MENDELIOME GENE PANEL DG 2.9

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
A2M	134.5	99%	99%	Alpha-2-macroglobulin deficiency, 614036 {Alzheimer disease, susceptibility to}, 104300
A4GALT	165.1	100%	100%	NOR polyagglutination syndrome, 111400 [Blood group, P1Pk system, P phenotype], 111400 [Blood group, P1Pk system, p phenotype], 111400
AAAS	108.6	99%	99%	Achalasia-addisonianism-alacrimia syndrome, 231550
AAGAB	165.5	100%	100%	Keratoderma, palmoplantar, punctate type IA, 148600
AARS	145.1	100%	99%	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 Epileptic encephalopathy, early infantile, 29, 616339
AARS2	141.5	99%	99%	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889
AASS	169.7	100%	99%	Hyperlysinemia, 238700 Saccharopinuria, 268700
ABAT	103.8	100%	99%	GABA-transaminase deficiency, 613163
ABCA1	130.4	100%	99%	HDL deficiency, type 2, 604091 Tangier disease, 205400 {Coronary artery disease in familial hypercholesterolemia, protection against}, 143890
ABCA12	179.2	99%	98%	Ichthyosis, autosomal recessive 4B (harlequin), 242500 Ichthyosis, congenital, autosomal recessive 4A, 601277
ABCA3	141.2	99%	99%	Surfactant metabolism dysfunction, pulmonary, 3, 610921
ABCA4	138.6	99%	99%	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	185.5	99%	99%	Cholestasis, benign recurrent intrahepatic, 2, 605479 Cholestasis, progressive familial intrahepatic 2, 601847
ABCB4	159.6	99%	98%	Cholestasis, intrahepatic, of pregnancy, 3, 614972

				Cholestasis, progressive familial intrahepatic 3, 602347 Gallbladder disease 1, 600803
ABCB6	141.4	100%	99%	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	151.3	99%	98%	Anemia, sideroblastic, with ataxia, 301310
ABCC2	153.5	100%	99%	Dubin-Johnson syndrome, 237500
ABCC6	121.4	93%	92%	Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850
ABCC8	161.8	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	175	100%	99%	Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia, 239850
ABCD1	94.7	76%	69%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABCD4	152.7	99%	98%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	167.3	100%	99%	Sitosterolemia, 210250
ABCG8	166.5	99%	97%	Sitosterolemia, 210250 {Gallbladder disease 4}, 611465
ABHD12	113.2	98%	90%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ABHD5	256.2	100%	99%	Chanarin-Dorfman syndrome, 275630
ABL1	164.5	100%	100%	Leukemia, Philadelphia chromosome-positive, resistant to imatinib
ACAD8	145.6	100%	100%	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	157.1	99%	97%	Mitochondrial complex I deficiency due to ACAD9 deficiency, 611126
ACADM	135.5	99%	98%	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	149.2	99%	98%	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	139.7	98%	94%	2-methylbutyrylglycinuria, 610006
ACADVL	127.9	99%	97%	VLCAD deficiency, 201475

ACAN	136.5	92%	85%	Osteochondritis dissecans, short stature, and early-onset osteoarthritis, 165800 Spondyloepimetaphyseal dysplasia, aggrecan type, 612813 Spondyloepiphyseal dysplasia, Kimberley type, 608361
ACAT1	139	99%	97%	Alpha-methylacetoacetic aciduria, 203750
ACE	133.8	93%	92%	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	141.2	97%	94%	Infantile cerebellar-retinal degeneration, 614559 ?Optic atrophy 9, 616289
ACOX1	167.6	100%	100%	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACP5	231.8	100%	99%	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACSF3	146.1	100%	99%	Combined malonic and methylmalonic aciduria, 614265
ACSL4	128.1	97%	93%	Mental retardation, X-linked 63, 300387
ACSL6	140.6	99%	98%	Myelodysplastic syndrome Myelogenous leukemia, acute
ACTA1	104	99%	97%	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	140.7	100%	99%	Aortic aneurysm, familial thoracic 6, 611788 Moyamoya disease 5, 614042 Multisystemic smooth muscle dysfunction syndrome, 613834
ACTB	118.8	98%	94%	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ACTC1	161.2	100%	99%	Atrial septal defect 5, 612794 Cardiomyopathy, dilated, 1R, 613424 Cardiomyopathy, hypertrophic, 11, 612098 Left ventricular noncompaction 4, 613424
ACTG1	129.7	100%	100%	Baraitser-Winter syndrome 2, 614583 Deafness, autosomal dominant 20/26, 604717
ACTG2	147.5	99%	98%	Visceral myopathy, 155310

ACTN1	164.8	100%	99%	Bleeding disorder, platelet-type, 15, 615193
ACTN2	179.1	100%	100%	Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158 Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158
ACTN4	161.1	100%	99%	Glomerulosclerosis, focal segmental, 1, 603278
ACVR1	184.8	100%	99%	Fibrodysplasia ossificans progressiva, 135100
ACVR1B	185.9	99%	97%	Pancreatic cancer, somatic
ACVR2B	171.9	99%	95%	Heterotaxy, visceral, 4, autosomal, 613751
ACVRL1	138.6	100%	98%	Telangiectasia, hereditary hemorrhagic, type 2, 600376
ACY1	162.7	99%	98%	Aminoacylase 1 deficiency, 609924
ADA	128.2	99%	98%	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADAM10	158.7	99%	98%	Reticulate acropigmentation of Kitamura, 615537 {Alzheimer disease 18, susceptibility to}, 615590
ADAM9	168.8	99%	97%	Cone-rod dystrophy 9, 612775
ADAMTS10	124.3	99%	99%	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS13	119.3	96%	93%	Thrombotic thrombocytopenic purpura, familial, 274150
ADAMTS17	135.3	91%	88%	Weill-Marchesani-like syndrome, 613195
ADAMTS18	168.8	99%	99%	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
ADAMTS2	131.5	99%	98%	Ehlers-Danlos syndrome, type VIIC, 225410
ADAMTSL2	124.8	96%	91%	Geleophysic dysplasia 1, 231050
ADAMTSL4	108.6	99%	98%	Ectopia lentis et pupillae, 225200 Ectopia lentis, isolated, autosomal recessive, 225100
ADAR	136.8	100%	99%	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
ADAT3	82.8	99%	97%	Mental retardation, autosomal recessive 36, 615286
ADCK3	161	99%	99%	Coenzyme Q10 deficiency, primary, 4, 612016
ADCK4	104.6	100%	99%	Nephrotic syndrome, type 9, 615573
ADCY5	148.6	93%	91%	Dyskinesia, familial, with facial myokymia, 606703
ADD3	181.4	100%	99%	Cerebral palsy,spastic quadriplegic,3, 617008
ADGRG6	193.6	99%	99%	Lethal congenital contracture syndrome 9, 616503
ADIPOQ	135.5	100%	98%	Adiponectin deficiency, 612556
ADK	118.9	99%	100%	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADNP	276.9	100%	100%	Helsmoortel-van der Aa syndrome, 615873
ADRA2B	226.7	100%	100%	Epilepsy, myoclonic, familial adult, 2, 607876

ADSL	196.2	100%	90%	Adenylosuccinase deficiency, 103050
ADSSL1	138.7	96%	99%	Myopathy, distal, 5, 617030
AFF2	145.1	99%	98%	Mental retardation, X-linked, FRAXE type, 309548
AFF4	141.5	99%	87%	CHOPS syndrome, 616368
AFG3L2	124	94%	100%	Spastic ataxia 5, autosomal recessive, 614487 Spinocerebellar ataxia 28, 610246
AGA	175.8	100%	99%	Aspartylglucosaminuria, 208400
AGBL1	145.6	100%	99%	Corneal dystrophy, Fuchs endothelial, 8, 615523
AGBL5	126.7	100%	97%	Retinitis pigmentosa 75,617023
AGK	152.2	99%	99%	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AGL	184.8	99%	95%	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGPAT2	117.9	99%	91%	Lipodystrophy, congenital generalized, type 1, 608594
AGPS	69.6	98%	91%	Rhizomelic chondrodysplasia punctata, type 3, 600121
AGRN	130.4	96%	100%	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120
AGT	214.3	100%	100%	Renal tubular dysgenesis, 267430 {Hypertension, essential, susceptibility to}, 145500 {Preeclampsia, susceptibility to}
AGTR1	187.9	100%	100%	Renal tubular dysgenesis, 267430 {Hypertension, essential}, 145500
AGXT	168.5	100%	98%	Hyperoxaluria, primary, type 1, 259900
AHCY	132.6	100%	97%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHDC1	137.5	99%	98%	Xia-Gibbs syndrome, 615829
AHI1	176.9	99%	90%	Joubert syndrome-3, 608629
AICDA	149.7	98%	99%	Immunodeficiency with hyper-IgM, type 2, 605258
AIFM1	126	100%	94%	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Deafness, X-linked 5, 300614
AIMP1	109.1	99%	99%	Leukodystrophy, hypomyelinating, 3, 260600
AIP	161.9	100%	100%	Pituitary adenoma, ACTH-secreting, 219090 Pituitary adenoma, growth hormone-secreting, 102200 Pituitary adenoma, prolactin-secreting, 600634
AIPL1	133.9	100%	97%	Cone-rod dystrophy, 604393

				Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393
AIRE	83.4	99%	99%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AK1	141.8	99%	94%	Hemolytic anemia due to adenylate kinase deficiency, 612631
AK2	110.8	99%	90%	Reticular dysgenesis, 267500
AKR1C2	201.9	96%	95%	46XY sex reversal 8, 614279 Obesity, hyperphagia, and developmental delay
AKR1D1	131	99%	99%	Bile acid synthesis defect, congenital, 2, 235555
AKT1	182.3	100%	99%	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	172.9	100%	95%	Diabetes mellitus, type II, 125853 Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900
AKT3	97.6	99%	98%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937
ALAD	106.6	100%	95%	Porphyria, acute hepatic, 612740 {Lead poisoning, susceptibility to}, 612740
ALAS2	99.8	98%	99%	Anemia, sideroblastic, 1, 300751 Protoporphyria, erythropoietic, X-linked, 300752
ALB	204.6	100%	100%	Analbuminemia, 616000 [Dysalbuminemic hyperthyroxinemia], 615999
ALDH18A1	143.9	100%	90%	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	122.3	95%	99%	Microphthalmia, isolated 8, 615113
ALDH2	137.6	100%	99%	Alcohol sensitivity, acute, 610251 {Esophageal cancer, alcohol-related, susceptibility to} {Hangover, susceptibility to}, 610251 {Sublingual nitroglycerin, susceptibility to poor response to}
ALDH3A2	153.1	100%	99%	Sjogren-Larsson syndrome, 270200
ALDH4A1	137.2	100%	84%	Hyperprolinemia, type II, 239510

ALDH5A1	105.3	92%	99%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	143.6	100%	91%	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	88.5	97%	96%	Epilepsy, pyridoxine-dependent, 266100
ALDOA	199.4	99%	98%	Glycogen storage disease XII, 611881
ALDOB	168.3	99%	49%	Fructose intolerance, 229600
ALG1	57.5	53%	99%	Congenital disorder of glycosylation, type Ik, 608540
ALG11	189.4	100%	100%	Congenital disorder of glycosylation, type Ip, 613661
ALG12	171.3	100%	96%	Congenital disorder of glycosylation, type Ig, 607143
ALG13	104.2	99%	99%	Epileptic encephalopathy, early infantile, 36, 300884
ALG2	115.3	100%	99%	Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 ?Congenital disorder of glycosylation, type Ii, 607906
ALG3	116.7	100%	95%	Congenital disorder of glycosylation, type Id, 601110
ALG6	126.3	98%	95%	Congenital disorder of glycosylation, type Ic, 603147
ALG8	147.2	96%	99%	Congenital disorder of glycosylation, type Ih, 608104
ALG9	128.6	99%	99%	Congenital disorder of glycosylation, type Il, 608776 Gillessen-Kaesbach-Nishimura syndrome, 263210
ALMS1	208.7	99%	98%	Alstrom syndrome, 203800
ALOX12B	139.6	99%	99%	Ichthyosis, congenital, autosomal recessive 2, 242100
ALOXE3	131.6	100%	100%	Ichthyosis, congenital, autosomal recessive 3, 606545
ALPL	164.9	100%	99%	Hypophosphatasia, adult, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500 Odontohypophosphatasia, 146300
ALS2	192.8	99%	72%	Amyotrophic lateral sclerosis 2, juvenile, 205100 Primary lateral sclerosis, juvenile, 606353 Spastic paralysis, infantile onset ascending, 607225
ALX3	123.3	81%	94%	Frontonasal dysplasia 1, 136760
ALX4	145.9	99%	99%	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597 {Craniosynostosis 5, susceptibility to}, 615529
AMACR	173.1	100%	92%	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
AMBN	200.2	97%	97%	Amelogenesis imperfecta, type IF, 616270
AMELX	104.2	99%	98%	Amelogenesis imperfecta, type 1E, 301200

AMER1	102.8	99%	85%	Osteopathia striata with cranial sclerosis, 300373
AMH	46	97%	99%	Persistent Mullerian duct syndrome, type I, 261550
AMHR2	172.3	100%	76%	Persistent Mullerian duct syndrome, type II, 261550
AMN	69.7	89%	99%	Megaloblastic anemia-1, Norwegian type, 261100
AMPD1	151.5	100%	99%	Myopathy due to myoadenylate deaminase deficiency, 615511
AMPD2	153.4	100%	100%	Pontocerebellar hypoplasia, type 9, 615809 ?Spastic paraplegia 63, 615686
AMT	187.9	100%	99%	Glycine encephalopathy, 605899
ANG	175.1	100%	95%	Amyotrophic lateral sclerosis 9, 611895
ANGPTL3	113.1	99%	99%	Hypobetalipoproteinemia, familial, 2, 605019
ANK1	152.6	100%	100%	Spherocytosis, type 1, 182900
ANK2	168.6	100%	100%	Cardiac arrhythmia, ankyrin-B-related, 600919 Long QT syndrome 4, 600919
ANKH	124.8	100%	99%	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000
ANKK1	177.3	100%	95%	Dopamine receptor D2, reduced brain density of
ANKRD11	118.5	98%	85%	KBG syndrome, 148050
ANKRD26	99.3	93%	90%	Thrombocytopenia 2, 188000
ANKS6	95.2	93%	95%	Nephronophthisis 16, 615382
ANLN	165.7	97%	98%	Focal segmental glomerulosclerosis 8, 616032
ANO10	136.7	99%	98%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANO3	167.4	99%	98%	Dystonia 24, 615034
ANO5	168	99%	96%	Gnathodiaphyseal dysplasia, 166260 Miyoshi muscular dystrophy 3, 613319 Muscular dystrophy, limb-girdle, type 2L, 611307
ANO6	155.8	98%	96%	Scott syndrome, 262890
ANTXR1	144.4	98%	96%	GAPO syndrome, 230740 {Hemangioma, capillary infantile, susceptibility to}, 602089
ANTXR2	112.4	99%	99%	Hyaline fibromatosis syndrome, 228600
AP1S1	114.6	100%	68%	MEDNIK syndrome, 609313
AP1S2	77	81%	89%	Mental retardation, X-linked syndromic 5, 304340
AP2S1	145.7	90%	95%	Hypocalciuric hypercalcemia, familial, type III, 600740
AP3B1	128.1	99%	99%	Hermansky-Pudlak syndrome 2, 608233
AP4B1	163.4	100%	98%	Spastic paraplegia 47, autosomal recessive, 614066

AP4E1	125.7	99%	97%	Spastic paraplegia 51, autosomal recessive, 613744 Stuttering, familial persistent, 1, 184450
AP4M1	133.2	99%	70%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	83.2	76%	99%	Spastic paraplegia 52, autosomal recessive, 614067
AP5Z1	104.5	100%	99%	Spastic paraplegia 48, autosomal recessive, 613647
APC	186.4	100%	97%	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	190.1	99%	100%	Hypotrichosis 1, 605389
APOA1	106.6	100%	81%	Amyloidosis, 3 or more types, 105200 ApoA-I and apoC-III deficiency, combined Corneal clouding, autosomal recessive Hypoalphalipoproteinemia, 604091
APOA2	125.8	84%	100%	Apolipoprotein A-II deficiency {Hypercholesterolemia, familial, modifier of}, 143890
APOA5	170.4	100%	99%	Hyperchylomicronemia, late-onset, 144650 {Hypertriglyceridemia, susceptibility to}, 145750
APOB	217.5	99%	100%	Hypercholesterolemia, due to ligand-defective apo B, 144010 Hypobetalipoproteinemia, 615558
APOC2	120.1	100%	100%	Hyperlipoproteinemia, type Ib, 207750
APOC3	96.1	100%	90%	Apolipoprotein C-III deficiency, 614028
APOE	68.2	97%	86%	Alzheimer disease-2, 104310 Hyperlipoproteinemia, type III Lipoprotein glomerulopathy, 611771 Sea-blue histiocyte disease, 269600 {?Macular degeneration, age-related}, 603075 {Coronary artery disease, severe, susceptibility to}, 617347
APOPT1	90.9	87%	99%	Mitochondrial complex IV deficiency, 220110
APP	159.8	100%	99%	Alzheimer disease 1, familial, 104300 Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants, 605714

APRT	73.3	100%	92%	Adenine phosphoribosyltransferase deficiency, 614723
APTX	127.2	94%	97%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
AQP2	132.9	99%	97%	Diabetes insipidus, nephrogenic, 125800
AQP5	123.9	99%	90%	Palmoplantar keratoderma, Bothnian type, 600231
AR	98.2	95%	99%	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	172.3	99%	100%	Periventricular heterotopia with microcephaly, 608097
ARG1	181	100%	100%	Argininemia, 207800
ARHGAP26	175.3	100%	98%	Leukemia, juvenile myelomonocytic, somatic, 607785
ARHGAP31	141.8	99%	100%	Adams-Oliver syndrome 1, 100300
ARHGDIA	174.4	100%	93%	Nephrotic syndrome, type 8, 615244
ARHGEF6	154.5	97%	98%	Mental retardation, X-linked 46, 300436
ARHGEF9	99.5	99%	91%	Epileptic encephalopathy, early infantile, 8, 300607
ARID1A	158.4	95%	92%	Coffin-Siris syndrome 2, 614607
ARID1B	159.9	96%	97%	Coffin-Siris syndrome 1, 135900
ARL13B	117.5	99%	82%	Joubert syndrome 8, 612291
ARL2BP	82.9	90%	97%	Retinitis pigmentosa with or without situs inversus, 615434
ARL6	121.4	100%	90%	Bardet-Biedl syndrome 3, 600151 ?Retinitis pigmentosa 55, 613575 {Bardet-Biedl syndrome 1, modifier of}, 209900
ARMC4	135.4	91%	99%	Ciliary dyskinesia, primary, 23, 615451
ARMC5	145.7	100%	99%	ACTH-independent macronodular adrenal hyperplasia 2, 615954
ARSA	114.5	100%	94%	Metachromatic leukodystrophy, 250100
ARSB	137.9	98%	93%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARSE	107.3	98%	99%	Chondrodysplasia punctata, X-linked recessive, 302950
ARV1	171.6	100%	67%	Epileptic encephalopathy, early infantile, 38, 617020
ARX	38.3	81%	97%	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	147.1	99%	99%	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASB10	110.1	100%	93%	Glaucoma 1, open angle, F, 603383
ASCC1	166.4	96%	84%	Barrett esophagus/esophageal adenocarcinoma, 614266 ?Spinal muscular atrophy with congenital bone fractures 2,616867
ASCL1	208.2	95%	98%	Central hypoventilation syndrome, congenital, 209880 Haddad syndrome, 209880
ASL	128.8	100%	94%	Argininosuccinic aciduria, 207900
ASNS	108.7	98%	94%	Asparagine synthetase deficiency, 615574
ASPA	144.7	99%	96%	Canavan disease, 271900
ASPH	142.1	99%	96%	Traboulsi syndrome, 601552
ASPM	134.5	99%	97%	Microcephaly 5, primary, autosomal recessive, 608716
ASPSCR1	109.2	99%	90%	Alveolar soft-part sarcoma, 606243
ASS1	118.2	96%	98%	Citrullinemia, 215700
ASXL1	172.5	99%	99%	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ASXL3	170	99%	99%	Bainbridge-Ropers syndrome, 615485
ATCAY	165.5	100%	99%	Ataxia, cerebellar, Cayman type, 601238
ATF6	161.2	100%	99%	Achromatopsia 7, 616517
ATIC	142.5	99%	98%	AICA-ribosiduria due to ATIC deficiency, 608688
ATL1	183.7	99%	96%	Neuropathy, hereditary sensory, type ID, 613708 Spastic paraplegia 3A, autosomal dominant, 182600
ATL3	134.1	99%	96%	Neuropathy, hereditary sensory, type IF, 615632
ATM	132.3	99%	97%	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic Lymphoma, mantle cell, somatic T-cell prolymphocytic leukemia, somatic {Breast cancer, susceptibility to}, 114480
ATN1	135.7	99%	86%	Dentatorubro-pallidoluysian atrophy, 125370
ATOH7	99.2	93%	99%	Persistent hyperplastic primary vitreous, autosomal recessive, 221900
ATP13A2	129.3	100%	99%	Kufor-Rakeb syndrome, 606693 ?Ceroid lipofuscinosis, neuronal, 12, 606693
ATP1A2	219	100%	100%	Alternating hemiplegia of childhood, 104290

				Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481
ATP1A3	204.3	100%	100%	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235
ATP2A1	176.4	100%	99%	Brody myopathy, 601003
ATP2A2	198.5	100%	99%	Acrokeratosis verruciformis, 101900 Darier disease, 124200
ATP2C1	156.9	100%	97%	Hailey-Hailey disease, 169600
ATP6AP1	119.5	99%	99%	Immunodeficiency 47, 300972
ATP6V0A2	160.2	99%	99%	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250
ATP6V0A4	139.7	100%	100%	Renal tubular acidosis, distal, autosomal recessive, 602722
ATP6V1B1	197.9	100%	99%	Renal tubular acidosis with deafness, 267300
ATP6V1B2	168.1	100%	98%	Deafness,congenital,with onychodystrophy,autosomal dominant, 124480 Zimmerman-Laband syndrome 2, 616455
ATP7A	148.2	99%	99%	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATP7B	170.2	100%	94%	Wilson disease, 277900
ATP8B1	160.4	96%	99%	Cholestasis, benign recurrent intrahepatic, 243300 Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, progressive familial intrahepatic 1, 211600
ATPAF2	115.8	100%	98%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
ATR	175.1	99%	95%	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
ATRX	105.7	98%	100%	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Alpha-thalassemia/mental retardation syndrome, 301040 Mental retardation-hypotonic facies syndrome, X-linked, 309580
ATXN1	175.9	100%	97%	Spinocerebellar ataxia 1, 164400
ATXN10	158.1	99%	85%	Spinocerebellar ataxia 10, 603516
ATXN2	100.2	92%	93%	Spinocerebellar ataxia 2, 183090 {Amyotrophic lateral sclerosis, susceptibility to, 13}, 183090 {Parkinson disease, late-onset, susceptibility to}, 168600
ATXN3	116.6	98%	94%	Machado-Joseph disease, 109150

ATXN7	140.5	97%	96%	Spinocerebellar ataxia 7, 164500
ATXN8OS	999999		97%	Spinocerebellar ataxia 8, 608768
AUH	103	99%	96%	3-methylglutaconic aciduria, type I, 250950
AURKC	91.2	99%	72%	Spermatogenic failure 5,243060
AUTS2	125.8	97%	96%	Mental retardation, autosomal dominant 26,615834
AVP	60.1	94%	98%	Diabetes insipidus, neurohypophyseal, 125700
AVPR2	149.9	99%	99%	Diabetes insipidus, nephrogenic, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539
AXIN1	162.5	99%	100%	?Caudal duplication anomaly, 607864 Hepatocellular carcinoma, somatic, 114550
AXIN2	132.5	100%	90%	Colorectal cancer, somatic, 114500 Oligodontia-colorectal cancer syndrome, 608615
B2M	331.7	100%	71%	Immunodeficiency 43, 241600 ?Amyloidosis, familial visceral, 105200
B3GALNT2	141.5	93%	95%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B3GALT6	56.4	76%	92%	Ehlers-Danlos syndrome, progeroid type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B3GALTL	120.7	98%	100%	Peters-plus syndrome, 261540
B3GAT3	105.3	98%	96%	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B3GNT1	125.5	100%	99%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 13, 615287
B4GALNT1	167.7	99%	95%	Spastic paraplegia 26, autosomal recessive, 609195
B4GALT1	124.3	100%	100%	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	122.7	97%	95%	Ehlers-Danlos syndrome with short stature and limb anomalies, 130070
B9D2	126.9	100%	99%	Meckel syndrome 10, 614175
BAAT	143	98%	86%	Hypercholanemia, familial, 607748
BAG3	140.5	100%	98%	Cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954
BANF1	62.6	95%	85%	Nestor-Guillermo progeria syndrome, 614008
BAP1	153.1	99%	100%	Tumor predisposition syndrome, 614327
BAX	109.3	86%	99%	Colorectal cancer, somatic, 114500 T-cell acute lymphoblastic leukemia, somatic, 613065
BBS1	178.6	100%	100%	Bardet-Biedl syndrome 1, 209900
BBS10	195.4	100%	99%	Bardet-Biedl syndrome 10, 615987

BBS12	237	100%	98%	Bardet-Biedl syndrome 12, 615989
BBS2	201.1	100%	96%	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	153.5	99%	95%	Bardet-Biedl syndrome 4, 615982
BBS5	144	98%	95%	Bardet-Biedl syndrome 5, 615983
BBS7	156.1	99%	81%	Bardet-Biedl syndrome 7, 615984
BBS9	139.9	97%	99%	Bardet-Biedl syndrome 9, 615986
BCAP31	75.6	94%	99%	Deafness, dystonia, and cerebral hypomyelination, 300475
BCHE	214.8	100%	84%	Apnea, postanesthetic
BCKDHA	195.9	100%	99%	Maple syrup urine disease, type Ia, 248600
BCKDHB	147.9	93%	99%	Maple syrup urine disease, type Ib, 248600
BCKDK	184.6	100%	97%	Branched-chain ketoacid dehydrogenase kinase deficiency, 614923
BCL10	132	100%	98%	?Immunodeficiency 37, 616098 Lymphoma, MALT, somatic, 137245 {Lymphoma, follicular, somatic}, 605027 {Male germ cell tumor, somatic}, 273300, {Mesothelioma, somatic}, 156240 {Sezary syndrome, somatic},
BCL11A	150.4	98%	100%	Intellectual development disorder with persistence of fetal hemoglobin, 617101
BCL2	177	99%	100%	Leukemia/lymphoma, B-cell, 2
BCL7A	169.3	100%	97%	B-cell non-Hodgkin lymphoma, high-grade
BCMO1	176.4	100%	85%	Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300
BCOR	117.5	99%	100%	Microphthalmia, syndromic 2, 300166
BCR	123.7	89%	100%	Leukemia, acute lymphocytic, somatic, 613065 Leukemia, chronic myeloid, somatic, 608232
BCS1L	199	100%	94%	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BDNF	204.2	100%	97%	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	148.3	98%	96%	Spinocerebellar ataxia 31,117210
BEST1	159.3	99%	97%	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	119.9	99%	99%	Cataract 33, 611391
BFSP2	101.3	99%	44%	Cataract 12, multiple types, 611597
BGN	155.8	99%	99%	Meester-Loeys syndrome, 300989 Spondyloepimetaphyseal dysplasia, X-linked, 300106
BHLHA9	17.3	65%	97%	Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432 ?Camptosynpolydactyly, complex, 607539
BICD2	162.1	100%	99%	Spinal muscular atrophy, lower extremity-predominant, 2, AD, 615290
BIN1	120.9	99%	97%	Myopathy, centronuclear, autosomal recessive, 255200
BLK	134.8	100%	90%	Maturity-onset diabetes of the young, type 11, 613375
BLM	139.5	99%	95%	Bloom syndrome, 210900
BLNK	120.2	95%	96%	Agammaglobulinemia 4, 613502
BLOC1S3	56	99%	99%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	111.3	99%	99%	Hermansky-pudlak syndrome 9, 614171
BLVRA	155.1	100%	98%	Hyperbiliverdinemia, 614156
BMP1	168.1	100%	100%	Osteogenesis imperfecta, type XIII, 614856
BMP15	128.6	100%	100%	Ovarian dysgenesis 2, 300510 Premature ovarian failure 4, 300510
BMP2	203.5	100%	99%	Brachydactyly, type A2, 112600 {HFE hemochromatosis, modifier of}, 235200
BMP4	164.2	100%	97%	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625
BMPER	163.9	100%	99%	Diaphanospondylodysostosis, 608022
BMPR1A	115.4	99%	99%	Juvenile polyposis syndrome, infantile form, 174900 Polyposis syndrome, hereditary mixed, 2, 610069 Polyposis, juvenile intestinal, 174900
BMPR1B	195.7	100%	83%	Acromesomelic dysplasia, Demirhan type, 609441 Brachydactyly, type A1, D, 616849

				Brachydactyly, type A2, 112600
BMPR2	220.4	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	63.6	92%	82%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BPGM	127.2	100%	98%	Erythrocytosis due to bisphosphoglycerate mutase deficiency, 222800
BRAF	86.4	91%	98%	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	121.6	99%	95%	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BRCA2	118.6	99%	98%	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	112.5	99%	94%	Cerebellofaciodental syndrome, 616202
BRIP1	149	99%	100%	Breast cancer, early-onset, 114480 Fanconi anemia, complementation group J, 609054
BRWD3	122.6	98%	100%	Mental retardation, X-linked 93, 300659
BSCL2	129.8	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	180.6	100%	99%	Bartter syndrome, type 4a, 602522 Sensorineural deafness with mild renal dysfunction, 602522
BTD	168.1	100%	99%	Biotinidase deficiency, 253260
BTK	139.7	100%	98%	Agammaglobulinemia and isolated hormone deficiency, 307200 Agammaglobulinemia, X-linked 1, 300755

BUB1	172.6	100%	99%	Colorectal cancer with chromosomal instability, somatic
BUB1B	158.2	98%	100%	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430
C10orf11	165.3	99%	88%	Albinism, oculocutaneous, type VII, 615179
C10orf2	192.9	100%	99%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Perrault syndrome 5, 616138 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286
C11orf73	85.5	95%	93%	Leukodystrophy,hypomyelinating,13,616881
C12orf57	150	100%	99%	Temtamy syndrome, 218340
C12orf65	94.5	98%	99%	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035
C15orf41	147	100%	98%	Dyserythropoietic anemia, congenital, type Ib, 615631
C19orf12	110.3	100%	98%	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
C1GALT1C1	167.1	99%	100%	Tn polyagglutination syndrome, somatic, 300622
C1QA	147.6	100%	99%	C1q deficiency, 613652
C1QB	225.6	100%	78%	C1q deficiency, 613652
C1QC	215.1	100%	99%	C1q deficiency, 613652
C1QTNF5	175.6	91%	41%	Retinal degeneration, late-onset, autosomal dominant, 605670
C1S	138	100%	96%	C1s deficiency, 613783
C2	20.1	82%	98%	C2 deficiency, 217000 {Macular degeneration, age-related, 14, reduced risk of}, 615489
C21orf59	164.8	99%	99%	Ciliary dyskinesia, primary, 26, 615500
C2orf71	137.3	99%	46%	Retinitis pigmentosa 54, 613428
C3	167.5	100%	43%	C3 deficiency, 613779 {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 {Macular degeneration, age-related, 9}, 611378
C4A	22.1	76%	100%	C4a deficiency, 614380 [Blood group, Rodgers], 614374
C4B	20.5	73%	96%	C4B deficiency, 614379
C4orf26	260.2	100%	97%	Amelogenesis imperfecta, type IIA4, 614832
C5	149.2	98%	99%	C5 deficiency, 609536

				[Eculizumab, poor response to], 615749
C5orf42	154.1	99%	96%	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
C6	185.4	100%	93%	C6 deficiency, 612446 Combined C6/C7 deficiency
C7	159	99%	99%	C7 deficiency, 610102
C7orf10	153.9	96%	99%	Glutaric aciduria III, 231690
C8A	134.3	100%	99%	C8 deficiency, type I, 613790
C8B	154.8	100%	99%	C8 deficiency, type II, 613789
C8orf37	149.6	100%	98%	Cone-rod dystrophy 16, 614500 Retinitis pigmentosa 64, 614500
C9	169.7	100%	99%	C9 deficiency, 613825 {Macular degeneration, age-related, 15, susceptibility to}, 615591
C9orf72	125	100%	97%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1, 105550
CA12	107.8	100%	99%	Hyperchlorhidrosis, isolated, 143860
CA2	178.8	99%	96%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA4	176.1	99%	93%	Retinitis pigmentosa 17, 600852
CA5A	131.6	99%	92%	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CA8	131.4	97%	97%	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CABP2	96.1	98%	92%	Deafness, autosomal recessive 93, 614899
CABP4	116.6	99%	99%	Cone-rod synaptic disorder, congenital nonprogressive, 610427
CACNA1A	104.4	95%	99%	Episodic ataxia, type 2, 108500 Migraine, familial hemiplegic, 1, 141500 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Spinocerebellar ataxia 6, 183086
CACNA1C	182.7	99%	97%	Brugada syndrome 3, 611875 Timothy syndrome, 601005
CACNA1D	178.3	100%	98%	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896
CACNA1F	105.9	99%	96%	Aland Island eye disease, 300600 Cone-rod dystrophy, X-linked, 3, 300476 Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071
CACNA1G	152.3	99%	99%	Spinocerebellar ataxia 42, 616795
CACNA1H	125.8	98%	97%	Hyperaldosteronism, familial, type IV, 617027

				{Epilepsy, childhood absence, susceptibility to, 6}, 611942 {Epilepsy, idiopathic generalized, susceptibility to, 6}, 611942
CACNA1S	155.5	100%	98%	Hypokalemic periodic paralysis, type 1, 170400 {Malignant hyperthermia susceptibility 5}, 601887 {Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580
CACNA2D4	119.3	98%	97%	Retinal cone dystrophy 4, 610478
CACNB2	165.2	99%	99%	Brugada syndrome 4, 611876
CACNB4	134.7	99%	100%	Episodic ataxia, type 5, 613855 {Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 {Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682
CACNG2	131.8	100%	66%	Mental retardation, autosomal dominant 10, 614256
CALM1	154.6	100%	95%	Long QT syndrome 14, 616247 Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916
CALM2	65.8	68%	99%	Long QT syndrome 15, 616249
CALR	113.7	99%	99%	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
CAMTA1	203.2	100%	100%	Cerebellar ataxia, nonprogressive, with mental retardation, 614756
CANT1	165.9	100%	98%	Desbuquois dysplasia 1, 251450
CAPN1	171.6	100%	99%	Spastic paraplegia 76, autosomal recessive, 616907
CAPN3	126.3	99%	97%	Muscular dystrophy, limb-girdle, type 2A, 253600
CAPN5	185.4	100%	98%	Vitreoretinopathy, neovascular inflammatory, 193235
CARD11	176.4	99%	96%	B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11, 615206
CARD14	130.2	99%	99%	Pityriasis rubra pilaris, 173200 Psoriasis 2, 602723
CARD9	129.1	99%	96%	Candidiasis, familial, 2, autosomal recessive, 212050
CARS2	132.6	100%	95%	Combined oxidative phosphorylation deficiency 27, 616672
CASC5	129.5	98%	97%	Microcephaly 4, primary, autosomal recessive, 604321
CASK	112.2	99%	99%	FG syndrome 4, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 Mental retardation, with or without nystagmus, 300422
CASP10	127.9	99%	99%	Autoimmune lymphoproliferative syndrome, type II, 603909 Gastric cancer, somatic, 613659 Lymphoma, non-Hodgkin, somatic, 605027

CASP8	172.1	100%	100%	Hepatocellular carcinoma, somatic, 114550 ?Autoimmune lymphoproliferative syndrome, type IIB, 607271 {Breast cancer, protection against}, 114480 {Lung cancer, protection against}, 211980
CASQ1	128	100%	99%	Myopathy, vacuolar, with CASQ1 aggregates, 616231
CASQ2	168.6	100%	95%	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CASR	192.1	99%	100%	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	136.7	98%	99%	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295
CAT	179.4	100%	100%	Acatalasemia, 614097
CATSPER1	141.3	100%	100%	Spermatogenic failure 7, 612997
CAV1	282.6	100%	98%	Pulmonary hypertension, primary, 3, 615343 ?Lipodystrophy, congenital generalized, type 3, 612526 ?Partial lipodystrophy, congenital cataracts, and neurodegeneration syndrome, 606721
CAV3	294	100%	94%	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	146	99%	99%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia,607785
CBS	134.1	98%	97%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CC2D1A	141.1	100%	94%	Mental retardation, autosomal recessive 3, 608443
CC2D2A	144.7	99%	99%	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284
CCBE1	79.9	99%	97%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CCDC103	118.3	100%	99%	Ciliary dyskinesia, primary, 17, 614679

CCDC11	185.9	99%	89%	Heterotaxy, visceral, 6, autosomal recessive, 614779
CCDC114	132	100%	99%	Ciliary dyskinesia, primary, 20, 615067
CCDC115	94.5	91%	95%	Congenital disorder of glycosylation, type IIo, 616828
CCDC151	134	100%	92%	Ciliary dyskinesia, primary, 30, 616037
CCDC174	133.9	99%	96%	Hypotonia, infantile, with psychomotor retardation, 616816
CCDC22	109.1	98%	98%	Ritscher-Schinzel syndrome 2, 300963
CCDC39	112.3	99%	96%	Ciliary dyskinesia, primary, 14, 613807
CCDC40	135.7	99%	96%	Ciliary dyskinesia, primary, 15, 613808
CCDC41	126.7	99%	99%	Nephronophthisis 18, 615862
CCDC65	101.2	99%	100%	Ciliary dyskinesia, primary, 27, 615504
CCDC78	120.3	100%	94%	Myopathy, centronuclear, 4, 614807
CCDC8	117.6	100%	98%	3-M syndrome 3, 614205
CCDC88A	109.5	98%	97%	PEHO syndrome, 260565
CCDC88C	119.9	99%	100%	Hydrocephalus, nonsyndromic, autosomal recessive, 236600 ?Spinocerebellar ataxia 40, 616053
CCM2	178.9	98%	96%	Cerebral cavernous malformations-2,603284
CCND2	169.6	100%	99%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938
CCNO	101.5	99%	100%	Ciliary dyskinesia, primary, 29, 615872
CCT5	171.1	99%	99%	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CD151	154.1	100%	99%	Nephropathy with pretibial epidermolysis bullosa and deafness, 609057 [Blood group, Raph], 179620
CD19	105.5	100%	97%	Immunodeficiency, common variable, 3, 613493
CD27	138.1	100%	98%	Lymphoproliferative syndrome 2, 615122
CD2AP	115.6	99%	98%	Glomerulosclerosis, focal segmental, 3, 607832
CD320	103.9	99%	100%	Methylmalonic aciduria due to transcobalamin receptor defect, 613646
CD36	150.9	99%	99%	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	211.9	100%	100%	Immunodeficiency 19, 615617
CD3E	171.6	100%	98%	Immunodeficiency 18, 615615 Immunodeficiency 18, SCID variant, 615615
CD3G	195.3	100%	99%	Immunodeficiency 17, CD3 gamma deficient, 615607

CD4	117.8	99%	90%	OKT4 epitope deficiency, 613949
CD40	196.1	100%	88%	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	150.6	98%	98%	Immunodeficiency, X-linked, with hyper-IgM, 308230
CD59	211.7	94%	100%	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD79A	155.7	99%	99%	Agammaglobulinemia 3, 613501
CD79B	268.6	100%	99%	Agammaglobulinemia 6, 612692
CD81	168.9	100%	99%	Immunodeficiency, common variable, 6, 613496
CD8A	117.9	99%	97%	CD8 deficiency, familial, 608957
CD96	204.8	100%	96%	C syndrome, 211750
CDAN1	108.7	98%	95%	Dyserythropoietic anemia, congenital, type Ia, 224120
CDC14A	194.4	99%	97%	Deafness,autosomal recessive 105,616958
CDC42	114.2	99%	99%	Takenouchi-Kosaki syndrome, 616737
CDC45	183.8	99%	98%	Meier-Gorlin syndrome 7, 617063
CDC6	172.4	100%	99%	?Meier-Gorlin syndrome 5, 613805
CDC73	122.3	99%	99%	Hyperparathyroidism, familial primary, 145000 Hyperparathyroidism-jaw tumor syndrome, 145001 Parathyroid adenoma with cystic changes, 145001 Parathyroid carcinoma, 608266
CDCA7	129.5	99%	97%	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910
CDH1	129.3	99%	99%	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	145.3	99%	98%	Mental retardation, autosomal dominant 3, 612580
CDH23	225.8	99%	98%	Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D, 601067 Usher syndrome, type 1D/F digenic, 601067
CDH3	164.1	99%	99%	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 Hypotrichosis, congenital, with juvenile macular dystrophy, 601553
CDHR1	168.7	99%	96%	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660
CDK5RAP2	142.1	99%	99%	Microcephaly 3, primary, autosomal recessive, 604804
CDKL5	135.2	99%	63%	Epileptic encephalopathy, early infantile, 2, 300672

CDKN1B	99	99%	92%	Multiple endocrine neoplasia, type IV, 610755
CDKN1C	37.4	77%	99%	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732
CDKN2A	80	93%	40%	Melanoma and neural system tumor syndrome, 155755 Orolaryngeal cancer, multiple, Pancreatic cancer/melanoma syndrome, 606719 {Melanoma, cutaneous malignant, 2}, 155601
CDON	147.2	100%	94%	Holoprosencephaly 11, 614226
CDSN	20.4	58%	99%	Hypotrichosis 2, 146520 Peeling skin syndrome 1, 270300
CDT1	106	98%	71%	Meier-Gorlin syndrome 4, 613804
CEACAM16	160.2	100%	99%	Deafness, autosomal dominant 4B, 614614
CEBPA	55.7	86%	98%	Leukemia, acute myeloid, somatic, 601626 ?Leukemia, acute myeloid, 601626
CEBPE	84.5	100%	83%	Specific granule deficiency, 245480
CECR1	106	99%	98%	Polyarteritis nodosa, childhood-onset, 615688 ?Sneddon syndrome, 182410
CEL	147	85%	99%	Maturity-onset diabetes of the young, type VIII, 609812
CENPF	159.3	99%	97%	Stromme syndrome, 243605
CENPJ	171.4	99%	99%	?Seckel syndrome 4, 613676 Microcephaly 6, primary, autosomal recessive, 608393
CEP104	150.1	99%	96%	Joubert syndrome 25, 616781
CEP120	163	100%	97%	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP152	204.7	98%	100%	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823
CEP164	100.3	99%	87%	Nephronophthisis 15, 614845
CEP19	245.1	100%	93%	Morbid obesity and spermatogenic failure, 615703
CEP290	92.8	95%	94%	Joubert syndrome 5, 610188 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Senior-Loken syndrome 6, 610189 ?Bardet-Biedl syndrome 14, 615991
CEP41	98.2	98%	96%	Joubert syndrome 15, 614464
CEP57	115.1	99%	99%	Mosaic variegated aneuploidy syndrome 2, 614114

CERKL	121.8	99%	98%	Retinitis pigmentosa 26, 608380
CERS3	143.1	100%	100%	Ichthyosis, congenital, autosomal recessive 9, 615023
CES1	145	99%	69%	Carboxylesterase 1 deficiency
CETP	152.4	100%	87%	Hyperalphalipoproteinemia, 143470 [High density lipoprotein cholesterol level QTL 10], 143470
CFC1	78.2	81%	97%	Heterotaxy, visceral, 2, autosomal, 605376
CFD	76.4	95%	95%	Complement factor D deficiency, 613912
CFH	216.6	99%	97%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814 {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 {Macular degeneration, age-related, 4}, 610698
CFHR5	127.2	98%	87%	Nephropathy due to CFHR5 deficiency, 614809
CFI	198	98%	95%	Complement factor I deficiency, 610984 {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 {Macular degeneration, age-related, 13, susceptibility to}, 615439
CFL2	134.5	95%	97%	Nemaline myopathy 7, autosomal recessive, 610687
CFP	109.2	99%	100%	Properdin deficiency,X-linked, 312060
CFTR	158.3	99%	87%	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	193.5	100%	42%	Mental retardation, autosomal dominant 40, 616579
CHAT	146.3	92%	90%	Myasthenic syndrome, congenital, 6, presynaptic, 254210
CHCHD10	27.8	65%	99%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 Spinal muscular atrophy, Jokela type, 615048 ?Myopathy, isolated mitochondrial, autosomal dominant, 616209
CHCHD2	93	97%	99%	Parkinson disease 22, autosomal dominant, 616710
CHD2	145.9	99%	80%	Epileptic encephalopathy, childhood-onset, 615369
CHD7	168.4	100%	97%	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CHEK2	115	84%	90%	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	102.6	99%	99%	Muscular dystrophy, congenital, megaconial type, 602541
CHM	123.8	97%	95%	Choroideremia, 303100
CHMP1A	127.1	100%	98%	Pontocerebellar hypoplasia, type 8, 614961
CHMP2B	112.3	99%	97%	Amyotrophic lateral sclerosis 17, 614696 Dementia, familial, nonspecific, 600795
CHMP4B	158.5	99%	99%	Cataract 31, multiple types, 605387
CHN1	176.8	99%	99%	Duane retraction syndrome 2, 604356
CHRDL1	125.3	99%	100%	Megalocornea 1, X-linked 309300
CHRNA1	134.9	100%	96%	Multiple pterygium syndrome, lethal type, 253290 Myasthenic syndrome, congenital, 1A, slow-channel, 601462 Myasthenic syndrome, congenital, 1B, fast-channel, 608930
CHRNA2	229.9	100%	97%	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA4	169.5	97%	98%	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction, susceptibility to}, 188890
CHRNB1	163.8	99%	99%	Myasthenic syndrome, congenital, 2A, slow-channel, 616313 ?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314
CHRNB2	250.6	99%	97%	Epilepsy, nocturnal frontal lobe, 3, 605375
CHRND	185.4	99%	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	140.3	99%	94%	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	198.1	100%	98%	Escobar syndrome, 265000 Multiple pterygium syndrome, lethal type, 253290
CHST14	194.6	96%	100%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	107.5	99%	94%	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHST6	338.2	100%	98%	Macular corneal dystrophy, 217800
CHSY1	142.1	96%	99%	Temtamy preaxial brachydactyly syndrome, 605282
CHUK	146.6	99%	99%	Cocoon syndrome, 613630
CIB2	235.2	100%	100%	Deafness, autosomal recessive 48, 609439

				Usher syndrome, type IJ, 614869
CIITA	140.9	100%	83%	Bare lymphocyte syndrome, type II, complementation group A, 209920 {Rheumatoid arthritis, susceptibility to}, 180300
CIRH1A	202.1	100%	99%	Cirrhosis, North American Indian childhood type, 604901
CISD2	149.4	83%	97%	Wolfram syndrome 2, 604928
CITED2	122.5	100%	97%	Atrial septal defect 8, 614433 Ventricular septal defect 2, 614431
CKAP2L	219	99%	99%	Filippi syndrome, 272440
CLCF1	94.3	99%	99%	Cold-induced sweating syndrome 2, 610313
CLCN1	163.7	100%	99%	Myotonia congenita, dominant, 160800 Myotonia congenita, recessive, 255700 Myotonia levior, recessive
CLCN2	116.5	99%	98%	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	131	100%	97%	Mental retardation, X-linked 49/15, 300114
CLCN5	149.8	99%	95%	Dent disease, 300009 Hypophosphatemic rickets, 300554 Nephrolithiasis, type I, 310468 Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990
CLCN7	156.7	99%	94%	Osteopetrosis, autosomal dominant 2, 166600 Osteopetrosis, autosomal recessive 4, 611490
CLCNKA	122.4	99%	100%	Bartter syndrome, type 4b, digenic, 613090
CLCNKB	113.9	99%	100%	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090
CLDN1	153.7	100%	100%	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626
CLDN14	132.4	100%	96%	Deafness, autosomal recessive 29, 614035
CLDN16	171.5	100%	100%	Hypomagnesemia 3, renal, 248250
CLDN19	144.2	99%	99%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLEC7A	177.5	100%	96%	Candidiasis, familial, 4, autosomal recessive, 613108 {Aspergillosis, susceptibility to}, 614079
CLMP	116.8	99%	97%	Congenital short bowel syndrome, 615237
CLN3	133	99%	94%	Ceroid lipofuscinosis, neuronal, 3, 204200

CLN5	155.4	99%	100%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	144.8	98%	100%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	273.8	100%	96%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLP1	185.2	100%	97%	Pontocerebellar hypoplasia, type 10, 615803
CLPB	153.8	96%	99%	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CLPP	140.5	99%	100%	Perrault syndrome 3, 614129
CLRN1	171.9	100%	85%	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902
CNBP	152.3	100%	99%	Myotonic dystrophy 2,602668
CNGA1	149.9	90%	97%	Retinitis pigmentosa 49, 613756
CNGA3	177.3	99%	92%	Achromatopsia-2, 216900
CNGB1	115.4	99%	99%	Retinitis pigmentosa 45, 613767
CNGB3	129.7	97%	98%	Achromatopsia-3, 262300 Macular degeneration, juvenile, 248200
CNNM2	228.4	100%	98%	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNNM4	212.9	98%	99%	Jalili syndrome, 217080
CNTNAP1	189.6	99%	92%	Lethal congenital contracture syndrome 7, 616286
CNTNAP2	168.7	100%	100%	Cortical dysplasia-focal epilepsy syndrome, 610042 Pitt-Hopkins like syndrome 1, 610042 {Autism susceptibility 15}, 612100
COA6	76.8	98%	99%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 4, 616501
COASY	173	100%	99%	Neurodegeneration with brain iron accumulation 6, 615643
COCH	215.4	100%	99%	Deafness, autosomal dominant 9, 601369
COG1	129.4	100%	95%	Congenital disorder of glycosylation, type IIg, 611209
COG4	139.6	100%	90%	Congenital disorder of glycosylation, type IIj, 613489
COG5	135.2	99%	100%	Congenital disorder of glycosylation, type IIi, 613612
COG6	103.3	97%	98%	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328
COG7	139.6	100%	97%	Congenital disorder of glycosylation, type IIe, 608779
COG8	118.4	99%	92%	Congenital disorder of glycosylation, type IIh, 611182
COL10A1	116.3	99%	23%	Metaphyseal chondrodysplasia, Schmid type, 156500

COL11A1	112.4	96%	98%	Fibrochondrogenesis 1, 228520 Marshall syndrome, 154780 Stickler syndrome, type II, 604841 {Lumbar disc herniation, susceptibility to}, 603932
COL11A2	14.6	59%	96%	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	167.3	99%	96%	Bethlem myopathy 2, 616471 ?Ullrich congenital muscular dystrophy 2, 616470
COL13A1	96.6	99%	89%	Myasthenic syndrome, congenital, 19, 616720
COL17A1	118.8	98%	96%	Epidermolysis bullosa, junctional, localisata variant, 226650 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epithelial recurrent erosion dystrophy, 122400
COL18A1	94.8	96%	94%	Knobloch syndrome, type 1, 267750
COL1A1	146	98%	97%	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	118.4	97%	99%	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	149.4	99%	93%	Fibrosis of extraocular muscles, congenital, 5, 616219
COL2A1	119.3	99%	94%	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	124.9	98%	96%	Ehlers-Danlos syndrome, type IV, 130050
COL4A1	103.5	98%	95%	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	111.5	99%	98%	Porencephaly 2, 614483 {Hemorrhage, intracerebral, susceptibility to}, 614519
COL4A3	104.5	98%	95%	Alport syndrome, autosomal dominant, 104200 Alport syndrome, autosomal recessive, 203780 Hematuria, benign familial, 141200
COL4A3BP	153.5	99%	81%	Mental retardation, autosomal dominant 34, 616351
COL4A4	97.9	98%	95%	Alport syndrome, autosomal recessive, 203780 Hematuria, familial benign
COL4A5	61	93%	97%	Alport syndrome, 301050
COL5A1	131.5	98%	99%	Ehlers-Danlos syndrome, classic type, 130000
COL5A2	106.4	99%	99%	Ehlers-Danlos syndrome, classic type, 130000
COL6A1	160.6	99%	99%	Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090
COL6A2	179.4	100%	98%	Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090 ?Myosclerosis, congenital, 255600

COL6A3	201.8	100%	78%	Bethlem myopathy 1, 158810 Dystonia 27, 616411 Ullrich congenital muscular dystrophy 1, 254090
COL7A1	148.7	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	42.4	90%	94%	Corneal dystrophy, Fuchs endothelial, 1, 136800 Corneal dystrophy, posterior polymorphous 2, 609140
COL9A1	143.9	99%	91%	Stickler syndrome, type IV, 614134 /?Epiphyseal dysplasia, multiple, 6, 614135
COL9A2	77.1	99%	99%	Epiphyseal dysplasia, multiple, 2, 600204 ?Stickler syndrome, type V, 614284 {Intervertebral disc disease, susceptibility to}, 603932
COL9A3	78.3	97%	99%	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 {Intervertebral disc disease, susceptibility to}, 603932
COLEC11	221.8	100%	93%	3MC syndrome 2, 265050
COLQ	123.1	99%	93%	Myasthenic syndrome, congenital, 5, 603034
COMP	143.3	96%	84%	Epiphyseal dysplasia, multiple, 1, 132400 Pseudoachondroplasia, 177170
COQ2	92.8	96%	97%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ4	103.5	87%	96%	Coenzyme Q10 deficiency, primary, 7, 616276
COQ6	155.4	99%	99%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ9	94.2	99%	96%	Coenzyme Q10 deficiency, primary, 5, 614654
CORIN	197.7	99%	99%	Preeclampsia/eclampsia 5, 614595
CORO1A	187	99%	98%	Immunodeficiency 8, 615401
COX10	250.6	100%	81%	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 Mitochondrial complex IV deficiency, 220110
COX15	103.8	100%	99%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119

				Leigh syndrome due to cytochrome c oxidase deficiency, 256000
COX20	60.1	95%	99%	Mitochondrial complex IV deficiency, 220110
COX4I2	123.1	100%	100%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX6A1	208.3	99%	56%	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
COX6B1	186.7	100%	91%	Mitochondrial complex IV deficiency, 220110
COX7B	53.1	83%	99%	Linear skin defects with multiple congenital anomalies, 300887
CP	151.7	94%	98%	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 [Hypoceruloplasminemia, hereditary], 604290
CPA6	153.2	100%	87%	Epilepsy, familial temporal lobe, 5, 614417 Febrile seizures, familial, 11, 614418
CPN1	122.6	99%	100%	Carboxypeptidase N deficiency, 212070
CPOX	136.6	93%	98%	Coproporphyria, 121300 Harderoporphyria, 121300
CPS1	183.3	100%	96%	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}
CPT1A	189.9	99%	99%	CPT deficiency, hepatic, type IA, 255120
CPT2	182	98%	97%	CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 CPT II deficiency, myopathic, stress-induced 255110 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212
CR2	192	100%	100%	Immunodeficiency, common variable, 7, 614699 {Systemic lupus erythematosus, susceptibility to, 9}, 610927
CRADD	131	99%	97%	Mental retardation, autosomal recessive 34, with variant lissencephaly 614499
CRB1	232	100%	98%	Leber congenital amaurosis 8, 613835 Pigmented paravenous chorioretinal atrophy, 172870 Retinitis pigmentosa-12, autosomal recessive, 600105
CRB2	116.5	99%	95%	Focal segmental glomerulosclerosis 9, 616220 Ventriculomegaly with cystic kidney disease, 219730
CRBN	175.6	100%	96%	Mental retardation, autosomal recessive 2, 607417
CREB1	153.1	99%	95%	Histiocytoma, angiomatoid fibrous, somatic, 612160
CREBBP	141.3	98%	88%	Rubinstein-Taybi syndrome, 180849
CRELD1	113.4	99%	90%	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217

				{Atrioventricular septal defect, susceptibility to, 2}, 606217
CRIPT	45.8	98%	97%	Short stature with microcephaly and distinctive facies, 615789
CRLF1	127.2	91%	96%	Cold-induced sweating syndrome 1, 272430
CRTAP	132.5	99%	99%	Osteogenesis imperfecta, type VII, 610682
CRTC1	160.1	99%	93%	Mucoepidermoid salivary gland carcinoma
CRX	147.6	100%	96%	Cone-rod retinal dystrophy-2, 120970 Leber congenital amaurosis 7, 613829
CRYAA	158.8	97%	99%	Cataract 9, multiple types, 604219
CRYAB	120.6	99%	100%	Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, 2, 608810 Myopathy, myofibrillar, fatal infantile hypertrophy, alpha-B crystallin-related, 613869
CRYBA1	142.5	100%	99%	Cataract 10, multiple types, 600881
CRYBA4	131.7	100%	100%	Cataract 23, 610425
CRYBB1	141.9	100%	99%	Cataract 17, multiple types, 611544
CRYBB2	175.3	100%	97%	Cataract 3, multiple types, 601547
CRYBB3	180.7	100%	99%	Cataract 22, autosomal recessive, 609741
CRYGB	106.7	100%	99%	Cataract 39, multiple types, autosomal dominant, 615188
CRYGC	136.8	100%	91%	Cataract 2, multiple types, 604307
CRYGD	112	100%	97%	Cataract 4, multiple types, 115700
CRYGS	131.8	97%	98%	Cataract 20, multiple types, 116100
CRYM	100	99%	88%	Deafness, autosomal dominant 40, 616357
CSF1R	147.8	99%	97%	Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
CSF2RA	68.1	90%	93%	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF2RB	108.4	99%	89%	Surfactant metabolism dysfunction, pulmonary, 5, 614370
CSNK1D	173.6	97%	98%	Advanced sleep-phase syndrome, familial, 2, 615224
CSNK2A1	144.7	95%	99%	Okur-Chung neurodevelopmental syndrome, 617062 Glaucoma, primary congenital (Lee (2011) Mol Vis 17,3583)
CSPP1	133.7	99%	79%	Joubert syndrome 21, 615636
CSRP3	113.2	100%	99%	Cardiomyopathy, hypertrophic, 12, 612124 ?Cardiomyopathy, dilated, 1M, 607482
CST3	116.1	93%	99%	Cerebral amyloid angiopathy, 105150 {Macular degeneration, age-related, 11}, 611953
CSTA	135.2	100%	99%	Peeling skin syndrome 4, 607936

CSTB	108.7	99%	97%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTC1	122.4	99%	84%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTCF	166.6	99%	99%	Mental retardation, autosomal dominant 21, 615502
CTDP1	120.5	91%	87%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTH	206.8	100%	100%	Cystathioninuria, 219500 Homocysteine, total plasma, elevated
CTHRC1	119.4	93%	99%	Barrett esophagus/esophageal adenocarcinoma, 614266
CTLA4	212.7	100%	99%	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	155.3	99%	100%	Macular dystrophy,patterned,608970 Gastric cancer, diffuse (Majewski (2012) J Pathol epub)
CTNNA3	167.9	100%	99%	Arrhythmogenic right ventricular dysplasia, familial, 13, 615616
CTNNB1	199.2	100%	99%	Colorectal cancer, somatic, 114500 Hepatocellular carcinoma, somatic, 114550 Mental retardation, autosomal dominant 19, 615075 Ovarian cancer, somatic, 167000 Pilomatricoma, somatic, 132600
CTNS	142.7	100%	99%	Cystinosis, atypical nephropathic, 219800 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750
CTPS1	159.6	100%	100%	Immunodeficiency 24, 615897
CTSA	153.2	99%	98%	Galactosialidosis, 256540
CTSC	142.5	100%	79%	Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000 Periodontitis 1, juvenile, 170650
CTSD	197.2	99%	99%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	119	87%	98%	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362
CTSK	118.2	100%	97%	Pycnodysostosis, 265800
CUBN	134	99%	92%	Megaloblastic anemia-1, Finnish type, 261100
CUL3	149.7	99%	98%	Pseudohypoaldosteronism, type IIE, 614496
CUL4B	88.6	98%	99%	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354

CUL7	166.4	99%	100%	3-M syndrome 1, 273750
CXCR4	182.7	100%	98%	Myelokathexis, isolated WHIM syndrome, 193670
CYB5A	141	100%	80%	Methemoglobinemia, type IV, 250790
CYB5R3	181.8	98%	99%	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYBA	101.8	88%	89%	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690
CYBB	132.1	99%	97%	Chronic granulomatous disease, X-linked, 306400 Immunodeficiency 34, mycobacteriosis, X-linked, 300645
CYC1	189.9	96%	95%	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYCS	89.9	99%	97%	Thrombocytopenia 4, 612004
CYLD	132.2	99%	99%	Brooke-Spiegler syndrome, 605041 Cylindromatosis, familial, 132700 Trichoepithelioma, multiple familial, 1, 601606
CYP11A1	142.4	99%	99%	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	187.7	100%	99%	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 Aldosteronism, glucocorticoid-remediable, 103900
CYP11B2	190.1	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	143.7	100%	99%	17,20-lyase deficiency, isolated, 202110 17-alpha-hydroxylase/17,20-lyase deficiency, 202110
CYP19A1	200.3	100%	32%	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300
CYP1B1	129.2	100%	99%	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 Peters anomaly, 604229
CYP21A2	15.8	56%	99%	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910
CYP24A1	194.4	100%	98%	Hypercalcemia, infantile, 143880
CYP26B1	188	100%	96%	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
CYP26C1	96.5	99%	99%	Focal facial dermal dysplasia 4, 614974
CYP27A1	206.9	98%	99%	Cerebrotendinous xanthomatosis, 213700
CYP27B1	147.5	100%	97%	Vitamin D-dependent rickets, type I, 264700

CYP2A6	175.8	100%	94%	Coumarin resistance, 122700 {Lung cancer, resistance to}, 211980 {Nicotine addiction, protection from}, 188890
CYP2C8	121.9	99%	93%	Rhabdomyolysis, cerivastatin-induced
CYP2R1	156	98%	99%	Rickets due to defect in vitamin D 25-hydroxylation, 600081
CYP2U1	162.9	95%	99%	Spastic paraplegia 56, autosomal recessive, 615030
CYP4F22	134.6	100%	92%	Ichthyosis, congenital, autosomal recessive 5, 604777
CYP4V2	193.8	100%	96%	Bietti crystalline corneoretinal dystrophy, 210370
CYP7B1	129.5	97%	100%	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, autosomal recessive, 270800
D2HGDH	153.2	98%	95%	D-2-hydroxyglutaric aciduria, 600721
DAG1	236.1	100%	99%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818
DARS	129	99%	100%	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2	155.1	100%	96%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBH	173.8	100%	97%	Dopamine beta-hydroxylase deficiency, 223360 [Dopamine-beta-hydroxylase activity levels, plasma]
DBT	138.6	99%	99%	Maple syrup urine disease, type II, 248600
DCAF17	125.5	99%	99%	Woodhouse-Sakati syndrome, 241080
DCC	160.1	100%	99%	Colorectal cancer, somatic, 114500 Esophageal carcinoma, somatic 133239 Mirror movements 1, 157600
DCDC2	180	100%	95%	Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212
DCHS1	172.3	99%	95%	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390
DCLRE1C	158.5	98%	99%	Omenn syndrome, 603554 Severe combined immunodeficiency, Athabascan type, 602450
DCN	167.6	95%	98%	Corneal dystrophy, congenital stromal, 610048
DCPS	174.5	100%	99%	Al-Raqad syndrome, 616459
DCTN1	142.6	99%	99%	Neuropathy, distal hereditary motor, type VIIB, 607641 Perry syndrome, 168605 {Amyotrophic lateral sclerosis, susceptibility to}, 105400
DCX	123.8	100%	97%	Lissencephaly, X-linked, 300067

				Subcortical laminal heteropia, X-linked, 300067
DDB2	181.7	100%	95%	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DDC	121.6	99%	98%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	168.6	97%	99%	Spastic paraplegia 28, autosomal recessive, 609340
DDHD2	175.7	99%	74%	Spastic paraplegia 54, autosomal recessive, 615033
DDR2	172.2	100%	95%	Spondylometaepiphyseal dysplasia, short limb-hand type, 271665
DDX11	104.7	79%	98%	Warsaw breakage syndrome, 613398
DDX3X	109.8	98%	99%	Mental retardation, X-linked 102, 300958
DDX58	147.7	99%	84%	Singleton-Merten syndrome 2, 616298
DDX59	200.6	100%	99%	Orofaciodigital syndrome V, 174300
DEAF1	147.6	90%	99%	Mental retardation, autosomal dominant 24, 615828
DEPDC5	164.8	99%	99%	Epilepsy, familial focal, with variable foci, 604364
DES	145.8	99%	99%	Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 ?Muscular dystrophy, limb-girdle, type 2R, 615325
DFNA5	123.3	99%	97%	Deafness, autosomal dominant 5, 600994
DFNB59	168.9	100%	99%	Deafness, autosomal recessive 59, 610220
DGKE	160	99%	100%	Nephrotic syndrome, type 7, 615008 {Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008
DGUOK	141.9	99%	100%	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHCR24	205.7	100%	94%	Desmosterolosis, 602398
DHCR7	173	100%	81%	Smith-Lemli-Opitz syndrome, 270400
DHDDS	103.9	96%	99%	Retinitis pigmentosa 59, 613861
DHFR	61.8	95%	99%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHH	134.9	100%	98%	46XY partial gonadal dysgenesis, with minifascicular neuropathy, 607080 46XY sex reversal 7, 233420
DHODH	102.8	100%	99%	Miller syndrome, 263750
DHTKD1	156.7	99%	98%	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DIABLO	248.1	100%	86%	Deafness, autosomal dominant 64, 614152
DIAPH1	131.4	99%	95%	Deafness, autosomal dominant 1, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632
DIAPH2	79.1	95%	98%	Premature ovarian failure, 300511

DIAPH3	91.2	99%	99%	Auditory neuropathy, autosomal dominant, 1, 609129
DICER1	172	99%	99%	Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800 Pleuropulmonary blastoma, 601200 Rhabdomyosarcoma, embryonal, 2, 180295
DIP2B	183.3	99%	98%	Mental retardation, FRA12A type, 136630
DIS3L2	185.3	100%	98%	Perlman syndrome, 267000
DKC1	122.5	100%	99%	Dyskeratosis congenita, X-linked, 305000
DLAT	104.8	99%	98%	Pyruvate dehydrogenase E2 deficiency, 245348
DLC1	211.3	99%	94%	Colorectal cancer, somatic, 114500
DLD	157	99%	83%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLG3	102.3	99%	98%	Mental retardation, X-linked 90, 300850
DLL3	82.5	92%	96%	Spondylocostal dysostosis 1, autosomal recessive, 277300
DLL4	207	99%	98%	Adams-Oliver syndrome 6, 616589
DLX3	128.9	99%	97%	Amelogenesis imperfecta, type IV, 104510 Trichodontoosseous syndrome, 190320
DMD	140.4	99%	99%	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200
DMGDH	188.5	99%	98%	Dimethylglycine dehydrogenase deficiency, 605850
DMP1	151.2	99%	99%	Hypophosphatemic rickets, AR, 241520
DMPK	132.8	99%	97%	Myotonic dystrophy 1, 160900
DNA2	161.2	99%	97%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156 ?Seckel syndrome 8, 615807
DNAAF1	125.5	100%	94%	Ciliary dyskinesia, primary, 13, 613193
DNAAF2	113.2	99%	99%	Ciliary dyskinesia, primary, 10, 612518
DNAAF3	95.4	98%	99%	Ciliary dyskinesia, primary, 2, 606763
DNAH11	163.9	99%	98%	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH5	151.6	99%	92%	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAI1	133.4	99%	99%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	166	96%	78%	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJB2	126.6	100%	94%	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881
DNAJB6	68.1	91%	98%	Muscular dystrophy, limb-girdle, type 1E, 603511
DNAJC19	115.5	98%	99%	3-methylglutaconic aciduria, type V, 610198

DNAJC21	150.2	99%	98%	Bone marrow failure syndrome 3, 617052
DNAJC5	212	100%	92%	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350
DNAJC6	187	99%	100%	Parkinson disease 19, juvenile-onset, 615528
DNAL1	105.1	98%	90%	Ciliary dyskinesia, primary, 16, 614017
DNASE1L3	161.2	100%	98%	Systemic lupus erythematosus 16, 614420
DNM1	167.4	94%	95%	Epileptic encephalopathy, early infantile, 31, 616346
DNM1L	143.6	100%	98%	Encephalopahty, lethal, due to defective mitochondrial peroxisomal fission, 614388
DNM2	145	99%	96%	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	128.6	99%	99%	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 Neuropathy, hereditary sensory, type IE, 614116
DNMT3A	127.7	99%	99%	Tatton-Brown-Rahman syndrome, 615879
DNMT3B	143.6	99%	97%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK2	165.9	100%	96%	Immunodeficiency 40, 616433
DOCK6	134.7	99%	99%	Adams-Oliver syndrome 2, 614219
DOCK7	147.1	98%	92%	Epileptic encephalopathy, early infantile, 23, 615859
DOCK8	148.8	100%	99%	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
DOK7	93.6	94%	100%	Myasthenic syndrome, congenital, 10, 254300 ?Fetal akinesia deformation sequence, 208150
DOLK	189.5	100%	99%	Congenital disorder of glycosylation, type Im, 610768
DPAGT1	119.1	100%	87%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPH1	180	100%	96%	Developmental delay with short stature,dysmorphic features and sparse hair,616901
DPM1	149.2	92%	100%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	99	99%	95%	Congenital disorder of glycosylation, type Iu, 615042
DPM3	181.4	100%	68%	Congenital disorder of glycosylation, type Io, 612937
DPP6	157.8	98%	95%	Mental retardation, autosomal dominant 33, 616311 {Ventricular fibrillation, paroxysmal familial, 2}, 612956
DPY19L2	96.6	73%	98%	Spermatogenic failure 9, 613958
DPYD	190.3	97%	100%	5-fluorouracil toxicity, 274270 Dihydropyrimidine dehydrogenase deficiency, 274270
DPYS	144.7	99%	98%	Dihydropyrimidinuria, 222748

DRAM2	164.6	100%	69%	Cone-rod dystrophy 21, 616502
DRC1	108	99%	99%	Ciliary dyskinesia, primary, 21, 615294
DRD4	79.8	81%	97%	Autonomic nervous system dysfunction [Novelty seeking personality], 601696 {Attention deficit-hyperactivity disorder}, 143465
DRD5	112.3	100%	97%	Dystonia, primary cervical {Attention deficit-hyperactivity disorder, susceptibility to}, 143465 {Blepharospasm, primary benign}, 606798
DSC2	158.7	99%	99%	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 Arrhythmogenic right ventricular dysplasia 11, 610476
DSG1	182.9	98%	97%	Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508 Keratosis palmoplantaris striata I, AD, 148700
DSG2	149	99%	99%	Arrhythmogenic right ventricular dysplasia 10, 610193 Cardiomyopathy, dilated, 1BB, 612877
DSG4	228.3	99%	99%	Hypotrichosis 6, 607903
DSP	161.7	100%	99%	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	150.9	99%	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	201.8	99%	97%	Epidermolysis bullosa simplex, autosomal recessive 2, 615425 ?Neuropathy, hereditary sensory and autonomic, type VI, 614653
DTNA	173.4	100%	94%	Left ventricular noncompaction 1, with or without congenital heart defects, 604169
DTNBP1	127.3	99%	99%	Hermansky-Pudlak syndrome 7, 614076
DUOX2	157	96%	99%	Thryoid dyshormonogenesis 6, 607200
DUOXA2	137.5	100%	96%	Thyroid dyshormonogenesis 5, 274900
DUSP6	172.7	100%	100%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
DVL1	119	99%	96%	Robinow syndrome, autosomal dominant 2, 616331
DVL3	167.6	100%	99%	Robinow syndrome, autosomal dominant 3,616894

DYM	116.7	97%	91%	Dyggve-Melchior-Clausen disease, 223800 Smith-McCort dysplasia, 607326
DYNC1H1	189.4	100%	98%	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600
DYNC2H1	110.6	98%	99%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYNC2LI1	122.4	99%	93%	Short-rib thoracic dysplasia 15 with polydactyly, 617088
DYRK1A	168	100%	99%	Mental retardation, autosomal dominant 7, 614104
DYRK1B	110.1	97%	96%	Abdominal obesity-metabolic syndrome 3, 615812
DYSF	155.1	100%	97%	Miyoshi muscular dystrophy 1, 254130 Muscular dystrophy, limb-girdle, type 2B, 253601 Myopathy, distal, with anterior tibial onset, 606768
DYX1C1	101.6	99%	96%	Ciliary dyskinesia, primary, 25, 615482 {Dyslexia, susceptibility to, 1}, 127700
EARS2	110.3	99%	97%	Combined oxidative phosphorylation deficiency 12, 614924
EBP	89.2	99%	82%	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
ECE1	170.1	97%	99%	Hirschsprung disease, cardiac defects, and autonomic dysfunction, 613870 {Hypertension, essential, susceptibility to}, 145500
ECEL1	108.1	90%	99%	Arthrogryposis, distal, type 5D, 615065
ECHS1	123.3	100%	82%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ECM1	186	100%	99%	Urbach-Wiethe disease, 247100
EDA	99.3	92%	96%	Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100 Tooth agenesis, selective, X-linked 1, 313500
EDAR	151.6	99%	100%	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490 Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900 [Hair morphology 1, hair thickness], 612630
EDARADD	105.1	99%	99%	Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940 Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941
EDN1	166.6	100%	100%	Auriculocondylar syndrome 3, 615706 Question mark ears, isolated, 612798 {High density lipoprotein cholesterol level QTL 7}
EDN3	145.1	100%	93%	Central hypoventilation syndrome, congenital, 209880 Waardenburg syndrome, type 4B, 613265 {Hirschsprung disease, susceptibility to, 4}, 613712

EDNRA	229.1	100%	98%	Mandibulofacial dysostosis with alopecia, 616367 {Migraine, resistance to}, 157300
EDNRB	151.4	98%	99%	ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580 {Hirschsprung disease, susceptibility to, 2}, 600155
EEF1A2	213.2	100%	99%	Epileptic encephalopathy, early infantile, 33, 616409 Mental retardation, autosomal dominant 38, 616393
EFEMP1	210.5	99%	99%	Doyne honeycomb degeneration of retina, 126600
EFEMP2	146.1	100%	99%	Cutis laxa, autosomal recessive, type IB, 614437
EFNB1	144.7	100%	99%	Craniofrontonasal dysplasia, 304110
EFTUD2	127.2	99%	99%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EGF	165.2	100%	81%	Hypomagnesemia 4, renal, 611718
EGFR	178.5	100%	100%	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	63.9	88%	98%	Erythrocytosis, familial, 3, 609820 [Hemoglobin, high altitude adaptation], 609070
EGR2	117.6	100%	91%	Charcot-Marie-Tooth disease, type 1D, 607678 Dejerine-Sottas disease, 145900 Neuropathy, congenital hypomyelinating, 1, 605253
EHMT1	163	99%	99%	Kleefstra syndrome, 610253
EIF2AK3	177.3	96%	99%	Wolcott-Rallison syndrome, 226980
EIF2AK4	168.8	99%	99%	Pulmonary venoocclusive disease 2, 234810
EIF2B1	151	100%	100%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B2	155.4	100%	99%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B3	186.7	100%	98%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B4	159.3	100%	99%	Leukoencephaly with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B5	133.6	99%	99%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF4A3	115.1	100%	98%	Robin sequence with cleft mandible and limb abnormalities,268305
ELAC2	133	100%	98%	Combined oxidative phosphorylation deficiency 17, 615440

				{Prostate cancer, hereditary, 2, susceptibility to}, 614731
ELANE	115.1	99%	99%	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
ELN	103.3	99%	99%	Cutis laxa, AD, 123700 Supravalvar aortic stenosis, 185500
ELOVL4	115.5	100%	99%	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Stargardt disease 3, 600110 ?Spinocerebellar ataxia 34, 133190
ELOVL5	129.4	100%	94%	Spinocerebellar ataxia 38, 615957
EMC1	133.2	100%	100%	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EMD	117.4	98%	99%	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
EMG1	143.9	100%	96%	Bowen-Conradi syndrome, 211180
EMP2	101.9	100%	100%	Nephrotic syndrome, type 10, 615861
EMR2	173	96%	100%	Vibratory urticaria, 125630
EMX2	132.8	100%	96%	Schizencephaly, 269160
ENAM	154.1	100%	88%	Amelogenesis imperfecta, type IB, 104500 Amelogenesis imperfecta, type IC, 204650
ENG	148.5	99%	99%	Telangiectasia, hereditary hemorrhagic, type 1, 187300
ENPP1	174.9	94%	98%	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	170.4	99%	98%	Spastic paraplegia 64, autosomal recessive, 615683
EOGT	173.6	99%	97%	Adams-Oliver syndrome 4, 615297
EP300	204.1	99%	97%	Colorectal cancer, somatic, 114500 Rubinstein-Taybi syndrome 2, 613684
EPAS1	161.8	99%	99%	Erythrocytosis, familial, 4, 611783
EPB41	142.3	99%	85%	Elliptocytosis-1, 611804
EPB42	178.5	100%	98%	Spherocytosis, type 5, 612690
EPCAM	80.6	96%	97%	Colorectal cancer, hereditary nonpolyposis, type 8, 613244 Diarrhea 5, with tufting enteropathy, congenital, 613217
EPG5	138.8	99%	96%	Vici syndrome, 242840
EPHA2	193.2	99%	84%	Cataract 6, multiple types, 116600

EPHX1	139.7	98%	97%	Diphenylhydantoin toxicity Hypercholanemia, familial, 607748 ?Fetal hydantoin syndrome {Preeclampsia, susceptibility to}, 189800
EPM2A	121.8	85%	99%	Epilepsy, progressive myoclonic 2A (Lafora), 254780
ERBB2	157.1	98%	99%	Adenocarcinoma of lung, somatic, 211980 Gastric cancer, somatic, 613659 Glioblastoma, somatic, 137800 Ovarian cancer, somatic,
ERBB3	145.7	100%	96%	Lethal congenital contractural syndrome 2, 607598
ERBB4	172.4	99%	99%	Amyotrophic lateral sclerosis 19, 615515
ERCC1	102	99%	99%	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	143	100%	99%	Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy 1, photosensitive, 601675 Xeroderma pigmentosum, group D, 278730
ERCC3	120.8	100%	99%	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651
ERCC4	165.7	99%	99%	Fanconi anemia, complementation group Q, 615272 Xeroderma pigmentosum, group F, 278760 Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 ?XFE progeroid syndrome, 610965
ERCC5	151.3	100%	98%	Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	196.3	100%	87%	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	143.8	99%	98%	Bone marrow failure syndrome 2, 615715
ERCC8	109.4	96%	100%	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621
ERF	127.9	99%	99%	Chitayat syndrome,617180

				Craniosynostosis 4, 600775
ERLIN1	179.6	100%	94%	Spastic Paraplegia 62, 615681
ERLIN2	172.1	100%	59%	Spastic paraplegia 18, autosomal recessive, 611225
ESCO2	135.2	98%	99%	Roberts syndrome, 268300 SC phocomelia syndrome, 269000
ESPN	47.3	75%	97%	Deafness, autosomal recessive 36, 609006 Deafness, neurosensory, without vestibular involvement, autosomal dominant
ESR1	130.8	100%	99%	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	133	99%	100%	Deafness, autosomal recessive 35, 608565
ETFA	163.2	100%	99%	Glutaric acidemia IIA, 231680
ETFB	128.4	100%	97%	Glutaric acidemia IIB, 231680
ETFDH	127.9	100%	100%	Glutaric acidemia IIC, 231680
ETHE1	92	99%	91%	Ethylmalonic encephalopathy, 602473
ETV6	152.2	100%	94%	Leukemia, acute myeloid, somatic, 601626 Thrombocytopenia 5, 616216
EVC	126.1	94%	86%	Ellis-van Creveld syndrome, 225500 Weyers acrodental dysostosis, 193530
EVC2	137.8	96%	91%	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530
EWSR1	82	94%	85%	Ewing sarcoma, 612219 Neuroepithelioma, 612219
EXOSC3	90.8	97%	100%	Pontocerebellar hypoplasia, type 1B, 614678
EXOSC8	102.4	93%	97%	Pontocerebellar hypoplasia, type 1C, 616081
EXPH5	229.7	100%	99%	Epidermolysis bullosa, nonspecific, autosomal recessive, 615028
EXT1	105.5	99%	99%	Chondrosarcoma, 215300 Exostoses, multiple, type 1, 133700
EXT2	178.8	99%	99%	Exostoses, multiple, type 2, 133701 ?Seizures, scoliosis, and macrocephaly syndrome, 616682
EYA1	167.2	100%	97%	Anterior segment anomalies with or without cataract, 113650

				Branchiootic syndrome 1, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650 ?Otofaciocervical syndrome, 166780
EYA4	189.6	100%	98%	Cardiomyopathy, dilated, 1J, 605362 Deafness, autosomal dominant 10, 601316
EYS	172.3	99%	97%	Retinitis pigmentosa 25, 602772
EZH2	166.9	99%	99%	Weaver syndrome, 277590
F10	187.1	99%	99%	Factor X deficiency, 227600
F11	170.5	100%	99%	Factor XI deficiency, autosomal dominant, 612416 Factor XI deficiency, autosomal recessive, 612416
F12	131.4	100%	92%	Angioedema, hereditary, type III, 610618 Factor XII deficiency, 234000
F13A1	182.8	100%	99%	Factor XIIIA deficiency, 613225 {Myocardial infarction, protection against}, 608446 {Venous thrombosis, protection against}, 188050
F13B	156.6	97%	98%	Factor XIIIB deficiency, 613235
F2	138.3	100%	99%	Dysprothrombinemia, 613679 Hypoprothrombinemia, 613679 Thrombophilia due to thrombin defect, 188050 {Pregnancy loss, recurrent, susceptibility to, 2}, 614390 {Stroke, ischemic, susceptibility to}, 601367
F5	196.8	99%	98%	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	178.3	100%	97%	Factor VII deficiency, 227500 {Myocardial infarction, decreased susceptibility to}, 608446
F8	138.1	99%	89%	Hemophilia A, 306700
F9	156	99%	99%	Hemophilia B, 306900 Thrombophilia, X-linked, due to factor IX defect, 300807 {Deep venous thrombosis, protection against}, 300807 {Warfarin sensitivity}, 122700
FA2H	101.9	95%	99%	Spastic paraplegia 35, autosomal recessive, 612319

FADD	140.3	99%	89%	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovasuclar malformations, 613759
FAH	176.7	100%	99%	Tyrosinemia, type I, 276700
FAM105B	177	95%	99%	Autoinflammation,panniculitis, and dermatosis syndrome, 617099
FAM111A	292.7	100%	96%	Gracile bone dysplasia, 602361 Kenny-Caffey syndrome, type 2, 127000
FAM111B	185.5	100%	90%	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704
FAM126A	161.3	98%	98%	Leukodystrophy, hypomyelinating, 5, 610532
FAM134B	132.9	96%	94%	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
FAM161A	148.8	99%	98%	Retinitis pigmentosa 28, 606068
FAM20A	115.2	98%	78%	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM20C	118.7	99%	91%	Raine syndrome, 259775
FAM58A	88.6	82%	99%	STAR syndrome, 300707
FAM83H	93.9	96%	98%	Amelogenesis imperfecta, type III, 130900
FAN1	168.9	100%	90%	Interstitial nephritis, karyomegalic, 614817
FANCA	129.9	99%	98%	Fanconi anemia, complementation group A, 227650
FANCB	84.3	97%	96%	Fanconi anemia, complementation group B, 300514
FANCC	118.8	99%	85%	Fanconi anemia, complementation group C, 227645
FANCD2	156.1	99%	100%	Fanconi anemia, complementation group D2, 227646
FANCE	127.7	88%	99%	Fanconi anemia, complementation group E, 600901
FANCF	179.5	100%	98%	Fanconi anemia, complementation group F, 603467
FANCG	158	100%	97%	Fanconi anemia, complementation group G, 614082
FANCI	174.9	99%	93%	Fanconi anemia, complementation group I, 609053
FANCL	105.1	99%	100%	Fanconi anemia, complementation group L, 614083
FAR1	102.4	96%	99%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154
FARS2	234.6	100%	98%	Combined oxidative phosphorylation deficiency 14, 614946 ?Spastic paraplegia 77, autosomal recessive, 617046
FAS	294.7	100%	100%	Autoimmune lymphoproliferative syndrome, type IA, 601859 Squamous cell carcinoma, burn scar-related, somatic {Autoimmune lymphoproliferative syndrome}, 601859
FASLG	98.6	99%	91%	Autoimmune lymphoproliferative syndrome, type IB, 601859 {Lung cancer, susceptibility to}, 211980
FAT4	259.2	100%	99%	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006

				Van Maldergem syndrome 2, 615546
FBLN5	116.4	91%	99%	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	177.6	99%	99%	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	191.3	100%	100%	Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118
FBP1	132.9	100%	99%	Fructose-1,6-bisphosphatase deficiency, 229700
FBXL4	231	100%	98%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FBXO38	207.1	99%	99%	Neuronopathy, distal hereditary motor, type IID, 615575
FBXO7	224.2	99%	98%	Parkinson disease 15, autosomal recessive, 260300
FCGR3A	242.2	100%	99%	Immunodeficiency 20, 615707
FCGR3B	180.2	99%	92%	Neutropenia, alloimmune neonatal
FCN3	145.2	100%	99%	Immunodeficiency due to ficolin 3 deficiency, 613860
FDPS	66.8	98%	96%	Porokeratosis 9, multiple types, 616631
FECH	137.3	100%	98%	Protoporphyria, erythropoietic, autosomal recessive, 177000
FERMT1	109.7	98%	99%	Kindler syndrome,173650
FERMT3	139.2	99%	97%	Leukocyte adhesion deficiency,type III,612840
FEZF1	170.4	100%	97%	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FGA	178.9	99%	88%	Afibrinogenemia, congenital, 202400 Amyloidosis, familial visceral, 105200 Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, congenital, 616004
FGB	198.5	99%	96%	Afibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Hypofibrinogenemia, congenital, 202400

FGD1	98.4	94%	100%	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400
FGD4	117.7	98%	99%	Charcot-Marie-Tooth disease, type 4H, 609311
FGF10	176	100%	98%	Aplasia of lacrimal and salivary glands, 180920 LADD syndrome, 149730
FGF14	225.2	100%	100%	Spinocerebellar ataxia 27, 609307
FGF16	130.2	99%	98%	Metacarpal 4-5 fusion, 309630
FGF17	167.7	100%	87%	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
FGF23	122.5	99%	99%	Hypophosphatemic rickets, autosomal dominant, 193100 Osteomalacia, tumor-induced Tumoral calcinosis, hyperphosphatemic, familial, 211900
FGF3	86.6	96%	80%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGF5	129.1	100%	98%	Trichomegaly, 190330
FGF8	137.4	87%	96%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGFR1	165.2	99%	99%	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	155.6	97%	98%	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	129.1	100%	89%	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	178.7	99%	93%	Afibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, 616004 Hypofibrinogenemia, congenital, 202400
FH	183.5	93%	99%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FHL1	98.4	98%	89%	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	214.4	100%	95%	Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228 Yunis-Varon syndrome, 216340 ?Polymicrogyria, bilateral temporooccipital, 612691
FIGLA	105.4	95%	99%	Premature ovarian failure 6, 612310
FKBP10	177.3	98%	100%	Bruck syndrome 1, 259450 Osteogenesis imperfecta, type XI, 610968
FKBP14	96.5	99%	95%	Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss, 614557
FKRP	103.2	100%	97%	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	155.6	99%	99%	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	200.1	99%	99%	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100
FLCN	176.6	100%	99%	Birt-Hogg-Dube syndrome, 135150 Colorectal cancer, somatic, 114500 Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700
FLG	237.1	100%	99%	Ichthyosis vulgaris, 146700 {Dermatitis, atopic, susceptibility to, 2}, 605803
FLNA	160.7	100%	99%	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	169.5	100%	100%	Atelosteogenesis, type I, 108720 Atelosteogenesis, type III, 108721 Boomerang dysplasia, 112310 Larsen syndrome, 150250 Spondylocarpotarsal synostosis syndrome, 272460
FLNC	188.5	100%	98%	Cardiomyopathy, familial hypertrophic, 26 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Myopathy, myofibrillar, 5, 609524
FLRT3	242.5	100%	98%	Hypogonadotropic hypogonadism 21 with anosmia, 615271

FLT3	147.6	99%	98%	Leukemia, acute lymphoblastic, somatic, 613065 Leukemia, acute myeloid, reduced survival in, somatic, 601626 Leukemia, acute myeloid, somatic, 601626
FLT4	179	99%	100%	Hemangioma, capillary infantile, somatic, 602089 Lymphedema, hereditary, IA, 153100
FLVCR1	166.7	99%	79%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FLVCR2	196.2	100%	99%	Proliferative vasculopathy and hydraencephaly-hydrocephaly syndrome, 225790
FMN2	107.3	84%	90%	Mental retardation, autosomal recessive 47, 616193
FMO3	195.2	100%	99%	Trimethylaminuria, 602079
FMR1	90.5	97%	100%	Fragile X syndrome, 300624 Fragile X tremor/ataxia syndrome, 300623 Premature ovarian failure 1, 311360
FN1	156.3	100%	79%	Glomerulopathy with fibronectin deposits 2, 601894 Plasma fibronectin deficiency, 614101
FOLR1	155.8	100%	88%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXC1	39.3	94%	62%	Axenfeld-Rieger syndrome, type 3, 602482 Iridogoniodysgenesis, type 1, 601631 Iris hypoplasia and glaucoma, 601631 Rieger or Axenfeld anomalies, 602482
FOXC2	64.1	98%	55%	Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400 Lymphedema-distichiasis syndrome, 153400
FOXE1	37.4	81%	93%	Bamforth-Lazarus syndrome, 241850 {Thyroid cancer, nonmedullary, 4}, 616534
FOXE3	24.7	73%	81%	Anterior segment mesenchymal dysgenesis, 107250 Aphakia, congenital primary, 610256 Cataract 34,multiple types, 612968
FOXF1	93.8	99%	100%	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380
FOXG1	139.8	87%	78%	Rett syndrome, congenital variant, 613454
FOXI1	180.6	100%	99%	Enlarged vestibular aqueduct, 600791
FOXL2	43	94%	91%	Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 Premature ovarian failure 3, 608996
FOXN1	127.1	100%	100%	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXO1	147.2	95%	98%	Rhabdomyosarcoma,alveolar,268220
FOXP1	155.1	100%	92%	Mental retardation with language impairment and with or without autistic features, 613670

FOXP2	184.8	99%	99%	Speech-language disorder-1, 602081
FOXP3	130.9	98%	99%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790 {Diabetes mellitus, type I, susceptibility to}, 222100
FOXRED1	144.3	100%	99%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
FRAS1	168.5	100%	99%	Fraser syndrome, 219000
FREM1	149.2	99%	99%	Bifid nose with or without anorectal and renal anomalies, 608980 Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485
FREM2	194.3	99%	62%	Fraser syndrome, 219000
FRMD7	140.9	99%	100%	Nystagmus 1, congenital, X-linked, 310700 Nystagmus, infantile periodic alternating, X-linked, 310700
FRRS1L	130.4	70%	100%	Epileptic encephalopathy, early infantile, 37, 616981
FSCN2	149.5	100%	98%	Retinitis pigmentosa 30, 607921
FSHB	158.9	100%	89%	Hypogonadotropic hypogonadism 24 without anosmia, 229070
FSHR	146.4	99%	93%	Ovarian dysgenesis 1, 233300 Ovarian hyperstimulation syndrome, 608115 Ovarian response to FSH stimulation, 276400
FTCD	104.6	95%	99%	Glutamate formiminotransferase deficiency, 229100
FTL	149.2	99%	96%	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159
FTO	148.3	100%	99%	Growth retardation, developmental delay, facial dysmorphism, 612938 {Obesity, susceptibility to, BMIQ14}, 612460
FTSJ1	143.1	99%	94%	Mental retardation, X-linked 9, 309549
FUCA1	151.4	100%	99%	Fucosidosis, 230000
FUS	148.1	97%	99%	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 Tremor, hereditary essential, 4, 614782
FUT6	168.7	100%	76%	Fucosyltransferase 6 deficiency, 613852
FUZ	125	100%	99%	Neural tube defects, 182940
FXN	84.8	86%	99%	Friedreich ataxia with retained reflexes, 229300 Friedreich ataxia, 229300
FXYD2	104.4	100%	99%	Hypomagnesemia 2, renal, 154020
FYCO1	141.9	100%	100%	Cataract 18, autosomal recessive, 610019

FZD4	226.6	99%	100%	Exudative vitreoretinopathy 1, 133780 Retinopathy of prematurity, 133780
FZD6	260.7	100%	100%	Nail disorder, nonsyndromic congenital, 10, (claw-shaped nails), 614157
G6PC	201.1	100%	96%	Glycogen storage disease Ia, 232200
G6PC3	143.5	100%	99%	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
G6PD	138.1	99%	100%	Favism, 134700 Hemolytic anemia due to G6PD deficiency, 300908 {Resistance to malaria due to G6PD deficiency}, 611162
GAA	136.8	100%	92%	Glycogen storage disease II, 232300
GABRA1	219.1	100%	96%	Epileptic encephalopathy, early infantile, 19, 615744 {Epilepsy, childhood absence, susceptibility to, 4}, 611136 {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136
GABRG2	196.1	94%	99%	Epilepsy, generalized, with febrile seizures plus, type 3, 611277 Febrile seizures, familial, 8, 611277 {Epilepsy, childhood absence, susceptibility to, 2}, 607681
GALC	115.9	99%	96%	Krabbe disease, 245200
GALE	182.8	100%	95%	Galactose epimerase deficiency, 230350
GALK1	127.8	99%	98%	Galactokinase deficiency with cataracts, 230200
GALNS	108.4	99%	100%	Mucopolysaccharidosis IVA, 253000
GALNT3	166.7	99%	91%	Tumoral calcinosis, hyperphosphatemic, familial, 211900
GALT	182.8	100%	99%	Galactosemia, 230400
GAMT	123	98%	98%	Cerebral creatine deficiency syndrome 2, 612736
GAN	199	100%	98%	Giant axonal neuropathy-1, 256850
GANAB	125.1	99%	99%	Polycystic kidney disease 3, 600666
GARS	154.1	99%	94%	Charcot-Marie-Tooth disease, type 2D, 601472 Neuropathy, distal hereditary motor, type VA, 600794
GAS8	165.1	99%	99%	Ciliary dyskinesia, primary, 33, 616726
GATA1	100.8	99%	100%	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	145.4	100%	63%	Emberger syndrome, 614038 Immunodeficiency 21, 614172

				{Leukemia, acute myeloid, susceptibility to}, 601626 {Myelodysplastic syndrome, susceptibility to}, 614286
GATA3	205.9	100%	73%	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GATA4	98.9	74%	99%	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	61.9	86%	99%	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	140.6	100%	100%	Mental retardation, autosomal dominant 18, 615074
GATM	173	100%	99%	Cerebral creatine deficiency syndrome 3, 612718
GBA	227.1	100%	98%	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	185.6	100%	91%	Spastic paraplegia 46, autosomal recessive, 614409
GBE1	189.8	99%	89%	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GCDH	139.2	93%	100%	Glutaricaciduria, type I, 231670
GCH1	92.5	97%	99%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GCK	151.4	100%	100%	Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, permanent neonatal, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485 MODY, type II, 125851
GCLC	161.4	100%	100%	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 {Myocardial infarction, susceptibility to}, 608446
GCM2	173.7	100%	70%	Hypoparathyroidism, familial isolated, 146200

GCNT2	201.6	100%	98%	Adult i phenotype without cataract, 110800 Cataract 13 with adult i phenotype, 116700 [Blood group, Ii], 110800
GCSH	41.7	85%	56%	Glycine encephalopathy, 605899
GDAP1	210.8	99%	100%	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	26	77%	100%	Double-outlet right ventricle, 217095 Right atrial isomerism, 208530 Tetralogy of Fallot, 187500 Transposition of great arteries, dextro-looped 3, 613854
GDF2	185.5	100%	100%	Telangiectasia, hereditary hemorrhagic, type 5, 615506
GDF3	137.7	100%	92%	Klippel-Feil syndrome 3, autosomal dominant, 613702 Microphthalmia with coloboma 6, 613703 Microphthalmia, isolated 7, 613704
GDF5	179.6	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	88.9	98%	98%	Klippel-Feil syndrome 1, autosomal dominant, 118100 Leber congenital amaurosis 17, 615360 Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094
GDI1	160.8	100%	98%	Mental retardation, X-linked 41, 300849
GDNF	213.9	99%	92%	Central hypoventilation syndrome, 209880 {Hirschsprung disease, susceptibility to, 3}, 613711 {Pheochromocytoma, modifier of}, 171300
GFAP	119.9	99%	96%	Alexander disease, 203450
GFER	91.5	99%	100%	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay,

				613076
GFI1	99.4	99%	97%	Neutropenia, nonimmune chronic idiopathic, of adults, 607847 Neutropenia, severe congenital 2, autosomal dominant, 613107
GFI1B	204.5	100%	99%	Bleeding disorder, platelet-type, 17, 187900
GFM1	125.6	99%	99%	Combined oxidative phosphorylation deficiency 1, 609060
GFPT1	171.9	99%	100%	Myasthenia, congenital, 12, with tubular aggregates, 610542
GGCX	124.5	100%	99%	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842 Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450
GH1	229.6	100%	99%	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	245.4	99%	98%	Growth hormone insensitivity, partial, 604271 Increased responsiveness to growth hormone Laron dwarfism, 262500 {Hypercholesterolemia, familial, modifier of}, 143890
GHRHR	148.2	100%	99%	Growth hormone deficiency, isolated, type IB, 612781
GHSR	198.6	99%	88%	Growth hormone deficiency, isolated partial, 615925
GIF	169.4	100%	100%	Intrinsic factor deficiency, 261000
GIPC3	125.6	92%	99%	Deafness, autosomal recessive 15, 601869
GJA1	238.2	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	172.5	99%	99%	Cataract 14, multiple types, 601885
GJA5	319.7	100%	100%	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic (GJA5/SCN5A), 108770
GJA8	179.7	99%	100%	Cataract 1, multiple types, 116200
GJB1	245.6	100%	100%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GJB2	209.4	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	343.4	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	396.3	100%	71%	Erythrokeratodermia variabilis with erythema gyratum repens, 133200
GJB6	214	100%	68%	Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290 Ectodermal dysplasia 2, Clouston type, 129500
GJC2	50	88%	97%	Leukodystrophy, hypomyelinating, 2, 608804 Lymphedema, hereditary, IC, 613480 Spastic paraplegia 44, autosomal recessive, 613206
GK	57.5	84%	97%	Glycerol kinase deficiency, 307030
GLA	83.5	99%	84%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	97.3	99%	99%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLDC	88.5	92%	97%	Glycine encephalopathy, 605899
GLE1	121.2	99%	99%	Arthrogryposis, lethal, with anterior horn cell disease, 611890 Lethal congenital contracture syndrome 1, 253310
GLI2	148.1	99%	97%	Culler-Jones syndrome, 615849 Holoprosencephaly-9, 610829
GLI3	168.4	99%	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	108.7	99%	94%	Nephronophthisis 7, 611498
GLIS3	147.4	100%	99%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GLMN	95.5	99%	93%	Glomuvenous malformations,138000
GLRA1	124.8	100%	84%	Hyperekplexia, hereditary 1, autosomal dominant or recessive, 149400
GLRB	119.6	98%	85%	Hyperekplexia 2, autosomal recessive, 614619
GLRX5	105.8	91%	97%	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GLUD1	84.5	94%	99%	Hyperinsulinism-hyperammonemia syndrome, 606762
GLUL	104.4	99%	100%	Glutamine deficiency, congenital, 610015
GLYCTK	230.9	99%	90%	D-glyceric aciduria, 220120
GM2A	154.1	100%	100%	GM2-gangliosidosis, AB variant, 272750
GMNN	139.5	97%	100%	Meier-Gorlin syndrome 6, 616835
GMPPA	162.5	100%	99%	Alacrima, achalasia, and mental retardation syndrome, 615510
GMPPB	276.3	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	185.8	100%	97%	Hypocalcemia, autosomal dominant 2, 615361 Hypocalciuric hypercalcemia, type II, 145981
GNAI2	152.3	100%	93%	Pituitary ACTH-secreting adenoma Ventricular tachycardia, idiopathic, 192605
GNAI3	132	99%	100%	Auriculocondylar syndrome 1, 602483
GNAL	151.4	97%	75%	Dystonia 25, 615073
GNAO1	197.4	100%	97%	Epileptic encephalopathy, early infantile, 17, 615473
GNAQ	90.6	89%	100%	Capillary malformations, congenital, 1, somatic, mosaic, 163000 Sturge-Weber syndrome, somatic, mosaic, 185300
GNAS	154.4	99%	99%	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	999999		100%	Pseudohypoparathyroidism, type IB, 603233

GNAT1	180.9	100%	99%	Night blindness, congenital stationary, autosomal dominant 3, 610444 ?Night blindness, congenital stationary, type 1G, 616389
GNAT2	157.8	100%	100%	Achromatopsia-4, 613856
GNB1	225.2	100%	99%	Leukemia,acute lymphoblastic,somatic, 613065 Mental retardation, autosomal dominant 42, 616973
GNB3	211.1	100%	94%	Night blindness, congenital stationary, type 1H, 617024 {Hypertension, essential, susceptibility to}, 145500
GNB4	189.5	100%	97%	Charcot-Marie-Tooth disease, dominant intermediate F, 615185
GNE	167.4	100%	97%	Nonaka myopathy, 605820 Sialuria, 269921
GNMT	176.1	99%	92%	Glycine N-methyltransferase deficiency, 606664
GNPAT	176.5	99%	100%	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	190.4	99%	93%	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GNPTG	169.7	96%	97%	Mucolipidosis III gamma, 252605
GNRHR	218.3	100%	96%	Hypogonadotropic hypogonadism 7 without anosmia, 146110
GNS	119.6	97%	98%	Mucopolysaccharidosis type IIID, 252940
GORAB	203.4	99%	95%	Geroderma osteodysplasticum, 231070
GOSR2	143.2	97%	69%	Epilepsy, progressive myoclonic 6, 614018
GOT1	135.7	99%	100%	Aspartate aminotransferase, serum level of, QTL1, 614419
GP1BA	175.2	97%	93%	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	41.4	80%	94%	Bernard-Soulier syndrome, type B, 231200 Giant platelet disorder, isolated, 231200
GP6	140.1	100%	100%	Bleeding disorder, platelet-type, 11, 614201
GP9	100.8	98%	99%	Bernard-Soulier syndrome, type C, 231200
GPC3	106.4	98%	99%	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPC6	160	100%	97%	Omodysplasia 1, 258315
GPD1	100.2	100%	99%	Hypertriglyceridemia, transient infantile, 614480
GPD1L	184.1	100%	98%	Brugada syndrome 2, 611777
GPHN	186.4	99%	100%	Molybdenum cofactor deficiency C, 615501

GPI	162.7	100%	98%	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470
GPIHBP1	123.3	99%	83%	Hyperlipoproteinemia, type 1D, 615947
GPR101	129.9	100%	99%	Pituitary adenoma, growth hormone-secreting 2, 300943
GPR143	73.2	90%	100%	Nystagmus 6, congenital, X-linked, 300814 Ocular albinism, type I, Nettleship-Falls type, 300500
GPR179	160.1	100%	98%	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
GPR56	177.1	100%	98%	Polymicrogyria, bilateral frontoparietal, 606854 Polymicrogyria, bilateral perisylvian, 615752
GPR98	172.4	99%	85%	Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472 ?Febrile seizures, familial, 4, 604352
GPSM2	135.2	99%	100%	Chudley-McCullough syndrome, 604213
GPX4	195.5	87%	100%	Spondylometaphyseal dysplasia, Sedaghatian type, 250220
GRHL2	152.8	100%	81%	Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029
GRHL3	162.5	100%	95%	Van der Woude syndrome 2, 606713
GRHPR	118	86%	99%	Hyperoxaluria, primary, type II, 260000
GRIA3	102.9	99%	95%	Mental retardation, X-linked 94, 300699
GRID2	216.8	100%	99%	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRIK2	170.5	96%	100%	Mental retardation, autosomal recessive, 6, 611092
GRIN1	169.2	100%	99%	Mental retardation, autosomal dominant 8, 614254
GRIN2A	170.4	100%	99%	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570
GRIN2B	194.9	99%	99%	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation, autosomal dominant 6, 613970
GRIP1	148.9	100%	99%	Fraser syndrome, 219000
GRK1	137.1	100%	87%	Oguchi disease-2, 613411
GRM1	194.2	100%	100%	Spinocerebellar ataxia, autosomal recessive 13, 614831
GRM6	166.2	94%	100%	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270
GRN	214.6	100%	89%	Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
GRXCR1	232.7	100%	92%	Deafness, autosomal recessive 25, 613285
GSC	127	98%	99%	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471
GSN	141.9	97%	96%	Amyloidosis, Finnish type, 105120

GSS	117	100%	99%	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900
GTF2E2	105.9	99%	99%	Trichothiodystrophy 6,nonphotosensitive,616943
GTF2H5	152.5	100%	100%	Trichothiodystrophy 3, photosensitive, 616395
GTPBP3	139.5	100%	99%	Combined oxidative phosphorylation deficiency 23, 616198
GUCA1A	195.7	100%	98%	Cone dystrophy-3, 602093 Cone-rod dystrophy 14, 602093
GUCA1B	154.9	100%	99%	Retinitis pigmentosa 48, 613827
GUCY1A3	187.6	99%	94%	Moyamoya 6 with achalasia, 615750
GUCY2C	151.6	100%	87%	Diarrhea 6, 614616 Meconium ileus, 614665
GUCY2D	104.8	99%	99%	Cone-rod dystrophy 6, 601777 Leber congenital amaurosis 1, 204000
GUSB	126.8	91%	98%	Mucopolysaccharidosis VII, 253220
GYG1	168.1	100%	97%	Polyglucosan body myopathy 2, 616199 ?Glycogen storage disease XV, 613507
GYS1	120.2	99%	99%	Glycogen storage disease 0, muscle, 611556
GYS2	172	99%	98%	Glycogen storage disease 0, liver, 240600
H19	999999		94%	Beckwith-Wiedemann syndrome, 130650 Silver-Russell syndrome, 180860 Wilms tumor 2, 194071
H6PD	194.2	99%	92%	Cortisone reductase deficiency 1, 604931
HACE1	167.6	99%	89%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADH	126.2	97%	100%	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HADHA	96.1	97%	100%	Fatty liver, acute, of pregnancy, 609016 HELLP syndrome, maternal, of pregnancy, 609016 LCHAD deficiency, 609016 Trifunctional protein deficiency, 609015
HADHB	106.7	96%	100%	Trifunctional protein deficiency, 609015
HAMP	196.3	100%	99%	Hemochromatosis, type 2B, 613313
HARS	167	100%	88%	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504
HAX1	157.7	100%	100%	Neutropenia, severe congenital 3, autosomal recessive, 610738

HBA1	133.9	100%	100%	Erythremias, alpha- Heinz body anemias, alpha-, 140700 Hemoglobin H disease, nondeletional, 613978 Methemoglobinemias, alpha- Thalassemias, alpha-, 604131
HBA2	129.7	97%	94%	Erythrocytosis Heinz body anemia, 140700 Hemoglobin H disease, nondeletional, 613978 Hypochromic microcytic anemia Thalassemia, alpha-, 604131
HBB	187.2	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	244.9	100%	98%	Thalassemia due to Hb Lepore Thalassemia, delta-
HBG1	168.4	97%	96%	Fetal hemoglobin quantitative trait locus 1, 141749
HBG2	321.7	100%	99%	Cyanosis, transient neonatal, 613977 Fetal hemoglobin quantitative trait locus 1, 141749
HCCS	115.6	99%	96%	Linear skin defects with multiple congenital anomalies 1, 309801
HCFC1	113.8	99%	99%	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541
HCN1	155.1	100%	83%	Epileptic encephalopathy, early infantile, 24, 615871
HCN4	94.6	99%	90%	Brugada syndrome 8, 613123 Sick sinus syndrome 2, 163800
HDAC8	142.5	100%	78%	Cornelia de Lange syndrome 5, 300882
HEATR2	123.9	90%	99%	Ciliary dyskinesia, primary, 18, 614874
HELLS	110.7	96%	76%	Immunodeficiency-centromeric instability-facial anomalies syndrome 4,616911
HEPACAM	156.4	86%	66%	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926

HERC1	191.4	99%	96%	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
HERC2	120.8	79%	98%	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	44.3	79%	95%	Spondylocostal dysostosis 4, autosomal recessive, 613686
HESX1	96.7	99%	99%	Growth hormone deficiency with pituitary anomalies, 182230 Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230
HEXA	133.7	100%	100%	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800 [Hex A pseudodeficiency], 272800
HEXB	166.2	99%	85%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HFE	148.7	100%	99%	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	135.7	100%	98%	Hemochromatosis type 2A,602390
HFM1	57.7	93%	81%	Premature ovarian failure 9, 615724
HGD	144.2	99%	86%	Alkaptonuria, 203500
HGF	173.2	99%	90%	Deafness, autosomal recessive 39, 608265
HGSNAT	122.6	81%	100%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544
HIBCH	87.8	97%	99%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HINT1	60.9	98%	100%	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200
HIVEP2	214.5	100%	99%	Mental retardation, autosomal dominant 43, 616977
HK1	152.9	100%	99%	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285
HLCS	182.6	100%	100%	Holocarboxylase synthetase deficiency, 253270
HMBS	117.7	100%	90%	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000
HMGCL	152.8	100%	51%	HMG-CoA lyase deficiency, 246450
HMGCS2	143.1	100%	99%	HMG-CoA synthase-2 deficiency, 605911

HMOX1	152.2	96%	97%	Heme oxygenase-1 deficiency, 614034 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963
HMX1	25.4	72%	99%	Oculoauricular syndrome, 612109
HNF1A	164.4	99%	99%	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	137.9	99%	90%	Diabetes mellitus, noninsulin-dependent, 125853 Renal cysts and diabetes syndrome, 137920 {Renal cell carcinoma}, 144700
HNF4A	159.4	100%	87%	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026 MODY, type I, 125850 {Diabetes mellitus, noninsulin-dependent}, 125853
HNMT	169.7	100%	81%	Mental retardation, autosomal recessive 51, 616739 {Asthma, susceptibility to}, 600807
HNRNPA1	86.5	98%	98%	Amyotrophic lateral sclerosis 20, 615426 ?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3, 615424
HNRNPDL	79.4	97%	100%	Muscular dystrophy, limb-girdle, type 1G, 609115
HNRNPK	78.5	87%	87%	Au-Kline syndrome, 616580
HOGA1	156.5	99%	66%	Hyperoxaluria, primary, type III, 613616
HOXA1	193.3	100%	100%	Athabaskan brainstem dysgenesis syndrome, 601536 Bosley-Salih-Alorainy syndrome, 601536
HOXA11	86.4	97%	93%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
HOXA13	58	73%	99%	Guttmacher syndrome, 176305 Hand-foot-uterus syndrome, 140000
HOXB1	113.6	100%	92%	Facial paresis, hereditary congenital, 3, 614744
HOXC13	133.7	98%	100%	Ectodermal dysplasia 9, hair/nail type, 614931
HOXD10	130.3	99%	99%	Charcot-Marie-Tooth disease, foot deformity of, 192950 Vertical talus, congenital, 192950
HOXD13	128.2	96%	98%	Brachydactyly, type D, 113200 Brachydactyly, type E, 113300 Syndactyly, type V, 186300 Synpolydactyly 1, 186000

				?Brachydactyly-syndactyly syndrome, 610713
HPCA	296	100%	89%	Dystonia 2, torsion, autosomal recessive, 224500
HPD	159.2	100%	99%	Hawkinsinuria, 140350 Tyrosinemia, type III, 276710
HPGD	110.1	99%	99%	Cranioosteoarthropathy, 259100 Digital clubbing, isolated congenital, 119900 Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100
HPRT1	82.8	96%	99%	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322
HPS1	136.6	100%	98%	Hermansky-Pudlak syndrome 1, 203300
HPS3	175.7	100%	90%	Hermansky-Pudlak syndrome 3, 614072
HPS4	151.7	100%	93%	Hermansky-Pudlak syndrome 4, 614073
HPS5	160.7	99%	95%	Hermansky-Pudlak syndrome 5, 614074
HPS6	156.4	97%	99%	Hermansky-Pudlak syndrome 6, 614075
HPSE2	127	97%	95%	Urofacial syndrome 1, 236730
HR	110.5	98%	99%	Alopecia universalis, 203655 Atrichia with papular lesions, 209500 Hypotrichosis 4, 146550
HRAS	204.3	100%	84%	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	187.4	97%	98%	Thrombophilia due to elevated HRG, 613116 Thrombophilia due to HRG deficiency, 613116
HSD11B1	182.6	100%	100%	Cortisone reductase deficiency 2, 614662
HSD11B2	179.5	88%	93%	Apparent mineralocorticoid excess, 218030
HSD17B10	119.7	100%	100%	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 ?Mental retardation, X-linked syndromic 10, 300220
HSD17B3	161.7	100%	96%	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	125.3	95%	95%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B2	181.3	100%	87%	3-beta-hydroxysteroid dehydrogenase, type II, deficiency, 201810

HSD3B7	166.7	99%	83%	Bile acid synthesis defect, congenital, 1, 607765
HSF4	119.8	98%	100%	Cataract 5, multiple types, 116800
HSPA9	105.5	93%	93%	Anemia, sideroblastic, 4, 182170 Even-plus syndrome, 616854
HSPB1	42.3	96%	98%	Charcot-Marie-Tooth disease, axonal, type 2F, 606595 Neuropathy, distal hereditary motor, type IIB, 608634
HSPB8	167.5	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2L, 608673 Neuropathy, distal hereditary motor, type IIA, 158590
HSPD1	105.4	98%	81%	Leukodystrophy, hypomyelinating, 4, 612233 Spastic paraplegia 13, autosomal dominant, 605280
HSPG2	133.9	99%	97%	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
HTR1A	196.1	100%	96%	Periodic fever, menstrual cycle dependent, 614674
HTRA1	116.9	86%	99%	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	171.9	98%	100%	Huntington disease, 143100
HUWE1	107.1	99%	99%	Mental retardation, X-linked syndromic, Turner type, 300706
HYDIN	143.3	99%	100%	Ciliary dyskinesia, primary, 5, 608647
HYLS1	174.4	100%	98%	Hydrolethalus syndrome, 236680
ICK	145.5	99%	100%	Endocrine-cerebroosteodysplasia, 612651
ICOS	186.3	100%	97%	Immunodeficiency, common variable, 1, 607594
IDH2	108	99%	86%	D-2-hydroxyglutaric aciduria 2, 613657
IDH3B	170.6	100%	80%	Retinitis pigmentosa 46, 612572
IDS	113.9	99%	98%	Mucopolysaccharidosis II, 309900
IDUA	120.5	92%	97%	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016
IER3IP1	72.1	92%	98%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	143.7	99%	93%	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFITM5	84.9	99%	99%	Osteogenesis imperfecta, type V, 610967

IFNGR1	171.1	99%	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	164	94%	99%	Immunodeficiency 28, mycobacteriosis, 614889
IFT122	165.7	100%	100%	Cranioectodermal dysplasia 1, 218330
IFT140	124.5	99%	79%	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	125.7	100%	96%	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT43	128	100%	100%	Cranioectodermal dysplasia 3, 614099
IFT80	79.6	92%	99%	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IGBP1	126.1	99%	96%	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472
IGF1	157.6	100%	99%	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IGF1R	153.4	100%	86%	Insulin-like growth factor I, resistance to, 270450
IGF2R	156.1	98%	65%	Hepatocellular carcinoma, somatic, 114550
IGFALS	83.7	100%	100%	Acid-labile subunit, deficiency of, 615961
IGFBP7	76.8	95%	95%	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis, 614224
IGHG2	38	83%	100%	IgG2 deficiency, selective
IGHM	204.2	100%	95%	Agammaglobulinemia 1, 601495
IGHMBP2	123	98%	96%	Charcot-Marie-Tooth disease, axonal, type 2S, 616155 Neuronopathy, distal hereditary motor, type VI, 604320
IGKC	171.9	100%	100%	Kappa light chain deficiency, 614102
IGLL1	82.2	99%	99%	Agammaglobulinemia 2, 613500
IGSF1	96.1	99%	95%	Hypothyroidism, central, and testicular enlargement, 300888
IHH	147.1	100%	72%	Acrocapitofemoral dysplasia, 607778 Brachydactyly, type A1, 112500
IKBKAP	152.8	99%	99%	Dysautonomia, familial, 223900
IKBKB	129.9	98%	99%	Immunodeficiency 15, 615592
IKBKG	57.4	83%	97%	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	202.5	100%	99%	Immunodeficiency,common variable, 1,616873
IL10RA	164.2	100%	99%	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	214.6	99%	96%	Inflammatory bowel disease 25, early onset, autosomal recessive, 612567 {Hepatitis B virus, susceptibility to}, 610424
IL11RA	155.9	100%	98%	Craniosynostosis and dental anomalies, 614188
IL12B	122.6	100%	98%	Immunodeficiency 29, mycobacteriosis, 614890
IL12RB1	138.8	98%	98%	Immunodeficiency 30, 614891
IL17RC	108.9	99%	100%	Candidiasis, familial, 9, 616445
IL17RD	146.4	99%	100%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
IL1RAPL1	135.1	100%	99%	Mental retardation, X-linked 21/34, 300143
IL1RN	174.7	100%	97%	Interleukin 1 receptor antagonist deficiency, 612852 {Gastric cancer risk after H. pylori infection}, 137215 {Microvascular complications of diabetes 4}, 612628
IL21R	149.6	100%	99%	Immunodeficiency, primary, autosomal recessive, IL21R-related, 615207 [IgE, elevated level of], 147050
IL2RA	140	100%	99%	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367 {Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942
IL2RG	75.4	99%	99%	Combined immunodeficiency, X-linked, moderate, 312863 Severe combined immunodeficiency, X-linked, 300400
IL31RA	151	100%	99%	Amyloidosis, primary localized cutaneous, 2, 613955
IL36RN	94.7	100%	99%	Psoriasis 14, pustular, 614204
IL7R	162.6	100%	82%	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
ILDR1	111.6	100%	98%	Deafness, autosomal recessive 42, 609646
IMPAD1	152.8	100%	97%	Chondrodysplasia with joint dislocations, GPAPP type, 614078
IMPDH1	59.3	93%	91%	Leber congenital amaurosis 11, 613837 Retinitis pigmentosa 10, 180105
IMPG1	125.5	99%	98%	Macular dystrophy, vitelliform, 4, 616151
IMPG2	191.4	99%	92%	Macular dystrophy, vitelliform, 5, 616152 Retinitis pigmentosa 56, 613581
INF2	110.1	93%	95%	Charcot-Marie-Tooth disease, dominant intermediate E, 614455 Glomerulosclerosis, focal segmental, 5, 613237
ING1	144.7	99%	99%	Squamous cell carcinoma, head and neck, somatic, 275355

INPP5E	109	97%	80%	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INPPL1	142.3	98%	94%	Opsismodysplasia, 258480
INS	97.3	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	85.3	81%	81%	Cryptorchidism, 219050
INSR	151.6	97%	86%	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968 Leprechaunism, 246200 Rabson-Mendenhall syndrome, 262190
INVS	166	100%	95%	Nephronophthisis 2, infantile, 602088
IQCB1	122	92%	99%	Senior-Loken syndrome 5, 609254
IQSEC2	72.6	94%	98%	Mental retardation, X-linked 1/78, 309530
IRAK4	110.5	99%	98%	Invasive pneumococcal disease, recurrent isolated, 1, 610799 IRAK4 deficiency, 607676
IRF1	187.1	100%	100%	Gastric cancer, somatic, 613659 Myelodysplastic syndrome, preleukemic Myelogenous leukemia, acute Nonsmall cell lung cancer, somatic, 211980
IRF6	134.4	99%	88%	Popliteal pterygium syndrome 1, 119500 van der Woude syndrome, 119300 {Orofacial cleft 6}, 608864
IRF8	124.4	99%	93%	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893 Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 614894
IRGM	159.6	100%	99%	Inflammatory bowel disease 19, 612278 {Mycobacterium tuberculosis, protection against}, 607948
IRX5	90.6	95%	100%	Hamamy syndrome, 611174
ISCA2	91.2	98%	92%	Multiple mitochondrial dysfunctions syndrome 4, 616370
ISCU	143.3	100%	95%	Myopathy with lactic acidosis, hereditary, 255125
ISG15	183.3	100%	97%	Immunodeficiency 38, 616126
ISPD	130.8	97%	98%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052
ITCH	143	95%	98%	Autoimmune disease, multisystem, with facial dysmorphism, 613385

ITGA2B	136.6	99%	96%	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Glanzmann thrombasthenia, 273800 Thrombocytopenia, neonatal alloimmune, BAK antigen related
ITGA3	159.6	99%	99%	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748
ITGA6	174.6	99%	99%	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730
ITGA7	145.6	99%	99%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
ITGA8	142.8	99%	95%	Renal hypodysplasia/aplasia 1, 191830
ITGB2	181.5	100%	95%	Leukocyte adhesion deficiency, 116920
ITGB3	151.9	99%	99%	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Glanzmann thrombasthenia, 273800 Purpura, posttransfusion Thrombocytopenia, neonatal alloimmune {Myocardial infarction, susceptibility to}, 608446
ITGB4	173.9	98%	99%	Epidermolysis bullosa of hands and feet, 131800 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa, junctional, with pyloric atresia, 226730
ITGB6	179.8	96%	100%	Amelogenesis imperfecta, type IH, 616221
ITK	150.8	100%	99%	Lymphoproliferative syndrome 1, 613011
ITM2B	137.9	100%	99%	Dementia, familial British, 176500 Dementia, familial Danish, 117300 ?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079
ITPA	144.8	100%	97%	Epileptic encephalopathy, early infantile, 35, 616647 [Inosine triphosphatase deficiency], 613850
ITPR1	177.7	100%	98%	Gillespie syndrome,206700 Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360
IVD	123.5	100%	100%	Isovaleric acidemia, 243500
IYD	141.6	99%	94%	Thyroid dyshormonogenesis 4, 274800
JAG1	160.7	99%	95%	Alagille syndrome, 118450 Tetralogy of Fallot, 187500 ?Deafness, congenital heart defects, and posterior embryotoxon
JAGN1	166.5	100%	98%	Neutropenia, severe congenital, 6, autosomal recessive, 616022
JAK2	113.7	96%	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	113.3	98%	99%	SCID, autosomal recessive, T-negative/B-positive type, 600802
JAM3	158.3	99%	99%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
JPH2	106.7	97%	87%	Cardiomyopathy, hypertrophic, 17, 613873
JPH3	159.2	100%	100%	Huntington disease-like 2, 606438
JUP	160.5	100%	99%	Arrhythmogenic right ventricular dysplasia 12, 611528 Naxos disease, 601214
KAL1	102.2	89%	89%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
KANK1	163.5	100%	99%	Cerebral palsy, spastic quadriplegic, 2, 612900
KANK2	171.3	100%	99%	Palmoplantar keratoderma and woolly hair, 616099
KANSL1	90.1	95%	99%	Koolen-De Vries syndrome, 610443
KARS	141.9	100%	100%	Deafness, autosomal recessive 89, 613916 ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641
KAT6A	202	100%	97%	Mental retardation, autosomal dominant 32, 616268
KAT6B	194.1	99%	99%	Genitopatellar syndrome, 606170 SBBYSS syndrome, 603736
KATNB1	166.2	100%	100%	Lissencephaly 6, with microcephaly, 616212
KBTBD13	118.6	99%	99%	Nemaline myopathy 6, autosomal dominant, 609273
KCNA1	179.6	100%	99%	Episodic ataxia/myokymia syndrome, 160120
KCNA2	178.9	100%	100%	Epileptic encephalopathy, early infantile, 32, 616366
KCNA5	160.1	100%	58%	Atrial fibrillation, familial, 7, 612240
KCNB1	150.8	100%	98%	Epileptic encephalopathy, early infantile, 26, 616056
KCNC1	218.2	100%	100%	Epilepsy, progressive myoclonic 7, 616187
KCNC3	148.5	72%	99%	Spinocerebellar ataxia 13, 605259
KCND3	210.5	100%	100%	Brugada syndrome 9, 616399 Spinocerebellar ataxia 19, 607346
KCNE1	538.9	100%	99%	Jervell and Lange-Nielsen syndrome 2, 612347 Long QT syndrome 5, 613695
KCNE2	165.4	100%	89%	Atrial fibrillation, familial, 4, 611493 Long QT syndrome 6, 613693
KCNE3	197	100%	100%	Brugada syndrome 6, 613119
KCNH1	195.4	100%	99%	Temple-Baraitser syndrome, 611816

				Zimmermann-Laband syndrome 1, 135500
KCNH2	118.6	95%	100%	Long QT syndrome 2, 613688 Short QT syndrome 1, 609620 {Long QT syndrome 2, acquired, susceptibility to}, 613688
KCNJ1	235	100%	99%	Bartter syndrome, type 2, 241200
KCNJ10	219.9	100%	100%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ11	281.8	100%	99%	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	204.7	100%	100%	Leber congenital amaurosis 16, 614186 Snowflake vitreoretinal degeneration, 193230
KCNJ2	210.8	100%	97%	Andersen syndrome, 170390 Atrial fibrillation, familial, 9, 613980 Short QT syndrome 3, 609622
KCNJ5	219.7	100%	100%	Hyperaldosteronism, familial, type III, 613677 Long QT syndrome 13, 613485
KCNJ6	192.3	100%	99%	Keppen-Lubinsky syndrome, 614098
KCNK3	178.5	99%	99%	Pulmonary hypertension, primary, 4, 615344
KCNK9	214.4	100%	89%	Birk-Barel mental retardation dysmorphism syndrome, 612292
KCNMA1	150.1	100%	95%	Generalized epilepsy and paroxysmal dyskinesia, 609446
KCNN4	155.8	100%	96%	Dehydrated hereditary stomatocytosis 2, 616689
KCNQ1	127.6	92%	92%	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	999999		94%	Beckwith-Wiedemann syndrome, 130650
KCNQ2	114.3	98%	99%	Epileptic encephalopathy, early infantile, 7, 613720 Myokymia, 121200 Seizures, benign neonatal, 1, 121200
KCNQ3	118.7	99%	85%	Seizures, benign neonatal, type 2, 121201

KCNQ4	155.5	94%	92%	Deafness, autosomal dominant 2A, 600101
KCNT1	133.4	95%	93%	Epilepsy, nocturnal frontal lobe, 5, 615005 Epileptic encephalopathy, early infantile, 14, 614959
KCNV2	154.5	100%	97%	Retinal cone dystrophy 3B, 610356
KCTD1	159	94%	95%	Scalp-ear-nipple syndrome, 181270
KCTD17	106.5	98%	89%	Dystonia 26, myoclonic, 616398
KCTD7	157.8	94%	99%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM1A	166	99%	100%	Cleft palate, psychomotor retardation, and distinctive facial features, 616728
KDM5C	123.7	98%	99%	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
KDM6A	118	95%	98%	Kabuki syndrome 2, 300867
KDR	170.7	100%	94%	Hemangioma, capillary infantile, somatic, 602089 {Hemangioma, capillary infantile, susceptibility to}, 602089
KERA	208.7	100%	100%	Cornea plana congenita, recessive, 217300
KHDC3L	151.4	100%	99%	Hydatidiform mole, recurrent, 2, 614293
KIAA0196	157.5	99%	95%	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, autosomal dominant, 603563
KIAA0586	133.4	98%	97%	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546
KIAA1279	191.4	100%	99%	Goldberg-Shprintzen megacolon syndrome, 609460
KIAA2022	176.1	100%	99%	Mental retardation, X-linked 98, 300912
KIF11	98	97%	98%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF1A	130.8	99%	99%	Mental retardation, autosomal dominant 9, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal recessive, 610357
KIF1B	186.4	99%	95%	Pheochromocytoma, 171300 ?Charcot-Marie-Tooth disease, type 2A1, 118210 {Neuroblastoma, susceptibility to, 1}, 256700
KIF1C	131.8	99%	99%	Spastic ataxia 2, autosomal recessive, 611302
KIF21A	147.3	99%	98%	Fibrosis of extraocular muscles, congenital, 1, 135700 Fibrosis of extraocular muscles, congenital, 3B, 135700
KIF22	179.3	100%	89%	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546
KIF2A	126.5	99%	99%	Cortical dysplasia, complex, with other brain malformations 3, 615411
KIF5A	143.4	100%	98%	Spastic paraplegia 10, autosomal dominant, 604187
KIF5C	133.7	99%	99%	Cortical dysplasia, complex, with other brain malformations 2, 615282

KIF7	95.3	95%	92%	Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120
KIRREL3	157.3	99%	97%	Mental retardation, autosomal dominant 4, 612581
KISS1R	97.2	99%	96%	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 ?Precocious puberty, central, 1, 176400
KIT	195.4	100%	98%	Gastrointestinal stromal tumor, familial, 606764 Germ cell tumors, 273300 Leukemia, acute myeloid, 601626 Mast cell disease, 154800 Piebaldism, 172800
KITLG	95.5	96%	87%	Deafness, congenital, unilateral or asymmetric, 616697 Hyperpigmentation with or without hypopigmentation, 145250 [Skin/hair/eye pigmentation 7, blond/brown hair], 611664
KIZ	174.5	99%	99%	Retinitis pigmentosa 69, 615780
KL	185.9	97%	100%	Tumoral calcinosis, hyperphosphatemic, 211900 {Coronary artery disease, susceptibility to}
KLC2	135.9	99%	99%	Spastic paraplegia, optic atrophy, and neuropathy, 609541
KLF1	61.9	95%	99%	Blood group--Lutheran inhibitor, 111150 Dyserythropoietic anemia, congenital, type IV, 613673 [Hereditary persistence of fetal hemoglobin], 613566
KLF11	198.8	100%	100%	Maturity-onset diabetes of the young, type VII, 610508
KLF6	147	100%	99%	Gastric cancer, somatic, 613659 Prostate cancer, somatic, 176807
KLHL10	206.1	100%	97%	Spermatogenic failure 11, 615081
KLHL3	156.5	99%	99%	Pseudohypoaldosteronism, type IID, 614495
KLHL40	160.3	100%	97%	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	228.9	100%	100%	Nemaline myopathy 9, 615731
KLHL7	154.6	99%	98%	Cold induced sweating syndrome 3, 617055 Retinitis pigmentosa 42, 612943
KLK4	210	99%	99%	Amelogenesis imperfecta, type IIA1, 204700
KLKB1	194.2	99%	99%	Fletcher factor (prekallikrein) deficiency, 612423
KLLN	143.7	100%	99%	Cowden syndrome 4, 615107
KMT2A	165.3	99%	99%	Leukemia, myeloid/lymphoid or mixed-lineage, 159555

				Wiedemann-Steiner syndrome, 605130
KMT2D	158.6	100%	98%	Kabuki syndrome 1, 147920
KPTN	122.9	100%	95%	Mental retardation, autosomal recessive 41, 615637
KRAS	89.6	99%	95%	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	118.2	100%	98%	Cavernous malformations of CNS and retina, 116860 Cerebral cavernous malformations-1, 116860 Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations, 116860
KRT1	142.2	100%	81%	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	120.2	98%	55%	Epidermolytic hyperkeratosis, 113800 Ichthyosis with confetti, 609165 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602
KRT12	130.8	98%	36%	Meesmann corneal dystrophy, 122100
KRT13	151.9	99%	67%	White sponge nevus 2, 615785
KRT14	56.7	89%	99%	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	39.1	72%	100%	Pachyonychia congenita 1, 167200

				Palmoplantar keratoderma, nonepidermolytic, focal, 613000
KRT17	23.6	55%	99%	Pachyonychia congenita 2, 167210 Steatocystoma multiplex, 184500
KRT18	38.2	83%	99%	Cirrhosis, cryptogenic, 215600 {Cirrhosis, noncryptogenic, susceptibility to}, 215600
KRT2	149	100%	99%	Ichthyosis bullosa of Siemens, 146800
KRT25	167.5	100%	88%	Woolly hair, autosomal recessive 3, 616760
KRT3	112.1	100%	90%	Meesmann corneal dystrophy, 122100
KRT4	138.6	100%	80%	White sponge nevus 1, 193900
KRT5	146.8	100%	99%	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	186.4	95%	76%	Pachyonychia congenita 3, 615726
KRT6B	187.8	97%	95%	Pachyonychia congenita 4, 615728
KRT6C	171.6	88%	94%	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735
KRT74	164	99%	97%	?Ectodermal dysplasia 7, hair/nail type, 614929 ?Hypotrichosis 3, 613981 Woolly hair, autosomal dominant, 194300
KRT8	43.1	92%	95%	Cirrhosis, cryptogenic, 215600 {Cirrhosis, noncryptogenic, susceptibility to}, 215600
KRT81	99.1	99%	97%	Monilethrix, 158000
KRT85	123.2	99%	97%	Ectodermal dysplasia 4, hair/nail type, 602032
KRT86	108.6	99%	99%	Monilethrix, 158000
KRT9	90.4	97%	99%	Palmoplantar keratoderma, epidermolytic, 144200
L1CAM	152.5	99%	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	139.7	98%	99%	L-2-hydroxyglutaric aciduria, 236792

LAMA1	154	100%	99%	Poretti-Boltshauser syndrome, 615960
LAMA2	176.7	100%	99%	Muscular dystrophy, congenital merosin-deficient, 607855 Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855
LAMA3	165.5	99%	99%	Epidermolysis bullosa, generalized atrophic benign, 226650 Epidermolysis bullosa, junctional, Herlitz type, 226700 Laryngoonychocutaneous syndrome, 245660
LAMA4	159.3	100%	97%	Cardiomyopathy, dilated, 1JJ, 615235
LAMB1	199.1	100%	96%	Lissencephaly 5, 615191
LAMB2	220.8	100%	91%	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 Pierson syndrome, 609049
LAMB3	134.9	100%	99%	Amelogenesis imperfecta, type IA, 104530 Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LAMC2	137.9	99%	99%	Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LAMC3	151.5	98%	70%	Cortical malformations, occipital, 614115
LAMP2	120.4	93%	100%	Danon disease, 300257
LAMTOR2	204.7	100%	97%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LARGE	142.4	100%	90%	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	68.5	82%	96%	Alazami syndrome, 615071
LARS2	157.5	100%	96%	Perrault syndrome 4, 615300 ?Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LAS1L	107	99%	98%	Wilson-Turner syndrome, 309585
LBR	97.8	95%	93%	Greenberg skeletal dysplasia, 215140 Pelger-Huet anomaly, 169400 ?Reynolds syndrome, 613471
LCA5	178	98%	89%	Leber congenital amaurosis 5, 604537
LCAT	166.1	99%	98%	Fish-eye disease, 136120 Norum disease, 245900
LCT	145.1	99%	96%	Lactase deficiency, congenital, 223000
LDB3	146.5	95%	99%	Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 Cardiomyopathy, hypertrophic, 24, 601493 Left ventricular noncompaction 3, 601493 Myopathy, myofibrillar, 4, 609452

LDHA	69.6	94%	76%	Glycogen storage disease XI, 612933
LDLR	192.2	99%	87%	Hypercholesterolemia, familial, 143890 LDL cholesterol level QTL2, 143890
LDLRAP1	188	98%	93%	Hypercholesterolemia, familial, autosomal recessive, 603813
LEF1	140	100%	99%	Sebaceous tumors, somatic
LEFTY2	46.5	89%	92%	Left-right axis malformations (Koasaki (1999) Am J Hum Genet 64, 712)
LEMD2	80.2	95%	99%	Cataract 46, juvenile-onset, 212500
LEMD3	116.1	98%	97%	Buschke-Ollendorff syndrome, 166700 Melorheostosis with osteopoikilosis, 155950 Osteopoikilosis, 166700
LEP	228.8	100%	98%	Obesity, morbid, due to leptin deficiency, 614962
LEPR	138.5	94%	64%	Obesity, morbid, due to leptin receptor deficiency, 614963
LEPRE1	149.8	100%	93%	Osteogenesis imperfecta, type VIII, 610915
LEPREL1	117.6	99%	100%	Myopia, high, with cataract and vitreoretinal degeneration, 614292
LGI1	207	99%	91%	Epilepsy, familial temporal lobe, 1, 600512
LHB	26.1	95%	99%	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300
LHCGR	188	97%	99%	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	306.5	100%	95%	Deafness, autosomal recessive 67, 610265
LHX3	107.4	98%	98%	Pituitary hormone deficiency, combined, 3, 221750
LHX4	158.6	100%	99%	Pituitary hormone deficiency, combined, 4, 262700
LIAS	175	100%	98%	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIFR	156.7	98%	92%	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LIG1	106.1	99%	99%	DNA ligase I deficiency
LIG4	207.5	100%	95%	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500
LIM2	98.4	99%	99%	Cataract 19, multiple types, 615277
LIMS2	129.2	93%	97%	Muscular dystrophy, limb-girdle, type 2W, 616827
LINS	158	99%	99%	Mental retardation, autosomal recessive 27, 614340
LIPA	128	97%	98%	Cholesteryl ester storage disease, 278000 Wolman disease, 278000

LIPC	130.1	100%	99%	Hepatic lipase deficiency, 614025 [High density lipoprotein cholesterol level QTL 12], 612797 {Diabetes mellitus, noninsulin-dependent}, 125853
LIPE	121.8	99%	92%	Lipodystrophy, familial partial, type 6, 615980
LIPH	154.3	100%	98%	Hypotrichosis 7, 604379 Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379
LIPN	153.5	99%	94%	Ichthyosis, congenital, autosomal recessive 8, 613943
LIPT1	269	100%	88%	Lipoyltransferase 1 deficiency, 616299
LITAF	136.8	96%	98%	Charcot-Marie-Tooth disease, type 1C, 601098
LMAN1	156.5	99%	90%	Combined factor V and VIII deficiency, 227300
LMBR1	128.9	98%	98%	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	96.5	93%	99%	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LMF1	150.1	99%	96%	Lipase deficiency, combined, 246650
LMNA	95.6	97%	97%	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	148.6	99%	56%	Leukodystrophy, adult-onset, autosomal dominant, 169500
LMOD3	197.4	100%	99%	Nemaline myopathy 10, 616165
LMX1B	131.5	99%	98%	Nail-patella syndrome, 161200
LONP1	164.9	98%	96%	CODAS syndrome, 600373

LOR	22.5	91%	99%	Vohwinkel syndrome with ichthyosis, 604117
LOXHD1	146.9	100%	99%	Deafness, autosomal recessive 77, 613079
LPAR6	123.4	100%	100%	Hypotrichosis 8, 278150 Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150
LPIN1	151.2	99%	100%	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	124.6	100%	98%	Majeed syndrome, 609628
LPL	175.7	100%	98%	Combined hyperlipidemia, familial, 144250 Lipoprotein lipase deficiency, 238600 [High density lipoprotein cholesterol level QTL 11]
LPP	143.4	100%	93%	Leukemia, acute myeloid, 601626 Lipoma
LRAT	345.6	100%	99%	Leber congenital amaurosis 14, 613341 Retinal dystrophy, early-onset severe, 613341 Retinitis pigmentosa, juvenile, 613341
LRBA	164.3	99%	98%	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRIG2	181.2	99%	98%	Urofacial syndrome 2, 615112
LRIT3	160.4	94%	99%	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRP2	205.6	100%	97%	Donnai-Barrow syndrome, 222448
LRP4	184.3	99%	97%	Cenani-Lenz syndactyly syndrome, 212780 Sclerosteosis 2, 614305 ?Myasthenic syndrome, congenital, 17, 616304
LRP5	210	98%	95%	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	178.9	100%	99%	Tooth agenesis, selective, 7, 616724 {Coronary artery disease, autosomal dominant, 2}, 610947
LRPAP1	156.3	99%	99%	Myopia 23, autosomal recessive, 615431
LRPPRC	157.1	99%	96%	Leigh syndrome, French-Canadian type, 220111
LRRC6	183.1	98%	99%	Ciliary dyskinesia, primary, 19, 614935
LRRC8A	300.4	100%	97%	Agammaglobulinemia 5, 613506

LRSAM1	157.9	100%	97%	Charcot-Marie-Toothe disease, axonal, type 2P, 614436
LRTOMT	147.8	99%	97%	Deafness, autosomal recessive 63, 611451
LSS	137.3	100%	78%	Cataract 44, 616509
LTBP2	121.8	99%	96%	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	128.7	98%	100%	Dental anomalies and short stature, 601216
LTBP4	137.6	99%	98%	Cutis laxa, autosomal recessive, type IC, 613177
LYRM7	65.3	92%	99%	Mitochondrial complex III deficiency, nuclear type 8, 615838
LYST	171	98%	100%	Chediak-Higashi syndrome, 214500
LYZ	187	100%	100%	Amyloidosis, renal, 105200
LZTFL1	142	99%	97%	Bardet-Biedl syndrome 17, 615994
LZTR1	166.5	100%	74%	Noonan syndrome 10, 616564 {Schwannomatosis-2, susceptibility to}, 615670
LZTS1	114.6	100%	99%	Esophageal squamous cell carcinoma, 133239
MAB21L2	261.2	100%	99%	Microphthalmia, syndromic 14, 615877
MAD1L1	112.4	99%	96%	Lymphoma, somatic Prostate cancer, somatic, 176807
MAF	67	79%	100%	Ayme-Gripp syndrome, 601088 Cataract 21, multiple types, 610202
MAFB	125.4	99%	97%	Duane retraction syndrome 3, 617041 Multicentric carpotarsal osteolysis syndrome, 166300
MAG	154.4	100%	94%	Spastic paraplegia 75, autosomal recessive, 616680
MAGED2	84.1	99%	87%	Bartter syndrome, type 5,antenatal,transient,300971
MAGEL2	136.6	100%	99%	Schaaf-Yang syndrome, 615547
MAGT1	118.7	98%	98%	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853
MAK	181.6	96%	99%	Retinitis pigmentosa 62, 614181
MALT1	153.1	90%	95%	Immunodeficiency 12, 615468
MAML2	132.5	100%	98%	Mucoepidermoid salivary gland carcinoma
MAMLD1	145.9	99%	99%	Hypospadias 2, X-linked, 300758
MAN1B1	162.6	100%	97%	Mental retardation, autosomal recessive 15, 614202
MAN2B1	134.6	98%	95%	Mannosidosis, alpha-, types I and II, 248500
MANBA	153.6	99%	89%	Mannosidosis, beta, 248510

MAOA	130.2	100%	99%	Brunner syndrome, 300615 {Antisocial behavior},300615
MAP2K1	106.2	99%	99%	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	122.2	99%	93%	Cardiofaciocutaneous syndrome 4, 615280
MAP3K1	187.2	94%	98%	46XY sex reversal 6, 613762
MAP3K8	166.9	100%	100%	Lung cancer, somatic, 211980
MAPRE2	221.3	100%	96%	Symmetric circumferential skin creases, congenital, 2, 616734
MAPT	100	98%	99%	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	140.1	99%	99%	Charcot-Marie-Tooth disease, axonal, type 2U, 616280 Interstitial lung and liver disease, 615486
MARS2	179.7	100%	97%	Spastic ataxia 3, autosomal recessive, 611390 ?Combined oxidative phosphorylation deficiency 25, 616430
MARVELD2	184.2	98%	84%	Deafness, autosomal recessive 49, 610153
MASP1	160.1	99%	93%	3MC syndrome 1, 257920
MASP2	179.7	100%	99%	MASP2 deficiency, 613791
MAT1A	206.1	99%	98%	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MATN3	125.7	84%	99%	Epiphyseal dysplasia, multiple, 5, 607078 Spondyloepimetaphyseal dysplasia, 608728 {Osteoarthritis susceptibility 2}, 140600
MATR3	107.4	97%	100%	Amyotrophic lateral sclerosis 21, 606070
MBD5	204.8	100%	99%	Mental retardation, autosomal dominant 1, 156200
MBTPS2	147.5	99%	99%	IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800 ?Olmsted syndrome, X-linked, 300918
MC2R	201.9	100%	99%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MC4R	287	100%	100%	Obesity, autosomal dominant, 601665
MCC	172.7	100%	99%	Colorectal cancer, somatic, 114500
MCCC1	176.8	100%	99%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200

MCCC2	159.5	100%	99%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	134.1	100%	100%	Methylmalonyl-CoA epimerase deficiency, 251120
MCFD2	114.3	100%	97%	Factor V and factor VIII, combined deficiency of, 613625
MCM4	175.6	100%	98%	Natural killer cell and glucocorticoid deficiency with DNA repair defect, 609981
MCM6	182.1	100%	99%	Lactase persistence/nonpersistence, 223100
MCM9	175	100%	95%	Ovarian dysgenesis 4, 616185
MCOLN1	168.5	99%	94%	Mucolipidosis IV, 252650
MCPH1	156.6	99%	99%	Microcephaly 1, primary, autosomal recessive, 251200
MECOM	179.9	100%	97%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MECP2	99.4	99%	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	111.4	98%	95%	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Opitz-Kaveggia syndrome, 305450
MED13L	141.7	99%	96%	Mental retardation and distinctive facial features with or without cardiac defects, 616789 Transposition of the great arteries, dextro-looped 1, 608808
MED17	156.8	99%	92%	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	162.9	99%	100%	Mental retardation, autosomal recessive 18, 614249
MED25	124.1	98%	98%	Basel-Vanagait-Smirin-Yosef syndrome, 616449 ?Charcot-Marie-Tooth disease, type 2B2, 605589
MEF2C	150.6	99%	97%	Chromosome 5q14.3 deletion syndrome, 613443 Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
MEFV	135.7	96%	95%	Familial Mediterranean fever, AD, 134610 Familial Mediterranean fever, AR, 249100
MEGF10	172.9	100%	99%	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399
MEGF8	142.7	99%	86%	Carpenter syndrome 2, 614976
MEN1	140.1	99%	99%	Adrenal adenoma, somatic Angiofibroma, somatic Carcinoid tumor of lung

				Lipoma, somatic Multiple endocrine neoplasia 1, 131100 Parathyroid adenoma, somatic
MEOX1	110.2	98%	100%	Klippel-Feil syndrome 2, 214300
MERTK	182.5	100%	98%	Retinitis pigmentosa 38, 613862
MESP2	82.3	93%	100%	Spondylocostal dysostosis 2, autosomal recessive, 608681
MET	223.1	100%	100%	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	154	100%	99%	Mental retardation, autosomal recessive 44, 615942
MFAP5	126.6	100%	99%	Aortic aneurysm, familial thoracic 9, 616166
MFN2	157.5	100%	100%	Charcot-Marie-Tooth disease, type 2A2A, 609260 Charcot-Marie-Tooth disease, type 2A2B, 617087 Hereditary motor and sensory neuropathy VIA, 601152
MFRP	140.1	100%	100%	Microphthalmia, isolated 5, 611040 Nanophthalmos 2, 609549
MFSD2A	134.8	99%	92%	Microcephaly 15, primary, autosomal recessive, 616486
MFSD8	143.4	100%	99%	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170
MGAT2	177.1	100%	93%	Congenital disorder of glycosylation, type IIa, 212066
MGME1	178.2	100%	98%	Mitochondrial DNA depletion syndrome 11, 615084
MGP	161.6	95%	97%	Keutel syndrome, 245150
MIB1	160.1	100%	95%	Left ventricular noncompaction 7, 615092
MICU1	134.3	97%	100%	Myopathy with extrapyramidal signs, 615673
MID1	177.2	99%	89%	Opitz GBBB syndrome, type I, 300000
MINPP1	165.6	99%	95%	Thyroid carcinoma, follicular, 188470
MIP	134.2	99%	99%	Cataract 15, multiple types, 615274
MIR17HG	999999		99%	Feingold syndrome 2, 614326
MIR184	999999		99%	EDICT syndrome, 614303
MIR96	999999		99%	Deafness, autosomal dominant 50, 613074
MITF	173.3	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	216.9	89%	93%	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKL1	108.9	99%	98%	Megakaryoblastic leukemia, acute
MKRN3	117.6	100%	96%	Precocious puberty, central, 2, 615346
MKS1	114.6	99%	91%	Bardet-Biedl syndrome 13, 615990 Meckel syndrome 1, 249000
MLC1	114.2	100%	99%	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MLH1	187.4	100%	99%	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MLH3	192.4	100%	100%	Colorectal cancer, hereditary nonpolyposis, type 7, 614385 Colorectal cancer, somatic, 114500 {Endometrial cancer, susceptibility to}, 608089
MLLT10	151.1	95%	80%	Leukemia,acute myeloid,601626
MLLT11	79.2	99%	95%	Leukemia, acute myelomonocytic, somatic, 607785
MLPH	110.2	99%	98%	Griscelli syndrome, type 3, 609227
MLYCD	94.5	95%	92%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	222.3	100%	98%	Methylmalonic aciduria, vitamin B12-responsive, 251100
MMAB	118.2	100%	99%	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110
MMACHC	227.3	100%	98%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	94.4	90%	88%	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410
MME	129.2	99%	97%	Charcot-Marie-Tooth disease, axonal, type 2T, 617017 ?Spinocerebellar ataxia 43, 617018
MMP1	187.1	99%	96%	COPD, rate of decline of lung function in, 606963 {Epidermolysis bullosa dystrophica, autosomal recessive, modifier of}, 226600
MMP13	145.7	92%	57%	Metaphyseal anadysplasia 1, 602111 Spondyloepimetaphyseal dysplasia, Missouri type, 602111
MMP19	146.9	99%	98%	Cavitary optic disc anomalies, 611543
MMP2	173.5	100%	92%	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP20	112.5	99%	99%	Amelogenesis imperfecta, type IIA2, 612529

MMP21	115.9	93%	98%	Heterotaxy, visceral, 7, autosomal, 616749
MMP9	134.7	99%	99%	Metaphyseal anadysplasia 2, 613073
MN1	110.4	99%	99%	Meningioma, 607174
MNX1	38.4	68%	99%	Currarino syndrome, 176450
MOCOS	196.4	99%	97%	Xanthinuria, type II, 603592
MOCS1	92.7	98%	100%	Molybdenum cofactor deficiency A, 252150
MOCS2	183	99%	97%	Molybdenum cofactor deficiency B, 252160
MOGS	128	99%	91%	Congenital disorder of glycosylation, type IIb, 606056
MORC2	147.6	100%	99%	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688
MPC1	160.7	100%	99%	Mitochondrial pyruvate carrier deficiency,614741
MPDU1	129.4	100%	99%	Congenital disorder of glycosylation, type If, 609180
MPDZ	171.5	98%	100%	Hydrocephalus, nonsyndromic, autosomal recessive 2, 615219
MPI	142	100%	89%	Congenital disorder of glycosylation, type Ib, 602579
MPL	163.9	99%	82%	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498
MPLKIP	103.8	99%	99%	Trichothiodystrophy 4, nonphotosensitive, 234050
MPO	160.6	100%	93%	Myeloperoxidase deficiency, 254600 {Alzheimer disease, susceptibility to}, 104300 {Lung cancer, protection against, in smokers}
MPV17	112.5	100%	97%	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MPZ	128.1	99%	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	204	100%	97%	Glucocorticoid deficiency 2, 607398
MRE11A	64.3	97%	99%	Ataxia-telangiectasia-like disorder, 604391
MRPL3	74.4	92%	82%	Combined oxidative phosphorylation deficiency 9, 614582
MRPS16	159.3	100%	99%	Combined oxidative phosphorylation deficiency 2, 610498
MRPS22	167.4	97%	98%	Combined oxidative phosphorylation deficiency 5, 611719
MS4A1	145.2	99%	99%	Immunodeficiency, common variable, 5, 613495

MSH2	137.3	99%	94%	Colorectal cancer, hereditary nonpolyposis, type 1, 120435 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MSH3	142.5	99%	96%	Endometrial carcinoma, somatic, 608089 Familial adenomatous polyposis 4,617100
MSH6	190.9	100%	88%	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 Endometrial cancer, familial, 608089 Mismatch repair cancer syndrome, 276300
MSMO1	57.3	93%	96%	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MSR1	211.6	100%	100%	Barrett esophagus/esophageal adenocarcinoma, 614266 Prostate cancer, hereditary, 176807
MSRB3	160	99%	93%	Deafness, autosomal recessive 74, 613718
MSTN	215.3	99%	99%	Muscle hypertrophy, 614160
MSX1	93.5	98%	88%	Ectodermal dysplasia 3, Witkop type, 189500 Orofacial cleft 5, 608874 Tooth agenesis, selective, 1, with or without orofacial cleft, 106600
MSX2	116.3	99%	99%	Craniosynostosis, type 2, 604757 Parietal foramina 1, 168500 Parietal foramina with cleidocranial dysplasia, 168550
MTAP	113.9	95%	94%	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250
MTFMT	148.9	99%	99%	Combined oxidative phosphorylation deficiency 15, 614947
MTHFR	161.7	100%	99%	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	108.7	98%	99%	Myotubular myopathy, X-linked, 310400
MTMR2	130	100%	94%	Charcot-Marie-Tooth disease, type 4B1, 601382
MTO1	185.3	90%	100%	Combined oxidative phosphorylation deficiency 10, 614702
MTOR	152.6	100%	98%	Smith-Kingsmore syndrome, 616638
MTPAP	133.3	98%	99%	Ataxia, spastic, 4, 613672
MTR	174.9	99%	98%	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTRR	152.5	99%	99%	Homocystinuria-megaloblastic anemia, cbl E type, 236270 {Neural tube defects, folate-sensitive, susceptibility to}, 601634

MTTP	162.6	100%	87%	Abetalipoproteinemia, 200100 {Metabolic syndrome, protection against}, 605552
MUC1	105	98%	99%	Medullary cystic kidney disease 1, 174000
MUSK	178.8	100%	97%	Fetal akinesia deformation sequence, 208150 Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325
MUT	154.5	99%	100%	Methylmalonic aciduria, mut(0) type, 251000
MUTYH	178	100%	86%	Adenomas, multiple colorectal, 608456 Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600 Gastric cancer, somatic, 613659
MVD	114.6	99%	99%	Porokeratosis 7, multiple types, 614714
MVK	167.2	100%	100%	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900
MXI1	129.8	91%	99%	Neurofibrosarcoma {Prostate cancer, susceptibility to}, 176807
MYBPC1	172.4	99%	93%	Arthrogryposis, distal, type 1B, 614335 Lethal congenital contracture syndrome 4, 614915
MYBPC3	161.5	99%	99%	Cardiomyopathy, dilated, 1MM, 615396 Cardiomyopathy, hypertrophic, 4, 115197 Left ventricular noncompaction 10, 615396
MYC	208	100%	98%	Burkitt lymphoma, 113970
MYCN	117	95%	97%	Feingold syndrome, 164280
MYD88	195.9	100%	96%	Macroglobulinemia, Waldenstrom, somatic, 153600 Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260
MYF6	130.1	100%	99%	Myopathy, centronuclear, 3, 614408
MYH11	152.8	100%	98%	Aortic aneurysm, familial thoracic 4, 132900
MYH14	118.4	98%	96%	Deafness, autosomal dominant 4A, 600652 ?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369
MYH2	146.3	99%	100%	Proximal myopathy and ophthalmoplegia, 605637
MYH3	123.5	99%	99%	Arthrogryposis, distal, type 2A, 193700 Arthrogryposis, distal, type 2B, 601680 Arthrogryposis, distal, type 8, 178110
MYH6	127.6	99%	99%	Atrial septal defect 3, 614089 Cardiomyopathy, dilated, 1EE, 613252 Cardiomyopathy, hypertrophic, 14, 613251

				{Sick sinus syndrome 3}, 614090
MYH7	123.2	99%	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	152.7	100%	98%	Carney complex variant, 608837 Trismus-pseudocamptodactyly syndrome, 158300
MYH9	152.2	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	153.4	99%	94%	Cardiomyopathy, hypertrophic, 10, 608758
MYL3	124.4	100%	98%	Cardiomyopathy, hypertrophic, 8, 608751
MYLK	168.7	99%	96%	Aortic aneurysm, familial thoracic 7, 613780
MYLK2	121.5	100%	94%	Cardiomyopathy, hypertrophic, 1, digenic, 192600
MYO15A	136.6	97%	98%	Deafness, autosomal recessive 3, 600316
MYO18B	143	99%	99%	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549
MYO1E	146.8	99%	98%	Glomerulosclerosis, focal segmental, 6, 614131
MYO3A	133.3	98%	100%	Deafness, autosomal recessive 30, 607101
MYO5A	138.4	99%	98%	Griscelli syndrome, type 1, 214450
MYO5B	154.8	98%	99%	Microvillus inclusion disease, 251850
MYO6	108.6	98%	96%	Deafness, autosomal dominant 22, 606346 Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346 Deafness, autosomal recessive 37, 607821
MYO7A	149.2	99%	100%	Deafness, autosomal dominant 11, 601317 Deafness, autosomal recessive 2, 600060 Usher syndrome, type 1B, 276900
MYOC	196.1	100%	91%	Glaucoma 1A, primary open angle, 137750
MYOT	172.4	99%	92%	Muscular dystrophy, limb-girdle, type 1A, 159000 Myopathy, myofibrillar, 3, 609200

				Myopathy, spheroid body, 182920
MYOZ2	194.6	100%	98%	Cardiomyopathy, hypertrophic, 16, 613838
MYPN	165.3	99%	81%	Cardiomyopathy, dilated, 1KK, 615248 Cardiomyopathy, familial restrictive, 4, 615248 Cardiomyopathy, hypertrophic, 22, 615248
MYT1L	199.4	100%	99%	Mental retardation, autosomal dominant 39, 616521
NAA10	109.4	99%	97%	Ogden syndrome, 300855 ?Microphthalmia, syndromic 1, 309800
NAGA	159.7	100%	98%	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241
NAGLU	131.8	94%	99%	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491
NAGS	84	97%	98%	N-acetylglutamate synthase deficiency, 237310
NALCN	147	99%	22%	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419
NANOS1	44.4	93%	97%	Spermatogenic failure 12, 615413
NANS	115	100%	100%	Sponyloepimetaphyseal dysplasia, Genevieve type,610442
NARS2	153	97%	99%	Combined oxidative phosphorylation deficiency 24, 616239
NBAS	176.4	99%	99%	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NBEAL2	189.5	99%	88%	Gray platelet syndrome, 139090
NBN	106	99%	100%	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260
NCF1	23.9	26%	99%	Chronic granulomatous disease due to deficiency of NCF-1, 233700
NCF2	145.9	99%	100%	Chronic granulomatous disease due to deficiency of NCF-2, 233710
NCF4	188.3	100%	100%	?Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III, 613960
NCSTN	129.2	100%	95%	Acne inversa, familial, 1, 142690
NDE1	106.3	100%	100%	Lissencephaly 4 (with microcephaly), 614019 ?Microhydranencephaly, 605013
NDN	98.5	96%	100%	Prader-Willi syndrome, 176270
NDP	126.5	100%	97%	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600

NDRG1	144.6	100%	100%	Charcot-Marie-Tooth disease, type 4D, 601455
NDST1	231.1	100%	76%	Mental retardation, autosomal recessive 46, 616116
NDUFA1	229	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFA11	95.2	99%	97%	Mitochondrial complex I deficiency, 252010
NDUFA12	175	100%	98%	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000
NDUFA2	146.8	100%	98%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFA9	154	99%	87%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFAF1	128.7	100%	76%	Mitochondrial complex I deficiency, 252010
NDUFAF2	48.3	87%	99%	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFAF3	130.4	100%	99%	Mitochondrial complex I deficiency, 252010
NDUFAF4	125.1	99%	90%	Mitochondrial complex I deficiency, 252010
NDUFAF5	108.8	99%	100%	Mitochondrial complex 1 deficiency, 252010
NDUFAF6	106.3	99%	99%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFB11	108.3	95%	99%	Linear skin defects with multiple congenital anomalies 3, 300952
NDUFB3	29.3	96%	99%	Mitochondrial complex I deficiency, 252010
NDUFS1	165.9	100%	97%	Mitochondrial complex I deficiency, 252010
NDUFS2	121.6	100%	73%	Mitochondrial complex I deficiency, 252010
NDUFS3	149.4	90%	82%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
NDUFS4	200.6	100%	95%	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFS6	146.2	100%	98%	Mitochondrial complex I deficiency, 252010
NDUFS7	141.6	100%	96%	Leigh syndrome, 256000
NDUFS8	146.8	100%	98%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFV1	158	99%	43%	Mitochondrial complex I deficiency, 252010
NDUFV2	88.7	90%	100%	Mitochondrial complex I deficiency, 252010
NEB	154.8	83%	99%	Nemaline myopathy 2, autosomal recessive, 256030
NEFH	114.3	98%	83%	Charcot-Marie-Tooth disease,axonal,type 2CC,616924 ?{Amyotrophic lateral sclerosis, susceptibility to}, 105400
NEFL	175.9	99%	91%	Charcot-Marie-Tooth disease, type 1F, 607734 Charcot-Marie-Tooth disease, type 2E, 607684
NEK1	142.4	99%	99%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NEK9	150	99%	96%	Lethal congenital contracture syndrome 10, 617022

				Nevus comedonicus, somatic, 617025 ?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262
NEU1	20.4	72%	96%	Sialidosis, type I, 256550 Sialidosis, type II, 256550
NEUROD1	166.6	100%	96%	Maturity-onset diabetes of the young 6, 606394 {Diabetes mellitus, noninsulin-dependent}, 125853
NEUROG3	128.5	100%	93%	Diarrhea 4, malabsorptive, congenital, 610370
NEXN	87.1	93%	86%	Cardiomyopathy, dilated, 1CC, 613122 Cardiomyopathy, hypertrophic, 20, 613876
NF1	146.2	93%	100%	Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321 Watson syndrome, 193520
NF2	113.8	100%	99%	Meningioma, NF2-related, somatic, 607174 Neurofibromatosis, type 2, 101000 Schwannomatosis, 162091
NFIX	166.6	98%	99%	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753
NFKB1	124.4	99%	99%	Immunodeficiency, common variable, 12, 616576
NFKB2	146.9	99%	99%	Immunodeficiency, common variable, 10, 615577
NFKBIA	126	98%	93%	Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency, 612132
NFU1	59.7	96%	99%	Multiple mitochondrial dysfunctions syndrome 1, 605711
NGF	268.3	100%	98%	Neuropathy, hereditary sensory and autonomic, type V, 608654
NGLY1	162.3	100%	96%	Congenital disorder of deglycosylation, 615273
NHEJ1	85.5	100%	95%	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
NHLRC1	192.8	100%	92%	Epilepsy, progressive myoclonic 2B (Lafora), 254780
NHP2	101.6	100%	99%	Dyskeratosis congenita, autosomal recessive 2, 613987
NHS	138.9	96%	98%	Cataract 40, X-linked, 302200 Nance-Horan syndrome, 302350
NIN	173	99%	71%	Seckel syndrome 7, 614851
NIPA1	179.3	99%	98%	Spastic paraplegia 6, autosomal dominant, 600363
NIPAL4	152.5	99%	99%	Ichthyosis, congenital, autosomal recessive 6, 612281

NIPBL	142.5	97%	99%	Cornelia de Lange syndrome 1, 122470
NKX2-1	58.2	99%	99%	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 {Thyroid cancer, monmedullary, 1}, 188550
NKX2-5	96.3	100%	98%	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	126.1	100%	99%	Conotruncal heart malformations, 217095 Persistent truncus arteriosus, 217095
NKX3-2	60.3	90%	93%	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330
NLGN4X	189	99%	99%	Mental retardation, X-linked, 300495 {Asperger syndrome susceptibility, X-linked 2}, 300497 {Autism susceptibility, X-linked 2}, 300495
NLRC4	202.8	100%	97%	Autoinflammation with infantile enterocolitis, 616050 ?Familial cold autoinflammatory syndrome 4, 616115
NLRP12	184.3	99%	96%	Familial cold autoinflammatory syndrome 2, 611762
NLRP3	162.9	100%	99%	CINCA syndrome, 607115 Familial cold-induced inflammatory syndrome 1, 120100 Muckle-Wells syndrome, 191900
NLRP7	145.5	99%	100%	Hydatidiform mole,recurrent, 1,231090
NME1	114.4	100%	100%	Neuroblastoma, 256700
NME8	128.5	97%	88%	Ciliary dyskinesia, primary, 6, 610852
NMNAT1	144.4	100%	96%	Leber congenital amaurosis 9, 608553
NNT	166.9	99%	99%	Glucocorticoid deficiency 4, 614736
NOBOX	91.9	99%	97%	Premature ovarian failure 5, 611548
NOD2	149.9	100%	98%	Blau syndrome, 186580 Yao syndrome, 617321 {Inflammatory bowel disease 1}, 266600 {Psoriatic arthritis, susceptibility to}, 607507
NODAL	155.4	100%	99%	Heterotaxy, visceral, 5, 270100
NOG	219.2	100%	91%	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	99.9	95%	99%	Myoclonus, familial cortical, 614937
NONO	110.4	99%	99%	Mental retardation, X-linked, syndromic 34, 300967
NOP10	159.6	100%	98%	Dyskeratosis congenita, autosomal recessive 1, 224230
NOP56	131.8	99%	97%	Spinocerebellar ataxia 36, 614153
NOTCH1	154.9	99%	99%	Adams-Oliver syndrome 5, 616028 Aortic valve disease 1, 109730
NOTCH2	180.3	100%	98%	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NOTCH3	129.3	95%	96%	Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310 Lateral meningocele syndrome, 130720 ?Myofibromatosis, infantile 2, 615293
NPC1	153.4	99%	82%	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220 {Nasopharyngeal carcinoma 1}
NPC2	148.6	100%	100%	Niemann-pick disease, type C2, 607625
NPHP1	154.6	99%	99%	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NPHP3	146	99%	98%	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540
NPHP4	152.6	100%	99%	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
NPHS1	114.9	99%	95%	Nephrotic syndrome, type 1, 256300
NPHS2	111	99%	100%	Nephrotic syndrome, type 2, 600995
NPM1	96.7	94%	98%	Leukemia, acute myeloid, somatic, 601626
NPPA	124	100%	95%	Atrial fibrillation, familial, 6, 612201 Atrial standstill 2, 615745
NPR2	178.1	100%	100%	Acromesomelic dysplasia, Maroteaux type, 602875 Epiphyseal chondrodysplasia, Miura type, 615923 Short stature with nonspecific skeletal abnormalities, 616255
NR0B1	130	99%	97%	46XY sex reversal 2, dosage-sensitive, 300018

				Adrenal hypoplasia, congenital, with hypogonadotropic hypogonadism, 300200
NR0B2	115.6	100%	96%	Obesity, mild, early-onset, 601665
NR1H4	152.1	98%	99%	Cholestasis, progressive familial intrahepatic 5, 617049
NR2E3	117.7	100%	100%	Enhanced S-cone syndrome, 268100 Retinitis pigmentosa 37, 611131
NR2F1	216.5	99%	96%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NR2F2	255.8	99%	98%	Congenital heart defects, multiple types, 4, 615779
NR3C1	165.1	100%	100%	Glucocorticoid resistance, 615962
NR3C2	176.6	99%	99%	Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115 Pseudohypoaldosteronism type I, autosomal dominant, 177735
NR4A3	124.3	99%	95%	Chondrosarcoma,extraskeletal myxoid,612237
NR5A1	102.1	100%	94%	46XY sex reversal 3, 612965 Adrenocortical insufficiency Premature ovarian failure 7, 612964 Spermatogenic failure 8, 613957
NRAS	203.3	100%	95%	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	67.2	99%	82%	Retinal degeneration, autosomal recessive, clumped pigment type Retinitis pigmentosa 27, 613750
NRXN1	193.5	99%	99%	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332
NSD1	181.1	100%	84%	Beckwith-Wiedemann syndrome, 130650 Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550
NSDHL	196.4	100%	94%	CHILD syndrome, 308050 CK syndrome, 300831
NSMF	116.6	96%	97%	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
NSUN2	133.7	95%	88%	Mental retardation, autosomal recessive 5, 611091
NT5C2	148.4	97%	99%	Spastic paraplegia 45, autosomal recessive, 613162

NT5C3A	80.9	92%	97%	Anemia, hemolytic, due to UMPH1 deficiency, 266120
NT5E	198.7	100%	99%	Calcification of joints and arteries, 211800
NTF4	66.7	97%	99%	Glaucoma 1, open angle, 1O, 613100
NTHL1	118.4	98%	95%	Familial adenomatous polyposis 3, 616415
NTRK1	160.6	99%	95%	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma, familial, 155240
NUBPL	102.9	92%	71%	Mitochondrial complex I deficiency, 252010
NUMA1	135.7	100%	97%	Leukemia, acute promyelocytic, somatic, 612376
NUP107	147	99%	97%	Nephrotic syndrome, type 11, 616730
NUP214	176.5	99%	100%	Leukemia, acute myeloid, somatic, 601626 Leukemia, T-cell acute lymphoblastic, somatic, 613065
NUP62	113.7	100%	98%	Striatonigral degeneration, infantile, 271930
NUP93	154.5	97%	75%	Nephrotic syndrome, type 12,616892
NYX	81.5	98%	99%	Night blindness, congenital stationary (complete), 1A, X-linked, 310500
OAT	96.2	80%	97%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OBSL1	152.4	99%	97%	3-M syndrome 2, 612921
OCA2	141.6	99%	97%	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	239.3	100%	98%	Band-like calcification with simplified gyration and polymicrogyria, 251290
OCRL	140.5	99%	62%	Dent disease 2, 300555 Lowe syndrome, 309000
OFD1	59.2	87%	58%	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424
OGG1	162.2	100%	100%	Renal cell carcinoma, clear cell, somatic, 144700
OPA1	146.6	99%	99%	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	125.8	99%	92%	3-methylglutaconic aciduria, type III, 258501

				Optic atrophy 3 with cataract, 165300
OPHN1	103.5	99%	98%	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
OPLAH	124.4	99%	89%	5-oxoprolinase deficiency, 260005
OPN1LW	85.7	68%	100%	Blue cone monochromacy, 303700 Colorblindness, protan, 303900
OPN1MW	69.7	65%	99%	Blue cone monochromacy, 303700 Colorblindness, deutan, 303800
OPN1SW	141	100%	99%	Colorblindness, tritan, 190900
OPTN	120	100%	89%	Amyotrophic lateral sclerosis 12, 613435 Glaucoma 1, open angle, E, 137760 {Glaucoma, normal tension, susceptibility to}, 606657
ORAI1	243	94%	99%	Immunodeficiency 9, 612782 Myopathy, tubular aggregate, 2, 615883
ORC1	129.5	99%	96%	Meier-Gorlin syndrome 1, 224690
ORC4	81.4	97%	99%	Meier-Gorlin syndrome 2, 613800
ORC6	152.4	100%	99%	Meier-Gorlin syndrome 3, 613803
OSBPL2	157	100%	96%	Deafness, autosomal dominant 67, 616340
OSMR	173.4	100%	99%	Amyloidosis, primary localized cutaneous, 1, 105250
OSTM1	96.5	92%	95%	Osteopetrosis, autosomal recessive 5, 259720
OTC	128.2	99%	99%	Ornithine transcarbamylase deficiency, 311250
OTOA	134.4	98%	99%	Deafness, autosomal recessive 22, 607039
OTOF	150.8	100%	99%	Auditory neuropathy, autosomal recessive, 1, 601071 Deafness, autosomal recessive 9, 601071
OTOG	160.5	100%	100%	Deafness, autosomal recessive 18B, 614945
OTOGL	139.5	99%	94%	Deafness, autosomal recessive 84B, 614944
OTX2	140.5	100%	61%	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125
OVOL2	137.5	98%	95%	Corneal dystrophy, posterior polymorphous, 1, 122000
OXCT1	146.5	99%	87%	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050
P2RX1	134.3	100%	100%	Bleeding disorder due to P2RX1 defect, somatic, 609821
P2RX2	139.1	99%	93%	Deafness, autosomal dominant 41, 608224
P2RY12	240.6	100%	99%	Bleeding disorder, platelet-type, 8, 609821
P4HB	123.6	94%	64%	Cole-Carpenter syndrome 1, 112240

PABPN1	88.3	66%	98%	Oculopharyngeal muscular dystrophy, 164300
PACS1	130.4	97%	99%	Schuss-Hoeijmakers-syndrome, 615009
PAFAH1B1	125	92%	99%	Lissencephaly 1, 607432 Subcortical laminar heterotopia, 607432
PAH	186.6	100%	98%	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600
PAK3	94.1	98%	99%	Mental retardation, X-linked 30/47, 300558
PALB2	180	100%	99%	Fanconi anemia, complementation group N, 610832 {Breast cancer, susceptibility to}, 114480 {Pancreatic cancer, susceptibility to, 3}, 613348
PAM16	57.4	65%	99%	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320
PANK2	178.4	99%	97%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PAPSS2	113.6	99%	100%	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847
PARK2	142.8	99%	100%	Adenocarcinoma of lung, somatic, 211980 Adenocarcinoma, ovarian, somatic, 167000 Parkinson disease, juvenile, type 2, 600116 {Leprosy, susceptibility to}, 607572
PARK7	88.1	99%	98%	Parkinson disease 7,autosomal recessive early-onset,606324
PARN	151.5	100%	99%	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
PAX2	200.3	100%	97%	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330
PAX3	128.2	100%	99%	Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220 Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820
PAX4	96.8	99%	93%	Diabetes mellitus, type 2, 125853 Maturity-onset diabetes of the young, type IX, 612225 {Diabetes mellitus, ketosis-prone, susceptibility to}, 612227
PAX6	135.8	100%	96%	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	124.9	100%	99%	Rhabdomyosarcoma 2, alveolar, 268220
PAX8	101.3	99%	99%	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700
PAX9	266	99%	96%	Tooth agenesis, selective, 3, 604625
PC	166.1	99%	99%	Pyruvate carboxylase deficiency, 266150
PCBD1	129.9	99%	92%	Hyperphenylalaninemia, BH4-deficient, D, 264070
PCCA	120.1	98%	96%	Propionicacidemia, 606054
PCCB	167.8	98%	88%	Propionicacidemia, 606054
PCDH15	179.5	99%	99%	Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1F, 602083
PCDH19	219.8	99%	99%	Epileptic encephalopathy, early infantile, 9, 300088
PCNT	128.9	99%	99%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PCSK1	175.4	100%	96%	Obesity with impaired prohormone processing, 600955 {Obesity, susceptibility to, BMIQ12}, 612362
PCSK9	111.2	97%	99%	Hypercholesterolemia, familial, 3, 603776 {Low density lipoprotein cholesterol level QTL 1}, 603776
PCYT1A	131.4	99%	100%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDCD10	109.1	97%	96%	Cerebral cavernous malformations 3, 603285
PDE10A	172.5	99%	97%	Dyskinesia,limb and orofacial,infantile-onset,616921 Striatal degeneration,autosomal dominant,616922
PDE11A	191.5	100%	65%	Pigmented nodular adrenocortical disease, primary, 2, 610475
PDE3A	138	99%	99%	Hypertension and brachydactyly syndrome, 112410
PDE4D	132.3	98%	100%	Acrodysostosis 2, with or without hormone resistance, 614613 {Stroke, susceptibility to, 1}, 606799
PDE6A	140.4	100%	100%	Retinitis pigmentosa 43, 613810
PDE6B	174.5	100%	95%	Night blindness, congenital stationary, autosomal dominant 2, 163500 Retinitis pigmentosa-40, 613801
PDE6C	148.4	98%	99%	Cone dystrophy 4, 613093
PDE6G	113.4	99%	92%	Retinitis pigmentosa 57, 613582
PDE6H	74.7	93%	97%	Achromatopsia 6, 610024

				Retinal cone dystrophy 3, 610024
PDE8B	125	99%	97%	Pigmented nodular adrenocortical disease, primary, 3, 614190 Striatal degeneration, autosomal dominant, 609161
PDGFB	110.3	100%	100%	Basal ganglia calcification, idiopathic, 5, 615483 Dermatofibrosarcoma protuberans, 607907 Meningioma, SIS-related, 607174
PDGFRA	172.3	100%	87%	Gastrointestinal stromal tumor, somatic, 606764 Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685
PDGFRB	160.1	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	162	100%	75%	Colorectal cancer, somatic, 114500 Hepatocellular cancer, somatic, 114550
PDHA1	112.7	98%	100%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	148.5	99%	98%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	133.8	99%	98%	Lacticacidemia due to PDX1 deficiency,245349
PDP1	197.1	100%	99%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	140.2	93%	86%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	146.3	98%	97%	Coenzyme Q10 deficiency, primary, 3, 614652
PDX1	39.1	93%	92%	MODY, type IV, 606392 Pancreatic agenesis 1, 260370 {Diabetes mellitus, type II, susceptibility to}, 125853
PDYN	136.3	100%	99%	Spinocerebellar ataxia 23, 610245
PDZD7	113.2	99%	99%	Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472 {Retinal disease in Usher syndrome type IIA, modifier of}, 276901
PEPD	127	99%	99%	Prolidase deficiency, 170100
PER2	114.2	100%	98%	Advanced sleep phase syndrome, familial, 1, 604348
PET100	98.1	98%	93%	Mitochondrial complex IV deficiency, 220110
PEX1	139.9	98%	99%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	124.7	97%	100%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871

PEX11B	103.8	100%	99%	Peroxisome biogenesis disorder 14B, 614920
PEX12	157.9	100%	97%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	240	100%	98%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	145.5	99%	85%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	146.3	97%	87%	Peroxisome biogenesis disorder 8A, (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	118.8	100%	99%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	187.6	100%	100%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	87.2	100%	100%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	129.6	99%	91%	Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	132.1	99%	100%	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	95.9	91%	94%	Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863
PEX7	152.7	90%	80%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PFKM	165.3	100%	99%	Glycogen storage disease VII, 232800
PFN1	177.4	100%	100%	Amyotrophic lateral sclerosis 18, 614808
PGAM2	177.3	100%	97%	Glycogen storage disease X, 261670
PGAP1	125.3	97%	86%	Mental retardation, autosomal recessive 42, 615802
PGAP2	173.9	100%	97%	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGAP3	126.1	98%	99%	Hyperphosphatasia with mental retardation syndrome 4, 615716
PGK1	57.8	92%	95%	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	149.5	100%	99%	Congenital disorder of glycosylation, type It, 614921
PGM3	225	100%	99%	Immunodeficiency 23, 615816
PHEX	138.1	98%	100%	Hypophosphatemic rickets, X-linked dominant, 307800
PHF6	74.9	94%	45%	Borjeson-Forssman-Lehmann syndrome, 301900
PHF8	102.5	99%	88%	Mental retardation syndrome, X-linked, Siderius type, 300263

PHGDH	138	100%	93%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHKA1	120.6	99%	89%	Muscle glycogenosis, 300559
PHKA2	119.9	99%	97%	Glycogen storage disease, type IXa1, 306000 Glycogen storage disease, type IXa2, 306000
PHKB	147.7	100%	98%	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750
PHKG2	173.9	100%	99%	Cirrhosis due to liver phosphorylase kinase deficiency Glycogen storage disease IXc, 613027
PHOX2A	49.8	71%	86%	Fibrosis of extraocular muscles, congenital, 2, 602078
PHOX2B	106.6	94%	99%	Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, 209880 Neuroblastoma with Hirschsprung disease, 613013 {Neuroblastoma, susceptibility to, 2}, 613013
PHYH	90	98%	99%	Refsum disease, 266500
PI4KA	118	93%	100%	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531
PICALM	120.8	99%	94%	Leukemia, acute myeloid, somatic, 601626
PIEZO1	158.2	99%	99%	Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380 Lymphedema, hereditary, III, 616843
PIEZO2	135.3	99%	99%	Arthrogryposis, distal, type 3, 114300 Arthrogryposis, distal, type 5, 108145 Arthrogryposis, distal, with impaired proproception and touch, 617146 ?Marden-Walker syndrome, 248700
PIGA	97	94%	100%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGG	192.9	100%	99%	Mental retardation,autosomal recessive 53,616917
PIGL	125.2	100%	99%	CHIME syndrome, 280000
PIGM	159.5	100%	97%	Glycosylphosphatidylinositol deficiency, 610293
PIGN	130.2	99%	98%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	143.9	100%	87%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGT	195.8	100%	99%	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 ?Paroxysmal nocturnal hemoglobinuria 2, 615399
PIGV	168	100%	98%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIGY	121.8	100%	89%	Hyperphosphatasia with mental retardation syndrome 6, 616809
PIK3CA	155.4	100%	94%	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	159.5	99%	97%	Immunodeficiency 14, 615513
PIK3R1	158.1	99%	88%	Immunodeficiency 36, 616005 SHORT syndrome, 269880 ?Agammaglobulinemia 7, autosomal recessive, 615214
PIK3R2	104.3	90%	98%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PIK3R5	117.8	100%	95%	Ataxia-oculomotor apraxia 3, 615217
PIKFYVE	177.4	99%	33%	Corneal fleck dystrophy, 121850
PINK1	98	93%	88%	Parkinson disease 6, early onset, 605909
PIP5K1C	131.7	96%	99%	Lethal congenital contractural syndrome 3, 611369
PITPNM3	128	99%	99%	Cone-rod dystrophy 5, 600977
PITX1	149	93%	99%	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800 Liebenberg syndrome, 186550
PITX2	146.1	99%	91%	Axenfeld-Rieger syndrome, type 1, 180500 Iridogoniodysgenesis, type 2, 137600 Peters anomaly, 604229 Ring dermoid of cornea, 180550
PITX3	64.5	99%	99%	Anterior segment mesenchymal dysgenesis, 107250 Cataract 11, multiple types, 610623 Cataract 11, syndromic, 610623
PKD1	30.1	41%	97%	Polycystic kidney disease, adult type I, 173900
PKD2	129.6	92%	99%	Polycystic kidney disease 2, 613095
PKHD1	176.8	100%	100%	Polycystic kidney and hepatic disease, 263200
PKLR	195.6	100%	99%	Adenosine triphosphate, elevated, of erythrocytes, 102900 Pyruvate kinase deficiency, 266200
PKP1	140	100%	99%	Ectodermal dysplasia/skin fragility syndrome, 604536

PKP2	112.7	96%	97%	Arrhythmogenic right ventricular dysplasia 9, 609040
PLA2G4A	170.7	100%	98%	Phospholipase A2, group IV A, deficiency of
PLA2G6	135.5	99%	98%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953
PLA2G7	158.1	100%	99%	Platelet-activating factor acetylhydrolase deficiency, 614278 {Asthma, susceptibility to}, 600807 {Atopy, susceptibility to}, 147050
PLAG1	237.3	100%	99%	Adenomas, salivary gland pleomorphic, somatic, 181030
PLAU	123.7	99%	99%	Quebec platelet disorder, 601709 {Alzheimer disease, late-onset, susceptibility to}, 104300
PLCB1	173.5	100%	92%	Epileptic encephalopathy, early infantile, 12, 613722
PLCB4	139.4	99%	86%	Auriculocondylar syndrome 2, 614669
PLCD1	122.7	99%	87%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCE1	166.9	99%	90%	Nephrotic syndrome, type 3, 610725
PLCG2	130.6	100%	97%	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468
PLEC	122.1	99%	100%	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	153.2	100%	96%	Leukodystrophy and acquired microcephaly with or without dystonia, 616763
PLEKHG5	99.5	98%	92%	Charcot-Marie-Tooth disease, recessive intermediate C, 615376 Spinal muscular atrophy, distal, autosomal recessive, 4, 611067
PLEKHM1	68	90%	99%	Osteopetrosis, autosomal recessive 6, 611497
PLG	138.6	87%	98%	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PLIN1	89.6	98%	99%	Lipodystrophy, familial partial, type 4, 613877
PLK4	172.8	99%	99%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLN	252.7	100%	99%	Cardiomyopathy, dilated, 1P, 609909 Cardiomyopathy, hypertrophic, 18, 613874
PLOD1	155.4	99%	92%	Ehlers-Danlos syndrome, type VI, 225400
PLOD2	143.1	97%	95%	Bruck syndrome 2, 609220

PLOD3	117.6	99%	81%	Lysyl hydroxylase 3 deficiency, 612394
PLP1	155	99%	99%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PLS3	156.3	99%	99%	Bone mineral density QTL18, osteoporosis, 300910
PML	142.6	100%	97%	Leukemia, acute promyelocytic, PML/RARA type
PMM2	171.3	100%	99%	Congenital disorder of glycosylation, type Ia, 212065
PMP22	119.5	96%	99%	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	135.9	98%	96%	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMS2	109.8	83%	98%	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 Mismatch repair cancer syndrome, 276300
PMVK	128.4	100%	99%	Porokeratosis 1, multiple types, 175800
PNKD	126.7	100%	87%	Paroxysmal nonkinesigenic dyskinesia,118800
PNKP	100.7	99%	100%	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNP	147.3	100%	93%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA1	192.9	99%	85%	Ichthyosis, congenital, autosomal recessive 10, 615024
PNPLA2	139.3	99%	99%	Neutral lipid storage disease with myopathy, 610717
PNPLA6	150.4	99%	96%	Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 Spastic paraplegia 39, autosomal recessive, 612020 ?Laurence-Moon syndrome, 245800
PNPO	80.9	100%	98%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
PNPT1	67.9	95%	95%	Combined oxidative phosphorylation deficiency 13, 614932 Deafness, autosomal recessive 70, 614934
POC1A	144.7	100%	92%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POC1B	99.5	97%	99%	Cone-rod dystrophy 20, 615973
POF1B	93.7	93%	99%	Premature ovarian failure 2B,300604
POFUT1	162.7	100%	97%	Dowling-Degos disease 2, 615327
POGLUT1	150.1	99%	99%	Dowling-Degos disease 4, 615696

POGZ	172.9	99%	98%	White-Sutton syndrome, 616364
POLA1	133.4	99%	95%	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220
POLD1	117.3	95%	100%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381 {Colorectal cancer, susceptibility to, 10}, 612591
POLE	158.9	99%	99%	FILS syndrome, 615139 {Colorectal cancer, susceptibility to, 12}, 615083
POLG	128.1	100%	99%	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	189.4	99%	99%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131
POLH	170.4	100%	97%	Xeroderma pigmentosum, variant type, 278750
POLR1A	137.5	99%	100%	Acrofacial dysostosis, Cincinnati type, 616462
POLR1C	117.5	99%	100%	Leukodystrophy, hypomyelinating, 11, 616494 Treacher Collins syndrome 3, 248390
POLR1D	193.8	100%	91%	Treacher Collins syndrome 2, 613717
POLR3A	153	100%	98%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	163.8	100%	96%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMC	105.7	100%	99%	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734 {Obesity, early-onset, susceptibility to}, 601665
POMGNT1	136.7	99%	99%	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	258.7	100%	97%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830
POMK	223.3	100%	99%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249 ?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094
POMP	167	98%	100%	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952
POMT1	184.3	99%	99%	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	117.5	99%	100%	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	182	100%	99%	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571
PORCN	142.4	100%	97%	Focal dermal hypoplasia, 305600
POU1F1	141.9	99%	97%	Pituitary hormone deficiency, combined, 1, 613038
POU3F4	159.7	100%	99%	Deafness, X-linked 2, 304400
POU4F3	295.2	100%	93%	Deafness, autosomal dominant 15, 602459
PPARG	171.7	100%	99%	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	148.1	100%	98%	Osteogenesis imperfecta, type IX, 259440
PPM1D	183.5	100%	99%	Breast cancer, 114480
PPOX	104.3	99%	100%	Porphyria variegata, 176200
PPP1R15B	156	99%	96%	Microcephaly, short stature, and impaired glucose metabolism 2, 616817
PPP1R3A	195.2	99%	96%	Insulin resistance, severe, digenic, 604367
PPP2R1A	158.2	93%	100%	Mental retardation, autosomal dominant 36, 616362
PPP2R1B	179.1	100%	85%	Lung cancer, 211980
PPP2R2B	172.3	99%	99%	Spinocerebellar ataxia 12, 604326
PPP2R5D	166.9	100%	98%	Mental retardation, autosomal dominant 35, 616355
PPT1	203.5	100%	81%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	180.7	97%	99%	Renpenning syndrome, 309500
PRCC	150.5	99%	90%	Renal cell carcinoma, papillary, 605074
PRCD	103.1	100%	100%	Retinitis pigmentosa 36, 610599
PRDM12	112.2	89%	94%	Neuropathy, hereditary sensory and autonomic, type VIII, 616488
PRDM16	180	99%	95%	Cardiomyopathy, dilated, 1LL, 615373 Left ventricular noncompaction 8, 615373
PRDM5	163.3	99%	92%	Brittle cornea syndrome 2, 614170
PRDM6	107.1	95%	100%	Patent ductus arteriosus 3, 617039
PRF1	128.8	100%	100%	Aplastic anemia, 609135 Hemophagocytic lymphohistiocytosis, familial, 2, 603553

				Lymphoma, non-Hodgkin, 605027
PRG4	162.1	98%	95%	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250
PRICKLE1	138.6	100%	93%	Epilepsy, progressive myoclonic 1B, 612437
PRIMPOL	132	97%	96%	Myopia 22,autosomal dominant,615420
PRKAG2	146.1	99%	96%	Cardiomyopathy, hypertrophic 6, 600858 Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200
PRKAR1A	104.2	97%	99%	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	183.2	100%	100%	Pituitary tumor, invasive
PRKCD	204.8	100%	98%	Autoimmune lymphoproliferative syndrome, type III, 615559
PRKCG	137.2	99%	97%	Spinocerebellar ataxia 14, 605361
PRKCSH	148.9	99%	83%	Polycystic liver disease, 174050
PRKDC	120.2	99%	92%	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PRKG1	133.1	99%	100%	Aortic aneurysm, familial thoracic 8, 615436
PRKRA	179.8	99%	93%	Dystonia 16, 612067
PRLR	170.1	100%	86%	Multiple fibroadenomas of the breast, 615554 ?Hyperprolactinemia, 615555
PRNP	147.8	100%	93%	Cerebral amyloid angiopathy, PRNP-related, 137440 Creutzfeldt-Jakob disease, 123400 Gerstmann-Straussler disease, 137440 Huntington disease-like 1, 603218 Insomnia, fatal familial, 600072 Prion disease with protracted course, 606688 {Kuru, susceptibility to}, 245300
PROC	142.6	99%	95%	Thrombophilia due to protein C deficiency, autosomal dominant, 176860 Thrombophilia due to protein C deficiency, autosomal recessive, 612304
PRODH	96.4	89%	93%	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850
PROK2	116.6	99%	99%	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	387.8	100%	99%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROM1	141	96%	99%	Cone-rod dystrophy 12, 612657

				Macular dystrophy, retinal, 2, 608051 Retinitis pigmentosa 41, 612095 Stargardt disease 4, 603786
PROP1	92.2	94%	100%	Pituitary hormone deficiency, combined, 2, 262600
PROS1	104.9	97%	100%	Thrombophilia due to protein S deficiency, autosomal dominant, 612336 Thrombophilia due to protein S deficiency, autosomal recessive, 614514
PRPF3	92	98%	99%	Retinitis pigmentosa 18, 601414
PRPF31	130.5	98%	99%	Retinitis pigmentosa 11, 600138
PRPF4	160.8	99%	100%	Retinitis pigmentosa 70, 615922
PRPF6	130.5	100%	99%	Retinitis pigmentosa 60, 613983
PRPF8	143.2	99%	91%	Retinitis pigmentosa 13, 600059
PRPH2	249.9	100%	99%	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	178.9	100%	99%	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	91.4	100%	77%	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751
PRRX1	119.4	99%	97%	Agnathia-otocephaly complex, 202650
PRSS1	210.5	100%	99%	Pancreatitis, hereditary, 167800 Trypsinogen deficiency, 614044
PRSS12	178.3	99%	100%	Mental retardation, autosomal recessive 1, 249500
PRSS56	59.9	98%	25%	Microphthalmia, isolated 6, 613517
PRX	127.5	99%	99%	Charcot-Marie-Tooth disease, type 4F, 614895 Dejerine-Sottas disease, 145900
PSAP	129.6	99%	98%	Combined SAP deficiency, 611721 Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722

				Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PSAT1	53.4	91%	97%	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992
PSEN1	156.6	99%	96%	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	142.9	100%	97%	Alzheimer disease-4, 606889 Cardiomyopathy, dilated, 1V, 613697
PSENEN	102.7	100%	100%	Acne inversa, familial, 2, 613736
PSMB8	16	61%	99%	Autoinflammation, lipodystrophy, and dermatosis syndrome, 256040
PSMC3IP	123.1	100%	78%	Ovarian dysgenesis 3, 614324
PSPH	164.6	99%	95%	Phosphoserine phosphatase deficiency, 614023
PSTPIP1	104.7	99%	98%	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416
PTCH1	127.7	98%	98%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly-7, 610828
PTCH2	131.6	99%	98%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Medulloblastoma, 155255
PTDSS1	162.9	100%	93%	Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	169.6	100%	96%	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	81.5	89%	97%	Pancreatic agenesis 2, 615935 Pancreatic and cerebellar agenesis, 609069
PTGIS	149.3	98%	89%	Hypertension, essential, 145500
PTH	137.2	100%	96%	Hypoparathyroidism, autosomal dominant, 146200 Hypoparathyroidism, autosomal recessive, 146200
PTH1R	118.4	99%	99%	Chondrodysplasia, Blomstrand type, 215045 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Metaphyseal chondrodysplasia, Murk Jansen type, 156400
PTHLH	153.3	99%	91%	Brachydactyly, type E2, 613382 Humoral hypercalcemia of malignancy
PTPN11	105.7	98%	99%	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
PTPN12	181.4	98%	100%	Colon cancer, somatic, 114500
PTPN14	199.8	99%	96%	Choanal atresia and lymphedema, 613611
PTPRC	127.6	96%	98%	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971 {Hepatitic C virus, susceptibility to}, 609532
PTPRJ	189.1	97%	93%	Colon cancer, somatic, 114500
PTPRO	175	100%	97%	Nephrotic syndrome, type 6, 614196
PTPRQ	125.9	93%	99%	Deafness, autosomal recessive 84A, 613391
PTRF	162.4	99%	99%	Lipodystrophy, congenital generalized, type 4, 613327
PTRH2	305.6	100%	99%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PTS	133.3	99%	95%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUF60	197.3	99%	96%	Verheij syndrome, 615583
PURA	135.6	98%	100%	Mental retardation, autosomal dominant 31, 616158
PUS1	123.1	99%	100%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PVRL1	166.5	100%	99%	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060
PVRL4	157.6	100%	99%	Ectodermal dysplasia-syndactyly syndrome 1, 613573
PXDN	172.6	99%	91%	Corneal opacification and other ocular anomalies, 269400
PYCR1	103.5	99%	100%	Cutis laxa, autosomal recessive, type IIB, 612940

				Cutis laxa, autosomal recessive, type IIIB, 614438
PYCR2	142.2	99%	100%	Leukodystrophy, hypomyelinating, 10, 616420
PYGL	177.1	100%	93%	Glycogen storage disease VI, 232700
PYGM	156.7	100%	100%	McArdle disease, 232600
QARS	163.6	100%	99%	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	91.4	100%	99%	Hyperphenylalaninemia, BH4-deficient, C, 261630
RAB18	108.6	99%	96%	Warburg micro syndrome 3, 614222
RAB23	141.3	100%	100%	Carpenter syndrome, 201000
RAB27A	171.6	100%	99%	Griscelli syndrome, type 2, 607624
RAB28	68.8	98%	97%	Cone-rod dystrophy 18, 615374
RAB33B	282.1	100%	89%	Smith-McCort dysplasia 2, 615222
RAB39B	125.7	100%	89%	Mental retardation, X-linked 72, 300271 ?Waisman syndrome, 311510
RAB3GAP1	155.9	99%	99%	Warburg micro syndrome 1, 600118
RAB3GAP2	116.8	99%	96%	Martsolf syndrome, 212720 Warburg micro syndrome 2, 614225
RAB7A	152.4	100%	98%	Charcot-Marie-Tooth disease,type 2B, 600882
RAC2	135.4	100%	99%	Neutrophil immunodeficiency syndrome, 608203
RAD21	109	99%	100%	Cornelia de Lange syndrome 4, 614701
RAD50	119.9	94%	100%	Nijmegen breakage syndrome-like disorder, 613078
RAD51	141.7	89%	99%	Mirror movements 2, 614508 {Breast cancer, susceptibility to}, 114480 ?Fanconi anemia, complementation group R, 617244
RAD51C	165.7	100%	96%	Fanconi anemia, complementation group O, 613390 {Breast-ovarian cancer, familial, susceptibility to, 3}, 613399
RAD54B	131.9	99%	95%	Colon cancer, somatic, 114500 Lymphoma, non-Hodgkin, somatic, 605027
RAD54L	130.6	99%	100%	Adenocarcinoma, colonic, somatic Lymphoma, non-Hodgkin, somatic, 605027 {Breast cancer, invasive ductal}, 114480
RAF1	144.9	100%	95%	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553
RAG1	208.9	100%	99%	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	271.4	100%	92%	Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457
RAI1	159.2	100%	78%	Smith-Magenis syndrome, 182290
RAP1GDS1	125.6	99%	91%	Lymphocytic leukemia, acute T-cell
RAPSN	151.1	99%	83%	Fetal akinesia deformation sequence, 208150 Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326
RARB	147.3	100%	94%	Microphthalmia, syndromic 12, 615524
RARS	114.4	99%	98%	Leukodystrophy, hypomyelinating, 9, 616140
RARS2	137.6	100%	95%	Pontocerebellar hypoplasia, type 6, 611523
RASA1	129	97%	98%	Basal cell carcinoma, somatic, 605462 Capillary malformation-arteriovenous malformation, 608354 Parkes Weber syndrome, 608355
RAX	94.3	91%	97%	Microphthalmia, isolated 3, 611038
RAX2	73.8	98%	97%	?Macular degeneration, age-related, 6, 613757 Cone-rod dystrophy 11, 610381
RB1	97.9	94%	95%	Bladder cancer, somatic, 109800 Osteosarcoma, somatic, 259500 Retinoblastoma, 180200 Retinoblastoma, trilateral, 180200 Small cell cancer of the lung, somatic, 182280
RB1CC1	130.4	98%	88%	Breast cancer, somatic, 114480
RBBP8	121.9	99%	100%	Jawad syndrome, 251255 Pancreatic carcinoma, somatic Seckel syndrome 2, 606744
RBCK1	112	98%	94%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RBM10	130.8	99%	99%	TARP syndrome, 311900
RBM20	194.8	99%	69%	Cardiomyopathy, dilated, 1DD, 613172
RBM8A	109.4	99%	98%	Thrombocytopenia-absent radius syndrome, 274000
RBP4	124.5	99%	95%	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147

RBPJ	94.2	95%	99%	Adams-Oliver syndrome 3, 614814
RD3	182.9	100%	99%	Leber congenital amaurosis 12, 610612
RDH12	102.7	99%	93%	Leber congenital amaurosis 13, 612712
RDH5	174.3	100%	98%	Fundus albipunctatus, 136880
RDX	49.9	87%	98%	Deafness, autosomal recessive 24, 611022
RECQL4	152.3	99%	96%	Baller-Gerold syndrome, 218600 RAPADILINO syndrome, 266280 Rothmund-Thomson syndrome, 268400
REEP1	113.6	97%	99%	Spastic paraplegia 31, autosomal dominant, 610250 ?Neuronopathy, distal hereditary motor, type VB, 614751
RELN	184.8	100%	99%	Lissencephaly 2 (Norman-Roberts type), 257320 {Epilepsy, familial temporal lobe, 7}, 616436
REN	163.2	100%	94%	Hyperuricemic nephropathy, familial juvenile 2, 613092 Renal tubular dysgenesis, 267430 [Hyperproreninemia]
RERE	86	96%	99%	Neurodevelopmental disorder with or without anomalies of the brain,eye or heart, 616975
RET	169	99%	99%	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	114.8	99%	99%	Congenital disorder of glycosylation, type In, 612015
RFX5	131.9	98%	99%	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFX6	192.6	100%	98%	Mitchell-Riley syndrome, 615710
RFXANK	121.5	100%	96%	MHC class II deficiency, complementation group B, 209920
RFXAP	124.3	96%	100%	Bare lymphocyte syndrome, type II, complementation group D, 209920
RGR	145.7	100%	95%	Retinitis pigmentosa 44, 613769
RGS9	117.1	100%	99%	Bradyopsia, 608415
RGS9BP	89.6	99%	99%	Bradyopsia, 608415
RHAG	183	100%	100%	Anemia, hemolytic, Rh-null, regulator type, 268150 Overhydrated hereditary stomatocytosis, 185000 Rh-mod syndrome

RHBDF2	107.7	99%	99%	Tylosis with esophageal cancer, 148500
RHCE	210.8	97%	98%	Rh-null disease, amorph type [Blood group, Rhesus], 111690
RHO	233	100%	98%	Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 Retinitis punctata albescens, 136880
RIMS1	140.2	98%	94%	Cone-rod dystrophy 7, 603649
RIN2	125	100%	99%	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075
RIPK4	163.1	100%	92%	Popliteal pterygium syndrome, Bartsocas-Papas type, 263650
RIT1	190.3	100%	98%	Noonan syndrome 8, 615355
RLBP1	143.9	100%	99%	Bothnia retinal dystrophy, 607475 Fundus albipunctatus, 136880 Newfoundland rod-cone dystrophy, 607476 Retinitis punctata albescens, 136880
RLIM	142.8	100%	86%	Mental Retardation, X-linked 61, 300978
RMND1	168.4	99%	99%	Combined oxidative phosphorylation deficiency 11, 614922
RMRP	999999		88%	Anauxetic dysplasia, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RNASEH1	113.9	98%	100%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479
RNASEH2A	157.1	100%	99%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	128.3	98%	95%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	244.3	99%	97%	Aicardi-Goutieres syndrome 3, 610329
RNASEL	170.8	100%	98%	Prostate cancer 1, 601518
RNASET2	99.3	93%	99%	Leukoencephalopathy, cystic, without megalencephaly, 612951
RNF125	181.7	100%	97%	Tenorio syndrome, 616260
RNF135	93.8	92%	96%	Macrocephaly, macrosomia, facial dysmorphism syndrome, 614192
RNF139	246.4	100%	95%	Renal cell carcinoma, 144700
RNF168	293.7	100%	99%	RIDDLE syndrome, 611943
RNF170	143.3	99%	99%	Ataxia, sensory, 1, autosomal dominant, 608984
RNF212	121.6	99%	100%	Recombination rate QTL 1, 612042
RNF216	158.3	99%	99%	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840
RNF6	216.7	100%	99%	Esophageal carcinoma, somatic, 133239

RNU4ATAC	999999		99%	Microcephalic osteodysplastic primordial dwarfism, type I, 210710 Roifman syndrome, 616651
ROBO2	167.1	99%	99%	Vesicoureteral reflux 2, 610878
ROBO3	107	99%	78%	Gaze palsy, horizontal, with progressive scoliosis, 607313
ROGDI	127.1	97%	99%	Kohlschutter-Tonz syndrome, 226750
ROM1	128.9	100%	95%	Retinitis pigmentosa 7, digenic, 608133
ROR2	190.4	99%	99%	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RORC	156.7	100%	64%	Immunodeficiency 42, 616622
RP1	145.9	100%	88%	Retinitis pigmentosa 1, 180100
RP1L1	109.5	100%	71%	Occult macular dystrophy, 613587
RP2	189.5	100%	91%	Retinitis pigmentosa 2, 312600
RPE65	165.8	100%	96%	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794
RPGR	115.9	85%	70%	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	158.2	100%	94%	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826
RPGRIP1L	160.8	96%	91%	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561
RPL11	111.8	100%	78%	Diamond-Blackfan anemia 7, 612562
RPL21	85.4	80%	91%	Hypotrichosis 12, 615885
RPL35A	90.3	97%	94%	Diamond-Blackfan anemia 5, 612528
RPL5	48.5	86%	89%	Diamond-Blackfan anemia 6, 612561
RPS10	131.7	97%	68%	Diamond-Blackfan anemia 9, 613308
RPS14	131.2	99%	99%	Macrocytic anemia, refractory, due to 5q deletion, somatic, 153550
RPS17	51.1	83%	77%	Diamond-Blackfan anemia 4, 612527
RPS19	97.5	99%	98%	Diamond-Blackfan anemia 1, 105650
RPS24	131.5	97%	90%	Diamond-blackfan anemia 3, 610629
RPS26	95.3	91%	100%	Diamond-Blackfan anemia 10, 613309
RPS28	43.5	99%	98%	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164

RPS29	62.8	98%	96%	Diamond-Blackfan anemia 13, 615909
RPS6KA3	97.9	96%	98%	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844
RPS7	118.7	84%	100%	Diamond-Blackfan anemia 8, 612563
RPSA	89	100%	99%	Asplenia, isolated congenital, 271400
RRAS2	90.1	89%	100%	Ovarian carcinoma
RRM2B	163	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	65.5	99%	96%	Retinoschisis, 312700
RSPH1	172.2	100%	97%	Ciliary dyskinesia, primary, 24, 615481
RSPH3	145.1	99%	96%	Ciliary dyskinesia, primary, 32, 616481
RSPH4A	173	98%	92%	Ciliary dyskinesia, primary, 11, 612649
RSPH9	156.2	99%	74%	Ciliary dyskinesia, primary, 12, 612650
RSPO1	128.8	100%	95%	Palmoplantar hyperkeratosis and true hermaphroditism, 610644 Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644
RSPO4	130.2	100%	99%	Anonychia congenita, 206800
RSPRY1	186.1	100%	95%	Spondylopeimetaphyseal dsyplasia, Faden-Alkuraya type, 616723
RTEL1	137.2	99%	99%	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373
RTN2	121.4	98%	100%	Spastic paraplegia 12, autosomal dominant, 604805
RTN4IP1	109.9	99%	98%	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
RTTN	144.1	98%	96%	Microcephaly,short stature,and polymicrogyria with seizures,614833
RUNX1	111.4	97%	99%	Leukemia, acute myeloid, 601626 Platelet disorder, familial, with associated myeloid malignancy, 601399
RUNX2	111.8	74%	100%	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	136.1	98%	98%	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	167.3	99%	88%	Arrhythmogenic right ventricular dysplasia 2, 600996 Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772
S1PR2	244.8	98%	95%	Deafness, autosomal recessive 68, 610419
SACS	187.5	100%	94%	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAG	159.1	100%	99%	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758
SALL1	144	99%	96%	Townes-Brocks branchiootorenal-like syndrome, 107480 Townes-Brocks syndrome, 107480
SALL4	161.4	98%	98%	Duane-radial ray syndrome, 607323 IVIC syndrome, 147750
SAMD9	226.4	100%	99%	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455
SAMD9L	218.7	100%	99%	Ataxia-pancytopenia syndrome, 159550
SAMHD1	154.9	99%	84%	Aicardi-Goutieres syndrome 5, 612952 ?Chilblain lupus 2, 614415
SAR1B	172.4	92%	99%	Chylomicron retention disease, 246700
SARS2	120.4	96%	98%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SATB2	122.4	99%	99%	Glass syndrome, 612313
SBDS	231.2	100%	96%	Shwachman-Diamond syndrome, 260400 {Aplastic anemia, susceptibility to}, 609135
SBF1	114.9	99%	98%	Charcot-Marie-Tooth disease, type 4B3, 615284
SBF2	132.7	99%	100%	Charcot-Marie-Tooth disease, type 4B2, 604563
SC5D	237.6	99%	100%	Lathosterolosis, 607330
SCARB2	141.5	100%	99%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCARF2	88.8	94%	93%	Van den Ende-Gupta syndrome, 600920
SCN10A	187	99%	99%	Episodic pain syndrome, familial, 2, 615551
SCN11A	165	99%	99%	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN1A	170.2	99%	97%	Dravet syndrome, 607208 Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Febrile seizures, familial, 3A, 604403 Migraine, familial hemiplegic, 3, 609634

SCN1B	189.8	97%	95%	Atrial fibrillation, familial, 13, 615377 Brugada syndrome 5, 612838 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233
SCN2A	186.5	99%	99%	Epileptic encephalopathy, early infantile, 11, 613721 Seizures, benign familial infantile, 3, 607745
SCN2B	211.9	100%	97%	Atrial fibrillation, familial, 14, 615378
SCN3B	163.5	100%	94%	Atrial fibrillation, familial, 16, 613120 Brugada syndrome 7, 613120
SCN4A	229.7	100%	100%	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	81.8	99%	98%	Atrial fibrillation, familial, 17, 611819 Long QT syndrome-10, 611819
SCN5A	181.2	100%	97%	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	209.5	99%	98%	Epileptic encephalopathy, early infantile, 13, 614558 ?Cognitive impairment with or without cerebellar ataxia, 614306
SCN9A	180.4	98%	79%	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	147.9	97%	97%	Bronchiectasis with or without elevated sweat chloride 2, 613021 Pseudohypoaldosteronism, type I, 264350
SCNN1B	172.3	100%	93%	Bronchiectasis with or without elevated sweat chloride 1, 211400 Liddle syndrome, 177200 Pseudohypoaldosteronism, type I, 264350
SCNN1G	160.7	99%	99%	Bronchiectasis with or without elevated sweat chloride 3, 613071 Liddle syndrome, 177200 Pseudohypoaldosteronism, type I, 264350
SCO1	122.6	98%	96%	Mitochondrial complex IV deficiency, 220110
SCO2	126.5	100%	58%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908
SCP2	138	100%	96%	Leukoencephalopathy with dystonia and motor neuropathy, 613724
SCYL1	176.6	99%	97%	Spinocerebellar ataxia, autosomal recessive 21, 616719
SDCCAG8	155	99%	98%	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SDHA	123.2	84%	100%	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165
SDHAF1	50.5	99%	80%	Mitochondrial complex II deficiency, 252011
SDHAF2	151.4	94%	97%	Paragangliomas 2, 601650
SDHB	146.3	100%	99%	Cowden syndrome 2, 612359 Gastrointestinal stromal tumor, 606764 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 4, 115310 Pheochromocytoma, 171300
SDHC	117.8	99%	83%	Gastrointestinal stromal tumor, 606764 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 3, 605373
SDHD	59.2	63%	100%	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	149.6	98%	95%	Craniolenticulosutural dysplasia, 607812
SEC23B	169	97%	98%	Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SEC24D	159.3	99%	96%	Cole-Carpenter syndrome 2, 616294
SEC61A1	165.8	100%	100%	Hyperuricemic nephropathy, familial juvenile, 4,617056
SEC63	83	88%	100%	Polycystic liver disease, 174050
SECISBP2	132.9	99%	100%	Thyroid hormone metabolism, abnormal, 609698
SEMA4A	153.4	99%	99%	Cone-rod dystrophy 10, 610283 Retinitis pigmentosa 35, 610282
SEPN1	133.1	86%	100%	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310
SEPSECS	198.4	100%	100%	Pontocerebellar hypoplasia type 2D, 613811
SEPT12	109.2	98%	99%	Spermatogenic failure 10, 614822
SEPT9	137.1	99%	99%	Amyotrophy, hereditary neuralgic, 162100 Leukemia, acute myeloid, therapy-related Ovarian carcinoma
SERAC1	139.3	99%	99%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPINA1	136.3	100%	94%	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	145.8	100%	97%	Alpha-1-antichymotrypsin deficiency Cerebrovascular disease, occlusive
SERPINA6	196.4	100%	96%	Corticosteroid-binding globulin deficiency,611489
SERPINB7	154.7	100%	99%	Palmoplantar keratoderma, Nagashima type, 615598
SERPINC1	152.9	100%	99%	Thrombophilia due to antithrombin III deficiency, 613118
SERPIND1	157.3	100%	99%	Thrombophilia due to heparin cofactor II deficiency,612356
SERPINE1	171	100%	98%	Plasminogen activator inhibitor-1 deficiency, 613329 {Transcription of plasminogen activator inhibitor, modulator of}
SERPINF1	117.3	100%	93%	Osteogenesis imperfecta, type VI, 613982
SERPINF2	166.3	100%	99%	Alpha-2-plasmin inhibitor deficiency,262850
SERPING1	98.9	98%	100%	Angioedema, hereditary, types I and II, 106100 Complement component 4, partial deficiency of, 120790

SERPINI1	127.6	99%	99%	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218
SETBP1	142.5	97%	95%	Mental retardation, autosomal dominant 29, 616078 Schinzel-Giedion midface retraction syndrome, 269150
SETD2	182.8	100%	99%	Luscan-Lumish syndrome, 616831
SETD5	200.5	100%	99%	Mental retardation, autosomal dominant 23, 615761
SETX	197.6	100%	96%	Amyotrophic lateral sclerosis 4, juvenile, 602433 Spinocerebellar ataxia, autosomal recessive 1, 606002
SF3B1	162.2	99%	98%	Myelodysplastic syndrome, somatic, 614286
SF3B4	85.7	99%	92%	Acrofacial dysostosis 1, Nager type, 154400
SFRP4	167.1	100%	99%	Pyle disease, 265900
SFTPA2	182.9	100%	96%	Pulmonary fibrosis, idiopathic, 178500
SFTPB	114.2	100%	94%	Surfactant metabolism dysfunction, pulmonary, 1, 265120
SFTPC	108	99%	84%	Surfactant metabolism dysfunction, pulmonary, 2, 610913
SFXN4	145.1	99%	89%	Combined oxidative phosphorylation deficiency 18, 615578
SGCA	173.2	100%	91%	Muscular dystrophy, limb-girdle, type 2D, 608099
SGCB	192.5	97%	99%	Muscular dystrophy, limb-girdle, type 2E, 604286
SGCD	109.9	99%	99%	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, type 2F, 601287
SGCE	110.4	95%	76%	Dystonia-11, myoclonic, 159900
SGCG	141.7	100%	95%	Muscular dystrophy, limb-girdle, type 2C, 253700
SGOL1	130.1	99%	99%	Chronic atrial and intestinal dysrhythmia, 616201
SGSH	140.9	96%	59%	Mucopolysaccharidisis type IIIA (Sanfilippo A), 252900
SH2B3	113.1	95%	94%	Erythrocytosis, somatic, 133100 Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
SH2D1A	127.3	92%	96%	Lymphoproliferative syndrome, X-linked, 1, 308240
SH3BP2	126.1	91%	97%	Cherubism, 118400
SH3PXD2B	158.6	100%	99%	Frank-ter Haar syndrome, 249420
SH3TC2	122.7	99%	96%	Charcot-Marie-Tooth disease, type 4C, 601596 Mononeuropathy of the median nerve, mild, 613353
SHANK3	90.5	86%	98%	Phelan-McDermid syndrome, 606232 {Schizophrenia 15}, 613950
SHH	114.4	99%	85%	Holoprosencephaly-3, 142945 Microphthalmia with coloboma 5, 611638

				Schizencephaly, 269160 Single median maxillary central incisor, 147250
SHOC2	150.5	100%	99%	Noonan-like syndrome with loose anagen hair, 607721
SHOX	28.6	78%	96%	Langer mesomelic dysplasia, 249700 Leri-Weill dyschondrosteosis, 127300 Short stature, idiopathic familial, 300582
SI	125.9	98%	57%	Sucrase-isomaltase deficiency, congenital, 222900
SIK1	111.9	98%	99%	Epileptic encephalopathy, early infantile, 30, 616341
SIL1	175.7	99%	99%	Marinesco-Sjogren syndrome, 248800
SIM1	160.7	100%	99%	Obesity, severe, 601665
SIX1	125.4	99%	100%	Brachiootic syndrome 3, 608389 Deafness, autosomal dominant 23, 605192
SIX3	172.7	99%	97%	Holoprosencephaly-2, 157170 Schizencephaly, 269160
SIX5	57.7	93%	100%	Branchiootorenal syndrome 2, 610896
SIX6	251.7	100%	99%	Optic disc anomalies with retinal and/or macular dystrophy, 212550
SKI	96.2	98%	98%	Shprintzen-Goldberg syndrome, 182212
SKIV2L	27	81%	100%	Trichohepatoenteric syndrome 2, 614602
SLC10A2	167.5	100%	87%	Bile acid malabsorption, primary, 613291
SLC11A2	131.7	100%	95%	Anemia, hypochromic microcytic, with iron overload 1, 206100
SLC12A1	185.9	100%	100%	Bartter syndrome, type 1, 601678
SLC12A3	164.5	100%	95%	Gitelman syndrome, 263800
SLC12A5	177.8	99%	98%	Epileptic encephalopathy, early infantile, 34, 616645 {Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685
SLC12A6	162.9	100%	100%	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC13A5	167.9	100%	99%	Epileptic encephalopathy, early infantile, 25, 615905
SLC16A1	180.7	99%	100%	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia, familial, 7, 610021 Monocarboxylate transporter 1 deficiency, 616095
SLC16A12	179.4	100%	98%	Cataract, juvenile, with microcornea and glucosuria, 612018
SLC16A2	70.9	96%	97%	Allan-Herndon-Dudley syndrome, 300523
SLC17A5	142.3	99%	99%	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC17A8	161	100%	88%	Deafness, autosomal dominant 25, 605583

SLC17A9	134.6	95%	100%	Porokeratosis 8, disseminated superficial actinic type, 616063
SLC19A2	128.3	99%	100%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	185.8	100%	98%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A1	215.5	100%	97%	Dicarboxylic aminoaciduria, 222730 {?Schizophrenia susceptibility 18}, 615232
SLC1A3	153.2	100%	92%	Episodic ataxia, type 6, 612656
SLC1A4	190.5	99%	99%	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC20A2	126.4	99%	96%	Basal ganglia calcification, idiopathic, 1, 213600
SLC22A12	127.3	100%	96%	Hypouricemia, renal, 220150
SLC22A18	108.4	96%	96%	Breast cancer, somatic, 114480 Lung cancer, somatic, 211980 Rhabdomyosarcoma, somatic, 268210
SLC22A5	171.6	100%	99%	Carnitine deficiency, systemic primary, 212140
SLC24A1	222.7	100%	97%	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
SLC24A4	133.3	99%	97%	Amelogenesis imperfecta, type IIA5, 615887 [Skin/hair/eye pigmentation 6, blond/brown hair], 210750 [Skin/hair/eye pigmentation 6, blue/green eyes], 210750
SLC24A5	130.5	99%	97%	Albinism, oculocutaneous, type VI, 113750 [Skin/hair/eye pigmentation 4, fair/dark skin], 113750
SLC25A1	90.7	98%	97%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A12	183.5	99%	100%	Epileptic encephalopathy, early infantile, 39, 612949
SLC25A13	138.2	99%	90%	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A15	238.4	98%	100%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A19	79.7	99%	99%	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A20	109.7	100%	99%	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A22	120.5	99%	99%	Epileptic encephalopathy, early infantile, 3, 609304
SLC25A26	114.1	99%	99%	Combined oxidative phosphorylation deficiency 28, 616794
SLC25A3	160.6	99%	99%	Mitochondrial phosphate carrier deficiency, 610773
SLC25A38	124.7	99%	99%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC25A4	142.3	100%	99%	Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184 Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418

				Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283
SLC25A46	206.3	97%	99%	Neuropathy, hereditary motor and sensory, type VIB, 616505
SLC26A2	289.4	100%	98%	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	198.1	100%	99%	Diarrhea 1, secretory chloride, congenital, 214700
SLC26A4	160.6	100%	99%	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791 Pendred syndrome, 274600
SLC26A8	150.4	100%	94%	Spermatogenic failure 3, 606766
SLC27A4	184.8	99%	99%	Ichthyosis prematurity syndrome, 608649
SLC29A3	236.3	99%	99%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC2A1	191.5	100%	96%	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	187.1	100%	99%	Arterial tortuosity syndrome, 208050
SLC2A2	205.9	100%	97%	Fanconi-Bickel syndrome, 227810 {Diabetes mellitus, noninsulin-dependent}, 125853
SLC2A9	145.8	99%	98%	Hypouricemia, renal, 2, 612076 {Uric acid concentration, serum, QTL 2}, 612076
SLC30A10	194.2	100%	93%	Hypermanganesemia with dystonia 1, 613280
SLC30A2	147.9	100%	100%	Zinc deficiency, transient neonatal, 608118
SLC33A1	152.6	98%	100%	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539
SLC34A1	163.1	100%	95%	Fanconi renotubular syndrome 2, 613388 Hypercalcemia, infantile 2, 616963 Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286
SLC34A2	182.7	100%	99%	Pulmonary alveolar microlithiasis, 265100
SLC34A3	120.6	99%	97%	Hypophosphatemic rickets with hypercalciuria, 241530
SLC35A1	169.7	100%	96%	Congenital disorder of glycosylation, type IIf, 603585

SLC35A2	111.5	99%	99%	Congenital disorder of glycosylation, type IIm, 300896
SLC35C1	244.4	99%	100%	Congenital disorder of glycosylation, type IIc, 266265
SLC35D1	144.4	97%	99%	Schneckenbecken dysplasia, 269250
SLC36A2	123.5	100%	99%	Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC37A4	139.2	100%	99%	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240
SLC38A8	80.4	99%	95%	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218
SLC39A13	134.6	99%	99%	Spondylocheirodysplasia, Ehlers-Danlos syndrome-like, 612350
SLC39A14	120.5	99%	99%	Hypermanganesemia with dystonia 2, 617013
SLC39A4	93.7	99%	99%	Acrodermatitis enteropathica, 201100
SLC39A5	146.8	100%	100%	Myopia 24, autosomal dominant, 615946
SLC39A8	160.7	100%	100%	Congenital disorder of glycosylation, type IIn, 616721
SLC3A1	205.3	100%	99%	Cystinuria, 220100
SLC40A1	167.8	99%	100%	Hemochromatosis, type 4, 606069
SLC45A2	152.5	99%	99%	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	103.2	99%	98%	Folate malabsorption, hereditary, 229050
SLC4A1	154.1	100%	100%	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	160.3	100%	99%	Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy, autosomal recessive, 217700 Corneal endothelial dystrophy and perceptive deafness, 217400

SLC4A4	159.1	99%	99%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC52A1	231.4	100%	99%	Riboflavin deficiency, 615026
SLC52A2	206.8	100%	99%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	108.8	99%	99%	Brown-Vialetto-Van Laere syndrome 1, 211530 Fazio-Londe disease, 211500
SLC5A1	149.1	100%	99%	Glucose/galactose malabsorption, 606824
SLC5A2	139.8	100%	99%	Renal glucosuria, 233100
SLC5A5	109.9	99%	81%	Thyroid dyshormonogenesis 1, 274400
SLC5A7	144.4	100%	100%	Neuronopathy, distal hereditary motor, type VIIA, 158580
SLC6A1	167	100%	99%	Myoclonic-atonic epilepsy, 616421
SLC6A17	219.8	100%	99%	Mental retardation, autosomal recessive 48, 616269
SLC6A19	177.6	99%	97%	Hartnup disorder, 234500 Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC6A2	173.9	100%	99%	Orthostatic intolerance, 604715
SLC6A20	189.1	100%	94%	Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC6A3	150.7	100%	91%	Parkinsonism-dystonia, infantile, 613135 {Nicotine dependence, protection against}, 188890
SLC6A5	155.8	100%	87%	Hyperekplexia 3, 614618
SLC6A8	59	91%	98%	Cerebral creatine deficiency syndrome 1, 300352
SLC7A14	198.4	100%	100%	Retinitis pigmentosa 68, 615725
SLC7A7	114.5	100%	100%	Lysinuric protein intolerance, 222700
SLC7A9	144.1	99%	100%	Cystinuria, 220100
SLC9A3	173.3	99%	98%	Diarrhea 8, secretory sodium, congenital, 616868
SLC9A3R1	134.2	100%	99%	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
SLC9A6	119.2	98%	99%	Mental retardation, X-linked syndromic, Christianson type, 300243
SLCO1B1	65.1	95%	99%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO1B3	65.3	97%	83%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO2A1	111.8	99%	100%	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
SLFN14	230.7	100%	96%	Bleeding disorder,platelet-type,20,616913
SLITRK1	146.4	100%	99%	Tourette syndrome, 137580 ?Trichotillomania, 613229
SLITRK6	263.8	100%	97%	Deafness and myopia, 221200

SLURP1	117	100%	99%	Meleda disease, 248300
SLX4	129.5	100%	100%	Fanconi anemia, complementation group P, 613951
SMAD3	151.7	99%	90%	Loeys-Dietz syndrome 3, 613795
SMAD4	136.5	99%	98%	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Polyposis, juvenile intestinal, 174900
SMAD6	111	91%	88%	Aortic valve disease 2, 614823
SMAD9	149.8	100%	94%	Pulmonary hypertension,primary,615342 Polyposis & gastrointestinal ganglioneuromas (Ngeow (2015) Gastroenterology 149,886)
SMARCA2	127.7	97%	96%	Nicolaides-Baraitser syndrome, 601358
SMARCA4	165.4	100%	94%	Coffin-Siris syndrome 4, 614609 {Rhabdoid tumor predisposition syndrome 2}, 613325
SMARCAD1	103.7	99%	97%	Adermatoglyphia, 136000
SMARCAL1	153.8	100%	93%	Schimke immunoosseous dysplasia, 242900
SMARCB1	265.7	100%	98%	Coffin-Siris syndrome 3, 614608 Rhabdoid tumors, somatic, 609322 {Rhabdoid predisposition syndrome 1}, 609322 {Schwannomatosis-1, susceptibility to}, 162091
SMARCE1	97.3	97%	98%	Coffin-Siris syndrome 5,616938 {Meningioma, familial, susceptibility to}, 607174
SMC1A	114.2	100%	77%	Cornelia de Lange syndrome 2,300590
SMC3	96.1	94%	99%	Cornelia de Lange syndrome 3, 610759
SMCHD1	114.1	98%	100%	Fascioscapulohumeral muscular dystrophy 2, digenic, 158901
SMN1	119.9	99%	100%	Spinal muscular atrophy-1, 253300 Spinal muscular atrophy-2, 253550 Spinal muscular atrophy-3, 253400 Spinal muscular atrophy-4, 271150
SMO	177.8	98%	100%	Basal cell carcinoma, somatic, 605462 Curry-Jones syndrome,somatic mosaic, 601707
SMOC1	130.7	99%	97%	Microphthalmia with limb anomalies, 206920
SMOC2	135	97%	99%	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SMPD1	151.8	99%	97%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616

SMPX	84.6	100%	97%	Deafness, X-linked 4, 300066
SMS	79.4	89%	99%	Mental retardation, X-linked, Snyder-Robinson type, 309583
SNAI2	128.7	100%	77%	Piebaldism, 172800 Waardenburg syndrome, type 2D, 608890
SNAP29	149.1	100%	99%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNCA	158.8	100%	88%	Dementia, Lewy body, 127750 Parkinson disease 1, 168601 Parkinson disease 4, 605543
SNCB	81.9	100%	89%	Dementia, Lewy body, 127750
SNIP1	164.9	99%	99%	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501
SNRNP200	165.8	99%	95%	Retinitis pigmentosa 33, 610359
SNRPB	91	99%	97%	Cerebrocostomandibular syndrome, 117650
SNRPE	92.4	99%	99%	Hypotrichosis 11, 615059
SNRPN	128.6	100%	95%	Prader-Willi syndrome, 176270
SNTA1	104.9	85%	98%	Long QT syndrome 12, 612955
SNX10	142	100%	93%	Osteopetrosis, autosomal recessive 8, 615085
SNX14	89.1	96%	58%	Spinocerebellar ataxia, autosomal recessive 20, 616354
SOBP	136.1	95%	98%	Mental retardation, anterior maxillary protrusion, and strabismus, 613671
SOD1	150.2	100%	81%	Amyotrophic lateral sclerosis 1, 105400
SOS1	120.1	98%	99%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOS2	115.8	99%	93%	Noonan syndrome 9, 616559
SOST	133.2	100%	100%	Craniodiaphyseal dysplasia, autosomal dominant, 122860 Sclerosteosis 1, 269500 Van Buchem disease, 239100
SOX10	74.6	98%	91%	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266
SOX11	129.8	99%	100%	Mental retardation, autosomal dominant, 27, 615866
SOX17	83.7	99%	88%	Vesicoureteral reflux 3, 613674
SOX18	27.6	78%	99%	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823 Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940
SOX2	127.1	99%	95%	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900

SOX3	47	94%	99%	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000
SOX5	125.3	100%	89%	Lamb-Shaffer syndrome, 616803
SOX9	138.7	97%	98%	Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia with autosomal sex reversal, 114290 Campomelic dysplasia, 114290
SP110	148.5	100%	98%	Hepatic venoocclusive disease with immunodeficiency, 235550 {Mycobacterium tuberculosis, susceptibility to}, 607948
SPAG1	103.9	97%	98%	Ciliary dyskinesia, primary, 28, 615505
SPARC	172.7	100%	92%	Osteogenesis imperfecta, type XVII, 616507
SPAST	82.7	97%	99%	Spastic paraplegia 4, autosomal dominant, 182601
SPATA5	153.8	100%	98%	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPATA7	149.2	98%	91%	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, autosomal recessive, 604232
SPECC1L	164.4	100%	90%	Opitz GBBB syndrome, type II, 145410 ?Facial clefting, oblique, 1, 600251
SPEG	106.8	96%	96%	Centronuclear myopathy 5, 615959
SPG11	145	99%	100%	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360
SPG20	163.8	99%	100%	Troyer syndrome, 275900
SPG21	141.5	99%	99%	Mast syndrome, 248900
SPG7	138.5	96%	99%	Spastic paraplegia 7, autosomal recessive, 607259
SPINK1	95.1	100%	100%	Pancreatitis, hereditary, 167800 Tropical calcific pancreatitis, 608189 {Fibrocalculous pancreatic diabetes, susceptibility to}, 608189
SPINK5	182.2	99%	99%	Atopy, 147050 Netherton syndrome, 256500
SPINT2	66	99%	96%	Diarrhea 3, secretory sodium, congenital, syndromic, 270420
SPR	194	99%	99%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRED1	196.7	99%	96%	Legius syndrome, 611431
SPRTN	182.3	100%	98%	Ruijs-Aalfs syndrome, 616200
SPRY4	146.5	100%	98%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266

SPTA1	144.3	99%	97%	Elliptocytosis-2, 130600 Pyropoikilocytosis, 266140 Spherocytosis, type 3, 270970
SPTAN1	137.5	99%	99%	Epileptic encephalopathy, early infantile, 5, 613477
SPTB	166.8	100%	88%	Anemia, neonatal hemolytic, fatal and near-fatal Elliptocytosis-3 Spherocytosis, type 2, 616649
SPTBN2	127.6	99%	50%	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia, autosomal recessive 14, 615386
SPTLC1	131.7	99%	99%	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	174.4	100%	98%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SQSTM1	142.3	99%	99%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 Paget disease of bone 3, 167250
SRC	124.7	99%	94%	Colon cancer, advanced, somatic ?Thrombocytopenia 6,616937
SRCAP	165.3	99%	99%	Floating-Harbor syndrome, 136140
SRD5A2	96.1	99%	97%	Pseudovaginal perineoscrotal hypospadias, 264600
SRD5A3	172	100%	100%	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713
SRP72	82.3	95%	96%	Bone marrow failure syndrome 1, 614675
SRY	58.9	50%	99%	46XX sex reversal 1, 400045 46XY sex reversal 1, 400046
SSTR5	192.2	99%	99%	Somatostatin analog, resistance to
ST14	177.8	99%	96%	Ichthyosis, congenital, autosomal recessive 11, 602400
ST3GAL3	195.5	100%	99%	Epileptic encephalopathy, early infantile, 15, 615006 Mental retardation, autosomal recessive 12, 611090
ST3GAL5	144.8	95%	97%	Amish infantile epilepsy syndrome, 609056
STAC3	131.7	100%	97%	Native American myopathy, 255995
STAMBP	125	99%	99%	Microcephaly-capillary malformation syndrome, 614261
STAR	147.6	100%	89%	Lipoid adrenal hyperplasia, 201710
STAT1	161.2	98%	99%	Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796 Immunodeficiency 31C, autosomal dominant, 614162
STAT2	125.3	100%	99%	Immunodeficiency 44, 616636

STAT3	132.1	100%	97%	Autoimmune disease, multisystem, infantile-onset, 615952 Hyper-IgE recurrent infection syndrome, 147060
STAT5B	140.5	99%	95%	Growth hormone insensitivity with immunodeficiency, 245590 Leukemia, acute promyelocytic, somatic, 102578
STIL	205.1	99%	99%	Microcephaly 7, primary, autosomal recessive, 612703
STIM1	144	99%	100%	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 1 160565 Stormorken syndrome, 185070
STK11	127.1	99%	98%	Melanoma, malignant, somatic Pancreatic cancer, 260350 Peutz-Jeghers syndrome, 175200 Testicular tumor, somatic, 273300
STK4	162.5	100%	99%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STOX1	196.2	89%	99%	Preeclampsia/eclampsia 4, 609404
STRA6	120.5	100%	98%	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186
STRADA	144.2	100%	86%	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087
STRC	111.3	99%	99%	Deafness, autosomal recessive 16, 603720
STS	104	99%	97%	Ichthyosis, X-linked, 308100
STUB1	186.3	100%	94%	Spinocerebellar ataxia, autosomal recessive 16, 615768
STX11	348.1	100%	45%	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STX16	153.7	99%	100%	Pseudohypoparathyroidism, type IB, 603233
STX1B	191.4	100%	95%	Generalized epilepsy with febrile seizures plus, type 9, 616172
STXBP1	146.2	100%	71%	Epileptic encephalopathy, early infantile, 4, 612164
STXBP2	144.3	99%	99%	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
SUCLA2	78.2	94%	97%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	120	100%	95%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUFU	146	99%	86%	Basal cell nevus syndrome, 109400 Medulloblastoma, desmoplastic, 155255 {Meningioma, familial, susceptibility to}, 607174
SUMF1	137.1	99%	97%	Multiple sulfatase deficiency, 272200
SUMO1	22.3	63%	96%	Orofacial cleft 10, 613705

SUOX	221.3	100%	87%	Sulfite oxidase deficiency, 272300
SYCP3	107.4	99%	99%	Pregnancy loss, recurrent, 4, 270960 Spermatogenic failure 4, 270960
SYN1	73.7	84%	99%	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNE1	172	99%	97%	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 Spinocerebellar ataxia, autosomal recessive 8, 610743
SYNE2	135.1	99%	98%	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999
SYNE4	80.7	99%	90%	Deafness, autosomal recessive 76, 615540
SYNGAP1	77.1	95%	93%	Mental retardation, autosomal dominant 5, 612621
SYNJ1	138.6	99%	99%	Parkinson disease 20, early-onset, 615530
SYP	81.3	99%	97%	Mental retardation, X-linked 96, 300802
SYT14	208	96%	97%	Spinocerebellar ataxia, autosomal recessive 11, 614229
SYT2	116.6	100%	97%	Myasthenic syndrome, congenital, 7, presynaptic, 616040
SZT2	158.8	99%	68%	Epileptic encephalopathy, early infantile, 18, 615476
T	162.4	99%	100%	Sacral agenesis with vertebral anomalies, 615709 {Neural tube defects, susceptibility to}, 182940
TAB2	232.8	99%	99%	Congenital heart defects, nonsyndromic, 2, 614980
TAC3	80.1	98%	100%	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACO1	103.3	97%	18%	Mitochondrial complex IV deficiency, 220110
TACR3	178.6	100%	20%	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
TACSTD2	232.7	99%	53%	Corneal dystrophy, gelatinous drop-like, 204870
TAF1	133.9	99%	86%	Dystonia-Parkinsonism, X-linked, 314250 Mental retardation, X-linked, syndromic 33, 300966
TAF2	139.3	99%	100%	Mental retardation, autosomal recessive 40, 615599
TAL1	55	85%	100%	Leukemia, T-cell acute lymphocytic, somatic, 613065
TAL2	101.9	100%	98%	Leukemia, T-cell acute lymphocytic, somatic, 613065
TALDO1	153.4	100%	94%	Transaldolase deficiency, 606003
TANGO2	158.8	100%	99%	Metabolic encephalomyopathic crises,recurrent,with rhabdomyolysis,cardiac arrhythmias and neurodegeneration,616878
TAP1	13.1	52%	97%	Bare lymphocyte syndrome, type I, 604571
TAP2	12.6	50%	99%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571 Wegener-like granulomatosis
TAPBP	23.6	83%	93%	Bare lymphocyte syndrome, type I, 604571
TAPT1	113.6	89%	95%	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type,616897

TARDBP	176.5	100%	82%	Amyotrophic lateral sclerosis 10, with or without FTD, 612069 Frontotemporal lobar degeneration, TARDBP-related, 612069
TAT	140.9	100%	99%	Tyrosinemia, type II, 276600
TAZ	123.4	99%	70%	Barth syndrome, 302060
TBC1D20	165.4	94%	99%	Warburg micro syndrome 4, 615663
TBC1D24	203.2	100%	95%	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	118.4	99%	99%	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000
TBCE	162.8	99%	99%	Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Kenny-Caffey syndrome, type 1, 244460
TBCK	111.7	97%	90%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3,616900
TBK1	119.5	99%	96%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439
TBL1XR1	103.7	95%	97%	Mental retardation, autosomal dominant 41,616944 Piermont syndrome,602342
TBP	137.1	100%	94%	Spinocerebellar ataxia 17, 607136 {Parkinson disease, susceptibility to}, 168600
TBX1	90.6	79%	100%	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Velocardiofacial syndrome, 192430
TBX15	127.2	100%	86%	Cousin syndrome, 260660
TBX18	89.8	98%	100%	Congenital anomalies of kidney and urinary tract 2, 143400
TBX19	204.3	100%	97%	Adrenocorticotropic hormone deficiency, 201400
TBX20	137.1	100%	100%	Atrial septal defect 4, 611363
TBX21	97	95%	96%	Asthma and nasal polyps, 208550 {Asthma, aspirin-induced, susceptibility to}, 208550
TBX22	139.9	99%	99%	Cleft palate with ankyloglossia, 303400 ?Abruzzo-Erickson syndrome, 302905
TBX3	95.1	99%	89%	Ulnar-mammary syndrome, 181450
TBX4	198	96%	100%	Ischiocoxopodopatellar syndrome, 147891
TBX5	145.9	100%	97%	Holt-Oram syndrome, 142900

TBX6	132.1	96%	93%	Spondylocostal dysostosis 5, 122600
TBXAS1	170.1	100%	97%	Ghosal hematodiaphyseal syndrome, 231095 ?Thromboxane synthase deficiency, 614158
TCAP	99.8	99%	99%	Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, type 2G, 601954
TCF12	173.7	100%	95%	Craniosynostosis 3, 615314
TCF3	90.7	99%	98%	Agammaglobulinemia 8,autosomal dominant,616941
TCF4	150.3	100%	99%	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954
TCIRG1	127.8	96%	98%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	201.5	100%	99%	Transcobalamin II deficiency, 275350
TCOF1	110.5	99%	99%	Treacher Collins syndrome 1, 154500
TCTN1	116.8	96%	98%	Joubert syndrome 13, 614173
TCTN2	166.2	99%	100%	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	133.6	100%	100%	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TDGF1	170.5	99%	95%	Forebrain defects
TDP1	128.3	99%	99%	Spinocerebellar ataxia, autosomal recessive with axonal neuropathy, 607250
TDP2	203.7	100%	99%	Spinocerebellar ataxia, autosomal recessive, 616949
TDRD7	176.2	99%	99%	Cataract 36, 613887
TEAD1	173.5	100%	86%	Sveinsson choreoretinal atrophy, 108985
TECPR2	169.7	100%	100%	Spastic paraplegia 49, autosomal recessive, 615031
TECR	108.3	99%	99%	Mental retardation, autosomal recessive 14, 614020
TECTA	226.1	100%	96%	Deafness, autosomal dominant 8/12, 601543 Deafness, autosomal recessive 21, 603629
TEK	200.5	100%	96%	Glaucoma 3,primary congenital, E, 617272 Venous malformations, multiple cutaneous and mucosal, 600195
TELO2	121.1	98%	94%	You-Hoover-Fong syndrome, 616954
TENM3	217.1	99%	96%	Microphthalmia, isolated, with coloboma 9, 615145
TENM4	165	100%	99%	Tremor, hereditary essential, 5, 616736
TERC	999999		99%	Dyskeratosis congenita, autosomal dominant 1, 127550 {Aplastic anemia}, 614743 {Pulmonary fibrosis, idiopathic, susceptibility to}, 614743

TET2	226.1	100%	93%	Myelodysplastic syndrome, somatic, 614286
TEX11	94.5	92%	98%	Spermatogenic failure, X-linked, 2, 309120
TF	141.5	100%	99%	Atransferrinemia, 209300
TFAP2A	126.8	100%	100%	Branchiooculofacial syndrome, 113620
TFAP2B	145.4	98%	94%	Char syndrome, 169100 Patent ductus arteriosus 2, 617035
TFE3	85.4	99%	93%	Renal cell carcinoma, papillary, 1, 300854
TFG	131.7	99%	100%	Hereditary motor and sensory neuropathy, Okinawa type, 604484 ?Spastic paraplegia 57, autosomal recessive, 615658
TFR2	107.4	99%	99%	Hemochromatosis, type 3, 604250
TFRC	190.2	100%	100%	Immunodeficiency 46, 616740
TG	148.9	100%	99%	Thyroid dyshormonogenesis 3, 274700 {Autoimmune thyroid disease, susceptibility to, 3}, 608175
TGDS	112.7	99%	98%	Catel-Manzke syndrome, 616145
TGFB1	84.7	99%	93%	Camurati-Engelmann disease, 131300 {Cystic fibrosis lung disease, modifier of}, 219700
TGFB2	203	100%	99%	Loeys-Dietz syndrome 4, 614816
TGFB3	177.7	100%	98%	Arrhythmogenic right ventricular dysplasia 1, 107970 Loeys-Dietz syndrome 5, 615582
TGFBI	152.7	99%	91%	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	204.7	94%	100%	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800
TGFBR2	212.4	100%	100%	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome 2, 610168
TGIF1	151.7	100%	99%	Holoprosencephaly-4, 142946
TGM1	185.6	100%	99%	Ichthyosis, congenital, autosomal recessive 1, 242300
TGM5	184.6	100%	91%	Peeling skin syndrome 2, 609796

TGM6	163.5	99%	71%	Spinocerebellar ataxia 35, 613908
TH	87.6	97%	100%	Segawa syndrome, recessive, 605407
THAP1	162.3	100%	100%	Dystonia 6, torsion, 602629
THBD	129.5	99%	99%	Thrombophilia due to thrombomodulin defect, 614486 {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
THOC2	95	97%	90%	Mental retardation, X-linked 12/35, 300957
THOC6	269.9	100%	97%	Beaulieu-Boycott-Innes syndrome, 613680
THPO	95.6	100%	99%	Thrombocythemia 1, 187950
THRA	198.8	99%	100%	Hypothyroidism, congenital, nongoitrous, 6, 614450
THRB	204.9	99%	96%	Thyroid hormone resistance, 188570 Thyroid hormone resistance, autosomal recessive, 274300 Thyroid hormone resistance, selective pituitary, 145650
TIA1	155.1	97%	97%	Welander distal myopathy, 604454
TIMM8A	43.7	88%	94%	Jensen syndrome, 311150 Mohr-Tranebjaerg syndrome, 304700
TIMP3	169.1	100%	99%	Sorsby fundus dystrophy, 136900
TINF2	208.7	100%	88%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TJP2	129.6	99%	98%	Cholestasis, progressive familial intrahepatic 4, 615878 Hypercholanemia, familial, 607748
TK2	105.4	94%	99%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560
TKT	131.2	98%	98%	Short stature, developmental delay, and congenital heart defects, 617044
TLE6	121.6	100%	96%	Preimplantation embryonic lethality, 616814
TLL1	173.9	100%	99%	Atrial septal defect 6, 613087
TMC1	148.4	98%	99%	Deafness, autosomal dominant 36, 606705 Deafness, autosomal recessive 7, 600974
TMC6	88.2	99%	98%	Epidermodysplasia verruciformis, 226400
TMC8	128.1	98%	98%	Epidermodysplasia verruciformis, 226400
TMCO1	114.3	100%	99%	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980
TMEM126A	112.9	98%	100%	Optic atrophy 7, 612989
TMEM126B	111.8	99%	99%	Mitochondrial complex I deficiency,252010
TMEM138	128	100%	93%	Joubert syndrome 16, 614465
TMEM165	136.2	99%	89%	Congenital disorder of glycosylation, type IIk, 614727
TMEM173	106.3	99%	93%	STING-associated vasculopathy, infantile-onset, 615934

TMEM199	140.3	100%	94%	Congenital disorder of glycosylation, type IIp, 616829
TMEM216	147.1	99%	94%	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	105.5	99%	92%	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	126	99%	99%	Joubert syndrome 14, 614424
TMEM240	136.1	100%	99%	Spinocerebellar ataxia 21, 607454
TMEM38B	124.3	100%	99%	Osteogenesis imperfecta, type XIV, 615066
TMEM43	145.6	99%	100%	Arrhythmogenic right ventricular dysplasia 5, 604400 Emery-Dreifuss muscular dystrophy 7, AD, 614302
TMEM5	138.9	96%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
TMEM67	93.3	95%	91%	COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991
TMEM70	172	96%	100%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMEM98	158.7	98%	99%	Nanophthalmos 4, 615972
TMIE	129	99%	71%	Deafness, autosomal recessive 6, 600971
TMPRSS15	140	97%	89%	Enterokinase deficiency,226200
TMPRSS3	126.7	99%	96%	Deafness, autosomal recessive 8/10, 601072
TMPRSS6	119.3	99%	99%	Iron-refractory iron deficiency anemia, 206200
TNC	199.3	100%	100%	Deafness, autosomal dominant 56, 615629
TNFAIP3	159.5	100%	100%	Autoinflammatory syndrome, familial, Behcet-like, 616744
TNFRSF10B	145.6	100%	95%	Squamous cell carcinoma, head and neck, 275355
TNFRSF11A	152.6	94%	95%	Osteolysis, familial expansile, 174810 Osteopetrosis, autosomal recessive 7, 612301 {Paget disease of bone 2, early-onset}, 602080
TNFRSF11B	258.6	100%	99%	Paget disease of bone 5, juvenile-onset, 239000
TNFRSF13B	115.4	100%	98%	Immunodeficiency, common variable, 2, 240500 Immunoglobulin A deficiency 2, 609529
TNFRSF13C	57.8	89%	99%	Immunodeficiency, common variable, 4, 613494
TNFRSF1A	96.7	91%	32%	Periodic fever, familial, 142680 {Multiple sclerosis, susceptibility to, 5}, 614810

TNFSF11	177.2	99%	100%	Osteopetrosis, autosomal recessive 2, 259710
TNIK	139.9	100%	99%	Mental retardation, autosomal recessive 54, 617028
TNNC1	208.9	100%	97%	Cardiomyopathy, dilated, 1Z, 611879 Cardiomyopathy, hypertrophic, 13, 613243
TNNI2	135.4	100%	100%	Arthrogryposis multiplex congenita, distal, type 2B, 601680
TNNI3	122.5	99%	95%	Cardiomyopathy, dilated, 1FF, 613286 Cardiomyopathy, familial restrictive, 1, 115210 Cardiomyopathy, hypertrophic, 7, 613690 ?Cardiomyopathy, dilated, 2A, 611880
TNNT1	103.2	99%	99%	Nemaline myopathy 5, Amish type, 605355
TNNT2	117.4	100%	98%	Cardiomyopathy, dilated, 1D, 601494 Cardiomyopathy, familial restrictive, 3, 612422 Cardiomyopathy, hypertrophic, 2, 115195 Left ventricular noncompaction 6, 601494
TNNT3	135.1	99%	99%	Arthyrgryposis, distal, type 2B, 601680
TNPO3	163	100%	87%	Muscular dystrophy, limb-girdle, type 1F, 608423
TNXB	17.8	58%	97%	Ehlers-Danlos syndrome due to tenascin X deficiency, 606408 Vesicoureteral reflux 8, 615963
TOPORS	261.3	100%	100%	Retinitis pigmentosa 31, 609923
TOR1A	210.4	100%	75%	Dystonia-1, torsion, 128100 {Dystonia-1, modifier of}
TP53	100.3	99%	100%	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	226.8	100%	93%	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	104.9	98%	100%	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512
TPK1	126.3	100%	97%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TPM1	155.6	99%	77%	Cardiomyopathy, dilated, 1Y, 611878 Cardiomyopathy, hypertrophic, 3, 115196 Left ventricular noncompaction 9, 611878
TPM2	120.8	100%	99%	Arthrogryposis multiplex congenita, distal, type 1, 108120 Arthrogryposis, distal, type 2B, 601680 CAP myopathy 2, 609285 Nemaline myopathy 4, autosomal dominant, 609285
TPM3	96.1	89%	79%	CAP myopathy 1, 609284 Myopathy, congenital, with fiber-type disproportion, 255310 Nemaline myopathy 1, autosomal dominant or recessive, 609284
TPO	165	99%	98%	Thyroid dyshormonogenesis 2A, 274500
TPP1	155.4	100%	100%	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TPRN	82.9	83%	98%	Deafness, autosomal recessive 79, 613307
TRAC	164.6	100%	100%	Immunodeficiency 7, TCR-alpha/beta deficient, 615387
TRAF3IP1	97.5	96%	99%	Senior-Loken syndrome 9, 616629
TRAIP	148.3	100%	96%	Seckel syndrome 9, 616777
TRAPPC11	156	99%	96%	Muscular dystrophy, limb-girdle, type 2S, 615356
TRAPPC2	113.4	91%	92%	Spondyloepiphyseal dysplasia tarda, 313400
TRAPPC9	164.3	100%	99%	Mental retardation, autosomal recessive 13, 613192
TRDN	85.6	90%	99%	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441
TREM2	149.5	99%	98%	Nasu-Hakola disease, 221770
TREX1	302.9	100%	96%	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700
TRIM2	186.5	99%	99%	Charcot-Marie-Tooth disease, type 2R, 615490
TRIM32	147.3	100%	94%	Muscular dystrophy, limb-girdle, type 2H, 254110 ?Bardet-Biedl syndrome 11, 615988

TRIM37	137.3	100%	88%	Mulibrey nanism, 253250
TRIO	158.3	98%	96%	Mental retardation, autosomal dominant 44,617061
TRIOBP	150.3	97%	99%	Deafness, autosomal recessive 28, 609823
TRIP11	114.2	97%	98%	Achondrogenesis, type IA, 200600
TRIP4	123.9	100%	99%	Spinal muscular atrophy with congenital bone fractures 1,616866 ?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066
TRMT10A	176.3	100%	100%	Microcephaly, short stature, and impaired glucose metabolism 1, 616033
TRMT10C	144.7	99%	99%	Combined oxidative phosphorylation deficiency 30, 616974
TRMT5	228.2	99%	99%	Combined oxidative phosphorylation deficiency 26, 616539
TRMU	118.6	100%	97%	Liver failure, transient infantile, 613070 {Deafness, mitochondrial, modifier of}, 580000
TRNT1	123.4	98%	99%	Retinitis pigmentosa and erythrocytic microcytosis, 616959 Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084
TRPA1	111.4	93%	93%	Episodic pain syndrome, familial, 615040
TRPC6	127.5	98%	99%	Glomerulosclerosis, focal segmental, 2, 603965
TRPM1	185.9	100%	86%	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TRPM4	128	99%	93%	Progressive familial heart block, type IB, 604559
TRPM6	166.1	100%	99%	Hypomagnesemia 1, intestinal, 602014
TRPS1	196.7	100%	99%	Trichorhinophalangeal syndrome, type I, 190350 Trichorhinophalangeal syndrome, type III, 190351
TRPV3	165.4	100%	100%	Olmsted syndrome, 614594 ?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400
TRPV4	188.6	99%	98%	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	140.4	99%	98%	Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100
TSC2	150.1	99%	99%	Lymphangioleiomyomatosis, somatic, 606690

				Tuberous sclerosis-2, 613254
TSEN15	97.3	99%	99%	Pontocerebellar hypoplasia, type 2F, 617026
TSEN2	130.5	100%	99%	Pontocerebellar hypoplasia type 2B, 612389
TSEN34	62.1	93%	99%	Pontocerebellar hypoplasia type 2C, 612390
TSEN54	98.8	96%	99%	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753 ?Pontocerebellar hypoplasia type 5, 610204
TSFM	135.9	100%	82%	Combined oxidative phosphorylation deficiency 3, 610505
TSG101	109.9	100%	98%	Breast cancer, somatic, 114480
TSHB	372.8	100%	99%	Hypothryoidism, congenital, nongoitrous 4, 275100
TSHR	226.1	99%	98%	Hyperthyroidism, familial gestational, 603373 Hyperthyroidism, nonautoimmune, 609152 Hypothyroidism, congenital, nongoitrous, 1 275200 Thyroid adenoma, hyperfunctioning, somatic Thyroid carcinoma with thyrotoxicosis
TSHZ1	178.2	98%	98%	Aural atresia, congenital, 607842
TSPAN12	163.4	100%	98%	Exudative vitreoretinopathy 5, 613310
TSPAN7	137.2	100%	99%	Mental retardation, X-linked 58, 300210
TSPEAR	153.1	100%	97%	Deafness, autosomal recessive 98, 614861 Ectodermal dysplasia (Peled et al. (2016) PLOS Genetics online)
TSPYL1	154	100%	97%	Sudden infant death with dysgenesis of the testes syndrome, 608800
TTBK2	165.1	100%	86%	Spinocerebellar ataxia 11, 604432
TTC19	105.8	90%	100%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTC21B	132.4	99%	97%	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
TTC25	102.4	99%	100%	Ciliary dyskinesia, primary, 35, 617092
TTC37	159.2	99%	99%	Trichohepatoenteric syndrome 1, 222470
TTC7A	131.2	99%	49%	Gastrointestinal defects and immunodeficiency syndrome, 243150
TTC8	118.3	99%	100%	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
TTI2	109.9	100%	96%	Mental retardation, autosomal recessive 39, 615541
TTLL5	169.1	99%	100%	Cone-rod dystrophy 19, 615860
TTN	236.7	98%	98%	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	128.8	94%	95%	Ataxia with isolated vitamin E deficiency, 277460
TTR	184.7	100%	75%	Amyloidosis, hereditary, transthyretin-related, 105210 Carpal tunnel syndrome, familial, 115430 [Dystransthyretinemic hyperthyroxinemia], 145680
TUBA1A	115.3	99%	100%	Lissencephaly 3, 611603
TUBA4A	237.6	100%	96%	Amyotrophic lateral sclerosis 22 with or without frontotemoral dementia, 616208
TUBA8	171.3	100%	99%	Polymicrogyria with optic nerve hypoplasia, 613180
TUBB	21.1	77%	99%	Cortical dysplasia, complex, with other brain malformations 6, 615771 Symmetric circumferential skin creases, congenital, 1, 156610
TUBB1	191	100%	95%	Macrothrombocytopenia, autosomal dominant, TUBB1-related, 613112
TUBB2A	111.8	99%	99%	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB2B	104.4	100%	85%	Polymicrogyria, symmetric or asymmetric, 610031
TUBB3	212.4	99%	98%	Cortical dysplasia, complex, with other brain malformations 1, 614039 Fibrosis of extraocular muscles, congenital, 3A, 600638
TUBB4A	135.6	96%	99%	Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438
TUBB8	32.9	96%	99%	Oocyte maturation defect 2, 616780
TUBG1	190.6	100%	89%	Cortical dysplasia, complex, with other brain malformations 4, 615412
TUBGCP4	149	99%	100%	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TUBGCP6	176.6	99%	100%	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
TUFM	151	100%	100%	Combined oxidative phosphorylation deficiency 4, 610678
TULP1	116.2	98%	98%	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132
TUSC3	166.8	100%	98%	Mental retardation, autosomal recessive 7, 611093
TWIST1	135.7	94%	99%	Craniosynostosis, type 1, 123100 Robinow-Sorauf syndrome, 180750 Saethre-Chotzen syndrome with eyelid anomalies, 101400 Saethre-Chotzen syndrome, 101400
TWIST2	124.4	99%	96%	Ablepharon-macrostomia syndrome, 200110 Barber-Say syndrome, 209885

				Focal facial dermal dysplasia 3, Setleis type, 227260
TXNL4A	132.6	100%	99%	Burn-McKeown syndrome, 608572
TYK2	136.2	99%	97%	Immunodeficiency 35, 611521
TYMP	104.7	98%	98%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYR	205.9	100%	98%	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	90.7	100%	100%	Nasu-Hakola disease, 221770
TYRP1	200.1	100%	96%	Albinism, oculocutaneous, type III, 203290 [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271
UBA1	176.9	99%	99%	Spinal muscular atrophy, X-linked 2, infantile, 301830
UBE2A	110.5	99%	97%	Mental retardation, X-linked syndromic, Nascimento-type, 300860
UBE2T	129.6	99%	99%	Fanconi anemia, complementation group T, 616435
UBE3A	117.2	99%	96%	Angelman syndrome, 105830
UBE3B	145.7	100%	100%	Kaufman oculocerebrofacial syndrome, 244450
UBIAD1	263.9	99%	87%	Corneal dystrophy, Schnyder type, 121800
UBQLN2	138.3	99%	97%	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857
UBR1	144.2	99%	99%	Johanson-Blizzard syndrome, 243800
UGT1A1	263	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	131.5	97%	97%	Glomerulocystic kidney disease with hyperuricemia and isosthenuria, 609886 Hyperuricemic nephropathy, familial juvenile 1, 162000 Medullary cystic kidney disease 2, 603860
UMPS	204.3	100%	100%	Orotic aciduria, 258900
UNC13D	111.2	99%	97%	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC80	151.4	99%	99%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801
UNG	84.6	99%	96%	Immunodeficiency with hyper IgM, type 5, 608106
UPB1	184.6	100%	99%	Beta-ureidopropionase deficiency, 613161

UPF3B	68.3	95%	94%	Mental retardation, X-linked, syndromic 14, 300676
UQCRB	124	99%	43%	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC2	150.8	100%	99%	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRQ	180.8	100%	97%	Mitochondrial complex III deficiency, nuclear type 4, 615159
UROD	174	99%	100%	Porphyria cutanea tarda, 176100 Porphyria, hepatoerythropoietic, 176100
UROS	126.7	100%	100%	Porphyria, congenital erythropoietic, 263700
USB1	157.7	99%	99%	Poikiloderma with neutropenia,604173
USH1C	119.4	100%	98%	Deafness, autosomal recessive 18A, 602092 Usher syndrome, type 1C, 276904
USH1G	185.8	98%	43%	Usher syndrome, type 1G, 606943
USH2A	178.6	100%	100%	Retinitis pigmentosa 39, 613809 Usher syndrome, type 2A, 276901
USP9X	135.2	98%	99%	Mental retardation, X-linked 99, 300919 Mental retardation, X-linked 99, syndromic, female-restricted, 300968
USP9Y	36.5	48%	99%	Spermatogenic failure, Y-linked, 2, 415000
UVSSA	138.1	100%	96%	UV-sensitive syndrome 3, 614640
VAC14	126.5	99%	99%	Striatonigral degeneration,childhood-onset,617054
VAMP1	156.6	100%	90%	Spastic ataxia 1, autosomal dominant, 108600
VANGL1	190.2	100%	100%	Caudal regression syndrome, 600145 {Neural tube defects, susceptibility to}, 182940
VANGL2	185.6	100%	99%	Neural tube defects, 182940
VAPB	119.5	99%	99%	Amyotrophic lateral sclerosis 8, 608627 Spinal muscular atrophy, late-onset, Finkel type, 182980
VARS2	20.5	68%	87%	Combined oxidative phosphorylation deficiency 20, 615917
VCAN	213.9	100%	99%	Wagner syndrome 1, 143200
VCL	117.4	100%	91%	Cardiomyopathy, dilated, 1W, 611407 Cardiomyopathy, hypertrophic, 15, 613255
VCP	155.7	100%	97%	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	124.6	99%	94%	Rickets, vitamin D-resistant, type IIA, 277440 ?Osteoporosis, involutional, 166710
VEGFC	194	100%	99%	Lymphedema, hereditary, ID, 615907

VHL	126	97%	76%	Erythrocytosis, familial, 2, 263400 Hemangioblastoma, cerebellar, somatic Pheochromocytoma, 171300 Renal cell carcinoma, somatic, 144700 von Hippel-Lindau syndrome, 193300
VIPAS39	156.9	100%	95%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VKORC1	161.5	100%	90%	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473 Warfarin resistance, 122700
VLDLR	217.6	100%	97%	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VMA21	56.6	96%	79%	Myopathy, X-linked, with excessive autophagy, 310440
VPS11	170.6	100%	96%	Leukodystrophy, hypomyelinating, 12, 616683
VPS13A	89.6	97%	99%	Choreoacanthocytosis, 200150
VPS13B	170.3	99%	98%	Cohen syndrome, 216550
VPS13C	130.4	98%	80%	Parkinson disease 23, autosomal recessive, early onset, 616840
VPS33B	142.9	100%	96%	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
VPS37A	86.4	93%	99%	Spastic paraplegia 53, autosomal recessive, 614898
VPS45	158.8	97%	96%	Neutropenia, severe congenital, 5, autosomal recessive, 615285
VPS53	145	91%	98%	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	157.2	99%	96%	Pontocerebellar hypoplasia type 1A, 607596
VSX1	67.7	92%	89%	Keratoconus 1, 148300 ?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195
VSX2	85.2	99%	97%	Microphthalmia with coloboma 3, 610092 Microphthalmia, isolated 2, 610093
VWF	129.6	100%	99%	von Willebrand disease, type 1, 193400 von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 von Willibrand disease, type 3, 277480
WAC	178.2	99%	98%	Desanto-Shinawi syndrome, 616708
WAS	72	89%	99%	Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, 313900 Thrombocytopenia, X-linked, intermittent, 313900 Wiskott-Aldrich syndrome, 301000
WDR11	142.6	97%	99%	Hypogonadotropic hypogonadism 14 with or without anosmia, 614858
WDR19	170	100%	99%	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307

				?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WDR34	122	99%	99%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
WDR35	186.3	99%	99%	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091
WDR36	144.1	99%	99%	Glaucoma 1, open angle, G, 609887
WDR45	85.2	95%	98%	Neurodegeneration with brain iron accululation 5, 300894
WDR60	131.2	99%	99%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WDR62	178	99%	99%	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
WDR72	161.3	99%	99%	Amelogenesis imperfecta, type IIA3, 613211
WDR73	147.2	100%	92%	Galloway-Mowat syndrome, 251300
WDR81	187.7	100%	99%	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185
WFS1	260.1	99%	99%	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	145.5	100%	100%	Leukemia, acute myeloid, 601626
WISP3	132	100%	96%	Arthropathy, progressive pseudorheumatoid, of childhood, 208230 Spondyloepiphyseal dysplasia tarda with progressive arthropathy, 208230
WNK1	178.6	99%	89%	Neuropathy, hereditary sensory and autonomic, type II, 201300 Pseudohypoaldosteronism, type IIC, 614492
WNK4	142.3	99%	99%	Pseudohypoaldosteronism, type IIB, 614491
WNT1	225.8	100%	99%	Osteogenesis imperfecta, type XV, 615220 {Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221
WNT10A	135.1	99%	90%	Odontoonychodermal dysplasia, 257980 Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400
WNT10B	146.6	100%	99%	Split-hand/foot malformation 6, 225300
WNT4	284.8	93%	89%	Mullerian aplasia and hyperandrogenism, 158330 SERKAL syndrome, 611812
WNT5A	181.5	100%	99%	Robinow syndrome, autosomal dominant 1, 180700
WNT7A	230.2	100%	97%	Fuhrmann syndrome, 228930 Ulna and fibula, absence of, with severe limb deficiency, 276820

WRAP53	175.2	100%	99%	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	149.6	99%	98%	Werner syndrome, 277700
WT1	94.2	96%	88%	Denys-Drash syndrome, 194080 Frasier syndrome, 136680 Meacham syndrome, 608978 Mesothelioma, somatic, 156240 Nephrotic syndrome, type 4, 256370 Wilms tumor, type 1, 194070
WWOX	143.3	100%	96%	Epileptic encephalopathy, early infantile, 28, 616211 Esophageal squamous cell carcinoma, somatic, 133239 Spinocrebellar ataxia, autosomal recessive 12, 614322
XDH	118.7	100%	84%	Xanthinuria, type I, 278300
XIAP	123	93%	99%	Lymphoproliferative syndrome, X-linked, 2, 300635
XIST	999999		99%	X-inactivation,familial skewed,300087
XK	107.1	99%	99%	McLeod syndrome with or without chronic granulomatous disease, 300842
XPA	69.2	97%	100%	Xeroderma pigmentosum, group A, 278700
XPC	176.3	100%	100%	Xeroderma pigmentosum, group C, 278720
XPNPEP3	142.9	98%	100%	Nephronophthisis-like nephropathy 1, 613159
XPR1	171.5	99%	97%	Basal ganglia calcification, idiopathic, 6, 616413
XRCC4	134.1	99%	90%	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	144.5	92%	99%	Desbuquois dysplasia 2, 615777 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
XYLT2	161.9	98%	99%	Spondyloocular syndrome, 605822 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
YAP1	112.2	90%	32%	Coloboma, ocular, 120433 Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433
YARS	144.8	100%	99%	Charcot-Marie-Tooth disease, dominant intermediate C, 608323
YARS2	199.3	99%	99%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
ZAP70	207.8	100%	99%	Autoimmune disease,multisystem,infantile-onset,2,617006 Immunodeficiency 48,269840
ZBTB16	165.9	100%	100%	Leukemia, acute promyelocytic, PL2F/RARA type Skeletal defects, genital hypoplasia, and mental retardation, 612447
ZBTB20	226.5	100%	84%	Primrose syndrome, 259050

ZBTB24	213.6	100%	99%	Immunodeficiency-centromeric instability-facial anomalies syndrome-2, 614069
ZC4H2	93.9	99%	99%	Wieacker-Wolff syndrome, 314580
ZDHHC9	63.6	99%	99%	Mental retardation, X-linked syndromic, Raymond type, 300799
ZEB1	197.2	100%	99%	Corneal dystrophy, Fuchs endothelial, 6, 613270 Corneal dystrophy, posterior polymorphous, 3, 609141
ZEB2	195.1	100%	100%	Mowat-Wilson syndrome, 235730
ZFP57	16.3	76%	99%	Diabetes mellitus, transient neonatal, 1, 601410
ZFPM2	208.3	100%	100%	46XY sex reversal 9, 616067 Diaphragmatic hernia 3, 610187 Tetralogy of Fallot, 187500
ZFYVE26	130.7	100%	97%	Spastic paraplegia 15, autosomal recessive, 270700
ZFYVE27	133.5	100%	99%	Spastic paraplegia 33, autosomal dominant, 610244
ZIC1	213	100%	99%	Craniosynostosis 6, 616602
ZIC2	116.8	92%	100%	Holoprosencephaly-5, 609637
ZIC3	127.1	100%	100%	Congenital heart defects, nonsyndromic, 1, X-linked, 306955 Heterotaxy, visceral, 1, X-linked 306955 VACTERL association, X-linked, 314390
ZMPSTE24	151.9	99%	97%	Mandibuloacral dysplasia with type B lipodystrophy, 608612 Restrictive dermopathy, lethal, 275210
ZMYND10	161.7	100%	100%	Ciliary dyskinesia, primary, 22, 615444
ZMYND11	146.6	100%	96%	Mental retardation, autosomal dominant 30, 616083
ZNF408	151.6	100%	99%	Retinitis pigmentosa 72, 616469 ?Exudative vitreoretinopathy 6, 616468
ZNF41	127.2	100%	91%	Mental retardation, X-linked 89, 300848
ZNF423	268.6	100%	100%	Joubert syndrome 19, 614844 Nephronophthisis 14, 614844
ZNF469	102.9	99%	99%	Brittle cornea syndrome 1, 229200
ZNF513	122.2	100%	100%	Retinitis pigmentosa 58, 613617
ZNF644	213.7	100%	100%	Myopia 21, autosomal dominant, 614167
ZNF674	140.4	100%	100%	Mental retardation, X-linked 92, 300851
ZNF687	195.1	100%	100%	Paget disease of bone 6, 616833
ZNF711	155.1	99%	99%	Mental retardation, X-linked 97, 300803
ZNF750	144.3	100%	100%	Seborrhea-like dermatitis with psoriasiform elements, 610227
ZNF81	93.1	99%	99%	Mental retardation, X-linked 45, 300498

ZP1	224.1	100%	100%	Oocyte maturation defect 1, 615774
ZSWIM6	176.5	94%	94%	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 : April 14th 2017
This list is accurate for panel version DG 2.9

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