MENDELIOME GENE PANEL DGD141114

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM ID
A4GALT	116.3	100%	100%	[Blood group, P1Pk system, p phenotype], 111400 [Blood group, P1Pk system, P(2) phenotype], 111400 NOR polyagglutination syndrome, 111400
AAAS	98.2	100%	99%	Achalasia-addisonianism-alacrimia syndrome, 231550
AAGAB	121.3	99%	95%	Keratoderma, palmoplantar, punctate type IA, 148600
AARS	91.7	98%	95%	Charcot-Marie-Tooth disease, axonal, type 2N, 613287
AARS2	86.4	99%	96%	Combined oxidative phosphorylation deficiency 8, 614096
AASS	101.1	100%	99%	Hyperlysinemia, 238700 Saccharopinuria, 268700 (1)
ABAT	66.2	100%	92%	GABA-transaminase deficiency, 613163
ABCA1	86.2	100%	99%	Tangier disease, 205400 HDL deficiency, type 2, 604091 {Coronary artery disease in familial hypercholesterolemia, protection against}, 143890
ABCA12	104.6	100%	99%	Ichthyosis, congenital, autosomal recessive 4A, 601277 Ichthyosis, autosomal recessive 4B (harlequin), 242500
ABCA3	91.4	99%	96%	Surfactant metabolism dysfunction, pulmonary, 3, 610921
ABCA4	87.8	99%	96%	Stargardt disease 1, 248200 Retinitis pigmentosa 19, 601718 Cone-rod dystrophy 3, 604116 Macular degeneration, age-related, 2, 153800 Fundus flavimaculatus, 248200 Retinal dystrophy, early-onset severe, 248200
ABCB11	92.4	99%	97%	Cholestasis, progressive familial intrahepatic 2, 601847 Cholestasis, benign recurrent intrahepatic, 2, 605479
ABCB4	96.9	98%	95%	Cholestasis, progressive familial intrahepatic 3, 602347 Cholestasis, intrahepatic, of pregnancy, 3, 614972 Gallbladder disease 1, 600803
ABCB6	118.6	100%	98%	Microphthalmia, isolated, with coloboma 7, 614497 [Blood group, Langereis system], 111600 Dyschromatosis universalis hereditaria 3, 615402

ABCB7	57.9	100%	94%	Anemia, sideroblastic, with ataxia, 301310
ABCC2	102.8	100%	99%	Dubin-Johnson syndrome, 237500
ABCC6	51.1	71%	67%	Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850 Arterial calcification, generalized, of infancy, 2, 614473
ABCC8	83.1	100%	97%	Hyperinsulinemic hypoglycemia, familial, 1, 256450 Hypoglycemia of infancy, leucine-sensitive, 240800 Diabetes mellitus, transient neonatal 2, 610374 Diabetes mellitus, noninsulin-dependent, 125853 Diabetes mellitus, permanent neonatal, 6
ABCC9	107.1	100%	98%	Cardiomyopathy, dilated, 1O, 608569 Atrial fibrillation, familial, 12, 614050 Hypertrichotic osteochondrodysplasia, 239850
ABCD1	31.8	73%	64%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABCD4	100.4	100%	97%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	135.1	99%	87%	Sitosterolemia, 210250
ABCG8	90.7	97%	96%	Sitosterolemia, 210250 Gallbladder disease 4, 611465
ABHD12	61.0	98%	83%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ABHD5	121.2	100%	100%	Chanarin-Dorfman syndrome, 275630
ABL1	96.2	99%	97%	Leukemia, Philadelphia chromosome-positive, resistant to imatinib
ACAD8	92.1	99%	94%	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	91.0	100%	99%	ACAD9 deficiency, 611126
ACADM	147.3	100%	100%	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	107.7	100%	100%	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	85.7	98%	96%	2-methylbutyrylglycinuria, 610006
ACADVL	87.5	100%	98%	VLCAD deficiency, 201475
ACAN	116.2	93%	90%	Spondyloepiphyseal dysplasia, Kimberley type, 608361 Spondyloepimetaphyseal dysplasia, aggrecan type, 612813 Osteochondritis dissecans, short stature, and early-onset osteoarthritis, 165800
ACAT1	109.9	100%	98%	Alpha-methylacetoacetic aciduria, 203750

ACE	90.1	97%	95%	{Myocardial infarction, susceptibility to} {Alzheimer disease, susceptibility to}, 104300 {Microvascular complications of diabetes 3}, 612624 [Angiotensin I-converting enzyme, benign serum increase] {SARS, progression of} Renal tubular
ACO2	77.9	90%	83%	Infantile cerebellar-retinal degeneration, 614559
ACOX1	75.3	99%	93%	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACP5	98.3	100%	100%	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACSF3	77.6	100%	99%	Combined malonic and methylmalonic aciduria, 614265
ACSL4	67.4	100%	97%	Mental retardation, X-linked 63, 300387
ACSL6	86.9	100%	99%	Myelodysplastic syndrome Myelogenous leukemia, acute
ACTA1	73.6	99%	94%	Nemaline myopathy 3, autosomal dominant or recessive, 161800 Myopathy, actin, congenital, with excess of thin myofilaments, 161800 Myopathy, actin, congenital, with cores, 161800 Myopathy, congenital, with fiber-type disproportion 1, 255310
ACTA2	84.6	100%	99%	Aortic aneurysm, familial thoracic 6, 611788 Multisystemic smooth muscle dysfunction syndrome, 613834 Moyamoya disease 5, 614042
ACTB	60.1	97%	90%	Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACTC1	82.8	100%	93%	Cardiomyopathy, dilated, 1R, 613424 Cardiomyopathy, familial hypertrophic, 11, 612098 Atrial septal defect 5, 612794 Left ventricular noncompaction 4, 613424
ACTG1	56.9	99%	93%	Deafness, autosomal dominant 20/26, 604717 Baraitser-Winter syndrome 2, 614583
ACTN1	92.2	99%	97%	Bleeding disorder, platelet-type, 15, 615193
ACTN4	95.0	99%	96%	Glomerulosclerosis, focal segmental, 1, 603278
ACVR1	91.8	100%	97%	Fibrodysplasia ossificans progressiva, 135100
ACVR1B	105.2	98%	92%	Pancreatic cancer, somatic
ACVR2B	84.4	96%	96%	Heterotaxy, visceral, 4, autosomal, 613751
ACVRL1	55.3	93%	86%	Telangiectasia, hereditary hemorrhagic, type 2, 600376
ACY1	87.8	100%	97%	Aminoacylase 1 deficiency, 609924
ADA	70.7	100%	95%	Severe combined immunodeficiency due to ADA deficiency, 102700 Adenosine deaminase deficiency, partial, 102700

ADAM10	126.6	100%	100%	Reticulate acropigmentation of Kitamura, 615537 {Alzheimer disease 18, susceptibility to}
ADAM17	112.3	99%	98%	Inflammatory skin and bowel disease, neonatal, 614328
ADAM9	118.1	100%	98%	Cone-rod dystrophy 9, 612775
ADAMTS10	74.9	97%	92%	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS13	57.7	94%	84%	Thrombotic thrombocytopenic purpura, familial, 274150
ADAMTS17	75.2	92%	84%	Weill-Marchesani-like syndrome, 613195
ADAMTS18	99.3	100%	98%	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
ADAMTS2	95.1	97%	92%	Ehlers-Danlos syndrome, type VIIC, 225410
ADAMTSL2	64.7	96%	79%	Geleophysic dysplasia 1, 231050
ADAMTSL4	95.1	100%	96%	Ectopia lentis, isolated, autosomal recessive, 225100 Ectopia lentis et pupillae, 225200
ADAR	137.0	99%	98%	Dyschromatosis symmetrica hereditaria, 127400 Aicardi-Goutieres syndrome 6, 615010
ADAT3	48.3	100%	99%	Mental retardation, autosomal recessive 36, 615286
ADCK3	97.8	100%	95%	Coenzyme Q10 deficiency primary 4
ADCK4	68.6	100%	91%	Nephrotic syndrome, type 9, 615573 (3)
ADCY5	83.2	98%	94%	Dyskinesia, familial, with facial myokymia, 606703
ADIPOQ	144.6	100%	100%	Adiponectin deficiency, 612556
ADK	123.9	94%	94%	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADRB2	145.3	100%	100%	{Asthma, nocturnal, susceptibility to}, 600807 {Obesity, susceptibility to}, 601665 Beta-2-adrenoreceptor agonist, reduced response to
ADSL	127.7	100%	98%	ade(-)I bifunctional Adenylosuccinase deficiency, 103050
AFF2	70.4	98%	95%	Mental retardation, X-linked, FRAXE type, 309548
AFG3L2	76.4	95%	91%	Spinocerebellar ataxia 28, 610246 Ataxia, spastic, 5, autosomal recessive, 614487
AGA	117.2	100%	89%	Aspartylglucosaminuria
AGBL1	96.1	100%	99%	Corneal dystrophy, Fuchs endothelial, 8, 615523 (3)
AGK	106.6	100%	100%	Sengers syndrome, 212350 Cataract 38, autosomal recessive, 614691

AGL	145.6	100%	100%	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGPAT2	62.9	95%	82%	Lipodystrophy, congenital generalized, type 1, 608594
AGPS	116.3	100%	100%	Rhizomelic chondrodysplasia punctata, type 3, 600121
AGRN	80.7	97%	90%	Myasthenia, limb-girdle, familial, 254300
AGT	122.9	100%	100%	{Hypertension, essential, susceptibility to}, 145500 {Preeclampsia, susceptibility to} Renal tubular dysgenesis, 267430
AGTR1	147.4	97%	97%	Hypertension, essential, 145500 Renal tubular dysgenesis, 267430
AGXT	88.5	97%	90%	Hyperoxaluria, primary, type 1, 259900
AHCY	70.6	92%	73%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHI1	114.6	100%	99%	Joubert syndrome-3, 608629
AICDA	81.0	100%	99%	Immunodeficiency with hyper-IgM, type 2, 605258
AIFM1	57.2	99%	88%	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490
AIMP1	125.6	100%	100%	Leukodystrophy, hypomyelinating, 3, 260600
AIP	94.0	96%	91%	Pituitary adenoma, growth hormone-secreting, 102200 Pituitary adenoma, prolactin-secreting, 600634 Pituitary adenoma, ACTH-secreting, 219090
AIPL1	88.9	100%	100%	Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393 Cone-rod dystrophy, 604393
AIRE	70.1	98%	91%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AK1	86.6	100%	98%	Hemolytic anemia due to adenylate kinase deficiency, 612631
AK2	73.5	77%	75%	Reticular dysgenesis, 267500
AKAP9	131.6	100%	99%	Long QT syndrome-11, 611820
AKR1C2	76.9	96%	75%	Obesity, hyperphagia, and developmental delay 46XY sex reversal 8, 614279
AKR1D1	98.0	100%	100%	Bile acid synthesis defect, congenital, 2, 235555

AKT1	121.7	100%	97%	IGH Breast cancer, somatic, 114480 Colorectal cancer, somatic, 114500 Ovarian cancer, somatic, 167000 {Schizophrenia, susceptibility to}, 181500 (2) Proteus syndrome, somatic, 176920 Cowden syndrome 6, 615109
AKT2	117.1	98%	96%	Diabetes mellitus, type II, 125853 Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900
AKT3	115.6	100%	94%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome, 603387
ALAD	83.5	100%	95%	Porphyria, acute hepatic, 612740 {Lead poisoning, susceptibility to}, 612740
ALAS2	41.0	87%	79%	Anemia, sideroblastic, X-linked, 300751 Protoporphyria, erythropoietic, X-linked, 300752
ALB	106.9	100%	97%	Analbuminemia [Dysalbuminemic hyperthyroxinemia] [Dysalbuminemic hyperzincemia], 194470 (1)
ALDH18A1	91.9	97%	91%	Cutis laxa, autosomal recessive, type IIIA, 219150
ALDH1A3	83.5	99%	93%	Microphthalmia, isolated 8, 615113
ALDH2	87.4	98%	93%	Alcohol sensitivity, acute, 610251 {Hangover, susceptibility to}, 610251 {Sublingual nitroglycerin, susceptibility to poor response to} {Esophageal cancer, alcohol-related, susceptibility to}
ALDH3A2	97.3	100%	100%	Sjogren-Larsson syndrome, 270200
ALDH4A1	69.4	94%	89%	Hyperprolinemia, type II, 239510
ALDH5A1	71.8	97%	95%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	100.9	100%	100%	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	64.9	90%	87%	Epilepsy, pyridoxine-dependent, 266100
ALDOA	111.4	98%	94%	Glycogen storage disease XII, 611881
ALDOB	106.5	100%	98%	Fructose intolerance, 229600
ALG1	47.7	45%	45%	Congenital disorder of glycosylation, type Ik, 608540
ALG11	154.3	100%	100%	Congenital disorder of glycosylation, type Ip, 613661
ALG12	103.4	100%	97%	Congenital disorder of glycosylation, type Ig, 607143
ALG13	58.2	93%	85%	Congenital disorder of glycosylation, type Is, 300884
ALG2	115.0	100%	97%	Congenital disorder of glycosylation, type Ii, 607906
ALG3	83.7	100%	93%	Congenital disorder of glycosylation, type Id, 601110

ALG6	104.1	100%	100%	Congenital disorder of glycosylation, type Ic, 603147
ALG8	85.2	96%	95%	Congenital disorder of glycosylation, type Ih, 608104
ALG9	83.5	100%	99%	Congenital disorder of glycosylation, type Il, 608776
ALMS1	195.4	98%	98%	Alstrom syndrome, 203800
ALOX12B	101.5	100%	99%	Ichthyosis, congenital, autosomal recessive 2, 242100
ALOXE3	84.2	100%	96%	Ichthyosis, congenital, autosomal recessive 3, 606545
ALPL	80.7	100%	100%	Hypophosphatasia, infantile, 241500 Hypophosphatasia, childhood, 241510 Odontohypophosphatasia, 146300 Hypophosphatasia, adult, 146300
ALS2	143.8	99%	97%	Amyotrophic lateral sclerosis 2, juvenile, 205100 Primary lateral sclerosis, juvenile, 606353 Spastic paralysis, infantile onset ascending, 607225
ALX1	161.4	100%	100%	Frontonasal dysplasia 3, 613456
ALX3	75.4	85%	80%	Frontonasal dysplasia 1, 136760
ALX4	80.4	100%	98%	Parietal foramina 2, 609597 Frontonasal dysplasia 2, 613451
AMACR	82.8	100%	100%	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
AMELX	62.2	100%	96%	Amelogenesis imperfecta, hypoplastic/hypomaturation type 1E, 301200 -3
AMER1	67.4	100%	98%	Osteopathia striata with cranial sclerosis
AMH	33.2	84%	71%	Persistent Mullerian duct syndrome, type I, 261550
AMHR2	119.0	100%	99%	Persistent Mullerian duct syndrome, type II, 261550
AMN	65.8	89%	85%	Megaloblastic anemia-1, Norwegian type, 261100
AMPD1	106.6	100%	99%	Myoadenylate deaminase deficiency
AMT	127.5	100%	99%	Glycine encephalopathy, 605899
ANG	155.1	100%	99%	Amyotrophic lateral sclerosis 9, 611895
ANGPTL3	128.0	100%	99%	Hypobetalipoproteinemia, familial, 2, 605019
ANK1	95.6	99%	96%	Spherocytosis, type 1, 182900
ANK2	125.4	100%	99%	Long QT syndrome-4, 600919 Cardiac arrhythmia, ankyrin-B-related, 600919
ANKH	110.5	100%	99%	Craniometaphyseal dysplasia, 123000 Chondrocalcinosis 2, 118600
ANKK1	100.2	100%	99%	Dopamine receptor D2, reduced brain density of
ANKRD11	118.3	91%	87%	KBG syndrome, 148050
ANKRD26	119.9	100%	97%	Thrombocytopenia 2, 188000

ANKS6	62.7	96%	85%	Nephronophthisis 16, 615382
ANO10	96.1	99%	97%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANO3	109.4	100%	100%	Dystonia 24, 615034
ANO5	103.4	100%	100%	Gnathodiaphyseal dysplasia, 166260 Muscular dystrophy, limb-girdle, type 2L, 611307 Miyoshi muscular dystrophy 3, 613319
ANO6	92.8	98%	95%	Scott syndrome, 262890
ANTXR1	83.3	97%	91%	{Hemangioma, capillary infantile, susceptibility to}, 602089 GAPO syndrome, 230740
ANTXR2	120.7	100%	100%	Hyaline fibromatosis syndrome, 228600
AP1S1	79.9	100%	99%	MEDNIK syndrome, 609313
AP1S2	95.3	100%	100%	Mental retardation, X-linked syndromic, Fried type, 300630
AP2S1	84.7	100%	100%	Hypocalciuric hypercalcemia, familial, type III, 600740
AP3B1	109.5	100%	99%	Hermansky-Pudlak syndrome 2, 608233
AP4B1	107.5	100%	100%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	136.2	100%	99%	Spastic paraplegia 51, autosomal recessive, 613744
AP4M1	102.6	100%	98%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	76.7	99%	87%	Spastic paraplegia 52, autosomal recessive, 614067
AP5Z1	75.3	96%	90%	Spastic paraplegia 48, autosomal recessive, 613647
APC	150.5	100%	100%	Adenomatous polyposis coli, 175100 Gastric cancer, somatic, 613659 Adenoma, periampullary, somatic Hepatoblastoma, somatic, 114550 Desmoid disease, hereditary, 135290 Colorectal cancer, somatic, 114500 Brain tumor-polyposis syndrom
APCDD1	123.4	100%	99%	Hypotrichosis simplex, 605389
APOA1	80.5	100%	95%	ApoA-I and apoC-III deficiency, combined Hypoalphalipoproteinemia, 604091 Corneal clouding, autosomal recessive Amyloidosis, 3 or more types, 105200
APOA2	83.7	100%	100%	Apolipoprotein A-II deficiency {Hypercholesterolemia, familial, modification of}, 143890
APOA5	129.6	100%	100%	{Hypertriglyceridemia, susceptibility to}, 145750 Hyperchylomicronemia, late-onset, 144650

APOB	162.5	99%	99%	Ag linked Hypobetalipoproteinemia Hypobetalipoproteinemia, normotriglyceridemic Hypercholesterolemia, due to ligand-defective apo B, 144010
APOC2	156.8	100%	100%	Hyperlipoproteinemia, type Ib, 207750
APOC3	99.8	100%	100%	Hyperalphalipoproteinemia 2, 614028
APOE	43.5	82%	73%	{Myocardial infarction, susceptibility to}, 608446
APOE	43.5	82%	73%	Hyperlipoproteinemia, type III {Myocardial infarction susceptibility} Sea-blue histiocyte disease, 269600 Alzheimer disease-2, 104310 {?Macular degeneration, age-related}, 603075 Lipoprotein glomerulopathy, 611771
APP	89.0	100%	99%	Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants, 605714 Alzheimer disease 1, familial, 104300
APRT	47.5	100%	91%	Adenine phosphoribosyltransferase deficiency, 614723
APTX	120.8	96%	94%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
AQP2	87.6	91%	87%	Diabetes insipidus, nephrogenic, 125800
AQP5	103.4	100%	94%	Palmoplantar keratoderma, Bothnian type, 600231
AR	48.5	99%	92%	Androgen insensitivity, 300068 Spinal and bulbar muscular atrophy of Kennedy, 313200 Androgen insensitivity, partial, with or without breast cancer, 312300 {Prostate cancer, susceptibility to}, 176807 Hypospadias 1, X-linked, 300633
ARFGEF2	114.4	100%	99%	Periventricular heterotopia with microcephaly, 608097
ARG1	135.0	98%	91%	Argininemia, 207800
ARHGAP26	118.8	100%	99%	Leukemia, juvenile myelomonocytic, 607785
ARHGAP31	134.5	100%	99%	Adams-Oliver syndrome 1, 100300
ARHGEF10	92.9	98%	94%	Slowed nerve conduction velocity, AD, 608236
ARHGEF12	121.4	99%	99%	Leukemia, acute myeloid, 601626
ARHGEF6	54.8	97%	93%	Mental retardation, X-linked 46, 300436

ARHGEF9	50.1	96%	87%	Epileptic encephalopathy, early infantile, 8, 300607
ARID1A	102.5	99%	95%	Mental retardation, autosomal dominant 14, 614607
ARID1B	122.4	100%	98%	Mental retardation, autosomal dominant 12, 614562
ARL13B	126.0	99%	95%	Joubert syndrome 8, 612291
ARL2BP	85.2	100%	96%	Retinitis pigmentosa with or without situs inversus, 615434
ARL6	160.1	100%	100%	Bardet-Biedl syndrome 3, 209900 {Bardet-Biedl syndrome 1, modifier of}, 209900 Retinitis pigmentosa 55, 613575
ARMC4	97.3	87%	85%	Ciliary dyskinesia, primary, 23, 615451
ARNT	82.0	96%	92%	Leukemia, acute myeloblastic
ARSA	87.1	97%	94%	Metachromatic leukodystrophy, 250100
ARSB	91.1	100%	99%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARSE	49.9	90%	75%	Chondrodysplasia punctata, X-linked recessive, 302950
ARX	31.5	76%	62%	Epileptic encephalopathy, early infantile, 1, 308350 Lissencephaly, X-linked 2, 300215 Mental retardation, X-linked 29 and others, 300419 Proud syndrome, 300004 Partington syndrome, 309510 Hydranencephaly with abnormal genitalia, 30021
ASAH1	99.1	100%	100%	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASB10	73.1	92%	86%	Glaucoma 1, open angle, F, 603383
ASCC1	96.7	93%	88%	Barrett esophagus/esophageal adenocarcinoma, 614266
ASCL1	154.4	100%	100%	Central hypoventilation syndrome, congenital, 209880 Haddad syndrome, 209880
ASL	81.3	99%	96%	Argininosuccinic aciduria, 207900
ASNS	64.5	92%	87%	temperature sensitive G1 mutant
ASPA	112.8	100%	99%	Canavan disease, 271900
ASPM	141.0	100%	99%	Microcephaly 5, primary, autosomal recessive, 608716
ASPSCR1	75.3	97%	93%	Alveolar soft-part sarcoma, 606243
ASS1	40.4	85%	60%	Citrullinemia, 215700
ASXL1	143.4	98%	96%	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ASXL3	157.3	100%	99%	Bainbridge-Ropers syndrome, 615485
ATCAY	104.3	100%	100%	Ataxia, cerebellar, Cayman type, 601238

ATIC	115.4	100%	99%	AICA-ribosiduria due to ATIC deficiency, 608688
ATL1	108.5	100%	99%	Spastic paraplegia 3A, autosomal dominant, 182600 Neuropathy, hereditary sensory, type ID, 613708
ATL3	100.4	99%	97%	Neuropathy, hereditary sensory, type IF, 615632
ATM	118.0	100%	99%	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic {Breast cancer, susceptibility to}, 114480 Lymphoma, mantle cell T-cell prolymphocytic leukemia, somatic
ATN1	119.7	97%	97%	Dentatorubro-pallidoluysian atrophy, 125370
ATP13A2	81.3	97%	91%	Parkinson disease 9, 606693
ATP1A2	100.1	100%	98%	Migraine, familial hemiplegic, 2, 602481 Alternating hemiplegia of childhood, 104290 Migraine, familial basilar, 602481
ATP1A3	110.1	100%	98%	Dystonia-12, 128235 Alternating hemiplegia of childhood 2, 614820
ATP2A1	121.2	100%	99%	Brody myopathy, 601003
ATP2A2	120.5	100%	100%	Darier disease, 124200 Acrokeratosis verruciformis, 101900
ATP2C1	119.6	100%	100%	Hailey-Hailey disease, 169600
ATP5E	159.8	100%	100%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053
ATP6V0A2	106.5	100%	99%	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250
ATP6V0A4	77.6	99%	93%	Renal tubular acidosis, distal, autosomal recessive, 602722
ATP6V1B1	98.0	100%	98%	Renal tubular acidosis with deafness
ATP7A	60.3	100%	97%	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATP7B	125.7	100%	98%	Wilson disease, 277900
ATP8B1	113.5	99%	98%	Cholestasis, progressive familial intrahepatic 1, 211600 Cholestasis, benign recurrent intrahepatic, 243300 Cholestasis, intrahepatic, of pregnancy, 1, 147480
ATPAF2	70.9	100%	98%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273

ATR	119.4	100%	99%	Seckel syndrome 1, 210600 Cutaneous telangiectasia and cancer syndrome, familial, 614564
ATRX	73.1	100%	98%	Alpha-thalassemia/mental retardation syndrome, 301040 Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Mental retardation-hypotonic facies syndrome, X-linked, 309580
ATXN1	89.1	100%	100%	Spinocerebellar ataxia 1, 164400
ATXN10	118.4	100%	100%	Spinocerebellar ataxia 10, 603516
ATXN2	97.6	89%	80%	Spinocerebellar ataxia 2, 183090 {Amyotrophic lateral sclerosis, susceptibility to, 13}, 183090
ATXN3	125.6	100%	100%	Machado-Joseph disease, 109150
ATXN7	129.0	97%	95%	Spinocerebellar ataxia 7, 164500
AUH	114.5	100%	100%	3-methylglutaconic aciduria, type I, 250950
AURKC	102.7	100%	99%	Spermatogenic failure 5
AVP	39.8	93%	68%	Diabetes insipidus, neurohypophyseal, 125700
AVPR2	44.8	95%	89%	Diabetes insipidus, nephrogenic, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539
AXIN1	127.5	97%	92%	Hepatocellular carcinoma, somatic, 114550 Caudal duplication anomaly, 607864
AXIN2	100.1	98%	92%	Oligodontia-colorectal cancer syndrome, 608615 Colorectal cancer, somatic, 114500
B2M	163.4	100%	100%	Hypoproteinemia, hypercatabolic, 241600
B3GALNT2	79.9	90%	87%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies type A 11
B3GALT6	64.0	77%	75%	Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640 Ehlers-Danlos syndrome, progeroid type, 2, 615349
B3GALTL	107.6	95%	95%	Peters-plus syndrome, 261540
B3GAT3	58.8	97%	83%	Multiple joint dislocations, short stature, craniofacial dysmorphism, and congenital heart defects, 245600
B3GNT1	106.0	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
B4GALNT1	80.3	95%	86%	Spastic paraplegia 26, autosomal recessive, 609195
B4GALT1	78.9	100%	100%	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	89.7	100%	95%	Ehlers-Danlos syndrome, progeroid type, 1, 130070
B9D1	83.9	100%	93%	Meckel syndrome 9, 614209
B9D2	49.9	100%	98%	Meckel syndrome 10, 614175

BAAT	123.0	100%	99%	Hypercholanemia, familial, 607748
BAG3	151.0	100%	100%	Myopathy, myofibrillar, 6, 612954 Cardiomyopathy, dilated, 1HH, 613881
BANF1	54.6	86%	54%	Nestor-Guillermo progeria syndrome, 614008
BAP1	88.2	100%	98%	Tumor predisposition syndrome, 614327
BAX	88.3	99%	95%	Colorectal cancer T-cell acute lymphoblastic leukemia
BBS1	114.8	100%	98%	Bardet-Biedl syndrome 1, 209900
BBS10	126.9	100%	100%	Bardet-Biedl syndrome 10, 209900
BBS12	144.1	100%	100%	Bardet-Biedl syndrome 12, 209900
BBS2	115.0	100%	99%	Bardet-Biedl syndrome 2, 209900
BBS4	91.7	95%	91%	Bardet-Biedl syndrome 4, 209900
BBS5	150.6	100%	100%	Bardet-Biedl syndrome 5, 209900
BBS7	130.1	100%	99%	Bardet-Biedl syndrome 7, 209900
BBS9	124.4	100%	100%	Bardet-Biedl syndrome 9
BCAP31	40.7	78%	72%	Deafness, dystonia, and cerebral hypomyelination, 300475 (3)
BCHE	164.2	100%	100%	Apnea, postanesthetic
BCKDHA	102.4	100%	98%	Maple syrup urine disease, type Ia, 248600
BCKDHB	95.9	98%	84%	Maple syrup urine disease, type Ib, 248600
BCKDK	128.2	100%	100%	Branched-chain ketoacid dehydrogenase kinase deficiency, 614923
BCL10	110.6	95%	89%	Lymphoma, MALT, somatic, 137245 {Lymphoma, follicular, somatic}, 613024 {Male germ cell tumor, somatic}, 273300, {Sezary syndrome, somatic}, {Mesothelioma, somatic}, 156240
BCL2	159.4	100%	99%	Leukemia/lymphoma, B-cell, 2
BCL7A	73.1	96%	86%	B-cell non-Hodgkin lymphoma, high-grade
BCO1	131.9	100%	99%	Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300
BCOR	61.1	99%	93%	Microphthalmia, syndromic 2, 300166
BCR	76.7	84%	83%	Leukemia, chronic myeloid, 608232 Leukemia, acute lymphocytic, 613065
BCS1L	148.6	100%	100%	Mitochondrial complex III deficiency, nuclear type 1, 124000 Leigh syndrome, 256000 Bjornstad syndrome, 262000 GRACILE syndrome, 603358

BDNF	189.2	100%	96%	{Memory impairment, susceptibility to} Central hypoventilation syndrome, congenital, 209880 {Obsessive-compulsive disorder, protection against}, 164230 {Bulimia nervosa, age of onset of weight loss in},607499 {Anorexia nervosa, susceptibi
BEAN1	79.0	100%	100%	Spinocerebellar ataxia 31
BEST1	106.5	100%	97%	Best macular dystrophy, 153700 Maculopathy, bull's-eye Vitelliform macular dystrophy, adult-onset, 608161 Bestrophinopathy, 611809 Vitreoretinochoroidopathy, 193220 Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma, 1
BFSP1	124.0	100%	100%	Cataract 33, 611391
BFSP2	62.7	94%	92%	Cataract 12, multiple types, 611597
BICD2	93.3	99%	96%	Spinal muscular atrophy, lower extremity-predominant, 2, AD, 615290 -3
BIN1	54.9	85%	75%	Myopathy, centronuclear, autosomal recessive, 255200
BLK	103.2	100%	99%	Maturity-onset diabetes of the young, type 11, 613375
BLM	118.7	100%	100%	Bloom syndrome
BLNK	102.7	100%	100%	Agammaglobulinemia 4, 613502
BLOC1S3	31.0	79%	68%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	117.5	99%	92%	Hermansky-pudlak syndrome 9, 614171
BLVRA	85.8	100%	99%	Hyperbiliverdinemia, 614156
BMP1	98.8	97%	95%	Osteogenesis imperfecta, type XIII, 614856
BMP15	72.2	100%	100%	Ovarian dysgenesis 2, 300510 Premature ovarian failure 4, 300510
BMP2	115.8	100%	100%	{HFE hemochromatosis, modifier of}, 235200 Brachydactyly, type A2, 112600
BMP4	114.2	100%	100%	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625 -3
BMPER	122.1	97%	97%	Diaphanospondylodysostosis, 608022
BMPR1A	59.0	78%	63%	Polyposis, juvenile intestinal, 174900 Polyposis syndrome, hereditary mixed, 2, 610069 Juvenile polyposis syndrome, infantile form, 174900
BMPR1B	104.1	100%	96%	Brachydactyly, type A2, 112600 Chrondrodysplasia, acromesomelic, with genital anomalies, 609441

BMPR2	150.9	100%	99%	Pulmonary hypertension, familial primary, 1, with or without HHT, 178600 Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600 Pulmonary venoocclusive disease, 265450
BOLA3	54.7	100%	99%	Multiple mitochondrial dysfunctions syndrome 2, 614299
BPGM	148.3	100%	100%	Erythrocytosis due to bisphosphoglycerate mutase deficiency, 222800
BRAF	78.5	100%	99%	Melanoma, malignant, somatic Colorectal cancer, somatic Adenocarcinoma of lung, somatic, 211980 Nonsmall cell lung cancer, somatic Cardiofaciocutaneous syndrome, 115150 Noonan syndrome 7, 613706 LEOPARD syndrome 3, 613707
BRAT1	76.2	100%	98%	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BRCA2	151.7	100%	100%	{Breast-ovarian cancer, familial, 2}, 612555 Fanconi anemia, complementation group D1, 605724 Prostate cancer, 176807 {Breast cancer, male, susceptibility to}, 114480 Wilms tumor, 194070 {Medulloblastoma}, 155255 {Glioblastoma 3},
BRIP1	130.4	100%	100%	Breast cancer early-onset
BRWD3	62.9	98%	96%	Mental retardation, X-linked 93, 300659
BSCL2	110.4	100%	100%	Lipodystrophy, congenital generalized, type 2, 269700 Silver spastic paraplegia syndrome, 270685 Neuropathy, distal hereditary motor, type V, 600794
BSND	116.3	100%	98%	Bartter syndrome, type 4a, 602522 Sensorineural deafness with mild renal dysfunction, 602522
BTD	141.7	100%	100%	Biotinidase deficiency, 253260
BTK	53.4	99%	95%	Agammaglobulinemia, X-linked 1, 300755 Agammaglobulinemia and isolated hormone deficiency, 307200
BUB1	118.0	98%	94%	Colorectal cancer with chromosomal instability
BUB1B	117.8	100%	98%	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430
C10orf11	91.1	100%	100%	Albinism, oculocutaneous, type VII, 615179

C10orf2	138.3	100%	100%	Progressive external ophthalmoplegia, autosomal dominant, 3, 609286 Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245
C12orf57	77.6	100%	96%	Temtamy syndrome, 218340
C12orf65	174.3	100%	100%	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035
C15orf41	85.9	87%	82%	Dyserythropoietic anemia, congenital, type Ib, 615631 (3)
C19orf12	67.0	100%	93%	Neurodegeneration with brain iron accumulation 4, 614298
C1GALT1C1	86.6	100%	99%	Tn polyagglutination syndrome, somatic, 300622
C1QA	126.3	100%	98%	C1q deficiency, 613652
C1QB	101.5	96%	86%	C1q deficiency, 613652
C1QC	137.1	94%	80%	C1q deficiency, 613652
C1QTNF5	101.7	98%	91%	Retinal degeneration, late-onset, autosomal dominant, 605670
C1S	105.2	100%	99%	C1s deficiency, 613783
C2	19.2	80%	46%	C2 deficiency, 217000 {Macular degeneration, age-related, reduced risk of}, 603075
C21orf59	98.1	100%	97%	Ciliary dyskinesia, primary, 26, 615500 (3)
C2orf71	105.5	99%	96%	Retinitis pigmentosa 54, 613428
C3	99.1	99%	95%	C3 deficiency, 613779 {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 {Macular degeneration, age-related, 9}, 611378
C4A	1.2	3%	2%	C4a deficiency, 614380 [Blood group, Rodgers], 614374 ?Systemic lupus erythematosus, susceptibility to or protection against}, 152700 (2)
C4B	1.4	4%	2%	C4B deficiency, 614379
C4orf26	162.2	100%	100%	Amelogenesis imperfecta, hypomaturation type, IIA4, 614832
C5	100.4	100%	99%	C5 deficiency, 609536
C5orf42	129.8	100%	99%	Joubert syndrome 17, 614615
C6	114.8	100%	99%	C6 deficiency, 612446 Combined C6/C7 deficiency
C7	95.3	98%	93%	C7 deficiency, 610102
C8A	88.6	100%	99%	C8 deficiency, type I, 613790
C8B	96.3	100%	97%	C8 deficiency, type II, 613789
C8orf37	95.2	100%	100%	Retinitis pigmentosa 64, 614500 Cone-rod dystrophy 16, 614500 -3

C9	112.3	100%	100%	C9 deficiency Macular degeneration, age-related, 15, susceptibility to
C9orf72	86.8	100%	100%	Amyotrophic lateral sclerosis and/or frontotemporal dementia, 105550 -3
CA12	74.1	100%	100%	Hyperchlorhidrosis, isolated, 143860
CA2	143.7	100%	100%	Osteopetrosis autosomal recessive 3 with renal tubular acidosis
CA4	83.8	98%	92%	Retinitis pigmentosa 17, 600852
CA5A	28.4	43%	36%	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751 (3)
CA8	83.5	100%	100%	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CABP2	47.4	84%	72%	Deafness, autosomal recessive 93, 614899
CABP4	64.3	100%	100%	Night blindness, congenital stationary (incomplete), 2B, autosomal recessive, 610427
CACNA1A	80.9	98%	90%	Migraine, familial hemiplegic, 1, 141500 Episodic ataxia, type 2, 108500 Spinocerebellar ataxia 6, 183086 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500
CACNA1C	97.3	98%	95%	Timothy syndrome, 601005 Brugada syndrome 3, 611875
CACNA1D	115.7	100%	98%	Sinoatrial node dysfunction and deafness, 614896
CACNA1F	42.5	94%	85%	Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071 Cone-rod dystropy, X-linked, 3, 300476 Aland Island eye disease, 300600
CACNA1S	96.0	100%	99%	Hypokalemic periodic paralysis, type 1, 170400 {Malignant hyperthermia susceptibility 5}, 601887 {Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580
CACNA2D4	78.1	98%	94%	Retinal cone dystrophy 4, 610478
CACNB2	127.4	100%	100%	Brugada syndrome 4, 611876
CACNB4	92.9	100%	93%	{Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682 {Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 Episodic ataxia, type 5, 613855
CACNG2	96.4	100%	100%	Mental retardation, autosomal dominant 10, 614256
CALM1	114.1	100%	100%	Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916 -3
CALR	140.5	96%	90%	distal to C3, near LDLR
CALR3	96.1	100%	99%	Cardiomyopathy, familial hypertrophic, 19, 613875

CAMTA1	124.8	97%	97%	Cerebellar ataxia, nonprogressive, with mental retardation, 614756 -3
CANT1	93.1	99%	97%	Desbuquois dysplasia, 251450
CAPN3	115.4	99%	97%	Muscular dystrophy, limb-girdle, type 2A, 253600
CAPN5	78.6	100%	95%	Vitreoretinopathy, neovascular inflammatory, 193235
CARD11	97.9	100%	98%	Persistent polyclonal B-cell lymphocytosis, 606445 Immunodeficiency 11, 615206
CARD14	62.6	97%	89%	{Psoriasis susceptibility 2}, 602723 Pityriasis rubra pilaris, 173200
CARD9	60.9	100%	97%	Candidiasis, familial, 2, autosomal recessive, 212050
CASC5	145.0	98%	98%	Microcephaly 4, primary, autosomal recessive, 604321
CASK	51.6	98%	94%	Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 FG syndrome 4, 300422 Mental retardation, with or without nystagmus, 300422
CASP10	110.7	100%	100%	Autoimmune lymphoproliferative syndrome, type II, 603909 Non-Hodgkin lymphoma, somatic, 605027 Gastric cancer, somatic, 613659
CASP8	118.6	100%	98%	Immunodeficiency due to CASP8 deficiency, 607271 Hepatocellular carcinoma, somatic, 114550 {Breast cancer, protection against}, 114480 {Lung cancer, protection against}, 211980
CASQ2	87.8	100%	96%	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CASR	118.8	100%	99%	Hypocalciuric hypercalcemia, type I, 145980 Hyperparathyroidism, neonatal, 239200 Hypocalcemia, autosomal dominant, 601198 Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 {Epilepsy idiopathic generalized, susceptibility to,
CAT	91.7	98%	91%	Acatalasemia, 614097
CATSPER1	102.9	99%	98%	Spermatogenic failure 7, 612997
CAV1	163.1	100%	100%	Lipodystrophy, congenital generalized, type 3, 612526 Pulmonary hypertension, primary, 3, 615343
CAV3	146.7	100%	100%	Muscular dystrophy, limb-girdle, type IC, 607801 Rippling muscle disease, 606072 Creatine phosphokinase, elevated serum, 123320 Myopathy, distal, Tateyama type, 614321 Cardiomyopathy, familial hypertrophic, 192600

				Long QT syndrome-9, 6
CBL	127.5	100%	99%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563
CBS	73.6	98%	89%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CBX2	135.4	100%	99%	46XY sex reversal 5, 613080
CC2D1A	96.2	100%	97%	Mental retardation, autosomal recessive 3, 608443
CC2D2A	92.9	98%	97%	Joubert syndrome 9, 612285 Meckel syndrome 6, 612284 COACH syndrome, 216360
CCBE1	93.2	97%	89%	Hennekam lymphangiectasia-lymphedema syndrome, 235510
CCDC103	106.0	100%	100%	Ciliary dyskinesia, primary, 17, 614679
CCDC114	77.5	100%	98%	Ciliary dyskinesia, primary, 20, 615067
CCDC39	104.7	100%	100%	Ciliary dyskinesia, primary, 14, 613807
CCDC40	84.4	97%	93%	Ciliary dyskinesia, primary, 15, 613808
CCDC50	131.9	99%	96%	Deafness, autosomal dominant 44, 607453
CCDC65	74.5	100%	96%	Ciliary dyskinesia, primary, 27, 615504
CCDC78	90.8	100%	100%	Myopathy, centronuclear, 4, 614807
CCDC8	130.3	100%	100%	Three M syndrome 3, 614205
CCDC88C	90.9	99%	96%	Hydrocephalus, nonsyndromic, autosomal recessive, 236600
CCM2	96.5	95%	92%	Cerebral cavernous malformations-2
CCT5	80.0	94%	81%	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CD151	87.3	100%	96%	Nephropathy with pretibial epidermolysis bullosa and deafness, 609057 [Blood group, Raph], 179620
CD19	78.8	100%	98%	Immunodeficiency, common variable, 3, 613493
CD247	87.8	100%	100%	Immunodeficiency due to defect in CD3-zeta, 610163
CD27	80.9	100%	98%	Lymphoproliferative syndrome 2
CD2AP	116.4	100%	99%	Glomerulosclerosis, focal segmental, 3, 607832
CD320	79.5	97%	88%	Methylmalonic aciduria due to transcobalamin receptor defect, 613646

CD36	138.8	100%	100%	[Macrothrombocytopenia] (1) Platelet glycoprotein IV deficiency, 608404 {Malaria, cerebral, susceptibility to}, 611162 {Malaria, cerebral, reduced risk of}, 611162 {Coronary heart disease, susceptibility to, 7}, 610938
CD3D	83.6	100%	99%	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971
CD3E	93.8	95%	86%	Immunodeficiency due to defect in CD3-epsilon Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971
CD3G	87.9	100%	100%	Immunodeficiency due to defect in CD3-gamma
CD4	84.0	97%	96%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
CD4	84.0	97%	96%	OKT4 epitope deficiency, 613949
CD40	103.3	95%	91%	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	63.0	91%	83%	Immunodeficiency X-linked with hyper-IgM
CD59	115.7	100%	100%	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD79A	75.4	98%	85%	Agammaglobulinemia 3, 613501
CD79B	126.4	100%	99%	Agammaglobulinemia 6, 612692
CD81	61.1	97%	87%	Immunodeficiency, common variable, 6, 613496
CD8A	82.1	95%	93%	CD8 deficiency, familial, 608957
CD96	119.4	100%	100%	C syndrome, 211750
CDAN1	91.2	99%	96%	Anemia, congenital dyserythropoietic, type I, 224120
CDC6	103.5	100%	99%	Meier-Gorlin syndrome 5, 613805
CDC73	150.1	100%	100%	Hyperparathyroidism, familial primary, 145000 Hyperparathyroidism-jaw tumor syndrome, 145001 Parathyroid adenoma with cystic changes, 145001 Parathyroid carcinoma, 608266
CDH1	117.6	100%	99%	Endometrial carcinoma, somatic, 608089 Ovarian carcinoma, somatic, 167000 {Breast cancer, lobular}, 114480 Gastric cancer, familial diffuse, with or without cleft lip and/or palate, 137215 {Prostate cancer, susceptibility to}, 176807
CDH15	80.2	100%	98%	Mental retardation, autosomal dominant 3, 612580

CDH23	93.7	99%	98%	Usher syndrome, type 1D, 601067 Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D/F digenic, 601067
CDH3	96.9	97%	92%	i Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280
CDHR1	110.9	98%	98%	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660 -3
CDK5RAP2	109.5	99%	95%	Microcephaly 3, primary, autosomal recessive, 604804
CDKL5	65.8	98%	95%	Epileptic encephalopathy, early infantile, 2, 300672 Angelman syndrome-like, 105830
CDKN1B	130.0	100%	100%	Multiple endocrine neoplasia, type IV, 610755
CDKN1C	39.5	91%	82%	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732
CDKN2A	139.7	100%	100%	{Melanoma, cutaneous malignant, 2}, 155601 Melanoma and neural system tumor syndrome, 155755 Pancreatic cancer/melanoma syndrome, 606719 Orolaryngeal cancer, multiple, -3
CDON	117.2	100%	98%	Holoprosencephaly 11, 614226
CDSN	17.0	81%	39%	Hypotrichosis simplex of scalp 1, 146520 Peeling skin syndrome, 270300
CDT1	44.7	91%	73%	Meier-Gorlin syndrome 4, 613804
CEACAM16	92.3	97%	90%	Deafness, autosomal dominant 4B, 614614
CEBPA	43.1	99%	70%	Leukemia, acute myeloid, 601626
CEBPE	101.1	100%	100%	Specific granule deficiency, 245480
CECR1	89.9	98%	97%	Polyarteritis nodosa, 615688 (3)
CEL	57.3	64%	61%	Maturity-onset diabetes of the young, type VIII, 609812
CENPJ	139.2	100%	100%	Microcephaly 6, primary, autosomal recessive, 608393 Seckel syndrome 4, 613676
CEP135	121.5	100%	99%	Microcephaly 8, primary, autosomal recessive, 614673
CEP152	131.9	99%	99%	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823
CEP164	76.6	98%	92%	Nephronophthisis 15, 614845
CEP19	165.5	100%	100%	Morbid obesity and spermatogenic failure, 615703 (3)

CEP290	101.5	100%	98%	Joubert syndrome 5, 610188 Senior-Loken syndrome 6, 610189 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Bardet-Biedl syndrome 14, 209900
CEP41	88.3	100%	99%	Joubert syndrome 15, 614464
CEP57	88.6	100%	97%	Mosaic variegated aneuploidy syndrome 2, 614114
CERKL	137.2	100%	100%	Retinitis pigmentosa 26, 608380
CERS3	83.6	100%	100%	Ichthyosis, congenital, autosomal recessive 9, 615023
CES1	59.0	60%	57%	Carboxylesterase 1 deficiency
CETP	101.0	100%	100%	Hyperalphalipoproteinemia, 143470 [High density lipoprotein cholesterol level QTL 10], 143470
CFAP53	183.1	99%	98%	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFC1	1.0	%	%	Heterotaxy, visceral, 2, autosomal, 605376 Double-outlet right ventricle, 217095 Transposition of the great arteries, dextro-looped 2, 613853
CFD	50.3	96%	86%	Complement factor D deficiency, 613912
CFH	107.3	95%	92%	Basal laminar drusen
CFHR5	107.7	94%	92%	Nephropathy due to CFHR5 deficiency, 614809
CFI	137.0	100%	100%	Complement factor I deficiency, 610984 {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 {Macular degeneration, age-related, 13, susceptibility to}, 615439
CFL2	133.6	100%	100%	Nemaline myopathy 7, autosomal recessive, 610687
CFP	48.6	97%	85%	Properdin deficiency X-linked
CFTR	123.1	95%	95%	Cystic fibrosis, 219700 Congenital bilateral absence of vas deferens, 277180 Sweat chloride elevation without CF {Pancreatitis, idiopathic}, 167800 {Hypertrypsinemia, neonatal} {Bronchiectasis with or without elevated sweat chloride 1,
CHAT	69.3	91%	77%	Myasthenic syndrome, congenital, associated with episodic apnea, 254210
CHD2	128.0	99%	98%	Epileptic encephalopathy, childhood-onset, 615369
CHD7	121.0	100%	99%	CHARGE syndrome, 214800 {Scoliosis, idiopathic 3}, 608765 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370

CHEK2	55.5	70%	65%	Li-Fraumeni syndrome, 609265 Osteosarcoma, somatic, 259500 {Breast cancer, susceptibility to}, 114480 {Prostate cancer, familial, susceptibility to}, 176807 {Breast and colorectal cancer, susceptibility to}
CHKB	91.1	93%	91%	Muscular dystrophy, congenital, megaconial type, 602541
CHM	52.7	98%	91%	Choroideremia, 303100
CHMP1A	81.5	95%	89%	Pontocerebellar hypoplasia, type 8, 614961
CHMP2B	124.5	100%	100%	Dementia, familial, nonspecific, 600795 Amyotrophic lateral sclerosis 17, 614696
CHMP4B	114.9	100%	100%	Cataract 31, multiple types, 605387
CHN1	140.2	100%	96%	Duane retraction syndrome 2, 604356
CHRDL1	63.2	99%	94%	Megalocornea 1, X-linked 309300
CHRM3	161.3	100%	100%	Eagle-Barrett syndrome, 100100
CHRNA1	101.6	100%	98%	Myasthenic syndrome, slow-channel congenital, 601462 Myasthenic syndrome, fast-channel congenital, 608930 Multiple pterygium syndrome, lethal type, 253290
CHRNA2	115.4	100%	100%	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA4	93.2	99%	98%	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction, susceptibility to}, 188890
CHRNB1	102.5	98%	94%	Myasthenic syndrome, slow-channel congenital, 601462 Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931
CHRNB2	142.5	95%	93%	Epilepsy, nocturnal frontal lobe, 3, 605375
CHRND	94.6	97%	91%	Myasthenic syndrome, slow-channel congenital, 601462 Myasthenic syndrome, fast-channel congenital, 608930 Multiple pterygium syndrome, lethal type, 253290
CHRNE	158.8	100%	100%	Myasthenic syndrome, slow-channel congenital, 601462 Myasthenic syndrome, fast-channel congenital, 608930 Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931
CHRNG	95.7	100%	97%	Myasthenia gravis, neonatal transient (2) Escobar syndrome, 265000 Multiple pterygium syndrome, lethal type, 253290
CHST14	130.2	100%	100%	Ehlers-Danlos syndrome, musculocontractural type , 601776
CHST3	67.2	100%	100%	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHST6	123.3	100%	100%	Macular corneal dystrophy, 217800

CHSY1	147.3	98%	97%	Temtamy preaxial brachydactyly syndrome, 605282
CHUK	92.4	100%	99%	Cocoon syndrome, 613630
CIB2	103.6	100%	100%	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CIITA	99.1	98%	96%	Bare lymphocyte syndrome type II complementation group A
CIRH1A	107.2	100%	100%	Cirrhosis, North American Indian childhood type, 604901
CISD2	190.3	77%	77%	Wolfram syndrome 2, 604928
CITED2	108.4	99%	97%	Ventricular septal defect 2, 614431 Atrial septal defect 8, 614433
CLCF1	43.5	81%	75%	Cold-induced sweating syndrome 1, 610313
CLCN1	91.2	100%	97%	Myotonia congenita, recessive, 255700 Myotonia congenita, dominant, 160800 Myotonia levior, recessive
CLCN2	107.0	100%	99%	{Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628 {Epilepsy, juvenile absence, susceptibility to, 2}, 607628 {Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628
CLCN5	84.2	99%	94%	Dent disease, 300009 Nephrolithiasis, type I, 310468 Hypophosphatemic rickets, 300554 Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990
CLCN7	80.7	100%	98%	Osteopetrosis, autosomal recessive 4, 611490 Osteopetrosis, autosomal dominant 2, 166600
CLCNKA	87.2	83%	79%	Bartter syndrome, type 4b, digenic, 613090
CLCNKB	74.1	86%	80%	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090
CLDN1	104.5	100%	100%	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626
CLDN14	66.2	100%	99%	Deafness, autosomal recessive 29, 614035
CLDN16	126.8	98%	95%	Hypomagnesemia 3, renal, 248250
CLDN19	69.3	100%	93%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLEC7A	103.1	100%	100%	Candidiasis, familial, 4, autosomal recessive, 613108 {Aspergillosis, susceptibility to}, 614079
CLIC2	33.1	92%	63%	Mental retardation, X-linked, syndromic 32, 300886
CLK1	151.4	100%	99%	3MC syndrome 2
CLMP	99.8	97%	97%	Congenital short bowel syndrome, 615237
CLN3	86.5	100%	99%	Ceroid lipofuscinosis, neuronal, 3, 204200

CLN5	139.7	100%	90%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	63.7	98%	81%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	133.8	100%	100%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLPP	90.1	97%	91%	Perrault syndrome 3, 614129
CLRN1	165.5	100%	100%	Usher syndrome, type 3A, 276902 Retinitis pigmentosa 61, 614180 -3
CNBP	106.4	100%	95%	Myotonic dystrophy 2
CNGA1	119.8	91%	90%	Retinitis pigmentosa 49, 613756
CNGA3	146.7	100%	99%	Achromatopsia-2, 216900
CNGB1	84.8	96%	90%	Retinitis pigmentosa 45, 613767
CNGB3	108.7	100%	98%	Achromatopsia-3, 262300 Macular degeneration, juvenile, 248200 -3
CNNM2	136.5	100%	99%	Hypomagnesemia 6, renal, 613882
CNNM4	170.4	99%	96%	Jalili syndrome, 217080
CNTN1	109.4	100%	99%	Myopathy, congenital, Compton-North, 612540
CNTNAP2	100.2	100%	99%	Cortical dysplasia-focal epilepsy syndrome, 610042 {Autism susceptibility 15}, 612100 Pitt-Hopkins like syndrome 1, 610042
COA5	84.9	99%	96%	Mitochondrial complex IV deficiency, 220110
COASY	123.7	100%	100%	Neurodegeneration with brain iron accumulation 6, 615643
COCH	119.2	100%	98%	Deafness, autosomal dominant 9, 601369
COG1	121.0	100%	98%	Congenital disorder of glycosylation, type IIg, 611209
COG4	86.4	98%	95%	Congenital disorder of glycosylation, type IIj, 613489
COG5	101.6	97%	94%	Congenital disorder of glycosylation, type IIi, 613612
COG6	101.9	97%	95%	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328
COG7	77.0	100%	96%	Congenital disorder of glycosylation, type IIe, 608779
COG8	116.5	100%	100%	Congenital disorder of glycosylation, type IIh, 611182
COL10A1	93.3	100%	99%	Metaphyseal chondrodysplasia, Schmid type, 156500
COL11A1	99.4	98%	97%	Stickler syndrome, type II, 604841 Marshall syndrome, 154780 {Lumbar disc herniation, susceptibility to}, 603932 Fibrochondrogenesis, 228520

COL11A2	14.4	61%	17%	Stickler syndrome, type III, 184840 Otospondylomegaepiphyseal dysplasia, 215150 Weissenbacher-Zweymuller syndrome, 277610 Deafness, autosomal dominant 13, 601868 Deafness, autosomal recessive 53, 609706 Fibrochondrogenesis 2, 614524
COL17A1	82.4	97%	89%	Epidermolysis bullosa, junctional, non-Herlitz type, 226650
COL18A1	86.0	96%	90%	Knobloch syndrome, type 1, 267750
COL1A1	112.0	99%	97%	Osteogenesis imperfecta, type I, 166200 OI type II, 166210 OI type III, 259420 OI type IV, 166220 Ehlers-Danlos syndrome, type I, 130000 Ehlers-Danlos syndrome, type VIIA, 130060 {Osteoporosis}, 166710 Caffey disease, 114000
COL1A2	93.1	99%	96%	Ehlers-Danlos syndrome, type VIIB, 130060 Osteogenesis imperfecta, type IV, 166220 Osteogenesis imperfecta, type III, 259420 Osteogenesis imperfecta, type II, 166210 {Osteoporosis, postmenopausal}, 166710 Ehlers-Danlos syndrome, cardi
COL2A1	85.2	99%	95%	Stickler syndrome, type I, 108300 Kniest dysplasia, 156550 Achondrogenesis, type II or hypochondrogenesis, 200610 SED congenita, 183900 SMED Strudwick type, 184250 Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 Sp
COL3A1	72.7	97%	95%	Ehlers-Danlos syndrome, type IV, 130050 Ehlers-Danlos syndrome, type III, 130020
COL4A1	87.4	99%	96%	Porencephaly 1, 175780 Brain small vessel disease with hemorrhage, 607595 Angiopathy, hereditary, with nephropathy, aneurysms, and muscle, 611773 Brain small vessel disease with Axenfeld-Rieger anomaly, 607595 {Hemorrhage, intracerebral, s

COL4A2	80.1	99%	97%	Porencephaly 2, 614483 {Hemorrhage, intracerebral, susceptibility to}, 614519
COL4A3	74.2	97%	94%	Alport syndrome, autosomal recessive, 203780 Hematuria, benign familial, 141200 Alport syndrome, autosomal dominant, 104200
COL4A4	92.0	100%	97%	Alport syndrome, autosomal recessive, 203780 Hematuria,familial benign
COL4A5	38.2	94%	78%	Alport syndrome, 301050
COL5A1	97.1	98%	95%	Ehlers-Danlos syndrome, type II, 130010 Ehlers-Danlos syndrome, type I, 130000
COL5A2	85.0	97%	92%	Ehlers-Danlos syndrome, type I, 130000
COL6A1	81.8	98%	96%	Bethlem myopathy, 158810 Ullrich congenital muscular dystrophy, 254090 {Ossification of the posterior longitudinal spinal ligaments}, 602475 (2)
COL6A2	82.7	99%	97%	Bethlem myopathy, 158810 Ullrich congenital muscular dystrophy, 254090 Myosclerosis, congenital, 255600
COL6A3	123.2	99%	99%	Bethlem myopathy, 158810 Ullrich congenital muscular dystrophy, 254090
COL7A1	101.1	100%	98%	Epidermolysis bullosa dystrophica, AD, 131750 Epidermolysis bullosa dystrophica, AR, 226600 Epidermolysis bullosa, pretibial, 131850 EBD, Bart type, 132000 EBD, localisata variant Transient bullous of the newborn, 131705 Epidermoly
COL8A2	62.8	97%	94%	Corneal dystrophy, Fuchs endothelial, 1, 136800 Corneal dystrophy polymorphous posterior, 2, 609140
COL9A1	108.1	100%	96%	Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	72.9	98%	93%	Epiphyseal dysplasia, multiple, 2, 600204 {Intervertebral disc disease, susceptibility to}, 603932 Stickler syndrome, type V, 614284
COL9A3	70.4	99%	89%	Epiphyseal dysplasia, multiple, 3, 600969 Epiphyseal dysplasia, multiple, with myopathy {Intervertebral disc disease, susceptibility to}, 603932
COLQ	73.9	100%	90%	Endplate acetylcholinesterase deficiency, 603034

COMP	86.2	100%	98%	Pseudoachondroplasia, 177170 Epiphyseal dysplasia, multiple 1, 132400
COQ2	75.5	99%	96%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ6	109.2	99%	95%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ9	86.6	91%	83%	Coenzyme Q10 deficiency, primary, 5, 614654
CORIN	114.4	100%	99%	Preeclampsia/eclampsia 5, 614595
CORO1A	88.6	85%	84%	Immunodeficiency 8, 615401
COX10	130.3	100%	97%	Encephalopathy, progressive mitochondrial, with proximal renal tubulopathy due to cytochrome c oxidase deficiency
COX14	127.5	100%	100%	Mitochondrial complex IV deficiency, 220110
COX15	75.0	100%	93%	Leigh syndrome due to cytochrome c oxidase deficiency, 256000 Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119
COX20	59.4	88%	88%	Mitochondrial complex IV deficiency, 220110
COX4I2	56.1	99%	89%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX7B	32.8	76%	67%	Aplasia cutis congenita, reticulolinear, with mmicrocephaly, facial dysmorphism and other congenital anomalies, 300887
CP	90.4	99%	92%	[Hypoceruloplasminemia, hereditary], 604290 Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290
CPA6	122.8	100%	100%	Epilepsy, familial temporal lobe, 5, 614417 Febrile seizures, familial, 11, 614418
CPN1	77.9	100%	99%	Carboxypeptidase N deficiency, 212070
CPOX	85.2	100%	97%	Coproporphyria, 121300 Harderoporphyria, 121300
CPS1	107.5	100%	99%	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}
CPT1A	96.1	100%	98%	CPT deficiency, hepatic, type IA, 255120
CPT2	109.4	92%	91%	Myopathy due to CPT II deficiency, 255110 CPT deficiency, hepatic, type II, 600649 CPT II deficiency, lethal neonatal, 608836 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212
CR2	119.6	100%	100%	{Systemic lupus erythematosus, susceptibility to, 9}, 610927 Immunodeficiency, common variable, 7, 614699

CRADD	128.0	100%	98%	Mental retardation, autosomal recessive 34, 614499
CRB1	165.3	100%	100%	Retinitis pigmentosa-12, autosomal recessive, 600105 Leber congenital amaurosis 8, 613835 Pigmented paravenous chorioretinal atrophy, 172870
CRBN	145.0	100%	100%	Mental retardation, autosomal recessive 2, 607417
CREB1	97.2	100%	100%	Histiocytoma, angiomatoid fibrous, somatic, 612160
CREBBP	83.1	99%	97%	Rubinstein-Taybi syndrome, 180849
CRELD1	83.7	100%	93%	{Atrioventricular septal defect, susceptibility to, 2}, 606217 Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217
CRLF1	56.0	90%	82%	Cold-induced sweating syndrome, 272430
CRTAP	98.4	100%	100%	Osteogenesis imperfecta, type VII, 610682
CRTC1	89.7	97%	91%	Mucoepidermoid salivary gland carcinoma
CRX	152.8	100%	100%	Cone-rod retinal dystrophy-2, 120970 Leber congenital amaurosis 7, 613829
CRYAA	108.4	100%	100%	Cataract 9, multiple types, 604219
CRYAB	133.3	100%	100%	Myopathy, myofibrillar, 2, 608810 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, fatal infantile hypertrophy, alpha-B crystallin-related, 613869 Cardiomyopathy, dilated, 1II, 615184
CRYBA1	91.0	100%	100%	Cataract 10, multiple types, 600881
CRYBA4	79.0	100%	100%	Cataract 23, 610425
CRYBB1	56.9	100%	94%	Cataract 17, multiple types, 611544
CRYBB2	111.3	100%	100%	Cataract 3, multiple types, 601547
CRYBB3	110.5	100%	100%	Cataract 22, autosomal recessive, 609741
CRYGB	69.4	100%	93%	Cataract 39, multiple types, autosomal dominant, 615188
CRYGC	84.1	100%	96%	Cataract 2, multiple types, 604307
CRYGD	76.2	90%	77%	Cataract 4, multiple types, 115700
CRYGS	92.6	98%	90%	Cataract 20, multiple types, 116100
CRYM	75.1	100%	99%	Deafness, autosomal dominant 40
CSF1R	79.2	99%	95%	Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
CSF2RA	.0	%	%	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF2RB	102.4	100%	98%	Surfactant metabolism dysfunction, pulmonary, 5, 614370
CSF3R	81.0	100%	98%	Neutrophilia, hereditary, 162830
CSNK1D	97.8	88%	85%	Advanced sleep-phase syndrome, familial, 2, 615224
CSPP1	121.7	100%	99%	Joubert syndrome 21, 615636

CSRP3	107.3	100%	95%	Cardiomyopathy, dilated, 1M, 607482 Cardiomyopathy, familial hypertrophic, 12, 612124
CST3	56.2	100%	99%	Cerebral amyloid angiopathy, 105150 Macular degeneration, age-related, 11, 611953
CSTA	105.8	100%	100%	Exfoliative ichthyosis, autosomal recessive, ichthyosis bullosa of Siemens-like, 607936
CSTB	166.9	100%	99%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTC1	95.5	100%	99%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTCF	113.2	100%	98%	Mental retardation, autosomal dominant 21, 615502
CTDP1	69.8	89%	87%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTH	130.2	100%	100%	Cystathioninuria, 219500 Homocysteine, total plasma, elevated -3
CTHRC1	87.2	100%	92%	Barrett esophagus/esophageal adenocarcinoma, 614266
CTNNA3	116.2	98%	97%	Arrhythmogenic right ventricular dysplasia, familial, 13, 615616
CTNNB1	113.6	100%	99%	Mental retardation, autosomal dominant 19, 615075 Colorectal cancer, somatic, 114500 Pilomatricoma, somatic, 132600 Ovarian cancer, somatic, 167000 Hepatocellular carcinoma, somatic, 114550
CTNS	114.9	97%	94%	? Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, atypical nephropathic, 219800 -3
CTSA	92.8	100%	98%	Galactosialidosis, 256540
CTSC	86.4	100%	100%	Papillon-Lefevre syndrome, 245000 Haim-Munk syndrome, 245010 Periodontitis 1, juvenile, 170650
CTSD	96.2	100%	100%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	109.8	96%	82%	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362
CTSK	129.9	100%	100%	Pycnodysostosis, 265800
CUBN	88.4	99%	96%	Megaloblastic anemia-1, Finnish type, 261100
CUL3	103.8	99%	96%	Pseudohypoaldosteronism, type IIE, 614496
CUL4B	61.8	100%	92%	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
CUL7	99.0	100%	99%	3-M syndrome 1, 273750
CXCR4	226.9	100%	100%	WHIM syndrome, 193670 Myelokathexis, isolated
CYB5A	48.4	100%	94%	Methemoglobinemia, type IV, 250790

CYB5R3	77.9	97%	94%	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYBA	43.3	93%	84%	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690
CYBB	53.3	88%	81%	Chronic granulomatous disease, X-linked, 306400 Atypical mycobacteriosis, familial, X-linked 2, 300645
CYC1	81.4	95%	79%	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYCS	49.5	100%	92%	Thrombocytopenia 4, 612004
CYLD	114.3	100%	100%	Cylindromatosis, familial, 132700 Brooke-Spiegler syndrome, 605041 Trichoepithelioma, multiple familial, 1, 601606
CYP11A1	89.8	100%	97%	Adrenal insufficiency congenital with 46XY sex reversal partial or complete
CYP11B1	128.0	98%	94%	anti-Lepore-like Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 Aldosteronism, glucocorticoid-remediable, 103900
CYP11B2	104.9	98%	91%	Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 {Low renin hypertension, susceptibility to} Aldosterone to renin ratio raised
CYP17A1	105.5	99%	96%	17-alpha-hydroxylase/17,20-lyase deficiency, 202110 17,20-lyase deficiency, isolated, 202110
CYP19A1	131.0	100%	100%	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300 -3
CYP1B1	103.3	100%	99%	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 Peters anomaly, 604229
CYP21A2	5.2	17%	10%	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910
CYP24A1	105.8	100%	97%	Hypercalcemia, infantile, 143880
CYP26B1	66.2	100%	97%	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
CYP26C1	59.2	100%	91%	Focal facial dermal dysplasia 4, 614974
CYP27A1	118.1	100%	96%	Cerebrotendinous xanthomatosis, 213700
CYP27B1	107.1	100%	98%	Vitamin D-dependent rickets, type I, 264700

CYP2A6	29.3	54%	42%	Coumarin resistance, 122700 {Nicotine addiction, protection from}, 188890 {Lung cancer, resistance to}, 211980
CYP2B6	105.6	88%	88%	Efavirenz, poor metabolism of, 614546 {Efavirenz central nervous system toxicity, susceptibility to}, 614546
CYP2C19	103.4	98%	96%	Clopidogrel impaired responsiveness to
CYP2C8	133.7	100%	100%	Rhabdomyolysis, cerivastatin-induced
CYP2C9	108.6	100%	97%	Tolbutamide poor metabolizer Warfarin sensitivity, 122700
CYP2R1	107.1	99%	96%	Rickets due to defect in vitamin D 25-hydroxylation, 600081
CYP2U1	108.4	100%	99%	Spastic paraplegia 56, autosomal recessive, 615030
CYP4F22	98.9	100%	100%	Ichthyosis, congenital, autosomal recessive 5, 604777
CYP4V2	124.7	100%	99%	Bietti crystalline corneoretinal dystrophy, 210370
CYP7B1	97.1	100%	96%	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, autosomal recessive, 270800
D2HGDH	57.7	96%	86%	D-2-hydroxyglutaric aciduria, 600721
DAG1	145.4	100%	100%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818
DARS	131.4	100%	100%	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2	116.2	100%	100%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBH	99.0	100%	97%	[Dopamine-beta-hydroxylase activity levels, plasma] Dopamine beta-hydroxylase deficiency, 223360
DBT	111.7	100%	100%	Maple syrup urine disease, type II, 248600
DCAF17	102.4	99%	94%	Woodhouse-Sakati syndrome, 241080
DCC	120.1	99%	99%	Mirror movements 1, 157600 Colorectal cancer, somatic, 114500 Esophageal carcinoma, somatic 133239
DCHS1	99.4	100%	98%	Van Maldergem syndrome 1, 601390 (3)
DCLRE1C	106.1	97%	97%	Severe combined immunodeficiency, Athabascan type, 602450 Omenn syndrome, 603554
DCN	121.3	100%	100%	Corneal dystrophy, congenital stromal, 610048
DCTN1	112.7	99%	95%	Neuropathy, distal hereditary motor, type VIIB, 607641 {Amyotrophic lateral sclerosis, susceptibility to}, 105400 Perry syndrome, 168605
DCX	60.2	100%	94%	Lissencephaly, X-linked, 300067 Subcortical laminal heteropia, X-linked, 300067

DDB2	97.1	100%	99%	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DDC	88.8	100%	98%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	141.9	100%	97%	Spastic paraplegia 28, autosomal recessive, 609340
DDHD2	105.4	100%	100%	Spastic paraplegia 54, autosomal recessive, 615033
DDOST	97.6	100%	98%	Congenital disorder of glycosylation, type Ir, 614507
DDR2	127.4	100%	100%	Spondylometaepiphyseal dysplasia, short limb-hand type, 271665
DDX11	10.2	18%	13%	Warsaw breakage syndrome, 613398
DDX59	153.0	100%	100%	Orofaciodigital syndrome V, 174300
DEPDC5	114.0	99%	99%	Epilepsy, familial focal, with variable foci, 604364
DES	92.8	96%	88%	Myopathy, myofibrillar, 1, 601419 Cardiomyopathy, dilated, 1I, 604765 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 ?Muscular dystrophy, limb-girdle, type 2R, 615325
DFNA5	100.4	99%	95%	Deafness, autosomal dominant 5, 600994
DFNB31	85.0	100%	98%	Deafness autosomal recessive 31
DFNB59	127.4	100%	100%	Deafness autosomal recessive 59
DGKE	111.9	100%	99%	Nephrotic syndrome, type 7, 615008
DGUOK	99.3	100%	100%	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHCR24	86.6	99%	97%	Desmosterolosis, 602398
DHCR7	115.9	100%	97%	Smith-Lemli-Opitz syndrome, 270400
DHDDS	70.3	100%	90%	Retinitis pigmentosa 59, 613861
DHFR	51.9	81%	58%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHH	77.9	100%	100%	46XY partial gonadal dysgenesis, with minifascicular neuropathy, 607080 46XY sex reversal 7, 233420
DHODH	100.6	100%	99%	Miller syndrome, 263750
DHTKD1	103.3	100%	98%	2-aminoadipic 2-oxoadipic aciduria, 204750 Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DIABLO	114.0	97%	90%	Deafness autosomal dominant 64
DIAPH1	80.8	99%	91%	Deafness, autosomal dominant 1, 124900
DIAPH2	57.7	95%	93%	Premature ovarian failure, 300511
DIAPH3	111.8	97%	93%	Auditory neuropathy, autosomal dominant, 1, 609129
DICER1	119.3	100%	100%	Pleuropulmonary blastoma, 601200 Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800
DIP2B	100.8	100%	97%	Mental retardation, FRA12A type, 136630
DIS3L2	133.8	99%	94%	Perlman syndrome, 267000
DKC1	50.0	99%	91%	Dyskeratosis congenita, X-linked, 305000

DLAT	107.0	100%	100%	Pyruvate dehydrogenase E2 deficiency, 245348
DLC1	148.3	100%	99%	Colorectal cancer, somatic
DLD	143.7	100%	100%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLG3	39.5	90%	77%	Mental retardation, X-linked 90, 300850
DLL3	70.3	94%	81%	Spondylocostal dysostosis, autosomal recessive, 1, 277300
DLX3	75.6	98%	91%	Trichodontoosseous syndrome, 190320 Amelogenesis imperfecta, hypomaturation-hypoplastic type, with taurodontism, 104510
DMD	57.8	98%	95%	Duchenne muscular dystrophy, 310200 Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045
DMGDH	116.6	97%	96%	Dimethylglycine dehydrogenase deficiency, 605850
DMP1	110.0	100%	100%	Hypophosphatemic rickets, AR, 241520
DMPK	94.8	100%	99%	Myotonic dystrophy 1, 160900
DNA2	115.7	100%	100%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 6, 615156
DNAAF1	120.8	100%	99%	Ciliary dyskinesia primary 13
DNAAF2	113.0	100%	100%	Ciliary dyskinesia primary 10
DNAAF3	73.3	96%	81%	Ciliary dyskinesia, primary, 2, 606763
DNAH11	115.8	100%	99%	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH5	96.0	99%	98%	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAI1	124.5	100%	100%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	109.7	98%	93%	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJB2	107.5	100%	96%	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881
DNAJB6	42.4	85%	78%	Muscular dystrophy, limb-girdle, type 1E, 603511
DNAJC19	57.8	79%	78%	3-methylglutaconic aciduria, type V, 610198
DNAJC5	69.7	91%	79%	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350
DNAJC6	95.2	100%	97%	Parkinson disease 19, juvenile-onset, 615528
DNAL1	118.6	100%	100%	Ciliary dyskinesia, primary, 16, 614017
DNASE1L3	86.9	100%	100%	Systemic lupus erythematosus 16, 614420
DNM1L	104.9	100%	100%	Encephalopahty, lethal, due to defective mitochondrial peroxisomal fission, 614388
DNM2	80.0	100%	96%	Charcot-Marie-Tooth disease, dominant intermediate B, 606482 Myopathy, centronuclear, 160150 Charcot-Marie-Tooth disease, axonal, type 2M, 606482 Lethal congenital contracture syndrome 5, 615368

DNMT1	101.5	99%	96%	Neuropathy, hereditary sensory, type IE, 614116 Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121
DNMT3B	93.8	100%	99%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK6	89.4	99%	95%	Adams-Oliver syndrome 2, 614219
DOCK8	83.4	100%	98%	Mental retardation, autosomal dominant 2, 614113 Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
DOK7	54.1	95%	85%	Myasthenia, limb-girdle, familial, 254300 Fetal akinesia deformation sequence, 208150
DOLK	145.3	100%	100%	Congenital disorder of glycosylation type Im
DPAGT1	90.9	99%	95%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, with tubular aggregates 2, 614750
DPM1	157.7	100%	100%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	59.9	98%	88%	Congenital disorder of glycosylation, type Iu, 615042
DPM3	101.1	100%	100%	Congenital disorder of glycosylation, type Io, 612937
DPP6	91.8	98%	90%	Ventricular fibrillation, paroxysmal familial, 2, 612956
DPY19L2	22.8	24%	20%	Spermatogenic failure 9, 613958
DPYD	119.1	98%	96%	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270
DPYS	60.6	100%	98%	Dihydropyrimidinuria, 222748
DRC1	75.1	100%	97%	Ciliary dyskinesia, primary, 21, 615294
DRD2	112.3	100%	98%	Dystonia, myoclonic, 159900
DRD4	40.7	92%	67%	Autonomic nervous system dysfunction [Novelty seeking personality], 601696 (1) {Attention deficit-hyperactivity disorder}, 143465
DRD5	18.0	60%	47%	{Blepharospasm, primary benign}, 606798 Dystonia, primary cervical {Attention deficit-hyperactivity disorder, susceptibility to}, 143465
DSC2	104.3	100%	99%	Arrhythmogenic right ventricular dysplasia 11, 610476 Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476
DSC3	100.6	99%	99%	Hypotrichosis and recurrent skin vesicles, 613102
DSG1	150.0	100%	100%	pemphigus foliaceous antigen Keratosis palmoplantaris striata I, 148700
DSG2	132.4	100%	98%	Arrhythmogenic right ventricular dysplasia 10, 610193 Cardiomyopathy, dilated, 1BB, 612877
DSG4	134.5	99%	96%	Hypotrichosis, localized, autosomal recessive, 607903

DSP	130.6	99%	98%	Keratosis palmoplantaris striata II, 612908 Dilated cardiomyopathy with woolly hair and keratoderma, 605676 Arrhythmogenic right ventricular dysplasia 8, 607450 Skin fragility-woolly hair syndrome, 607655 Epidermolysis bullosa, lethal acan
DSPP	139.2	98%	96%	Dentinogenesis imperfecta, Shields type II, 125490 Deafness, autosomal dominant 36, with dentinogenesis, 605594 Dentinogenesis imperfecta, Shields type III, 125500 Dentin dysplasia, type II, 125420 -3
DST	145.9	100%	99%	Neuropathy, hereditary sensory and autonomic, type VI, 614653 Epidermolysis bullosa simplex, sutosomal recessive 2, 615425
DTNA	102.0	100%	97%	Left ventricular noncompaction 1, with or without congenital heart defects, 604169
DTNBP1	112.3	100%	100%	{Schizophrenia}, 181500 (2) Hermansky-Pudlak syndrome 7, 614076
DUOX2	92.8	94%	92%	Thryoid dyshormonogenesis 6, 607200
DUOXA2	93.5	100%	99%	Thyroid dyshormonogenesis 5, 274900
DUSP6	146.1	100%	100%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
DYM	102.2	100%	99%	Dyggve-Melchior-Clausen disease, 223800 Smith-McCort dysplasia, 607326
DYNC1H1	117.4	99%	96%	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant, AD, 158600
DYNC2H1	116.8	100%	99%	Asphyxiating thoracic dystrophy 3, 613091 Short rib-polydactyly syndrome, type III, 263510 Short rib-polydactyly syndrome, type IIB, 615087
DYRK1A	146.9	99%	99%	Mental retardation, autosomal dominant 7, 614104
DYSF	98.6	99%	99%	Muscular dystrophy, limb-girdle, type 2B, 253601 Myopathy, distal, with anterior tibial onset, 606768 Miyoshi muscular dystrophy 1, 254130
DYX1C1	93.9	100%	100%	{Dyslexia, susceptibility to, 1}, 127700
EARS2	70.5	93%	91%	Combined oxidative phosphorylation deficiency 12, 614924
EBP	43.9	94%	73%	Chondrodysplasia punctata, X-linked dominant, 302960
ECE1	93.4	97%	97%	Hirschsprung disease, cardiac defects, and autonomic dysfunction, 613870 {Hypertension, essential, susceptibility to}, 145500
ECEL1	62.0	96%	79%	Arthrogryposis, distal, type 5D, 615065

ECM1	108.5	100%	99%	Urbach-Wiethe disease, 247100
EDA	47.2	96%	86%	Ectodermal dysplasia 1 hypohidrotic X-linked
EDAR	80.1	100%	99%	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 -3
EDARADD	115.9	100%	95%	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941 Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, 614940
EDN1	129.1	100%	100%	[High density lipoprotein cholesterol level QTL 7]
EDN3	93.8	100%	100%	Waardenburg syndrome, type 4B, 613265 Central hypoventilation syndrome, congenital, 209880 {Hirschsprung disease, susceptibility to, 4}, 613712
EDNRA	122.3	100%	100%	Migraine, resistance to, 157300
EDNRB	161.7	100%	99%	{Hirschsprung disease, susceptibility to, 2}, 600155 ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580
EFEMP1	123.5	100%	94%	Doyne honeycomb degeneration of retina, 126600
EFEMP2	100.1	100%	100%	Cutis laxa, autosomal recessive, type IB, 614437
EFNB1	60.2	100%	96%	? Craniofrontonasal dysplasia, 304110
EFTUD2	89.7	98%	97%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EGF	113.7	99%	98%	Hypomagnesemia 4, renal, 611718
EGFR	97.6	100%	100%	Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980 Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980 {Nonsmall cell lung cancer, susceptibility to}, 211980
EGLN1	88.7	80%	74%	Erythrocytosis, familial, 3, 609820
EGR2	73.9	100%	98%	Neuropathy, congenital hypomyelinating, 1, 605253 Charcot-Marie-Tooth disease, type 1D, 607678 Dejerine-Sottas disease, 145900
EHMT1	99.8	98%	95%	Kleefstra syndrome, 610253
EIF2AK3	115.2	93%	92%	Wolcott-Rallison syndrome, 226980
EIF2AK4	107.6	99%	98%	Pulmonary venoocclusive disease 2, 234810 (3)
EIF2B1	113.5	97%	93%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B2	89.8	100%	98%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896

EIF2B3	86.3	100%	99%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B4	121.8	100%	100%	Leukoencephaly with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B5	96.4	100%	99%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF4A3	71.3	100%	98%	Robin sequence with cleft mandible and limb anomalies, 268305 (3)
EIF4G1	103.1	100%	99%	Parkinson disease 18, 614251
ELAC2	85.6	100%	100%	{Prostate cancer, hereditary, 2, susceptibility to}, 614731 Combined oxidative phosphorylation deficiency 17, 615440
ELANE	113.7	97%	80%	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
ELN	66.9	99%	96%	Supravalvar aortic stenosis, 185500 Cutis laxa, AD, 123700
ELOVL4	102.9	100%	100%	Stargardt disease 3, 600110 Macular dystrophy, autosomal dominant, chromosome 6-linked, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
EMD	84.8	100%	95%	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
EMG1	87.5	100%	100%	Bowen-Conradi syndrome, 211180
EMX2	106.1	100%	100%	Schizencephaly, 269160
ENAM	130.4	100%	100%	Amelogenesis imperfecta, type IB, 104500 Amelogenesis imperfecta, type IC, 204650
ENG	67.2	99%	94%	Telangiectasia, hereditary hemorrhagic, type 1, 187300
ENO3	107.7	99%	96%	Glycogen storage disease XIII, 612932
ENPP1	113.3	95%	93%	Ossification of posterior longitudinal ligament of spine, 602475 {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853 {Obesity, susceptibility to}, 601665 Arterial calcification, generalized, of infancy, 1, 208000 Hypophos
ENTPD1	116.2	100%	99%	Spastic paraplegia 64, 615683
EOGT	105.5	100%	99%	Adams-Oliver syndrome 4, 615297
EP300	139.2	99%	98%	Rubinstein-Taybi syndrome 2, 613684 Colorectal cancer, somatic, 114500
EPAS1	85.5	98%	92%	Erythrocytosis, familial, 4, 611783
EPB41	125.7	100%	100%	Elliptocytosis-1, 611804
EPB42	94.9	99%	95%	Spherocytosis, hereditary, type 5, 612690
EPCAM	117.1	100%	99%	Diarrhea 5, with tufting enteropathy, congenital, 613217

				Colorectal cancer, hereditary nonpolyposis, type 8, 613244
EPG5	89.4	100%	99%	Vici syndrome, 242840
EPHA2	89.5	97%	94%	Cataract 6, multiple types, 116600
EPHB2	115.6	97%	97%	Prostate cancer, progression and metastasis of, 603688
EPHX1	94.0	95%	91%	?Fetal hydantoin syndrome (1) Diphenylhydantoin toxicity (1) Hypercholanemia, familial, 607748 {Preeclampsia, susceptibility to}, 189800
EPM2A	54.7	80%	73%	Epilepsy, progressive myoclonic 2A (Lafora), 254780
EPX	101.2	100%	97%	Eosinophil peroxidase deficiency, 261500
ERBB2	100.5	98%	98%	Adenocarcinoma of lung, somatic, 211980 Glioblastoma, somatic, 137800 Gastric cancer, somatic, 613659 Ovarian cancer, somatic,
ERBB3	112.3	100%	99%	Lethal congenital contractural syndrome 2, 607598
ERBB4	121.0	100%	100%	Amyotrophic lateral sclerosis 19, 615515
ERCC1	73.0	100%	96%	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	88.6	99%	96%	Xeroderma pigmentosum, group D, 278730 Trichothiodystrophy, 601675 Cerebrooculofacioskeletal syndrome 2, 610756
ERCC3	123.3	100%	100%	Xeroderma pigmentosum, group B, 610651 Trichothiodystrophy, 601675
ERCC4	141.5	97%	94%	Xeroderma pigmentosum, group F, 278760 XFE progeroid syndrome, 610965 Fanconi anemia, complementation group Q, 615272 Xeroderma pigmentosum, type F/Cockayne syndrome, 278760
ERCC5	122.5	99%	97%	Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	153.2	100%	98%	Cockayne syndrome, type B, 133540 Cerebrooculofacioskeletal syndrome 1, 214150 De Sanctis-Cacchione syndrome, 278800 {Macular degeneration, age-related, susceptibility to 5}, 613761 UV-sensitive syndrome 1, 600630 {Lung cancer, suscept
ERCC6L2	130.4	100%	100%	Bone marrow failure syndrome 2, 615715 (3)

ERCC8	86.0	100%	100%	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621
ERF	109.2	100%	100%	Craniosynostosis 4, 600775
ERLIN2	115.7	100%	98%	Spastic paraplegia 18, autosomal recessive, 611225
ESCO2	96.3	100%	99%	Roberts syndrome, 268300 SC phocomelia syndrome, 269000
ESPN	42.7	77%	52%	Deafness, autosomal recessive 36, 609006 Deafness, neurosensory, without vestibular involvement, autosomal dominant
ESR1	115.7	100%	100%	Estrogen resistance, 615363 {HDL response to hormone replacement, augmented} {Migraine, susceptibility to}, 157300 {Atherosclerosis, susceptibility to} {Myocardial infarction, susceptibility to}, 608446 {Breast cancer}, 114480 (1)
ESRRB	63.0	84%	75%	Deafness, autosomal recessive 35, 608565
ETFA	115.4	100%	100%	Glutaric acidemia IIA, 231680
ETFB	107.8	100%	99%	Glutaric acidemia IIB, 231680
ETFDH	132.6	100%	100%	Glutaric acidemia IIC, 231680
ETHE1	61.0	100%	96%	Ethylmalonic encephalopathy, 602473
ETV6	109.8	100%	98%	Leukemia, acute myeloid, somatic, 601626
EVC	81.0	91%	88%	Ellis-van Creveld syndrome, 225500 Weyers acrodental dysostosis, 193530
EVC2	102.4	94%	92%	Ellis-van Creveld syndrome
EWSR1	63.3	91%	73%	Ewing sarcoma, 612219 Neuroepithelioma, 612219
EXOSC3	64.3	94%	80%	Pontocerebellar hypoplasia, type 1B, 614678
EXPH5	147.5	100%	100%	Epidermolysis bullosa, nonspecific, autosomal recessive, 615028
EXT1	104.7	98%	95%	Exostoses, multiple, type 1, 133700 Chondrosarcoma, 215300
EXT2	117.3	100%	96%	Exostoses, multiple, type 2, 133701
EYA1	113.1	100%	99%	Branchiootorenal syndrome 1, with or without cataracts, 113650 Anterior segment anomalies with or without cataract, 113650 Branchiootic syndrome 1, 602588 Otofaciocervical syndrome, 166780
EYA4	124.0	100%	100%	Deafness, autosomal dominant 10, 601316 Cardiomyopathy, dilated, 1J, 605362

EYS	127.6	100%	99%	Retinitis pigmentosa 25, 602772
EZH2	89.2	99%	93%	Weaver syndrome, 277590
F10	98.8	100%	100%	Factor X deficiency, 227600
F11	107.6	96%	91%	Factor XI deficiency, autosomal recessive, 612416 Factor XI deficiency, autosomal dominant, 612416
F12	94.8	100%	96%	Factor XII deficiency, 234000 Angioedema, hereditary, type III, 610618
F13A1	101.2	99%	97%	Factor XIIIA deficiency, 613225 {Myocardial infarction, protection against}, 608446 {Venous thrombosis, protection against}, 188050
F13B	91.1	100%	98%	Factor XIIIB deficiency, 613235
F2	87.8	96%	91%	Hypoprothrombinemia, 613679 Dysprothrombinemia, 613679 Thrombophilia due to thrombin defect, 188050 {Stroke, ischemic, susceptibility to}, 601367 {Pregnancy loss, recurrent, susceptibility to, 2}, 614390
F5	134.3	100%	99%	Factor V deficiency, 227400 {Thrombophilia, susceptibility to, due to factor V Leiden}, 188055 {Stroke, ischemic, susceptibility to}, 601367 {Budd-Chiari syndrome}, 600880 Thrombophilia due to activated protein C resistance, 188055 {Pr
F7	87.1	100%	100%	Factor VII deficiency, 227500 {Myocardial infarction, decreased susceptibility to}, 608446
F8	68.5	99%	95%	Hemophilia A, 306700
F9	82.4	100%	100%	Hemophilia B, 306900 {Warfarin sensitivity}, 122700 Thrombophilia, X-linked, due to factor IX defect, 300807 {Deep venous thrombosis, protection against}, 300807
FA2H	77.3	98%	94%	Spastic paraplegia 35 autosomal recessive
FADD	110.3	100%	100%	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovasuclar malformations, 613759
FAH	108.6	100%	98%	Tyrosinemia, type I, 276700
FAM111A	175.3	100%	100%	Kenny-Caffey syndrome, type 2, 127000 Gracile bone dysplasia, 602361
FAM111B	187.0	100%	99%	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704 (3)

FAM126A	143.6	100%	100%	Leukodystrophy, hypomyelinating, 5, 610532
FAM134B	89.2	100%	99%	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
FAM161A	148.2	100%	100%	Retinitis pigmentosa 28, 606068
FAM20A	70.9	99%	94%	Amelogenesis imperfecta and gingival fibromatosis syndrome, 614253
FAM20C	78.4	90%	84%	Raine syndrome, 259775
FAM58A	20.3	48%	45%	STAR syndrome, 300707
FAM83H	81.0	100%	99%	Amelogenesis imperfecta, type 3, 130900
FAN1	111.6	100%	99%	Interstitial nephritis, karyomegalic, 614817
FANCA	86.0	100%	96%	Fanconi anemia, complementation group A, 227650
FANCB	65.5	96%	88%	Fanconi anemia complementation group B
FANCC	78.9	99%	96%	Fanconi anemia, complementation group C, 227645
FANCD2	98.9	87%	85%	Fanconi anemia, complementation group D2, 227646
FANCE	84.3	90%	87%	Fanconi anemia, complementation group E, 600901
FANCF	152.2	100%	100%	Fanconi anemia, complementation group F, 603467
FANCG	118.1	100%	96%	Fanconi anemia complementation group G
FANCI	126.2	100%	99%	Fanconi anemia, complementation group I, 609053
FANCL	95.2	100%	99%	Fanconi anemia complementation group L
FANCM	122.3	100%	99%	Fanconi anemia, complementation group M, 614087
FARS2	97.6	98%	94%	Combined oxidative phosphorylation deficiency 14, 614946
FAS	181.5	100%	100%	Autoimmune lymphoproliferative syndrome type IA
FASLG	83.8	97%	92%	Autoimmune lymphoproliferative syndrome type IB
FAT4	160.3	100%	100%	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 Van Maldergem syndrome 2, 615546
FBLN1	104.1	98%	95%	Synpolydactyly, 3/3'4, associated with metacarpal and metatarsal synostoses, 608180
FBLN5	81.8	100%	99%	Cutis laxa, autosomal recessive, type IA, 219100 Cutis laxa, autosomal dominant 2, 614434 Macular degeneration, age-related, 3, 608895
FBN1	98.5	100%	99%	Marfan syndrome, 154700 Ectopia lentis, familial, 129600 MASS syndrome, 604308 Weill-Marchesani syndrome 2, dominant, 608328 Aortic aneurysm, ascending, and dissection Stiff skin syndrome, 184900 Acromicric dysplasia, 102370 Ge

FBN2	104.7	99%	98%	Contractural arachnodactyly, congenital, 121050
FBP1	90.3	100%	96%	Fructose-1,6-bidphosphatase deficiency, 229700
FBXL4	154.9	100%	100%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FBXO38	120.7	100%	96%	Neuronopathy, distal hereditary motor, type IID, 65575
FBXO7	159.5	100%	100%	Parkinson disease 15, autosomal recessive, 260300
FCGR3A	50.6	50%	49%	{Viral infections, recurrent}
FCGR3B	57.7	57%	53%	Neutropenia, alloimmune neonatal
FCN3	105.6	99%	95%	Immunodeficiency due to ficolin 3 deficiency, 613860
FECH	106.9	100%	98%	Protoporphyria, erythropoietic, autosomal recessive, 177000
FERMT1	99.0	100%	97%	Kindler syndrome
FERMT3	104.3	100%	97%	Leukocyte adhesion deficiency type III
FGA	169.8	100%	99%	Dysfibrinogenemia, alpha type, causing bleeding diathesis Dysfibrinogenemia, alpha type, causing recurrent thrombosis Amyloidosis, hereditary renal, 105200 Afibrinogenemia, congenital, 202400
FGB	116.1	99%	97%	Dysfibrinogenemia, beta type Afibrinogenemia, congenital, 202400 Thrombophilia, dysfibrinogenemic
FGD1	47.0	94%	88%	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400
FGD4	124.3	100%	100%	Charcot-Marie-Tooth disease, type 4H, 609311
FGF10	124.1	100%	100%	Aplasia of lacrimal and salivary glands, 180920 LADD syndrome, 149730
FGF14	113.5	100%	100%	Spinocerebellar ataxia 27, 609307
FGF16	66.8	100%	98%	Metacarpal 4-5 fusion, 609630
FGF17	93.2	100%	100%	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
FGF23	75.3	96%	92%	Hypophosphatemic rickets, autosomal dominant, 193100 Osteomalacia, tumor-induced (1) Tumoral calcinosis, hyperphosphatemic, familial, 211900
FGF3	72.2	100%	93%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGF8	42.8	83%	61%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGF9	150.9	100%	100%	Multiple synostoses syndrome 3, 612961

FGFR1	123.9	100%	100%	Pfeiffer syndrome, 101600 Jackson-Weiss syndrome, 123150 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Osteoglophonic dysplasia, 166250 Trigonocephaly 1, 190440 Hartsfield syndrome, 615465
FGFR2	122.8	100%	98%	Crouzon syndrome, 123500 Jackson-Weiss syndrome, 123150 Beare-Stevenson cutis gyrata syndrome, 123790 Pfeiffer syndrome, 101600 Apert syndrome, 101200 Saethre-Chotzen syndrome, 101400 Craniosynostosis, nonspecific Gastric cance
FGFR3	72.8	94%	91%	Achondroplasia, 100800 Hypochondroplasia, 146000 Thanatophoric dysplasia, type I, 187600 Crouzon syndrome with acanthosis nigricans, 612247 Muenke syndrome, 602849 Bladder cancer, somatic, 109800 Colorectal cancer, somatic, 114500
FGG	119.4	100%	97%	Dysfibrinogenemia, gamma type Hypofibrinogenemia, gamma type Thrombophilia, dysfibrinogenemic
FH	85.7	98%	89%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FHL1	45.6	96%	81%	Hemophagocytic lymphohistiocytosis, familial, 1 (2)
FIG4	132.9	100%	99%	Charcot-Marie-Tooth disease, type 4J, 611228 Amyotrophic lateral sclerosis 11, 612577 Yunis-Varon syndrome, 216340
FIGLA	82.1	97%	91%	Premature ovarian failure 6, 612310
FKBP10	85.2	100%	99%	Osteogenesis imperfecta, type XI, 610968
FKBP14	141.6	100%	100%	Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss, 614557

FKRP	80.2	100%	98%	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
FKTN	108.3	100%	99%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Cardiomyopathy, dilated, 1X, 611615 Muscular dy
FLCN	119.1	100%	98%	Birt-Hogg-Dube syndrome, 135150 Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700 Colorectal cancer, somatic, 114500
FLG	49.9	99%	82%	Ichthyosis vulgaris, 146700 {Dermatitis, atopic, susceptibility to, 2}, 605803
FLNA	60.2	98%	90%	Heterotopia, periventricular, 300049 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Frontometaphyseal dysplasia, 3056
FLNB	88.2	100%	98%	Spondylocarpotarsal synostosis syndrome, 272460 Larsen syndrome, 150250 Atelosteogenesis, type I, 108720 Atelosteogenesis, type III, 108721 Boomerang dysplasia, 112310
FLNC	93.2	96%	94%	Myopathy, myofibrillar, 5, 609524 Myopathy, distal, 4, 614065 -3
FLRT3	217.0	100%	100%	Hypogonadotropic hypogonadism 21 with anosmia, 615271
FLT3	105.7	99%	96%	Leukemia, acute myeloid, reduced survival in Leukemia, acute myeloid, 601626 Leukemia, acute lymphoblastic
FLT4	98.2	99%	99%	Lymphedema, hereditary I, 153100 Hemangioma, capillary infantile, somatic, 602089
FLVCR1	95.8	100%	100%	Ataxia, posterior column, with retinitis pigmentosa, 609033

FLVCR2	134.9	100%	100%	Proliferative vasculopathy and hydraencephaly-hydrocephaly syndrome, 225790
FMO3	107.4	100%	100%	Trimethylaminuria, 602079
FMR1	56.6	98%	95%	Fragile X syndrome, 300624 Fragile X tremor/ataxia syndrome, 300623 Premature ovarian failure 1, 311360
FN1	88.7	99%	96%	Glomerulopathy with fibronectin deposits 2, 601894 Plasma fibronectin deficiency, 614101 (1)
FOLR1	78.4	100%	97%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXC1	51.1	100%	92%	Iridogoniodysgenesis, type 1, 601631 Rieger or Axenfeld anomalies, 602482 Axenfeld-Rieger syndrome, type 3, 602482 Iris hypoplasia and glaucoma, 601631
FOXC2	80.3	100%	90%	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXE1	111.5	100%	100%	Bamforth-Lazarus syndrome
FOXE3	14.2	70%	38%	Anterior segment mesenchymal dysgenesis, 107250 Aphakia, congenital primary, 610256
FOXF1	107.1	100%	100%	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380
FOXG1	91.1	93%	80%	Rett syndrome, congenital variant, 613454
FOXI1	100.5	100%	100%	Enlarged vestibular aqueduct, 600791
FOXL2	86.1	100%	100%	Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 Premature ovarian failure 3, 608996
FOXN1	108.5	100%	99%	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXO1	123.7	100%	88%	Rhabdomyosarcoma alveolar
FOXP1	102.1	99%	97%	Mental retardation with language impairment and autistic features, 613670
FOXP2	113.9	100%	100%	Speech-language disorder-1, 602081
FOXP3	31.8	89%	69%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790 {Diabetes mellitus, type I, susceptibility to}, 222100
FOXRED1	95.2	100%	97%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
FRAS1	99.0	98%	95%	Fraser syndrome, 219000

FREM1	108.6	100%	98%	Bifid nose with or without anorectal and renal anomalies, 608980 Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485
FREM2	127.7	100%	99%	Fraser syndrome, 219000
FRMD7	64.4	100%	95%	Nystagmus 1, congenital, X-linked, 310700 Nystagmus, infantile periodic alternating, X-linked, 310700
FSCN2	82.2	100%	100%	Retinitis pigmentosa 30, 607921
FSHB	81.4	100%	100%	Follicle-stimulating hormone deficiency, isolated, 229070
FSHR	92.8	100%	99%	Ovarian dysgenesis 1, 233300 Ovarian response to FSH stimulation, 276400 Ovarian hyperstimulation syndrome, 608115
FTCD	58.1	92%	81%	Glutamate formiminotransferase deficiency, 229100
FTL	95.3	100%	95%	Hyperferritinemia-cataract syndrome, 600886 Neurodegeneration with brain iron accumulation 3, 606159
FTO	126.9	97%	97%	Growth retardation, developmental delay, coarse facies, and early death, 612938
FTSJ1	53.2	92%	77%	Mental retardation, X-linked 9, 309549
FUCA1	74.3	100%	96%	Fucosidosis, 230000
FUS	79.5	100%	95%	Amyotrophic lateral sclerosis 6, autosomal recessive, with or without frontotemporal dementia, 608030 Tremor, hereditary essential, 4, 614782
FUT6	77.0	81%	73%	Fucosyltransferase 6 deficiency, 613852
FUZ	79.4	100%	100%	Neural tube defects, 182940
FXN	87.0	93%	87%	Friedreich ataxia, 229300 Friedreich ataxia with retained reflexes, 229300
FXYD2	64.3	99%	80%	Hypomagnesemia-2, renal, 154020
FYCO1	86.2	99%	98%	Cataract 18, autosomal recessive, 610019
FZD4	142.2	100%	100%	Exudative vitreoretinopathy, 133780 Retinopathy of prematurity, 133780
FZD6	154.0	100%	100%	Nail disorder, nonsyndromic congenital, 10, (claw-shaped nails), 614157
G6PC	140.5	100%	100%	Glycogen storage disease Ia, 232200
G6PC3	116.3	100%	100%	Neutropenia, severe congenital 4, autosomal recessive, 612541 Dursun syndrome, 612541
G6PD	57.0	95%	92%	Hemolytic anemia due to G6PD deficiency Favism, 134700 {Resistance to malaria due to G6PD deficiency}, 611162
GAA	96.6	100%	98%	Glycogen storage disease II, 232300

GABRA1	135.1	100%	96%	{Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136 {Epilepsy, childhood absence, susceptibility to, 4}, 611136
GABRB3	113.7	100%	97%	Insomnia {Epilepsy, childhood absence, susceptibility to, 5}, 612269
GABRG2	135.7	98%	94%	Epilepsy, generalized, with febrile seizures plus, type 3, 611277 {Epilepsy, childhood absence, susceptibility to, 2}, 607681 Febrile seizures, familial, 8, 611277
GAD1	96.2	100%	98%	Cerebral palsy, spastic quadriplegic, 1, 603513
GALC	96.4	100%	96%	Krabbe disease, 245200
GALE	115.6	100%	100%	Galactose epimerase deficiency, 230350
GALK1	96.4	100%	100%	Galactokinase deficiency with cataracts, 230200
GALNS	67.4	92%	92%	Mucopolysaccharidosis IVA, 253000
GALNT3	108.3	100%	100%	Tumoral calcinosis, hyperphosphatemic, familial, 211900
GALT	113.2	100%	100%	Galactosemia, 230400
GAMT	91.8	98%	92%	Cerebral creatine deficiency syndrome 2, 612736
GAN	147.9	100%	99%	Giant axonal neuropathy-1, 256850
GARS	135.8	100%	100%	Charcot-Marie-Tooth disease type 2D
GATA1	53.3	96%	87%	Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367 Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 Thrombocytopenia with beta-thalassemia, X-linked, 314050 Anemia, X-linked, with/without neu
GATA2	95.5	99%	93%	Dendritic cell, monocyte, B lymphocyte, and natural killer lymphocyte deficiency, 614172 Emberger syndrome, 614038 {Myelodysplastic syndrome, susceptibility to}, 614286 {Leukemia, acute myeloid, susceptibility to}, 601626
GATA3	126.7	100%	98%	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GATA4	58.6	78%	64%	Atrial septal defect 2, 607941 Ventricular septal defect 1, 614429 Atrioventricular septal defect 4, 614430
GATA6	49.9	92%	71%	Atrioventricular septal defect 5, 614474 Atrial septal defect 9, 614475 Pancreatic agenesis and congenital heart defects, 600001 Persistent truncus arteriosus, 217095 Tetralogy of Fallot, 187500

GATAD1	81.4	93%	89%	Cardiomyopathy, dilated, 2B, 614672
GATAD2B	112.9	100%	98%	Mental retardation, autosomal dominant 18, 615074
GATM	88.8	100%	94%	Cerebral creatine deficiency syndrome 3, 612718
GBA	62.1	63%	58%	Gaucher disease, type I, 230800 Gaucher disease, type II, 230900 Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 Gaucher disease, perinatal lethal, 608013 {Parkinson disease, late-onset, susceptibility to}, 168600
GBA2	122.9	100%	100%	Spastic paraplegia 46, autosomal recessive, 614409
GBE1	104.3	98%	94%	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GCDH	80.6	92%	91%	Glutaricaciduria, type I, 231670
GCH1	99.7	100%	100%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GCK	82.5	100%	97%	MODY, type II, 125851 Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, gestational, 125851 Hyperinsulinemic hypoglycemia, familial, 3, 602485 Diabetes mellitus, permanent neonatal, 606176
GCLC	137.3	100%	100%	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 {Myocardial infarction, susceptibility to}, 608446
GCM2	118.7	100%	100%	Hypoparathyroidism familial isolated
GCNT2	161.3	100%	100%	[Blood group, Ii], 110800 Cataract 13 with adult i phenotype, 110800 Adult i phenotype without cataract, 110800
GCSH	14.8	52%	39%	Glycine encephalopathy, 605899
GDAP1	106.2	100%	100%	Charcot-Marie-Tooth disease, type 4A, 214400 Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706 Charcot-Marie-Tooth disease, axonal, type 2K, 607831 Charcot-Marie-Tooth disease, recessive intermediate, A, 608340
GDF1	25.4	95%	71%	Double-outlet right ventricle, 217095 Tetralogy of Fallot, 187500 Transposition of great arteries, dextro-looped 3, 613854 Right atrial isomerism, 208530

GDF2	143.9	100%	100%	Telangiectasia, hereditary hemorraghic, type 5, 615506
GDF3	127.8	100%	100%	Klippel-Feil syndrome 3, autosomal dominant, 613702 Microphthalmia with coloboma 6, 613703 Microphthalmia, isolated 7, 613704
GDF5	99.5	100%	100%	Acromesomelic dysplasia, Hunter-Thompson type, 201250 Brachydactyly, type C, 113100 Chondrodysplasia, Grebe type, 200700 Du Pan syndrome, 228900 Brachydactyly, type A2, 112600 Symphalangism, proximal, 1B, 615298 Multiple synostoses
GDF6	135.5	100%	100%	Klippel-Feil syndrome 1, autosomal dominant, 118100 Microphthalmia, isolated 4, 613094 Microphthalmia with coloboma 6, digenic, 613703 Leber congenital amaurosis 17, 615360
GDI1	70.5	99%	96%	Mental retardation, X-linked 41, 300849
GDNF	164.3	100%	99%	Central hypoventilation syndrome, 209880 {Pheochromocytoma, modifier of}, 171300 {Hirschsprung disease, susceptibility to, 3}, 613711
GFAP	83.0	100%	98%	Alexander disease, 203450
GFER	63.9	99%	94%	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076
GFI1	61.8	100%	93%	Neutropenia, severe congenital 2, autosomal dominant, 613107 Neutropenia, nonimmune chronic idiopathic, of adults, 607847
GFI1B	96.5	100%	100%	Bleeding disorder, platelet-type,17, 187900
GFM1	121.8	100%	100%	Combined oxidative phosphorylation deficiency 1, 609060
GFPT1	105.6	100%	96%	Myasthenia, congenital, with tubular aggregates 1, 610542
GGCX	92.4	99%	99%	Vitamin K-dependent coagulation defect, 277450 Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842
GH1	66.9	69%	67%	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	132.4	99%	99%	Laron dwarfism, 262500 Short stature, 604271 {Hypercholesterolemia, familial, modification of}, 143890 Increased responsiveness to growth hormone
GHRHR	104.0	100%	98%	Growth hormone deficiency, isolated, type IB, 612781
GHSR	128.8	100%	100%	Short stature, 604271
GIF	108.9	100%	100%	Intrinsic factor deficiency, 261000
GIGYF2	106.4	99%	97%	Parkinson disease 11, 607688
GIPC3	107.2	95%	92%	Deafness, autosomal recessive 15, 601869
GJA1	63.1	91%	79%	Oculodentodigital dysplasia, 164200 Syndactyly, type III, 186100 Hypoplastic left heart syndrome 1, 241550 Atrioventricular septal defect 3, 600309 Oculodentodigital dysplasia, autosomal recessive, 257850 Craniometaphyseal dysplasia, a
GJA3	100.0	100%	99%	Cataract 14, multiple types, 601885
GJA5	124.4	100%	100%	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic, 108770
GJA8	135.9	100%	99%	Cataract 1, multiple types, 116200
GJB1	85.4	100%	100%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GJB2	172.7	100%	100%	Deafness, autosomal recessive 1A, 220290 Deafness, autosomal dominant 3A, 601544 Vohwinkel syndrome, 124500 Keratoderma, palmoplantar, with deafness, 148350 Keratitis-ichthyosis-deafness syndrome, 148210 Hystrix-like ichthyosis with de
GJB3	149.1	100%	100%	Erythrokeratodermia variabilis et progressiva, 133200 Deafness, autosomal dominant 2B, 612644 Deafness, autosomal recessive Deafness, autosomal dominant, with peripheral neuropathy Deafness, digenic, GJB2/GJB3, 220290
GJB4	136.7	100%	100%	Erythrokeratodermia variabilis with erythema gyratum repens, 133200
GJB6	157.2	100%	100%	Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290 Ectodermal dysplasia 2, Clouston type, 129500

GJC2	57.0	99%	83%	Leukodystrophy, hypomyelinating, 2, 608804 Spastic paraplegia 44, autosomal recessive, 613206 Lymphedema, hereditary, IC, 613480
GK	24.5	77%	56%	Glycerol kinase deficiency, 307030
GLA	47.2	95%	86%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	76.5	99%	94%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLDC	60.1	98%	85%	Glycine encephalopathy, 605899
GLE1	103.8	98%	94%	Lethal congenital contracture syndrome 1, 253310 Arthrogryposis, lethal, with anterior horn cell disease, 611890
GLI2	107.0	100%	97%	Holoprosencephaly-9, 610829
GLI3	121.7	100%	99%	Greig cephalopolysyndactyly syndrome, 175700 Pallister-Hall syndrome, 146510 Polydactyly, preaxial, type IV, 174700 Polydactyly, postaxial, types A1 and B, 174200 {Hypothalamic hamartomas, somatic}, 241800
GLIS2	88.9	100%	98%	Nephronophthisis 7, 611498
GLIS3	95.7	100%	98%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199 -3
GLMN	99.6	100%	100%	Glomuvenous malformations
GLRA1	113.9	100%	98%	Hyperekplexia, hereditary 1, autosomal dominant or recessive, 149400
GLRB	135.0	100%	99%	Hyperekplexia 2, autosomal recessive, 614619
GLRX5	29.9	72%	46%	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive, 205950
GLUD1	111.8	88%	88%	Hyperinsulinism-hyperammonemia syndrome, 606762
GLUL	28.1	60%	49%	Glutamine deficiency, congenital, 610015
GLYCTK	83.8	99%	98%	D-glyceric aciduria, 220120
GM2A	127.8	100%	100%	GM2-gangliosidosis, AB variant, 272750
GMPPA	115.1	100%	99%	Alacrima, achalasia, and mental retardation syndrome, 615510 (3)
GMPPB	118.5	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation),
GMPS	116.3	100%	100%	Leukemia, acute myelogenous, 601626

GNA11	95.5	100%	99%	Hypocalciuric hypercalcemia, type II, 145981 Hypocalcemia, autosomal dominant 2, 615361
GNAI2	76.6	100%	99%	GNAI2L Pituitary ACTH-secreting adenoma Ventricular tachycardia, idiopathic, 192605
GNAI3	125.5	100%	100%	Auriculocondylar syndrome 1, 602483
GNAL	91.1	100%	97%	Dystonia 25, 615073
GNAO1	109.4	100%	99%	Epileptic encephalopathy, early infantile, 17, 615473
GNAQ	69.0	99%	93%	Sturge-Weber syndrome, somatic, mosaic, 185300 Capillary malformations, congenital, 1, somatic, mosaic, 163000
GNAS	125.3	100%	99%	Pseudohypoparathyroidism Ia, 103580 McCune-Albright syndrome, 174800 Pseudohypoparathyroidism Ic, 612462 Osseous heteroplasia, progressive, 166350 Pseudohypoparathyroidism Ib, 603233 Prolonged bleeding time, brachydactyly and mental re
GNAT1	81.7	100%	95%	Night blindness, congenital stationary, autosomal dominant 3, 610444
GNAT2	125.7	100%	100%	Achromatopsia-4, 613856
GNB4	144.4	100%	100%	Charcot-Marie-Tooth disease, dominant intermediate F, 615185
GNE	99.5	100%	99%	Sialuria, 269921 Inclusion body myopathy, autosomal recessive, 600737 Nonaka myopathy, 605820
GNMT	85.8	100%	99%	Glycine N-methyltransferase deficiency, 606664
GNPAT	115.9	100%	100%	Chondrodysplasia punctata, rhizomelic, type 2, 222765
GNPTAB	135.6	100%	100%	Mucolipidosis III alpha/beta, 252600 Mucolipidosis II alpha/beta, 252500
GNPTG	89.9	86%	80%	Mucolipidosis III gamma
GNRH1	34.0	100%	91%	Hypogonadotropic hypogonadism 12 with or without anosmia, 614841
GNRHR	169.5	100%	100%	Hypogonadotropic hypogonadism 7 with or without anosmia, 146110 Fertile eunuch syndrome, 228300
GNS	80.0	94%	85%	Mucopolysaccharidosis type IIID, 252940
GOLGA5	124.3	100%	100%	Thyroid carcinoma, papillary, 188550
GORAB	150.9	100%	98%	Geroderma osteodysplasticum, 231070
GOSR2	111.9	100%	100%	Epilepsy, progressive myoclonic 6, 614018
GOT1	100.1	100%	100%	Aspartate aminotransferase, serum level of, QTL1, 614419

GP1BA	117.2	96%	95%	Bernard-Soulier syndrome, type A1 (recessive), 231200 {Nonarteritic anterior ischemic optic neuropathy, susceptibility to}, 258660 Bernard-Soulier syndrome, type A2 (dominant), 153670 von Willebrand disease, platelet-type, 177820
GP1BB	27.3	92%	61%	Bernard-Soulier syndrome, type B, 231200 Giant platelet disorder, isolated, 231200
GP6	100.4	100%	97%	Bleeding disorder, platelet-type, 11, 614201
GP9	56.9	95%	89%	Bernard-Soulier syndrome, type C, 231200
GPC3	51.4	100%	97%	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPC6	110.0	100%	100%	Omodysplasia 1, 258315
GPD1	72.9	100%	99%	Hypertriglyceridemia, transient infantile, 614480
GPD1L	105.6	100%	100%	Brugada syndrome 2, 611777
GPHN	119.4	100%	100%	Molybdenum cofactor deficiency, type C, 252150
GPI	91.9	100%	97%	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470
GPR143	26.6	83%	62%	Ocular albinism, type I, Nettleship-Falls type, 300500 Nystagmus 6, congenital, X-linked, 300814
GPR179	144.9	100%	99%	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
GPR56	94.4	100%	99%	Polymicrogyria, bilateral frontoparietal, 606854
GPR98	115.4	99%	98%	Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472
GPSM2	146.5	100%	100%	Chudley-McCullough syndrome, 604213
GRHL2	100.4	100%	98%	Deafness, autosomal dominant 28, 608641
GRHL3	106.2	100%	98%	Van der Woude syndrome 2, 606713 (3)
GRHPR	88.2	97%	78%	Hyperoxaluria, primary, type II, 260000
GRIA3	54.6	92%	84%	Mental retardation, X-linked 94, 300699
GRIK2	121.1	100%	99%	Mental retardation, autosomal recessive, 6, 611092
GRIN1	84.1	99%	93%	Mental retardation, autosomal dominant 8, 614254
GRIN2A	130.9	99%	98%	Epilepsy with neurodevelopmental defects, 613971
GRIN2B	140.6	99%	98%	Mental retardation, autosomal dominant 6, 613970
GRIP1	95.9	99%	95%	Fraser syndrome, 219000
GRK1	95.1	100%	99%	Oguchi disease-2, 613411
GRM1	152.4	100%	99%	Spinocerebellar ataxia, autosomal recessive 13, 614831

GRM6	96.1	95%	92%	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270
GRN	115.7	100%	99%	Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706
GRXCR1	189.0	100%	100%	Deafness, autosomal recessive 25, 613285
GSC	48.7	99%	81%	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471
GSN	70.7	93%	85%	Amyloidosis, Finnish type, 105120
GSS	80.0	98%	94%	Hemolytic anemia due to glutathione synthetase deficiency, 231900 Glutathione synthetase deficiency, 266130
GTF2H5	97.0	100%	100%	Trichothiodystrophy, complementation group A, 601675
GUCA1A	67.7	65%	52%	Cone dystrophy-3, 602093 Cone-rod dystrophy 14, 602093
GUCA1B	118.6	100%	100%	Retinitis pigmentosa 48, 613827
GUCY1A3	149.8	100%	100%	Moyamoya 6 with achalasia, 615750
GUCY2C	96.2	100%	99%	Diarrhea 6, 614616 Meconium ileus, 614665
GUCY2D	82.5	99%	96%	Leber congenital amaurosis 1, 204000 Cone-rod dystrophy 6, 601777
GUSB	61.9	89%	81%	Mucopolysaccharidosis VII, 253220
GYG1	45.5	83%	60%	Glycogen storage disease XV, 613507
GYS1	66.9	95%	82%	Glycogen storage disease 0, muscle, 611556
GYS2	92.1	100%	100%	Glycogen storage disease, type 0, 240600
H6PD	125.6	100%	100%	Cortisone reductase deficiency 1, 604931
HADH	91.5	100%	100%	3-hydroxyacyl-CoA dehydrogenase deficiency
HADHA	93.0	96%	87%	LCHAD deficiency, 609016 Trifunctional protein deficiency, 609015 HELLP syndrome, maternal, of pregnancy, 609016 Fatty liver, acute, of pregnancy, 609016
HADHB	94.9	100%	99%	Trifunctional protein deficiency, 609015
HAMP	95.8	100%	99%	Hemochromatosis, type 2B, 613313
HARS	119.1	100%	100%	Usher syndrome type 3B, 614504
HARS2	135.4	100%	100%	Perrault syndrome 2, 614926
HAX1	140.7	100%	100%	Neutropenia, severe congenital 3, autosomal recessive, 610738

HBA1	49.2	56%	49%	5'-zeta-pseudozeta-pseudoalpha-alpha-2-alpha-1-3' Thalassemias, alpha-, 604131 Methemoglobinemias, alpha- Erythremias, alpha- Heinz body anemias, alpha-, 140700 Hemoglobin H disease, nondeletional, 613978
HBA2	54.9	62%	49%	Thalassemia, alpha-, 604131 Heinz body anemia, 140700 Erythrocytosis Hypochromic microcytic anemia Hemoglobin H disease, nondeletional, 613978
HBB	151.6	100%	100%	Sickle cell anemia, 603903 Thalassemias, beta-, 613985 Erythremias, beta- Methemoglobinemias, beta- Heinz body anemias, beta-, 140700 Thalassemia-beta, dominant inclusion-body, 603902 Hereditary persistence of fetal hemoglobin, 141
HBD	208.3	100%	100%	Thalassemia, delta- Thalassemia due to Hb Lepore
HBG1	14.2	41%	21%	Fetal hemoglobin quantitative trait locus 1, 141749
HBG2	36.7	78%	65%	Fetal hemoglobin quantitative trait locus 1, 141749 Cyanosis, transient neonatal, 613977
HCCS	63.9	100%	96%	Microphthalmia, syndromic 7, 309801
HCFC1	35.7	93%	78%	Mental retardation, X-linked 3, 309541
HCN4	66.1	100%	95%	Sick sinus syndrome 2, 163800 Brugada syndrome 8, 613123
HCRT	55.8	93%	81%	Narcolepsy 1, 161400
HDAC4	70.2	95%	88%	Brachydactyly-mental retardation syndrome, 600430
HDAC6	58.0	92%	84%	Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863
HDAC8	47.9	99%	82%	Wilson-Turner syndrome, 309585 Cornelia de Lange syndrome 5, 300882
HEATR2	66.8	83%	78%	Ciliary dyskinesia, primary, 18, 614874
HEPACAM	70.4	89%	84%	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926

HERC2	62.9	63%	60%	[Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220
HES7	36.2	89%	76%	Spondylocostal dysostosis 4, autosomal recessive, 613686
HESX1	99.0	100%	99%	Septooptic dysplasia, 182230 Pituitary hormone deficiency, combined, 5, 182230 Growth hormone deficiency with pituitary anomalies, 182230
HEXA	90.7	100%	100%	Tay-Sachs disease, 272800 GM2-gangliosidosis, several forms, 272800 [Hex A pseudodeficiency], 272800
HEXB	114.8	100%	100%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HFE	101.2	100%	98%	Hemochromatosis, 235200 {Microvascular complications of diabetes 7}, 612635 {Porphyria variegata, susceptibility to}, 176200 {Porphyria cutanea tarda, susceptibility to}, 176100 {Alzheimer disease, susceptibility to}, 104300 [Transferr
HFE2	113.5	97%	96%	Hemochromatosis type 2A
HFM1	109.5	98%	98%	Split hand/foot malformation 1 (4)
HGD	89.4	100%	100%	Alkaptonuria, 203500
HGF	116.0	100%	100%	Deafness, autosomal recessive 39, 608265
HGSNAT	105.4	93%	93%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930
HIBCH	67.3	100%	99%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HINT1	80.6	96%	85%	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200
HK1	116.1	100%	100%	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285
HLCS	141.8	100%	100%	Holocarboxylase synthetase deficiency, 253270
HMBS	99.6	99%	98%	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000
HMGCL	103.5	100%	99%	HMG-CoA lyase deficiency, 246450
HMGCS2	114.9	100%	99%	HMG-CoA synthase-2 deficiency, 605911
HMOX1	65.9	100%	95%	Heme oxygenase-1 deficiency, 614034 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963
HMX1	37.6	99%	78%	Oculoauricular syndrome, 612109

HNF1A	81.4	98%	95%	MODY, type III, 600496 {Diabetes mellitus, noninsulin-dependent, 2}, 125853 {Diabetes mellitus, insulin-dependent}, 222100 Hepatic adenoma, somatic, 142330 Renal cell carcinoma, 144700 Diabetes mellitus, insulin-dependent, 20, 612520
HNF1B	78.7	98%	96%	Renal cysts and diabetes syndrome, 137920 Diabetes mellitus, noninsulin-dependent, 125853 {Renal cell carcinoma}, 144700
HNF4A	69.2	100%	93%	MODY, type I, 125850 {Diabetes mellitus, noninsulin-dependent}, 125853
HNRNPA1	46.4	92%	86%	?Inclusion body myopathy wtih early-onset Paget disease without frontotemporal dementia 3, 615424 Amyotrophic lateral sclerosis 19, 615426
HOGA1	72.6	100%	94%	Hyperoxaluria, primary, type III, 613616
HOXA1	142.1	100%	100%	Bosley-Salih-Alorainy syndrome, 601536 Athabaskan brainstem dysgenesis syndrome, 601536
HOXA11	113.8	100%	100%	Radioulnar synostosis with amegakaryocytic thrombocytopenia, 605432 -3
HOXA13	62.9	97%	69%	Hand-foot-uterus syndrome, 140000 Guttmacher syndrome, 176305 -3
HOXB1	100.4	100%	100%	Facial paresis, hereditary congenital, 3
HOXC13	86.5	99%	96%	Ectodermal dysplasia 9, hair/nail type, 614931
HOXD10	151.6	100%	100%	Vertical talus, congenital, 192950 Charcot-Marie-Tooth disease, foot deformity of, 192950
HOXD13	117.0	100%	98%	Synpolydactyly, type II, 186000 Brachydactyly, type E, 113300 Brachydactyly, type D, 113200 Synpolydactyly with foot anomalies, 186000 Syndactyly, type V, 186300 Brachydactyly-syndactyly syndrome, 610713 VACTERL association, 192350
HPD	98.3	100%	100%	Tyrosinemia, type III, 276710 Hawkinsinuria, 140350
HPGD	68.5	92%	76%	Cranioosteoarthropathy, 259100 Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100 Digital clubbing, isolated congenital, 119900

HPRT1	50.8	100%	78%	Lesch-Nyhan syndrome, 300322 HPRT-related gout, 300323
HPS1	75.8	96%	92%	Hermansky-Pudlak syndrome 1, 203300
HPS3	119.5	100%	100%	Hermansky-Pudlak syndrome 3, 614072
HPS4	107.2	98%	97%	Hermansky-Pudlak syndrome 4, 614073
HPS5	102.3	100%	100%	Hermansky-Pudlak syndrome 5, 614074
HPS6	85.2	100%	93%	Hermansky-Pudlak syndrome 6, 614075
HPSE2	77.2	100%	97%	Urofacial syndrome 1, 236730
HR	84.8	98%	93%	Alopecia universalis, 203655 Atrichia with papular lesions, 209500 Hypotrichosis, hereditary, Marie Unna type, 1, 146550
HRAS	87.1	100%	99%	{Bladder cancer, somatic}, 109800 Costello syndrome, 218040 {Thyroid carcinoma, follicular, somatic}, 188470 Congenital myopathy with excess of muscle spindles, 218040 {Nevus sebaceous, somatic}, 162900 Schimmelpenning-Feuerstein-Mims s
HRG	141.4	94%	94%	Thrombophilia due to HRG deficiency, 613116 Thrombophilia due to elevated HRG, 613116 (1)
HSD11B1	109.1	100%	100%	Cortisone reductase deficiency 2, 614662
HSD11B2	113.2	78%	78%	Apparent mineralocorticoid excess, 218030
HSD17B10	54.1	95%	91%	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 Mental retardation, X-linked syndromic 10, 300220 Mental retardation, X-linked 17/31, microduplication, 300705
HSD17B3	105.0	99%	96%	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	98.2	100%	99%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B2	44.0	93%	78%	3-beta-hydroxysteroid dehydrogenase, type II, deficiency, 201810
HSD3B7	72.7	92%	82%	Bile acid synthesis defect, congenital, 1, 607765
HSF4	100.0	100%	98%	Cataract 5, multiple types, 116800
HSPB1	50.1	98%	80%	Neuropathy, distal hereditary motor, type IIB, 608634 Charcot-Marie-Tooth disease, axonal, type 2F, 606595
HSPB3	208.7	100%	100%	Neuronopathy, distal hereditary motor, type IIC, 613376
HSPB8	98.5	100%	100%	Neuropathy, distal hereditary motor, type IIA, 158590 Charcot-Marie-Tooth disease, axonal, type 2L, 608673
HSPD1	14.8	61%	36%	Spastic paraplegia 13, autosomal dominant, 605280

				Leukodystrophy, hypomyelinating, 4, 612233
HSPG2	73.3	98%	93%	Schwartz-Jampel syndrome, type 1, 255800 Dyssegmental dysplasia, Silverman-Handmaker type, 224410
HTR1A	121.1	100%	100%	Periodic fever, menstrual cycle dependent, 614674
HTRA1	69.0	84%	78%	{Macular degeneration, age-related, 7}, 610149 {Macular degeneration, age-related, neovascular type}, 610149 CARASIL syndrome, 600142
HTRA2	141.2	100%	99%	Parkinson disease 13, 610297
HTT	99.8	99%	97%	Huntington disease, 143100
HUWE1	48.2	96%	86%	Mental retardation, X-linked syndromic, Turner type, 300706
HYAL1	97.4	99%	96%	Mucopolysaccharidosis type IX, 601492
HYDIN	93.5	88%	85%	Ciliary dyskinesia, primary, 5, 608647
HYLS1	154.2	100%	100%	Hydrolethalus syndrome, 236680
ICK	103.8	100%	100%	Endocrine-cerebroosteodysplasia, 612651
ICOS	133.8	100%	100%	Immunodeficiency, common variable, 1, 607594
IDH2	106.8	100%	94%	D-2-hydroxyglutaric aciduria 2, 613657
IDH3B	116.8	95%	95%	Retinitis pigmentosa 46, 612572
IDS	55.1	84%	77%	Mucopolysaccharidosis II, 309900
IDUA	86.0	95%	84%	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Is, 607016 Mucopolysaccharidosis Ih/s, 607015
IER3IP1	67.1	100%	99%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFITM5	95.2	100%	86%	Osteogenesis imperfecta, type V, 610967
IFNGR1	145.7	100%	100%	Mycobacterial infection, atypical, familial disseminated, 209950 BCG infection, generalized familial, 209950 {H. pylori infection, susceptibility to}, 600263 {Tuberculosis, susceptibility to}, 607948 {Mycobacterium tuberculosis infection,
IFT122	79.1	96%	95%	Cranioectodermal dysplasia 1, 218330
IFT140	83.3	99%	95%	Mainzer-Saldino syndrome, 266920
IFT172	97.6	100%	97%	Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 Mainzer-Saldino syndrome (Halbritter (2013) Am J Hum Genet 93, 915) Asphyxiating thoracic dystrophy with or without Joubert Syndrome (Halbritter (2013) Am J Hum Genet 93, 915)
IFT43	84.7	100%	100%	Cranioectodermal dysplasia 3, 614099
IFT80	84.4	100%	93%	Asphyxiating thoracic dystrophy 2, 611263

IGBP1	49.0	84%	74%	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472
IGF1	132.1	100%	100%	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IGF1R	104.7	100%	99%	Insulin-like growth factor I, resistance to, 270450
IGF2R	93.7	98%	95%	Hepatocellular carcinoma
IGFALS	68.9	100%	98%	Acid-labile subunit, deficiency of
IGFBP7	36.0	61%	32%	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis, 614224
IGHMBP2	70.4	97%	88%	Neuronopathy, distal hereditary motor, type VI, 604320
IGLL1	22.0	72%	39%	Agammaglobulinemia 2, 613500
IGSF1	59.9	98%	92%	Hypothyroidism, central, and testicular enlargement, 300888
IHH	104.4	100%	100%	Acrocapitofemoral dysplasia, 607778 Brachydactyly, type A1, 112500
IKBKAP	106.1	100%	99%	Dysautonomia, familial, 223900
IKBKB	91.0	98%	94%	Immunodeficiency 15, 615592 (3)
IKBKG	11.4	23%	22%	Incontinentia pigmenti, type II, 308300 Ectodermal dysplasia, hypohidrotic, with immune deficiency, 300291 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 Immunodeficiency, isolated, 300584 {Atypical mycobacterio
IKZF1	101.4	100%	99%	Leukemia, acute lymphoblastic
IL10RA	99.1	100%	100%	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	111.2	98%	94%	Inflammatory bowel disease 25 early onset autosomal recessive
IL11RA	84.6	100%	96%	Craniosynostosis and dental anomalies, 614188
IL17F	90.0	97%	93%	Candidiasis, familial, 6, autosomal dominant, 613956
IL17RA	90.8	100%	92%	Candidiasis, familial, 5, autosomal recessive, 613953
IL17RD	99.5	98%	96%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
IL1RAPL1	68.7	100%	99%	Mental retardation, X-linked 21/34, 300143
IL1RN	106.9	100%	100%	{Gastric cancer risk after H. pylori infection}, 137215 {Microvascular complications of diabetes 4}, 612628 Interleukin 1 receptor antagonist deficiency, 612852
IL21R	122.5	100%	99%	[IgE, elevated level of], 147050 Immunodeficiency, primary, autosomal recessive, IL21R-related, 615207
IL2RA	101.4	100%	99%	Interleukin-2 receptor, alpha chain, deficiency of, 606367 {Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942

IL2RG	46.6	98%	87%	Severe combined immunodeficiency, X-linked, 300400 Combined immunodeficiency, X-linked, moderate, 312863
IL31RA	120.5	100%	96%	Amyloidosis, primary localized cutaneous, 2, 613955
IL36RN	98.7	100%	100%	Psoriasis, generalized pustular, 614204
IL7R	99.3	100%	100%	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
ILDR1	57.4	100%	99%	Deafness, autosomal recessive 42, 609646
IMPAD1	130.4	100%	100%	Chondrodysplasia with joint dislocations, GRAPP type, 614078
IMPDH1	35.8	83%	60%	Retinitis pigmentosa 10, 180105 Leber congenital amaurosis 11, 613837
IMPG2	125.8	99%	97%	Retinitis pigmentosa 56, 613581 Maculopathy, IMPG2-related, 613581
INF2	67.9	93%	85%	Glomerulosclerosis, focal segmental, 5, 613237 Charcot-Marie-Tooth disease, dominant intermediate E, 614455
ING1	126.1	100%	98%	Squamous cell carcinoma, head and neck, somatic, 275355
INPP5E	74.2	100%	99%	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300
INPPL1	94.9	98%	95%	Opsismodysplasia, 258480
INS	42.2	100%	85%	Hyperproinsulinemia, familial, with or without diabetes Maturity-onset diabetes of the young, type 10, 613370 Diabetes mellitus, permanent neonatal, 606176 Diabetes mellitus, type 1, 125852 Diabetes mellitus, insulin-dependent, 2, 125852
INSL3	43.2	88%	85%	Cryptorchidism, 219050
INSR	120.8	96%	93%	Leprechaunism, 246200 Rabson-Mendenhall syndrome, 262190 Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968
INVS	124.2	100%	99%	Nephronophthisis 2, infantile, 602088
IQCB1	95.1	99%	94%	Senior-Loken syndrome 5, 609254
IQSEC2	40.8	84%	67%	Mental retardation, X-linked 1, 309530
IRAK4	103.5	100%	100%	IRAK4 deficiency, 607676 Invasive pneumococcal disease, recurrent isolated, 1, 610799

IRF1	103.3	100%	100%	Myelodysplastic syndrome, preleukemic Myelogenous leukemia, acute Gastric cancer, somatic, 613659 Nonsmall cell lung cancer, somatic, 211980
IRF4	110.2	100%	100%	Multiple myeloma, 254500
IRF6	101.5	99%	94%	van der Woude syndrome, 119300 Popliteal pterygium syndrome 1, 119500 Orofacial cleft 6, 608864
IRF8	69.1	100%	98%	Monocyte and dendritic cell deficiency, recessive, 614894 CD11C+/CD1C+ dendritic cell deficiency, dominant, 614893
IRGM	162.7	100%	100%	{Mycobacterium tuberculosis, protection against}, 607948 Inflammatory bowel disease 19, 612278
IRX5	56.3	97%	88%	Hamamy syndrome, 611174
ISCU	85.5	100%	96%	Myopathy with lactic acidosis, hereditary, 255125
ISPD	79.5	93%	90%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
ITCH	98.4	95%	95%	Autoimmune disease, syndromic multisystem, 613385
ITGA2B	71.7	97%	91%	BAK platelet antigen Glanzmann thrombasthenia, 273800 Thrombocytopenia, neonatal alloimmune, BAK antigen related Bleeding disorder, platelet-type, 16, autosomal dominant, 187800
ITGA3	107.4	100%	94%	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748
ITGA6	133.2	100%	98%	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730 -3
ITGA7	86.0	98%	93%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
ITGA8	93.6	99%	98%	Renal hypodysplasia/aplasia 1, 191830 (3)
ITGB2	84.1	100%	98%	Leukocyte adhesion deficiency, 116920
ITGB3	91.8	99%	98%	PL(A) platelet antigen Glanzmann thrombasthenia, 273800 Thrombocytopenia, neonatal alloimmune {Myocardial infarction, susceptibility to}, 608446 Purpura, posttransfusion Bleeding disorder, platelet-type, 16, autosomal dominant, 187800
ITGB4	82.1	97%	92%	Epidermolysis bullosa, junctional, with pyloric atresia, 226730 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa of hands and feet, 131800
ITK	100.2	100%	100%	Lymphoproliferative syndrome 1, 613011
ITM2B	99.6	100%	100%	Dementia, familial British, 176500 Dementia, familial Danish, 117300

ITPR1	107.1	99%	98%	Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360
IVD	96.0	100%	96%	Isovaleric acidemia, 243500
IYD	78.4	100%	99%	Thyroid dyshormonogenesis 4, 274800
JAG1	110.2	99%	96%	Alagille syndrome, 118450 Tetralogy of Fallot, 187500 Deafness, congenital heart defects, and posterior embryotoxon
JAK2	115.9	99%	98%	Polycythemia vera, 263300 Thrombocythemia 3, 614521 Myelofibrosis, somatic, 254450 {Budd-Chiari syndrome}, 600880 Leukemia, acute myelogenous, 601626 Erythrocytosis, somatic, 133100
JAK3	89.3	99%	96%	SCID, autosomal recessive, T-negative/B-positive type, 600802
JAM3	69.8	94%	89%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
JPH2	67.6	100%	94%	Cardiomyopathy, familial hypertrophic 17, 613873
JPH3	109.9	97%	97%	Huntington disease-like 2, 606438
JUP	78.6	99%	95%	Naxos disease, 601214 Arrhythmogenic right ventricular dysplasia 12, 611528
KAL1	47.6	93%	81%	hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
KANK1	137.4	100%	99%	Cerebral palsy, spastic quadriplegic, 2, 612900
KANSL1	49.7	78%	65%	Koolen-De Vries syndrome, 610443
KARS	116.2	100%	100%	Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness, autosomal recessive 89, 613916
KAT6B	148.4	100%	100%	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KBTBD13	54.0	100%	96%	Nemaline myopathy 6, autosomal dominant, 609273
KCNA1	112.2	100%	100%	Episodic ataxia/myokymia syndrome, 160120
KCNA5	152.0	99%	98%	Atrial fibrillation, familial, 7, 612240
KCNC3	78.5	76%	68%	Spinocerebellar ataxia 13, 605259
KCND3	128.2	99%	97%	Spinocerebellar ataxia 19, 607346
KCNE1	206.9	100%	100%	Jervell and Lange-Nielsen syndrome 2, 612347 Long QT syndrome-5, 613695
KCNE2	132.4	100%	100%	Long QT syndrome-6, 613693 Atrial fibrillation, familial, 4, 611493

KCNE3	90.8	100%	100%	Brugada syndrome 6, 613119
KCNH2	64.4	94%	85%	Long QT syndrome-2, 613688 {Long QT syndrome-2, acquired, susceptibility to}, 613688 Short QT syndrome-1, 609620
KCNJ1	146.1	97%	97%	Bartter syndrome, type 2, 241200
KCNJ10	151.4	100%	100%	SESAME syndrome, 612780 Enlarged vestibular aqueduct, digenic, 600791
KCNJ11	144.0	100%	100%	Hyperinsulinemic hypoglycemia, familial, 2, 601820 Diabetes, permanent neonatal, 606176 Diabetes mellitus, permanent neonatal, with neurologic features, 606176 {Diabetes mellitus, type 2, susceptibility to}, 125853 Diabetes mellitus, trans
KCNJ13	211.1	100%	100%	Snowflake vitreoretinal degeneration, 193230 Leber congenital amaurosis 16, 614186
KCNJ2	110.4	97%	92%	Andersen syndrome, 170390 Short QT syndrome-3, 609622 Atrial fibrillation, familial, 9, 613980
KCNJ5	175.5	100%	100%	Long QT syndrome 13, 613485 Hyperaldosteronism, familial, type III, 613677
KCNK3	105.2	96%	92%	Pulmonary hypertension, primary, 4, 615344
KCNMA1	75.8	94%	89%	Generalized epilepsy and paroxysmal dyskinesia, 609446
KCNQ1	62.5	93%	81%	Long QT syndrome-1, 192500 Jervell and Lange-Nielsen syndrome, 220400 Atrial fibrillation, familial, 3, 607554 Short QT syndrome-2, 609621 {Long QT syndrome 1, acquired, susceptibility to}, 192500
KCNQ2	76.3	100%	98%	Seizures, benign neonatal, 1, 121200 Myokymia, 121200 Epileptic encephalopathy, early infantile, 7, 613720
KCNQ3	100.3	100%	98%	Seizures, benign neonatal, type 2, 121201
KCNQ4	111.9	94%	90%	Deafness, autosomal dominant 2A, 600101
KCNT1	77.9	99%	93%	Epileptic encephalopathy, early infantile, 14, 614959 Epilepsy, nocturnal frontal lobe, 5, 615005
KCNV2	84.3	100%	99%	Retinal cone dystrophy 3B, 610356
KCTD1	121.8	99%	97%	Scalp-ear-nipple syndrome, 181270
KCTD7	106.4	92%	87%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726

KDM5C	53.9	100%	93%	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534 -3
KDM6A	64.4	99%	92%	Kabuki syndrome 2, 300867
KDR	110.4	100%	99%	Hemangioma, capillary infantile, somatic, 602089 {Hemangioma, capillary infantile, susceptibility to}, 602089
KERA	145.8	100%	100%	Cornea plana congenita, recessive, 217300
KHDC3L	119.4	100%	100%	Hydatidiform mole, recurrent, 2, 614293
KIAA0196	102.3	99%	98%	Spastic paraplegia 8, autosomal dominant, 603563
KIAA1279	105.1	99%	95%	Goldberg-Shprintzen megacolon syndrome, 609460
KIAA2022	78.3	100%	99%	?Mental retardation, nonsyndromic, X-linked (2)
KIF11	98.6	99%	96%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF1A	71.6	98%	92%	Spastic paraplegia 30, autosomal recessive, 610357 Neuropathy, hereditary sensory, type IIC, 614213 Mental retardation, autosomal dominant 9, 614255
KIF1B	128.9	100%	100%	Charcot-Marie-Tooth disease, type 2A1, 118210 Pheochromocytoma, 171300 {Neuroblastoma, susceptibility to, 1}, 256700
KIF1C	115.5	100%	96%	Spastic ataxia 2, autosomal recessive, 611302
KIF21A	108.2	100%	98%	Fibrosis of extraocular muscles, congenital, 1, 135700 Fibrosis of extraocular muscles, congenital, 3B, 135700
KIF22	104.9	100%	97%	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546
KIF2A	123.4	100%	97%	Cortical dysplasia, complex, with other brain malformations 3, 615411
KIF5A	94.4	99%	98%	Spastic paraplegia 10, autosomal dominant, 604187
KIF7	67.0	93%	86%	Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990
KIRREL3	75.7	98%	91%	Mental retardation, autosomal dominant 4, 612581
KISS1	36.9	89%	69%	Hypogonadotropic hypogonadism 13 with or without anosmia, 614842
KISS1	36.9	89%	69%	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 Precocious puberty, central, 1, 176400
KISS1R	37.5	99%	90%	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 Precocious puberty, central, 1, 176400
KIT	110.8	100%	96%	Piebaldism, 172800 Gastrointestinal stromal tumor, familial, 606764 Mast cell disease, 154800 Leukemia, acute myeloid, 601626

				Germ cell tumors, 273300
KITLG	80.0	100%	99%	[Skin/hair/eye pigmentation 7, blond/brown hair], 611664 Hyperpigmentation, familial progressive, 2, 145250
KL	139.6	97%	96%	{Coronary artery disease, susceptibility to} Tumoral calcinosis, hyperphosphatemic, 211900
KLF1	47.6	100%	99%	Blood group--Lutheran inhibitor, 111150 [Hereditary persistence of fetal hemoglobin], 613566 Anemia, dyserythropoietic congenital, type IV, 613673
KLF11	164.4	97%	97%	Maturity-onset diabetes of the young, type VII, 610508
KLF6	150.2	100%	100%	Prostate cancer, somatic, 176807 Gastric cancer, somatic, 613659
KLHDC8B	58.3	97%	72%	Hodgkin lymphoma, 236000
KLHL10	131.3	100%	100%	Spermatogenic failure 11, 615081
KLHL3	85.5	99%	91%	Pseudohypoaldosteronism, type IID, 614495
KLHL40	73.8	100%	100%	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	147.8	100%	100%	Nemaline myopathy 9, 615731 (3)
KLHL7	118.9	100%	100%	Retinitis pigmentosa 42, 612943
KLK4	146.3	100%	100%	Amelogenesis imperfecta, type IIA1, 204700
KLKB1	128.4	100%	100%	Fletcher factor deficiency, 612423
KLLN	104.9	100%	100%	Cowden syndrome 4, 615107
KMT2A	142.0	99%	98%	Wiedemann-Steiner syndrome
KMT2D	100.9	99%	98%	Kabuki syndrome 1
KPTN	83.5	100%	95%	Mental retardation, autosomal recessive 41, 615637 (3)
KRAS	65.6	100%	88%	Lung cancer, somatic, 211980 Bladder cancer, somatic, 109800 Pancreatic carcinoma, somatic, 260350 Gastric cancer, somatic, 137215 Leukemia, acute myelogenous Noonan syndrome 3, 609942 Cardiofaciocutaneous syndrome 2, 615278 Br
KRIT1	105.0	100%	99%	Cavernous malformations of CNS and retina

KRT1	100.4	100%	100%	Epidermolytic hyperkeratosis, 113800 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602 Ichthyosis histrix, Curth-Macklin type, 146590 Palmoplantar keratoderma, nonepidermolytic, 600962 Palmoplantar keratoderma, epidermolytic, 1
KRT10	96.5	89%	88%	Epidermolytic hyperkeratosis, 113800 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602 Ichthyosis with confetti, 609165
KRT12	108.2	98%	95%	Meesmann corneal dystrophy, 122100
KRT13	86.7	100%	99%	White sponge nevus, 193900
KRT14	28.2	74%	44%	Epidermolysis bullosa simplex, Dowling-Meara type, 131760 Epidermolysis bullosa simplex, Koebner type, 131900 Epidermolysis bullosa simplex, recessive 1, 601001 Naegeli-Franceschetti-Jadassohn syndrome, 161000 Dermatopathia pigmentosa reti
KRT16	6.4	25%	7%	Pachyonychia congenita, Jadassohn-Lewandowsky type, 167200 Palmoplantar keratoderma, nonepidermolytic, focal, 613000
KRT17	7.4	30%	5%	Pachyonychia congenita, Jackson-Lawler type, 167210 Steatocystoma multiplex, 184500
KRT18	20.4	79%	40%	Cirrhosis, cryptogenic {Cirrhosis, noncryptogenic, susceptibility to}, 215600
KRT2	117.3	99%	97%	Ichthyosis bullosa of Siemens, 146800
KRT3	70.1	100%	100%	Meesmann corneal dystrophy, 122100
KRT4	81.4	100%	95%	White sponge nevus, 193900
KRT5	73.3	100%	98%	Epidermolysis bullosa simplex, Dowling-Meara type, 131760 Epidermolysis bullosa simplex, Koebner type, 131900 Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 Epidermolysis bullosa simplex with mottled pigmentation, 131960 Dowlin
KRT6A	32.5	64%	44%	Pachyonychia congenita, Jadassohn-Lewandowsky type, 167200
KRT6B	33.8	66%	49%	Pachyonychia congenita, Jackson-Lawler type, 167210
KRT6C	22.7	50%	35%	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735
KRT74	86.2	96%	90%	Woolly hair, autosomal dominant, 194300 Hypotrichosis simplex of the scalp 2, 613981
KRT8	31.1	90%	60%	Cirrhosis, cryptogenic {Cirrhosis, noncryptogenic, susceptibility to}, 215600
KRT81	20.4	63%	43%	Monilethrix, 158000

KRT83	26.1	75%	42%	Monilethrix, 158000
KRT85	37.9	77%	57%	Ectodermal dysplasia 4, hair/nail type, 602032
KRT86	24.8	63%	42%	Monilethrix, 158000
KRT9	102.4	96%	90%	Palmoplantar keratoderma, epidermolytic, 144200
L1CAM	62.0	98%	92%	Hydrocephalus due to aqueductal stenosis, 307000 MASA syndrome, 303350 CRASH syndrome, 303350 Hydrocephalus with Hirschsprung disease, 307000 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 Corpus callosum
L2HGDH	81.2	100%	97%	L-2-hydroxyglutaric aciduria, 236792
LAMA2	103.3	100%	98%	Muscular dystrophy, congenital merosin-deficient, 607855 Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855
LAMA4	101.2	100%	99%	Cardiomyopathy dilated 1JJ
LAMB1	115.7	100%	99%	Lissencephaly 5, 615191
LAMB2	111.3	100%	99%	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 Pierson syndrome, 609049
LAMB3	79.0	98%	93%	Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LAMC2	100.0	100%	99%	Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LAMC3	95.8	99%	94%	Cortical malformations, occipital, 614115
LAMP2	61.4	98%	90%	Danon disease, 300257
LAMTOR2	80.1	100%	100%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798 -3
LARGE	108.7	99%	96%	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	118.1	100%	100%	Alazami syndrome, 615071
LARS2	116.2	100%	99%	Perrault syndrome 4, 615300
LBR	100.1	100%	100%	Pelger-Huet anomaly, 169400 HEM skeletal dysplasia, 215140 Reynolds syndrome, 613471
LCA5	142.1	100%	98%	Leber congenital amaurosis 5, 604537
LCAT	107.1	93%	88%	Norum disease, 245900 Fish-eye disease, 136120

LCT	130.0	100%	99%	Lactase deficiency, congenital, 223000
LDB3	87.3	95%	93%	Myopathy, myofibrillar, 4, 609452 Cardiomyopathy, dilated 1C, 601493 Left ventricular noncompaction 3, with or without dilated cardiomyopathy, 601493
LDHA	45.9	76%	58%	Glycogen storage disease XI, 612933
LDHB	89.5	100%	100%	Lactate dehydrogenase-B deficiency, 614128
LDLR	108.2	100%	99%	C3 Hypercholesterolemia, familial, 143890 LDL cholesterol level QTL2, 143890
LDLRAP1	84.7	100%	96%	Hypercholesterolemia, familial, autosomal recessive, 603813
LEF1	91.0	100%	99%	Sebaceous tumors, somatic
LEFTY2	52.2	86%	66%	-
LEMD3	110.4	100%	100%	Osteopoikilosis, 166700 Buschke-Ollendorff syndrome, 166700 Melorheostosis with osteopoikilosis, 155950
LEP	105.3	100%	100%	Obesity, morbid, due to leptin deficiency, 614962
LEPR	124.4	94%	93%	Obesity, morbid, due to leptin receptor deficiency, 614963
LEPRE1	91.5	100%	99%	Osteogenesis imperfecta, type VIII, 610915
LEPREL1	70.5	99%	88%	Myopia, high, with cataract and vitreoretinal degeneration, 614292 -3
LFNG	58.5	84%	79%	Spondylocostal dysostosis, autosomal recessive 3, 609813
LGI1	136.6	100%	100%	Epilepsy, familial temporal lobe, 1, 600512
LHB	16.6	57%	50%	Hypogonadism, hypergonadotropic ?Male pseudohermaphroditism due to defective LH (1)
LHCGR	147.6	99%	94%	Precocious puberty, male, 176410 Leydig cell hypoplasia with pseudohermaphroditism, 238320 Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320 Luteinizing hormone resistance, female, 238320 Leydig cell adenoma, somatic, with
LHFPL5	176.4	100%	100%	Deafness, autosomal recessive 67, 610265
LHX3	46.2	100%	90%	Pituitary hormone deficiency, combined, 3, 221750
LHX4	88.9	100%	100%	Pituitary hormone deficiency, combined, 4, 262700
LIAS	100.3	100%	100%	Pyruvate dehydrogenase lipoic acid synthetase deficiency, 614462
LIFR	114.5	98%	97%	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LIG1	77.8	99%	93%	DNA ligase I deficiency

LIG4	190.0	100%	100%	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500 Severe combined immunodeficiency with sensitivity to ionizing radiation, 602450
LIM2	70.4	82%	81%	Cataract 19, 615277
LINS	124.2	100%	100%	Mental retardation, autosomal recessive 27, 614340 (3)
LIPA	109.4	100%	100%	? Wolman disease, 278000 Cholesteryl ester storage disease, 278000 -3
LIPC	84.0	100%	95%	[High density lipoprotein cholesterol level QTL 12], 612797 {Diabetes mellitus, noninsulin-dependent}, 125853 Hepatic lipase deficiency, 614025
LIPH	122.7	100%	100%	Hypotrichosis, localized, autosomal recessive 2, 604379 Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379
LIPN	124.7	100%	94%	Ichthyosis, congenital, autosomal recessive 8, 613943
LITAF	88.6	100%	100%	Charcot-Marie-Tooth disease, type 1C, 601098
LMAN1	125.5	100%	100%	Combined factor V and VIII deficiency, 227300
LMBR1	100.8	100%	98%	Acheiropody, 200500 Polydactyly, preaxial type II, 174500 Triphalangeal thumb, type I, 174500 Triphalangeal thumb-polysyndactyly syndrome, 174500 Syndactyly, type IV, 186200
LMBRD1	108.9	100%	100%	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LMF1	94.1	100%	98%	Lipase deficiency, combined, 246650
LMNA	71.1	97%	89%	Emery-Dreifuss muscular dystrophy 2, AD, 181350 Cardiomyopathy, dilated, 1A, 115200 Lipodystrophy, familial partial, 2, 151660 Emery-Dreifuss muscular dystrophy 3, AR, 181350 Charcot-Marie-Tooth disease, type 2B1, 605588 Muscular dystr
LMNB1	78.8	96%	85%	Leukodystrophy, adult-onset, autosomal dominant, 169500
LMX1B	86.7	100%	98%	Nail-patella syndrome, 161200
LOR	48.0	100%	95%	Vohwinkel syndrome with ichthyosis, 604117
LOXHD1	103.8	100%	99%	Deafness, autosomal recessive 77, 613079
LPAR6	131.1	100%	100%	Hypotrichosis 8, 278150 Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150
LPIN1	100.3	100%	99%	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	75.5	100%	97%	Majeed syndrome, 609628

LPL	109.4	100%	100%	Lipoprotein lipase deficiency, 238600 Combined hyperlipidemia, familial, 144250 [High density lipoprotein cholesterol level QTL 11]
LPP	133.5	100%	99%	Leukemia, acute myeloid, 601626
LRAT	196.5	100%	100%	Retinal dystrophy, early-onset severe, 613341 Leber congenital amaurosis 14, 613341 Retinitis pigmentosa, juvenile, 613341
LRBA	111.3	99%	98%	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRIG2	124.0	100%	98%	Urofacial syndrome 2, 615112
LRIT3	140.3	94%	94%	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRP2	107.4	100%	99%	Donnai-Barrow syndrome, 222448
LRP4	93.9	99%	96%	Cenani-Lenz syndactyly syndrome, 212780 Sclerosteosis 2, 614305
LRP5	89.4	98%	94%	Osteoporosis-pseudoglioma syndrome, 259770 [Bone mineral density variability 1], 601884 Hyperostosis, endosteal, 144750 van Buchem disease, type 2, 607636 Osteosclerosis, 144750 {Osteoporosis}, 166710 Exudative vitreoretinopathy 4,
LRPAP1	85.4	100%	96%	Myopia 23, autosomal recessive, 615431
LRPPRC	100.6	98%	96%	Leigh syndrome, French-Canadian type, 220111
LRRC6	112.2	100%	100%	Ciliary dyskinesia, primary, 19, 614935
LRRC8A	121.1	100%	100%	Agammaglobulinemia 5, 613506
LRRK2	112.8	99%	99%	Parkinson disease 8, 607060
LRSAM1	81.9	100%	97%	Charcot-Marie-Toothe disease, axonal, type 2P, 614436
LRTOMT	92.6	93%	90%	Deafness, autosomal recessive 63, 611451
LTBP2	71.1	98%	94%	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	72.8	99%	95%	Tooth agenesis, selective, 6, 613097
LTBP4	89.2	97%	90%	Cutis laxa, autosomal recessive, type IC, 613177
LYST	120.5	99%	96%	Chediak-Higashi syndrome
LYZ	108.4	100%	100%	Amyloidosis, renal, 105200
LZTFL1	90.2	100%	100%	Bardet-Biedl syndrome 17, 615994

LZTS1	142.2	100%	97%	Esophageal squamous cell carcinoma
MAD1L1	69.4	96%	92%	Lymphoma, somatic Prostate cancer, somatic, 176807
MAF	79.8	81%	75%	Cataract, pulverulent or cerulean, with or without microcornea, 610202
MAFB	87.4	100%	100%	Multicentric carpotarsal osteolysis syndrome, 166300
MAGEL2	84.1	95%	91%	Prader-Willi-like syndrome, 615547
MAGT1	54.0	100%	100%	Mental retardation, X-linked 95, 300716 Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853
MAK	85.9	96%	95%	REtinitis pigmentosa 62, 614181
MAML2	110.0	100%	100%	Mucoepidermoid salivary gland carcinoma
MAMLD1	63.9	100%	100%	Hypospadias 2, X-linked, 300758
MAN1B1	96.6	100%	99%	Mental retardation, autosomal recessive 15, 614202
MAN2B1	80.2	97%	92%	Mannosidosis, alpha-, types I and II, 248500
MANBA	92.3	100%	99%	Mannosidosis, beta, 248510
MAOA	53.7	100%	95%	Brunner syndrome, 300615
MAP2K1	103.7	95%	88%	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	101.9	100%	98%	Cardiofaciocutaneous syndrome 4, 615280
MAP3K1	114.5	97%	95%	46XY sex reversal 6, 613762
MAP3K8	127.6	100%	99%	Lung cancer, somatic, 211980
MAPT	21.1	57%	37%	Dementia, frontotemporal, with or without parkinsonism, 600274 Pick disease, 172700 Supranuclear palsy, progressive, 601104 Supranuclear palsy, progressive atypical, 260540 {Parkinson disease, susceptibility to}, 168600 Tauopathy and r
MARS2	156.2	100%	100%	Spastic ataxia 3, autosomal recessive, 611390
MARVELD2	146.7	98%	95%	Deafness, autosomal recessive 49, 610153
MASP1	112.0	100%	99%	3MC syndrome 1, 257920
MASP2	111.7	98%	92%	MASP2 deficiency, 613791
MASTL	127.7	100%	100%	Thrombocytopenia-2, 188000
MAT1A	94.5	100%	99%	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850

MATN3	96.9	93%	84%	Epiphyseal dysplasia, multiple, 5, 607078 {Osteoarthritis susceptibility 2}, 140600 Spondyloepimetaphyseal dysplasia, 608728
MATR3	115.3	95%	93%	Myopathy, distal 2, 606070
MBD5	140.8	100%	99%	Mental retardation, autosomal dominant 1, 156200
MBTPS2	71.3	100%	98%	IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800
MC2R	149.2	100%	100%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MC4R	173.9	100%	100%	Obesity, autosomal dominant, 601665
MCC	88.2	99%	97%	Colorectal cancer
MCCC1	97.0	99%	98%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	113.1	98%	92%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	79.6	100%	100%	Methylmalonyl-CoA epimerase deficiency, 251120
MCFD2	59.5	100%	96%	Factor V and factor VIII, combined deficiency of, 613625
MCM4	105.2	99%	97%	Natural killer cell and glucocorticoid deficiency with DNA repair defect, 609981
MCM6	108.7	100%	97%	Lactase persistance/nonpersistance, 223100
MCOLN1	96.7	97%	92%	Mucolipidosis IV, 252650
MCPH1	122.8	100%	100%	Microcephaly 1, primary, autosomal recessive, 251200
MECOM	123.7	100%	98%	Myelodysplasia syndrome-1
MECP2	89.1	97%	90%	Rett syndrome, 312750 Mental retardation, X-linked, syndromic 13, 300055 Rett syndrome, preserved speech variant, 312750 Encephalopathy, neonatal severe, 300673 {Autism susceptibility, X-linked 3}, 300496 Angelman syndrome, 105830
MED12	62.1	94%	85%	Opitz-Kaveggia syndrome, 305450 Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895
MED13L	116.1	100%	99%	Transposition of the great arteries, dextro-looped 1, 608808
MED17	149.7	100%	97%	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	113.6	100%	100%	Mental retardation, autosomal recessive 18, 614249
MED25	98.4	99%	87%	Charcot-Marie-Tooth disease, type 2B2, 605589
MEF2C	117.4	100%	100%	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443 Chromosome 5q14.3 deletion syndrome, 613443 (4)

MEFV	116.1	100%	100%	Familial Mediterranean fever, AR, 249100 Familial Mediterranean fever, AD, 134610
MEGF10	107.7	100%	99%	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399
MEGF8	90.5	99%	93%	Carpenter syndrome 2, 614976
MEN1	111.0	99%	96%	Multiple endocrine neoplasia 1, 131100 Carcinoid tumor of lung Parathyroid adenoma, somatic Lipoma, somatic Angiofibroma, somatic Adrenal adenoma, somatic
MEOX1	74.6	100%	99%	Klippel-Feil syndrome 2, 214300
MERTK	114.9	100%	99%	Retinitis pigmentosa 38, 613862
MESP2	72.8	100%	100%	Spondylocostal dysostosis, autosomal recessive 2, 608681
MET	124.5	100%	100%	Renal cell carcinoma, papillary, familial and somatic, 605074 Hepatocellular carcinoma, childhood type, 114550 {Autism suseptibility 9}, 611015
MFN2	102.2	100%	97%	Charcot-Marie-Tooth disease, type 2A2, 609260 Hereditary motor and sensory neuropathy VI, 601152
MFRP	87.8	100%	99%	Microphthalmia, isolated 5, 611040 Nanophthalmos 2, 609549
MFSD8	120.0	100%	100%	Ceroid lipofuscinosis, neuronal, 7, 610951
MGAT2	217.2	100%	100%	Congenital disorder of glycosylation, type IIa, 212066
MGME1	159.2	100%	100%	Mitochondrial DNA depletion syndrome 11, 615084
MGP	72.8	100%	98%	Keutel syndrome, 245150 {Natural teeth remaining intact} (2)
MIB1	107.6	100%	100%	Left ventricular noncompaction 7, 615092
MICU1	94.9	100%	98%	Myopathy with extrapyramidal signs, 615673 (3)
MID1	93.3	99%	96%	Opitz GBBB syndrome, type I, 300000
MINPP1	165.5	100%	100%	Thyroid carcinoma, follicular, 188470
MIP	76.1	100%	97%	Cataract 15, multiple types, 615274
MITF	131.8	100%	100%	Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 Tietz albinism-deafness syndrome, 103500 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456

MKKS	148.8	100%	100%	McKusick-Kaufman syndrome, 236700 Bardet-Biedl syndrome 6, 209900
MKL1	70.5	97%	91%	Megakaryoblastic leukemia, acute
MKRN3	119.1	100%	100%	Precocious puberty, central, 2, 615346
MKS1	110.6	100%	99%	Meckel syndrome 1, 249000 Bardet-Biedl syndrome 13, 209900
MLC1	95.0	100%	100%	Megalencephalic leukoencephalopathy with subcortical cysts, 604004 -3
MLH1	101.0	100%	99%	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MLH3	169.3	100%	99%	Colorectal cancer, somatic, 114500 Colorectal cancer, hereditary nonpolyposis, type 7, 614385 Endometrial cancer, 608089
MLLT10	114.1	96%	95%	Leukemia acute myeloid
MLLT11	150.0	100%	100%	Leukemia, acute myelomonocytic
MLPH	83.9	99%	95%	Griscelli syndrome, type 3, 609227
MLYCD	78.3	94%	81%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	174.5	100%	100%	Methylmalonic aciduria, vitamin B12-responsive, 251100
MMAB	79.8	100%	92%	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110
MMACHC	177.9	100%	100%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	79.5	100%	100%	Homocystinuria cblD type variant 1
MMP1	119.6	100%	100%	COPD, rate of decline of lung function in, 606963 {Epidermolysis bullosa dystrophica, autosomal recessive, modifier of}, 226600
MMP13	148.0	100%	100%	Spondyloepimetaphyseal dysplasia, Missouri type, 602111 Metaphyseal anadysplasia 1, 602111
MMP2	102.8	100%	100%	Torg-Winchester syndrome, 259600
MMP20	100.2	100%	98%	Amelogenesis imperfecta, type IIA2, 612529
MMP9	91.8	95%	91%	Metaphyseal anadysplasia 2, 613073
MN1	76.0	100%	100%	Meningioma, 607174
MNX1	52.1	76%	65%	Currarino syndrome, 176450
MOCS1	68.9	98%	92%	Molybdenum cofactor deficiency, type A, 252150
MOCS2	116.2	100%	100%	Molybdenum cofactor deficiency, type B, 252150
MOG	19.1	70%	42%	Narcolepsy 7, 614250
MOGS	125.1	100%	100%	Congenital disorder of glycosylation type IIb

MPC1	89.7	100%	100%	Mitochondrial pyruvate carrier deficiency
MPDU1	113.2	100%	99%	Congenital disorder of glycosylation, type If, 609180
MPDZ	103.8	98%	97%	Hydrocephalus, nonsyndromic, autosomal recessive 2, 615219
MPI	92.1	100%	97%	Congenital disorder of glycosylation, type Ib, 602579
MPL	119.3	99%	94%	Thrombocytopenia, congenital amegakaryocytic, 604498 Thrombocythemia 2, 601977 Myelofibrosis with myeloid metaplasia, somatic, 254450
MPLKIP	91.9	100%	100%	Trichothiodystrophy, nonphotosensitive 1, 234050
MPO	89.2	100%	97%	Myeloperoxidase deficiency, 254600 {Alzheimer disease, susceptibility to}, 104300 {Lung cancer, protection against, in smokers}
MPV17	114.3	100%	100%	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810 -3
MPZ	103.2	100%	95%	Charcot-Marie-Tooth disease, type 1B, 118200 Dejerine-Sottas disease, 145900 Neuropathy, congenital hypomyelinating, 605253 Charcot-Marie-Tooth disease, type 2J, 607736 Roussy-Levy syndrome, 180800 Charcot-Marie-Tooth disease, type 2I,
MRAP	75.0	99%	95%	Glucocorticoid deficiency 2
MRE11A	97.1	100%	100%	Ataxia-telangiectasia-like disorder, 604391
MRPL3	74.9	96%	92%	Combined oxidative phosphorylation deficiency 9, 614582
MRPS16	141.6	100%	100%	Combined oxidative phosphorylation deficiency 2, 610498
MRPS22	90.7	100%	100%	Combined oxidative phosphorylation deficiency 5, 611719
MS4A1	152.1	100%	100%	Immunodeficiency, common variable, 5, 613495
MSH2	99.7	99%	96%	Colorectal cancer, hereditary nonpolyposis, type 1, 120435 Muir-Torre syndrome, 158320 Mismatch repair cancer syndrome, 276300
MSH3	109.4	99%	98%	Endometrial carcinoma
MSH6	148.5	100%	100%	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 Endometrial cancer, familial, 608089 Mismatch repair cancer syndrome, 276300
MSR1	115.3	99%	98%	Prostate cancer, hereditary, 176807 Barrett esophagus/esophageal adenocarcinoma, 614266
MSRB3	115.5	100%	98%	Deafness, autosomal recessive 74, 613718
MSTN	196.3	100%	100%	Muscle hypertrophy

MSX1	60.2	100%	89%	Tooth agenesis, selective, 1, with or without orofacial cleft, 106600 Orofacial cleft 5, 608874 Ectodermal dysplasia 3, Witkop type, 189500
MSX2	38.8	82%	62%	Craniosynostosis, type 2, 604757 Parietal foramina 1, 168500 Parietal foramina with cleidocranial dysplasia, 168550
MTAP	84.1	100%	100%	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250
MTFMT	100.3	100%	100%	Combined oxidative phosphorylation deficiency 15, 614947
MTHFR	93.8	100%	98%	Homocystinuria due to MTHFR deficiency, 236250 {Schizophrenia, susceptibility to}, 181500 {Vascular disease, susceptibility to} {Neural tube defects, susceptibility to}, 601634 {Thromboembolism, susceptibility to}, 188050
MTM1	55.5	100%	98%	Myotubular myopathy, X-linked, 310400
MTMR2	106.9	100%	100%	Charcot-Marie-Tooth disease, type 4B1, 601382
MTO1	129.0	100%	98%	Combined oxidative phosphorylation deficiency 10, 614702
MTPAP	119.0	91%	91%	Ataxia, spastic, 4, 613672
MTR	104.3	100%	99%	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTRR	105.2	100%	98%	Homocystinuria-megaloblastic anemia, cbl E type, 236270 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTTP	102.2	100%	98%	Abetalipoproteinemia
MUC1	84.2	93%	87%	Medullary cystic kidney disease 1, 174000
MUSK	130.9	100%	98%	Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931
MUT	124.8	100%	100%	Methylmalonic aciduria, mut(0) type, 251000
MUTYH	112.8	100%	100%	Adenomas, multiple colorectal, 608456 Gastric cancer, somatic, 613659 Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600
MVK	88.3	100%	97%	Mevalonic aciduria, 610377 Hyper-IgD syndrome, 260920 Porokeratosis 3, disseminated superficial actinic, 175900
MXI1	108.4	100%	94%	Neurofibrosarcoma {Prostate cancer, susceptibility to}, 176807

MYBPC1	96.3	100%	98%	Arthrogryposis, distal, type 1B, 614335 Lethal congenital contracture syndrome 4, 614915
MYBPC3	90.7	95%	94%	Cardiomyopathy, familial hypertrophic, 4, 115197 Cardiomyopathy, dilated, 1MM, 615396 Left ventricular noncompaction 10, 615396
MYCN	107.3	97%	94%	Feingold syndrome, 164280
MYD88	165.8	100%	96%	Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260 Macroglobulinemia, Waldenstrom, somatic, 153600
MYF6	145.5	100%	100%	Myopathy, centronuclear, 3, 614408
MYH11	118.8	100%	97%	Aortic aneurysm, familial thoracic 4, 132900
MYH14	62.4	96%	85%	Deafness, autosomal dominant 4A, 600652 Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369
MYH2	100.8	97%	93%	Inclusion body myopathy-3, 605637
MYH3	111.2	97%	95%	Arthrogryposis, distal, type 2A, 193700 Arthrogryposis, distal, type 2B, 601680
MYH6	97.4	95%	89%	Cardiomyopathy, familial hypertrophic, 14, 613251 Atrial septal defect 3, 614089 Cardiomyopathy, dilated, 1EE, 613252 {Sick sinus syndrome 3}, 614090
MYH7	89.3	95%	88%	Cardiomyopathy, familial hypertrophic, 1, 192600 Cardiomyopathy, dilated, 1S, 613426 Myopathy, myosin storage, 608358 Laing distal myopathy, 160500 Scapuloperoneal syndrome, myopathic type, 181430 Left ventricular noncompaction 5, 6134
MYH8	104.4	95%	86%	Carney complex variant, 608837 Trismus-pseudocamptodactyly syndrome, 158300
MYH9	96.5	100%	98%	May-Hegglin anomaly, 155100 Fechtner syndrome, 153640 Sebastian syndrome, 605249 Deafness, autosomal dominant 17, 603622 Epstein syndrome, 153650 Macrothrombocytopenia and progressive sensorineural deafness, 600208
MYL2	101.4	100%	100%	Cardiomyopathy, familial hypertrophic, 10, 608758
MYL3	86.1	100%	99%	Cardiomyopathy, familial hypertrophic, 8, 608751
MYLK	111.5	99%	96%	Aortic aneurysm, familial thoracic 7, 613780
MYLK2	89.2	98%	97%	Cardiomyopathy, hypertrophic, midventricular, digenic, 192600

MYO15A	89.7	98%	92%	Deafness, autosomal recessive 3, 600316
MYO1A	102.3	100%	97%	Deafness, autosomal dominant 48, 607841
MYO1E	82.6	97%	93%	Glomerulosclerosis focal segmental 6
MYO3A	109.8	99%	97%	Deafness, autosomal recessive 30, 607101
MYO5A	95.6	99%	97%	Griscelli syndrome, type 1, 214450
MYO5B	83.5	95%	92%	Microvillus inclusion disease, 251850
MYO6	108.0	100%	99%	Deafness, autosomal dominant 22, 606346 Deafness, autosomal recessive 37, 607821 Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346
MYO7A	77.0	95%	91%	Usher syndrome, type 1B, 276900 Deafness, autosomal recessive 2, 600060 Deafness, autosomal dominant 11, 601317
MYOC	182.9	100%	100%	Glaucoma 1A, primary open angle, 137750
MYOT	140.1	100%	100%	Muscular dystrophy, limb-girdle, type 1A, 159000 Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
MYOZ2	105.2	100%	100%	Cardiomyopathy, familial hypertrophic, 16, 613838
MYPN	109.9	100%	96%	Cardiomyopathy, dilated, 1KK, 615248 Cardiomypathy, familial hypertrophic, 22, 615248 Cardiomyopaty, familial restrictive 4, 615248
NAA10	50.5	94%	86%	N-terminal acetyltransferase deficiency, 300855
NAGA	78.6	100%	99%	Schindler disease, type I, 609241 Kanzaki disease, 609242 Schindler disease, type III, 609241
NAGLU	67.1	94%	89%	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NAGS	52.2	85%	75%	N-acetylglutamate synthase deficiency, 237310
NALCN	109.7	99%	97%	?Neuroaxonal neurodegeneration, infantile, with facial dysmophism, 615419
NANOS1	39.1	94%	80%	Spermatogenic failure 12, 615413
NBAS	103.6	100%	99%	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NBEAL2	109.7	99%	98%	Gray platelet syndrome, 139090
NBN	121.4	98%	97%	Leukemia
NCF1	.9	%	%	Chronic granulomatous disease due to deficiency of NCF-1, 233700
NCF2	101.4	100%	98%	Chronic granulomatous disease due to deficiency of NCF-2, 233710
NCF4	79.4	97%	96%	Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III, 613960

NCOA4	36.4	72%	62%	Thyroid carcinoma, papillary, 188550
NCSTN	80.1	97%	90%	Acne inversa, familial, 1, 142690
NDE1	97.3	100%	96%	Lissencephaly 4 (with microcephaly), 614019
NDN	41.1	100%	100%	Prader-Willi syndrome, 176270
NDP	52.2	86%	78%	Norrie disease, 310600 Exudative vitreoretinopathy, X-linked, 305390
NDRG1	82.9	97%	92%	Charcot-Marie-Tooth disease, type 4D, 601455
NDUFA1	116.6	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFA10	88.8	100%	97%	previous assignment to chr. 12 Leigh syndrome, 256000
NDUFA11	102.0	99%	84%	Mitochondrial complex I deficiency, 252010
NDUFA12	105.7	100%	100%	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000
NDUFA2	160.5	100%	100%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFA9	99.3	100%	100%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 -3
NDUFAF1	116.9	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF2	55.3	100%	98%	Mitochondrial complex I deficiency, 252010 Leigh syndrome, 256000
NDUFAF3	131.2	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF4	81.2	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF5	136.0	100%	100%	Mitochondrial complex 1 deficiency, 252010
NDUFAF6	101.9	100%	97%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFB3	1.5	%	%	Mitochondrial complex I deficiency, 252010
NDUFS1	80.4	100%	98%	Mitochondrial complex I deficiency, 252010
NDUFS2	122.5	100%	97%	Mitochondrial complex I deficiency, 252010
NDUFS3	153.9	100%	100%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
NDUFS4	127.6	100%	100%	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFS6	118.7	90%	77%	Complex I, mitochondrial respiratory chain, deficiency of, 252010
NDUFS7	100.0	100%	100%	Leigh syndrome, 256000
NDUFS8	107.7	100%	96%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFV1	63.4	100%	92%	Mitochondrial complex I deficiency, 252010
NDUFV2	124.6	100%	100%	Mitochondrial complex I deficiency, 252010
NEB	89.1	82%	80%	Nemaline myopathy 2, autosomal recessive, 256030
NEFL	133.8	100%	100%	Charcot-Marie-Tooth disease, type 2E, 607684 Charcot-Marie-Tooth disease, type 1F, 607734
NEK1	129.4	100%	99%	Short rib-polydactyly syndrome, type IIA, 263520

NEU1	13.5	64%	20%	Sialidosis, type I, 256550 Sialidosis, type II, 256550
NEUROD1	130.4	100%	100%	{Diabetes mellitus, noninsulin-dependent}, 125853 Maturity-onset diabetes of the young 6, 606394
NEUROG3	105.6	100%	100%	Diarrhea 4, malabsorptive, congenital, 610370
NEXN	136.1	100%	100%	Cardiomyopathy, dilated, 1CC, 613122 Cardiomyopathy, familial hypertrophic, 20, 613876
NF1	82.4	84%	81%	Neurofibromatosis, type 1, 162200 Leukemia, juvenile myelomonocytic, 607785 Melanoma, desmoplastic neurotrophic (2) Neurofibromatosis, familial spinal, 162210 Neurofibromatosis-Noonan syndrome, 601321 Watson syndrome, 193520
NF2	82.2	100%	98%	loss of heterozygosity Neurofibromatosis, type 2, 101000 Meningioma, NF2-related, somatic, 607174 Schwannomatosis, 162091
NFIX	129.0	98%	97%	Sotos syndrome 2, 614753 Marshall-Smith syndrome, 602535
NFKB2	87.4	100%	98%	Immunodeficiency, common variable, 10, 615577
NFKBIA	100.8	100%	100%	Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency, 612132
NFU1	86.0	98%	91%	Multiple mitochondrial dysfunctions syndrome 1, 605711
NGF	180.0	100%	100%	Neuropathy hereditary sensory and autonomic type V
NHEJ1	74.6	100%	99%	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
NHLRC1	109.3	100%	100%	Epilepsy, progressive myoclonic 2B (Lafora), 254780
NHP2	52.5	100%	97%	Dyskeratosis congenita autosomal recessive 2
NHS	67.6	93%	89%	Nance-Horan syndrome, 302350 Cataract 40, X-linked, 302200
NIN	132.5	100%	99%	Seckel syndrome 7, 614851
NIPA1	98.4	91%	82%	Spastic paraplegia 6, autosomal dominant, 600363
NIPAL4	131.8	100%	98%	Ichthyosis, congenital, autosomal recessive 6, 612281
NIPBL	122.1	98%	98%	Cornelia de Lange syndrome 1, 122470
NKX2-1	89.6	100%	100%	Goiter, familial, due to TTF-1 defect (1) Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978

NKX2-5	113.0	100%	99%	Atrial septal defect 7, with or without AV conduction defects, 108900 Tetrology of Fallot, 187500 Hypothyroidism, congenital nongoitrous, 5, 225250 Ventricular septal defect 3, 614432 Hypoplastic left heart syndrome 2, 614435 Conotrunc
NKX2-6	82.4	100%	93%	Persistent truncus arteriosus, 217095
NKX3-2	71.0	100%	98%	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330
NLGN4X	34.1	66%	56%	Mental retardation X-linked
NLRP12	99.6	100%	98%	Familial cold autoinflammatory syndrome 2, 611762
NLRP3	124.2	100%	99%	Cold-induced autoinflammatory syndrome, familial, 120100 Muckle-Wells syndrome, 191900 CINCA syndrome, 607115
NLRP7	134.0	100%	100%	Hydatidiform mole
NME8	102.2	100%	100%	Ciliary dyskinesia, primary, 6, 610852
NMNAT1	96.0	100%	100%	Leber congenital amaurosis 9, 608553
NNT	96.8	100%	100%	Glucocorticoid deficiency 4, 614736
NOBOX	84.4	96%	93%	Premature ovarian failure 5, 611548
NOD2	96.2	100%	98%	{Inflammatory bowel disease 1}, 266600 Blau syndrome, 186580 {Psoriatic arthritis, susceptibility to}, 607507 Sarcoidosis, early-onset, 609464
NODAL	126.5	94%	83%	Heterotaxy, visceral, 5, 270100
NOG	110.9	100%	100%	Symphalangism, proximal, 185800 Multiple synostosis syndrome 1, 186500 Tarsal-carpal coalition syndrome, 186570 Stapes ankylosis with broad thumb and toes, 184460 Brachydactyly, type B2, 611377
NOL3	173.8	100%	100%	Myoclonus familial cortical
NOP10	193.8	100%	100%	Dyskeratosis congenita autosomal recessive 1
NOP56	95.7	99%	96%	Spinocerebellar ataxia 36, 614153
NOTCH1	71.3	97%	89%	Aortic valve disease, 109730 Leukemia, T-cell acute lymphoblastic (2)
NOTCH2	89.5	91%	89%	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500

NOTCH3	69.6	93%	84%	Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy, 125310 ?Myofibromatosis, infantile 2, 615293
NPC1	90.2	99%	98%	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220
NPC2	74.4	100%	100%	Niemann-pick disease, type C2, 607625
NPHP1	167.1	100%	100%	Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900 Joubert syndrome 4, 609583
NPHP3	110.4	100%	99%	Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540 Meckel syndrome 7, 267010
NPHP4	89.5	98%	93%	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
NPHS1	87.3	100%	96%	Nephrotic syndrome, type 1, 256300
NPHS2	128.4	100%	100%	Nephrotic syndrome type 2
NPM1	45.0	82%	79%	Leukemia, acute promyelocytic, NPM/RARA type Leukemia, acute myeloid, 601626
NPPA	154.3	100%	98%	Atrial fibrillation, familial, 6, 612201
NPR2	152.7	100%	100%	Acromesomelic dysplasia, Maroteaux type, 602875
NR0B1	61.6	100%	100%	46XY sex reversal 2 dosage-sensitive
NR0B2	69.5	100%	100%	Obesity, mild, early-onset, 601665
NR2E3	82.8	98%	93%	Enhanced S-cone syndrome, 268100 Retinitis pigmentosa 37, 611131
NR2F1	164.8	100%	100%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NR3C1	108.1	98%	85%	Cortisol resistance
NR3C2	140.9	100%	98%	Pseudohypoaldosteronism type I, autosomal dominant, 177735 Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115
NR4A3	97.2	100%	96%	Chondrosarcoma extraskeletal myxoid
NR5A1	69.4	92%	85%	46XY sex reversal 3
NRAS	129.6	100%	100%	Autoimmune lymphoproliferative syndrome type IV, 614470 Noonan syndrome 6, 613224 Epidermal nevus, somatic, 162900 Thyroid carcinoma, follicular, somatic, 188470 Colorectal cancer, somatic, 114500

NRL	47.6	100%	98%	Retinitis pigmentosa 27, 613750 Retinal degeneration, autosomal recessive, clumped pigment type
NRXN1	117.0	99%	98%	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332
NSD1	124.8	100%	100%	Sotos syndrome 1, 117550 Leukemia, acute myeloid, 601626 (1) Beckwith-Wiedemann syndrome, 130650
NSDHL	52.1	97%	94%	CHILD syndrome, 308050 CK syndrome, 300831
NSMF	84.3	93%	93%	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
NSUN2	124.4	99%	90%	Mental retardation, autosomal recessive 5, 611091
NT5C2	122.9	100%	100%	Spastic paraplegia 45, 613162
NT5C3A	73.3	96%	88%	Anemia hemolytic due to UMPH1 deficiency
NT5E	111.7	100%	100%	Calcification of joints and arteries, 211800
NTF4	43.9	99%	92%	Glaucoma 1, open angle, 1O, 613100
NTRK1	67.5	99%	94%	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma, familial, 155240
NTRK2	99.9	98%	97%	Obesity, hyperphagia, and developmental delay, 613886
NUBPL	86.1	100%	100%	Mitochondrial complex I deficiency, 252010
NUMA1	90.2	97%	96%	Leukemia, acute promyelocytic, NUMA/RARA type
NUP214	129.1	100%	99%	Leukemia, acute myeloid, 601626 Leukemia, T-cell acute lymphoblastic
NUP62	86.1	100%	97%	Striatonigral degeneration, infantile, 271930
NYX	47.7	97%	94%	Night blindness, congenital stationary (complete), 1A, X-linked, 310500
OAT	44.9	83%	71%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OBSL1	83.1	99%	94%	3-M syndrome 2, 612921
OCA2	96.0	100%	99%	?hypopigmentation in PWS and AS Albinism, oculocutaneous, type II, 203200 Albinism, brown oculocutaneous, 203200 [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220
OCLN	92.4	74%	73%	Band-like calcification with simplified gyration and polymicrogyria, 251290
OCRL	59.4	97%	94%	Lowe syndrome, 309000 Dent disease 2, 300555
OFD1	37.0	88%	78%	Oral-facial-digital syndrome 1, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 Joubert syndrome 10, 300804

OGG1	94.0	100%	98%	Renal cell carcinoma, clear cell, somatic, 144700
OPA1	129.7	99%	99%	Optic atrophy 1, 165500 {Glaucoma, normal tension, susceptibility to}, 606657 Optic atrophy plus syndrome, 125250
OPA3	108.0	100%	100%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPHN1	48.4	99%	88%	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
OPLAH	97.3	99%	96%	5-oxoprolinase deficiency, 260005
OPN1LW	.4	%	%	Colorblindness, protan, 303900 Blue cone monochromacy, 303700 -3
OPN1MW	.5	%	%	Colorblindness, deutan, 303800 Blue cone monochromacy, 303700 -3
OPN1SW	91.8	96%	92%	Colorblindness, tritan, 190900
OPTN	95.9	100%	100%	Glaucoma 1, open angle, E, 137760 {Glaucoma, normal tension, susceptibility to}, 606657 Amyotrophic lateral sclerosis 12, 613435
ORAI1	79.1	99%	87%	Immunodeficiency 9, 612782
ORC1	103.4	100%	99%	Meier-Gorlin syndrome 1, 224690
ORC4	104.9	100%	100%	Meier-Gorlin syndrome 2, 613800
ORC6	88.7	100%	98%	Meier-Gorlin syndrome 3, 613803
OSMR	134.3	100%	100%	Amyloidosis, primary localized cutaneous, 1, 105250
OSTM1	106.3	100%	100%	Osteopetrosis, autosomal recessive 5, 259720
OTC	54.5	100%	94%	Ornithine transcarbamylase deficiency, 311250
OTOA	65.3	67%	66%	Deafness, autosomal recessive 22, 607039
OTOF	92.6	99%	96%	Deafness, autosomal recessive 9, 601071 Auditory neuropathy, autosomal recessive, 1, 601071
OTOG	89.0	99%	94%	Deafness, autosomal recessive 18B, 614945
OTOGL	119.0	100%	99%	Deafness, autosomal recessive 84B, 614944
OTX2	153.3	100%	100%	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, and pituitary dysfunction, 610125
OXCT1	96.3	100%	98%	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050
P2RX1	76.4	100%	98%	Bleeding disorder due to P2RX1 defect
P2RX2	96.6	100%	99%	Deafness, autosomal dominant 41, 608224
P2RY12	183.0	100%	100%	Bleeding disorder, platelet-type, 8, 609821

PABPN1	55.9	67%	54%	Oculopharyngeal muscular dystrophy, 164300
PACS1	104.1	98%	97%	Mental retardation, autosomal dominant 17, 615009
PAFAH1B1	71.2	89%	80%	Lissencephaly 1, 607432 Subcortical laminar heterotopia, 607432 -3
PAH	86.5	100%	98%	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600
PAK3	51.4	100%	97%	Mental retardation, X-linked 30/47, 300558
PALB2	136.6	100%	97%	Fanconi anemia, complementation group N, 610832 {Breast cancer, susceptibility to}, 114480 {Pancreatic cancer, susceptibility to, 3}, 613348
PANK2	112.1	90%	86%	Neurodegeneration with brain iron accumulation 1, 234200 HARP syndrome, 607236
PAPSS2	93.4	100%	99%	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847 -3
PARK2	66.8	98%	95%	Adenocarcinoma of lung somatic
PARK7	101.0	100%	100%	Parkinson disease 7 autosomal recessive early-onset
PAX2	88.9	93%	85%	Papillorenal syndrome, 120330 Renal hypoplasia, isolated, 191830 -3
PAX3	111.9	99%	96%	Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820 Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220
PAX4	64.4	100%	93%	Maturity-onset diabetes of the young, type IX, 612225 Diabetes mellitus, type 2, 125853 Diabetes mellitus, ketosis-prone, 612227
PAX6	83.7	100%	100%	Aniridia, 106210 Peters anomaly, 604229 Cataract with late-onset corneal dystrophy, 106210 Keratitis, 148190 Foveal hyperplasia, 136520 Morning glory disc anomaly, 120430 Optic nerve hypoplasia, 165550 Coloboma, ocular, 120200
PAX8	67.5	100%	86%	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700
PAX9	206.0	99%	99%	Tooth agenesis, selective, 3, 604625
PC	96.7	95%	92%	Pyruvate carboxylase deficiency, 266150
PCBD1	60.2	100%	98%	Hyperphenylalaninemia BH4-deficient D

PCCA	96.2	96%	94%	Propionicacidemia, 606054
PCCB	114.2	100%	100%	Propionicacidemia, 606054
PCDH15	134.8	100%	100%	Usher syndrome, type 1F, 602083 Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1D/F digenic, 601067
PCDH19	72.6	100%	98%	Epileptic encephalopathy, early infantile, 9, 300088
PCM1	123.2	100%	99%	Thyroid carcinoma, papillary, 188550
PCNT	94.7	98%	94%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720 -3
PCSK1	110.5	100%	98%	Obesity with impaired prohormone processing, 600955 {Obesity, susceptibility to, BMIQ12}, 612362
PCSK9	68.4	97%	93%	Hypercholesterolemia, familial, 3, 603776 {Low density lipoprotein cholesterol level QTL 1}, 603776
PCYT1A	90.8	100%	98%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDCD10	85.0	97%	79%	Cerebral cavernous malformations 3, 603285
PDE11A	120.4	99%	97%	Pigmented nodular adrenocortical disease, primary, 2, 610475
PDE4D	109.4	97%	91%	{Stroke, susceptibility to, 1}, 606799 Acrodysostosis 2, with or without hormone resistance, 614613
PDE6A	100.7	99%	95%	Retinitis pigmentosa 43, 613810
PDE6B	101.0	100%	99%	Night blindness, congenital stationary, autosomal dominant 2, 163500 Retinitis pigmentosa-40, 613801
PDE6C	109.6	100%	99%	Cone dystrophy 4, 613093
PDE6G	99.1	100%	100%	Retinitis pigmentosa 57, 613582
PDE6H	40.0	87%	75%	Retinal cone dystrophy 3, 610024 Achromatopsia 6, 610024
PDE8B	96.6	100%	100%	Pigmented nodular adrenocortical disease, primary, 3, 614190 Striatal degeneration, autosomal dominant, 609161
PDGFB	66.7	100%	100%	Meningioma, SIS-related, 607174 Dermatofibrosarcoma protuberans, 607907
PDGFRA	126.1	100%	99%	Gastrointestinal stromal tumor, somatic, 606764 Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685
PDGFRB	86.9	100%	98%	Myeloproliferative disorder with eosinophilia, 131440 (4) Basal ganglia calcification, idiopathic, 4, 615007 Myofibromatosis, infantile, 1, 228550
PDGFRL	108.6	99%	96%	Hepatocellular cancer, somatic, 114550 Colorectal cancer, somatic, 114500
PDHA1	60.8	98%	90%	Pyruvate dehydrogenase E1-alpha deficiency, 312170

				Leigh syndrome, X-linked, 308930
PDHB	97.2	100%	100%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDP1	155.3	100%	100%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	94.4	90%	86%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	90.7	100%	99%	Coenzyme Q10 deficiency, primary, 3, 614652
PDX1	46.2	100%	98%	Lacticacidemia due to PDX1 deficiency, 245349
PDX1	46.2	100%	98%	MODY type IV
PDYN	136.3	100%	100%	Spinocerebellar ataxia 23, 610245
PDZD7	74.0	96%	86%	{Retinal disease in Usher syndrome type IIA, modifier of}, 276901 Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472
PEPD	66.4	97%	89%	Prolidase deficiency, 170100
PER2	80.1	100%	99%	Advanced sleep phase syndrome, familial, 1, 604348
PET100	70.5	100%	99%	Mitochondrial complex IV deficiency, 220110
PEX1	124.5	100%	100%	Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	73.0	95%	86%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX11B	157.2	100%	100%	Peroxisome biogenesis disorder 14B, 614920
PEX12	133.0	100%	100%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	134.6	100%	95%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	92.3	100%	100%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	91.8	94%	84%	Peroxisome biogenesis disorder 8A, (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	104.7	100%	99%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	159.1	100%	100%	Peroxisome biogenesis disorder 5A (Zellweger)
PEX26	113.0	100%	100%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	141.1	100%	100%	Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	88.7	97%	96%	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370
PEX6	91.3	94%	85%	Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863
PEX7	102.2	99%	93%	Rhizomelic chondrodysplasia punctata, type 1, 215100

				Peroxisome biogenesis disorder 9B, 614879
PFKM	102.2	100%	100%	Glycogen storage disease VII, 232800
PFN1	51.6	100%	79%	Amyotrophic lateral sclerosis 18, 614808
PGAM2	96.8	100%	100%	Glycogen storage disease X, 261670
PGAP2	117.1	100%	99%	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGAP3	61.6	98%	92%	Hyperphosphatasia with mental retardation syndrome 4, 615716 (3)
PGK1	42.7	75%	67%	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	109.2	100%	99%	Glycogen storage disease XIV, 612934 Congenital disorder of glycosylation, type It, 614921
PHEX	63.1	100%	98%	Hypophosphatemic rickets, X-linked dominant, 307800
PHF6	78.0	100%	98%	Borjeson-Forssman-Lehmann syndrome, 301900
PHF8	51.1	97%	85%	Mental retardation syndrome, X-linked, Siderius type, 300263
PHGDH	84.6	100%	97%	Phosphoglycerate dehydrogenase deficiency, 601815
PHKA1	47.3	94%	88%	Muscle glycogenosis, 300559
PHKA2	49.6	97%	85%	Glycogen storage disease, type IXa1, 306000 Glycogen storage disease, type IXa2, 306000
PHKB	119.0	97%	97%	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750
PHKG2	143.0	100%	100%	Glycogen storage disease IXc, 613027 Cirrhosis due to liver phosphorylase kinase deficiency
PHOX2A	30.2	81%	64%	Fibrosis of extraocular muscles, congenital, 2, 602078
PHOX2B	58.6	100%	96%	Central hypoventilation syndrome congenital with or without Hirschsprung disease
PHYH	88.8	100%	99%	Refsum disease, 266500
PICALM	105.1	100%	98%	Leukemia, acute myeloid, 601626 Leukemia, acute T-cell lymphoblastic
PIEZO1	85.0	98%	91%	Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380
PIEZO2	101.1	99%	98%	?Marden-Walker syndrome, 248700 Arthrogryposis, distal, type 3, 114300 Arthrogryposis, distal, type 5, 108145
PIGA	66.6	99%	98%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818 Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868
PIGL	95.7	100%	100%	CHIME syndrome, 280000
PIGM	105.4	100%	100%	Glycosylphosphatidylinositol deficiency, 610293
PIGN	107.0	100%	100%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080

PIGO	104.6	100%	99%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGV	182.7	100%	100%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIK3CA	126.7	93%	90%	Ovarian cancer, somatic, 167000 Breast cancer, somatic, 114480 Colorectal cancer, somatic, 114500 Gastric cancer, somatic, 613659 Hepatocellular carcinoma, somatic, 114550 Nonsmall cell lung cancer, somatic, 211980 Keratosis, sebor
PIK3CD	90.4	98%	90%	Immunodeficiency 14, 615513
PIK3R1	144.1	100%	100%	Agammaglobulinemia 7, autosomal recessive, 615214 SHORT syndrome, 269880
PIK3R2	80.8	96%	87%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome, 603387
PIK3R5	72.9	100%	98%	Ataxia-oculomotor apraxia 3, 615217
PIKFYVE	138.0	100%	100%	Corneal fleck dystrophy, 121850
PINK1	78.0	93%	87%	Parkinson disease 6, early onset, 605909
PIP5K1C	58.1	87%	83%	Lethal congenital contractural syndrome 3, 611369
PITPNM3	77.8	99%	93%	Cone-rod dystrophy 5, 600977
PITX1	69.5	99%	92%	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800 Liebenberg syndrome, 186550 (4)
PITX2	109.7	95%	83%	Axenfeld-Rieger syndrome, type 1, 180500 Iridogoniodysgenesis, type 2, 137600 Ring dermoid of cornea, 180550 Peters anomaly, 604229
PITX3	37.2	99%	84%	Anterior segment mesenchymal dysgenesis, 107250 Cataract 11, multiple types, 610623 Cataract 11, syndromic, 610623
PKD1	13.9	19%	18%	Polycystic kidney disease, adult type I, 173900
PKD2	101.4	98%	92%	Polycystic kidney disease 2, 613095
PKHD1	103.8	98%	96%	Polycystic kidney and hepatic disease
PKLR	123.2	100%	97%	Pyruvate kinase deficiency, 266200 Adenosine triphosphate, elevated, of erythrocytes, 102900
PKP1	82.7	96%	86%	Ectodermal dysplasia/skin fragility syndrome, 604536
PKP2	62.9	87%	73%	Arrhythmogenic right ventricular dysplasia 9, 609040
PLA2G4A	133.0	100%	98%	Phospholipase A2, group IV A, deficiency of

PLA2G5	108.2	100%	100%	Fleck retina, familial benign, 228980
PLA2G6	77.6	99%	93%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, 612953
PLA2G7	122.6	100%	100%	Platelet-activating factor acetylhydrolase deficiency, 614278 {Asthma, susceptibility to}, 600807 {Atopy, susceptibility to}, 147050
PLAG1	153.3	100%	99%	Adenomas, salivary gland pleomorphic, 181030
PLAU	92.1	100%	91%	{Alzheimer disease, late-onset, susceptibility to}, 104300 Quebec platelet disorder, 601709
PLCB1	113.0	100%	98%	Epileptic encephalopathy, early infantile, 12, 613722
PLCB4	93.3	100%	98%	Auriculocondylar syndrome 2, 614669
PLCD1	95.1	97%	92%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCE1	123.8	99%	97%	Nephrotic syndrome, type 3, 610725
PLCG2	107.3	99%	98%	Familial cold autoinflammatory syndrome 3, 614468 Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878
PLEC	97.5	99%	96%	Epidermolysis bullosa simplex with pyloric atresia
PLEKHG5	80.3	96%	93%	Spinal muscular atrophy, distal, autosomal recessive, 4, 611067 Charcot-Marie-Tooth disease, recessive intermediate C, 615376
PLEKHM1	6.7	25%	15%	Osteopetrosis, autosomal recessive 6, 611497
PLG	65.3	74%	70%	Plasminogen Tochigi disease Thrombophilia, dysplasminogenemic (1) Plasminogen deficiency, types I and II (1) Conjunctivitis, ligneous, 217090
PLIN1	54.5	90%	78%	Lipodystrophy, familial partial, type 4, 613877
PLN	156.1	100%	100%	Cardiomyopathy, dilated, 1P, 609909 Cardiomyopathy, familial hypertrophic, 18, 613874
PLOD1	78.4	100%	96%	Ehlers-Danlos syndrome, type VI, 225400
PLOD2	111.0	100%	100%	Bruck syndrome 2, 609220
PLOD3	79.6	95%	85%	Lysyl hydroxylase 3 deficiency, 612394
PLP1	48.9	100%	95%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PLS3	66.7	100%	100%	Bone mineral density QTL18, osteoporosis, 300910
PML	103.1	99%	97%	Leukemia, acute promyelocytic, PML/RARA type
PMM2	92.4	100%	100%	Congenital disorder of glycosylation, type Ia, 212065

PMP22	94.8	100%	100%	Charcot-Marie-Tooth disease, type 1A, 118220 Dejerine-Sottas disease, 145900 Neuropathy, recurrent, with pressure palsies, 162500 Charcot-Marie-Tooth disease, type 1E, 118300 Roussy-Levy syndrome, 180800 Neuropathy, inflammatory demyel
PMS2	62.1	56%	55%	Mismatch repair cancer syndrome, 276300 Colorectal cancer, hereditary nonpolyposis, type 4, 614337
PNKP	70.4	100%	99%	Epileptic encephalopathy, early infantile, 10, 613402
PNP	124.9	100%	100%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA1	122.3	100%	100%	Ichthyosis, congenital, autosomal recessive 10, 615024
PNPLA2	78.5	100%	91%	Neutral lipid storage disease with myopathy, 610717
PNPLA6	81.5	100%	96%	Spastic paraplegia 39, autosomal recessive, 612020
PNPO	71.8	100%	97%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
PNPT1	103.6	100%	100%	Combined oxidative phosphorylation deficiency 13, 614932 Deafness, autosomal recessive 70, 614934
POC1A	102.0	96%	87%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POF1B	62.4	98%	90%	Premature ovarian failure 2B
POFUT1	118.3	100%	100%	Dowling-Degos disease 2, 615327
POGLUT1	107.5	100%	98%	Dowling-Degos disease 4, 615696 (3)
POLD1	78.7	94%	86%	{Colorectal cancer, susceptibility to, 10}, 612591 Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381
POLE	104.0	100%	98%	FILS syndrome
POLG	90.5	98%	92%	Progressive external ophthalmoplegia, autosomal recessive, 258450 Progressive external ophthalmoplegia, autosomal dominant, 157640 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial DNA depletion syndrome 4A (Alpers typ
POLG2	129.6	100%	100%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131
POLH	138.3	99%	96%	Xeroderma pigmentosum, variant type, 278750
POLR1C	122.6	100%	96%	Treacher Collins syndrome 3, 248390
POLR1D	190.6	100%	100%	Treacher Collins syndrome 2, 613717
POLR3A	90.6	99%	96%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694

POLR3B	104.2	100%	99%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMC	58.2	80%	63%	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734 {Obesity, early-onset, susceptibility to}, 601665
POMGNT1	96.7	100%	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), t
POMGNT2	135.3	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies type A 8
POMP	176.3	100%	100%	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952
POMT1	99.9	100%	98%	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), t
POMT2	74.5	100%	93%	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), t
POR	101.0	100%	100%	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571
PORCN	55.0	85%	76%	Focal dermal hypoplasia, 305600
POU1F1	105.7	100%	100%	Pituitary hormone deficiency, combined, 1, 613038
POU3F4	68.1	100%	100%	Deafness, X-linked 2, 304400
POU4F3	173.5	100%	100%	Deafness, autosomal dominant 15, 602459
PPARG	113.8	99%	96%	Obesity, severe, 601665 [Obesity, resistance to] Insulin resistance, severe, digenic, 604367 Lipodystrophy, familial partial, type 3, 604367 Carotid intimal medial thickness 1, 609338

				{Diabetes, type 2}, 125853
PPIB	90.7	100%	100%	Osteogenesis imperfecta, type IX, 259440
PPM1D	147.2	100%	99%	Breast cancer, 114480
PPM1K	99.8	100%	94%	Maple syrup urine disease, mild variant, 615135
PPOX	90.4	100%	97%	Porphyria variegata, 176200
PPP1R3A	191.6	100%	100%	Insulin resistance, severe, digenic, 604367
PPP2R1B	116.8	100%	100%	Lung cancer, 211980
PPP2R2B	98.4	94%	85%	Spinocerebellar ataxia 12, 604326
PPT1	66.2	100%	93%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	66.2	100%	99%	Renpenning syndrome, 309500
PRCC	92.6	100%	99%	Renal cell carcinoma, papillary, 605074
PRCD	87.0	100%	100%	Retinitis pigmentosa 36, 610599
PRDM16	111.0	99%	97%	Left ventricular noncompaction 8, 615373 Cardiomyopathy, dilated, 1LL, 615373
PRDM5	108.1	100%	100%	Brittle cornea syndrome 2, 614170
PRF1	94.1	100%	98%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027
PRG4	99.9	96%	85%	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250
PRICKLE1	104.2	99%	98%	Epilepsy, progressive myoclonic 1B, 612437
PRICKLE2	109.8	100%	100%	Epilepsy, progressive myoclonic 5, 613832
PRIMPOL	123.2	100%	97%	Myopia 22, autosomal dominant, 615420 (3)
PRKAG2	90.4	100%	100%	Wolff-Parkinson-White syndrome, 194200 Cardiomyopathy, familial hypertrophic 6, 600858 Glycogen storage disease of heart, lethal congenital, 261740
PRKAR1A	104.7	97%	89%	Carney complex, type 1, 160980 Myxoma, intracardiac, 255960 Thyroid carcinoma, papillary, somatic, 188550 Pigmented nodular adrenocortical disease, primary, 1, 610489 Adrenocortical tumor, somatic, Acrodysostosis 1, with or without hor

PRKCA	107.8	100%	99%	Pituitary tumor, invasive
PRKCG	94.5	98%	92%	Spinocerebellar ataxia 14, 605361
PRKCSH	86.2	100%	96%	Polycystic liver disease, 174050
PRKG1	94.8	100%	97%	Aortic aneurysm, familial thoracic 8, 615436
PRKRA	90.3	91%	76%	Dystonia 16
PRLR	91.2	100%	100%	?Hyperprolactinemia, 615555 Multiple fibroadenomas of the breast, 615554
PRNP	97.8	100%	100%	Creutzfeldt-Jakob disease, 123400 Gerstmann-Straussler disease, 137440 Insomnia, fatal familial, 600072 Prion disease with protracted course, 606688 Huntington disease-like 1, 603218 {Kuru, susceptibility to}, 245300
PROC	93.7	99%	94%	Thrombophilia due to protein C deficiency, autosomal dominant, 176860 Thrombophilia due to protein C deficiency, autosomal recessive, 612304
PRODH	51.5	84%	66%	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850
PROK2	86.2	100%	98%	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	177.3	100%	100%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROM1	87.8	99%	93%	Retinitis pigmentosa 41, 612095 Cone-rod dystrophy 12, 612657 Stargardt disease 4, 603786 Macular dystrophy, retinal, 2, 608051
PROP1	66.3	100%	70%	Pituitary hormone deficiency, combined, 2, 262600
PROS1	58.0	85%	72%	Thrombophilia due to protein S deficiency, autosomal dominant, 612336 Thrombophilia due to protein S deficiency, autosomal recessive, 614514
PRPF3	96.8	100%	100%	Retinitis pigmentosa 18
PRPF31	82.2	86%	83%	Retinitis pigmentosa 11, 600138
PRPF6	88.8	100%	99%	Retinitis pigmentosa 60, 613983
PRPF8	120.7	99%	98%	Retinitis pigmentosa 13, 600059
PRPH2	147.4	100%	100%	Retinitis pigmentosa 7, 608133 Retinitis punctata albescens, 136880 Macular dystrophy, patterned, 169150 Macular dystrophy, vitelliform, 608161 Foveomacular dystrophy, adult-onset, with choroidal neovascularization, 608161 Macular dyst

PRPS1	63.6	99%	97%	Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Arts syndrome, 301835 Deafness, X-linked 1, 304500
PRRT2	77.9	100%	100%	Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751 Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066
PRRX1	76.7	99%	94%	Agnathia-otocephaly complex, 202650
PRSS1	99.7	78%	78%	Preeclampsia/eclampsia 5, 614595
PRSS1	99.7	78%	78%	Pancreatitis, hereditary, 167800 Trypsinogen deficiency, 614044 (1)
PRSS12	99.7	98%	96%	Mental retardation, autosomal recessive 1, 249500
PRSS56	60.5	97%	85%	Microphthalmia, isolated 6, 613517
PRX	113.1	99%	98%	Dejerine-Sottas disease, autosomal recessive, 145900 Charcot-Marie-Tooth disease, type 4F, 614895
PSAP	80.2	100%	98%	Metachromatic leukodystrophy due to SAP-b deficiency, 249900 Gaucher disease, atypical, 610539 Combined SAP deficiency, 611721 Krabbe disease, atypical, 611722
PSAT1	41.1	67%	54%	Phosphoserine aminotransferase deficiency, 610992
PSEN1	96.5	100%	93%	Alzheimer disease, type 3, 607822 Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 Dementia, frontotemporal, 600274 Pick disease, 172700
PSEN2	86.9	100%	99%	Alzheimer disease-4, 606889 Cardiomyopathy, dilated, 1V, 613697 -3
PSENEN	130.5	100%	100%	Acne inversa, familial, 2, 613736
PSMB8	9.8	35%	11%	Autoinflammation, lipodystrophy, and dermatosis syndrome, 256040
PSMC3IP	142.5	100%	94%	Ovarian dysgenesis 3, 614324
PSTPIP1	61.6	99%	85%	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416 -3
PTCH1	87.7	99%	96%	Basal cell nevus syndrome, 109400 Basal cell carcinoma, somatic, 605462 Holoprosencephaly-7, 610828

PTCH2	77.8	98%	94%	Medulloblastoma, 155255 Basal cell carcinoma, somatic, 605462 -3
PTDSS1	120.9	100%	100%	Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	137.7	100%	97%	Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Bannayan-Riley-Ruvalcaba syndrome, 153480 {Meningioma}, 607174 {Glioma susceptibility 2}, 613028 Macrocephaly/autism syndrome, 605309 PTEN hamartoma tumor syndrome VAT
PTF1A	36.0	87%	58%	Diabetes mellitus, permanent neonatal, with cerebellar agenesis, 609069
PTGIS	56.1	100%	89%	Hypertension, essential, 145500
PTH	199.9	100%	100%	Hypoparathyroidism, autosomal dominant, 146200 Hypoparathyroidism, autosomal recessive, 146200
PTH1R	83.8	97%	95%	Chondrodysplasia Blomstrand type
PTHLH	131.2	100%	100%	Humoral hypercalcemia of malignancy (1) Brachydactyly, type E2, 613382
PTPN11	46.9	88%	66%	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, 607785 Metachondromatosis, 156250
PTPN12	118.8	100%	100%	Colon cancer
PTPN14	122.3	100%	98%	Choanal atresia and lymphedema, 613611
PTPRC	101.1	98%	94%	{Hepatitic C virus, susceptibility to}, 609532 Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971
PTPRJ	112.1	97%	96%	Colon cancer, somatic, 114500
PTPRO	106.4	98%	97%	Nephrotic syndrome, type 6, 614196
PTPRQ	118.2	100%	98%	Deafness, autosomal recessive 84A, 613391
PTRF	151.4	100%	100%	Lipodystrophy, congenital generalized, type 4, 613327
PTS	118.4	100%	100%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUF60	123.9	97%	94%	Verheij syndrome, 615583 (3)
PUS1	65.8	100%	98%	Mitochondrial myopathy and sideroblastic anemia 1, 600462
PVRL1	71.7	100%	97%	Cleft lip/palate-ectodermal dysplasia syndrome
PVRL4	140.1	100%	100%	Ectodermal dysplasia-syndactyly syndrome 1

PYCR1	83.6	100%	96%	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
PYGL	102.8	100%	99%	Glycogen storage disease VI, 232700
PYGM	97.3	100%	98%	McArdle disease, 232600
QARS	119.4	100%	99%	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	77.2	100%	98%	Hyperphenylalaninemia, BH4-deficient, C, 261630
RAB18	105.5	100%	100%	Warburg micro syndrome 3, 614222
RAB23	134.0	100%	100%	Carpenter syndrome, 201000
RAB27A	112.7	100%	100%	Griscelli syndrome, type 2, 607624
RAB28	79.0	96%	95%	Cone-rod dystrophy 18, 615374
RAB33B	108.1	100%	99%	Smith-McCort dysplasia 2
RAB39B	86.9	100%	100%	Mental retardation, X-linked 72, 300271
RAB3GAP1	124.9	99%	97%	Warburg micro syndrome 1, 600118
RAB3GAP2	110.7	99%	99%	Martsolf syndrome, 212720 Warburg micro syndrome 2, 614225
RAB40AL	16.2	69%	39%	Mental retardation, X-linked, syndromic, Martin-Probst type, 300519 -3
RAB7A	78.9	100%	100%	Charcot-Marie-Tooth disease type 2B
RAC2	53.0	100%	94%	Neutrophil immunodeficiency syndrome, 608203
RAD21	97.2	99%	95%	Cornelia de Lange syndrome 4, 614701
RAD50	115.0	100%	99%	Nijmegen breakage syndrome-like disorder, 613078
RAD51	88.5	100%	96%	Mirror movements 2
RAD51C	105.5	100%	100%	Fanconi anemia, complementation group O, 613390 {Breast-ovarian cancer, familial, susceptibility to, 3}, 613399
RAD54B	128.3	100%	100%	Lymphoma, non-Hodgkin Colon adenocarcinoma
RAD54L	88.6	99%	96%	{Breast cancer, invasive ductal}, 114480 Lymphoma, non-Hodgkin,somatic, 605027 Adenocarcinoma, colonic, somatic
RAF1	90.7	100%	100%	Noonan syndrome 5, 611553 LEOPARD syndrome 2, 611554
RAG1	149.1	100%	100%	Severe combined immunodeficiency, B cell-negative, 601457 Omenn syndrome, 603554 Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889 Combined cellular and humoral immune

RAG2	206.1	100%	100%	Severe combined immunodeficiency, B cell-negative, 601457 Omenn syndrome, 603554 Combined cellular and humoral immune defects with granulomas, 233650
RAI1	137.2	99%	99%	Smith-Magenis syndrome, 182290
RAP1GDS1	88.6	99%	99%	Lymphocytic leukemia, acute T-cell
RAPSN	92.2	92%	86%	Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931 Myasthenic syndrome, congenital, associated with facial dysmorphism and acetylcholine receptor deficiency, 608931 Fetal akinesia deformation sequence, 20815
RARB	144.6	98%	98%	Microphthalmia, syndromic 12, 615524
RARS2	83.0	100%	98%	Pontocerebellar hypoplasia, type 6, 611523
RASA1	100.0	100%	99%	Parkes Weber syndrome, 608355 Capillary malformation-arteriovenous malformation, 608354 Basal cell carcinoma, somatic, 605462
RAX2	57.2	100%	99%	Cone-rod dystrophy 11
RB1	112.4	98%	98%	Retinoblastoma, 180200 Osteosarcoma, somatic, 259500 Bladder cancer, somatic, 109800 Small cell cancer of the lung, somatic, 182280 Retinoblastoma, trilateral, 180200
RB1CC1	130.4	100%	100%	Breast cancer, somatic, 114480
RBBP8	115.5	100%	100%	Pancreatic carcinoma, somatic Seckel syndrome 2, 606744 Jawad syndrome, 251255
RBM10	49.0	91%	82%	TARP syndrome, 311900
RBM20	111.0	98%	96%	Cardiomyopathy, dilated, 1DD, 613172
RBM28	101.9	100%	99%	Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RBM8A	85.9	100%	100%	Thrombocytopenia-absent radius syndrome, 274000
RBP4	80.1	95%	90%	Retinol dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RBPJ	76.2	99%	95%	Adams-Oliver syndrome 3, 614814
RD3	56.9	100%	100%	Leber congenital amaurosis 12, 610612
RDH12	63.6	91%	80%	Leber congenital amaurosis 13, 612712
RDH5	107.3	100%	100%	Fundus albipunctatus, 136880
RDX	56.3	91%	81%	Deafness, autosomal recessive 24, 611022

RECQL4	87.7	98%	95%	Rothmund-Thomson syndrome, 268400 RAPADILINO syndrome, 266280 Baller-Gerold syndrome, 218600
REEP1	96.3	100%	94%	Spastic paraplegia 31, autosomal dominant, 610250 Neuronopathy, distal hereditary motor, type VB, 614751
RELN	105.7	99%	98%	Lissencephaly 2 (Norman-Roberts type), 257320
REN	104.6	100%	100%	[Hyperproreninemia] Renal tubular dysgenesis, 267430 Hyperuricemic nephropathy, familial juvenile 2, 613092
RET	90.8	98%	94%	Multiple endocrine neoplasia IIA, 171400 Medullary thyroid carcinoma, 155240 Multiple endocrine neoplasia IIB, 162300 Central hypoventilation syndrome, congenital, 209880 Pheochromocytoma, 171300 Renal agenesis, 191830 {Hirschsprun
RFT1	78.3	100%	97%	Congenital disorder of glycosylation, type In, 612015
RFX5	116.0	99%	97%	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFX6	126.4	100%	100%	Martinez-Frias syndrome, 601346
RFXANK	94.4	97%	95%	MHC class II deficiency, complementation group B, 209920
RFXAP	86.6	93%	88%	Bare lymphocyte syndrome, type II, complementation group D, 209920
RGR	87.0	95%	87%	Retinitis pigmentosa 44, 613769
RGS9	104.9	98%	93%	Bradyopsia, 608415
RGS9BP	37.4	100%	94%	Bradyopsia, 608415
RHAG	85.6	100%	100%	Anemia, hemolytic, Rh-null, regulator type, 268150 Rh-mod syndrome
RHBDF2	60.4	99%	88%	Tylosis with esophageal cancer, 148500
RHCE	96.3	73%	69%	[Blood group, Rhesus], 111690 Rh-null disease, amorph type -3
RHO	125.6	100%	98%	Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis punctata albescens, 136880
RIMS1	103.9	100%	100%	Cone-rod dystrophy 7, 603649
RIN2	126.8	100%	98%	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075
RIPK4	104.8	99%	94%	Popliteal pterygium syndrome 2, lethal type, 263650

RIT1	162.5	100%	100%	Noonan syndrome 8, 615355
RLBP1	101.2	100%	98%	Fundus albipunctatus, 136880 Retinitis punctata albescens, 136880 Newfoundland rod-cone dystrophy, 607476 Bothnia retinal dystrophy, 607475
RMND1	86.7	95%	92%	Combined oxidative phosphorylation deficiency 11, 614922
RMRP	146.5	100%	100%	Anauxetic dysplasia
RNASEH2A	90.2	100%	88%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	115.0	100%	100%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	161.5	100%	100%	Aicardi-Goutieres syndrome 3, 610329
RNASEL	139.3	99%	96%	Prostate cancer 1, 601518
RNASET2	102.0	100%	98%	Leukoencephalopathy, cystic, without megalencephaly, 612951
RNF135	95.0	95%	85%	Macrocephaly, macrosomia, facial dysmorphism syndrome, 614192
RNF139	158.8	100%	100%	Renal cell carcinoma, 144700
RNF168	204.4	100%	100%	RIDDLE syndrome, 611943
RNF170	121.0	100%	100%	Ataxia, sensory, 1, autosomal dominant, 608984
RNF212	97.7	99%	98%	Recombination rate QTL 1, 612042
RNF216	78.8	95%	91%	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840
RNF6	155.7	100%	100%	Esophageal carcinoma, somatic, 133239
ROBO2	108.8	100%	99%	Vesicoureteral reflux 2, 610878
ROBO3	81.8	97%	87%	Gaze palsy, horizontal, with progressive scoliosis, 607313
ROGDI	94.4	96%	95%	Kohlschutter-Tonz syndrome, 226750
ROM1	92.5	100%	100%	Retinitis pigmentosa 7, digenic, 608133
ROR2	109.8	99%	95%	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RP1	179.8	100%	100%	Retinitis pigmentosa 1, 180100 {Hypertriglyceridemia, susceptibility to}, 145750
RP1L1	128.3	100%	100%	Occult macular dystrophy, 613587
RP2	60.2	100%	98%	Retinitis pigmentosa 2, 312600
RPE65	114.7	100%	98%	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794

RPGR	51.9	83%	75%	Retinitis pigmentosa 3, 300029 Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 Macular degeneration, X-linked atrophic, 300834 Cone-rod dystrophy, X-linked, 1, 304020
RPGRIP1	118.5	100%	98%	Leber congenital amaurosis 6, 613826 Cone-rod dystrophy 13, 608194
RPGRIP1L	101.4	98%	96%	Joubert syndrome 7, 611560 Meckel syndrome 5, 611561 COACH syndrome, 216360
RPIA	72.0	100%	95%	Ribose 5-phosphate isomerase deficiency, 608611
RPL11	75.5	100%	99%	Diamond-Blackfan anemia 7, 612562
RPL35A	30.2	87%	56%	Diamond-Blackfan anemia 5, 612528
RPL5	31.5	86%	60%	Diamond-Blackfan anemia 6, 612561
RPS10	30.7	89%	77%	Diamond-Blackfan anemia 9, 613308
RPS14	33.0	77%	60%	Macrocytic anemia, refractory, due to 5q deletion, somatic, 153550 -3
RPS17	.0	%	%	Diamond-Blackfan anemia 4, 612527
RPS19	38.6	55%	42%	Diamond-Blackfan anemia 1, 105650
RPS24	91.1	100%	94%	Diamond-blackfan anemia 3, 610629
RPS26	35.7	63%	58%	Diamond-Blackfan anemia 10, 613309
RPS6KA3	50.9	98%	91%	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844
RPS7	18.4	76%	37%	Diamond-Blackfan anemia 8, 612563
RPSA	25.5	75%	54%	Asplenia, isolated congenital
RRAS2	102.7	98%	93%	Ovarian carcinoma
RRM2B	111.9	100%	100%	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 5, 613077 Mitochondrial DNA depletion syndrome 8B (MNGIE ty
RS1	39.8	92%	82%	25cM from XG Retinoschisis, 312700
RSPH1	101.7	100%	98%	Ciliary dyskinesia, primary, 24, 615481
RSPH4A	146.2	100%	100%	Ciliary dyskinesia, primary, 11, 612649
RSPH9	85.6	100%	97%	Ciliary dyskinesia, primary, 12, 612650
RSPO1	43.3	98%	74%	Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644

				Palmoplantar hyperkeratosis and true hermaphroditism, 610644
RSPO4	90.4	100%	100%	Anonychia congenita, 206800
RTEL1	86.0	99%	96%	Dyskeratosis congenita, autosomal recessive 5, 615190 Dyskeratosis congenita, autosomal dominant 4, 615190
RTN2	73.4	97%	95%	Spastic paraplegia 12, autosomal dominant, 604805
RTTN	90.5	99%	98%	Polymicrogyria with seizures, 614833
RUNX1	70.5	100%	91%	Leukemia, acute myeloid, 601626 Platelet disorder, familial, with associated myeloid malignancy, 601399
RUNX2	129.2	100%	100%	Cleidocranial dysplasia, 119600 Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600 Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyl
RXFP2	128.5	100%	100%	Cryptorchidism
RYR1	80.7	97%	92%	{Malignant hyperthermia susceptibility 1}, 145600 Central core disease, 117000 Minicore myopathy with external ophthalmoplegia, 255320 Neuromuscular disease, congenital, with uniform type 1 fiber, 117000 King-Denborough syndrome, 145600
RYR2	115.1	100%	99%	Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772 Arrhythmogenic right ventricular dysplasia 2, 600996
SACS	159.3	100%	100%	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAG	111.5	100%	100%	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758
SALL1	145.0	100%	98%	Townes-Brocks syndrome, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480
SALL4	97.7	97%	95%	Duane-radial ray syndrome, 607323 IVIC syndrome, 147750
SAMD9	201.0	100%	100%	Tumoral calcinosis, familial, normophosphatemic, 610455
SAMHD1	122.5	100%	99%	Aicardi-Goutieres syndrome 5, 612952 Chilblain lupus 2, 614415 -3
SAR1B	100.4	100%	100%	Chylomicron retention disease, 246700
SARS2	66.5	98%	91%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SART3	93.6	100%	98%	Porokeratosis, disseminated superficial actinic, 1, 175900

SAT1	74.9	100%	96%	Keratosis follicularis spinulosa decalvans, 308800
SATB2	105.8	97%	94%	Cleft palate and mental retardation, 119540
SBDS	78.8	100%	97%	Shwachman-Bodian-Diamond syndrome
SBF2	104.6	99%	97%	Charcot-Marie-Tooth disease, type 4B2, 604563
SC5D	175.4	100%	100%	Lathosterolosis
SCARB2	88.9	100%	95%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCARF2	53.1	93%	89%	Van den Ende-Gupta syndrome, 600920
SCN10A	125.2	99%	98%	Episodic pain syndrome, familial, 2, 615551
SCN11A	124.9	99%	98%	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN1A	118.3	99%	98%	Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Dravet syndrome, 607208 Migraine, familial hemiplegic, 3, 609634 Febrile seizures, familial, 3A, 604403
SCN1B	100.1	96%	96%	Epilepsy, generalized, with febrile seizures plus, type 1, 604233 Brugada syndrome 5, 612838 Cardiac conduction defect, nonspecific, 612838 Atrial fibrillation, familial, 13, 615377
SCN2A	126.7	100%	98%	Seizures, benign familial infantile, 3, 607745 Epileptic encephalopathy, early infantile, 11, 613721
SCN2B	98.2	100%	99%	Atrial fibrillation, familial, 14, 615378
SCN3B	99.0	100%	100%	Brugada syndrome 7, 613120
SCN4A	132.7	100%	99%	Hyperkalemic periodic paralysis, type 2, 170500 Paramyotonia congenita, 168300 Myotonia congenita, atypical, acetazolamide-responsive, 608390 Myasthenic syndrome, acetazolamide-responsive, 614198 Hypokalemic periodic paralysis, type 2, 613
SCN4B	79.7	100%	98%	Long QT syndrome-10, 611819
SCN5A	112.0	100%	99%	Long QT syndrome-3, 603830 Brugada syndrome 1, 601144 Heart block, progressive, type IA, 113900 Heart block, nonprogressive, 113900 Ventricular fibrillation, familial, 1, 603829 Sick sinus syndrome 1, 608567 Cardiomyopathy, dilated
SCN8A	142.4	100%	99%	Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy, early infantile, 13, 614558

SCN9A	113.8	100%	99%	Erythermalgia, primary, 133020 Insensitivity to pain, channelopathy-associated, 243000 Paroxysmal extreme pain disorder, 167400 Febrile seizures, familial, 3B, 613863 Epilepsy, generalized, with febrile seizures plus, type 7, 613863 Sm
SCNN1A	94.5	97%	93%	Pseudohypoaldosteronism, type I, 264350 Bronchiectasis with or without elevated sweat chloride 2, 613021
SCNN1B	89.4	98%	96%	Liddle syndrome, 177200 Pseudohypoaldosteronism, type I, 264350 Bronchiectasis with or without elevated sweat chloride 1, 211400
SCNN1G	129.4	100%	99%	Liddle syndrome, 177200 Pseudohypoaldosteronism, type I, 264350 Bronchiectasis with or without elevated sweat chloride 3, 613071
SCO1	94.2	96%	95%	Hepatic failure early onset
SCO2	85.4	100%	100%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908
SCP2	98.4	99%	97%	Leukoencephalopathy with dystonia and motor neuropathy, 613724
SDCCAG8	105.0	100%	100%	Senior-Loken syndrome 7, 613615
SDHA	9.1	30%	16%	Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Cardiomyopathy, dilated, 1GG, 613642 Paragangliomas 5, 614165
SDHAF1	91.8	98%	93%	Mitochondrial complex II deficiency, 252011
SDHAF2	107.4	100%	100%	Paragangliomas 2, 601650
SDHB	85.5	100%	100%	Pheochromocytoma, 171300 Paraganglioma and gastric stromal sarcoma, 606864 Cowden syndrome 2, 612359 Gastrointestinal stromal tumor, 606764
SDHC	21.1	39%	33%	Paragangliomas 3, 605373 Paraganglioma and gastric stromal sarcoma, 606864 Gastrointestinal stromal tumor, 606764

SDHD	10.7	33%	16%	Paragangliomas 1, with or without deafness, 168000 Pheochromocytoma, 171300 Carcinoid tumors, intestinal, 114900 Merkel cell carcinoma, somatic Paraganglioma and gastric stromal sarcoma, 606864 Cowden syndrome 3, 615106
SEC23A	114.2	100%	100%	Craniolenticulosutural dysplasia, 607812
SEC23B	121.3	100%	100%	Anemia, dyserythropoietic congenital, type II, 224100
SEC63	89.3	93%	91%	Polycystic liver disease, 174050
SECISBP2	94.9	100%	99%	Thyroid hormone metabolism, abnormal, 609698
SEMA3E	108.7	100%	100%	CHARGE syndrome, 214800
SEMA4A	120.6	99%	96%	Retinitis pigmentosa 35, 610282 Cone-rod dystrophy 10, 610283 -3
SEPN1	74.4	88%	79%	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310
SEPSECS	89.6	100%	90%	Pontocerebellar hypoplasia type 2D
SEPT12	75.7	100%	98%	Spermatogenic failure 10
SEPT9	102.9	88%	88%	Amyotrophy hereditary neuralgic
SERAC1	89.8	100%	100%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPINA1	132.6	100%	100%	Emphysema due to AAT deficiency, 613490 Emphysema-cirrhosis, due to AAT deficiency, 613490 Hemorrhagic diathesis due to \'antithrombin\' Pittsburgh, 613490 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963 (1)
SERPINA3	157.3	100%	98%	Cerebrovascular disease, occlusive
SERPINA6	118.6	100%	100%	Corticosteroid-binding globulin deficiency
SERPINA7	73.5	100%	99%	Thyroxine-binding globulin deficiency
SERPINB6	133.2	93%	93%	Deafness, autosomal recessive 91, 613453
SERPINB7	105.8	100%	100%	Palmoplantar keratoderma, Nagashima type, 615598
SERPINC1	130.8	100%	100%	Thrombophilia due to antithrombin III deficiency, 613118

SERPIND1	116.4	100%	97%	Thrombophilia due to heparin cofactor II deficiency
SERPINE1	94.5	97%	90%	Plasminogen activator inhibitor-1 deficiency
SERPINF1	109.8	92%	84%	Osteogenesis imperfecta, type VI, 613982
SERPINF2	134.7	100%	99%	Alpha-2-plasmin inhibitor deficiency
SERPING1	117.4	97%	95%	Angioedema hereditary types I and II
SERPINH1	137.6	100%	100%	{Preterm premature rupture of the membranes, susceptibility to}, 610504 Osteogenesis imperfecta, type X, 613848
SERPINI1	91.5	97%	90%	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218 -3
SETBP1	147.4	98%	96%	Schinzel-Giedion midface retraction syndrome, 269150
SETD5	162.3	100%	98%	Mental retardation, autosomal dominant 23, 615761 (3)
SETX	152.7	100%	100%	Ataxia-ocular apraxia-2, 606002 Amyotrophic lateral sclerosis 4, juvenile, 602433
SF3B1	119.3	100%	99%	Myelodysplastic syndrome, somatic, 614286
SF3B4	73.2	100%	99%	Acrofacial dysostosis 1, Nager type, 154400
SFTPA2	30.2	51%	44%	contiguous with SFTPA1 Pulmonary fibrosis, idiopathic, 178500
SFTPB	61.2	95%	86%	Surfactant metabolism dysfunction, pulmonary, 1, 265120
SFTPC	67.2	100%	99%	Surfactant metabolism dysfunction, pulmonary, 2, 610913
SFXN4	92.5	100%	100%	Combined oxidative phosphorylation deficiency 18, 615578 (3)
SGCA	83.4	90%	81%	Muscular dystrophy, limb-girdle, type 2D, 608099
SGCB	138.7	96%	96%	Muscular dystrophy, limb-girdle, type 2E, 604286
SGCD	100.0	100%	100%	Muscular dystrophy, limb-girdle, type 2F, 601287 Cardiomyopathy, dilated, 1L, 606685
SGCE	89.6	95%	94%	Dystonia-11, myoclonic, 159900
SGCG	105.5	100%	100%	Muscular dystrophy, limb-girdle, type 2C, 253700
SGSH	76.0	94%	94%	Mucopolysaccharidisis type IIIA (Sanfilippo A), 252900
SH2B3	95.5	96%	94%	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950 Erythrocytosis, somatic, 133100
SH2D1A	52.7	94%	83%	Lymphoproliferative syndrome, X-linked, 308240
SH3BP2	82.8	87%	87%	Cherubism, 118400
SH3PXD2B	116.7	99%	96%	Frank-ter Haar syndrome, 249420

SH3TC2	97.8	97%	96%	Charcot-Marie-Tooth disease, type 4C, 601596 Mononeuropathy of the median nerve, mild, 613353
SHANK3	66.6	93%	77%	Phelan-McDermid syndrome, 606232 {Schizophrenia 15}, 613950
SHH	95.9	99%	85%	Holoprosencephaly-3, 142945 Single median maxillary central incisor, 147250 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160
SHOC2	117.3	100%	99%	Noonan-like syndrome with loose anagen hair, 607721
SHOX	.6	%	%	Short stature, idiopathic familial, 300582 Leri-Weill dyschondrosteosis, 127300 Langer mesomelic dysplasia, 249700
SHROOM4	63.7	98%	94%	Stocco dos Santos X-linked mental retardation syndrome, 300434
SI	107.2	100%	99%	Sucrase-isomaltase deficiency, congenital, 222900
SIGMAR1	93.3	100%	99%	Amyotrophic lateral sclerosis 16, juvenile, 614373
SIL1	93.0	100%	93%	Marinesco-Sjogren syndrome, 248800
SIM1	120.6	100%	98%	Obesity, severe, 601665
SIX1	80.7	100%	100%	Brachiootic syndrome 3, 608389 Deafness, autosomal dominant 23, 605192
SIX3	119.8	100%	100%	Holoprosencephaly-2, 157170 Schizensephaly, 269160
SIX5	44.7	94%	81%	Branchiootorenal syndrome 2, 610896
SIX6	128.1	100%	97%	Microphthalmia with cataract 2, 212550
SKI	59.7	82%	79%	Shprintzen-Goldberg syndrome, 182212
SKIV2L	17.7	72%	39%	Trichohepatoenteric syndrome 2, 614602
SLC10A2	140.8	100%	100%	Bile acid malabsorption, primary, 613291
SLC11A2	88.8	99%	97%	Anemia hypochromic microcytic
SLC12A1	142.0	100%	100%	Bartter syndrome, type 1, 601678
SLC12A3	85.6	99%	97%	Gitelman syndrome, 263800
SLC12A6	99.5	100%	99%	Agenesis of the corpus callosum with peripheral neuropathy, 218000 -3
SLC16A1	139.9	100%	99%	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia, familial, 7, 610021
SLC16A12	110.4	99%	95%	Cataract, juvenile, with microcornea and glucosuria, 612018
SLC16A2	50.4	99%	92%	Allan-Herndon-Dudley syndrome, 300523

SLC17A5	100.9	100%	99%	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC17A8	124.0	100%	100%	Deafness, autosomal dominant 25, 605583
SLC19A2	94.7	100%	100%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	110.7	100%	100%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A3	113.7	100%	100%	Episodic ataxia, type 6, 612656
SLC20A2	88.3	99%	96%	Basal ganglia cancification, idiopathic, 3, 614540
SLC22A12	85.5	97%	92%	Hypouricemia, renal, 220150
SLC22A18	96.4	100%	99%	Breast cancer somatic
SLC22A5	123.3	100%	99%	Carnitine deficiency, systemic primary, 212140
SLC24A1	143.1	100%	98%	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
SLC24A5	117.8	100%	96%	[Skin/hair/eye pigmentation 4, fair/dark skin], 113750 Albinism, oculocutaneous, type VI, 113750
SLC25A1	73.0	89%	82%	No OMIM phenotype
SLC25A12	111.6	100%	100%	Hypomyelination, global cerebral, 612949
SLC25A13	99.0	100%	99%	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A15	100.6	88%	83%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 -3
SLC25A19	79.7	100%	97%	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A20	75.1	100%	100%	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A22	75.4	100%	92%	Epileptic encephalopathy, early infantile, 3, 609304
SLC25A3	95.1	100%	100%	Mitochondrial phosphate carrier deficiency, 610773
SLC25A38	74.4	100%	96%	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive, 205950
SLC25A4	117.8	100%	98%	Progressive external ophthalmoplegia with mitochondrial DNA deletions 3, 609283 Mitochondrial DNA depletion syndrome 12 (cardiomyopathic type), 615418

SLC26A2	137.6	100%	100%	Diastrophic dysplasia, 222600 Atelosteogenesis II, 256050 Achondrogenesis Ib, 600972 Epiphyseal dysplasia, multiple, 4, 226900 Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 De la Chapelle dysplasia, 256050
SLC26A3	113.4	100%	98%	?Colon cancer (1) Chloride diarrhea, congenital, Finnish type, 214700
SLC26A4	95.4	99%	95%	Pendred syndrome, 274600 Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791
SLC26A5	93.4	100%	99%	Deafness, autosomal recessive 61, 613865
SLC26A8	106.9	100%	97%	Spermatogenic failure 3, 606766
SLC27A4	78.2	89%	86%	Ichthyosis prematurity syndrome, 608649
SLC29A3	143.7	100%	99%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC2A1	94.3	100%	100%	GLUT1 deficiency syndrome 1, 606777 GLUT1 deficiency syndrome 2, 612126 {Epilepsy, idiopathic generalized, suscpetibility to, 12}, 614847 Dystonia 9, 601042
SLC2A10	88.5	99%	98%	Arterial tortuosity syndrome, 208050
SLC2A2	125.4	100%	100%	{Diabetes mellitus, noninsulin-dependent} Fanconi-Bickel syndrome, 227810
SLC2A9	67.1	100%	94%	{Uric acid concentration, serum, QTL 2}, 612076 Hypouricemia, renal, 2, 612076
SLC30A10	133.4	100%	100%	Hypermanganesemia with dystonia, polycythemia, and cirrhosis, 613280
SLC30A2	85.8	100%	97%	Zinc deficiency, transient neonatal, 608118
SLC33A1	107.9	100%	100%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC34A1	84.3	99%	96%	Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286 Fanconi renotubular syndrome 2, 613388
SLC34A2	129.4	100%	100%	Pulmonary alveolar microlithiasis, 265100 ?Testicular microlithiasis, 610441
SLC34A3	74.0	98%	90%	Hypophosphatemic rickets with hypercalciuria, 241530
SLC35A1	117.6	100%	100%	Congenital disorder of glycosylation, type IIf, 603585
SLC35A2	57.1	97%	96%	Congenital disorder of glycosylation, type IIm, 300896
SLC35C1	97.8	100%	100%	Congenital disorder of glycosylation, type IIc, 266265

SLC35D1	111.7	100%	100%	Schneckenbecken dysplasia, 269250
SLC36A2	126.6	100%	100%	Iminoglycinuria, digenic, 242600 Hyperglycinuria, 138500
SLC37A4	86.8	99%	97%	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240
SLC38A8	65.5	98%	86%	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218 (3)
SLC39A13	104.4	100%	98%	Spondylocheirodysplasia, Ehlers-Danlos syndrome-like, 612350
SLC39A4	65.8	100%	98%	Acrodermatitis enteropathica, 201100
SLC3A1	134.0	100%	100%	Cystinuria, 220100
SLC40A1	117.9	99%	97%	Hemochromatosis, type 4, 606069
SLC45A2	99.4	100%	92%	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	79.2	100%	98%	Folate malabsorption, hereditary, 229050
SLC4A1	91.9	99%	96%	Ovalocytosis Spherocytosis, type 4, 612653 [Malaria, resistance to], 611162 Renal tubular acidosis, distal, AD, 179800 Renal tubular acidosis, distal, AR, 611590 [Blood group, Diego], 110500 [Blood group, Waldner], 112010 [Bloo
SLC4A11	110.0	99%	98%	Corneal endothelial dystrophy 2, autosomal recessive, 217700 Corneal endothelial dystrophy and perceptive deafness, 217400 Corneal dystrophy, Fuchs endothelial, 4, 613268
SLC4A4	110.4	100%	100%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC52A1	132.0	100%	100%	Riboflavin deficiency, 615026
SLC52A2	116.3	100%	100%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	69.1	100%	95%	Brown-Vialetto-Van Laere syndrome 1, 211530 Fazio-Londe disease, 211500
SLC5A1	106.2	100%	99%	Glucose/galactose malabsorption, 606824
SLC5A2	81.4	98%	95%	Renal glucosuria, 233100
SLC5A5	58.1	99%	92%	Thyroid dyshormonogenesis 1, 274400
SLC5A7	109.8	100%	100%	Neuronopathy, distal hereditary motor, type VIIA, 158580

SLC6A19	93.4	98%	95%	Hartnup disorder, 234500 Iminoglycinuria, digenic, 242600 Hyperglycinuria, 138500
SLC6A2	174.3	100%	100%	Orthostatic intolerance
SLC6A20	93.8	92%	89%	Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC6A3	77.2	100%	100%	{Nicotine dependence, protection against}, 188890 Parkinsonism-dystonia, infantile, 613135
SLC6A5	107.1	100%	99%	Hyperekplexia 3, 614618
SLC6A8	4.4	13%	5%	Cerebral creatine deficiency syndrome 1, 300352
SLC7A14	142.2	100%	99%	Retinitis pigmentosa 68, 615725 (3)
SLC7A7	90.3	100%	100%	Lysinuric protein intolerance, 222700
SLC7A9	69.4	100%	96%	Cystinuria, 220100
SLC9A3R1	101.2	100%	95%	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
SLC9A6	62.5	98%	91%	Mental retardation, X-linked syndromic, Christianson type, 300243
SLCO1B1	113.3	100%	99%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO1B3	121.6	100%	97%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO2A1	70.3	100%	98%	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
SLITRK1	133.1	100%	100%	Tourette syndrome, 137580 Trichotillomania, 613229
SLITRK6	162.2	100%	100%	Deafness and myopia, 221200
SLURP1	38.7	96%	75%	Meleda disease, 248300
SLX4	134.1	99%	94%	Fanconi anemia, complementation group P, 613951
SMAD3	76.6	100%	90%	Loeys-Dietz syndrome, type 3, 613795
SMAD4	134.5	100%	99%	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome
SMAD6	78.5	95%	67%	Aortic valve disease 2, 614823
SMAD9	116.7	100%	100%	Pulmonary hypertension primary 2
SMARCA2	89.6	97%	94%	Nicolaides-Baraitser syndrome, 601358
SMARCA4	82.7	97%	92%	Rhabdoid tumor predisposition syndrome 2, 613325 Mental retardation, autosomal dominant 16, 614609
SMARCAD1	126.6	100%	100%	Adermatoglyphia, 136000
SMARCAL1	121.3	99%	96%	Schimke immunoosseous dysplasia, 242900

SMARCB1	126.2	100%	100%	Rhabdoid tumors, somatic, 609322 Rhabdoid predisposition syndrome 1, 609322 Mental retardation, autosomal dominant 15, 614608
SMC1A	70.7	97%	88%	Cornelia de Lange syndrome 2
SMC3	116.6	99%	98%	Cornelia de Lange syndrome 3
SMCHD1	116.4	100%	99%	Fascioscapulohumeral muscular dystrophy 2, digenic, 158901
SMN1	2.2	10%	%	Spinal muscular atrophy-1, 253300 Spinal muscular atrophy-2, 253550 Spinal muscular atrophy-3, 253400 Spinal muscular atrophy-4, 271150
SMO	98.4	100%	97%	Basal cell carcinoma
SMOC1	80.6	100%	96%	Microphthalmia with limb anomalies, 206920
SMOC2	79.2	99%	86%	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SMPD1	109.7	99%	91%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SMPX	61.5	100%	89%	Deafness, X-linked 4, 300066
SMS	12.7	54%	29%	Mental retardation, X-linked, Snyder-Robinson type, 309583
SNAI2	84.3	100%	100%	Waardenburg syndrome, type 2D, 608890 Piebaldism, 172800
SNAP29	128.5	100%	100%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNCA	113.6	100%	100%	Parkinson disease 4, 605543 Dementia, Lewy body, 127750 Parkinson disease 1, 168601
SNCB	65.4	100%	100%	Dementia, Lewy body, 127750
SNIP1	142.4	100%	99%	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501
SNRNP200	112.4	100%	98%	Retinitis pigmentosa 33, 610359
SNRPE	56.2	79%	77%	Hypotrichosis 11, 615059
SNRPN	80.7	100%	80%	Prader-Willi syndrome, 176270
SNTA1	57.7	95%	84%	Long QT syndrome 12
SNX10	108.0	100%	100%	Osteopetrosis, autosomal recessive 8, 615085
SOBP	118.7	100%	94%	Mental retardation, anterior maxillary protrusion, and strabismus, 613671
SOD1	100.5	100%	100%	Amyotrophic lateral sclerosis 1, 105400
SOS1	122.2	100%	100%	Fibromatosis, gingival, 135300 Noonan syndrome 4, 610733

SOST	120.9	100%	100%	Sclerosteosis, 269500 Van Buchem disease, 239100 Craniodiaphyseal dysplasia, autosomal dominant, 122860
SOX10	85.0	100%	100%	Waardenburg syndrome, type 4C, 613266 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 PCWH syndrome, 609136
SOX17	74.3	100%	100%	Vesicoureteral reflux 3, 613674
SOX18	26.0	84%	55%	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823
SOX2	129.8	100%	100%	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SOX3	49.4	91%	84%	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000
SOX9	115.5	100%	100%	Campomelic dysplasia with autosomal sex reversal, 114290 Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290
SP110	93.7	100%	100%	Hepatic venoocclusive disease with immunodeficiency, 235550 {Mycobacterium tuberculosis, susceptibility to}, 607948
SP7	75.5	100%	100%	Osteogenesis imperfecta, type XII, 613849
SPAG1	118.3	99%	97%	Ciliary dyskinesia, primary, 28, 615505
SPAST	113.7	100%	100%	Spastic paraplegia 4, autosomal dominant, 182601
SPATA16	127.9	98%	95%	Spermatogenic failure 6, 102530
SPATA7	132.6	100%	100%	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, autosomal recessive, 604232
SPECC1L	120.2	100%	98%	Facial clefting, oblique, 1, 600251
SPG11	109.6	100%	99%	Spastic paraplegia 11, autosomal recessive, 604360
SPG20	111.5	100%	100%	Troyer syndrome, 275900
SPG21	110.6	100%	100%	Mast syndrome
SPG7	83.8	97%	87%	Spastic paraplegia 7 autosomal recessive
SPINK1	124.5	100%	89%	Pancreatitis, hereditary, 167800 {Fibrocalculous pancreatic diabetes, susceptibility to}, 608189 Tropical calcific pancreatitis, 608189
SPINK5	99.5	100%	97%	Netherton syndrome, 256500 Atopy, 147050
SPINT2	55.8	90%	59%	Diarrhea 3, secretory sodium, congenital, syndromic, 270420

SPR	67.2	100%	99%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRED1	135.9	100%	100%	Legius syndrome, 611431
SPRY4	93.6	100%	100%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
SPTA1	102.8	99%	98%	Elliptocytosis-2, 130600 Pyropoikilocytosis, 266140 Spherocytosis, type 3, 270970
SPTAN1	97.8	100%	98%	Epileptic encephalopathy, early infantile, 5, 613477
SPTB	105.0	100%	99%	Elliptocytosis-3 Spherocytosis, type 2 Anemia, neonatal hemolytic, fatal and near-fatal
SPTBN2	94.6	99%	97%	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia, autosomal recessive 14, 615386
SPTLC1	79.2	95%	90%	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	110.0	100%	100%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SQSTM1	93.6	100%	98%	Paget disease of bone, 602080
SRC	80.1	94%	85%	?Colon cancer, advanced
SRCAP	134.9	100%	99%	Floating-Harbor syndrome, 136140
SRD5A2	66.8	100%	100%	Pseudovaginal perineoscrotal hypospadias, 264600
SRD5A3	130.2	100%	100%	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713
SRP72	91.6	99%	97%	Bone marrow failure, familial, 614675
SRPX2	46.7	97%	82%	Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643 -3
SRY	73.5	98%	98%	46XY sex reversal 1, 400044 46XX sex reversal 1, 400045
SSTR5	98.3	99%	91%	Somatostatin analog, resistance to, 102200
ST14	89.2	99%	94%	Ichthyosis with hypotrichosis, 610765
ST3GAL3	114.0	100%	100%	Mental retardation, autosomal recessive 12, 611090 Epileptic encephalopathy, early infantile, 15, 615006
ST3GAL5	103.7	93%	93%	Amish infantile epilepsy syndrome
STAC3	105.3	100%	100%	Native American myopathy, 255995 (3)
STAMBP	121.9	100%	100%	Microcephaly-capillary malformation syndrome, 614261
STAR	100.6	100%	100%	Lipoid adrenal hyperplasia, 201710
STAT1	89.5	100%	99%	Mycobacterial infection, atypical, familial disseminated, 209950 Mycobacterial and viral infections, susceptibility to, autosomal recessive, 613796 Candidiasis, familial, 7, 614162

STAT3	84.3	98%	93%	Hyper-IgE recurrent infection syndrome, 147060
STAT5B	73.1	84%	74%	Leukemia, acute promyelocytic, STAT5B/RARA type Growth hormone insensitivity with immunodeficiency, 245590
STIL	154.8	100%	100%	Microcephaly 7, primary, autosomal recessive, 612703
STIM1	80.5	93%	90%	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 160565
STK11	77.8	100%	90%	Peutz-Jeghers syndrome, 175200 Melanoma, malignant, somatic Pancreatic cancer, 260350 Testicular tumor, somatic, 273300
STK4	98.4	100%	100%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STOX1	138.2	90%	89%	Preeclampsia/eclampsia 4, 609404
STRA6	73.8	100%	96%	Microphthalmia, syndromic 9, 601186 Microphthalmia, isolated, with coloboma 8, 601186
STRADA	78.4	100%	95%	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087
STRC	11.6	17%	15%	Deafness, autosomal recessive 16, 603720
STS	69.1	98%	92%	nonlyonizing Ichthyosis, X-linked, 308100
STX11	173.6	100%	100%	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STX16	120.5	100%	100%	Pseudohypoparathyroidism, type IB, 603233
STXBP1	91.8	100%	100%	Epileptic encephalopathy, early infantile, 4, 612164 (2)
STXBP2	81.3	100%	96%	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
SUCLA2	81.6	94%	91%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	94.7	95%	91%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUFU	82.2	96%	84%	Medulloblastoma, desmoplastic, 155255 {Meningioma, familial, susceptibility to}, 607174
SUMF1	69.1	100%	95%	Multiple sulfatase deficiency, 272200
SUMO1	29.3	71%	66%	Orofacial cleft 10, 613705
SUOX	172.0	100%	100%	Sulfite oxidase deficiency, 272300
SURF1	91.8	88%	88%	Leigh syndrome, due to COX deficiency, 256000
SYCP3	129.9	100%	100%	Spermatogenic failure 4, 270960 {Pregnancy loss, susceptibility to}
SYN1	33.0	69%	52%	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491

SYNE1	110.6	99%	97%	Spinocerebellar ataxia, autosomal recessive 8, 610743 Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998
SYNE2	108.7	100%	98%	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999
SYNE4	81.5	100%	100%	Deafness, autosomal recessive 76, 615540 (3)
SYNGAP1	53.6	92%	77%	Mental retardation, autosomal dominant 5, 612621
SYNJ1	102.8	100%	97%	Parkinson disease 20, early-onset, 615530
SYP	50.2	99%	92%	Mental retardation, X-linked 96, 300802
SYT14	135.6	94%	94%	Spinocerebellar ataxia, autosomal recessive 11, 614229
SZT2	100.9	99%	95%	Epileptic encephalopathy, early infantile, 18, 615476
T	117.6	98%	96%	{Neural tube defects, susceptibility to}, 182940
TAB2	157.6	100%	100%	Congenital heart defects, nonsyndromic, 2, 614980
TAC3	74.8	100%	100%	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACR3	140.8	100%	100%	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
TACSTD2	182.1	100%	97%	Corneal dystrophy, gelatinous drop-like, 204870
TAF1	74.4	100%	99%	SVA retrotransposon insertion Dystonia-Parkinsonism, X-linked, 314250
TAF2	106.2	100%	100%	Mental retardation, autosomal recessive 40
TAL1	36.1	93%	81%	Leukemia-1, T-cell acute lymphocytic
TAL2	168.8	100%	100%	Leukemia-2, T-cell acute lymphoblastic
TALDO1	91.9	100%	100%	Transaldolase deficiency, 606003
TAP1	13.6	56%	21%	Bare lymphocyte syndrome, type I, 604571
TAP2	9.8	27%	17%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571 Wegener-like granulomatosis
TAPBP	21.6	69%	42%	Bare lymphocyte syndrome, type I, 604571
TARDBP	27.1	40%	33%	Amyotrophic lateral sclerosis 10, with or without FTD, 612069 Frontotemporal lobar degeneration, TARDBP-related, 612069
TAT	100.6	100%	100%	Tyrosinemia, type II, 276600
TAZ	50.6	100%	98%	Barth syndrome, 302060
TBC1D20	83.8	94%	91%	Warburg micro syndrome 4, 615663
TBC1D24	107.1	100%	100%	Myoclonic epilepsy, infantile, familial, 605021 Epileptic encephalopathy, early infantile, 16, 615338
TBCE	118.9	100%	100%	Kenny-Caffey syndrome-1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410
TBP	97.5	100%	99%	Spinocerebellar ataxia 17, 607136 {Parkinson disease, susceptibility to}, 168600

TBX1	64.9	77%	72%	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400 Velocardiofacial syndrome, 192430 Tetralogy of Fallot, 187500
TBX15	82.4	100%	96%	Cousin syndrome, 260660
TBX19	133.2	100%	99%	Adrenocorticotropic hormone deficiency, 201400
TBX20	47.4	72%	66%	Atrial septal defect 4, 611363
TBX21	109.7	88%	84%	{Asthma, aspirin-induced, susceptibility to}, 208550 Asthma and nasal polyps, 208550
TBX22	83.1	96%	93%	Cleft palate with ankyloglossia, 303400 ?Abruzzo-Erickson syndrome, 302905
TBX3	78.5	100%	95%	Ulnar-mammary syndrome, 181450
TBX4	115.7	95%	90%	Small patella syndrome, 147891
TBX5	86.6	99%	97%	Holt-Oram syndrome, 142900
TBXAS1	99.9	100%	99%	Ghosal hematodiaphyseal syndrome, 231095 ?Thromboxane synthase deficiency, 614158 (1)
TCAP	38.8	68%	48%	Muscular dystrophy, limb-girdle, type 2G, 601954 Cardiomyopathy, dilated, 1N, 607487
TCF12	114.0	100%	100%	Craniosynostosis 3, 615314
TCF4	99.0	97%	97%	Pitt-Hopkins syndrome, 610954
TCIRG1	78.3	95%	85%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	103.4	100%	97%	Transcobalamin II deficiency, 275350
TCOF1	95.5	99%	96%	Treacher Collins syndrome 1, 154500
TCTN1	103.9	96%	94%	Joubert syndrome 13
TCTN2	91.8	100%	98%	Meckel syndrome 8, 613885
TCTN3	103.8	100%	99%	Orofaciodigital syndrome IV, 258860 Joubert syndrome 18, 614815
TDGF1	79.9	100%	99%	Forebrain defects
TDP1	111.6	100%	100%	Spinocerebellar ataxia, autosomal recessive with axonal neuropathy, 607250
TDRD7	116.4	100%	100%	Cataract 36, 613887
TEAD1	88.6	100%	99%	Sveinsson choreoretinal atrophy, 108985
TECPR2	113.1	100%	99%	Spastic paraplegia 49, autosomal recessive, 615031
TECR	81.1	100%	89%	Mental retardation, autosomal recessive 14, 614020
TECTA	122.4	99%	98%	Deafness, autosomal dominant 8/12, 601543 Deafness, autosomal recessive 21, 603629

TEK	112.0	100%	99%	Venous malformations, multiple cutaneous and mucosal, 600195
TENM3	131.3	100%	99%	Microphthalmia isolated with coloboma 9
TET2	128.8	100%	99%	Myelodysplastic syndrome, somatic, 614286
TEX28	.3	%	%	No OMIM phenotype Mental retardation, x-linked 99 Blue cone monochromacy Achromatopsia Colorblindness
TF	94.1	98%	97%	Atransferrinemia, 209300
TFAP2A	72.6	95%	88%	Branchiooculofacial syndrome, 113620
TFAP2B	106.2	100%	100%	Char syndrome, 169100
TFE3	36.7	91%	74%	Renal cell carcinoma, papillary, 1, 300854
TFG	108.2	100%	97%	Hereditary motor and sensory neuropathy, proximal type, 604484 Chondrosarcoma, extraskeletal myxoid, 612237 (1)
TFR2	75.0	98%	94%	Hemochromatosis, type 3, 604250
TG	103.6	100%	98%	Thyroid dyshormonogenesis 3, 274700 {Autoimmune thyroid disease, susceptibility to, 3}, 608175
TGFB1	55.3	99%	84%	Camurati-Engelmann disease, 131300 {Cystic fibrosis lung disease, modifier of}, 219700
TGFB2	129.5	100%	98%	Loeys-Dietz syndrome, type 4, 614816
TGFB3	105.4	100%	100%	Arrhythmogenic right ventricular dysplasia 1, 107970
TGFBI	106.6	100%	100%	Corneal dystrophy, Groenouw type I, 121900 Corneal dystrophy, lattice type I, 122200 Corneal dystrophy, Reis-Bucklers type, 608470 Corneal dystrophy, Avellino type, 607541 Corneal dystrophy, lattice type IIIA, 608471 Corneal dystrophy,
TGFBR1	128.4	99%	94%	Loeys-Dietz syndrome, type 1A, 609192 Loeys-Dietz syndrome, type 2A, 608967 {Multiple self-healing squamous epithelioma, susceptiblity to}, 132800
TGFBR2	86.4	100%	100%	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome, type 1B, 610168 Loeys-Dietz syndrome, type 2B, 610380
TGIF1	192.6	99%	99%	Holoprosencephaly-4
TGM1	104.6	100%	95%	Ichthyosis, congenital, autosomal recessive 1, 242300
TGM5	98.5	100%	100%	Peeling skin syndrome, acral type, 609796

TGM6	65.1	91%	86%	Spinocerebellar ataxia 35, 613908
TH	77.8	96%	83%	Segawa syndrome, recessive, 605407
THAP1	127.9	100%	100%	Dystonia 6, torsion, 602629
THBD	75.2	100%	100%	Thrombophilia due to thrombomodulin defect, 614486 {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
THOC6	155.1	100%	98%	Beaulieu-Boycott-Innes syndrome, 613680
THPO	101.5	98%	89%	Thrombocythemia 1, 187950
THRA	121.6	100%	100%	Hypothyroidism, congenital, nongoitrous, 6, 614450
THRB	117.9	100%	100%	Thyroid hormone resistance, 188570 Thyroid hormone resistance, autosomal recessive, 274300 Thyroid hormone resistance, selective pituitary, 145650
TIA1	120.4	100%	100%	Welander distal myopathy, 604454
TIMM8A	26.0	75%	67%	Deafness, X-linked 1, progressive Mohr-Tranebjaerg syndrome, 304700 Jensen syndrome, 311150
TIMP3	123.0	100%	100%	Sorsby fundus dystrophy, 136900
TINF2	182.1	100%	100%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TJP2	86.5	100%	97%	Hypercholanemia, familial, 607748
TK2	89.7	100%	100%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560
TLL1	113.2	100%	100%	Atrial septal defect 6, 613087
TLR4	148.4	100%	99%	Endotoxin hyporesponsiveness {Macular degeneration, age-related, 10}, 611488 {Colorectal cancer, susceptibility to}, 114500
TMC1	111.4	100%	100%	Deafness, autosomal recessive 7, 600974 Deafness, autosomal dominant 36, 606705
TMC6	58.3	100%	93%	Epidermodysplasia verruciformis, 226400
TMC8	79.4	100%	98%	Epidermodysplasia verruciformis, 226400
TMCO1	73.3	100%	98%	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 614132
TMEM126A	78.5	100%	99%	Optic atrophy-7, 612989
TMEM138	97.1	100%	100%	Joubert syndrome 16, 614465
TMEM165	92.6	100%	100%	Congenital disorder of glycosylation, type IIk, 614727
TMEM216	71.0	97%	78%	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194

TMEM231	73.6	97%	90%	Joubert syndrome 20, 614970 Meckel syndrome, type 11, 615397 -3
TMEM237	101.8	100%	94%	Joubert syndrome 14, 614424
TMEM38B	114.6	100%	98%	Osteogenesis imperfecta, type XIV, 615066
TMEM43	83.1	100%	98%	Arrhythmogenic right ventricular dysplasia 5, 604400 Emery-Dreifuss muscular dystrophy 7, AD, 614302
TMEM5	168.4	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
TMEM67	115.6	100%	99%	Meckel syndrome 3, 607361 Joubert syndrome 6, 610688 {Bardet-Biedl syndrome 14, modifier of}, 209900 COACH syndrome, 216360 Nephronophthisis 11, 613550
TMEM70	210.6	100%	100%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMIE	57.4	99%	87%	Deafness, autosomal recessive 6, 600971
TMLHE	37.7	86%	77%	Epsilon-trimethyllysine hydroxylase deficiency, 300872
TMPRSS15	101.4	100%	99%	Enterokinase deficiency
TMPRSS3	91.3	100%	96%	Deafness, autosomal recessive 8/10, 601072
TMPRSS6	72.9	96%	92%	Iron-refractory iron deficiency anemia, 206200
TNC	130.3	100%	99%	Deafness, autosomal dominant 56, 615629
TNFRSF10B	91.0	100%	99%	Squamous cell carcinoma, head and neck, 275355
TNFRSF11A	101.8	96%	94%	Osteolysis, familial expansile, 174810 Paget disease of bone, 602080 Osteopetrosis, autosomal recessive 7, 612301
TNFRSF11B	181.2	100%	100%	Paget disease, juvenile, 239000
TNFRSF13B	60.5	100%	92%	Immunoglobulin A deficiency 2, 609529 Immunodeficiency, common variable, 2, 240500
TNFRSF13C	51.2	100%	73%	Immunodeficiency, common variable, 4, 613494
TNFRSF1A	69.3	94%	91%	Periodic fever, familial, 142680 {Multiple sclerosis, susceptibility to, 5}, 614810

TNFSF11	145.1	100%	100%	Osteopetrosis, autosomal recessive 2, 259710
TNNC1	106.7	100%	100%	Cardiomyopathy, dilated, 1Z, 611879 Cardiomyopathy, familial hypertrophic, 13, 613243
TNNI2	76.2	100%	98%	Arthrogryposis multiplex congenita, distal, type 2B, 601680
TNNI3	77.4	100%	92%	Cardiomyopathy, familial hypertrophic, 7, 613690 Cardiomyopathy, familial restrictive, 115210 Cardiomyopathy, dilated, 2A, 611880 Cardiomyopathy, dilated, 1FF, 613286
TNNT1	89.7	96%	93%	Nemaline myopathy 5, Amish type, 605355
TNNT2	94.0	100%	96%	Cardiomyopathy, familial hypertrophic, 2, 115195 Cardiomyopathy, dilated, 1D, 601494 Cardiomyopathy, familial restrictive, 3, 612422 Left ventricular noncompaction 6, 601494
TNNT3	80.5	100%	90%	Arthyrgryposis, distal, type 2B, 601680
TNXB	7.6	25%	9%	Ehlers-Danlos syndrome, autosomal recessive, due to tenascin X deficiency, 606408 Ehlers-Danlos syndrome, autosomal dominant, hypermobility type, 130020
TOP1	104.5	100%	96%	DNA topoisomerase I, camptothecin-resistant
TOP2A	128.0	99%	95%	DNA topoisomerase II, resistance to inhibition of, by amsacrine
TOPORS	159.3	100%	100%	Retinitis pigmentosa 31, 609923
TP53	77.3	100%	99%	Colorectal cancer, 114500 Li-Fraumeni syndrome, 151623 Hepatocellular carcinoma, 114550 Osteosarcoma, 259500 Choroid plexus papilloma, 260500 Nasopharyngeal carcinoma, 607107 Pancreatic cancer, 260350 Adrenal cortical carcinoma
TP63	128.7	100%	100%	Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 Split-hand/foot malformation 4, 605289 Hay-Wells syndrome, 106260 ADULT syndrome, 103285 Limb-mammary syndrome, 603543 Rapp-Hodgkin syndrome, 129400 Orofac
TPI1	68.0	100%	97%	Hemolytic anemia due to triosephosphate isomerase deficiency

TPK1	80.1	100%	100%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TPM1	85.7	100%	97%	Cardiomyopathy, familial hypertrophic, 3, 115196 Cardiomyopathy, dilated, 1Y, 611878 Left ventricular noncompaction 9, 611878
TPM2	99.8	100%	100%	Arthrogryposis multiplex congenita, distal, type 1, 108120 Arthrogryposis, distal, type 2B, 601680 Nemaline myopathy 4, autosomal dominant, 609285 CAP myopathy 2, 609285
TPM3	68.3	82%	80%	Nemaline myopathy 1, autosomal dominant or recessive, 609284 CAP myopathy 1, 609284 Myopathy congenital, with fiber-type disproportion, 255310
TPMT	95.7	100%	100%	6-mercaptopurine sensitivity, 610460
TPO	78.6	98%	95%	Thyroid dyshormonogenesis 2A, 274500
TPP1	135.3	100%	100%	Ceroid lipofuscinosis, neuronal, 2, 204500
TPRN	48.1	82%	78%	Deafness, autosomal recessive 79, 613307
TRAPPC11	116.5	100%	99%	Muscular dystrophy, limb-girdle, type 2S, 615356
TRAPPC2	29.9	75%	49%	Spondyloepiphyseal dysplasia tarda, 313400
TRAPPC9	68.6	98%	91%	Mental retardation, autosomal recessive 13, 613192
TRDN	80.2	100%	94%	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441
TREM2	100.0	100%	100%	Nasu-Hakola disease, 221770
TREX1	127.4	100%	100%	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700
TRHR	150.4	100%	100%	Thyrotropin-releasing hormone resistance, generalized
TRIM24	95.5	99%	98%	Thyroid carcinoma, papillary, 188550
TRIM32	106.2	100%	100%	Muscular dystrophy, limb-girdle, type 2H, 254110 Bardet-Biedl syndrome 11, 209900
TRIM33	97.8	97%	88%	Thyroid carcinoma, papillary, 188550
TRIM37	105.0	100%	98%	Mulibrey nanism, 253250
TRIOBP	93.9	95%	91%	Deafness, autosomal recessive 28, 609823
TRIP11	132.1	99%	98%	Achondrogenesis, type IA, 200600
TRMU	75.4	100%	99%	{Deafness, mitochondrial, modifier of}, 580000 Liver failure, transient infantile, 613070

TRPA1	67.7	83%	79%	Episodic pain syndrome, familial, 615040
TRPC6	79.3	94%	89%	Glomerulosclerosis, focal segmental, 2, 603965
TRPM1	129.2	98%	98%	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TRPM4	82.4	100%	99%	Progressive familial heart block, type IB, 604559
TRPM6	121.0	99%	98%	Hypomagnesemia 1, intestinal, 602014
TRPS1	136.2	100%	100%	Trichorhinophalangeal syndrome, type I, 190350 Trichorhinophalangeal syndrome, type III, 190351
TRPV3	99.3	99%	94%	Olmsted syndrome, 614594
TRPV4	93.1	100%	99%	Brachyolmia type 3, 113500 Spondylometaphyseal dysplasia, Kozlowski type, 184252 Metatropic dysplasia, 156530 Hereditary motor and sensory neuropathy, type IIc, 606071 Scapuloperoneal spinal muscular atrophy, 181405 [Sodium serum level
TSC1	95.7	99%	97%	Otosclerosis 1 (2)
TSC1	95.7	99%	97%	Tuberous sclerosis-1, 191100 Lymphangioleiomyomatosis, 606690 Focal cortical dysplasia, Taylor balloon cell type, 607341
TSC2	85.9	99%	95%	distal to PKD1 Tuberous sclerosis-2, 613254 Lymphangioleiomyomatosis, somatic, 606690
TSEN2	121.3	100%	100%	Pontocerebellar hypoplasia type 2B, 612389
TSEN34	66.2	100%	93%	Pontocerebellar hypoplasia type 2C, 612390
TSEN54	105.5	98%	96%	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753
TSFM	102.7	96%	94%	Combined oxidative phosphorylation deficiency 3, 610505
TSG101	107.3	100%	100%	Breast cancer, somatic, 114480
TSHB	171.4	100%	100%	Hypothryoidism, congenital, nongoitrous 4, 275100
TSHR	157.0	100%	98%	Hypothyroidism, congenital, nongoitrous, 1 275200 Thyroid adenoma, hyperfunctioning, somatic Hyperthyroidism, nonautoimmune, 609152 Thyroid carcinoma with thyrotoxicosis Hyperthyroidism, familial gestational, 603373
TSHZ1	124.1	99%	98%	Aural atresia, congenital, 607842
TSPAN12	117.9	100%	100%	Exudative vitreoretinopathy 5, 613310
TSPAN7	43.6	95%	71%	Mental retardation, X-linked 58, 300210
TSPEAR	120.9	100%	99%	Deafness, autosomal recessive 98, 614861

TSPYL1	191.4	100%	100%	Sudden infant death with dysgenesis of the testes syndrome, 608800 -3
TTBK2	131.5	100%	100%	Spinocerebellar ataxia 11, 604432
TTC19	72.4	87%	77%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTC21B	112.5	99%	98%	Nephronophthisis 12, 613820 Asphyxiating thoracic dystrophy 4, 613819
TTC37	116.5	100%	100%	Trichohepatoenteric syndrome 1, 222470
TTC7A	65.2	95%	95%	Intestinal atresia, multiple, 243150
TTC8	107.1	100%	100%	Bardet-Biedl syndrome 8, 209900 Retinitis pigmentosa 51, 613464
TTI2	112.9	100%	100%	Mental retardation, autosomal recessive 39, 615541
TTN	149.1	98%	97%	Cardiomyopathy, familial hypertrophic, 9, 613765 Cardiomyopathy, dilated, 1G, 604145 Tibial muscular dystrophy, tardive, 600334 Muscular dystrophy, limb-girdle, type 2J, 608807 Myopathy, proximal, with early respiratory muscle involvement,
TTPA	94.5	98%	92%	Ataxia with isolated vitamin E deficiency, 277460
TTR	87.4	100%	99%	Amyloidosis, hereditary, transthyretin-related, 105210 [Dystransthyretinemic hyperthyroxinemia], 145680 Carpal tunnel syndrome, familial, 115430
TUBA1A	20.1	84%	50%	Lissencephaly 3, 611603
TUBA8	100.5	100%	99%	Polymicrogyria with optic nerve hypoplasia, 613180
TUBB1	142.2	100%	100%	Macrothrombocytopenia, autosomal dominant, TUBB1-related, 613112
TUBB2A	50.8	100%	94%	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB2B	54.4	98%	92%	Polymicrogyria, symmetric or asymmetric, 610031
TUBB3	80.2	81%	79%	Fibrosis of extraocular muscles, congenital, 3A, 600638 Cortical dysplasia, complex, with other brain malformations 1, 614039
TUBB4A	62.8	85%	72%	?Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438
TUBG1	109.0	83%	81%	Cortical dysplasia, complex, with other brain malformations 4, 615412
TUBGCP6	116.4	100%	98%	Microcephaly and chorioretinopathy with or without mental retardation, 251270
TUFM	100.7	99%	94%	Combined oxidative phosphorylation deficiency 4, 610678
TULP1	84.8	100%	93%	Retinitis pigmentosa 14, 600132 Leber congenital amaurosis 15, 613843
TUSC3	126.6	100%	99%	Mental retardation, autosomal recessive 7, 611093

TWIST1	99.1	100%	99%	Saethre-Chotzen syndrome, 101400 Saethre-Chotzen syndrome with eyelid anomalies, 101400 Craniosynostosis, type 1, 123100 Robinow-Sorauf syndrome, 180750
TWIST2	74.9	100%	96%	Focal facial dermal dysplasia 3, Setleis type, 227260
TYK2	84.3	99%	95%	Tyrosine kinase 2 deficiency, 611521
TYMP	91.2	99%	92%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYR	132.9	74%	74%	Albinism, oculocutaneous, type IA, 203100 Waardenburg syndrome/albinism, digenic, 103470 Albinism, oculocutaneous, type IB, 606952 [Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 {Melanoma, cutaneous malignant, susceptibi
TYROBP	62.5	100%	100%	Nasu-Hakola disease, 221770
TYRP1	126.0	100%	100%	Albinism, oculocutaneous, type III, 203290 Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair), 612271
UBA1	66.0	100%	98%	Spinal muscular atrophy, X-linked 2, infantile, 301830
UBE2A	60.0	100%	100%	Mental retardation, X-linked syndromic, Nascimento-type, 300860
UBE3A	102.8	99%	99%	Angelman syndrome, 105830
UBE3B	107.4	99%	98%	Blepharophimosis-ptosis-intellectual disability syndrome, 615057 -3
UBIAD1	94.4	100%	100%	Corneal dystrophy, Schnyder type, 121800
UBQLN2	78.6	100%	100%	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857
UBR1	104.7	100%	99%	Johanson-Blizzard syndrome, 243800
UGT1A1	146.5	100%	98%	Crigler-Najjar syndrome, type I, 218800 [Gilbert syndrome], 143500 Crigler-Najjar syndrome, type II, 606785 Hyperbilirubinemia, familial transcient neonatal, 237900 [Bilirubin, serum level of, QTL1], 601816
UMOD	80.8	99%	96%	Hyperuricemic nephropathy, familial juvenile 1, 162000 Medullary cystic kidney disease 2, 603860 Glomerulocystic kidney disease with hyperuricemia and isosthenuria, 609886
UMPS	111.2	100%	100%	Orotic aciduria, 258900
UNC13D	151.8	100%	100%	Hemophagocytic lymphohistiocytosis familial 3
UNC93B1	41.3	55%	54%	Herpes simplex encephalitis, susceptibility to, 1, 610551
UNG	77.5	94%	92%	Immunodeficiency with hyper IgM, type 5, 608106

UPB1	110.4	100%	100%	Beta-ureidopropionase deficiency, 613161
UPF3B	58.0	98%	87%	Mental retardation, X-linked, syndromic 14, 300676
UQCRB	114.1	100%	100%	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC2	88.5	97%	94%	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRQ	63.3	100%	99%	Mitochondrial complex III deficiency, nuclear type 4, 615159
UROC1	73.3	98%	83%	Urocanase deficiency, 276880
UROD	84.5	97%	90%	Porphyria cutanea tarda, 176100 Porphyria, hepatoerythropoietic, 176100
UROS	71.7	99%	89%	Porphyria, congenital erythropoietic, 263700
USB1	59.4	93%	88%	Poikiloderma with neutropenia
USH1C	79.4	99%	92%	Acadian and Samaritan variety Usher syndrome, type 1C, 276904 Deafness, autosomal recessive 18A, 602092
USH1G	98.7	95%	88%	Usher syndrome type 1G
USH2A	116.3	100%	99%	Usher syndrome, type 2A, 276901 Retinitis pigmentosa 39, 613809 -3
USP9Y	66.9	99%	95%	Spermatogenic failure, Y-linked, 2, 415000
UVSSA	66.7	100%	96%	UV-sensitive syndrome 3, 614640
VANGL1	145.3	100%	100%	Caudal regression syndrome, 600145 Neural tube defects, 182940 -3
VANGL2	103.4	100%	95%	Neural tube defects, 182940
VAPB	153.5	100%	93%	Amyotrophic lateral sclerosis 8, 608627 Spinal muscular atrophy, late-onset, Finkel type, 182980
VAX1	79.7	100%	92%	Microphthalmia, syndromic 11, 614402
VCAN	146.6	100%	100%	Wagner syndrome 1, 143200
VCL	101.2	96%	92%	Cardiomyopathy, dilated, 1W, 611407 Cardiomyopathy, familial hypertrophic, 15, 613255
VCP	111.2	99%	96%	Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320 Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954
VDR	87.1	100%	100%	Rickets, vitamin D-resistant, type IIA, 277440 ?Osteoporosis, involutional, 166710 (1)
VHL	128.4	100%	100%	von Hippel-Lindau syndrome, 193300 Renal cell carcinoma, somatic, 144700 Pheochromocytoma, 171300 Hemangioblastoma, cerebellar, somatic Erythrocytosis, familial, 2, 263400

VIM	97.7	100%	100%	Cataract 30, pulverulent, 116300
VIPAS39	115.0	100%	97%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VKORC1	120.2	100%	100%	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473 Warfarin resistance, 122700
VLDLR	117.9	100%	100%	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VPS13A	122.9	100%	98%	Choreoacanthocytosis, 200150
VPS13B	113.3	99%	98%	Cohen syndrome, 216550
VPS33B	102.6	100%	97%	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
VPS35	78.0	96%	90%	Parkinson disease 17, 614203
VPS37A	84.0	100%	97%	Spastic paraplegia 53, autosomal recessive, 614898
VPS45	104.4	99%	97%	Neutropenia, severe congenital, 5, autosomal recessive, 615285
VRK1	133.6	100%	100%	Pontocerebellar hypoplasia type 1A, 607596
VSX1	55.8	99%	88%	Keratoconus 1, 148300 Corneal dystrophy, hereditary polymorphous posterior, 122000 Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195
VSX2	62.5	100%	94%	Microphthalmia with coloboma 3
VWF	59.3	78%	71%	von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 von Willebrand disease, type 1, 193400 von Willibrand disease, type 3, 277480
WAS	27.3	89%	69%	Wiskott-Aldrich syndrome, 301000 Thrombocytopenia, X-linked, 313900 Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, intermittent, 313900
WDPCP	93.8	100%	99%	Bardet-Biedl syndrome 15
WDR11	93.9	100%	99%	Hypogonadotropic hypogonadism 14 with or without anosmia, 614858
WDR19	123.9	100%	100%	Asphyxiating thoracic dystrophy 5, 614376 Nephronophthisis 13, 614377 Cranioectodermal dysplasia 4, 614378
WDR34	88.2	100%	97%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
WDR35	121.8	99%	97%	Cranioectodermal dysplasia 2, 613610 Short rib-polydactyly syndrome, type V, 614091
WDR36	121.3	99%	95%	Glaucoma 1, open angle, G, 609887
WDR45	42.3	100%	89%	Neurodegeneration with brain iron acculation 5, 300894
WDR60	101.9	99%	98%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503

WDR62	100.3	98%	94%	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
WDR72	111.0	96%	96%	Amelogenesis imperfecta, hypomaturation type, IIA3, 613211
WDR81	112.3	99%	98%	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185
WFS1	145.6	100%	98%	Wolfram syndrome, 222300 Deafness, autosomal dominant 6/14/38, 600965 Wolfram-like syndrome, autosomal dominant, 614296 {Diabetes mellitus, noninsulin-dependent, association with}, 125853
WHSC1L1	120.2	99%	96%	Leukemia, acute myeloid, 601626
WIPF1	92.9	100%	97%	Wiskott-Aldrich syndrome 2, 614493
WISP3	125.0	100%	100%	Arthropathy, progressive pseudorheumatoid, of childhood, 208230 Spondyloepiphyseal dysplasia tarda with progressive arthropathy, 208230
WNK1	144.2	100%	99%	Pseudohypoaldosteronism, type IIC, 614492 Neuropathy, hereditary sensory and autonomic, type II, 201300
WNK4	114.7	100%	99%	Pseudohypoaldosteronism, type IIB, 614491
WNT1	144.6	100%	96%	Osteogenesis imperfecta, type XV, 615220 {Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221
WNT10A	71.2	90%	85%	Odontoonychodermal dysplasia, 257980 Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400
WNT10B	92.9	100%	97%	Split-hand/foot malformation 6, 225300
WNT3	137.2	97%	94%	Tetra-amelia, autosomal recessive, 273395
WNT4	141.9	92%	92%	SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330
WNT5A	106.6	100%	99%	Robinow syndrome, autosomal dominant, 180700
WNT7A	132.1	100%	100%	Ulna and fibula, absence of, with sever limb deficiency, 276820 Fuhrmann syndrome, 228930
WRAP53	125.9	100%	99%	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	140.5	100%	99%	Werner syndrome
WT1	61.3	100%	98%	Wilms tumor, type 1, 194070 Denys-Drash syndrome, 194080 Nephrotic syndrome, type 4, 256370 Frasier syndrome, 136680 Meacham syndrome, 608978

				Mesothelioma, somatic, 156240
WWOX	105.4	100%	98%	Esophageal squamous cell carcinoma, 133239
XDH	89.2	100%	99%	Xanthinuria, type I, 278300
XIAP	69.4	84%	76%	Lymphoproliferative syndrome, X-linked, 2, 300635
XK	60.0	100%	84%	McLeod syndrome with or without chronic granulomatous disease, 300842
XPA	87.6	100%	98%	Xeroderma pigmentosum, group A, 278700
XPC	122.8	100%	98%	Xeroderma pigmentosum, group C, 278720
XPNPEP3	130.7	100%	99%	Nephronophthisis-like nephropathy 1, 613159
YAP1	84.1	97%	89%	Coloboma, ocular, 120433 Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433
YARS	107.6	100%	97%	Charcot-Marie-Tooth disease, dominant intermediate C, 608323
YARS2	106.9	100%	100%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
ZAP70	88.5	87%	87%	Selective T-cell defect, 269840
ZBTB16	124.4	100%	98%	Leukemia, acute promyelocytic, PL2F/RARA type Skeletal defects, genital hypoplasia, and mental retardation, 612447
ZBTB24	163.9	100%	100%	Immunodeficiency-centromeric instability-facial anomalies syndrome-2, 614069
ZC4H2	54.5	100%	98%	Wieacker-Wolf syndrome, 314580
ZDHHC9	36.6	97%	83%	Mental retardation, X-linked syndromic, Raymond type, 300799
ZEB1	146.4	98%	97%	Corneal dystrophy, posterior polymorphous, 3, 609141 Corneal dystrophy, Fuchs endothelial, 6, 613270
ZEB2	154.5	100%	99%	Mowat-Wilson syndrome, 235730
ZFP57	16.5	82%	30%	Diabetes mellitus, transient neonatal, 1, 601410
ZFPM2	183.8	98%	98%	Tetralogy of Fallot, 187500 Diaphragmatic hernia 3, 610187
ZFYVE26	88.6	96%	93%	Spastic paraplegia 15, autosomal recessive, 270700
ZFYVE27	78.6	96%	91%	Spastic paraplegia 33, autosomal dominant, 610244
ZIC2	58.5	93%	86%	Holoprosencephaly-5, 609637
ZIC3	54.2	100%	97%	Heterotaxy, visceral, 1, X-linked 306955 Congenital heart defects, nonsyndromic, 1, X-linked, 306955 VACTERL association, X-linked, 314390

ZMPSTE24	162.4	100%	100%	Mandibuloacral dysplasia with type B lipodystrophy, 608612 Restrictive dermopathy, lethal, 275210
ZMYND10	96.6	100%	92%	Ciliary dyskinesia, primary, 22, 615444
ZNF335	82.1	98%	94%	Microcephaly 10, primary, autosomal recessive, 615095
ZNF423	127.9	100%	99%	Nephronophthisis 14, 614844 Joubert syndrome 19, 614844
ZNF469	94.6	100%	99%	Brittle cornea syndrome, 229200
ZNF513	99.1	100%	96%	Retinitis pigmentosa 58, 613617
ZNF592	114.5	94%	92%	Spinocerebellar ataxia, autosomal recessive 5, 606937
ZNF644	168.2	100%	100%	Myopia 21, autosomal dominant, 614167
ZNF711	82.6	100%	100%	Mental retardation, X-linked 97, 300803
ZNF750	122.9	100%	99%	Seborrhea-like dermatitis with psoriasiform elements, 610227
ZNF81	46.3	99%	96%	Mental retardation, X-linked 45, 300498

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated February 2014
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
OMIM release used for OMIM disease identifiers and descriptions : 31 october 2014
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors