoencephalopathy with dystonia and motor neuropathy, 613724
SCYL1	138.3	100%	97%	Spinocerebellar ataxia, autosomal recessive 21, 616719
SDCCAG8	109.1	99%	91%	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SDHA	80.2	82%	77%	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165
SDHAF1	44.3	100%	99%	Mitochondrial complex II deficiency, 252011
SDHAF2	122.6	94%	92%	Paragangliomas 2, 601650
SDHB	123.6	100%	100%	Cowden syndrome 2, 612359 Gastrointestinal stromal tumor, 606764 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 4, 115310 Pheochromocytoma, 171300
SDHC	83.8	100%	93%	Gastrointestinal stromal tumor, 606764 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 3, 605373
SDHD	50.5	59%	59%	Carcinoid tumors, intestinal, 114900 Cowden syndrome 3, 615106 Merkel cell carcinoma, somatic Mitochondrial complex II deficiency, 252011 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 1, with or without deafness, 168000 Pheochromocytoma, 171300
SEC23A	116.6	98%	93%	Craniolenticulosutural dysplasia, 607812
SEC23B	146.5	97%	95%	Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SEC24D	122.0	99%	96%	Cole-Carpenter syndrome 2, 616294
SEC61A1	119.6	100%	100%	Hyperuricemic nephropathy, familial juvenile, 4,617056
SEC63	57.2	80%	72%	Polycystic liver disease, 174050
SECISBP2	109.8	99%	87%	Thyroid hormone metabolism, abnormal, 609698

SEMA4A	110.3	99%	98%	Cone-rod dystrophy 10, 610283 Retinitis pigmentosa 35, 610282
SEPN1	96.0	84%	83%	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310
SEPSECS	150.6	100%	100%	Pontocerebellar hypoplasia type 2D, 613811
SEPT12	84.6	98%	94%	Spermatogenic failure 10, 614822
SEPT9	107.3	99%	95%	Amyotrophy, hereditary neuralgic, 162100 Leukemia, acute myeloid, therapy-related Ovarian carcinoma
SERAC1	96.7	100%	93%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPINA1	127.0	100%	100%	Emphysema due to AAT deficiency, 613490 Emphysema-cirrhosis, due to AAT deficiency, 613490 Hemorrhagic diathesis due to \'antithrombin\' Pittsburgh, 613490 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963
SERPINA3	114.1	100%	100%	Alpha-1-antichymotrypsin deficiency Cerebrovascular disease, occlusive
SERPINA6	172.6	100%	100%	Corticosteroid-binding globulin deficiency,611489
SERPINB7	114.4	100%	99%	Palmoplantar keratoderma, Nagashima type, 615598
SERPINC1	118.5	100%	100%	Thrombophilia due to antithrombin III deficiency, 613118
SERPIND1	138.8	100%	100%	Thrombophilia due to heparin cofactor II deficiency,612356
SERPINE1	133.8	100%	100%	Plasminogen activator inhibitor-1 deficiency, 613329 {Transcription of plasminogen activator inhibitor, modulator of}
SERPINF1	83.9	100%	98%	Osteogenesis imperfecta, type VI, 613982
SERPINF2	114.9	99%	95%	Alpha-2-plasmin inhibitor deficiency,262850
SERPING1	85.3	96%	93%	Angioedema, hereditary, types I and II, 106100 Complement component 4, partial deficiency of, 120790
SERPINI1	98.9	100%	95%	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218
SETBP1	110.7	96%	95%	Mental retardation, autosomal dominant 29, 616078 Schinzel-Giedion midface retraction syndrome, 269150
SETD2	131.5	100%	99%	Luscan-Lumish syndrome, 616831
SETD5	154.3	100%	99%	Mental retardation, autosomal dominant 23, 615761
SETX	147.8	100%	98%	Amyotrophic lateral sclerosis 4, juvenile, 602433 Spinocerebellar ataxia, autosomal recessive 1, 606002
SF3B1	125.6	98%	96%	Myelodysplastic syndrome, somatic, 614286

SF3B4	65.3	100%	94%	Acrofacial dysostosis 1, Nager type, 154400
SFRP4	129.8	100%	97%	Pyle disease, 265900
SFTPA2	121.3	100%	100%	Pulmonary fibrosis, idiopathic, 178500
SFTPB	84.5	100%	99%	Surfactant metabolism dysfunction, pulmonary, 1, 265120
SFTPC	80.5	100%	95%	Surfactant metabolism dysfunction, pulmonary, 2, 610913
SFXN4	126.2	100%	99%	Combined oxidative phosphorylation deficiency 18, 615578
SGCA	129.3	100%	99%	Muscular dystrophy, limb-girdle, type 2D, 608099
SGCB	147.1	99%	96%	Muscular dystrophy, limb-girdle, type 2E, 604286
SGCD	83.4	100%	98%	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, type 2F, 601287
SGCE	81.9	95%	89%	Dystonia-11, myoclonic, 159900
SGCG	120.4	100%	100%	Muscular dystrophy, limb-girdle, type 2C, 253700
SGOL1	93.9	99%	89%	Chronic atrial and intestinal dysrhythmia, 616201
SGSH	111.1	94%	94%	Mucopolysaccharidisis type IIIA (Sanfilippo A), 252900
SH2B3	82.3	84%	74%	Erythrocytosis, somatic, 133100 Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
SH2D1A	137.8	89%	89%	Lymphoproliferative syndrome, X-linked, 1, 308240
SH3BP2	93.8	91%	89%	Cherubism, 118400
SH3PXD2B	123.4	100%	100%	Frank-ter Haar syndrome, 249420
SH3TC2	87.6	99%	96%	Charcot-Marie-Tooth disease, type 4C, 601596 Mononeuropathy of the median nerve, mild, 613353
SHANK3	61.7	76%	70%	Phelan-McDermid syndrome, 606232 {Schizophrenia 15}, 613950
SHH	90.0	99%	93%	Holoprosencephaly-3, 142945 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250
SHOC2	124.7	100%	99%	Noonan-like syndrome with loose anagen hair, 607721
SHOX	18.3	70%	48%	Langer mesomelic dysplasia, 249700 Leri-Weill dyschondrosteosis, 127300 Short stature, idiopathic familial, 300582
SI	92.9	96%	86%	Sucrase-isomaltase deficiency, congenital, 222900
SIK1	84.9	96%	92%	Epileptic encephalopathy, early infantile, 30, 616341

SIL1	134.6	99%	97%	Marinesco-Sjogren syndrome, 248800
SIM1	127.2	100%	99%	Obesity, severe, 601665
SIX1	96.2	100%	99%	Brachiootic syndrome 3, 608389 Deafness, autosomal dominant 23, 605192
SIX3	118.5	99%	96%	Holoprosencephaly-2, 157170 Schizencephaly, 269160
SIX5	38.0	83%	70%	Branchiootorenal syndrome 2, 610896
SIX6	179.8	100%	99%	Optic disc anomalies with retinal and/or macular dystrophy, 212550
SKI	73.7	99%	96%	Shprintzen-Goldberg syndrome, 182212
SKIV2L	21.0	69%	42%	Trichohepatoenteric syndrome 2, 614602
SLC10A2	127.1	100%	100%	Bile acid malabsorption, primary, 613291
SLC11A2	115.8	100%	99%	Anemia, hypochromic microcytic, with iron overload 1, 206100
SLC12A1	154.5	100%	99%	Bartter syndrome, type 1, 601678
SLC12A3	125.7	100%	99%	Gitelman syndrome, 263800
SLC12A5	139.4	99%	96%	Epileptic encephalopathy, early infantile, 34, 616645 {Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685
SLC12A6	134.2	100%	100%	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC13A5	141.5	100%	100%	Epileptic encephalopathy, early infantile, 25, 615905
SLC16A1	126.9	99%	94%	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia, familial, 7, 610021 Monocarboxylate transporter 1 deficiency, 616095
SLC16A12	168.9	100%	100%	Cataract, juvenile, with microcornea and glucosuria, 612018
SLC16A2	72.1	98%	89%	Allan-Herndon-Dudley syndrome, 300523
SLC17A5	103.5	100%	92%	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC17A8	122.7	100%	100%	Deafness, autosomal dominant 25, 605583
SLC17A9	102.3	95%	94%	Porokeratosis 8, disseminated superficial actinic type, 616063
SLC19A2	102.3	99%	95%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	151.6	100%	100%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A1	170.5	100%	100%	Dicarboxylic aminoaciduria, 222730 {?Schizophrenia susceptibility 18}, 615232
SLC1A3	116.2	100%	100%	Episodic ataxia, type 6, 612656
SLC1A4	141.2	100%	99%	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657

SLC20A2	83.6	98%	90%	Basal ganglia calcification, idiopathic, 1, 213600
SLC22A12	96.3	100%	99%	Hypouricemia, renal, 220150
SLC22A18	82.0	96%	77%	Breast cancer, somatic, 114480 Lung cancer, somatic, 211980 Rhabdomyosarcoma, somatic, 268210
SLC22A5	131.0	100%	100%	Carnitine deficiency, systemic primary, 212140
SLC24A1	174.9	100%	100%	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
SLC24A4	106.9	99%	96%	Amelogenesis imperfecta, type IIA5, 615887 [Skin/hair/eye pigmentation 6, blond/brown hair], 210750 [Skin/hair/eye pigmentation 6, blue/green eyes], 210750
SLC24A5	99.7	98%	87%	Albinism, oculocutaneous, type VI, 113750 [Skin/hair/eye pigmentation 4, fair/dark skin], 113750
SLC25A1	64.3	96%	88%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A12	126.8	96%	95%	Epileptic encephalopathy, early infantile, 39, 612949
SLC25A13	104.9	99%	91%	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A15	179.4	99%	96%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A19	61.8	95%	85%	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A20	82.6	100%	95%	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A22	93.7	100%	97%	Epileptic encephalopathy, early infantile, 3, 609304
SLC25A26	79.7	96%	93%	Combined oxidative phosphorylation deficiency 28, 616794
SLC25A3	125.0	96%	90%	Mitochondrial phosphate carrier deficiency, 610773
SLC25A38	93.8	100%	93%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC25A4	122.5	100%	100%	Mitochondrial DNA depletion syndrome 12 (cardiomyopathic type), 615418 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283
SLC25A46	149.0	92%	81%	Neuropathy, hereditary motor and sensory, type VIB, 616505
SLC26A2	206.3	100%	100%	Achondrogenesis Ib, 600972 Atelosteogenesis II, 256050 De la Chapelle dysplasia, 256050 Diastrophic dysplasia, 222600 Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 Epiphyseal dysplasia, multiple, 4, 226900
SLC26A3	144.5	98%	96%	Diarrhea 1, secretory chloride, congenital, 214700

SLC26A4	117.9	100%	97%	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791 Pendred syndrome, 274600
SLC26A8	118.9	100%	100%	Spermatogenic failure 3, 606766
SLC27A4	123.7	100%	97%	Ichthyosis prematurity syndrome, 608649
SLC29A3	171.4	100%	98%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC2A1	143.3	100%	100%	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	138.3	100%	100%	Arterial tortuosity syndrome, 208050
SLC2A2	151.5	100%	99%	Fanconi-Bickel syndrome, 227810 {Diabetes mellitus, noninsulin-dependent}, 125853
SLC2A9	124.1	100%	96%	Hypouricemia, renal, 2, 612076 {Uric acid concentration, serum, QTL 2}, 612076
SLC30A10	135.4	100%	100%	Hypermanganesemia with dystonia 1, 613280
SLC30A2	115.4	100%	95%	Zinc deficiency, transient neonatal, 608118
SLC33A1	117.8	95%	87%	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539
SLC34A1	121.3	100%	99%	Fanconi renotubular syndrome 2, 613388 Hypercalcemia, infantile 2, 616963 Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286
SLC34A2	139.9	100%	100%	Pulmonary alveolar microlithiasis, 265100
SLC34A3	96.3	94%	91%	Hypophosphatemic rickets with hypercalciuria, 241530
SLC35A1	123.3	100%	99%	Congenital disorder of glycosylation, type IIf, 603585
SLC35A2	118.5	100%	99%	Congenital disorder of glycosylation, type IIm, 300896
SLC35C1	172.5	97%	93%	Congenital disorder of glycosylation, type IIc, 266265
SLC35D1	110.7	93%	87%	Schneckenbecken dysplasia, 269250
SLC36A2	97.7	100%	99%	Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC37A4	114.9	100%	99%	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240
SLC38A8	71.9	99%	94%	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218
SLC39A13	102.5	100%	98%	Spondylocheirodysplasia, Ehlers-Danlos syndrome-like, 612350

SLC39A14	86.1	97%	94%	Hypermanganesemia with dystonia 2, 617013
SLC39A4	66.8	98%	91%	Acrodermatitis enteropathica, 201100
SLC39A5	106.0	100%	98%	Myopia 24, autosomal dominant, 615946
SLC39A8	114.6	100%	99%	Congenital disorder of glycosylation, type IIn, 616721
SLC3A1	148.1	100%	99%	Cystinuria, 220100
SLC40A1	118.7	98%	97%	Hemochromatosis, type 4, 606069
SLC45A2	103.8	100%	98%	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	80.2	99%	92%	Folate malabsorption, hereditary, 229050
SLC4A1	120.5	100%	99%	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	125.4	100%	100%	Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy 2, autosomal recessive, 217700 Corneal endothelial dystrophy and perceptive deafness, 217400
SLC4A4	123.1	100%	98%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC52A1	173.3	100%	100%	Riboflavin deficiency, 615026
SLC52A2	154.0	100%	100%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	99.7	100%	100%	Brown-Vialetto-Van Laere syndrome 1, 211530 Fazio-Londe disease, 211500
SLC5A1	120.1	100%	100%	Glucose/galactose malabsorption, 606824
SLC5A2	106.8	100%	94%	Renal glucosuria, 233100
SLC5A5	91.9	100%	91%	Thyroid dyshormonogenesis 1, 274400
SLC5A7	107.1	100%	99%	Neuronopathy, distal hereditary motor, type VIIA, 158580

SLC6A1	127.1	100%	100%	Myoclonic-atonic epilepsy, 616421
SLC6A17	172.0	100%	100%	Mental retardation, autosomal recessive 48, 616269
SLC6A19	144.9	100%	99%	Hartnup disorder, 234500 Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC6A2	123.9	100%	99%	Orthostatic intolerance, 604715
SLC6A20	135.5	99%	96%	Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC6A3	120.0	100%	98%	Parkinsonism-dystonia, infantile, 613135 {Nicotine dependence, protection against}, 188890
SLC6A5	127.5	100%	100%	Hyperekplexia 3, 614618
SLC6A8	57.9	92%	88%	Cerebral creatine deficiency syndrome 1, 300352
SLC7A14	149.1	100%	100%	Retinitis pigmentosa 68, 615725
SLC7A7	105.9	100%	100%	Lysinuric protein intolerance, 222700
SLC7A9	125.8	100%	98%	Cystinuria, 220100
SLC9A3	145.6	98%	96%	Diarrhea 8, secretory sodium, congenital, 616868
SLC9A3R1	104.1	100%	100%	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
SLC9A6	131.6	99%	88%	Mental retardation, X-linked syndromic, Christianson type, 300243
SLCO1B1	41.1	93%	87%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO1B3	44.3	95%	72%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO2A1	86.8	100%	98%	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
SLFN14	177.8	100%	100%	Bleeding disorder,platelet-type,20,616913
SLITRK1	113.2	100%	100%	Tourette syndrome, 137580 ?Trichotillomania, 613229
SLITRK6	193.5	100%	100%	Deafness and myopia, 221200
SLURP1	83.3	98%	86%	Meleda disease, 248300
SLX4	97.2	99%	98%	Fanconi anemia, complementation group P, 613951
SMAD3	109.5	100%	98%	Loeys-Dietz syndrome 3, 613795
SMAD4	97.9	99%	98%	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Polyposis, juvenile intestinal, 174900
SMAD6	83.5	88%	75%	Aortic valve disease 2, 614823

SMAD9	124.3	100%	100%	Pulmonary hypertension,primary,615342 Polyposis & gastrointestinal ganglioneuromas (Ngeow (2015) Gastroenterology 149,886)
SMARCA2	103.2	95%	93%	Nicolaides-Baraitser syndrome, 601358
SMARCA4	121.2	99%	95%	Coffin-Siris syndrome 4, 614609 {Rhabdoid tumor predisposition syndrome 2}, 613325
SMARCAD1	72.7	98%	92%	Adermatoglyphia, 136000
SMARCAL1	120.5	100%	99%	Schimke immunoosseous dysplasia, 242900
SMARCB1	195.8	100%	100%	Coffin-Siris syndrome 3, 614608 Rhabdoid tumors, somatic, 609322 {Rhabdoid predisposition syndrome 1}, 609322 {Schwannomatosis-1, susceptibility to}, 162091
SMARCE1	68.7	96%	82%	Coffin-Siris syndrome 5,616938 {Meningioma, familial, susceptibility to}, 607174
SMC1A	116.2	100%	100%	Cornelia de Lange syndrome 2,300590
SMC3	81.5	92%	82%	Cornelia de Lange syndrome 3, 610759
SMCHD1	83.5	96%	87%	Fascioscapulohumeral muscular dystrophy 2, digenic, 158901
SMN1	90.1	100%	92%	Spinal muscular atrophy-1, 253300 Spinal muscular atrophy-2, 253550 Spinal muscular atrophy-3, 253400 Spinal muscular atrophy-4, 271150
SMO	132.5	96%	92%	Basal cell carcinoma, somatic, 605462 Curry-Jones syndrome,somatic mosaic, 601707
SMOC1	104.0	98%	93%	Microphthalmia with limb anomalies, 206920
SMOC2	99.3	97%	93%	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SMPD1	104.5	98%	95%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SMPX	101.8	100%	99%	Deafness, X-linked 4, 300066
SMS	69.7	84%	78%	Mental retardation, X-linked, Snyder-Robinson type, 309583
SNAI2	124.1	100%	97%	Piebaldism, 172800 Waardenburg syndrome, type 2D, 608890
SNAP29	119.6	100%	100%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNCA	113.5	100%	100%	Dementia, Lewy body, 127750 Parkinson disease 1, 168601 Parkinson disease 4, 605543

SNCB	63.3	100%	100%	Dementia, Lewy body, 127750
SNIP1	122.2	100%	97%	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501
SNRNP200	130.9	99%	97%	Retinitis pigmentosa 33, 610359
SNRPB	71.8	100%	94%	Cerebrocostomandibular syndrome, 117650
SNRPE	90.3	99%	96%	Hypotrichosis 11, 615059
SNRPN	101.9	100%	98%	Prader-Willi syndrome, 176270
SNTA1	79.2	86%	79%	Long QT syndrome 12, 612955
SNX10	96.8	100%	96%	Osteopetrosis, autosomal recessive 8, 615085
SNX14	66.5	89%	79%	Spinocerebellar ataxia, autosomal recessive 20, 616354
SOBP	87.7	96%	86%	Mental retardation, anterior maxillary protrusion, and strabismus, 613671
SOD1	138.7	100%	100%	Amyotrophic lateral sclerosis 1, 105400
SOS1	85.5	92%	89%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOS2	86.7	98%	91%	Noonan syndrome 9, 616559
SOST	113.1	100%	100%	Craniodiaphyseal dysplasia, autosomal dominant, 122860 Sclerosteosis 1, 269500 Van Buchem disease, 239100
SOX10	61.5	97%	90%	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266
SOX11	99.5	100%	95%	Mental retardation, autosomal dominant, 27, 615866
SOX17	50.7	96%	83%	Vesicoureteral reflux 3, 613674
SOX18	19.8	74%	49%	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823 Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940
SOX2	83.7	100%	97%	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SOX3	51.9	94%	88%	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000
SOX5	88.3	99%	96%	Lamb-Shaffer syndrome, 616803
SOX9	95.7	95%	89%	Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia with autosomal sex reversal, 114290 Campomelic dysplasia, 114290
SP110	113.6	100%	99%	Hepatic venoocclusive disease with immunodeficiency, 235550 {Mycobacterium tuberculosis, susceptibility to}, 607948

SPAG1	74.2	96%	86%	Ciliary dyskinesia, primary, 28, 615505
SPARC	148.4	100%	100%	Osteogenesis imperfecta, type XVII, 616507
SPAST	54.8	87%	72%	Spastic paraplegia 4, autosomal dominant, 182601
SPATA5	116.5	99%	98%	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPATA7	116.6	97%	92%	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, autosomal recessive, 604232
SPECC1L	132.3	100%	100%	Opitz GBBB syndrome, type II, 145410 ?Facial clefting, oblique, 1, 600251
SPEG	85.3	92%	84%	Centronuclear myopathy 5, 615959
SPG11	117.4	98%	96%	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360
SPG20	131.7	97%	96%	Troyer syndrome, 275900
SPG21	116.4	100%	99%	Mast syndrome, 248900
SPG7	98.1	95%	91%	Spastic paraplegia 7, autosomal recessive, 607259
SPINK1	64.0	100%	95%	Pancreatitis, hereditary, 167800 Tropical calcific pancreatitis, 608189 {Fibrocalculous pancreatic diabetes, susceptibility to}, 608189
SPINK5	134.0	99%	92%	Atopy, 147050 Netherton syndrome, 256500
SPINT2	55.0	99%	88%	Diarrhea 3, secretory sodium, congenital, syndromic, 270420
SPR	150.6	95%	87%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRED1	135.7	96%	96%	Legius syndrome, 611431
SPRTN	142.9	100%	100%	Ruijs-Aalfs syndrome, 616200
SPRY4	123.7	100%	100%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
SPTA1	110.7	99%	98%	Elliptocytosis-2, 130600 Pyropoikilocytosis, 266140 Spherocytosis, type 3, 270970
SPTAN1	108.8	99%	97%	Epileptic encephalopathy, early infantile, 5, 613477
SPTB	126.3	100%	99%	Anemia, neonatal hemolytic, fatal and near-fatal Elliptocytosis-3 Spherocytosis, type 2, 616649
SPTBN2	100.1	99%	98%	Spinocerebellar ataxia 5, 600224

				Spinocerebellar ataxia, autosomal recessive 14, 615386
SPTLC1	104.4	96%	90%	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	139.9	100%	100%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SQSTM1	102.7	94%	91%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 Paget disease of bone 3, 167250
SRC	99.7	100%	99%	Colon cancer, advanced, somatic, 114500 ?Thrombocytopenia 6,616937
SRCAP	123.0	98%	97%	Floating-Harbor syndrome, 136140
SRD5A2	71.9	100%	99%	Pseudovaginal perineoscrotal hypospadias, 264600
SRD5A3	133.5	100%	97%	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713
SRP72	61.4	92%	85%	Bone marrow failure syndrome 1, 614675
SRY	1.7	1%	1%	46XX sex reversal, 400045 46XY sex reversal, 400044
SSTR5	144.7	99%	97%	Somatostatin analog, resistance to
ST14	130.8	100%	98%	Ichthyosis, congenital, autosomal recessive 11, 602400
ST3GAL3	152.1	100%	100%	Epileptic encephalopathy, early infantile, 15, 615006 Mental retardation, autosomal recessive 12, 611090
ST3GAL5	109.5	94%	94%	Amish infantile epilepsy syndrome, 609056
STAC3	113.2	100%	98%	Native American myopathy, 255995
STAMBP	103.4	100%	97%	Microcephaly-capillary malformation syndrome, 614261
STAR	103.0	100%	100%	Lipoid adrenal hyperplasia, 201710
STAT1	122.1	97%	96%	Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 ncy 31B, mycobacterial and viral infections, autosomal recessive, 613796 Immunodeficiency 31C, autosomal dominant, 614162
STAT2	100.3	100%	100%	Immunodeficiency 44, 616636
STAT3	110.2	100%	97%	Autoimmune disease, multisystem, infantile-onset, 615952 Hyper-IgE recurrent infection syndrome, 147060
STAT5B	113.3	100%	95%	Growth hormone insensitivity with immunodeficiency, 245590 Leukemia, acute promyelocytic, somatic, 102578
STIL	140.8	98%	98%	Microcephaly 7, primary, autosomal recessive, 612703
STIM1	107.5	96%	92%	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 1 160565 Stormorken syndrome, 185070

STK11	112.5	100%	91%	Melanoma, malignant, somatic Pancreatic cancer, 260350 Peutz-Jeghers syndrome, 175200 Testicular tumor, somatic, 273300
STK4	133.0	100%	98%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STOX1	171.4	89%	89%	Preeclampsia/eclampsia 4, 609404
STRA6	107.1	100%	99%	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186
STRADA	119.6	100%	100%	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087
STRC	95.5	100%	96%	Deafness, autosomal recessive 16, 603720
STS	113.2	100%	99%	Ichthyosis, X-linked, 308100
STUB1	139.6	100%	100%	Spinocerebellar ataxia, autosomal recessive 16, 615768
STX11	249.5	100%	100%	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STX16	129.4	98%	96%	Pseudohypoparathyroidism, type IB, 603233
STX1B	142.2	100%	100%	Generalized epilepsy with febrile seizures plus, type 9, 616172
STXBP1	121.9	100%	100%	Epileptic encephalopathy, early infantile, 4, 612164
STXBP2	111.7	100%	99%	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
SUCLA2	54.7	93%	79%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	80.5	100%	89%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUFU	112.8	100%	97%	Basal cell nevus syndrome, 109400 Medulloblastoma, desmoplastic, 155255 {Meningioma, familial, susceptibility to}, 607174
SUMF1	103.2	97%	86%	Multiple sulfatase deficiency, 272200
SUMO1	12.4	57%	21%	Orofacial cleft 10, 613705
SUOX	171.4	100%	100%	Sulfite oxidase deficiency, 272300
SYCP3	86.5	95%	67%	Pregnancy loss, recurrent, 4, 270960 Spermatogenic failure 4, 270960
SYN1	87.5	99%	80%	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNE1	125.0	99%	98%	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 Spinocerebellar ataxia, autosomal recessive 8, 610743
SYNE2	108.7	97%	95%	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999
SYNE4	67.1	98%	90%	Deafness, autosomal recessive 76, 615540

SYNGAP1	63.3	93%	84%	Mental retardation, autosomal dominant 5, 612621
SYNJ1	114.6	99%	96%	Parkinson disease 20, early-onset, 615530
SYP	85.2	100%	99%	Mental retardation, X-linked 96, 300802
SYT14	147.1	87%	84%	Spinocerebellar ataxia, autosomal recessive 11, 614229
SYT2	89.4	100%	98%	Myasthenic syndrome, congenital, 7, presynaptic, 616040
SZT2	121.0	99%	99%	Epileptic encephalopathy, early infantile, 18, 615476
T	142.4	100%	97%	Sacral agenesis with vertebral anomalies, 615709 {Neural tube defects, susceptibility to}, 182940
TAB2	176.7	99%	95%	Congenital heart defects, nonsyndromic, 2, 614980
TAC3	75.4	83%	75%	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACO1	90.8	94%	90%	Mitochondrial complex IV deficiency, 220110
TACR3	136.4	100%	99%	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
TACSTD2	186.9	98%	94%	Corneal dystrophy, gelatinous drop-like, 204870
TAF1	140.0	100%	98%	Dystonia-Parkinsonism, X-linked, 314250 Mental retardation, X-linked, syndromic 33, 300966
TAF2	113.6	99%	93%	Mental retardation, autosomal recessive 40, 615599
TAL1	50.8	89%	62%	Leukemia, T-cell acute lymphocytic, somatic, 613065
TAL2	89.0	100%	100%	Leukemia, T-cell acute lymphocytic, somatic, 613065
TALDO1	112.8	100%	96%	Transaldolase deficiency, 606003
TANGO2	134.3	100%	100%	Metabolic encephalomyopathic crises,recurrent,with rhabdomyolysis,cardiac arrhythmias and neurodegeneration,616878
TAP1	9.3	38%	4%	Bare lymphocyte syndrome, type I, 604571
TAP2	8.8	29%	11%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571 Wegener-like granulomatosis
TAPBP	16.4	72%	26%	Bare lymphocyte syndrome, type I, 604571
TAPT1	84.6	88%	85%	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type,616897
TARDBP	130.7	100%	100%	Amyotrophic lateral sclerosis 10, with or without FTD, 612069 Frontotemporal lobar degeneration, TARDBP-related, 612069
TAT	103.4	100%	100%	Tyrosinemia, type II, 276600
TAZ	118.5	100%	100%	Barth syndrome, 302060
TBC1D20	128.0	94%	92%	Warburg micro syndrome 4, 615663

TBC1D24	144.6	100%	99%	Deafness , autosomal recessive 86, 614617 Deafness, autosomal dominant 65, 616044 DOOR syndrome, 220500 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021
TBC1D7	94.9	93%	92%	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000
TBCE	125.3	99%	98%	Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Kenny-Caffey syndrome, type 1, 244460
TBCK	82.2	97%	87%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3,616900
TBK1	95.5	97%	87%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439
TBL1XR1	79.9	93%	69%	Mental retardation, autosomal dominant 41,616944 Piermont syndrome,602342
TBP	88.5	100%	98%	Spinocerebellar ataxia 17, 607136 {Parkinson disease, susceptibility to}, 168600
TBX1	65.9	74%	61%	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Velocardiofacial syndrome, 192430
TBX15	105.8	100%	94%	Cousin syndrome, 260660
TBX18	84.2	97%	92%	Congenital anomalies of kidney and urinary tract 2, 143400
TBX19	134.5	100%	100%	Adrenocorticotropic hormone deficiency, 201400
TBX20	110.0	100%	100%	Atrial septal defect 4, 611363
TBX21	71.4	95%	84%	Asthma and nasal polyps, 208550 {Asthma, aspirin-induced, susceptibility to}, 208550
TBX22	142.7	100%	99%	Cleft palate with ankyloglossia, 303400 ?Abruzzo-Erickson syndrome, 302905
TBX3	76.7	97%	92%	Ulnar-mammary syndrome, 181450
TBX4	140.5	96%	94%	Ischiocoxopodopatellar syndrome, 147891
TBX5	119.2	100%	99%	Holt-Oram syndrome, 142900
TBX6	112.8	99%	87%	Spondylocostal dysostosis 5, 122600
TBXAS1	119.9	100%	100%	Ghosal hematodiaphyseal syndrome, 231095 ?Thromboxane synthase deficiency, 614158
TCAP	85.2	100%	100%	Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, type 2G, 601954

TCF12	136.1	100%	99%	Craniosynostosis 3, 615314
TCF3	67.8	99%	95%	Agammaglobulinemia 8,autosomal dominant,616941
TCF4	127.3	100%	99%	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954
TCIRG1	102.5	96%	89%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	155.3	100%	100%	Transcobalamin II deficiency, 275350
TCOF1	75.8	98%	96%	Treacher Collins syndrome 1, 154500
TCTN1	94.0	95%	91%	Joubert syndrome 13, 614173
TCTN2	123.8	99%	93%	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	110.4	99%	97%	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TDGF1	115.7	86%	81%	Forebrain defects, 187395
TDP1	100.5	94%	92%	Spinocerebellar ataxia, autosomal recessive with axonal neuropathy, 607250
TDP2	142.3	100%	100%	Spinocerebellar ataxia, autosomal recessive, 616949
TDRD7	149.8	99%	97%	Cataract 36, 613887
TEAD1	139.7	97%	93%	Sveinsson choreoretinal atrophy, 108985
TECPR2	128.9	100%	100%	Spastic paraplegia 49, autosomal recessive, 615031
TECR	75.1	99%	94%	Mental retardation, autosomal recessive 14, 614020
TECTA	184.7	100%	100%	Deafness, autosomal dominant 8/12, 601543 Deafness, autosomal recessive 21, 603629
TEK	168.9	100%	99%	Venous malformations, multiple cutaneous and mucosal, 600195
TELO2	86.0	98%	87%	You-Hoover-Fong syndrome, 616954
TENM3	172.2	99%	98%	Microphthalmia, isolated, with coloboma 9, 615145
TENM4	131.5	99%	98%	Tremor, hereditary essential, 5, 616736
TERC	NC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550 {Aplastic anemia}, 614743 {Pulmonary fibrosis, idiopathic, susceptibility to}, 614743
TET2	178.3	100%	100%	Myelodysplastic syndrome, somatic, 614286
TEX11	104.9	94%	86%	Spermatogenic failure, X-linked, 2, 309120
TF	114.6	100%	100%	Atransferrinemia, 209300
TFAP2A	97.1	100%	99%	Branchiooculofacial syndrome, 113620
TFAP2B	106.5	98%	95%	Char syndrome, 169100 Patent ductus arteriosus 2, 617035

TFE3	80.4	100%	98%	Renal cell carcinoma, papillary, 1, 300854
TFG	96.4	93%	92%	Hereditary motor and sensory neuropathy, Okinawa type, 604484 ?Spastic paraplegia 57, autosomal recessive, 615658
TFR2	84.2	98%	90%	Hemochromatosis, type 3, 604250
TFRC	146.3	100%	98%	Immunodeficiency 46, 616740
TG	116.7	99%	98%	Thyroid dyshormonogenesis 3, 274700 {Autoimmune thyroid disease, susceptibility to, 3}, 608175
TGDS	91.1	99%	97%	Catel-Manzke syndrome, 616145
TGFB1	73.8	100%	95%	Camurati-Engelmann disease, 131300 {Cystic fibrosis lung disease, modifier of}, 219700
TGFB2	140.0	100%	98%	Loeys-Dietz syndrome 4, 614816
TGFB3	138.7	100%	100%	Arrhythmogenic right ventricular dysplasia 1, 107970 Loeys-Dietz syndrome 5, 615582
TGFBI	118.1	97%	89%	Corneal dystrophy, Avellino type, 607541 Corneal dystrophy, epithelial basement membrane, 121820 Corneal dystrophy, Groenouw type I, 121900 Corneal dystrophy, lattice type I, 122200 Corneal dystrophy, lattice type IIIA, 608471 Corneal dystrophy, Reis-Bucklers type, 608470 Corneal dystrophy, Thiel-Behnke type, 602082
TGFBR1	169.6	94%	93%	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800
TGFBR2	165.3	100%	100%	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome 2, 610168
TGIF1	122.9	100%	100%	Holoprosencephaly-4, 142946
TGM1	144.1	100%	100%	Ichthyosis, congenital, autosomal recessive 1, 242300
TGM5	140.5	100%	100%	Peeling skin syndrome 2, 609796
TGM6	118.9	100%	94%	Spinocerebellar ataxia 35, 613908
TH	71.1	93%	89%	Segawa syndrome, recessive, 605407
THAP1	119.1	100%	100%	Dystonia 6, torsion, 602629
THBD	96.4	100%	93%	Thrombophilia due to thrombomodulin defect, 614486 {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
THOC2	94.5	98%	91%	Mental retardation, X-linked 12/35, 300957

THOC6	206.9	100%	100%	Beaulieu-Boycott-Innes syndrome, 613680
THPO	88.6	100%	100%	Thrombocythemia 1, 187950
THRA	159.3	100%	99%	Hypothyroidism, congenital, nongoitrous, 6, 614450
THRB	149.6	100%	100%	Thyroid hormone resistance, 188570 Thyroid hormone resistance, autosomal recessive, 274300 Thyroid hormone resistance, selective pituitary, 145650
TIA1	108.4	92%	83%	Welander distal myopathy, 604454
TIMM8A	42.4	93%	74%	Jensen syndrome, 311150 Mohr-Tranebjaerg syndrome, 304700
TIMP3	134.5	100%	100%	Sorsby fundus dystrophy, 136900
TINF2	159.6	100%	100%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TJP2	108.0	99%	98%	Cholestasis, progressive familial intrahepatic 4, 615878 Hypercholanemia, familial, 607748
TK2	85.5	92%	88%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560
TKT	104.7	98%	96%	Short stature, developmental delay, and congenital heart defects, 617044
TLE6	91.8	100%	98%	Preimplantation embryonic lethality, 616814
TLL1	129.5	100%	100%	Atrial septal defect 6, 613087
TMC1	119.9	96%	93%	Deafness, autosomal dominant 36, 606705 Deafness, autosomal recessive 7, 600974
TMC6	67.0	99%	95%	Epidermodysplasia verruciformis, 226400
TMC8	98.3	98%	89%	Epidermodysplasia verruciformis, 226400
TMCO1	86.9	100%	99%	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980
TMEM126A	101.9	98%	77%	Optic atrophy 7, 612989
TMEM126B	79.1	97%	95%	Mitochondrial complex I deficiency,252010
TMEM138	106.2	100%	100%	Joubert syndrome 16, 614465
TMEM165	97.2	99%	94%	Congenital disorder of glycosylation, type IIk, 614727
TMEM173	86.6	100%	97%	STING-associated vasculopathy, infantile-onset, 615934
TMEM199	112.3	100%	99%	Congenital disorder of glycosylation, type IIp, 616829
TMEM216	144.8	100%	100%	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	84.9	100%	96%	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	90.8	97%	97%	Joubert syndrome 14, 614424

TMEM240	108.2	100%	100%	Spinocerebellar ataxia 21, 607454
TMEM38B	104.6	100%	100%	Osteogenesis imperfecta, type XIV, 615066
TMEM43	107.2	100%	96%	Arrhythmogenic right ventricular dysplasia 5, 604400 Emery-Dreifuss muscular dystrophy 7, AD, 614302
TMEM5	125.4	91%	89%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
TMEM67	63.7	92%	81%	COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991
TMEM70	113.2	91%	83%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMEM98	116.0	99%	95%	Nanophthalmos 4, 615972
TMIE	97.0	97%	91%	Deafness, autosomal recessive 6, 600971
TMPRSS15	114.8	95%	85%	Enterokinase deficiency,226200
TMPRSS3	103.4	99%	97%	Deafness, autosomal recessive 8/10, 601072
TMPRSS6	93.0	100%	100%	Iron-refractory iron deficiency anemia, 206200
TNC	148.0	100%	98%	Deafness, autosomal dominant 56, 615629
TNFAIP3	106.8	100%	100%	Autoinflammatory syndrome, familial, Behcet-like, 616744
TNFRSF10B	111.0	100%	100%	Squamous cell carcinoma, head and neck, 275355
TNFRSF11A	121.3	92%	90%	Osteolysis, familial expansile, 174810 Osteopetrosis, autosomal recessive 7, 612301 {Paget disease of bone 2, early-onset}, 602080
TNFRSF11B	221.2	100%	100%	Paget disease of bone 5, juvenile-onset, 239000
TNFRSF13B	87.9	100%	100%	Immunodeficiency, common variable, 2, 240500 Immunoglobulin A deficiency 2, 609529
TNFRSF13C	45.5	91%	58%	Immunodeficiency, common variable, 4, 613494
TNFRSF1A	77.4	91%	86%	Periodic fever, familial, 142680 {Multiple sclerosis, susceptibility to, 5}, 614810
TNFSF11	138.9	100%	84%	Osteopetrosis, autosomal recessive 2, 259710
TNIK	105.1	100%	97%	Mental retardation, autosomal recessive 54, 617028
TNNC1	185.5	100%	100%	Cardiomyopathy, dilated, 1Z, 611879 Cardiomyopathy, hypertrophic, 13, 613243
TNNI2	121.0	100%	100%	Arthrogryposis multiplex congenita, distal, type 2B, 601680

TNNI3	88.5	94%	80%	Cardiomyopathy, dilated, 1FF, 613286 Cardiomyopathy, familial restrictive, 1, 115210 Cardiomyopathy, hypertrophic, 7, 613690 ?Cardiomyopathy, dilated, 2A, 611880
TNNT1	79.1	98%	96%	Nemaline myopathy 5, Amish type, 605355
TNNT2	87.2	100%	100%	Cardiomyopathy, dilated, 1D, 601494 Cardiomyopathy, familial restrictive, 3, 612422 Cardiomyopathy, hypertrophic, 2, 115195 Left ventricular noncompaction 6, 601494
TNNT3	99.9	99%	95%	Arthyrgryposis, distal, type 2B, 601680
TNPO3	127.9	100%	99%	Muscular dystrophy, limb-girdle, type 1F, 608423
TNXB	13.0	50%	24%	Ehlers-Danlos syndrome due to tenascin X deficiency, 606408 Vesicoureteral reflux 8, 615963
TOPORS	199.0	100%	100%	Retinitis pigmentosa 31, 609923
TOR1A	196.2	100%	98%	Dystonia-1, torsion, 128100 {Dystonia-1, modifier of}
TP53	89.1	97%	91%	Adrenal cortical carcinoma, 202300 Breast cancer, 114480 Choroid plexus papilloma, 260500 Colorectal cancer, 114500 Hepatocellular carcinoma, 114550 Li-Fraumeni syndrome, 151623 Nasopharyngeal carcinoma, 607107 Osteosarcoma, 259500 Pancreatic cancer, 260350 {Basal cell carcinoma 7}, 614740 {Glioma susceptibility 1}, 137800
TP63	170.5	100%	100%	ADULT syndrome, 103285 Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 Hay-Wells syndrome, 106260 Limb-mammary syndrome, 603543 Orofacial cleft 8, 129400 Rapp-Hodgkin syndrome, 129400 Split-hand/foot malformation 4, 605289
TPI1	82.2	99%	94%	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512

TPK1	106.4	100%	94%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TPM1	135.1	100%	99%	Cardiomyopathy, dilated, 1Y, 611878 Cardiomyopathy, hypertrophic, 3, 115196 Left ventricular noncompaction 9, 611878
TPM2	94.4	100%	97%	Arthrogryposis multiplex congenita, distal, type 1, 108120 Arthrogryposis, distal, type 2B, 601680 CAP myopathy 2, 609285 Nemaline myopathy 4, autosomal dominant, 609285
TPM3	75.2	86%	84%	CAP myopathy 1, 609284 Myopathy, congenital, with fiber-type disproportion, 255310 Nemaline myopathy 1, autosomal dominant or recessive, 609284
TPO	113.8	98%	92%	Thyroid dyshormonogenesis 2A, 274500
TPP1	130.9	100%	100%	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TPRN	61.2	83%	76%	Deafness, autosomal recessive 79, 613307
TRAC	170.5	100%	100%	Immunodeficiency 7, TCR-alpha/beta deficient, 615387
TRAF3IP1	63.4	94%	92%	Senior-Loken syndrome 9, 616629
TRAIP	115.8	100%	100%	Seckel syndrome 9, 616777
TRAPPC11	110.1	97%	94%	Muscular dystrophy, limb-girdle, type 2S, 615356
TRAPPC2	117.9	85%	73%	Spondyloepiphyseal dysplasia tarda, 313400
TRAPPC9	123.3	100%	100%	Mental retardation, autosomal recessive 13, 613192
TRDN	62.6	83%	68%	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441
TREM2	110.7	100%	99%	Nasu-Hakola disease, 221770
TREX1	216.3	100%	100%	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700
TRIM2	138.7	99%	95%	Charcot-Marie-Tooth disease, type 2R, 615490
TRIM32	114.5	100%	100%	Muscular dystrophy, limb-girdle, type 2H, 254110 ?Bardet-Biedl syndrome 11, 615988
TRIM37	106.6	99%	96%	Mulibrey nanism, 253250
TRIO	123.7	97%	94%	Mental retardation, autosomal dominant 44,617061
TRIOBP	102.5	97%	94%	Deafness, autosomal recessive 28, 609823
TRIP11	74.0	93%	83%	Achondrogenesis, type IA, 200600

TRIP4	99.7	100%	98%	Spinal muscular atrophy with congenital bone fractures 1,616866 ?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066
TRMT10A	121.7	100%	100%	Microcephaly, short stature, and impaired glucose metabolism 1, 616033
TRMT10C	90.7	99%	95%	Combined oxidative phosphorylation deficiency 30, 616974
TRMT5	189.1	94%	86%	Combined oxidative phosphorylation deficiency 26, 616539
TRMU	94.2	100%	100%	Liver failure, transient infantile, 613070 {Deafness, mitochondrial, modifier of}, 580000
TRNT1	91.9	95%	89%	Retinitis pigmentosa and erythrocytic microcytosis, 616959 Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084
TRPA1	88.2	87%	75%	Episodic pain syndrome, familial, 615040
TRPC6	108.9	98%	96%	Glomerulosclerosis, focal segmental, 2, 603965
TRPM1	141.7	99%	97%	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TRPM4	99.4	100%	97%	Progressive familial heart block, type IB, 604559
TRPM6	132.6	100%	98%	Hypomagnesemia 1, intestinal, 602014
TRPS1	160.1	100%	100%	Trichorhinophalangeal syndrome, type I, 190350 Trichorhinophalangeal syndrome, type III, 190351
TRPV3	140.7	100%	97%	Olmsted syndrome, 614594 ?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400
TRPV4	159.5	100%	99%	Brachyolmia type 3, 113500 Digital arthropathy-brachydactyly, familial, 606835 Hereditary motor and sensory neuropathy, type IIc, 606071 Metatropic dysplasia, 156530 Parastremmatic dwarfism, 168400 Scapuloperoneal spinal muscular atrophy, 181405 SED, Maroteaux type, 184095 Spinal muscular atrophy, distal, congenital nonprogressive, 600175 Spondylometaphyseal dysplasia, Kozlowski type, 184252 [Sodium serum level QTL 1], 613508
TSC1	120.4	99%	97%	Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100
TSC2	118.0	100%	100%	Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254
TSEN15	65.7	97%	84%	Pontocerebellar hypoplasia, type 2F, 617026
TSEN2	101.7	100%	99%	Pontocerebellar hypoplasia type 2B, 612389

TSEN34	49.3	87%	85%	Pontocerebellar hypoplasia type 2C, 612390
TSEN54	77.1	94%	90%	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753 ?Pontocerebellar hypoplasia type 5, 610204
TSFM	121.3	100%	97%	Combined oxidative phosphorylation deficiency 3, 610505
TSG101	85.4	98%	95%	Breast cancer, somatic, 114480
TSHB	281.4	100%	100%	Hypothryoidism, congenital, nongoitrous 4, 275100
TSHR	176.5	99%	94%	Hyperthyroidism, familial gestational, 603373 Hyperthyroidism, nonautoimmune, 609152 Hypothyroidism, congenital, nongoitrous, 1 275200 Thyroid adenoma, hyperfunctioning, somatic Thyroid carcinoma with thyrotoxicosis
TSHZ1	134.7	98%	98%	Aural atresia, congenital, 607842
TSPAN12	117.5	100%	99%	Exudative vitreoretinopathy 5, 613310
TSPAN7	139.5	100%	100%	Mental retardation, X-linked 58, 300210
TSPEAR	125.7	100%	99%	Deafness, autosomal recessive 98, 614861
TSPYL1	116.6	100%	95%	Sudden infant death with dysgenesis of the testes syndrome, 608800
TTBK2	131.2	100%	99%	Spinocerebellar ataxia 11, 604432
TTC19	82.0	85%	79%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTC21B	92.0	99%	97%	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
TTC25	81.9	100%	99%	Ciliary dyskinesia, primary, 35, 617092
TTC37	113.5	98%	95%	Trichohepatoenteric syndrome 1, 222470
TTC7A	95.7	99%	97%	Gastrointestinal defects and immunodeficiency syndrome, 243150
TTC8	89.5	97%	89%	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
TTI2	91.0	98%	95%	Mental retardation, autosomal recessive 39, 615541
TTLL5	131.1	98%	94%	Cone-rod dystrophy 19, 615860
TTN	171.1	98%	97%	Cardiomyopathy, dilated, 1G, 604145 Cardiomyopathy, familial hypertrophic, 9, 613765 Muscular dystrophy, limb-girdle, type 2J, 608807 Myopathy, early-onset, with fatal cardiomyopathy, 611705 Myopathy, proximal, with early respiratory muscle involvement, 603689 Tibial muscular dystrophy, tardive, 600334

TTPA	108.9	90%	78%	Ataxia with isolated vitamin E deficiency, 277460
TTR	170.6	100%	100%	Amyloidosis, hereditary, transthyretin-related, 105210 Carpal tunnel syndrome, familial, 115430 [Dystransthyretinemic hyperthyroxinemia], 145680
TUBA1A	73.8	100%	92%	Lissencephaly 3, 611603
TUBA4A	169.1	100%	100%	Amyotrophic lateral sclerosis 22 with or without frontotemoral dementia, 616208
TUBA8	129.0	100%	96%	Polymicrogyria with optic nerve hypoplasia, 613180
TUBB	16.5	67%	39%	Cortical dysplasia, complex, with other brain malformations 6, 615771 Symmetric circumferential skin creases, congenital, 1, 156610
TUBB1	128.1	100%	100%	Macrothrombocytopenia, autosomal dominant, TUBB1-related, 613112
TUBB2A	85.4	100%	96%	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB2B	85.1	100%	100%	Polymicrogyria, symmetric or asymmetric, 610031
TUBB3	160.9	98%	97%	Cortical dysplasia, complex, with other brain malformations 1, 614039 Fibrosis of extraocular muscles, congenital, 3A, 600638
TUBB4A	111.6	96%	96%	Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438
TUBB8	30.0	81%	59%	Oocyte maturation defect 2, 616780
TUBG1	127.6	100%	100%	Cortical dysplasia, complex, with other brain malformations 4, 615412
TUBGCP4	123.1	96%	93%	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TUBGCP6	134.6	100%	99%	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
TUFM	117.0	100%	99%	Combined oxidative phosphorylation deficiency 4, 610678
TULP1	84.0	97%	91%	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132
TUSC3	128.3	100%	99%	Mental retardation, autosomal recessive 7, 611093
TWIST1	119.9	90%	79%	Craniosynostosis, type 1, 123100 Robinow-Sorauf syndrome, 180750 Saethre-Chotzen syndrome with eyelid anomalies, 101400 Saethre-Chotzen syndrome, 101400
TWIST2	101.2	100%	98%	Ablepharon-macrostomia syndrome, 200110 Barber-Say syndrome, 209885 Focal facial dermal dysplasia 3, Setleis type, 227260
TXNL4A	108.6	100%	99%	Burn-McKeown syndrome, 608572
TYK2	98.8	100%	99%	Immunodeficiency 35, 611521
TYMP	76.8	98%	85%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041

TYR	162.3	100%	100%	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	68.6	100%	100%	Nasu-Hakola disease, 221770
TYRP1	170.1	100%	100%	Albinism, oculocutaneous, type III, 203290 [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271
UBA1	181.1	99%	98%	Spinal muscular atrophy, X-linked 2, infantile, 301830
UBE2A	123.4	100%	99%	Mental retardation, X-linked syndromic, Nascimento-type, 300860
UBE2T	106.2	100%	100%	Fanconi anemia, complementation group T, 616435
UBE3A	84.2	98%	92%	Angelman syndrome, 105830
UBE3B	115.2	100%	98%	Kaufman oculocerebrofacial syndrome, 244450
UBIAD1	199.8	100%	96%	Corneal dystrophy, Schnyder type, 121800
UBQLN2	145.1	99%	97%	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857
UBR1	116.3	99%	95%	Johanson-Blizzard syndrome, 243800
UGT1A1	188.5	100%	100%	Crigler-Najjar syndrome, type I, 218800 Crigler-Najjar syndrome, type II, 606785 Hyperbilirubinemia, familial transient neonatal, 237900 [Bilirubin, serum level of, QTL1], 601816 [Gilbert syndrome], 143500
UMOD	109.1	97%	96%	Glomerulocystic kidney disease with hyperuricemia and isosthenuria, 609886 Hyperuricemic nephropathy, familial juvenile 1, 162000 Medullary cystic kidney disease 2, 603860
UMPS	164.2	100%	100%	Orotic aciduria, 258900
UNC13D	85.4	98%	96%	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC80	115.8	99%	98%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801
UNG	60.3	95%	90%	Immunodeficiency with hyper IgM, type 5, 608106
UPB1	143.6	100%	100%	Beta-ureidopropionase deficiency, 613161
UPF3B	57.5	94%	85%	Mental retardation, X-linked, syndromic 14, 300676
UQCRB	90.7	99%	93%	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC2	124.5	100%	100%	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRQ	131.9	100%	100%	Mitochondrial complex III deficiency, nuclear type 4, 615159

UROD	135.0	100%	100%	Porphyria cutanea tarda, 176100 Porphyria, hepatoerythropoietic, 176100
UROS	91.7	100%	100%	Porphyria, congenital erythropoietic, 263700
USB1	112.9	100%	97%	Poikiloderma with neutropenia,604173
USH1C	92.2	100%	99%	Deafness, autosomal recessive 18A, 602092 Usher syndrome, type 1C, 276904
USH1G	147.7	98%	96%	Usher syndrome, type 1G, 606943
USH2A	136.1	100%	99%	Retinitis pigmentosa 39, 613809 Usher syndrome, type 2A, 276901
USP9X	142.0	99%	96%	Mental retardation, X-linked 99, 300919 Mental retardation, X-linked 99, syndromic, female-restricted, 300968
USP9Y	.2	0%	0%	Spermatogenic failure, Y-linked, 2, 415000
UVSSA	111.5	100%	99%	UV-sensitive syndrome 3, 614640
VAC14	104.5	100%	99%	Striatonigral degeneration,childhood-onset,617054
VAMP1	123.8	100%	100%	Spastic ataxia 1, autosomal dominant, 108600
VANGL1	148.7	100%	99%	Caudal regression syndrome, 600145 {Neural tube defects, susceptibility to}, 182940
VANGL2	140.1	99%	98%	Neural tube defects, 182940
VAPB	96.3	99%	92%	Amyotrophic lateral sclerosis 8, 608627 Spinal muscular atrophy, late-onset, Finkel type, 182980
VARS2	12.2	48%	18%	Combined oxidative phosphorylation deficiency 20, 615917
VCAN	164.8	100%	100%	Wagner syndrome 1, 143200
VCL	98.4	100%	99%	Cardiomyopathy, dilated, 1W, 611407 Cardiomyopathy, hypertrophic, 15, 613255
VCP	124.6	100%	99%	Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954 Charcot-Marie-Tooth disease, type 2Y, 616687 Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320
VDR	103.3	99%	95%	Rickets, vitamin D-resistant, type IIA, 277440 ?Osteoporosis, involutional, 166710
VEGFC	150.2	100%	97%	Lymphedema, hereditary, ID, 615907
VHL	95.9	92%	81%	Erythrocytosis, familial, 2, 263400 Hemangioblastoma, cerebellar, somatic Pheochromocytoma, 171300 Renal cell carcinoma, somatic, 144700

				von Hippel-Lindau syndrome, 193300
VIPAS39	132.1	100%	100%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VKORC1	124.5	100%	100%	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473 Warfarin resistance, 122700
VLDLR	191.3	100%	100%	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VMA21	52.2	98%	85%	Myopathy, X-linked, with excessive autophagy, 310440
VPS11	134.7	99%	98%	Leukodystrophy, hypomyelinating, 12, 616683
VPS13A	64.0	93%	81%	Choreoacanthocytosis, 200150
VPS13B	126.1	99%	96%	Cohen syndrome, 216550
VPS13C	100.6	96%	88%	Parkinson disease 23, autosomal recessive, early onset, 616840
VPS33B	123.6	100%	100%	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
VPS37A	63.8	86%	66%	Spastic paraplegia 53, autosomal recessive, 614898
VPS45	116.3	95%	94%	Neutropenia, severe congenital, 5, autosomal recessive, 615285
VPS53	118.9	90%	89%	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	117.3	99%	92%	Pontocerebellar hypoplasia type 1A, 607596
VSX1	52.7	77%	70%	Keratoconus 1, 148300 ?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195
VSX2	59.5	100%	92%	Microphthalmia with coloboma 3, 610092 Microphthalmia, isolated 2, 610093
VWF	96.5	98%	96%	von Willebrand disease, type 1, 193400 von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 von Willibrand disease, type 3, 277480
WAC	151.8	98%	97%	Desanto-Shinawi syndrome, 616708
WAS	67.0	88%	79%	Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, 313900 Thrombocytopenia, X-linked, intermittent, 313900 Wiskott-Aldrich syndrome, 301000
WDR11	120.8	96%	96%	Hypogonadotropic hypogonadism 14 with or without anosmia, 614858
WDR19	126.0	99%	96%	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378

				?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WDR34	89.3	100%	91%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
WDR35	138.1	99%	94%	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091
WDR36	104.6	96%	86%	Glaucoma 1, open angle, G, 609887
WDR45	97.2	97%	92%	Neurodegeneration with brain iron accululation 5, 300894
WDR60	96.0	97%	93%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WDR62	128.8	99%	98%	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
WDR72	131.1	99%	98%	Amelogenesis imperfecta, type IIA3, 613211
WDR73	116.6	100%	99%	Galloway-Mowat syndrome, 251300
WDR81	133.7	99%	98%	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185
WFS1	193.9	99%	98%	Deafness, autosomal dominant 6/14/38, 600965 Wolfram syndrome, 222300 Wolfram-like syndrome, autosomal dominant, 614296 ?Cataract 41, 116400 {Diabetes mellitus, noninsulin-dependent, association with}, 125853
WHSC1L1	113.4	99%	98%	Leukemia, acute myeloid, 601626
WISP3	96.0	100%	100%	Arthropathy, progressive pseudorheumatoid, of childhood, 208230 Spondyloepiphyseal dysplasia tarda with progressive arthropathy, 208230
WNK1	137.9	99%	97%	Neuropathy, hereditary sensory and autonomic, type II, 201300 Pseudohypoaldosteronism, type IIC, 614492
WNK4	102.4	99%	97%	Pseudohypoaldosteronism, type IIB, 614491
WNT1	174.9	100%	99%	Osteogenesis imperfecta, type XV, 615220 {Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221
WNT10A	99.1	100%	99%	Odontoonychodermal dysplasia, 257980 Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400
WNT10B	109.4	100%	100%	Split-hand/foot malformation 6, 225300
WNT4	204.8	92%	92%	Mullerian aplasia and hyperandrogenism, 158330 SERKAL syndrome, 611812
WNT5A	145.5	100%	100%	Robinow syndrome, autosomal dominant 1, 180700

WNT7A	175.2	100%	100%	Fuhrmann syndrome, 228930 Ulna and fibula, absence of, with severe limb deficiency, 276820
WRAP53	136.1	100%	100%	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	116.6	97%	93%	Werner syndrome, 277700
WT1	78.9	93%	85%	Denys-Drash syndrome, 194080 Frasier syndrome, 136680 Meacham syndrome, 608978 Mesothelioma, somatic, 156240 Nephrotic syndrome, type 4, 256370 Wilms tumor, type 1, 194070
WWOX	121.4	100%	100%	Epileptic encephalopathy, early infantile, 28, 616211 Esophageal squamous cell carcinoma, somatic, 133239 Spinocrebellar ataxia, autosomal recessive 12, 614322
XDH	93.7	100%	99%	Xanthinuria, type I, 278300
XIAP	125.8	91%	91%	Lymphoproliferative syndrome, X-linked, 2, 300635
XIST	NC	NC	NC	X-inactivation,familial skewed,300087
XK	115.7	100%	100%	McLeod syndrome with or without chronic granulomatous disease, 300842
XPA	46.1	92%	69%	Xeroderma pigmentosum, group A, 278700
XPC	123.2	100%	99%	Xeroderma pigmentosum, group C, 278720
XPNPEP3	116.3	97%	97%	Nephronophthisis-like nephropathy 1, 613159
XPR1	124.9	99%	97%	Basal ganglia calcification, idiopathic, 6, 616413
XRCC4	83.0	100%	91%	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	114.7	93%	88%	Desbuquois dysplasia 2, 615777 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
XYLT2	115.2	98%	95%	Spondyloocular syndrome, 605822 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
YAP1	84.4	83%	72%	Coloboma, ocular, 120433 Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433
YARS	112.2	100%	98%	Charcot-Marie-Tooth disease, dominant intermediate C, 608323
YARS2	144.8	100%	96%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
ZAP70	165.6	99%	98%	Autoimmune disease,multisystem,infantile-onset,2,617006 Immunodeficiency 48,269840
ZBTB16	128.0	100%	99%	Leukemia, acute promyelocytic, PL2F/RARA type

				Skeletal defects, genital hypoplasia, and mental retardation, 612447
ZBTB20	185.2	100%	100%	Primrose syndrome, 259050
ZBTB24	156.7	100%	100%	Immunodeficiency-centromeric instability-facial anomalies syndrome-2, 614069
ZC4H2	93.7	100%	99%	Wieacker-Wolff syndrome, 314580
ZDHHC9	80.9	100%	99%	Mental retardation, X-linked syndromic, Raymond type, 300799
ZEB1	170.3	100%	98%	Corneal dystrophy, Fuchs endothelial, 6, 613270 Corneal dystrophy, posterior polymorphous, 3, 609141
ZEB2	145.2	100%	99%	Mowat-Wilson syndrome, 235730
ZFP57	10.3	46%	9%	Diabetes mellitus, transient neonatal, 1, 601410
ZFPM2	175.8	100%	100%	46XY sex reversal 9, 616067 Diaphragmatic hernia 3, 610187 Tetralogy of Fallot, 187500
ZFYVE26	101.8	99%	98%	Spastic paraplegia 15, autosomal recessive, 270700
ZFYVE27	106.0	100%	100%	Spastic paraplegia 33, autosomal dominant, 610244
ZIC1	147.0	100%	100%	Craniosynostosis 6, 616602
ZIC2	87.0	92%	78%	Holoprosencephaly-5, 609637
ZIC3	125.7	100%	100%	Congenital heart defects, nonsyndromic, 1, X-linked, 306955 Heterotaxy, visceral, 1, X-linked 306955 VACTERL association, X-linked, 314390
ZMPSTE24	110.8	100%	97%	Mandibuloacral dysplasia with type B lipodystrophy, 608612 Restrictive dermopathy, lethal, 275210
ZMYND10	119.9	100%	100%	Ciliary dyskinesia, primary, 22, 615444
ZMYND11	112.1	100%	99%	Mental retardation, autosomal dominant 30, 616083
ZNF408	100.6	100%	100%	Retinitis pigmentosa 72, 616469 ?Exudative vitreoretinopathy 6, 616468
ZNF41	137.3	100%	100%	Mental retardation, X-linked 89, 300848
ZNF423	218.5	100%	100%	Joubert syndrome 19, 614844 Nephronophthisis 14, 614844
ZNF469	76.0	99%	96%	Brittle cornea syndrome 1, 229200
ZNF513	95.9	100%	100%	Retinitis pigmentosa 58, 613617
ZNF644	143.8	100%	99%	Myopia 21, autosomal dominant, 614167
ZNF674	152.0	100%	100%	Mental retardation, X-linked 92, 300851
ZNF687	135.8	100%	100%	Paget disease of bone 6, 616833

ZNF711	162.4	100%	99%	Mental retardation, X-linked 97, 300803
ZNF750	126.2	100%	99%	Seborrhea-like dermatitis with psoriasiform elements, 610227
ZNF81	127.1	100%	98%	Mental retardation, X-linked 45, 300498
ZP1	166.7	100%	100%	Oocyte maturation defect 1, 615774
ZSWIM6	137.9	95%	90%	Acromelic frontonasal dysostosis, 603671

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan;43(Database issue):D1079-85.
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 : October 1st, 2016.
This list is accurate for panel versions DG 2.7 and DG 2.8 From DG 2.7 to DG 2.8 no changes were made to the content of the gene panels.

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors