

# MENDELIOME GENE PANEL DG 2.12 (3024 genes)

<i>Gene</i>	<i>Median</i>	<i>% covered &gt; 10x</i>	<i>% covered &gt; 20x</i>	<i>Associated Phenotype description and OMIM disease ID</i>
A2M	119.2	99	98	Alpha-2-macroglobulin deficiency, 614036 {Alzheimer disease, susceptibility to}, 104300
A4GALT	141	100	100	NOR polyagglutination syndrome, 111400 [Blood group, P1Pk system, P phenotype], 111400 [Blood group, P1Pk system, p phenotype], 111400
AAAS	106.6	100	99	Achalasia-addisonianism-alacrimia syndrome, 231550
AAGAB	151.7	100	98	Keratoderma, palmoplantar, punctate type IA, 148600
AARS	124.4	100	99	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 Epileptic encephalopathy, early infantile, 29, 616339
AARS2	126.4	99	99	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889
AASS	128.8	99	97	Hyperlysinemia, 238700 Saccharopinuria, 268700
ABAT	92.8	100	99	GABA-transaminase deficiency, 613163
ABCA1	123.6	100	99	HDL deficiency, type 2, 604091 Tangier disease, 205400 {Coronary artery disease in familial hypercholesterolemia, protection against}, 143890
ABCA12	140	99	97	Ichthyosis, autosomal recessive 4B (harlequin), 242500 Ichthyosis, congenital, autosomal recessive 4A, 601277
ABCA3	124.2	99	99	Surfactant metabolism dysfunction, pulmonary, 3, 610921
ABCA4	128.2	100	99	Cone-rod dystrophy 3, 604116 Fundus flavimaculatus, 248200 Retinal dystrophy, early-onset severe, 248200 Retinitis pigmentosa 19, 601718 Stargardt disease 1, 248200 {Macular degeneration, age-related, 2}, 153800
ABCB11	158.2	99	99	Cholestasis, benign recurrent intrahepatic, 2, 605479 Cholestasis, progressive familial intrahepatic 2, 601847

ABCB4	129.8	99	97	Cholestasis, intrahepatic, of pregnancy, 3, 614972 Cholestasis, progressive familial intrahepatic 3, 602347 Gallbladder disease 1, 600803
ABCB6	127.3	100	99	Dyschromatosis universalis hereditaria 3, 615402 Microphthalmia, isolated, with coloboma 7, 614497 Pseudohyperkalemia, familial, 2, due to red cell leak, 609153 [Blood group, Langereis system], 111600
ABCB7	132.4	99	98	Anemia, sideroblastic, with ataxia, 301310
ABCC2	135.7	100	100	Dubin-Johnson syndrome, 237500
ABCC6	116.6	93	92	Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850
ABCC8	146.9	100	99	Diabetes mellitus, noninsulin-dependent, 125853 Diabetes mellitus, permanent neonatal, 606176 Diabetes mellitus, transient neonatal 2, 610374 Hyperinsulinemic hypoglycemia, familial, 1, 256450 Hypoglycemia of infancy, leucine-sensitive, 240800
ABCC9	157.8	99	99	Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 10, 608569 Hypertrichotic osteochondrodysplasia, 239850
ABCD1	76.6	74	68	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABCD4	144.9	99	98	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	145.4	100	99	Sitosterolemia, 210250
ABCG8	148.7	99	96	Sitosterolemia, 210250 {Gallbladder disease 4}, 611465
ABHD12	107	97	87	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ABHD5	209.6	100	99	Chanarin-Dorfman syndrome, 275630
ABL1	142	100	99	Leukemia, Philadelphia chromosome-positive, resistant to imatinib
ACAD8	135.4	100	100	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	135.4	98	95	Mitochondrial complex I deficiency due to ACAD9 deficiency, 611126
ACADM	101.3	98	95	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	124.2	99	97	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	119.2	99	95	2-methylbutyrylglycinuria, 610006

ACADVL	118.9	98	95	VLCAD deficiency, 201475
ACAN	121.8	91	85	Osteochondritis dissecans, short stature, and early-onset osteoarthritis, 165800 Spondyloepimetaphyseal dysplasia, aggrecan type, 612813 Spondyloepiphyseal dysplasia, Kimberley type, 608361
ACAT1	123.7	99	94	Alpha-methylacetoacetic aciduria, 203750
ACE	114.8	93	92	Renal tubular dysgenesis, 267430 [Angiotensin I-converting enzyme, benign serum increase] {Microvascular complications of diabetes 3}, 612624 {Myocardial infarction, susceptibility to} {SARS, progression of} {Stroke, hemorrhagic}, 614519
ACO2	129.6	95	91	Infantile cerebellar-retinal degeneration, 614559 ?Optic atrophy 9, 616289
ACOX1	155.5	100	100	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACP5	196.7	100	99	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACSF3	128.8	99	99	Combined malonic and methylmalonic aciduria, 614265
ACSL4	105.2	97	92	Mental retardation, X-linked 63, 300387
ACSL6	119.5	99	98	Myelodysplastic syndrome Myelogenous leukemia, acute
ACTA1	100	99	95	Myopathy, actin, congenital, with cores, 161800 Myopathy, actin, congenital, with excess of thin myofilaments, 161800 Myopathy, congenital, with fiber-type disproportion 1, 255310 Nemaline myopathy 3, autosomal dominant or recessive, 161800 ?Myopathy, scapulohumeroperoneal, 616852
ACTA2	138	100	99	Aortic aneurysm, familial thoracic 6, 611788 Moyamoya disease 5, 614042 Multisystemic smooth muscle dysfunction syndrome, 613834
ACTB	129.3	99	94	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ACTC1	164.4	100	99	Atrial septal defect 5, 612794 Cardiomyopathy, dilated, 1R, 613424 Cardiomyopathy, hypertrophic, 11, 612098 Left ventricular noncompaction 4, 613424
ACTG1	149.4	100	100	Baraitser-Winter syndrome 2, 614583 Deafness, autosomal dominant 20/26, 604717

ACTG2	133.5	99	97	Visceral myopathy, 155310
ACTN1	143.9	100	99	Bleeding disorder, platelet-type, 15, 615193
ACTN2	159.6	100	100	Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158 Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158
ACTN4	143.9	99	97	Glomerulosclerosis, focal segmental, 1, 603278
ACVR1	165.2	100	100	Fibrodysplasia ossificans progressiva, 135100
ACVR1B	160.4	96	95	Pancreatic cancer, somatic
ACVR2B	140.6	97	94	Heterotaxy, visceral, 4, autosomal, 613751
ACVRL1	123	99	98	Telangiectasia, hereditary hemorrhagic, type 2, 600376
ACY1	130.4	100	98	Aminoacylase 1 deficiency, 609924
ADA	113.2	98	97	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADAM10	130	99	96	Reticulate acropigmentation of Kitamura, 615537 {Alzheimer disease 18, susceptibility to}, 615590
ADAM9	146.3	98	93	Cone-rod dystrophy 9, 612775
ADAMTS10	108	99	98	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS13	100	96	91	Thrombotic thrombocytopenic purpura, familial, 274150
ADAMTS17	117.3	88	86	Weill-Marchesani-like syndrome, 613195
ADAMTS18	147.6	99	98	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
ADAMTS2	117.5	98	96	Ehlers-Danlos syndrome, type VIIC, 225410
ADAMTSL2	112.4	96	91	Geleophysic dysplasia 1, 231050
ADAMTSL4	90.7	99	98	Ectopia lentis et pupillae, 225200 Ectopia lentis, isolated, autosomal recessive, 225100
ADAR	122.6	100	99	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
ADAT3	82	99	97	Mental retardation, autosomal recessive 36, 615286
ADCK3	134.7	100	99	Coenzyme Q10 deficiency, primary, 4, 612016
ADCK4	90.6	100	99	Nephrotic syndrome, type 9, 615573
ADCY5	129.5	92	89	Dyskinesia, familial, with facial myokymia, 606703
ADD3	169.6	100	99	Cerebral palsy, spastic quadriplegic, 3, 617008
ADIPOQ	139.6	100	100	Adiponectin deficiency, 612556
ADK	100.5	99	96	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADNP	242.5	100	100	Helsmoortel-van der Aa syndrome, 615873
ADRA2B	200.3	100	100	Epilepsy, myoclonic, familial adult, 2, 607876

ADSL	185.2	100	99	Adenylosuccinase deficiency, 103050
ADSSL1	113.8	89	85	Myopathy, distal, 5, 617030
AFF2	123.6	99	98	Mental retardation, X-linked, FRAXE type, 309548
AFF4	110.6	99	97	CHOPS syndrome, 616368
AFG3L2	121.1	91	84	Spastic ataxia 5, autosomal recessive, 614487 Spinocerebellar ataxia 28, 610246
AGA	130.2	100	100	Aspartylglucosaminuria, 208400
AGBL1	134.4	100	100	Corneal dystrophy, Fuchs endothelial, 8, 615523
AGBL5	111.2	100	99	Retinitis pigmentosa 75,617023
AGK	112.2	99	96	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AGL	145.9	99	98	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGPAT2	109.6	99	95	Lipodystrophy, congenital generalized, type 1, 608594
AGPS	51.7	96	84	Rhizomelic chondrodysplasia punctata, type 3, 600121
AGRN	115.1	95	89	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120
AGT	214.4	100	100	Renal tubular dysgenesis, 267430 {Hypertension, essential, susceptibility to}, 145500 {Preeclampsia, susceptibility to}
AGTR1	149.9	100	100	Renal tubular dysgenesis, 267430 {Hypertension, essential}, 145500
AGXT	139.7	99	99	Hyperoxaluria, primary, type 1, 259900
AHCY	124.6	100	99	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHDC1	121.2	98	96	Xia-Gibbs syndrome, 615829
AHI1	139.1	99	95	Joubert syndrome-3, 608629
AICDA	139.3	89	82	Immunodeficiency with hyper-IgM, type 2, 605258
AIFM1	107	100	99	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Deafness, X-linked 5, 300614
AIMP1	84.7	97	89	Leukodystrophy, hypomyelinating, 3, 260600
AIP	154.5	99	99	Pituitary adenoma, ACTH-secreting, 219090 Pituitary adenoma, growth hormone-secreting, 102200 Pituitary adenoma, prolactin-secreting, 600634

AIPL1	116.3	100	99	Cone-rod dystrophy, 604393 Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393
AIRE	72.7	99	93	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AK1	120.1	100	99	Hemolytic anemia due to adenylate kinase deficiency, 612631
AK2	110.3	99	96	Reticular dysgenesis, 267500
AKR1C2	179.7	96	89	46XY sex reversal 8, 614279 Obesity, hyperphagia, and developmental delay
AKR1D1	106.1	98	94	Bile acid synthesis defect, congenital, 2, 235555
AKT1	154.5	99	99	Breast cancer, somatic, 114480 Colorectal cancer, somatic, 114500 Cowden syndrome 6, 615109 Ovarian cancer, somatic, 167000 Proteus syndrome, somatic, 176920 {Schizophrenia, susceptibility to}, 181500
AKT2	149.1	100	99	Diabetes mellitus, type II, 125853 Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900
AKT3	79.5	97	88	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937
ALAD	100.7	99	97	Porphyria, acute hepatic, 612740 {Lead poisoning, susceptibility to}, 612740
ALAS2	90.3	99	97	Anemia, sideroblastic, 1, 300751 Protoporphyrin, erythropoietic, X-linked, 300752
ALB	156.5	100	98	Analbuminemia, 616000 [Dysalbuminemic hyperthyroxinemia], 615999
ALDH18A1	131.1	100	99	Cutis laxa, autosomal dominant 3, 616603 Cutis laxa, autosomal recessive, type IIIA, 219150 Spastic paraplegia 9A, autosomal dominant, 601162 Spastic paraplegia 9B, autosomal recessive, 616586
ALDH1A3	104.9	93	89	Microphthalmia, isolated 8, 615113
ALDH2	124.9	100	99	Alcohol sensitivity, acute, 610251 {Esophageal cancer, alcohol-related, susceptibility to} {Hangover, susceptibility to}, 610251 {Sublingual nitroglycerin, susceptibility to poor response to}
ALDH3A2	131.7	100	99	Sjogren-Larsson syndrome, 270200

ALDH4A1	116.3	100	98	Hyperprolinemia, type II, 239510
ALDH5A1	87.7	86	80	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	127.4	100	100	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	78.1	96	88	Epilepsy, pyridoxine-dependent, 266100
ALDOA	181.6	99	97	Glycogen storage disease XII, 611881
ALDOB	166	100	99	Fructose intolerance, 229600
ALG1	51	53	48	Congenital disorder of glycosylation, type Ik, 608540
ALG11	144.2	99	99	Congenital disorder of glycosylation, type Ip, 613661
ALG12	156.3	100	100	Congenital disorder of glycosylation, type Ig, 607143
ALG13	87.4	98	94	Epileptic encephalopathy, early infantile, 36, 300884
ALG2	115.9	100	100	Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 ?Congenital disorder of glycosylation, type li, 607906
ALG3	133	100	100	Congenital disorder of glycosylation, type Id, 601110
ALG6	96.3	96	93	Congenital disorder of glycosylation, type Ic, 603147
ALG8	126	96	95	Congenital disorder of glycosylation, type Ih, 608104
ALG9	117.1	100	99	Congenital disorder of glycosylation, type Il, 608776 Gillessen-Kaesbach-Nishimura syndrome, 263210
ALMS1	179.4	99	99	Alstrom syndrome, 203800
ALOX12B	130.9	100	99	Ichthyosis, congenital, autosomal recessive 2, 242100
ALOXE3	122.3	100	100	Ichthyosis, congenital, autosomal recessive 3, 606545
ALPL	156.7	100	100	Hypophosphatasia, adult, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500 Odontohypophosphatasia, 146300
ALS2	167.7	99	98	Amyotrophic lateral sclerosis 2, juvenile, 205100 Primary lateral sclerosis, juvenile, 606353 Spastic paralysis, infantile onset ascending, 607225
ALX3	103	73	70	Frontonasal dysplasia 1, 136760
ALX4	132.7	98	92	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597 {Craniosynostosis 5, susceptibility to}, 615529
AMACR	149.1	100	99	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
AMBN	175.5	94	88	Amelogenesis imperfecta, type IF, 616270

AMELX	98.7	99	95	Amelogenesis imperfecta, type 1E, 301200
AMER1	97.6	99	98	Osteopathia striata with cranial sclerosis, 300373
AMH	42.3	92	74	Persistent Mullerian duct syndrome, type I, 261550
AMHR2	158.2	100	99	Persistent Mullerian duct syndrome, type II, 261550
AMN	66.9	83	71	Megaloblastic anemia-1, Norwegian type, 261100
AMPD1	126.7	100	99	Myopathy due to myoadenylate deaminase deficiency, 615511
AMPD2	135.6	99	99	Pontocerebellar hypoplasia, type 9, 615809 ?Spastic paraplegia 63, 615686
AMT	173.3	100	100	Glycine encephalopathy, 605899
ANG	178.3	100	99	Amyotrophic lateral sclerosis 9, 611895
ANGPTL3	89.8	97	92	Hypobetalipoproteinemia, familial, 2, 605019
ANK1	136.7	99	99	Spherocytosis, type 1, 182900
ANK2	160.4	100	99	Cardiac arrhythmia, ankyrin-B-related, 600919 Long QT syndrome 4, 600919
ANKH	118.7	100	99	Chondrocalcinosis 2, 118600 Cranio metaphyseal dysplasia, 123000
ANKK1	165	100	99	Dopamine receptor D2, reduced brain density of
ANKRD11	96.4	97	94	KBG syndrome, 148050
ANKRD26	79.7	89	77	Thrombocytopenia 2, 188000
ANKS6	91.8	92	88	Nephronophthisis 16, 615382
ANLN	146.1	97	93	Focal segmental glomerulosclerosis 8, 616032
ANO10	116.6	98	96	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANO3	138.7	99	98	Dystonia 24, 615034
ANO5	142.1	99	96	Gnathodiaphyseal dysplasia, 166260 Miyoshi muscular dystrophy 3, 613319 Muscular dystrophy, limb-girdle, type 2L, 611307
ANO6	137.2	98	92	Scott syndrome, 262890
ANTXR1	123.2	98	95	GAP0 syndrome, 230740 {Hemangioma, capillary infantile, susceptibility to}, 602089
ANTXR2	96.1	98	94	Hyaline fibromatosis syndrome, 228600
AP1S1	111.3	99	99	MEDNIK syndrome, 609313
AP1S2	63.2	72	61	Mental retardation, X-linked syndromic 5, 304340
AP2S1	116.1	90	89	Hypocalciuric hypercalcemia, familial, type III, 600740
AP3B1	95	97	90	Hermansky-Pudlak syndrome 2, 608233



AP4B1	146.6	100	99	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	98.7	99	97	Spastic paraplegia 51, autosomal recessive, 613744 Stuttering, familial persistent, 1, 184450
AP4M1	127.3	99	96	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	65	71	68	Spastic paraplegia 52, autosomal recessive, 614067
AP5Z1	97	99	96	Spastic paraplegia 48, autosomal recessive, 613647
APC	159	99	98	Adenoma, periampullary, somatic Adenomatous polyposis coli, 175100 Brain tumor-polyposis syndrome 2, 175100 Colorectal cancer, somatic, 114500 Desmoid disease, hereditary, 135290 Gardner syndrome, 175100 Gastric cancer, somatic, 613659 Hepatoblastoma, somatic, 114550
APCDD1	179.9	100	99	Hypotrichosis 1, 605389
APOA1	99.3	100	100	Amyloidosis, 3 or more types, 105200 ApoA-I and apoC-III deficiency, combined Corneal clouding, autosomal recessive Hypoalphalipoproteinemia, 604091
APOA2	106.3	88	82	Apolipoprotein A-II deficiency {Hypercholesterolemia, familial, modifier of}, 143890
APOA5	152.7	100	100	Hyperchylomicronemia, late-onset, 144650 {Hypertriglyceridemia, susceptibility to}, 145750
APOB	181.6	99	99	Hypercholesterolemia, due to ligand-defective apo B, 144010 Hypobetalipoproteinemia, 615558
APOC2	99.8	100	100	Hyperlipoproteinemia, type Ib, 207750
APOC3	89	100	100	Apolipoprotein C-III deficiency, 614028
APOE	56.4	93	83	Alzheimer disease-2, 104310 Hyperlipoproteinemia, type III Lipoprotein glomerulopathy, 611771 Sea-blue histiocyte disease, 269600 {?Macular degeneration, age-related}, 603075 {Coronary artery disease, severe, susceptibility to}, 617347
APOPT1	67.9	86	83	Mitochondrial complex IV deficiency, 220110

APP	139	100	100	Alzheimer disease 1, familial, 104300 Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants, 605714
APRT	68.4	100	98	Adenine phosphoribosyltransferase deficiency, 614723
APTX	119.1	94	91	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
AQP2	117.5	99	95	Diabetes insipidus, nephrogenic, 125800
AQP5	111	99	97	Palmoplantar keratoderma, Bothnian type, 600231
AR	82.3	93	88	Androgen insensitivity, 300068 Androgen insensitivity, partial, with or without breast cancer, 312300 Hypospadias 1, X-linked, 300633 Spinal and bulbar muscular atrophy of Kennedy, 313200 {Prostate cancer, susceptibility to}, 176807
ARFGEF2	155	100	99	Periventricular heterotopia with microcephaly, 608097
ARG1	167.9	100	100	Argininemia, 207800
ARHGAP26	153.3	100	99	Leukemia, juvenile myelomonocytic, somatic, 607785
ARHGAP31	133.5	99	98	Adams-Oliver syndrome 1, 100300
ARHGDI1	143.9	100	99	Nephrotic syndrome, type 8, 615244
ARHGEF6	130.3	96	92	Mental retardation, X-linked 46, 300436
ARHGEF9	79.2	99	97	Epileptic encephalopathy, early infantile, 8, 300607
ARID1A	150.3	92	89	Coffin-Siris syndrome 2, 614607
ARID1B	157.6	94	89	Coffin-Siris syndrome 1, 135900
ARL13B	97.3	98	92	Joubert syndrome 8, 612291
ARL2BP	66.4	88	79	Retinitis pigmentosa with or without situs inversus, 615434
ARL6	85.2	99	95	Bardet-Biedl syndrome 3, 600151 ?Retinitis pigmentosa 55, 613575 {Bardet-Biedl syndrome 1, modifier of}, 209900
ARMC4	121.6	91	89	Ciliary dyskinesia, primary, 23, 615451
ARMC5	136.7	99	97	ACTH-independent macronodular adrenal hyperplasia 2, 615954
ARSA	97.9	100	99	Metachromatic leukodystrophy, 250100
ARSB	117.4	94	87	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARSE	102.9	99	93	Chondrodysplasia punctata, X-linked recessive, 302950
ARV1	133.5	100	99	Epileptic encephalopathy, early infantile, 38, 617020

ARX	29.3	76	59	Epileptic encephalopathy, early infantile, 1, 308350 Hydranencephaly with abnormal genitalia, 300215 Lissencephaly, X-linked 2, 300215 Mental retardation, X-linked 29 and others, 300419 Partington syndrome, 309510 Proud syndrome, 300004
ASAH1	116.8	99	95	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASB10	106	99	97	Glaucoma 1, open angle, F, 603383
ASCC1	139.7	96	93	Barrett esophagus/esophageal adenocarcinoma, 614266 ?Spinal muscular atrophy with congenital bone fractures 2,616867
ASCL1	213.7	90	79	Central hypoventilation syndrome, congenital, 209880 Haddad syndrome, 209880
ASL	114.6	99	98	Argininosuccinic aciduria, 207900
ASNS	105.6	97	90	Asparagine synthetase deficiency, 615574
ASPA	127.5	99	95	Canavan disease, 271900
ASPH	117.1	98	93	Traboulsi syndrome, 601552
ASPM	101	98	92	Microcephaly 5, primary, autosomal recessive, 608716
ASPSCR1	88.5	98	93	Alveolar soft-part sarcoma, 606243
ASS1	97.8	95	87	Citrullinemia, 215700
ASXL1	159.8	99	97	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ASXL3	162.8	99	98	Bainbridge-Ropers syndrome, 615485
ATCAY	149.5	100	99	Ataxia, cerebellar, Cayman type, 601238
ATF6	133.9	100	99	Achromatopsia 7, 616517
ATIC	119.6	99	99	AICA-ribosiduria due to ATIC deficiency, 608688
ATL1	160.3	99	97	Neuropathy, hereditary sensory, type ID, 613708 Spastic paraplegia 3A, autosomal dominant, 182600
ATL3	119.7	98	93	Neuropathy, hereditary sensory, type IF, 615632
ATM	109.8	99	94	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic Lymphoma, mantle cell, somatic T-cell prolymphocytic leukemia, somatic {Breast cancer, susceptibility to}, 114480

ATN1	121.2	99	97	Dentatorubro-pallidoluysian atrophy, 125370
ATOH7	102.8	95	89	Persistent hyperplastic primary vitreous, autosomal recessive, 221900
ATP13A2	117.5	100	98	Kufor-Rakeb syndrome, 606693 ?Ceroid lipofuscinosis, neuronal, 12, 606693
ATP1A2	191.1	100	99	Alternating hemiplegia of childhood, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481
ATP1A3	177.6	100	100	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235
ATP2A1	156.1	100	100	Brody myopathy, 601003
ATP2A2	175.4	100	99	Acrokeratosis verruciformis, 101900 Darier disease, 124200
ATP2C1	118.5	99	99	Hailey-Hailey disease, 169600
ATP6AP1	113.7	99	94	Immunodeficiency 47, 300972
ATP6V0A2	132.3	100	99	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250
ATP6V0A4	116.6	99	98	Renal tubular acidosis, distal, autosomal recessive, 602722
ATP6V1B1	176.8	100	100	Renal tubular acidosis with deafness, 267300
ATP6V1B2	136.8	99	98	Deafness, congenital, with onychodystrophy, autosomal dominant, 124480 Zimmerman-Laband syndrome 2, 616455
ATP7A	134.1	99	97	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATP7B	169.1	100	99	Wilson disease, 277900
ATP8B1	139.1	96	94	Cholestasis, benign recurrent intrahepatic, 243300 Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, progressive familial intrahepatic 1, 211600
ATPAF2	101.5	100	100	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
ATR	138.2	99	96	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
ATRX	83	98	92	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Alpha-thalassemia/mental retardation syndrome, 301040 Mental retardation-hypotonic facies syndrome, X-linked, 309580

ATXN1	160.1	100	100	Spinocerebellar ataxia 1, 164400
ATXN10	139.6	99	98	Spinocerebellar ataxia 10, 603516
ATXN2	89.3	85	77	Spinocerebellar ataxia 2, 183090 {Amyotrophic lateral sclerosis, susceptibility to, 13}, 183090 {Parkinson disease, late-onset, susceptibility to}, 168600
ATXN3	102.6	97	91	Machado-Joseph disease, 109150
ATXN7	131.2	96	94	Spinocerebellar ataxia 7, 164500
ATXN8OS	NC	NC	NC	Spinocerebellar ataxia 8, 608768
AUH	86.6	99	97	3-methylglutaconic aciduria, type I, 250950
AURKC	78.9	99	97	Spermatogenic failure 5,243060
AUTS2	110.4	96	95	Mental retardation, autosomal dominant 26,615834
AVP	53.5	76	57	Diabetes insipidus, neurohypophyseal, 125700
AVPR2	128.6	99	97	Diabetes insipidus, nephrogenic, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539
AXIN1	141.3	98	96	?Caudal duplication anomaly, 607864 Hepatocellular carcinoma, somatic, 114550
AXIN2	117.5	100	99	Colorectal cancer, somatic, 114500 Oligodontia-colorectal cancer syndrome, 608615
B2M	244	100	99	Immunodeficiency 43, 241600 ?Amyloidosis, familial visceral, 105200
B3GALNT2	115	92	89	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B3GALT6	47.6	76	71	Ehlers-Danlos syndrome, progeroid type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B3GALTL	101.2	97	93	Peters-plus syndrome, 261540
B3GAT3	93.5	99	95	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B3GNT1	120.6	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 13, 615287
B4GALNT1	151.3	95	90	Spastic paraplegia 26, autosomal recessive, 609195
B4GALT1	105.5	99	99	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	104.4	96	95	Ehlers-Danlos syndrome with short stature and limb anomalies, 130070
B9D2	111	100	100	Meckel syndrome 10, 614175
BAAT	121	98	95	Hypercholanemia, familial, 607748
BAG3	136.9	100	100	Cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954

BANF1	58.2	97	88	Nestor-Guillermo progeria syndrome, 614008
BAP1	135.5	99	98	Tumor predisposition syndrome, 614327
BAX	82.8	86	84	Colorectal cancer, somatic, 114500 T-cell acute lymphoblastic leukemia, somatic, 613065
BBS1	149.1	100	100	Bardet-Biedl syndrome 1, 209900
BBS10	172.3	100	100	Bardet-Biedl syndrome 10, 615987
BBS12	208.7	100	100	Bardet-Biedl syndrome 12, 615989
BBS2	181.9	100	99	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	135.2	99	97	Bardet-Biedl syndrome 4, 615982
BBS5	110.5	97	90	Bardet-Biedl syndrome 5, 615983
BBS7	120.5	98	91	Bardet-Biedl syndrome 7, 615984
BBS9	112.5	96	93	Bardet-Biedl syndrome 9, 615986
BCAP31	71.2	93	82	Deafness, dystonia, and cerebral hypomyelination, 300475
BCHE	162.9	100	99	Apnea, postanesthetic
BCKDHA	172.8	100	99	Maple syrup urine disease, type Ia, 248600
BCKDHB	112.6	88	81	Maple syrup urine disease, type Ib, 248600
BCKDK	179.3	100	100	Branched-chain ketoacid dehydrogenase kinase deficiency, 614923
BCL10	97.7	100	99	?Immunodeficiency 37, 616098 Lymphoma, MALT, somatic, 137245 {Lymphoma, follicular, somatic}, 605027 {Male germ cell tumor, somatic}, 273300, {Mesothelioma, somatic}, 156240 {Sezary syndrome, somatic},
BCL11A	141.2	98	97	Intellectual development disorder with persistence of fetal hemoglobin, 617101
BCL2	156.5	99	98	Leukemia/lymphoma, B-cell, 2
BCL7A	136.2	100	100	B-cell non-Hodgkin lymphoma, high-grade
BCMO1	165.1	100	100	Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300
BCOR	111	99	96	Microphthalmia, syndromic 2, 300166
BCR	110.1	88	84	Leukemia, acute lymphocytic, somatic, 613065 Leukemia, chronic myeloid, somatic, 608232
BCS1L	182.6	100	100	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000

				Mitochondrial complex III deficiency, nuclear type 1, 124000
BDNF	190.5	100	100	Central hypoventilation syndrome, congenital, 209880 {Anorexia nervosa, susceptibility to}, 610269 {Bulimia nervosa, age of onset of weight loss in}, 607499 {Memory impairment, susceptibility to} {Obsessive-compulsive disorder, protection against}, 164230
BEAN1	146.4	99	95	Spinocerebellar ataxia 31,117210
BEST1	144.6	99	97	Bestrophinopathy, autosomal recessive, 611809 Macular dystrophy, vitelliform, 2, 153700 Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma, 193220 Retinitis pigmentosa, concentric, 613194 Retinitis pigmentosa-50, 613194 Vitreoretinopathopathy, 193220
BFSP1	98.1	98	89	Cataract 33, 611391
BFSP2	89.5	99	97	Cataract 12, multiple types, 611597
BGN	129.9	100	99	Meester-Loeys syndrome, 300989 Spondyloepimetaphyseal dysplasia, X-linked, 300106
BHLHA9	13.9	58	41	Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432 ?Camptosynpolydactyly, complex, 607539
BICD2	159	100	99	Spinal muscular atrophy, lower extremity-predominant, 2, AD, 615290
BIN1	100.3	99	95	Myopathy, centronuclear, autosomal recessive, 255200
BLK	115.9	100	100	Maturity-onset diabetes of the young, type 11, 613375
BLM	116.3	99	96	Bloom syndrome, 210900
BLNK	94.2	94	91	Agammaglobulinemia 4, 613502
BLOC1S3	28.7	88	64	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	97.3	98	91	Hermansky-pudlak syndrome 9, 614171
BLVRA	125.3	100	99	Hyperbiliverdinemia, 614156
BMP1	141.6	99	99	Osteogenesis imperfecta, type XIII, 614856
BMP15	121.1	100	99	Ovarian dysgenesis 2, 300510 Premature ovarian failure 4, 300510
BMP2	173.7	100	99	Brachydactyly, type A2, 112600 {HFE hemochromatosis, modifier of}, 235200

BMP4	151.9	100	99	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625
BMPER	159.9	99	98	Diaphanospondylodysostosis, 608022
BMPR1A	98	99	94	Juvenile polyposis syndrome, infantile form, 174900 Polyposis syndrome, hereditary mixed, 2, 610069 Polyposis, juvenile intestinal, 174900
BMPR1B	172.1	100	98	Acromesomelic dysplasia, Demirhan type, 609441 Brachydactyly, type A1, D, 616849 Brachydactyly, type A2, 112600
BMPR2	195	100	99	Pulmonary hypertension, familial primary, 1, with or without HHT, 178600 Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600 Pulmonary venoocclusive disease 1, 265450
BOLA3	50.1	92	81	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BPGM	130.7	100	100	Erythrocytosis due to bisphosphoglycerate mutase deficiency, 222800
BRAF	74.3	87	77	Adenocarcinoma of lung, somatic, 211980 Cardiofaciocutaneous syndrome, 115150 Colorectal cancer, somatic LEOPARD syndrome 3, 613707 Melanoma, malignant, somatic Nonsmall cell lung cancer, somatic Noonan syndrome 7, 613706
BRAT1	108.7	99	97	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BRCA2	102.6	99	97	Fanconi anemia, complementation group D1, 605724 Wilms tumor, 194070 {Breast cancer, male, susceptibility to}, 114480 {Breast-ovarian cancer, familial, 2}, 612555 {Glioblastoma 3}, 613029 {Medulloblastoma}, 155255 {Pancreatic cancer 2}, 613347 {Prostate cancer}, 176807
BRF1	100.2	96	93	Cerebellofaciodental syndrome, 616202
BRIP1	117.8	99	97	Breast cancer, early-onset, 114480 Fanconi anemia, complementation group J, 609054
BRWD3	97.7	97	92	Mental retardation, X-linked 93, 300659



BSCL2	113.7	100	100	Encephalopathy, progressive, with or without lipodystrophy, 615924 Lipodystrophy, congenital generalized, type 2, 269700 Neuropathy, distal hereditary motor, type VA, 600794 Silver spastic paraplegia syndrome, 270685
BSND	137.5	100	100	Bartter syndrome, type 4a, 602522 Sensorineural deafness with mild renal dysfunction, 602522
BTD	166.9	100	99	Biotinidase deficiency, 253260
BTK	115.5	100	99	Agammaglobulinemia and isolated hormone deficiency, 307200 Agammaglobulinemia, X-linked 1, 300755
BUB1	136.4	99	97	Colorectal cancer with chromosomal instability, somatic
BUB1B	136.4	98	97	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430
C10orf11	154.5	99	99	Albinism, oculocutaneous, type VII, 615179
C10orf2	179	100	100	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Perrault syndrome 5, 616138 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286
C11orf73	61.7	91	79	Leukodystrophy, hypomyelinating, 13, 616881
C12orf57	152.5	100	100	Temtamy syndrome, 218340
C12orf65	88.1	97	91	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035
C15orf41	124.8	99	97	Dyserythropoietic anemia, congenital, type Ib, 615631
C19orf12	94.3	100	99	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
C1GALT1C1	148.1	99	98	Tn polyagglutination syndrome, somatic, 300622
C1QA	120.3	100	99	C1q deficiency, 613652
C1QB	183.5	100	99	C1q deficiency, 613652
C1QC	198.3	100	99	C1q deficiency, 613652
C1QTNF5	151.4	80	65	Retinal degeneration, late-onset, autosomal dominant, 605670
C1S	121.1	100	99	C1s deficiency, 613783
C2	20	15	15	C2 deficiency, 217000 {Macular degeneration, age-related, 14, reduced risk of}, 615489
C21orf59	145.8	98	94	Ciliary dyskinesia, primary, 26, 615500
C2orf71	124.7	99	98	Retinitis pigmentosa 54, 613428

C3	145.8	100	99	C3 deficiency, 613779 {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 {Macular degeneration, age-related, 9}, 611378
C4A	22.8	24	24	C4a deficiency, 614380 [Blood group, Rodgers], 614374
C4B	22.5	24	24	C4B deficiency, 614379
C4orf26	198.1	100	100	Amelogenesis imperfecta, type IIA4, 614832
C5	134.4	98	95	C5 deficiency, 609536 [Eculizumab, poor response to], 615749
C5orf42	122.4	98	95	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
C6	157.5	100	99	C6 deficiency, 612446 Combined C6/C7 deficiency
C7	132.2	99	94	C7 deficiency, 610102
C7orf10	125.4	94	88	Glutaric aciduria III, 231690
C8A	120.1	100	99	C8 deficiency, type I, 613790
C8B	135.9	99	99	C8 deficiency, type II, 613789
C8orf37	126.4	100	99	Cone-rod dystrophy 16, 614500 Retinitis pigmentosa 64, 614500
C9	133.5	100	98	C9 deficiency, 613825 {Macular degeneration, age-related, 15, susceptibility to}, 615591
C9orf72	108.5	99	94	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1, 105550
CA12	109.7	100	100	Hyperchlorhidrosis, isolated, 143860
CA2	140.5	100	99	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA4	142.6	100	99	Retinitis pigmentosa 17, 600852
CA5A	124.2	99	95	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CA8	114.5	96	92	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CABP2	83.3	92	81	Deafness, autosomal recessive 93, 614899
CABP4	98.5	99	97	Cone-rod synaptic disorder, congenital nonprogressive, 610427
CACNA1A	87.9	92	89	Episodic ataxia, type 2, 108500 Migraine, familial hemiplegic, 1, 141500 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Spinocerebellar ataxia 6, 183086
CACNA1C	155.1	99	99	Brugada syndrome 3, 611875

				Timothy syndrome, 601005
CACNA1D	152.9	100	99	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896
CACNA1F	100.2	99	98	Aland Island eye disease, 300600 Cone-rod dystrophy, X-linked, 3, 300476 Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071
CACNA1G	133	99	97	Spinocerebellar ataxia 42, 616795
CACNA1H	103.5	97	94	Hyperaldosteronism, familial, type IV, 617027 {Epilepsy, childhood absence, susceptibility to, 6}, 611942 {Epilepsy, idiopathic generalized, susceptibility to, 6}, 611942
CACNA1S	135.7	100	99	Hypokalemic periodic paralysis, type 1, 170400 {Malignant hyperthermia susceptibility 5}, 601887 {Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580
CACNA2D4	112.1	99	97	Retinal cone dystrophy 4, 610478
CACNB2	151	99	96	Brugada syndrome 4, 611876
CACNB4	107.6	97	95	Episodic ataxia, type 5, 613855 {Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 {Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682
CACNG2	114.6	100	99	Mental retardation, autosomal dominant 10, 614256
CALM1	114.2	100	99	Long QT syndrome 14, 616247 Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916
CALM2	54.1	67	65	Long QT syndrome 15, 616249
CALR	113.6	99	97	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
CAMTA1	186.2	99	98	Cerebellar ataxia, nonprogressive, with mental retardation, 614756
CANT1	142.4	100	99	Desbuquois dysplasia 1, 251450
CAPN1	146.3	100	100	Spastic paraplegia 76, autosomal recessive, 616907
CAPN3	109.9	98	96	Muscular dystrophy, limb-girdle, type 2A, 253600
CAPN5	166.4	100	99	Vitreoretinopathy, neovascular inflammatory, 193235
CARD11	154.9	99	98	B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11, 615206
CARD14	116.5	99	97	Pityriasis rubra pilaris, 173200 Psoriasis 2, 602723
CARD9	120.6	98	96	Candidiasis, familial, 2, autosomal recessive, 212050

CARS2	121.2	100	99	Combined oxidative phosphorylation deficiency 27, 616672
CASC5	113.7	98	95	Microcephaly 4, primary, autosomal recessive, 604321
CASK	93	98	93	FG syndrome 4, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 Mental retardation, with or without nystagmus, 300422
CASP10	117.6	99	98	Autoimmune lymphoproliferative syndrome, type II, 603909 Gastric cancer, somatic, 613659 Lymphoma, non-Hodgkin, somatic, 605027
CASP8	151.4	100	99	Hepatocellular carcinoma, somatic, 114550 ?Autoimmune lymphoproliferative syndrome, type IIB, 607271 {Breast cancer, protection against}, 114480 {Lung cancer, protection against}, 211980
CASQ1	122.1	100	99	Myopathy, vacuolar, with CASQ1 aggregates, 616231
CASQ2	143.2	99	99	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CASR	178	100	99	Hypercalciuric hypercalcemia Hyperparathyroidism, neonatal, 239200 Hypocalcemia, autosomal dominant, 601198 Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 Hypocalciuric hypercalcemia, type I, 145980 {Calcium, serum level of} {Epilepsy idiopathic generalized, susceptibility to, 8}, 612899
CAST	110.2	96	92	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295
CAT	148.5	100	100	Acatlasemia, 614097
CATSPER1	147.3	99	99	Spermatogenic failure 7, 612997
CAV1	265.7	100	100	Pulmonary hypertension, primary, 3, 615343 ?Lipodystrophy, congenital generalized, type 3, 612526 ?Partial lipodystrophy, congenital cataracts, and neurodegeneration syndrome, 606721
CAV3	305.2	100	100	Cardiomyopathy, familial hypertrophic, 192600 Creatine phosphokinase, elevated serum, 123320 Long QT syndrome 9, 611818 Muscular dystrophy, limb-girdle, type IC, 607801 Myopathy, distal, Tateyama type, 614321 Rippling muscle disease, 606072
CBL	133.2	99	98	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785

CBS	114.4	97	91	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CC2D1A	119.6	99	98	Mental retardation, autosomal recessive 3, 608443
CC2D2A	127.4	99	97	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284
CCBE1	76	99	95	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CCDC103	117	100	99	Ciliary dyskinesia, primary, 17, 614679
CCDC11	146.5	97	94	Heterotaxy, visceral, 6, autosomal recessive, 614779
CCDC114	120.7	100	99	Ciliary dyskinesia, primary, 20, 615067
CCDC115	76.3	90	88	Congenital disorder of glycosylation, type Ilo, 616828
CCDC151	113.2	100	99	Ciliary dyskinesia, primary, 30, 616037
CCDC174	133.7	98	93	Hypotonia, infantile, with psychomotor retardation, 616816
CCDC22	94	97	89	Ritscher-Schinzel syndrome 2, 300963
CCDC39	76.2	96	89	Ciliary dyskinesia, primary, 14, 613807
CCDC40	126.6	98	97	Ciliary dyskinesia, primary, 15, 613808
CCDC41	96.9	98	89	Nephronophthisis 18, 615862
CCDC65	105.9	99	97	Ciliary dyskinesia, primary, 27, 615504
CCDC78	115	100	100	Myopathy, centronuclear, 4, 614807
CCDC8	112.1	100	100	3-M syndrome 3, 614205
CCDC88A	78.8	94	84	PEHO syndrome, 260565
CCDC88C	101.6	99	97	Hydrocephalus, nonsyndromic, autosomal recessive, 236600 ?Spinocerebellar ataxia 40, 616053
CCM2	159.9	97	97	Cerebral cavernous malformations-2,603284
CCND2	152.5	100	100	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938
CCNO	103	99	95	Ciliary dyskinesia, primary, 29, 615872
CCT5	164.6	99	99	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CD151	132.5	100	100	Nephropathy with pretibial epidermolysis bullosa and deafness, 609057 [Blood group, Raph], 179620
CD19	88.8	99	98	Immunodeficiency, common variable, 3, 613493
CD27	118.4	100	99	Lymphoproliferative syndrome 2, 615122
CD2AP	98.1	99	96	Glomerulosclerosis, focal segmental, 3, 607832
CD320	92.1	100	99	Methylmalonic aciduria due to transcobalamin receptor defect, 613646

CD36	123.2	99	95	Platelet glycoprotein IV deficiency, 608404 [Macrothrombocytopenia] {Coronary heart disease, susceptibility to, 7}, 610938 {Malaria, cerebral, reduced risk of}, 611162 {Malaria, cerebral, susceptibility to}, 611162
CD3D	194	100	100	Immunodeficiency 19, 615617
CD3E	152.3	100	99	Immunodeficiency 18, 615615 Immunodeficiency 18, SCID variant, 615615
CD3G	157.1	100	100	Immunodeficiency 17, CD3 gamma deficient, 615607
CD4	120.1	100	99	OKT4 epitope deficiency, 613949
CD40	172.6	100	99	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	127.5	96	87	Immunodeficiency, X-linked, with hyper-IgM, 308230
CD59	200.8	93	86	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD79A	128.5	99	97	Agammaglobulinemia 3, 613501
CD79B	211	100	100	Agammaglobulinemia 6, 612692
CD81	143	99	98	Immunodeficiency, common variable, 6, 613496
CD8A	105.8	99	99	CD8 deficiency, familial, 608957
CD96	156.8	100	99	C syndrome, 211750
CDAN1	97.7	97	95	Dyserythropoietic anemia, congenital, type Ia, 224120
CDC14A	164.3	98	93	Deafness,autosomal recessive 105,616958
CDC42	94.3	97	91	Takenouchi-Kosaki syndrome, 616737
CDC45	160.7	99	97	Meier-Gorlin syndrome 7, 617063
CDC6	165.5	99	98	?Meier-Gorlin syndrome 5, 613805
CDC73	102.7	99	97	Hyperparathyroidism, familial primary, 145000 Hyperparathyroidism-jaw tumor syndrome, 145001 Parathyroid adenoma with cystic changes, 145001 Parathyroid carcinoma, 608266
CDCA7	109.2	100	99	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910
CDH1	125.3	99	99	Endometrial carcinoma, somatic, 608089 Gastric cancer, familial diffuse, with or without cleft lip and/or palate, 137215 Ovarian carcinoma, somatic, 167000 {Breast cancer, lobular}, 114480 {Prostate cancer, susceptibility to}, 176807
CDH15	120.8	99	97	Mental retardation, autosomal dominant 3, 612580

CDH23	195.1	100	99	Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D, 601067 Usher syndrome, type 1D/F digenic, 601067
CDH3	159.6	99	97	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 Hypotrichosis, congenital, with juvenile macular dystrophy, 601553
CDHR1	154.4	99	98	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660
CDK5RAP2	123.9	99	98	Microcephaly 3, primary, autosomal recessive, 604804
CDKL5	123.2	99	96	Epileptic encephalopathy, early infantile, 2, 300672
CDKN1B	93.2	100	99	Multiple endocrine neoplasia, type IV, 610755
CDKN1C	21.3	68	51	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732
CDKN2A	87.5	93	92	Melanoma and neural system tumor syndrome, 155755 Orolaryngeal cancer, multiple, Pancreatic cancer/melanoma syndrome, 606719 {Melanoma, cutaneous malignant, 2}, 155601
CDON	143.8	100	99	Holoprosencephaly 11, 614226
CDSN	19.9	16	16	Hypotrichosis 2, 146520 Peeling skin syndrome 1, 270300
CDT1	99.5	96	93	Meier-Gorlin syndrome 4, 613804
CEACAM16	145.3	100	99	Deafness, autosomal dominant 4B, 614614
CEBPA	46.8	75	65	Leukemia, acute myeloid, somatic, 601626 ?Leukemia, acute myeloid, 601626
CEBPE	71.2	99	95	Specific granule deficiency, 245480
CECR1	101.5	99	99	Polyarteritis nodosa, childhood-onset, 615688 ?Sneddon syndrome, 182410
CEL	130.3	85	82	Maturity-onset diabetes of the young, type VIII, 609812
CENPF	139.7	99	97	Stromme syndrome, 243605
CENPJ	141.7	99	97	?Seckel syndrome 4, 613676 Microcephaly 6, primary, autosomal recessive, 608393
CEP104	120.1	99	97	Joubert syndrome 25, 616781
CEP120	128.1	99	98	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP152	162.3	97	94	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823

CEP164	94.4	99	97	Nephronophthisis 15, 614845
CEP19	203	100	100	Morbid obesity and spermatogenic failure, 615703
CEP290	66	88	76	Joubert syndrome 5, 610188 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Senior-Loken syndrome 6, 610189 ?Bardet-Biedl syndrome 14, 615991
CEP41	83.6	97	89	Joubert syndrome 15, 614464
CEP57	92.7	99	93	Mosaic variegated aneuploidy syndrome 2, 614114
CERKL	100.2	98	92	Retinitis pigmentosa 26, 608380
CERS3	106.7	99	98	Ichthyosis, congenital, autosomal recessive 9, 615023
CES1	155.3	99	98	Carboxylesterase 1 deficiency
CETP	132.2	100	100	Hyperalphalipoproteinemia, 143470 [High density lipoprotein cholesterol level QTL 10], 143470
CFC1	76.7	83	73	Heterotaxy, visceral, 2, autosomal, 605376
CFD	80.9	89	81	Complement factor D deficiency, 613912
CFH	183.1	98	95	Basal laminar drusen, 126700 Complement factor H deficiency, 609814 {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 {Macular degeneration, age-related, 4}, 610698
CFHR5	99.6	98	93	Nephropathy due to CFHR5 deficiency, 614809
CFI	152.5	98	96	Complement factor I deficiency, 610984 {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 {Macular degeneration, age-related, 13, susceptibility to}, 615439
CFL2	117.3	94	86	Nemaline myopathy 7, autosomal recessive, 610687
CFP	96.5	98	93	Properdin deficiency,X-linked, 312060
CFTR	124	99	96	Congenital bilateral absence of vas deferens, 277180 Cystic fibrosis, 219700 Sweat chloride elevation without CF {Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400 {Hypertrypsinemia, neonatal} {Pancreatitis, idiopathic}, 167800
CHAMP1	160.6	100	100	Mental retardation, autosomal dominant 40, 616579
CHAT	129.9	89	86	Myasthenic syndrome, congenital, 6, presynaptic, 254210



CHCHD10	20	43	35	Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 Spinal muscular atrophy, Jokela type, 615048 ?Myopathy, isolated mitochondrial, autosomal dominant, 616209
CHCHD2	95.6	99	91	Parkinson disease 22, autosomal dominant, 616710
CHD2	138.7	99	99	Epileptic encephalopathy, childhood-onset, 615369
CHD7	150.9	99	98	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CHEK2	102.4	83	80	Li-Fraumeni syndrome, 609265 Osteosarcoma, somatic, 259500 {Breast and colorectal cancer, susceptibility to} {Breast cancer, susceptibility to}, 114480 {Prostate cancer, familial, susceptibility to}, 176807
CHKB	98.7	100	99	Muscular dystrophy, congenital, megaconial type, 602541
CHM	103.1	96	87	Choroideremia, 303100
CHMP1A	124.3	100	100	Pontocerebellar hypoplasia, type 8, 614961
CHMP2B	91.9	98	91	Amyotrophic lateral sclerosis 17, 614696 Dementia, familial, nonspecific, 600795
CHMP4B	139.8	99	99	Cataract 31, multiple types, 605387
CHN1	141.6	99	97	Duane retraction syndrome 2, 604356
CHRD1	110.6	100	99	Megalocornea 1, X-linked 309300
CHRNA1	128.6	100	99	Multiple pterygium syndrome, lethal type, 253290 Myasthenic syndrome, congenital, 1A, slow-channel, 601462 Myasthenic syndrome, congenital, 1B, fast-channel, 608930
CHRNA2	230.2	100	100	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA4	143.7	96	95	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction, susceptibility to}, 188890
CHRN1	132.1	98	96	Myasthenic syndrome, congenital, 2A, slow-channel, 616313 ?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314
CHRN2	248.7	98	94	Epilepsy, nocturnal frontal lobe, 3, 605375
CHRND	150.9	100	99	Multiple pterygium syndrome, lethal type, 253290 Myasthenic syndrome, congenital, 3B, fast-channel, 616322 ?Myasthenic syndrome, congenital, 3A, slow-channel, 616321 ?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323

CHRNE	127.7	99	95	Myasthenic syndrome, congenital, 4A, slow-channel, 605809 Myasthenic syndrome, congenital, 4B, fast-channel, 616324 Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931
CHRNA	155.4	100	100	Escobar syndrome, 265000 Multiple pterygium syndrome, lethal type, 253290
CHST14	165.7	95	93	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	91.8	100	97	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHST6	334.5	100	100	Macular corneal dystrophy, 217800
CHSY1	138.4	95	93	Temtamy preaxial brachydactyly syndrome, 605282
CHUK	131.6	100	98	Cocoon syndrome, 613630
CIB2	230.2	99	99	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CIITA	125.3	100	100	Bare lymphocyte syndrome, type II, complementation group A, 209920 {Rheumatoid arthritis, susceptibility to}, 180300
CIRH1A	170.8	100	99	Cirrhosis, North American Indian childhood type, 604901
CISD2	128	83	83	Wolfram syndrome 2, 604928
CITED2	114.1	100	99	Atrial septal defect 8, 614433 Ventricular septal defect 2, 614431
CKAP2L	161.3	98	96	Filippi syndrome, 272440
CLCF1	76.4	98	97	Cold-induced sweating syndrome 2, 610313
CLCN1	137.5	100	99	Myotonia congenita, dominant, 160800 Myotonia congenita, recessive, 255700 Myotonia levior, recessive
CLCN2	108.7	100	99	Leukoencephalopathy with ataxia, 615651 {Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628 {Epilepsy, juvenile absence, susceptibility to, 2}, 607628 {Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628
CLCN4	124	100	99	Mental retardation, X-linked 49/15, 300114
CLCN5	135.4	99	98	Dent disease, 300009 Hypophosphatemic rickets, 300554 Nephrolithiasis, type I, 310468 Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990
CLCN7	130	99	98	Osteopetrosis, autosomal dominant 2, 166600 Osteopetrosis, autosomal recessive 4, 611490

CLCNKA	108.7	99	94	Bartter syndrome, type 4b, digenic, 613090
CLCNKB	100.1	98	90	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090
CLDN1	137.8	100	100	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626
CLDN14	130.7	100	99	Deafness, autosomal recessive 29, 614035
CLDN16	136.3	100	99	Hypomagnesemia 3, renal, 248250
CLDN19	123.8	98	93	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLEC7A	150.6	100	99	Candidiasis, familial, 4, autosomal recessive, 613108 {Aspergillosis, susceptibility to}, 614079
CLMP	111.1	100	99	Congenital short bowel syndrome, 615237
CLN3	121.3	98	95	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	146.3	98	92	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	131.8	98	95	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	196.4	100	100	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLP1	181.9	100	99	Pontocerebellar hypoplasia, type 10, 615803
CLPB	135.7	96	95	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CLPP	115.5	99	96	Perrault syndrome 3, 614129
CLRN1	157.2	100	99	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902
CNBP	136.7	100	100	Myotonic dystrophy 2, 602668
CNGA1	127.2	89	84	Retinitis pigmentosa 49, 613756
CNGA3	167.8	100	99	Achromatopsia-2, 216900
CNGB1	102.7	98	94	Retinitis pigmentosa 45, 613767
CNGB3	101.3	97	93	Achromatopsia-3, 262300 Macular degeneration, juvenile, 248200
CNNM2	188.5	100	99	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNNM4	193.1	98	97	Jalili syndrome, 217080
CNTNAP1	161.4	99	97	Lethal congenital contracture syndrome 7, 616286
CNTNAP2	148.1	100	99	Cortical dysplasia-focal epilepsy syndrome, 610042 Pitt-Hopkins like syndrome 1, 610042 {Autism susceptibility 15}, 612100

COA6	78.9	98	91	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 4, 616501
COASY	168.5	100	100	Neurodegeneration with brain iron accumulation 6, 615643
COCH	190.2	99	99	Deafness, autosomal dominant 9, 601369
COG1	124	100	99	Congenital disorder of glycosylation, type IIg, 611209
COG4	123.5	100	99	Congenital disorder of glycosylation, type IIj, 613489
COG5	107	97	93	Congenital disorder of glycosylation, type Iii, 613612
COG6	78.6	94	85	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328
COG7	125.2	100	100	Congenital disorder of glycosylation, type Iie, 608779
COG8	122.7	99	98	Congenital disorder of glycosylation, type IIh, 611182
COL10A1	88.5	98	93	Metaphyseal chondrodysplasia, Schmid type, 156500
COL11A1	90.8	94	89	Fibrochondrogenesis 1, 228520 Marshall syndrome, 154780 Stickler syndrome, type II, 604841 {Lumbar disc herniation, susceptibility to}, 603932
COL11A2	13.2	14	14	Deafness, autosomal dominant 13, 601868 Deafness, autosomal recessive 53, 609706 Fibrochondrogenesis 2, 614524 Otospondylomegaepiphyseal dysplasia, 215150 Stickler syndrome, type III, 184840 Weissenbacher-Zweymuller syndrome, 277610
COL12A1	137.6	99	97	Bethlem myopathy 2, 616471 ?Ullrich congenital muscular dystrophy 2, 616470
COL13A1	85.1	99	97	Myasthenic syndrome, congenital, 19, 616720
COL17A1	108.1	99	96	Epidermolysis bullosa, junctional, localisata variant, 226650 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epithelial recurrent erosion dystrophy, 122400
COL18A1	88.8	93	87	Knobloch syndrome, type 1, 267750
COL1A1	135.1	98	96	Caffey disease, 114000 Ehlers-Danlos syndrome, classic, 130000 Ehlers-Danlos syndrome, type VIIA, 130060 Osteogenesis imperfecta, type I, 166200 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type III, 259420

				Osteogenesis imperfecta, type IV, 166220 {Bone mineral density variation QTL, osteoporosis}, 166710
COL1A2	101.9	96	93	Ehlers-Danlos syndrome, cardiac valvular form, 225320 Ehlers-Danlos syndrome, type VIIB, 130060 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type III, 259420 Osteogenesis imperfecta, type IV, 166220 {Osteoporosis, postmenopausal}, 166710
COL25A1	127.2	98	95	Fibrosis of extraocular muscles, congenital, 5, 616219
COL2A1	103.6	99	98	Achondrogenesis, type II or hypochondrogenesis, 200610 Avascular necrosis of the femoral head, 608805 Czech dysplasia, 609162 Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 Kniest dysplasia, 156550 Legg-Calve-Perthes disease, 150600 Osteoarthritis with mild chondrodysplasia, 604864 Otospondylomegaepiphyseal dysplasia, 215150 Platyspondylic skeletal dysplasia, Torrance type, 151210 SED congenita, 183900 SMED Strudwick type, 184250 Spondyloepiphyseal dysplasia, Stanescu type, 616583 Spondyloperipheral dysplasia, 271700 Stickler syndrome, type I, nonsyndromic ocular, 609508 Stickler syndrome, type I, 108300 Vitreoretinopathy with phalangeal epiphyseal dysplasia
COL3A1	104.2	97	92	Ehlers-Danlos syndrome, type IV, 130050
COL4A1	92.8	97	94	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Brain small vessel disease with or without ocular anomalies, 607595 Porencephaly 1, 175780 ?Retinal arteries, tortuosity of, 180000

				{Hemorrhage, intracerebral, susceptibility to}, 614519
COL4A2	96.9	98	93	Porencephaly 2, 614483 {Hemorrhage, intracerebral, susceptibility to}, 614519
COL4A3	89.5	97	95	Alport syndrome, autosomal dominant, 104200 Alport syndrome, autosomal recessive, 203780 Hematuria, benign familial, 141200
COL4A3BP	121.7	98	92	Mental retardation, autosomal dominant 34, 616351
COL4A4	85	97	93	Alport syndrome, autosomal recessive, 203780 Hematuria, familial benign
COL4A5	52.6	92	77	Alport syndrome, 301050
COL5A1	115.4	97	95	Ehlers-Danlos syndrome, classic type, 130000
COL5A2	89	99	97	Ehlers-Danlos syndrome, classic type, 130000
COL6A1	137.3	99	97	Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090
COL6A2	165.5	99	98	Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090 ?Myosclerosis, congenital, 255600
COL6A3	174.9	100	99	Bethlem myopathy 1, 158810 Dystonia 27, 616411 Ullrich congenital muscular dystrophy 1, 254090
COL7A1	129.8	99	97	EBD inversa, 226600 EBD, Bart type, 132000 EBD, localisata variant Epidermolysis bullosa dystrophica, AD, 131750 Epidermolysis bullosa dystrophica, AR, 226600 Epidermolysis bullosa pruriginosa, 604129 Epidermolysis bullosa, pretibial, 131850 Toenail dystrophy, isolated, 607523 Transient bullous of the newborn, 131705
COL8A2	37.4	84	69	Corneal dystrophy, Fuchs endothelial, 1, 136800 Corneal dystrophy, posterior polymorphous 2, 609140

COL9A1	120.5	99	96	Stickler syndrome, type IV, 614134 /?Epiphyseal dysplasia, multiple, 6, 614135
COL9A2	65.1	98	88	Epiphyseal dysplasia, multiple, 2, 600204 ?Stickler syndrome, type V, 614284 {Intervertebral disc disease, susceptibility to}, 603932
COL9A3	65.4	95	86	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 {Intervertebral disc disease, susceptibility to}, 603932
COLEC11	203.1	100	100	3MC syndrome 2, 265050
COLQ	117	99	98	Myasthenic syndrome, congenital, 5, 603034
COMP	121.8	93	92	Epiphyseal dysplasia, multiple, 1, 132400 Pseudoachondroplasia, 177170
COQ2	89.2	96	93	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ4	89.8	88	84	Coenzyme Q10 deficiency, primary, 7, 616276
COQ6	144.1	99	95	Coenzyme Q10 deficiency, primary, 6, 614650
COQ9	91.6	99	96	Coenzyme Q10 deficiency, primary, 5, 614654
CORIN	179.1	99	99	Preeclampsia/eclampsia 5, 614595
CORO1A	154.7	99	96	Immunodeficiency 8, 615401
COX10	241.9	100	99	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 Mitochondrial complex IV deficiency, 220110
COX15	98.7	100	99	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000
COX20	57.7	82	65	Mitochondrial complex IV deficiency, 220110
COX4I2	120.4	100	100	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX6A1	181.3	100	99	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
COX6B1	159.9	100	100	Mitochondrial complex IV deficiency, 220110
COX7B	48.2	73	41	Linear skin defects with multiple congenital anomalies, 300887
CP	120	93	89	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 [Hypoceruloplasminemia, hereditary], 604290
CPA6	118.3	99	98	Epilepsy, familial temporal lobe, 5, 614417 Febrile seizures, familial, 11, 614418
CPN1	118.2	100	98	Carboxypeptidase N deficiency, 212070
CPOX	116.9	95	88	Coproporphyrinuria, 121300

				Harderoporphyria, 121300
CPS1	143.8	100	99	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}
CPT1A	169.5	100	98	CPT deficiency, hepatic, type IA, 255120
CPT2	166.7	98	97	CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 CPT II deficiency, myopathic, stress-induced 255110 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212
CR2	160.7	100	99	Immunodeficiency, common variable, 7, 614699 {Systemic lupus erythematosus, susceptibility to, 9}, 610927
CRADD	115.1	99	98	Mental retardation, autosomal recessive 34, with variant lissencephaly 614499
CRB1	193.6	100	100	Leber congenital amaurosis 8, 613835 Pigmented paravenous chorioretinal atrophy, 172870 Retinitis pigmentosa-12, autosomal recessive, 600105
CRB2	111.4	99	94	Focal segmental glomerulosclerosis 9, 616220 Ventriculomegaly with cystic kidney disease, 219730
CRBN	148.2	99	95	Mental retardation, autosomal recessive 2, 607417
CREB1	144	98	93	Histiocytoma, angiomatoid fibrous, somatic, 612160
CREBBP	123.5	99	96	Rubinstein-Taybi syndrome, 180849
CRELD1	114.6	99	97	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217 {Atrioventricular septal defect, susceptibility to, 2}, 606217
CRIPT	34.4	96	74	Short stature with microcephaly and distinctive facies, 615789
CRLF1	106	90	89	Cold-induced sweating syndrome 1, 272430
CRTAP	110.6	99	97	Osteogenesis imperfecta, type VII, 610682
CRTC1	135.3	99	95	Mucoepidermoid salivary gland carcinoma
CRX	114.4	99	98	Cone-rod retinal dystrophy-2, 120970 Leber congenital amaurosis 7, 613829
CRYAA	135.7	92	86	Cataract 9, multiple types, 604219
CRYAB	125.9	99	98	Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, 2, 608810 Myopathy, myofibrillar, fatal infantile hypertrophy, alpha-B crystallin-related, 613869
CRYBA1	135.6	100	99	Cataract 10, multiple types, 600881



CRYBA4	117.7	100	100	Cataract 23, 610425
CRYBB1	129.3	100	99	Cataract 17, multiple types, 611544
CRYBB2	150.7	100	100	Cataract 3, multiple types, 601547
CRYBB3	144.4	100	100	Cataract 22, autosomal recessive, 609741
CRYGB	97.3	99	97	Cataract 39, multiple types, autosomal dominant, 615188
CRYGC	128.8	100	99	Cataract 2, multiple types, 604307
CRYGD	100.3	100	99	Cataract 4, multiple types, 115700
CRYGS	105.6	96	87	Cataract 20, multiple types, 116100
CRYM	97.6	99	98	Deafness, autosomal dominant 40, 616357
CSF1R	140	99	98	Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
CSF2RA	66.2	89	88	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF2RB	94.1	99	97	Surfactant metabolism dysfunction, pulmonary, 5, 614370
CSNK1D	148.4	94	90	Advanced sleep-phase syndrome, familial, 2, 615224
CSNK2A1	126.5	94	86	Okur-Chung neurodevelopmental syndrome, 617062 Glaucoma, primary congenital (Lee (2011) Mol Vis 17,3583)
CSPP1	111.9	99	97	Joubert syndrome 21, 615636
CSRP3	103	100	99	Cardiomyopathy, hypertrophic, 12, 612124 ?Cardiomyopathy, dilated, 1M, 607482
CST3	106.6	92	74	Cerebral amyloid angiopathy, 105150 {Macular degeneration, age-related, 11}, 611953
CSTA	119	99	99	Peeling skin syndrome 4, 607936
CSTB	100	98	87	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTC1	119	100	99	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTCF	158.2	98	96	Mental retardation, autosomal dominant 21, 615502
CTDP1	105.3	86	83	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTH	152.8	99	98	Cystathioninuria, 219500 Homocysteine, total plasma, elevated
CTHRC1	90.2	92	84	Barrett esophagus/esophageal adenocarcinoma, 614266
CTLA4	194.1	100	100	Autoimmune lymphoproliferative syndrome, type V, 616100 {Celiac disease, susceptibility to, 3}, 609755 {Diabetes mellitus, insulin-dependent, 12}, 601388 {Hashimoto thyroiditis}, 140300 {Systemic lupus erythematosus, susceptibility to}, 152700

CTNNA1	125.7	99	99	Macular dystrophy, patterned, 608970 Gastric cancer, diffuse (Majewski (2012) J Pathol epub)
CTNNA3	138.2	100	99	Arrhythmogenic right ventricular dysplasia, familial, 13, 615616
CTNNB1	163.8	100	99	Colorectal cancer, somatic, 114500 Hepatocellular carcinoma, somatic, 114550 Mental retardation, autosomal dominant 19, 615075 Ovarian cancer, somatic, 167000 Pilomatricoma, somatic, 132600
CTNS	120.3	100	99	Cystinosis, atypical nephropathic, 219800 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750
CTPS1	140.3	100	99	Immunodeficiency 24, 615897
CTSA	134.4	100	99	Galactosialidosis, 256540
CTSC	127.4	100	100	Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000 Periodontitis 1, juvenile, 170650
CTSD	164	97	95	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	113	84	80	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362
CTSK	105.1	100	99	Pycnodysostosis, 265800
CUBN	127.9	99	98	Megaloblastic anemia-1, Finnish type, 261100
CUL3	110.1	98	94	Pseudohypoadosteronism, type IIE, 614496
CUL4B	73.3	98	88	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
CUL7	149.9	99	97	3-M syndrome 1, 273750
CXCR4	202.8	100	99	Myelokathexis, isolated WHIM syndrome, 193670
CYB5A	134	100	100	Methemoglobinemia, type IV, 250790
CYB5R3	147.6	98	98	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYBA	97.4	78	71	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690
CYBB	111.5	99	99	Chronic granulomatous disease, X-linked, 306400 Immunodeficiency 34, mycobacteriosis, X-linked, 300645
CYC1	184.5	88	86	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYCS	72.1	99	94	Thrombocytopenia 4, 612004

CYLD	119.8	98	93	Brooke-Spiegler syndrome, 605041 Cylindromatosis, familial, 132700 Trichoepithelioma, multiple familial, 1, 601606
CYP11A1	124	99	97	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	176.1	100	100	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 Aldosteronism, glucocorticoid-remediable, 103900
CYP11B2	173.9	100	100	Aldosterone to renin ratio raised Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 {Low renin hypertension, susceptibility to}
CYP17A1	135	100	99	17,20-lyase deficiency, isolated, 202110 17-alpha-hydroxylase/17,20-lyase deficiency, 202110
CYP19A1	168.4	100	100	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300
CYP1B1	134.9	100	100	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 Peters anomaly, 604229
CYP21A2	15	15	13	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910
CYP24A1	165.3	100	100	Hypercalcemia, infantile, 143880
CYP26B1	178.4	100	99	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
CYP26C1	87.4	99	95	Focal facial dermal dysplasia 4, 614974
CYP27A1	175.2	98	96	Cerebrotendinous xanthomatosis, 213700
CYP27B1	137.4	100	99	Vitamin D-dependent rickets, type I, 264700
CYP2A6	168.2	100	99	Coumarin resistance, 122700 {Lung cancer, resistance to}, 211980 {Nicotine addiction, protection from}, 188890
CYP2C8	109	98	93	Rhabdomyolysis, cerivastatin-induced
CYP2R1	138.6	96	86	Rickets due to defect in vitamin D 25-hydroxylation, 600081
CYP2U1	119.2	93	91	Spastic paraplegia 56, autosomal recessive, 615030
CYP4F22	127.9	100	99	Ichthyosis, congenital, autosomal recessive 5, 604777
CYP4V2	147.7	99	98	Bietti crystalline corneoretinal dystrophy, 210370
CYP7B1	92.8	94	87	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, autosomal recessive, 270800
D2HGDH	132.7	97	95	D-2-hydroxyglutaric aciduria, 600721

DAG1	221.1	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818
DARS	98.5	98	93	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2	122.1	100	99	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBH	141	100	99	Dopamine beta-hydroxylase deficiency, 223360 [Dopamine-beta-hydroxylase activity levels, plasma]
DBT	102.1	97	93	Maple syrup urine disease, type II, 248600
DCAF17	91.9	95	89	Woodhouse-Sakati syndrome, 241080
DCC	139.3	100	99	Colorectal cancer, somatic, 114500 Esophageal carcinoma, somatic 133239 Mirror movements 1, 157600
DCDC2	150.6	99	99	Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212
DCHS1	160.4	99	99	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390
DCLRE1C	128.9	98	94	Omenn syndrome, 603554 Severe combined immunodeficiency, Athabaskan type, 602450
DCN	140.1	95	94	Corneal dystrophy, congenital stromal, 610048
DCPS	144.2	100	99	Al-Raqad syndrome, 616459
DCTN1	131.9	99	98	Neuropathy, distal hereditary motor, type VIIB, 607641 Perry syndrome, 168605 {Amyotrophic lateral sclerosis, susceptibility to}, 105400
DCX	116	100	99	Lissencephaly, X-linked, 300067 Subcortical laminar heteropia, X-linked, 300067
DDB2	162.8	100	99	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DDC	101.1	99	95	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	141.7	97	94	Spastic paraplegia 28, autosomal recessive, 609340
DDHD2	149.7	99	98	Spastic paraplegia 54, autosomal recessive, 615033
DDR2	155.2	100	99	Spondylometaphyseal dysplasia, short limb-hand type, 271665
DDX11	107.4	80	76	Warsaw breakage syndrome, 613398
DDX3X	89.7	98	94	Mental retardation, X-linked 102, 300958
DDX58	122.8	98	95	Singleton-Merten syndrome 2, 616298
DDX59	149.8	99	97	Orofaciodigital syndrome V, 174300
DEAF1	124.7	88	84	Mental retardation, autosomal dominant 24, 615828

DEPDC5	145.8	99	99	Epilepsy, familial focal, with variable foci, 604364
DES	121	99	98	Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419 Scapulooperoneal syndrome, neurogenic, Kaeser type, 181400 ?Muscular dystrophy, limb-girdle, type 2R, 615325
DFNA5	113	99	99	Deafness, autosomal dominant 5, 600994
DFNB59	123.6	100	99	Deafness, autosomal recessive 59, 610220
DGKE	142.3	99	95	Nephrotic syndrome, type 7, 615008 {Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008
DGUOK	125.1	100	100	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHCR24	177.4	100	100	Desmosterolosis, 602398
DHCR7	158.6	100	100	Smith-Lemli-Opitz syndrome, 270400
DHDDS	93.5	97	94	Retinitis pigmentosa 59, 613861
DHFR	48.5	91	71	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHH	117.7	100	100	46XY partial gonadal dysgenesis, with minifascicular neuropathy, 607080 46XY sex reversal 7, 233420
DHODH	92.3	100	99	Miller syndrome, 263750
DHTKD1	141.1	99	98	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DIABLO	206.4	100	99	Deafness, autosomal dominant 64, 614152
DIAPH1	120.5	99	97	Deafness, autosomal dominant 1, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632
DIAPH2	62.1	93	79	Premature ovarian failure, 300511
DIAPH3	74.8	97	90	Auditory neuropathy, autosomal dominant, 1, 609129
DICER1	145.4	99	98	Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800 Pleuropulmonary blastoma, 601200 Rhabdomyosarcoma, embryonal, 2, 180295
DIP2B	160.4	99	98	Mental retardation, FRA12A type, 136630
DIS3L2	157.1	99	99	Perlman syndrome, 267000
DKC1	112.6	99	98	Dyskeratosis congenita, X-linked, 305000
DLAT	91.6	99	96	Pyruvate dehydrogenase E2 deficiency, 245348
DLC1	179.3	99	99	Colorectal cancer, somatic, 114500
DLD	123.5	99	98	Dihydrolipoamide dehydrogenase deficiency, 246900
DLG3	90.7	99	94	Mental retardation, X-linked 90, 300850

DLL3	64.3	88	80	Spondylocostal dysostosis 1, autosomal recessive, 277300
DLL4	182	99	98	Adams-Oliver syndrome 6, 616589
DLX3	110.1	100	99	Amelogenesis imperfecta, type IV, 104510 Trichodontoosseous syndrome, 190320
DMD	113.7	99	97	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200
DMGDH	157.4	98	97	Dimethylglycine dehydrogenase deficiency, 605850
DMP1	159.9	99	99	Hypophosphatemic rickets, AR, 241520
DMPK	117.8	99	97	Myotonic dystrophy 1, 160900
DNA2	132	99	97	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156 ?Seckel syndrome 8, 615807
DNAAF1	115.8	100	99	Ciliary dyskinesia, primary, 13, 613193
DNAAF2	105.2	99	97	Ciliary dyskinesia, primary, 10, 612518
DNAAF3	91.8	97	90	Ciliary dyskinesia, primary, 2, 606763
DNAH11	134	99	98	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH5	123.8	99	98	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAI1	127.9	98	97	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	156.6	98	95	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJB2	102.1	100	100	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881
DNAJB6	58.4	90	77	Muscular dystrophy, limb-girdle, type 1E, 603511
DNAJC19	97.8	98	89	3-methylglutaconic aciduria, type V, 610198
DNAJC21	123.4	99	97	Bone marrow failure syndrome 3, 617052
DNAJC5	200.2	100	99	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350
DNAJC6	160.9	99	99	Parkinson disease 19, juvenile-onset, 615528
DNAL1	98.8	95	84	Ciliary dyskinesia, primary, 16, 614017
DNASE1L3	141.5	100	100	Systemic lupus erythematosus 16, 614420
DNM1	161.4	91	90	Epileptic encephalopathy, early infantile, 31, 616346
DNM1L	123.4	99	96	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission, 614388
DNM2	127.9	97	94	Charcot-Marie-Tooth disease, axonal, type 2M, 606482 Charcot-Marie-Tooth disease, dominant intermediate B, 606482 Lethal congenital contracture syndrome 5, 615368 Myopathy, centronuclear, 160150

DNMT1	113.5	99	98	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 Neuropathy, hereditary sensory, type IE, 614116
DNMT3A	115.7	98	95	Tatton-Brown-Rahman syndrome, 615879
DNMT3B	125	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK2	143.9	100	99	Immunodeficiency 40, 616433
DOCK6	121.2	98	96	Adams-Oliver syndrome 2, 614219
DOCK7	114.3	97	95	Epileptic encephalopathy, early infantile, 23, 615859
DOCK8	129.2	100	99	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
DOK7	99	94	92	Myasthenic syndrome, congenital, 10, 254300 ?Fetal akinesia deformation sequence, 208150
DOLK	203	100	99	Congenital disorder of glycosylation, type Im, 610768
DPAGT1	113	100	100	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPH1	157.3	100	99	Developmental delay with short stature, dysmorphic features and sparse hair, 616901
DPM1	131.2	91	86	Congenital disorder of glycosylation, type Ie, 608799
DPM2	102.2	100	99	Congenital disorder of glycosylation, type Iu, 615042
DPM3	184.3	100	100	Congenital disorder of glycosylation, type Io, 612937
DPP6	145.7	96	94	Mental retardation, autosomal dominant 33, 616311 {Ventricular fibrillation, paroxysmal familial, 2}, 612956
DPY19L2	94.5	72	66	Spermatogenic failure 9, 613958
DPYD	156.6	95	93	5-fluorouracil toxicity, 274270 Dihydropyrimidine dehydrogenase deficiency, 274270
DPYS	133.5	100	99	Dihydropyrimidinuria, 222748
DRAM2	131.7	100	99	Cone-rod dystrophy 21, 616502
DRC1	97.2	99	98	Ciliary dyskinesia, primary, 21, 615294
DRD4	79.2	80	70	Autonomic nervous system dysfunction [Novelty seeking personality], 601696 {Attention deficit-hyperactivity disorder}, 143465
DRD5	117.3	100	99	Dystonia, primary cervical {Attention deficit-hyperactivity disorder, susceptibility to}, 143465 {Blepharospasm, primary benign}, 606798
DSC2	128.3	99	96	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 Arrhythmogenic right ventricular dysplasia 11, 610476

DSG1	175	98	96	Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508 Keratosis palmoplantaris striata I, AD, 148700
DSG2	140.7	99	98	Arrhythmogenic right ventricular dysplasia 10, 610193 Cardiomyopathy, dilated, 1BB, 612877
DSG4	198.3	98	95	Hypotrichosis 6, 607903
DSP	154.1	100	99	Arrhythmogenic right ventricular dysplasia 8, 607450 Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821 Epidermolysis bullosa, lethal acantholytic, 609638 Keratosis palmoplantaris striata II, 612908 Skin fragility-woolly hair syndrome, 607655
DSPP	155.9	99	99	Deafness, autosomal dominant 39, with dentinogenesis, 605594 Dentin dysplasia, type II, 125420 Dentinogenesis imperfecta, Shields type II, 125490 Dentinogenesis imperfecta, Shields type III, 125500
DST	153.7	99	98	Epidermolysis bullosa simplex, autosomal recessive 2, 615425 ?Neuropathy, hereditary sensory and autonomic, type VI, 614653
DTNA	156.1	100	100	Left ventricular noncompaction 1, with or without congenital heart defects, 604169
DTNBP1	113.9	99	95	Hermansky-Pudlak syndrome 7, 614076
DUOX2	136.7	96	94	Thyroid dysmorphogenesis 6, 607200
DUOXA2	122.3	100	99	Thyroid dysmorphogenesis 5, 274900
DUSP6	176.2	100	99	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
DVL1	114.1	97	94	Robinow syndrome, autosomal dominant 2, 616331
DVL3	149.7	100	100	Robinow syndrome, autosomal dominant 3, 616894
DYM	101.2	97	94	Dyggve-Melchior-Clausen disease, 223800 Smith-McCort dysplasia, 607326
DYNC1H1	180	100	99	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600
DYNC2H1	90.4	96	87	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYNC2LI1	95.1	99	95	Short-rib thoracic dysplasia 15 with polydactyly, 617088
DYRK1A	158.4	100	100	Mental retardation, autosomal dominant 7, 614104
DYRK1B	105.3	93	84	Abdominal obesity-metabolic syndrome 3, 615812



DYSF	133.2	100	99	Miyoshi muscular dystrophy 1, 254130 Muscular dystrophy, limb-girdle, type 2B, 253601 Myopathy, distal, with anterior tibial onset, 606768
DYX1C1	79.6	96	84	Ciliary dyskinesia, primary, 25, 615482 {Dyslexia, susceptibility to, 1}, 127700
EARS2	103.5	99	98	Combined oxidative phosphorylation deficiency 12, 614924
EBP	83.8	100	97	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
ECE1	144.8	97	97	Hirschsprung disease, cardiac defects, and autonomic dysfunction, 613870 {Hypertension, essential, susceptibility to}, 145500
ECEL1	100.9	88	83	Arthrogryposis, distal, type 5D, 615065
ECHS1	112.7	99	97	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ECM1	171	100	99	Urbach-Wiethe disease, 247100
EDA	92.4	85	76	Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100 Tooth agenesis, selective, X-linked 1, 313500
EDAR	138.8	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
EDARADD	99.3	99	93	Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940 Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941
EDN1	145.4	100	100	Auriculocondylar syndrome 3, 615706 Question mark ears, isolated, 612798 {High density lipoprotein cholesterol level QTL 7}
EDN3	134.7	100	99	Central hypoventilation syndrome, congenital, 209880 Waardenburg syndrome, type 4B, 613265 {Hirschsprung disease, susceptibility to, 4}, 613712
EDNRA	218.7	100	99	Mandibulofacial dysostosis with alopecia, 616367 {Migraine, resistance to}, 157300
EDNRB	131	95	90	ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580 {Hirschsprung disease, susceptibility to, 2}, 600155
EEF1A2	178	98	93	Epileptic encephalopathy, early infantile, 33, 616409 Mental retardation, autosomal dominant 38, 616393
EFEMP1	167.9	100	99	Doyne honeycomb degeneration of retina, 126600

EFEMP2	121.1	100	99	Cutis laxa, autosomal recessive, type IB, 614437
EFNB1	119.4	100	99	Craniofrontonasal dysplasia, 304110
EFTUD2	124.4	100	99	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EGF	135.2	100	99	Hypomagnesemia 4, renal, 611718
EGFR	161.7	100	99	Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980 Non-small cell lung cancer, response to tyrosine kinase inhibitor in, 211980 ?Inflammatory skin and bowel disease, neonatal, 2, 616069 {Non-small cell lung cancer, susceptibility to}, 211980
EGLN1	39.2	79	66	Erythrocytosis, familial, 3, 609820 [Hemoglobin, high altitude adaptation], 609070
EGR2	124.6	100	100	Charcot-Marie-Tooth disease, type 1D, 607678 Dejerine-Sottas disease, 145900 Neuropathy, congenital hypomyelinating, 1, 605253
EHMT1	147.1	98	97	Kleefstra syndrome, 610253
EIF2AK3	147	95	91	Wolcott-Rallison syndrome, 226980
EIF2AK4	146.2	99	98	Pulmonary venoocclusive disease 2, 234810
EIF2B1	149.9	100	100	Leukoencephalopathy with vanishing white matter, 603896
EIF2B2	132	100	99	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B3	164.1	100	100	Leukoencephalopathy with vanishing white matter, 603896
EIF2B4	146.1	100	99	Leukoencephaly with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B5	128	99	98	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF4A3	106.8	100	99	Robin sequence with cleft mandible and limb abnormalities, 268305
ELAC2	124	100	99	Combined oxidative phosphorylation deficiency 17, 615440 {Prostate cancer, hereditary, 2, susceptibility to}, 614731
ELANE	80.9	99	95	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
ELN	91.2	99	97	Cutis laxa, AD, 123700 Supravalvar aortic stenosis, 185500
ELOVL4	91.9	99	98	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Stargardt disease 3, 600110 ?Spinocerebellar ataxia 34, 133190

ELOVL5	121.9	100	99	Spinocerebellar ataxia 38, 615957
EMC1	124.4	100	99	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EMD	101.1	99	97	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
EMG1	141.2	100	100	Bowen-Conradi syndrome, 211180
EMP2	101.3	99	96	Nephrotic syndrome, type 10, 615861
EMR2	160.6	96	96	Vibratory urticaria, 125630
EMX2	118.3	100	100	Schizencephaly, 269160
ENAM	148.8	100	99	Amelogenesis imperfecta, type IB, 104500 Amelogenesis imperfecta, type IC, 204650
ENG	128.9	97	93	Telangiectasia, hereditary hemorrhagic, type 1, 187300
ENPP1	134.9	92	83	Arterial calcification, generalized, of infancy, 1, 208000 Cole disease, 615522 Hypophosphatemic rickets, autosomal recessive, 2, 613312 {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853 {Obesity, susceptibility to}, 601665
ENTPD1	165.2	100	99	Spastic paraplegia 64, autosomal recessive, 615683
EOGT	150.4	100	98	Adams-Oliver syndrome 4, 615297
EP300	199.9	99	97	Colorectal cancer, somatic, 114500 Rubinstein-Taybi syndrome 2, 613684
EPAS1	137	99	97	Erythrocytosis, familial, 4, 611783
EPB41	135.5	98	95	Elliptocytosis-1, 611804
EPB42	161.2	100	99	Spherocytosis, type 5, 612690
EPCAM	64.4	93	79	Colorectal cancer, hereditary nonpolyposis, type 8, 613244 Diarrhea 5, with tufting enteropathy, congenital, 613217
EPG5	126.1	99	97	Vici syndrome, 242840
EPHA2	175.4	99	97	Cataract 6, multiple types, 116600
EPHX1	123	98	96	Diphenylhydantoin toxicity Hypercholanemia, familial, 607748 ?Fetal hydantoin syndrome {Preeclampsia, susceptibility to}, 189800
EPM2A	108.4	85	83	Epilepsy, progressive myoclonic 2A (Lafora), 254780
ERBB2	135.1	98	97	Adenocarcinoma of lung, somatic, 211980 Gastric cancer, somatic, 613659 Glioblastoma, somatic, 137800

				Ovarian cancer, somatic,
ERBB3	139.5	100	99	Lethal congenital contractural syndrome 2, 607598
ERBB4	144.4	99	99	Amyotrophic lateral sclerosis 19, 615515
ERCC1	76.6	100	97	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	123.8	100	99	Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy 1, photosensitive, 601675 Xeroderma pigmentosum, group D, 278730
ERCC3	113.3	99	98	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651
ERCC4	139.2	100	99	Fanconi anemia, complementation group Q, 615272 Xeroderma pigmentosum, group F, 278760 Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 ?XFE progeroid syndrome, 610965
ERCC5	139.8	100	99	Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	170.4	100	99	Cerebrooculofacioskeletal syndrome 1, 214150 Cockayne syndrome, type B, 133540 De Sanctis-Cacchione syndrome, 278800 Premature ovarian failure 11,616946 UV-sensitive syndrome 1, 600630 {Lung cancer, susceptibility to}, 211980 {Macular degeneration, age-related, susceptibility to 5}, 613761
ERCC6L2	124.2	99	97	Bone marrow failure syndrome 2, 615715
ERCC8	89.5	92	78	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621
ERF	107.7	99	97	Chitayat syndrome,617180 Craniosynostosis 4, 600775
ERLIN1	147	100	100	Spastic Paraplegia 62, 615681
ERLIN2	156.2	100	99	Spastic paraplegia 18, autosomal recessive, 611225
ESCO2	105.1	97	90	Roberts syndrome, 268300 SC phocomelia syndrome, 269000

ESPN	47.5	74	59	Deafness, autosomal recessive 36, 609006 Deafness, neurosensory, without vestibular involvement, autosomal dominant
ESR1	127.7	99	99	Estrogen resistance, 615363 {Atherosclerosis, susceptibility to} {Breast cancer}, 114480 {HDL response to hormone replacement, augmented} {Migraine, susceptibility to}, 157300 {Myocardial infarction, susceptibility to}, 608446
ESRRB	119.8	100	99	Deafness, autosomal recessive 35, 608565
ETFA	143.1	100	99	Glutaric acidemia IIA, 231680
ETFB	126.8	100	100	Glutaric acidemia IIB, 231680
ETFDH	105.5	100	99	Glutaric acidemia IIC, 231680
ETHE1	85.8	99	95	Ethylmalonic encephalopathy, 602473
ETV6	140.3	100	99	Leukemia, acute myeloid, somatic, 601626 Thrombocytopenia 5, 616216
EVC	110.7	93	89	Ellis-van Creveld syndrome, 225500 Weyers acrodental dysostosis, 193530
EVC2	119	96	94	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530
EWSR1	79.5	94	83	Ewing sarcoma, 612219 Neuroepithelioma, 612219
EXOSC3	88.4	97	89	Pontocerebellar hypoplasia, type 1B, 614678
EXOSC8	79.9	91	76	Pontocerebellar hypoplasia, type 1C, 616081
EXPH5	183.3	100	99	Epidermolysis bullosa, nonspecific, autosomal recessive, 615028
EXT1	105.5	99	98	Chondrosarcoma, 215300 Exostoses, multiple, type 1, 133700
EXT2	163.6	99	99	Exostoses, multiple, type 2, 133701 ?Seizures, scoliosis, and macrocephaly syndrome, 616682
EYA1	144.4	100	99	Anterior segment anomalies with or without cataract, 113650 Branchiootic syndrome 1, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650 ?Otofaciocervical syndrome, 166780
EYA4	160.5	100	99	Cardiomyopathy, dilated, 1J, 605362 Deafness, autosomal dominant 10, 601316

EYS	135.7	98	94	Retinitis pigmentosa 25, 602772
EZH2	139.1	99	97	Weaver syndrome, 277590
F10	185.4	99	98	Factor X deficiency, 227600
F11	155	100	100	Factor XI deficiency, autosomal dominant, 612416 Factor XI deficiency, autosomal recessive, 612416
F12	111.6	100	99	Angioedema, hereditary, type III, 610618 Factor XII deficiency, 234000
F13A1	147.4	100	99	Factor XIII A deficiency, 613225 {Myocardial infarction, protection against}, 608446 {Venous thrombosis, protection against}, 188050
F13B	113.4	96	87	Factor XIII B deficiency, 613235
F2	124.4	99	98	Dysprothrombinemia, 613679 Hypoprothrombinemia, 613679 Thrombophilia due to thrombin defect, 188050 {Pregnancy loss, recurrent, susceptibility to, 2}, 614390 {Stroke, ischemic, susceptibility to}, 601367
F5	173.6	99	97	Factor V deficiency, 227400 Thrombophilia due to activated protein C resistance, 188055 {Budd-Chiari syndrome}, 600880 {Pregnancy loss, recurrent, susceptibility to, 1}, 614389 {Stroke, ischemic, susceptibility to}, 601367 {Thrombophilia, susceptibility to, due to factor V Leiden}, 188055
F7	164.4	100	98	Factor VII deficiency, 227500 {Myocardial infarction, decreased susceptibility to}, 608446
F8	118.8	99	98	Hemophilia A, 306700
F9	145.5	99	95	Hemophilia B, 306900 Thrombophilia, X-linked, due to factor IX defect, 300807 {Deep venous thrombosis, protection against}, 300807 {Warfarin sensitivity}, 122700
FA2H	94.2	87	79	Spastic paraplegia 35, autosomal recessive, 612319
FADD	142.9	100	99	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations, 613759
FAH	151.6	100	99	Tyrosinemia, type I, 276700
FAM105B	149.7	90	86	Autoinflammation, panniculitis, and dermatosis syndrome, 617099

FAM111A	292	100	99	Gracile bone dysplasia, 602361 Kenny-Caffey syndrome, type 2, 127000
FAM111B	152.8	100	99	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704
FAM126A	124.9	97	95	Leukodystrophy, hypomyelinating, 5, 610532
FAM134B	126.1	95	89	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
FAM161A	115.3	98	95	Retinitis pigmentosa 28, 606068
FAM20A	104.8	98	91	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM20C	101.4	100	98	Raine syndrome, 259775
FAM58A	74.2	83	79	STAR syndrome, 300707
FAM83H	76.9	94	87	Amelogenesis imperfecta, type III, 130900
FAN1	144.1	100	99	Interstitial nephritis, karyomegalic, 614817
FANCA	123.5	99	98	Fanconi anemia, complementation group A, 227650
FANCB	68.8	96	88	Fanconi anemia, complementation group B, 300514
FANCC	116.5	99	96	Fanconi anemia, complementation group C, 227645
FANCD2	128.1	98	95	Fanconi anemia, complementation group D2, 227646
FANCE	108.1	86	84	Fanconi anemia, complementation group E, 600901
FANCF	166.9	100	100	Fanconi anemia, complementation group F, 603467
FANCG	147.9	100	100	Fanconi anemia, complementation group G, 614082
FANCI	152.2	99	97	Fanconi anemia, complementation group I, 609053
FANCL	87.9	99	94	Fanconi anemia, complementation group L, 614083
FAR1	80.3	96	92	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154
FARS2	207.8	100	100	Combined oxidative phosphorylation deficiency 14, 614946 ?Spastic paraplegia 77, autosomal recessive, 617046
FAS	271.9	100	99	Autoimmune lymphoproliferative syndrome, type IA, 601859 Squamous cell carcinoma, burn scar-related, somatic {Autoimmune lymphoproliferative syndrome}, 601859
FASLG	86.1	100	98	Autoimmune lymphoproliferative syndrome, type IB, 601859 {Lung cancer, susceptibility to}, 211980
FAT4	224.4	100	99	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 Van Maldergem syndrome 2, 615546
FBLN5	119.8	91	91	Cutis laxa, autosomal dominant 2, 614434 Cutis laxa, autosomal recessive, type IA, 219100 Macular degeneration, age-related, 3, 608895 Neuropathy, hereditary, with or without age-related macular degeneration, 608895

FBN1	159.8	99	99	Acromicric dysplasia, 102370 Aortic aneurysm, ascending, and dissection Ectopia lentis, familial, 129600 Geleophysic dysplasia 2, 614185 Marfan lipodystrophy syndrome,616914 Marfan syndrome, 154700 MASS syndrome, 604308 Stiff skin syndrome, 184900 Weill-Marchesani syndrome 2, dominant, 608328
FBN2	161.8	100	99	Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118
FBP1	126.9	100	98	Fructose-1,6-bisphosphatase deficiency, 229700
FBXL4	189.7	100	100	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FBXO38	179.8	99	98	Neuronopathy, distal hereditary motor, type IID, 615575
FBXO7	189.7	98	93	Parkinson disease 15, autosomal recessive, 260300
FCGR3A	244.9	100	99	Immunodeficiency 20, 615707
FCGR3B	176.7	99	98	Neutropenia, alloimmune neonatal
FCN3	127.8	100	99	Immunodeficiency due to ficolin 3 deficiency, 613860
FDPS	73	99	94	Porokeratosis 9, multiple types, 616631
FECH	122	99	99	Protoporphyrin, erythropoietic, autosomal recessive, 177000
FERMT1	105	98	96	Kindler syndrome,173650
FERMT3	122.6	100	98	Leukocyte adhesion deficiency,type III,612840
FEZF1	159	99	99	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FGA	157.2	99	96	Afibrinogenemia, congenital, 202400 Amyloidosis, familial visceral, 105200 Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, congenital, 616004
FGB	190.7	99	97	Afibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Hypofibrinogenemia, congenital, 202400
FGD1	86.4	92	86	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400
FGD4	110.2	98	96	Charcot-Marie-Tooth disease, type 4H, 609311
FGF10	142.2	100	100	Aplasia of lacrimal and salivary glands, 180920



				LADD syndrome, 149730
FGF14	190.3	100	99	Spinocerebellar ataxia 27, 609307
FGF16	126.4	99	97	Metacarpal 4-5 fusion, 309630
FGF17	139	100	100	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
FGF23	106.4	99	97	Hypophosphatemic rickets, autosomal dominant, 193100 Osteomalacia, tumor-induced Tumoral calcinosis, hyperphosphatemic, familial, 211900
FGF3	74	91	75	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGF5	109.2	99	97	Trichomegaly, 190330
FGF8	111.5	90	79	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGFR1	148.3	99	98	Encephalocraniocutaneous lipomatosis, 613001 Hartsfield syndrome, 615465 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Jackson-Weiss syndrome, 123150 Osteoglophonic dysplasia, 166250 Pfeiffer syndrome, 101600 Trigonocephaly 1, 190440
FGFR2	142.4	97	96	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Apert syndrome, 101200 Beare-Stevenson cutis gyrata syndrome, 123790 Bent bone dysplasia syndrome, 614592 Craniofacial-skeletal-dermatologic dysplasia, 101600 Craniosynostosis, nonspecific Crouzon syndrome, 123500 Gastric cancer, somatic, 613659 Jackson-Weiss syndrome, 123150 LADD syndrome, 149730 Pfeiffer syndrome, 101600 Saethre-Chotzen syndrome, 101400 Scaphocephaly and Axenfeld-Rieger anomaly Scaphocephaly, maxillary retrusion, and mental retardation, 609579

FGFR3	110.5	99	97	Achondroplasia, 100800 Bladder cancer, somatic, 109800 CATSHL syndrome, 610474 Cervical cancer, somatic, 603956 Colorectal cancer, somatic, 114500 Crouzon syndrome with acanthosis nigricans, 612247 Hypochondroplasia, 146000 LADD syndrome, 149730 Muenke syndrome, 602849 Nevus, epidermal, somatic, 162900 SADDAN, 616482 Spermatocytic seminoma, somatic, 273300 Thanatophoric dysplasia, type I, 187600 Thanatophoric dysplasia, type II, 187601
FGG	136.9	99	96	Afibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, 616004 Hypofibrinogenemia, congenital, 202400
FH	146.1	91	87	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FHL1	87.8	98	93	Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 Hemophagocytic lymphohistiocytosis, familial, 1 Myopathy, X-linked, with postural muscle atrophy, 300696 Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717 Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718 Scapuloperoneal myopathy, X-linked dominant, 300695
FIG4	154.9	99	98	Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228 Yunis-Varon syndrome, 216340 ?Polymicrogyria, bilateral temporooccipital, 612691
FIGLA	81.8	94	89	Premature ovarian failure 6, 612310
FKBP10	153.3	97	93	Bruck syndrome 1, 259450 Osteogenesis imperfecta, type XI, 610968
FKBP14	74	100	99	Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss, 614557

FKRP	94.6	100	99	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155
FKTN	119.9	99	94	Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588
FLAD1	191.8	100	98	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100
FLCN	160.8	100	99	Birt-Hogg-Dube syndrome, 135150 Colorectal cancer, somatic, 114500 Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700
FLG	234.3	100	99	Ichthyosis vulgaris, 146700 {Dermatitis, atopic, susceptibility to, 2}, 605803
FLNA	139.2	100	99	Cardiac valvular dysplasia, X-linked, 314400 Congenital short bowel syndrome, 300048 FG syndrome 2, 300321 Frontometaphyseal dysplasia, 305620 Heterotopia, periventricular, 300049 Heterotopia, periventricular, ED variant, 300537 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Terminal osseous dysplasia, 300244
FLNB	152.7	100	99	Atelosteogenesis, type I, 108720 Atelosteogenesis, type III, 108721 Boomerang dysplasia, 112310 Larsen syndrome, 150250 Spondylocarpotarsal synostosis syndrome, 272460
FLNC	165.3	100	99	Cardiomyopathy, familial hypertrophic, 26 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Myopathy, myofibrillar, 5, 609524
FLRT3	225.8	100	100	Hypogonadotropic hypogonadism 21 with anosmia, 615271

FLT3	134.7	99	98	Leukemia, acute lymphoblastic, somatic, 613065 Leukemia, acute myeloid, reduced survival in, somatic, 601626 Leukemia, acute myeloid, somatic, 601626
FLT4	156.1	98	97	Hemangioma, capillary infantile, somatic, 602089 Lymphedema, hereditary, IA, 153100
FLVCR1	139.4	99	95	Ataxia, posterior column, with retinitis pigmentosa, 609033
FLVCR2	159.9	100	100	Proliferative vasculopathy and hydraencephaly-hydrocephaly syndrome, 225790
FMN2	86.6	83	77	Mental retardation, autosomal recessive 47, 616193
FMO3	150.6	99	99	Trimethylaminuria, 602079
FMR1	81.3	96	86	Fragile X syndrome, 300624 Fragile X tremor/ataxia syndrome, 300623 Premature ovarian failure 1, 311360
FN1	145.5	100	99	Glomerulopathy with fibronectin deposits 2, 601894 Plasma fibronectin deficiency, 614101
FOLR1	150.5	100	100	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXC1	32.8	86	68	Axenveld-Rieger syndrome, type 3, 602482 Iridogoniodysgenesis, type 1, 601631 Iris hypoplasia and glaucoma, 601631 Rieger or Axenveld anomalies, 602482
FOXC2	44.3	95	79	Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400 Lymphedema-distichiasis syndrome, 153400
FOXE1	29.2	72	56	Bamforth-Lazarus syndrome, 241850 {Thyroid cancer, nonmedullary, 4}, 616534
FOXE3	20.8	69	48	Anterior segment mesenchymal dysgenesis, 107250 Aphakia, congenital primary, 610256 Cataract 34,multiple types, 612968
FOXF1	93.9	98	93	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380
FOXG1	158	84	81	Rett syndrome, congenital variant, 613454
FOXI1	152.6	100	100	Enlarged vestibular aqueduct, 600791
FOXL2	39.4	92	71	Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 Premature ovarian failure 3, 608996
FOXN1	112.9	100	99	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXO1	139.4	92	85	Rhabdomyosarcoma,alveolar,268220

FOXP1	132.2	100	99	Mental retardation with language impairment and with or without autistic features, 613670
FOXP2	163.2	99	97	Speech-language disorder-1, 602081
FOXP3	125.7	98	91	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790 {Diabetes mellitus, type I, susceptibility to}, 222100
FOXRED1	136.9	100	99	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
FRAS1	147.7	100	99	Fraser syndrome, 219000
FREM1	138.4	99	99	Bifid nose with or without anorectal and renal anomalies, 608980 Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485
FREM2	182.5	100	99	Fraser syndrome, 219000
FRMD7	114.9	99	98	Nystagmus 1, congenital, X-linked, 310700 Nystagmus, infantile periodic alternating, X-linked, 310700
FRRS1L	103.2	68	63	Epileptic encephalopathy, early infantile, 37, 616981
FSCN2	145.2	100	100	Retinitis pigmentosa 30, 607921
FSHB	148.8	100	100	Hypogonadotropic hypogonadism 24 without anosmia, 229070
FSHR	140.9	100	98	Ovarian dysgenesis 1, 233300 Ovarian hyperstimulation syndrome, 608115 Ovarian response to FSH stimulation, 276400
FTCD	90	94	89	Glutamate formiminotransferase deficiency, 229100
FTL	148.1	98	93	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159
FTO	141.8	99	98	Growth retardation, developmental delay, facial dysmorphism, 612938 {Obesity, susceptibility to, BMIQ14}, 612460
FTSJ1	129.4	98	94	Mental retardation, X-linked 9, 309549
FUCA1	134.9	100	99	Fucosidosis, 230000
FUS	137.7	97	94	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 Tremor, hereditary essential, 4, 614782
FUT6	165.4	100	100	Fucosyltransferase 6 deficiency, 613852
FUZ	116.2	100	100	Neural tube defects, 182940
FXN	75.3	85	75	Friedreich ataxia with retained reflexes, 229300 Friedreich ataxia, 229300
FXD2	96.9	99	99	Hypomagnesemia 2, renal, 154020

FYCO1	123.9	100	100	Cataract 18, autosomal recessive, 610019
FZD4	224.3	100	99	Exudative vitreoretinopathy 1, 133780 Retinopathy of prematurity, 133780
FZD6	208.7	100	100	Nail disorder, nonsyndromic congenital, 10, (claw-shaped nails), 614157
G6PC	180.8	100	100	Glycogen storage disease Ia, 232200
G6PC3	124	100	100	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
G6PD	119.4	99	97	Favism, 134700 Hemolytic anemia due to G6PD deficiency, 300908 {Resistance to malaria due to G6PD deficiency}, 611162
GAA	128.7	100	99	Glycogen storage disease II, 232300
GABRA1	183.2	100	100	Epileptic encephalopathy, early infantile, 19, 615744 {Epilepsy, childhood absence, susceptibility to, 4}, 611136 {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136
GABRG2	147.7	92	92	Epilepsy, generalized, with febrile seizures plus, type 3, 611277 Febrile seizures, familial, 8, 611277 {Epilepsy, childhood absence, susceptibility to, 2}, 607681
GALC	100.5	98	94	Krabbe disease, 245200
GALE	155.2	100	100	Galactose epimerase deficiency, 230350
GALK1	122.7	99	97	Galactokinase deficiency with cataracts, 230200
GALNS	93.1	99	95	Mucopolysaccharidosis IVA, 253000
GALNT3	128	99	96	Tumoral calcinosis, hyperphosphatemic, familial, 211900
GALT	155.5	100	99	Galactosemia, 230400
GAMT	93.7	90	80	Cerebral creatine deficiency syndrome 2, 612736
GAN	190.2	100	99	Giant axonal neuropathy-1, 256850
GANAB	120.3	99	98	Polycystic kidney disease 3, 600666
GARS	125.1	99	98	Charcot-Marie-Tooth disease, type 2D, 601472 Neuropathy, distal hereditary motor, type VA, 600794
GAS8	150.8	99	99	Ciliary dyskinesia, primary, 33, 616726
GATA1	84.2	99	95	Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835 Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 Thrombocytopenia with beta-thalassemia, X-linked, 314050 Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367

GATA2	119.6	99	98	Emberger syndrome, 614038 Immunodeficiency 21, 614172 {Leukemia, acute myeloid, susceptibility to}, 601626 {Myelodysplastic syndrome, susceptibility to}, 614286
GATA3	187.1	100	100	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GATA4	87.5	68	60	Atrial septal defect 2, 607941 Atrioventricular septal defect 4, 614430 Tetralogy of Fallot, 187500 Ventricular septal defect 1, 614429 ?Testicular anomalies with or without congenital heart disease, 615542
GATA6	61.6	83	71	Atrial septal defect 9, 614475 Atrioventricular septal defect 5, 614474 Pancreatic agenesis and congenital heart defects, 600001 Persistent truncus arteriosus, 217095 Tetralogy of Fallot, 187500
GATAD2B	129.8	100	99	Mental retardation, autosomal dominant 18, 615074
GATM	150.7	100	100	Cerebral creatine deficiency syndrome 3, 612718
GBA	240.7	100	100	Gaucher disease, perinatal lethal, 608013 Gaucher disease, type I, 230800 Gaucher disease, type II, 230900 Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 {Lewy body dementia, susceptibility to}, 127750 {Parkinson disease, late-onset, susceptibility to}, 168600
GBA2	176.3	99	99	Spastic paraplegia 46, autosomal recessive, 614409
GBE1	145.4	99	97	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GCDH	133.7	94	91	Glutaricaciduria, type I, 231670
GCH1	74.4	97	86	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GCK	141.7	100	100	Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, permanent neonatal, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485 MODY, type II, 125851

GCLC	133.2	99	98	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 {Myocardial infarction, susceptibility to}, 608446
GCM2	162	100	100	Hypoparathyroidism, familial isolated, 146200
GCNT2	167.9	100	100	Adult i phenotype without cataract, 110800 Cataract 13 with adult i phenotype, 116700 [Blood group, li], 110800
GCSH	34.2	83	67	Glycine encephalopathy, 605899
GDAP1	163	99	96	Charcot-Marie-Tooth disease, axonal, type 2K, 607831 Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706 Charcot-Marie-Tooth disease, recessive intermediate, A, 608340 Charcot-Marie-Tooth disease, type 4A, 214400
GDF1	19.4	64	48	Double-outlet right ventricle, 217095 Right atrial isomerism, 208530 Tetralogy of Fallot, 187500 Transposition of great arteries, dextro-looped 3, 613854
GDF2	163.3	100	100	Telangiectasia, hereditary hemorrhagic, type 5, 615506
GDF3	135.1	100	100	Klippel-Feil syndrome 3, autosomal dominant, 613702 Microphthalmia with coloboma 6, 613703 Microphthalmia, isolated 7, 613704
GDF5	141.9	100	100	Brachydactyly, type A1, C, 615072 Brachydactyly, type A2, 112600 Brachydactyly, type C, 113100 Chondrodysplasia, Grebe type, 200700 Du Pan syndrome, 228900 Multiple synostoses syndrome 2, 610017 Symphalangism, proximal, 1B, 615298 ?Acromesomelic dysplasia, Hunter-Thompson type, 201250 {Osteoarthritis-5}, 612400
GDF6	75	98	89	Klippel-Feil syndrome 1, autosomal dominant, 118100 Leber congenital amaurosis 17, 615360 Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094
GDI1	158.8	100	100	Mental retardation, X-linked 41, 300849



GDNF	186.1	99	98	Central hypoventilation syndrome, 209880 {Hirschsprung disease, susceptibility to, 3}, 613711 {Pheochromocytoma, modifier of}, 171300
GFAP	99.1	99	98	Alexander disease, 203450
GFER	76.3	93	75	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076
GFI1	83.1	99	92	Neutropenia, nonimmune chronic idiopathic, of adults, 607847 Neutropenia, severe congenital 2, autosomal dominant, 613107
GFI1B	189.6	100	100	Bleeding disorder, platelet-type, 17, 187900
GFM1	100.3	99	95	Combined oxidative phosphorylation deficiency 1, 609060
GFPT1	144.4	99	97	Myasthenia, congenital, 12, with tubular aggregates, 610542
GGCX	115.4	100	99	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842 Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450
GH1	175.4	100	100	Growth hormone deficiency, isolated, type IA, 262400 Growth hormone deficiency, isolated, type IB, 612781 Growth hormone deficiency, isolated, type II, 173100 Kowarski syndrome, 262650
GHR	213.4	99	99	Growth hormone insensitivity, partial, 604271 Increased responsiveness to growth hormone Laron dwarfism, 262500 {Hypercholesterolemia, familial, modifier of}, 143890
GHRHR	122.8	100	99	Growth hormone deficiency, isolated, type IB, 612781
GHSR	206.3	99	98	Growth hormone deficiency, isolated partial, 615925
GIF	141.3	100	99	Intrinsic factor deficiency, 261000
GIPC3	103.5	91	85	Deafness, autosomal recessive 15, 601869
GJA1	246.5	100	100	Atrioventricular septal defect 3, 600309 Craniometaphyseal dysplasia, autosomal recessive, 218400 Erythrokeratoderma variabilis et progressiva, 133200 Hypoplastic left heart syndrome 1, 241550 Oculodentodigital dysplasia, 164200 Oculodentodigital dysplasia, autosomal recessive, 257850 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100
GJA3	165.2	100	99	Cataract 14, multiple types, 601885

GJA5	268.2	100	100	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic (GJA5/SCN5A), 108770
GJA8	153.9	100	100	Cataract 1, multiple types, 116200
GJB1	231.2	100	99	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GJB2	205.6	100	100	Bart-Pumphrey syndrome, 149200 Deafness, autosomal dominant 3A, 601544 Deafness, autosomal recessive 1A, 220290 Hystrix-like ichthyosis with deafness, 602540 Keratitis-ichthyosis-deafness syndrome, 148210 Keratoderma, palmoplantar, with deafness, 148350 Vohwinkel syndrome, 124500
GJB3	309.1	100	100	Deafness, autosomal dominant 2B, 612644 Deafness, autosomal dominant, with peripheral neuropathy Deafness, autosomal recessive Deafness, digenic, GJB2/GJB3, 220290 Erythrokeratoderma variabilis et progressiva, 133200
GJB4	370.3	100	100	Erythrokeratoderma variabilis with erythema gyratum repens, 133200
GJB6	185.4	100	100	Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290 Ectodermal dysplasia 2, Clouston type, 129500
GJC2	42.1	68	58	Leukodystrophy, hypomyelinating, 2, 608804 Lymphedema, hereditary, IC, 613480 Spastic paraplegia 44, autosomal recessive, 613206
GK	43.9	74	54	Glycerol kinase deficiency, 307030
GLA	82	99	97	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	91.6	99	97	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLDC	80.8	90	82	Glycine encephalopathy, 605899
GLE1	111	100	99	Arthrogryposis, lethal, with anterior horn cell disease, 611890 Lethal congenital contracture syndrome 1, 253310

GLI2	138.7	99	97	Culler-Jones syndrome, 615849 Holoprosencephaly-9, 610829
GLI3	154.4	100	99	Greig cephalopolysyndactyly syndrome, 175700 Pallister-Hall syndrome, 146510 Polydactyly, postaxial, types A1 and B, 174200 Polydactyly, preaxial, type IV, 174700 {Hypothalamic hamartomas, somatic}, 241800
GLIS2	109.2	100	98	Nephronophthisis 7, 611498
GLIS3	133.5	99	99	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GLMN	66.7	97	86	Glomuvenous malformations,138000
GLRA1	123.4	100	100	Hyperekplexia, hereditary 1, autosomal dominant or recessive, 149400
GLRB	98.5	96	88	Hyperekplexia 2, autosomal recessive, 614619
GLRX5	108.1	92	83	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GLUD1	76.7	94	84	Hyperinsulinism-hyperammonemia syndrome, 606762
GLUL	109	99	98	Glutamine deficiency, congenital, 610015
GLYCK	203	100	99	D-glyceric aciduria, 220120
GM2A	139.7	100	100	GM2-gangliosidosis, AB variant, 272750
GMNN	101.2	92	83	Meier-Gorlin syndrome 6, 616835
GMPPA	137	100	99	Alacrima, achalasia, and mental retardation syndrome, 615510
GMPPB	229.1	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352
GNA11	150	99	96	Hypocalcemia, autosomal dominant 2, 615361 Hypocalciuric hypercalcemia, type II, 145981
GNAI2	139.1	100	99	Pituitary ACTH-secreting adenoma Ventricular tachycardia, idiopathic, 192605
GNAI3	110.6	99	92	Auriculocondylar syndrome 1, 602483
GNAL	134.8	94	91	Dystonia 25, 615073
GNAO1	180.4	100	100	Epileptic encephalopathy, early infantile, 17, 615473
GNAQ	82.6	81	69	Capillary malformations, congenital, 1, somatic, mosaic, 163000 Sturge-Weber syndrome, somatic, mosaic, 185300

GNAS	141.2	98	95	Acromegaly, somatic, 102200 ACTH-independent macronodular adrenal hyperplasia, 219080 McCune-Albright syndrome, somatic, mosaic 174800 Osseous heteroplasia, progressive, 166350 Pseudohypoparathyroidism Ia, 103580 Pseudohypoparathyroidism Ib, 603233 Pseudohypoparathyroidism Ic, 612462 Pseudopseudohypoparathyroidism, 612463
GNAS-AS1	NC	NC	NC	Pseudohypoparathyroidism, type IB, 603233
GNAT1	154.2	100	100	Night blindness, congenital stationary, autosomal dominant 3, 610444 ?Night blindness, congenital stationary, type 1G, 616389
GNAT2	130.9	99	99	Achromatopsia-4, 613856
GNB1	189.5	100	100	Leukemia,acute lymphoblastic,somatic, 613065 Mental retardation, autosomal dominant 42, 616973
GNB3	179.2	100	100	Night blindness, congenital stationary, type 1H, 617024 {Hypertension, essential, susceptibility to}, 145500
GNB4	152.5	100	99	Charcot-Marie-Tooth disease, dominant intermediate F, 615185
GNE	153.9	100	99	Nonaka myopathy, 605820 Sialuria, 269921
GNMT	147.3	99	96	Glycine N-methyltransferase deficiency, 606664
GNPAT	133.7	99	96	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	167.6	98	97	Mucopolysaccharidosis II alpha/beta, 252500 Mucopolysaccharidosis III alpha/beta, 252600
GNPTG	153.1	96	90	Mucopolysaccharidosis III gamma, 252605
GNRHR	162	100	100	Hypogonadotropic hypogonadism 7 without anosmia, 146110
GNS	107.9	96	92	Mucopolysaccharidosis type IIID, 252940
GORAB	176.3	99	97	Geroderma osteodysplasticum, 231070
GOSR2	123.2	97	96	Epilepsy, progressive myoclonic 6, 614018
GOT1	124.4	100	99	Aspartate aminotransferase, serum level of, QTL1, 614419
GP1BA	153.1	97	94	Bernard-Soulier syndrome, type A1 (recessive), 231200 Bernard-Soulier syndrome, type A2 (dominant), 153670 von Willebrand disease, platelet-type, 177820 {Nonarteritic anterior ischemic optic neuropathy, susceptibility to}, 258660
GP1BB	34.5	74	64	Bernard-Soulier syndrome, type B, 231200

				Giant platelet disorder, isolated, 231200
GP6	136.8	100	100	Bleeding disorder, platelet-type, 11, 614201
GP9	123.1	96	89	Bernard-Soulier syndrome, type C, 231200
GPC3	85.8	98	92	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPC6	142	100	100	Omodysplasia 1, 258315
GPD1	93.6	99	99	Hypertriglyceridemia, transient infantile, 614480
GPD1L	138.4	100	98	Brugada syndrome 2, 611777
GPHN	167.3	98	96	Molybdenum cofactor deficiency C, 615501
GPI	142.7	100	99	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470
GPIHBP1	120.1	99	99	Hyperlipoproteinemia, type 1D, 615947
GPR101	160.4	100	100	Pituitary adenoma, growth hormone-secreting 2, 300943
GPR126	147.6	99	97	Lethal congenital contracture syndrome 9, 616503
GPR143	60.5	85	74	Nystagmus 6, congenital, X-linked, 300814 Ocular albinism, type I, Nettleship-Falls type, 300500
GPR179	133.5	100	99	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
GPR56	150.9	100	100	Polymicrogyria, bilateral frontoparietal, 606854 Polymicrogyria, bilateral perisylvian, 615752
GPR98	139.9	99	96	Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472 ?Febrile seizures, familial, 4, 604352
GPSM2	113	99	97	Chudley-McCullough syndrome, 604213
GPX4	161.9	86	85	Spondylometaphyseal dysplasia, Sedaghatian type, 250220
GRHL2	134.6	100	100	Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029
GRHL3	140.8	100	99	Van der Woude syndrome 2, 606713
GRHPR	112.8	85	78	Hyperoxaluria, primary, type II, 260000
GRIA3	95.2	99	94	Mental retardation, X-linked 94, 300699
GRID2	175.4	100	99	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRIK2	133.1	96	94	Mental retardation, autosomal recessive, 6, 611092
GRIN1	148.7	100	99	Mental retardation, autosomal dominant 8, 614254
GRIN2A	159.3	100	100	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570
GRIN2B	189.7	99	99	Epileptic encephalopathy, early infantile, 27, 616139

				Mental retardation, autosomal dominant 6, 613970
GRIP1	130.8	100	99	Fraser syndrome, 219000
GRK1	126.4	100	99	Oguchi disease-2, 613411
GRM1	185.7	100	99	Spinocerebellar ataxia, autosomal recessive 13, 614831
GRM6	151.9	93	86	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270
GRN	184.8	100	100	Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
GRXCR1	183.6	100	99	Deafness, autosomal recessive 25, 613285
GSC	85.3	86	74	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471
GSN	120.7	97	91	Amyloidosis, Finnish type, 105120
GSS	104.2	100	99	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900
GTF2E2	83.5	96	91	Trichothiodystrophy 6, nonphotosensitive, 616943
GTF2H5	113.7	100	99	Trichothiodystrophy 3, photosensitive, 616395
GTPBP3	137.7	100	99	Combined oxidative phosphorylation deficiency 23, 616198
GUCA1A	162.6	100	100	Cone dystrophy-3, 602093 Cone-rod dystrophy 14, 602093
GUCA1B	144.2	100	99	Retinitis pigmentosa 48, 613827
GUCY1A3	171	99	97	Moyamoya 6 with achalasia, 615750
GUCY2C	135.2	100	99	Diarrhea 6, 614616 Meconium ileus, 614665
GUCY2D	91.6	98	91	Cone-rod dystrophy 6, 601777 Leber congenital amaurosis 1, 204000
GUSB	116.4	92	89	Mucopolysaccharidosis VII, 253220
GYG1	157.9	100	99	Polyglucosan body myopathy 2, 616199 ?Glycogen storage disease XV, 613507
GYS1	110.9	100	98	Glycogen storage disease 0, muscle, 611556
GYS2	150	98	94	Glycogen storage disease 0, liver, 240600
H19	NC	NC	NC	Beckwith-Wiedemann syndrome, 130650 Silver-Russell syndrome, 180860 Wilms tumor 2, 194071
H6PD	169.7	99	99	Cortisone reductase deficiency 1, 604931

HACE1	125.2	99	95	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADH	111.7	98	95	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HADHA	84.1	96	90	Fatty liver, acute, of pregnancy, 609016 HELLP syndrome, maternal, of pregnancy, 609016 LCHAD deficiency, 609016 Trifunctional protein deficiency, 609015
HADHB	80.4	92	79	Trifunctional protein deficiency, 609015
HAMP	175.8	100	100	Hemochromatosis, type 2B, 613313
HARS	159.6	100	100	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504
HAX1	136.6	100	100	Neutropenia, severe congenital 3, autosomal recessive, 610738
HBA1	142.3	100	100	Erythremias, alpha- Heinz body anemias, alpha-, 140700 Hemoglobin H disease, nondeletional, 613978 Methemoglobinemias, alpha- Thalassemias, alpha-, 604131
HBA2	134	97	91	Erythrocytosis Heinz body anemia, 140700 Hemoglobin H disease, nondeletional, 613978 Hypochromic microcytic anemia Thalassemia, alpha-, 604131
HBB	176.7	100	100	Delta-beta thalassemia, 141749 Erythremias, beta- Heinz body anemias, beta-, 140700 Hereditary persistence of fetal hemoglobin, 141749 Methemoglobinemias, beta- Sickle cell anemia, 603903 Thalassemia-beta, dominant inclusion-body, 603902 Thalassemias, beta-, 613985 {Malaria, resistance to}, 611162
HBD	242.5	100	100	Thalassemia due to Hb Lepore Thalassemia, delta-
HBG1	175	98	95	Fetal hemoglobin quantitative trait locus 1, 141749

HBG2	329.2	100	100	Cyanosis, transient neonatal, 613977 Fetal hemoglobin quantitative trait locus 1, 141749
HCCS	107.4	99	99	Linear skin defects with multiple congenital anomalies 1, 309801
HCFC1	106.7	99	96	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cbIX type ), 309541
HCN1	123.5	99	97	Epileptic encephalopathy, early infantile, 24, 615871
HCN4	79.6	98	91	Brugada syndrome 8, 613123 Sick sinus syndrome 2, 163800
HDAC8	131.2	100	99	Cornelia de Lange syndrome 5, 300882
HEATR2	106.5	85	79	Ciliary dyskinesia, primary, 18, 614874
HELLS	93.4	94	86	Immunodeficiency-centromeric instability-facial anomalies syndrome 4,616911
HEPACAM	142.1	81	76	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926
HERC1	173.7	99	99	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
HERC2	114.4	80	77	Mental retardation, autosomal recessive 38, 615516 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220
HES7	29.2	65	42	Spondylocostal dysostosis 4, autosomal recessive, 613686
HESX1	57.8	99	92	Growth hormone deficiency with pituitary anomalies, 182230 Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230
HEXA	126	100	98	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800 [Hex A pseudodeficiency], 272800
HEXB	129.7	99	93	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HFE	142	100	99	Hemochromatosis, 235200 [Transferrin serum level QTL2], 614193 {Alzheimer disease, susceptibility to}, 104300 {Microvascular complications of diabetes 7}, 612635 {Porphyria cutanea tarda, susceptibility to}, 176100 {Porphyria variegata, susceptibility to}, 176200
HFE2	117	100	100	Hemochromatosis type 2A,602390
HFM1	42.8	90	76	Premature ovarian failure 9, 615724
HGD	127.9	100	99	Alkaptonuria, 203500



HGF	146.7	99	96	Deafness, autosomal recessive 39, 608265
HGSNAT	95.2	81	80	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544
HIBCH	67.7	92	69	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HINT1	60	98	88	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200
HIVEP2	191.1	100	100	Mental retardation, autosomal dominant 43, 616977
HK1	140.9	100	99	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285
HLCS	172.9	100	100	Holocarboxylase synthetase deficiency, 253270
HMBS	109	100	99	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000
HMGCL	143.3	100	99	HMG-CoA lyase deficiency, 246450
HMGCS2	131.5	100	100	HMG-CoA synthase-2 deficiency, 605911
HMOX1	129	95	89	Heme oxygenase-1 deficiency, 614034 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963
HMX1	23	56	42	Oculoauricular syndrome, 612109
HNF1A	148.4	100	99	Diabetes mellitus, insulin-dependent, 20, 612520 Hepatic adenoma, somatic, 142330 MODY, type III, 600496 Renal cell carcinoma, 144700 {Diabetes mellitus, insulin-dependent}, 222100 {Diabetes mellitus, noninsulin-dependent, 2}, 125853
HNF1B	123.8	99	99	Diabetes mellitus, noninsulin-dependent, 125853 Renal cysts and diabetes syndrome, 137920 {Renal cell carcinoma}, 144700
HNF4A	142.1	99	99	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026 MODY, type I, 125850 {Diabetes mellitus, noninsulin-dependent}, 125853
HNMT	135.1	100	99	Mental retardation, autosomal recessive 51, 616739 {Asthma, susceptibility to}, 600807
HNRNPA1	72.5	97	87	Amyotrophic lateral sclerosis 20, 615426 ?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3, 615424
HNRNPDL	58.9	90	79	Muscular dystrophy, limb-girdle, type 1G, 609115
HNRNPK	71.6	86	78	Au-Kline syndrome, 616580

HOGA1	147.7	99	98	Hyperoxaluria, primary, type III, 613616
HOXA1	165.6	100	100	Athabaskan brainstem dysgenesis syndrome, 601536 Bosley-Salih-Alorainy syndrome, 601536
HOXA11	86.4	87	78	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
HOXA13	49.3	69	61	Guttmacher syndrome, 176305 Hand-foot-uterus syndrome, 140000
HOXB1	104.5	100	100	Facial paresis, hereditary congenital, 3, 614744
HOXC13	105.3	97	91	Ectodermal dysplasia 9, hair/nail type, 614931
HOXD10	137.8	100	99	Charcot-Marie-Tooth disease, foot deformity of, 192950 Vertical talus, congenital, 192950
HOXD13	142.2	93	89	Brachydactyly, type D, 113200 Brachydactyly, type E, 113300 Syndactyly, type V, 186300 Synpolydactyly 1, 186000 ?Brachydactyly-syndactyly syndrome, 610713
HPCA	283.9	100	100	Dystonia 2, torsion, autosomal recessive, 224500
HPD	138	100	100	Hawkinsinuria, 140350 Tyrosinemia, type III, 276710
HPGD	87.9	100	98	Cranioosteoarthropathy, 259100 Digital clubbing, isolated congenital, 119900 Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100
HPRT1	58.6	96	84	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322
HPS1	118	100	99	Hermansky-Pudlak syndrome 1, 203300
HPS3	135.2	99	96	Hermansky-Pudlak syndrome 3, 614072
HPS4	144.5	100	100	Hermansky-Pudlak syndrome 4, 614073
HPS5	133	99	98	Hermansky-Pudlak syndrome 5, 614074
HPS6	139.2	91	84	Hermansky-Pudlak syndrome 6, 614075
HPSE2	110.4	98	95	Urofacial syndrome 1, 236730
HR	95	97	94	Alopecia universalis, 203655 Atrichia with papular lesions, 209500 Hypotrichosis 4, 146550

HRAS	164.9	99	98	Congenital myopathy with excess of muscle spindles, 218040 Costello syndrome, 218040 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 {Bladder cancer, somatic}, 109800 {Nevus sebaceous or woolly hair nevus, somatic}, 162900 {Spitz nevus or nevus spilus, somatic}, 137550 {Thyroid carcinoma, follicular, somatic}, 188470
HRG	178.5	95	94	Thrombophilia due to elevated HRG, 613116 Thrombophilia due to HRG deficiency, 613116
HSD11B1	147	100	99	Cortisone reductase deficiency 2, 614662
HSD11B2	165.4	85	82	Apparent mineralocorticoid excess, 218030
HSD17B10	117.9	100	99	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 ?Mental retardation, X-linked syndromic 10, 300220
HSD17B3	156.8	100	100	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	94.9	93	90	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B2	189.3	100	100	3-beta-hydroxysteroid dehydrogenase, type II, deficiency, 201810
HSD3B7	136.6	98	91	Bile acid synthesis defect, congenital, 1, 607765
HSF4	103.1	97	94	Cataract 5, multiple types, 116800
HSPA9	91.6	91	85	Anemia, sideroblastic, 4, 182170 Even-plus syndrome, 616854
HSPB1	39.9	93	81	Charcot-Marie-Tooth disease, axonal, type 2F, 606595 Neuropathy, distal hereditary motor, type IIB, 608634
HSPB8	156.4	100	100	Charcot-Marie-Tooth disease, axonal, type 2L, 608673 Neuropathy, distal hereditary motor, type IIA, 158590
HSPD1	99.8	98	93	Leukodystrophy, hypomyelinating, 4, 612233 Spastic paraplegia 13, autosomal dominant, 605280
HSPG2	121.4	99	98	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
HTR1A	220.6	100	100	Periodic fever, menstrual cycle dependent, 614674
HTRA1	98.3	84	81	CARASIL syndrome, 600142 Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779 {Macular degeneration, age-related, 7}, 610149

				{Macular degeneration, age-related, neovascular type}, 610149
HTT	143.9	98	97	Huntington disease, 143100
HUWE1	99.1	99	97	Mental retardation, X-linked syndromic, Turner type, 300706
HYDIN	133.1	99	99	Ciliary dyskinesia, primary, 5, 608647
HYLS1	171.2	100	100	Hydrolethalus syndrome, 236680
ICK	131.7	99	98	Endocrine-cerebroosteodysplasia, 612651
ICOS	159.9	100	100	Immunodeficiency, common variable, 1, 607594
IDH2	103.7	99	96	D-2-hydroxyglutaric aciduria 2, 613657
IDH3B	173.6	100	100	Retinitis pigmentosa 46, 612572
IDS	110.7	99	98	Mucopolysaccharidosis II, 309900
IDUA	121.3	89	82	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016
IER3IP1	71.9	92	80	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	113.4	99	97	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFITM5	63.3	99	94	Osteogenesis imperfecta, type V, 610967
IFNGR1	133	99	97	Immunodeficiency 27A, mycobacteriosis, AR, 209950 Immunodeficiency 27B, mycobacteriosis, AD, 615978 {H. pylori infection, susceptibility to}, 600263 {Hepatitis B virus infection, susceptibility to}, 610424 {Tuberculosis infection, protection against}, 607948 {Tuberculosis, susceptibility to}, 607948
IFNGR2	142.2	93	93	Immunodeficiency 28, mycobacteriosis, 614889
IFT122	152.3	100	99	Cranioectodermal dysplasia 1, 218330
IFT140	115	100	99	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	116.5	100	99	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT43	114.8	100	100	Cranioectodermal dysplasia 3, 614099
IFT80	57.6	87	70	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IGBP1	119.2	99	97	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472
IGF1	122.7	100	100	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747

IGF1R	144.2	100	99	Insulin-like growth factor I, resistance to, 270450
IGF2R	139.6	98	96	Hepatocellular carcinoma, somatic, 114550
IGFALS	79.7	99	96	Acid-labile subunit, deficiency of, 615961
IGFBP7	71.6	91	83	Retinal arterial macroaneurysm with supra-valvular pulmonic stenosis, 614224
IGHG2	39	88	67	IgG2 deficiency, selective
IGHM	196.1	100	100	Agammaglobulinemia 1, 601495
IGHMBP2	107.9	99	96	Charcot-Marie-Tooth disease, axonal, type 2S, 616155 Neuronopathy, distal hereditary motor, type VI, 604320
IGKC	158.1	100	100	Kappa light chain deficiency, 614102
IGLL1	86.2	99	94	Agammaglobulinemia 2, 613500
IGSF1	85.5	99	96	Hypothyroidism, central, and testicular enlargement, 300888
IHH	129.4	100	100	Acrocapitofemoral dysplasia, 607778 Brachydactyly, type A1, 112500
IKBKAP	142.7	99	98	Dysautonomia, familial, 223900
IKBKB	123.7	98	94	Immunodeficiency 15, 615592
IKBKG	52.9	84	73	Ectodermal dysplasia, hypohidrotic, with immune deficiency, 300291 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 Immunodeficiency 33, 300636 Immunodeficiency, isolated, 300584 Incontinentia pigmenti, 308300 Invasive pneumococcal disease, recurrent isolated, 2, 300640
IKZF1	183.4	100	100	Immunodeficiency, common variable, 1, 616873
IL10RA	142.3	100	99	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	168.7	98	96	Inflammatory bowel disease 25, early onset, autosomal recessive, 612567 {Hepatitis B virus, susceptibility to}, 610424
IL11RA	140.1	100	99	Craniosynostosis and dental anomalies, 614188
IL12B	121.4	100	99	Immunodeficiency 29, mycobacteriosis, 614890
IL12RB1	124.5	97	94	Immunodeficiency 30, 614891
IL17RC	96.2	99	99	Candidiasis, familial, 9, 616445
IL17RD	133.9	99	97	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
IL1RAPL1	117.6	99	97	Mental retardation, X-linked 21/34, 300143
IL1RN	162.9	100	100	Interleukin 1 receptor antagonist deficiency, 612852 {Gastric cancer risk after H. pylori infection}, 137215 {Microvascular complications of diabetes 4}, 612628

IL21R	128.7	100	100	Immunodeficiency, primary, autosomal recessive, IL21R-related, 615207 [IgE, elevated level of], 147050
IL2RA	116.7	100	99	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367 {Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942
IL2RG	65.7	99	97	Combined immunodeficiency, X-linked, moderate, 312863 Severe combined immunodeficiency, X-linked, 300400
IL31RA	124.4	100	99	Amyloidosis, primary localized cutaneous, 2, 613955
IL36RN	99.1	100	100	Psoriasis 14, pustular, 614204
IL7R	129.2	99	99	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
ILD1	107.5	100	99	Deafness, autosomal recessive 42, 609646
IMPAD1	147.2	99	99	Chondrodysplasia with joint dislocations, GPAPP type, 614078
IMPDH1	61.3	87	83	Leber congenital amaurosis 11, 613837 Retinitis pigmentosa 10, 180105
IMPG1	103.3	99	97	Macular dystrophy, vitelliform, 4, 616151
IMPG2	154.4	99	97	Macular dystrophy, vitelliform, 5, 616152 Retinitis pigmentosa 56, 613581
INF2	88.2	93	90	Charcot-Marie-Tooth disease, dominant intermediate E, 614455 Glomerulosclerosis, focal segmental, 5, 613237
ING1	128	99	95	Squamous cell carcinoma, head and neck, somatic, 275355
INPP5E	89.3	95	90	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INPPL1	123.8	96	93	Opsismodysplasia, 258480
INS	87.8	100	99	Diabetes mellitus, insulin-dependent, 2, 125852 Diabetes mellitus, permanent neonatal, 606176 Hyperproinsulinemia, 616214 Maturity-onset diabetes of the young, type 10, 613370
INSL3	67.7	81	77	Cryptorchidism, 219050
INSR	141.1	97	94	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968 Leprechaunism, 246200 Rabson-Mendenhall syndrome, 262190
INVS	159.6	100	100	Nephronophthisis 2, infantile, 602088
IQCB1	92.2	89	75	Senior-Loken syndrome 5, 609254
IQSEC2	61.6	92	82	Mental retardation, X-linked 1/78, 309530

IRAK4	95.4	98	90	Invasive pneumococcal disease, recurrent isolated, 1, 610799 IRAK4 deficiency, 607676
IRF1	185.6	100	100	Gastric cancer, somatic, 613659 Myelodysplastic syndrome, preleukemic Myelogenous leukemia, acute Nonsmall cell lung cancer, somatic, 211980
IRF6	113.7	99	98	Popliteal pterygium syndrome 1, 119500 van der Woude syndrome, 119300 {Orofacial cleft 6}, 608864
IRF8	114.6	99	97	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893 Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 614894
IRGM	181.1	100	100	Inflammatory bowel disease 19, 612278 {Mycobacterium tuberculosis, protection against}, 607948
IRX5	73.9	94	86	Hamamy syndrome, 611174
ISCA2	92	99	96	Multiple mitochondrial dysfunctions syndrome 4, 616370
ISCU	111.2	100	99	Myopathy with lactic acidosis, hereditary, 255125
ISG15	160.6	100	100	Immunodeficiency 38, 616126
ISPD	104.2	95	84	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052
ITCH	124.7	95	94	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITGA2B	119.9	99	97	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Glanzmann thrombasthenia, 273800 Thrombocytopenia, neonatal alloimmune, BAK antigen related
ITGA3	142.8	99	98	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748
ITGA6	146.6	99	99	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730
ITGA7	130.4	99	97	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
ITGA8	122.6	99	98	Renal hypodysplasia/aplasia 1, 191830
ITGB2	152.5	100	99	Leukocyte adhesion deficiency, 116920
ITGB3	138.9	99	97	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Glanzmann thrombasthenia, 273800 Purpura, posttransfusion Thrombocytopenia, neonatal alloimmune {Myocardial infarction, susceptibility to}, 608446

ITGB4	150.4	97	94	Epidermolysis bullosa of hands and feet, 131800 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa, junctional, with pyloric atresia, 226730
ITGB6	136.9	96	95	Amelogenesis imperfecta, type IH, 616221
ITK	125.3	100	99	Lymphoproliferative syndrome 1, 613011
ITM2B	110.1	99	98	Dementia, familial British, 176500 Dementia, familial Danish, 117300 ?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079
ITPA	120.4	100	100	Epileptic encephalopathy, early infantile, 35, 616647 [Inosine triphosphatase deficiency], 613850
ITPR1	161.5	100	99	Gillespie syndrome, 206700 Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360
IVD	115.1	100	100	Isovaleric acidemia, 243500
IYD	117.4	99	97	Thyroid dyshormonogenesis 4, 274800
JAG1	148.6	98	97	Alagille syndrome, 118450 Tetralogy of Fallot, 187500 ?Deafness, congenital heart defects, and posterior embryotoxon
JAGN1	147.3	100	100	Neutropenia, severe congenital, 6, autosomal recessive, 616022
JAK2	90.5	95	94	Erythrocytosis, somatic, 133100 Leukemia, acute myeloid, somatic, 601626 Myelofibrosis, somatic, 254450 Polycythemia vera, somatic, 263300 Thrombocythemia 3, 614521 {Budd-Chiari syndrome, somatic}, 600800
JAK3	104.3	98	95	SCID, autosomal recessive, T-negative/B-positive type, 600802
JAM3	144.9	99	98	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
JPH2	87.6	90	75	Cardiomyopathy, hypertrophic, 17, 613873
JPH3	156.9	100	99	Huntington disease-like 2, 606438
JUP	145.3	100	99	Arrhythmogenic right ventricular dysplasia 12, 611528 Naxos disease, 601214
KAL1	90.9	89	87	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
KANK1	166.4	100	100	Cerebral palsy, spastic quadriplegic, 2, 612900
KANK2	151.7	99	99	Palmoplantar keratoderma and woolly hair, 616099



KANSL1	171.3	99	99	Koolen-De Vries syndrome, 610443
KARS	122.7	100	99	Deafness, autosomal recessive 89, 613916 ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641
KAT6A	169.5	100	99	Mental retardation, autosomal dominant 32, 616268
KAT6B	192.3	99	98	Genitopatellar syndrome, 606170 SBBYSS syndrome, 603736
KATNB1	141.9	100	100	Lissencephaly 6, with microcephaly, 616212
KBTBD13	107.1	99	96	Nemaline myopathy 6, autosomal dominant, 609273
KCNA1	167	100	99	Episodic ataxia/myokymia syndrome, 160120
KCNA2	162.2	100	100	Epileptic encephalopathy, early infantile, 32, 616366
KCNA5	143.7	99	96	Atrial fibrillation, familial, 7, 612240
KCNB1	145	100	99	Epileptic encephalopathy, early infantile, 26, 616056
KCNC1	199.4	100	100	Epilepsy, progressive myoclonic 7, 616187
KCNC3	129.7	63	54	Spinocerebellar ataxia 13, 605259
KCND3	182.9	99	99	Brugada syndrome 9, 616399 Spinocerebellar ataxia 19, 607346
KCNE1	463.7	100	100	Jervell and Lange-Nielsen syndrome 2, 612347 Long QT syndrome 5, 613695
KCNE2	181.5	100	100	Atrial fibrillation, familial, 4, 611493 Long QT syndrome 6, 613693
KCNE3	177.2	100	100	Brugada syndrome 6, 613119
KCNH1	188.2	100	99	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500
KCNH2	102.8	92	84	Long QT syndrome 2, 613688 Short QT syndrome 1, 609620 {Long QT syndrome 2, acquired, susceptibility to}, 613688
KCNJ1	233.8	100	100	Bartter syndrome, type 2, 241200
KCNJ10	238.8	100	99	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ11	300.2	100	100	Diabetes mellitus, permanent neonatal, with neurologic features, 606176 Diabetes mellitus, transient neonatal, 3, 610582 Diabetes, permanent neonatal, 606176 Hyperinsulinemic hypoglycemia, familial, 2, 601820 Maturity-onset diabetes of the young, type 13, 616329

				{Diabetes mellitus, type 2, susceptibility to}, 125853
KCNJ13	210.6	100	99	Leber congenital amaurosis 16, 614186 Snowflake vitreoretinal degeneration, 193230
KCNJ2	229.3	100	100	Andersen syndrome, 170390 Atrial fibrillation, familial, 9, 613980 Short QT syndrome 3, 609622
KCNJ5	194.5	100	99	Hyperaldosteronism, familial, type III, 613677 Long QT syndrome 13, 613485
KCNJ6	156	100	99	Keppen-Lubinsky syndrome, 614098
KCNK3	165.6	98	96	Pulmonary hypertension, primary, 4, 615344
KCNK9	194	100	100	Birk-Barel mental retardation dysmorphism syndrome, 612292
KCNMA1	131.4	100	99	Generalized epilepsy and paroxysmal dyskinesia, 609446
KCNN4	136.3	100	99	Dehydrated hereditary stomatocytosis 2, 616689
KCNQ1	114.9	93	90	Atrial fibrillation, familial, 3, 607554 Jervell and Lange-Nielsen syndrome, 220400 Long QT syndrome 1, 192500 Short QT syndrome 2, 609621 {Long QT syndrome 1, acquired, susceptibility to}, 192500
KCNQ1OT1	NC	NC	NC	Beckwith-Wiedemann syndrome, 130650
KCNQ2	93.2	98	94	Epileptic encephalopathy, early infantile, 7, 613720 Myokymia, 121200 Seizures, benign neonatal, 1, 121200
KCNQ3	111.1	98	95	Seizures, benign neonatal, type 2, 121201
KCNQ4	135.4	93	89	Deafness, autosomal dominant 2A, 600101
KCNT1	112.2	95	92	Epilepsy, nocturnal frontal lobe, 5, 615005 Epileptic encephalopathy, early infantile, 14, 614959
KCNV2	137.7	100	100	Retinal cone dystrophy 3B, 610356
KCTD1	138.8	89	82	Scalp-ear-nipple syndrome, 181270
KCTD17	93.9	95	87	Dystonia 26, myoclonic, 616398
KCTD7	148.9	94	92	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726

KDM1A	129.5	96	93	Cleft palate, psychomotor retardation, and distinctive facial features, 616728
KDM5C	113.5	97	95	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
KDM6A	109.6	93	84	Kabuki syndrome 2, 300867
KDR	137.8	100	99	Hemangioma, capillary infantile, somatic, 602089 {Hemangioma, capillary infantile, susceptibility to}, 602089
KERA	191.2	100	100	Cornea plana congenita, recessive, 217300
KHDC3L	110.1	99	99	Hydatidiform mole, recurrent, 2, 614293
KIAA0196	146.6	99	98	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, autosomal dominant, 603563
KIAA0586	114.7	98	92	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546
KIAA1279	166.1	100	100	Goldberg-Shprintzen megacolon syndrome, 609460
KIAA2022	140.1	99	99	Mental retardation, X-linked 98, 300912
KIF11	83.6	97	94	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF1A	114.2	99	96	Mental retardation, autosomal dominant 9, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal recessive, 610357
KIF1B	154.8	100	99	Pheochromocytoma, 171300 ?Charcot-Marie-Tooth disease, type 2A1, 118210 {Neuroblastoma, susceptibility to, 1}, 256700
KIF1C	121.5	100	99	Spastic ataxia 2, autosomal recessive, 611302
KIF21A	123.2	99	96	Fibrosis of extraocular muscles, congenital, 1, 135700 Fibrosis of extraocular muscles, congenital, 3B, 135700
KIF22	161.8	100	99	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546
KIF2A	105.3	97	88	Cortical dysplasia, complex, with other brain malformations 3, 615411
KIF5A	136.3	100	99	Spastic paraplegia 10, autosomal dominant, 604187
KIF5C	116.4	99	99	Cortical dysplasia, complex, with other brain malformations 2, 615282
KIF7	85.8	93	88	Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalnova syndrome, 607131 ?Hydroletharus syndrome 2, 614120
KIRREL3	136.4	100	99	Mental retardation, autosomal dominant 4, 612581
KISS1R	106.6	99	95	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 ?Precocious puberty, central, 1, 176400

KIT	152.9	100	99	Gastrointestinal stromal tumor, familial, 606764 Germ cell tumors, 273300 Leukemia, acute myeloid, 601626 Mast cell disease, 154800 Piebaldism, 172800
KITLG	81.9	96	91	Deafness, congenital, unilateral or asymmetric, 616697 Hyperpigmentation with or without hypopigmentation, 145250 [Skin/hair/eye pigmentation 7, blond/brown hair], 611664
KIZ	177.7	98	95	Retinitis pigmentosa 69, 615780
KL	181.3	97	96	Tumoral calcinosis, hyperphosphatemic, 211900 {Coronary artery disease, susceptibility to}
KLC2	116	99	98	Spastic paraplegia, optic atrophy, and neuropathy, 609541
KLF1	52	90	81	Blood group--Lutheran inhibitor, 111150 Dyserythropoietic anemia, congenital, type IV, 613673 [Hereditary persistence of fetal hemoglobin], 613566
KLF11	181.6	99	99	Maturity-onset diabetes of the young, type VII, 610508
KLF6	154.4	100	100	Gastric cancer, somatic, 613659 Prostate cancer, somatic, 176807
KLHL10	210.1	100	100	Spermatogenic failure 11, 615081
KLHL3	141.1	99	99	Pseudohypoaldosteronism, type IID, 614495
KLHL40	158.2	100	100	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	203.9	100	99	Nemaline myopathy 9, 615731
KLHL7	120.5	99	97	Cold induced sweating syndrome 3, 617055 Retinitis pigmentosa 42, 612943
KLK4	185.5	100	98	Amelogenesis imperfecta, type IIA1, 204700
KLKB1	143.5	99	96	Fletcher factor (prekallikrein) deficiency, 612423
KLLN	117.5	100	100	Cowden syndrome 4, 615107
KMT2A	152.5	99	98	Leukemia, myeloid/lymphoid or mixed-lineage, 159555 Wiedemann-Steiner syndrome, 605130
KMT2D	142.2	99	99	Kabuki syndrome 1, 147920
KPTN	112.2	100	99	Mental retardation, autosomal recessive 41, 615637

KRAS	64.6	99	98	Bladder cancer, somatic, 109800 Breast cancer, somatic, 114480 Cardiofaciocutaneous syndrome 2, 615278 Gastric cancer, somatic, 137215 Leukemia, acute myeloid, 601626 Lung cancer, somatic, 211980 Noonan syndrome 3, 609942 Pancreatic carcinoma, somatic, 260350 RAS-associated autoimmune leukoproliferative disorder, 614470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200
KRIT1	85	99	97	Cavernous malformations of CNS and retina, 116860 Cerebral cavernous malformations-1, 116860 Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations, 116860
KRT1	120	99	96	Epidermolytic hyperkeratosis, 113800 Ichthyosis histrix, Curth-Macklin type, 146590 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602 Keratosis palmoplantaris striata III, 607654 Palmoplantar keratoderma, epidermolytic, 144200 Palmoplantar keratoderma, nonepidermolytic, 600962
KRT10	103.1	98	93	Epidermolytic hyperkeratosis, 113800 Ichthyosis with confetti, 609165 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602
KRT12	126.2	98	95	Meesmann corneal dystrophy, 122100
KRT13	140.7	99	98	White sponge nevus 2, 615785
KRT14	59.4	89	82	Dermatopathia pigmentosa reticularis, 125595 Epidermolysis bullosa simplex, Dowling-Meara type, 131760 Epidermolysis bullosa simplex, Koebner type, 131900 Epidermolysis bullosa simplex, recessive 1, 601001 Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 Naegeli-Franceschetti-Jadassohn syndrome, 161000
KRT16	38.6	72	53	Pachyonychia congenita 1, 167200 Palmoplantar keratoderma, nonepidermolytic, focal, 613000
KRT17	21.6	47	31	Pachyonychia congenita 2, 167210 Steatocystoma multiplex, 184500

KRT18	40.3	84	67	Cirrhosis, cryptogenic, 215600 {Cirrhosis, noncryptogenic, susceptibility to}, 215600
KRT2	140.8	100	99	Ichthyosis bullosa of Siemens, 146800
KRT25	143.1	100	100	Woolly hair, autosomal recessive 3, 616760
KRT3	105.4	100	99	Meesmann corneal dystrophy, 122100
KRT4	130.1	100	99	White sponge nevus 1, 193900
KRT5	133.7	100	100	Dowling-Degos disease 1, 179850 Epidermolysis bullosa simplex, Dowling-Meara type, 131760 Epidermolysis bullosa simplex, Koebner type, 131900 Epidermolysis bullosa simplex, recessive 1, 601001 Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 Epidermolysis bullosa simplex-MP, 131960 Epidermylysis bullosa simplex-MCR, 609352
KRT6A	198.1	94	87	Pachyonychia congenita 3, 615726
KRT6B	195	95	89	Pachyonychia congenita 4, 615728
KRT6C	174.6	87	79	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735
KRT74	150.5	99	98	?Ectodermal dysplasia 7, hair/nail type, 614929 ?Hypotrichosis 3, 613981 Woolly hair, autosomal dominant, 194300
KRT8	39.7	92	73	Cirrhosis, cryptogenic, 215600 {Cirrhosis, noncryptogenic, susceptibility to}, 215600
KRT81	93.6	99	96	Monilethrix, 158000
KRT85	108.5	98	95	Ectodermal dysplasia 4, hair/nail type, 602032
KRT86	101.4	100	98	Monilethrix, 158000
KRT9	84.9	98	96	Palmoplantar keratoderma, epidermolytic, 144200
L1CAM	133.8	99	98	Corpus callosum, partial agenesis of, 304100 CRASH syndrome, 303350 Hydrocephalus due to aqueductal stenosis, 307000 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 Hydrocephalus with Hirschsprung disease, 307000 MASA syndrome, 303350
L2HGDH	129.1	98	97	L-2-hydroxyglutaric aciduria, 236792
LAMA1	137.3	100	99	Poretti-Boltshauser syndrome, 615960
LAMA2	143.6	99	99	Muscular dystrophy, congenital merosin-deficient, 607855

				Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855
LAMA3	147.7	99	99	Epidermolysis bullosa, generalized atrophic benign, 226650 Epidermolysis bullosa, junctional, Herlitz type, 226700 Laryngoonychocutaneous syndrome, 245660
LAMA4	132.6	100	99	Cardiomyopathy, dilated, 1JJ, 615235
LAMB1	170	100	99	Lissencephaly 5, 615191
LAMB2	202	100	99	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 Pierson syndrome, 609049
LAMB3	123.6	100	99	Amelogenesis imperfecta, type IA, 104530 Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LAMC2	117.7	99	98	Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LAMC3	130.8	98	96	Cortical malformations, occipital, 614115
LAMP2	106.8	92	91	Danon disease, 300257
LAMTOR2	167.3	100	99	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LARGE	142.6	100	99	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	56.2	80	63	Alazami syndrome, 615071
LARS2	143.2	100	100	Perrault syndrome 4, 615300 ?Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LAS1L	90.8	99	97	Wilson-Turner syndrome, 309585
LBR	87.9	93	83	Greenberg skeletal dysplasia, 215140 Pelger-Huet anomaly, 169400 ?Reynolds syndrome, 613471
LCA5	127.6	97	95	Leber congenital amaurosis 5, 604537
LCAT	145.9	97	91	Fish-eye disease, 136120 Norum disease, 245900
LCT	142	99	99	Lactase deficiency, congenital, 223000
LDB3	127.4	95	93	Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 Cardiomyopathy, hypertrophic, 24, 601493 Left ventricular noncompaction 3, 601493 Myopathy, myofibrillar, 4, 609452
LDHA	59.8	94	87	Glycogen storage disease XI, 612933

LDLR	171.4	99	97	Hypercholesterolemia, familial, 143890 LDL cholesterol level QTL2, 143890
LDLRAP1	156.5	95	91	Hypercholesterolemia, familial, autosomal recessive, 603813
LEF1	116	100	99	Sebaceous tumors, somatic
LEFTY2	42.3	91	77	Left-right axis malformations (Koasaki (1999) Am J Hum Genet 64, 712)
LEMD2	68.7	89	80	Cataract 46, juvenile-onset, 212500
LEMD3	96.5	95	88	Buschke-Ollendorff syndrome, 166700 Melorheostosis with osteopoikilosis, 155950 Osteopoikilosis, 166700
LEP	188.9	100	99	Obesity, morbid, due to leptin deficiency, 614962
LEPR	109.7	93	90	Obesity, morbid, due to leptin receptor deficiency, 614963
LEPRE1	137.1	100	99	Osteogenesis imperfecta, type VIII, 610915
LEPREL1	100.2	99	93	Myopia, high, with cataract and vitreoretinal degeneration, 614292
LGI1	167.3	99	96	Epilepsy, familial temporal lobe, 1, 600512
LHB	29	97	73	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300
LHCGR	154.4	95	92	Leydig cell adenoma, somatic, with precocious puberty, 176410 Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320 Leydig cell hypoplasia with pseudohermaphroditism, 238320 Luteinizing hormone resistance, female, 238320 Precocious puberty, male, 176410
LHFPL5	288.3	100	100	Deafness, autosomal recessive 67, 610265
LHX3	87.8	97	83	Pituitary hormone deficiency, combined, 3, 221750
LHX4	145.2	100	99	Pituitary hormone deficiency, combined, 4, 262700
LIAS	133.6	99	97	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIFR	123.3	97	92	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LIG1	95.4	100	99	DNA ligase I deficiency
LIG4	165.3	100	99	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500
LIM2	103.6	100	98	Cataract 19, multiple types, 615277
LIMS2	111	93	92	Muscular dystrophy, limb-girdle, type 2W, 616827
LINS	147.6	99	98	Mental retardation, autosomal recessive 27, 614340
LIPA	110.9	98	95	Cholesteryl ester storage disease, 278000 Wolman disease, 278000



LIPC	115.3	100	99	Hepatic lipase deficiency, 614025 [High density lipoprotein cholesterol level QTL 12], 612797 {Diabetes mellitus, noninsulin-dependent}, 125853
LIPE	113.5	99	98	Lipodystrophy, familial partial, type 6, 615980
LIPH	140.4	100	100	Hypotrichosis 7, 604379 Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379
LIPN	124.9	99	95	Ichthyosis, congenital, autosomal recessive 8, 613943
LIPT1	227.4	100	100	Lipoyltransferase 1 deficiency, 616299
LITAF	126.6	94	91	Charcot-Marie-Tooth disease, type 1C, 601098
LMAN1	129.9	99	94	Combined factor V and VIII deficiency, 227300
LMBR1	106.8	97	90	Acheiropody, 200500 Hypoplastic or aplastic tibia with polydactyly, 188740 Laurin-Sandrow syndrome, 135750 Polydactyly, preaxial type II, 174500 Syndactyly, type IV, 186200 Triphalangeal thumb, type I, 174500 Triphalangeal thumb-polysyndactyly syndrome, 174500
LMBRD1	80.1	91	82	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LMF1	132.8	99	97	Lipase deficiency, combined, 246650
LMNA	89.4	98	91	Cardiomyopathy, dilated, 1A, 115200 Charcot-Marie-Tooth disease, type 2B1, 605588 Emery-Dreifuss muscular dystrophy 2, AD, 181350 Emery-Dreifuss muscular dystrophy 3, AR, 616516 Heart-hand syndrome, Slovenian type, 610140 Hutchinson-Gilford progeria, 176670 Lipodystrophy, familial partial, 2, 151660 Malouf syndrome, 212112 Mandibuloacral dysplasia, 248370 Muscular dystrophy, congenital, 613205 Muscular dystrophy, limb-girdle, type 1B, 159001 Restrictive dermopathy, lethal, 275210
LMNB1	123.3	99	99	Leukodystrophy, adult-onset, autosomal dominant, 169500
LMOD3	141.7	99	98	Nemaline myopathy 10, 616165
LMX1B	111.5	96	92	Nail-patella syndrome, 161200
LONP1	141.7	98	96	CODAS syndrome, 600373

LOR	13.6	63	33	Vohwinkel syndrome with ichthyosis, 604117
LOXHD1	136.7	100	99	Deafness, autosomal recessive 77, 613079
LPAR6	103.8	99	98	Hypotrichosis 8, 278150 Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150
LPIN1	131.7	99	95	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	111.6	100	99	Majeed syndrome, 609628
LPL	147.1	100	100	Combined hyperlipidemia, familial, 144250 Lipoprotein lipase deficiency, 238600 [High density lipoprotein cholesterol level QTL 11]
LPP	132.1	100	100	Leukemia, acute myeloid, 601626 Lipoma
LRAT	298.3	100	100	Leber congenital amaurosis 14, 613341 Retinal dystrophy, early-onset severe, 613341 Retinitis pigmentosa, juvenile, 613341
LRBA	134.3	99	97	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRIG2	148.5	99	97	Urofacial syndrome 2, 615112
LRIT3	142.6	94	94	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRP2	176.4	100	99	Donnai-Barrow syndrome, 222448
LRP4	166.8	99	98	Cenani-Lenz syndactyly syndrome, 212780 Sclerosteosis 2, 614305 ?Myasthenic syndrome, congenital, 17, 616304
LRP5	190.1	98	97	Exudative vitreoretinopathy 4, 601813 Hyperostosis, endosteal, 144750 Osteopetrosis, autosomal dominant 1, 607634 Osteoporosis-pseudoglioma syndrome, 259770 Osteosclerosis, 144750 van Buchem disease, type 2, 607636 [Bone mineral density variability 1], 601884 {Osteoporosis}, 166710
LRP6	169.5	100	99	Tooth agenesis, selective, 7, 616724 {Coronary artery disease, autosomal dominant, 2}, 610947
LRPAP1	138.7	99	97	Myopia 23, autosomal recessive, 615431
LRPPRC	127.1	99	97	Leigh syndrome, French-Canadian type, 220111
LRRC6	138	94	91	Ciliary dyskinesia, primary, 19, 614935

LRRC8A	273.8	100	99	Agammaglobulinemia 5, 613506
LRSAM1	130.5	100	99	Charcot-Marie-Tooth disease, axonal, type 2P, 614436
LRTOMT	126.1	99	96	Deafness, autosomal recessive 63, 611451
LSS	128.1	100	99	Cataract 44, 616509
LTBP2	104.7	99	97	Glaucoma 3, primary congenital, D, 613086 Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750 Weill-Marchesani syndrome 3, recessive, 614819
LTBP3	113.8	98	94	Dental anomalies and short stature, 601216
LTBP4	117.5	98	95	Cutis laxa, autosomal recessive, type IC, 613177
LYRM7	49	87	72	Mitochondrial complex III deficiency, nuclear type 8, 615838
LYST	134.6	97	93	Chediak-Higashi syndrome, 214500
LYZ	165.5	100	100	Amyloidosis, renal, 105200
LZTFL1	108.7	99	95	Bardet-Biedl syndrome 17, 615994
LZTR1	134.3	100	99	Noonan syndrome 10, 616564 {Schwannomatosis-2, susceptibility to}, 615670
LZTS1	105.9	100	100	Esophageal squamous cell carcinoma, 133239
MAB21L2	245.9	100	100	Microphthalmia, syndromic 14, 615877
MAD1L1	97.9	99	97	Lymphoma, somatic Prostate cancer, somatic, 176807
MAF	60.2	77	72	Ayme-Gripp syndrome, 601088 Cataract 21, multiple types, 610202
MAFB	93.8	99	97	Duane retraction syndrome 3, 617041 Multicentric carpotarsal osteolysis syndrome, 166300
MAG	146.6	100	99	Spastic paraplegia 75, autosomal recessive, 616680
MAGED2	80.6	99	97	Bartter syndrome, type 5, antenatal, transient, 300971
MAGEL2	129.4	100	100	Schaaf-Yang syndrome, 615547
MAGT1	102.7	98	95	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853
MAK	142.6	95	94	Retinitis pigmentosa 62, 614181
MALT1	136.6	89	85	Immunodeficiency 12, 615468
MAML2	123	100	100	Mucoepidermoid salivary gland carcinoma
MAMLD1	132.4	99	98	Hypospadias 2, X-linked, 300758
MAN1B1	142.2	100	99	Mental retardation, autosomal recessive 15, 614202
MAN2B1	122.5	99	96	Mannosidosis, alpha-, types I and II, 248500

MANBA	119.9	99	97	Mannosidosis, beta, 248510
MAOA	114.5	99	98	Brunner syndrome, 300615 {Antisocial behavior},300615
MAP2K1	92.3	99	95	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	108	97	89	Cardiofaciocutaneous syndrome 4, 615280
MAP3K1	165.9	92	89	46XY sex reversal 6, 613762
MAP3K8	143.4	100	99	Lung cancer, somatic, 211980
MAPRE2	205.9	100	99	Symmetric circumferential skin creases, congenital, 2, 616734
MAPT	134.4	100	98	Dementia, frontotemporal, with or without parkinsonism, 600274 Pick disease, 172700 Supranuclear palsy, progressive atypical, 260540 Supranuclear palsy, progressive, 601104 {Parkinson disease, susceptibility to}, 168600
MARS	128	99	97	Charcot-Marie-Tooth disease, axonal, type 2U, 616280 Interstitial lung and liver disease, 615486
MARS2	173.3	100	100	Spastic ataxia 3, autosomal recessive, 611390 ?Combined oxidative phosphorylation deficiency 25, 616430
MARVELD2	159.6	97	93	Deafness, autosomal recessive 49, 610153
MASP1	148.9	100	99	3MC syndrome 1, 257920
MASP2	139.8	100	99	MASP2 deficiency, 613791
MAT1A	185.7	99	97	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MATN3	116.7	84	84	Epiphyseal dysplasia, multiple, 5, 607078 Spondyloepimetaphyseal dysplasia, 608728 {Osteoarthritis susceptibility 2}, 140600
MATR3	92.3	95	88	Amyotrophic lateral sclerosis 21, 606070
MBD5	196.7	100	99	Mental retardation, autosomal dominant 1, 156200
MBTPS2	114.2	99	97	IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800 ?Olmsted syndrome, X-linked, 300918
MC2R	213	100	100	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MC4R	272	100	100	Obesity, autosomal dominant, 601665
MCC	139.9	100	99	Colorectal cancer, somatic, 114500

MCCC1	151.8	100	99	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	131	100	99	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	99.9	100	100	Methylmalonyl-CoA epimerase deficiency, 251120
MCFD2	101.5	99	98	Factor V and factor VIII, combined deficiency of, 613625
MCM4	164.5	99	98	Natural killer cell and glucocorticoid deficiency with DNA repair defect, 609981
MCM6	154.2	100	99	Lactase persistence/nonpersistence, 223100
MCM9	151.5	100	99	Ovarian dysgenesis 4, 616185
MCOLN1	150.7	98	97	Mucopolidosis IV, 252650
MCPH1	148.7	99	98	Microcephaly 1, primary, autosomal recessive, 251200
MECOM	143.3	100	99	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MECP2	87.9	99	93	Encephalopathy, neonatal severe, 300673 Mental retardation, X-linked syndromic, Lubs type, 300260 Mental retardation, X-linked, syndromic 13, 300055 Rett syndrome, 312750 Rett syndrome, atypical, 312750 Rett syndrome, preserved speech variant, 312750 {Autism susceptibility, X-linked 3}, 300496
MED12	106.5	98	94	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Opitz-Kaveggia syndrome, 305450
MED13L	134.7	100	99	Mental retardation and distinctive facial features with or without cardiac defects, 616789 Transposition of the great arteries, dextro-looped 1, 608808
MED17	132.7	99	97	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	130.5	98	96	Mental retardation, autosomal recessive 18, 614249
MED25	104.1	99	95	Basel-Vanagait-Smirin-Yosef syndrome, 616449 ?Charcot-Marie-Tooth disease, type 2B2, 605589
MEF2C	137	97	93	Chromosome 5q14.3 deletion syndrome, 613443 Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
MEFV	108.8	94	91	Familial Mediterranean fever, AD, 134610 Familial Mediterranean fever, AR, 249100
MEGF10	154.5	100	99	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399
MEGF8	127.9	99	98	Carpenter syndrome 2, 614976

MEN1	123.4	99	96	Adrenal adenoma, somatic Angiofibroma, somatic Carcinoid tumor of lung Lipoma, somatic Multiple endocrine neoplasia 1, 131100 Parathyroid adenoma, somatic
MEOX1	76.8	96	91	Klippel-Feil syndrome 2, 214300
MERTK	170.1	100	99	Retinitis pigmentosa 38, 613862
MESP2	81.8	93	87	Spondylocostal dysostosis 2, autosomal recessive, 608681
MET	184.5	100	99	Hepatocellular carcinoma, childhood type, somatic, 114550 Renal cell carcinoma, papillary, 1, familial and somatic, 605074 ?Deafness, autosomal recessive 97, 616705 {Osteofibrous dysplasia,susceptibility to},607278
METTL23	145	100	100	Mental retardation, autosomal recessive 44, 615942
MFAP5	126.7	100	99	Aortic aneurysm, familial thoracic 9, 616166
MFN2	150.9	100	99	Charcot-Marie-Tooth disease, type 2A2A, 609260 Charcot-Marie-Tooth disease, type 2A2B, 617087 Hereditary motor and sensory neuropathy VIA, 601152
MFRP	121.5	100	100	Microphthalmia, isolated 5, 611040 Nanophthalmos 2, 609549
MFSD2A	115.3	98	95	Microcephaly 15, primary, autosomal recessive, 616486
MFSD8	124.8	99	98	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170
MGAT2	157.1	100	99	Congenital disorder of glycosylation, type IIa, 212066
MGME1	151.3	100	100	Mitochondrial DNA depletion syndrome 11, 615084
MGP	131.8	92	91	Keutel syndrome, 245150
MIB1	141.7	100	99	Left ventricular noncompaction 7, 615092
MICU1	134.1	96	89	Myopathy with extrapyramidal signs, 615673
MID1	176.5	99	98	Opitz GBBB syndrome, type I, 300000
MINPP1	147.8	98	94	Thyroid carcinoma, follicular, 188470
MIP	132.1	99	96	Cataract 15, multiple types, 615274
MIR17HG	NC	NC	NC	Feingold syndrome 2, 614326
MIR184	NC	NC	NC	EDICT syndrome, 614303
MIR96	NC	NC	NC	Deafness, autosomal dominant 50, 613074

MITF	155.2	100	99	Tietz albinism-deafness syndrome, 103500 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456
MKKS	225.1	89	89	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKL1	102.2	97	92	Megakaryoblastic leukemia, acute
MKRN3	162.2	100	99	Precocious puberty, central, 2, 615346
MKS1	115.6	99	98	Bardet-Biedl syndrome 13, 615990 Meckel syndrome 1, 249000
MLC1	103.6	100	99	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MLH1	162.3	100	99	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MLH3	155.3	100	100	Colorectal cancer, hereditary nonpolyposis, type 7, 614385 Colorectal cancer, somatic, 114500 {Endometrial cancer, susceptibility to}, 608089
MLLT10	142	94	91	Leukemia,acute myeloid,601626
MLLT11	92.2	100	99	Leukemia, acute myelomonocytic, somatic, 607785
MLPH	95.2	99	95	Griscelli syndrome, type 3, 609227
MLYCD	75.8	91	86	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	185.5	100	99	Methylmalonic aciduria, vitamin B12-responsive, 251100
MMAB	99.6	100	99	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110
MMACHC	206.1	100	100	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	77	89	74	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410
MME	100.6	98	93	Charcot-Marie-Tooth disease, axonal, type 2T, 617017 ?Spinocerebellar ataxia 43, 617018
MMP1	163.5	100	98	COPD, rate of decline of lung function in, 606963 {Epidermolysis bullosa dystrophica, autosomal recessive, modifier of}, 226600
MMP13	123.6	93	91	Metaphyseal anadysplasia 1, 602111 Spondyloepimetaphyseal dysplasia, Missouri type, 602111

MMP19	129.1	99	97	Cavitary optic disc anomalies, 611543
MMP2	164.8	100	100	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP20	100.5	100	98	Amelogenesis imperfecta, type IIA2, 612529
MMP21	93.4	90	84	Heterotaxy, visceral, 7, autosomal, 616749
MMP9	125.6	96	91	Metaphyseal anadysplasia 2, 613073
MN1	93.8	98	93	Meningioma, 607174
MNX1	29.3	63	51	Currarino syndrome, 176450
MOCOS	169.1	99	96	Xanthinuria, type II, 603592
MOCS1	87.4	98	93	Molybdenum cofactor deficiency A, 252150
MOCS2	139.5	99	99	Molybdenum cofactor deficiency B, 252160
MOGS	121.7	99	99	Congenital disorder of glycosylation, type IIb, 606056
MORC2	136.1	100	99	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688
MPC1	122.4	100	100	Mitochondrial pyruvate carrier deficiency, 614741
MPDU1	112	100	99	Congenital disorder of glycosylation, type If, 609180
MPDZ	148.9	98	96	Hydrocephalus, nonsyndromic, autosomal recessive 2, 615219
MPI	146.3	100	100	Congenital disorder of glycosylation, type Ib, 602579
MPL	135.2	99	97	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498
MPLKIP	72.4	97	79	Trichothiodystrophy 4, nonphotosensitive, 234050
MPO	151.1	100	99	Myeloperoxidase deficiency, 254600 {Alzheimer disease, susceptibility to}, 104300 {Lung cancer, protection against, in smokers}
MPV17	108.6	100	99	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MPZ	118.7	99	97	Charcot-Marie-Tooth disease, dominant intermediate D, 607791 Charcot-Marie-Tooth disease, type 1B, 118200 Charcot-Marie-Tooth disease, type 2I, 607677 Charcot-Marie-Tooth disease, type 2J, 607736 Dejerine-Sottas disease, 145900 Neuropathy, congenital hypomyelinating, 605253 Roussy-Levy syndrome, 180800
MRAP	161.8	100	100	Glucocorticoid deficiency 2, 607398
MRE11A	51.2	95	82	Ataxia-telangiectasia-like disorder, 604391
MRPL3	66.3	91	77	Combined oxidative phosphorylation deficiency 9, 614582



MRPS16	156.3	100	99	Combined oxidative phosphorylation deficiency 2, 610498
MRPS22	138.7	95	91	Combined oxidative phosphorylation deficiency 5, 611719
MS4A1	123.6	99	96	Immunodeficiency, common variable, 5, 613495
MSH2	113.4	98	93	Colorectal cancer, hereditary nonpolyposis, type 1, 120435 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MSH3	113.4	98	95	Endometrial carcinoma, somatic, 608089 Familial adenomatous polyposis 4,617100
MSH6	171.5	100	99	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 Endometrial cancer, familial, 608089 Mismatch repair cancer syndrome, 276300
MSMO1	45.6	92	78	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MSR1	170.9	100	99	Barrett esophagus/esophageal adenocarcinoma, 614266 Prostate cancer, hereditary, 176807
MSRB3	155.7	99	98	Deafness, autosomal recessive 74, 613718
MSTN	161.2	100	99	Muscle hypertrophy, 614160
MSX1	75.1	95	87	Ectodermal dysplasia 3, Witkop type, 189500 Orofacial cleft 5, 608874 Tooth agenesis, selective, 1, with or without orofacial cleft, 106600
MSX2	94.5	98	85	Craniosynostosis, type 2, 604757 Parietal foramina 1, 168500 Parietal foramina with cleidocranial dysplasia, 168550
MTAP	109.2	93	86	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250
MTFMT	124.7	99	96	Combined oxidative phosphorylation deficiency 15, 614947
MTHFR	131.3	100	100	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}
MTM1	92.6	98	91	Myotubular myopathy, X-linked, 310400
MTMR2	106.5	100	99	Charcot-Marie-Tooth disease, type 4B1, 601382
MTO1	173.8	89	87	Combined oxidative phosphorylation deficiency 10, 614702
MTOR	140.2	100	99	Smith-Kingsmore syndrome, 616638
MTPAP	107.7	99	93	Ataxia, spastic, 4, 613672

MTR	139.9	99	98	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTRR	139.2	100	99	Homocystinuria-megaloblastic anemia, cbl E type, 236270 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTTP	132.3	99	98	Abetalipoproteinemia, 200100 {Metabolic syndrome, protection against}, 605552
MUC1	106.2	96	92	Medullary cystic kidney disease 1, 174000
MUSK	158.5	100	100	Fetal akinesia deformation sequence, 208150 Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325
MUT	121.6	99	95	Methylmalonic aciduria, mut(0) type, 251000
MUTYH	165.2	100	99	Adenomas, multiple colorectal, 608456 Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600 Gastric cancer, somatic, 613659
MVD	101.5	100	99	Porokeratosis 7, multiple types, 614714
MVK	136.7	100	99	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900
MXI1	108.3	90	83	Neurofibrosarcoma {Prostate cancer, susceptibility to}, 176807
MYBPC1	150.8	99	99	Arthrogyrosis, distal, type 1B, 614335 Lethal congenital contracture syndrome 4, 614915
MYBPC3	142.8	98	95	Cardiomyopathy, dilated, 1MM, 615396 Cardiomyopathy, hypertrophic, 4, 115197 Left ventricular noncompaction 10, 615396
MYC	189.2	100	100	Burkitt lymphoma, 113970
MYCN	99.2	94	83	Feingold syndrome, 164280
MYD88	186.8	100	99	Macroglobulinemia, Waldenstrom, somatic, 153600 Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260
MYF6	121.6	100	100	Myopathy, centronuclear, 3, 614408
MYH11	132.8	100	99	Aortic aneurysm, familial thoracic 4, 132900
MYH14	102.1	97	91	Deafness, autosomal dominant 4A, 600652 ?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369
MYH2	129.4	99	98	Proximal myopathy and ophthalmoplegia, 605637

MYH3	110.4	99	98	Arthrogryposis, distal, type 2A, 193700 Arthrogryposis, distal, type 2B, 601680 Arthrogryposis, distal, type 8, 178110
MYH6	113.5	99	96	Atrial septal defect 3, 614089 Cardiomyopathy, dilated, 1EE, 613252 Cardiomyopathy, hypertrophic, 14, 613251 {Sick sinus syndrome 3}, 614090
MYH7	111.5	99	96	Cardiomyopathy, dilated, 1S, 613426 Cardiomyopathy, hypertrophic, 1, 192600 Left ventricular noncompaction 5, 613426 Liang distal myopathy, 160500 Myopathy, myosin storage, autosomal dominant, 608358 Myopathy, myosin storage, autosomal recessive, 255160 Scapuloperoneal syndrome, myopathic type, 181430
MYH8	134.9	100	99	Carney complex variant, 608837 Trismus-pseudocamptodactyly syndrome, 158300
MYH9	130.8	99	98	Deafness, autosomal dominant 17, 603622 Epstein syndrome, 153650 Fechtner syndrome, 153640 Macrothrombocytopenia and progressive sensorineural deafness, 600208 May-Hegglin anomaly, 155100 Sebastian syndrome, 605249
MYL2	134.9	98	90	Cardiomyopathy, hypertrophic, 10, 608758
MYL3	103.2	100	100	Cardiomyopathy, hypertrophic, 8, 608751
MYLK	148.6	99	99	Aortic aneurysm, familial thoracic 7, 613780
MYLK2	120.3	100	100	Cardiomyopathy, hypertrophic, 1, digenic, 192600
MYO15A	116.7	97	94	Deafness, autosomal recessive 3, 600316
MYO18B	132.7	100	99	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549
MYO1E	132.1	98	97	Glomerulosclerosis, focal segmental, 6, 614131
MYO3A	119.7	98	92	Deafness, autosomal recessive 30, 607101
MYO5A	124.8	99	97	Griscelli syndrome, type 1, 214450
MYO5B	137.8	98	95	Microvillus inclusion disease, 251850
MYO6	89.7	98	92	Deafness, autosomal dominant 22, 606346 Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346 Deafness, autosomal recessive 37, 607821

MYO7A	135	99	98	Deafness, autosomal dominant 11, 601317 Deafness, autosomal recessive 2, 600060 Usher syndrome, type 1B, 276900
MYOC	172.5	100	99	Glaucoma 1A, primary open angle, 137750
MYOT	139.4	99	95	Muscular dystrophy, limb-girdle, type 1A, 159000 Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
MYOZ2	146	100	100	Cardiomyopathy, hypertrophic, 16, 613838
MYPN	142.6	99	98	Cardiomyopathy, dilated, 1KK, 615248 Cardiomyopathy, familial restrictive, 4, 615248 Cardiomyopathy, hypertrophic, 22, 615248
MYT1L	178.5	100	99	Mental retardation, autosomal dominant 39, 616521
NAA10	103.3	98	96	Ogden syndrome, 300855 ?Microphthalmia, syndromic 1, 309800
NAGA	139.6	100	100	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241
NAGLU	108.9	92	90	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491
NAGS	81.2	97	91	N-acetylglutamate synthase deficiency, 237310
NALCN	139	99	97	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419
NANOS1	28.1	87	64	Spermatogenic failure 12, 615413
NANS	106.1	100	99	Sponyloepimetaphyseal dysplasia, Genevieve type, 610442
NARS2	120	97	97	Combined oxidative phosphorylation deficiency 24, 616239
NBAS	145.3	99	97	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NBEAL2	173	99	99	Gray platelet syndrome, 139090
NBN	80.5	99	94	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260
NCF1	23.8	25	22	Chronic granulomatous disease due to deficiency of NCF-1, 233700
NCF2	124.3	100	99	Chronic granulomatous disease due to deficiency of NCF-2, 233710
NCF4	158.9	100	100	?Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III, 613960

NCSTN	112.1	100	99	Acne inversa, familial, 1, 142690
NDE1	103.4	100	99	Lissencephaly 4 (with microcephaly), 614019 ?Microhydranencephaly, 605013
NDN	74.5	92	79	Prader-Willi syndrome, 176270
NDP	117.4	100	100	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600
NDRG1	128.7	99	98	Charcot-Marie-Tooth disease, type 4D, 601455
NDST1	201.7	100	100	Mental retardation, autosomal recessive 46, 616116
NDUFA1	167.6	100	99	Mitochondrial complex I deficiency, 252010
NDUFA11	87.1	99	95	Mitochondrial complex I deficiency, 252010
NDUFA12	159.7	100	100	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000
NDUFA2	134.2	100	100	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFA9	124.8	98	93	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFAF1	115.7	100	100	Mitochondrial complex I deficiency, 252010
NDUFAF2	58.6	85	70	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFAF3	121.1	100	100	Mitochondrial complex I deficiency, 252010
NDUFAF4	79.5	98	91	Mitochondrial complex I deficiency, 252010
NDUFAF5	95.8	98	94	Mitochondrial complex 1 deficiency, 252010
NDUFAF6	80.6	97	86	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFB11	110.4	94	88	Linear skin defects with multiple congenital anomalies 3, 300952
NDUFB3	22.6	91	59	Mitochondrial complex I deficiency, 252010
NDUFS1	132.1	99	98	Mitochondrial complex I deficiency, 252010
NDUFS2	117.8	100	100	Mitochondrial complex I deficiency, 252010
NDUFS3	142.6	90	90	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
NDUFS4	147.2	100	99	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFS6	119.1	99	99	Mitochondrial complex I deficiency, 252010
NDUFS7	118.5	100	99	Leigh syndrome, 256000
NDUFS8	141.6	100	99	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFV1	137	99	97	Mitochondrial complex I deficiency, 252010
NDUFV2	69.3	78	53	Mitochondrial complex I deficiency, 252010
NEB	124	82	81	Nemaline myopathy 2, autosomal recessive, 256030

NEFH	111.7	96	87	Charcot-Marie-Tooth disease, axonal, type 2CC, 616924 ?{Amyotrophic lateral sclerosis, susceptibility to}, 105400
NEFL	165	99	98	Charcot-Marie-Tooth disease, type 1F, 607734 Charcot-Marie-Tooth disease, type 2E, 607684
NEK1	103	98	93	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NEK9	136.8	99	98	Lethal congenital contracture syndrome 10, 617022 Nevus comedonicus, somatic, 617025 ?Arthrogyriposis, Perthes disease, and upward gaze palsy, 614262
NEU1	18.5	12	12	Sialidosis, type I, 256550 Sialidosis, type II, 256550
NEUROD1	166.9	100	100	Maturity-onset diabetes of the young 6, 606394 {Diabetes mellitus, noninsulin-dependent}, 125853
NEUROG3	119.7	100	99	Diarrhea 4, malabsorptive, congenital, 610370
NEXN	78	92	78	Cardiomyopathy, dilated, 1CC, 613122 Cardiomyopathy, hypertrophic, 20, 613876
NF1	129.8	93	90	Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321 Watson syndrome, 193520
NF2	100.3	100	99	Meningioma, NF2-related, somatic, 607174 Neurofibromatosis, type 2, 101000 Schwannomatosis, 162091
NFIX	165.5	97	94	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753
NFKB1	105.4	99	96	Immunodeficiency, common variable, 12, 616576
NFKB2	123.4	97	92	Immunodeficiency, common variable, 10, 615577
NFKBIA	116.5	98	93	Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency, 612132
NFU1	47.7	94	77	Multiple mitochondrial dysfunctions syndrome 1, 605711
NGF	258.2	100	100	Neuropathy, hereditary sensory and autonomic, type V, 608654
NGLY1	128	100	99	Congenital disorder of deglycosylation, 615273
NHEJ1	80.4	100	99	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
NHLRC1	174.1	100	100	Epilepsy, progressive myoclonic 2B (Lafora), 254780

NHP2	111.1	100	100	Dyskeratosis congenita, autosomal recessive 2, 613987
NHS	127.9	94	93	Cataract 40, X-linked, 302200 Nance-Horan syndrome, 302350
NIN	144.4	99	98	Seckel syndrome 7, 614851
NIPA1	174.6	99	99	Spastic paraplegia 6, autosomal dominant, 600363
NIPAL4	158.2	99	93	Ichthyosis, congenital, autosomal recessive 6, 612281
NIPBL	116	96	94	Cornelia de Lange syndrome 1, 122470
NKX2-1	52	96	83	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 {Thyroid cancer, monomedullary, 1}, 188550
NKX2-5	83.2	100	99	Atrial septal defect 7, with or without AV conduction defects, 108900 Conotruncal heart malformations, variable, 217095 Hypoplastic left heart syndrome 2, 614435 Hypothyroidism, congenital nongoitrous, 5, 225250 Tetrology of Fallot, 187500 Ventricular septal defect 3, 614432
NKX2-6	104	100	99	Conotruncal heart malformations, 217095 Persistent truncus arteriosus, 217095
NKX3-2	55.7	92	73	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330
NLGN4X	194.9	99	97	Mental retardation, X-linked, 300495 {Asperger syndrome susceptibility, X-linked 2}, 300497 {Autism susceptibility, X-linked 2}, 300495
NLRC4	180	100	99	Autoinflammation with infantile enterocolitis, 616050 ?Familial cold autoinflammatory syndrome 4, 616115
NLRP12	165.4	99	99	Familial cold autoinflammatory syndrome 2, 611762
NLRP3	150.5	100	100	CINCA syndrome, 607115 Familial cold-induced inflammatory syndrome 1, 120100 Muckle-Wells syndrome, 191900
NLRP7	135	99	98	Hydatidiform mole, recurrent, 1,231090
NME1	104.3	99	99	Neuroblastoma, 256700
NME8	105.7	97	91	Ciliary dyskinesia, primary, 6, 610852
NMNAT1	137.9	100	99	Leber congenital amaurosis 9, 608553
NNT	137	98	97	Glucocorticoid deficiency 4, 614736
NOBOX	79.7	99	97	Premature ovarian failure 5, 611548

NOD2	136	100	99	Blau syndrome, 186580 Yao syndrome, 617321 {Inflammatory bowel disease 1}, 266600 {Psoriatic arthritis, susceptibility to}, 607507
NODAL	160.9	100	99	Heterotaxy, visceral, 5, 270100
NOG	192.4	100	100	Brachydactyly, type B2, 611377 Multiple synostoses syndrome 1, 186500 Stapes ankylosis with broad thumb and toes, 184460 Symphalangism, proximal, 1A, 185800 Tarsal-carpal coalition syndrome, 186570
NOL3	76.2	93	83	Myoclonus, familial cortical, 614937
NONO	94.4	99	96	Mental retardation, X-linked, syndromic 34, 300967
NOP10	160.6	100	100	Dyskeratosis congenita, autosomal recessive 1, 224230
NOP56	137.4	99	98	Spinocerebellar ataxia 36, 614153
NOTCH1	137.8	99	98	Adams-Oliver syndrome 5, 616028 Aortic valve disease 1, 109730
NOTCH2	172.7	100	99	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NOTCH3	110.8	93	88	Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310 Lateral meningocele syndrome, 130720 ?Myofibromatosis, infantile 2, 615293
NPC1	148.1	99	97	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220 {Nasopharyngeal carcinoma 1}
NPC2	140.9	100	99	Niemann-pick disease, type C2, 607625
NPHP1	117.6	98	96	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NPHP3	114.3	99	95	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540
NPHP4	137.3	99	99	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
NPHS1	102.2	99	98	Nephrotic syndrome, type 1, 256300



NPHS2	117	99	95	Nephrotic syndrome, type 2, 600995
NPM1	70.1	88	77	Leukemia, acute myeloid, somatic, 601626
NPPA	114.8	100	100	Atrial fibrillation, familial, 6, 612201 Atrial standstill 2, 615745
NPR2	165	100	100	Acromesomelic dysplasia, Maroteaux type, 602875 Epiphyseal chondrodysplasia, Miura type, 615923 Short stature with nonspecific skeletal abnormalities, 616255
NR0B1	120.1	99	98	46XY sex reversal 2, dosage-sensitive, 300018 Adrenal hypoplasia, congenital, with hypogonadotropic hypogonadism, 300200
NR0B2	96.9	100	99	Obesity, mild, early-onset, 601665
NR1H4	142	96	92	Cholestasis, progressive familial intrahepatic 5, 617049
NR2E3	99.8	100	99	Enhanced S-cone syndrome, 268100 Retinitis pigmentosa 37, 611131
NR2F1	201.8	99	98	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NR2F2	246.2	98	94	Congenital heart defects, multiple types, 4, 615779
NR3C1	137.2	100	99	Glucocorticoid resistance, 615962
NR3C2	159.7	99	95	Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115 Pseudohypoaldosteronism type I, autosomal dominant, 177735
NR4A3	112.9	99	98	Chondrosarcoma, extraskeletal myxoid, 612237
NR5A1	80	100	98	46XY sex reversal 3, 612965 Adrenocortical insufficiency Premature ovarian failure 7, 612964 Spermatogenic failure 8, 613957
NRAS	188.2	100	100	Colorectal cancer, somatic, 114500 Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Thyroid carcinoma, follicular, somatic, 188470 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470
NRL	69.1	99	95	Retinal degeneration, autosomal recessive, clumped pigment type Retinitis pigmentosa 27, 613750
NRXN1	166.2	99	97	Pitt-Hopkins-like syndrome 2, 614325

				{Schizophrenia, susceptibility to, 17}, 614332
NSD1	155.4	100	100	Beckwith-Wiedemann syndrome, 130650 Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550
NSDHL	170.4	99	98	CHILD syndrome, 308050 CK syndrome, 300831
NSMF	94.8	96	95	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
NSUN2	114.9	95	92	Mental retardation, autosomal recessive 5, 611091
NT5C2	124.7	97	92	Spastic paraplegia 45, autosomal recessive, 613162
NT5C3A	62.1	89	78	Anemia, hemolytic, due to UMPH1 deficiency, 266120
NT5E	166.4	100	99	Calcification of joints and arteries, 211800
NTF4	66.7	94	79	Glaucoma 1, open angle, 10, 613100
NTHL1	98.6	99	95	Familial adenomatous polyposis 3, 616415
NTRK1	130.9	99	97	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma, familial, 155240
NUBPL	89.6	92	85	Mitochondrial complex I deficiency, 252010
NUMA1	127.1	100	99	Leukemia, acute promyelocytic, somatic, 612376
NUP107	122.9	99	94	Nephrotic syndrome, type 11, 616730
NUP214	167.5	99	99	Leukemia, acute myeloid, somatic, 601626 Leukemia, T-cell acute lymphoblastic, somatic, 613065
NUP62	111.6	100	99	Striatonigral degeneration, infantile, 271930
NUP93	140.8	97	94	Nephrotic syndrome, type 12, 616892
NYX	95.5	98	96	Night blindness, congenital stationary (complete), 1A, X-linked, 310500
OAT	89.2	77	70	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OBSL1	140.8	99	98	3-M syndrome 2, 612921
OCA2	139.8	99	97	Albinism, brown oculocutaneous, 203200 Albinism, oculocutaneous, type II, 203200 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220
OCLN	220.8	100	100	Band-like calcification with simplified gyration and polymicrogyria, 251290
OCRL	123	98	96	Dent disease 2, 300555 Lowe syndrome, 309000

OFD1	53	84	68	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424
OGG1	128.7	100	99	Renal cell carcinoma, clear cell, somatic, 144700
OPA1	122.5	99	94	Behr syndrome, 210000 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896 {Glaucoma, normal tension, susceptibility to}, 606657
OPA3	128.2	99	97	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPHN1	89.7	99	96	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
OPLAH	109.6	99	97	5-oxoprolinase deficiency, 260005
OPN1LW	77.5	67	62	Blue cone monochromacy, 303700 Colorblindness, protan, 303900
OPN1MW	67.6	68	60	Blue cone monochromacy, 303700 Colorblindness, deutan, 303800
OPN1SW	130.5	100	100	Colorblindness, tritan, 190900
OPTN	113.9	100	99	Amyotrophic lateral sclerosis 12, 613435 Glaucoma 1, open angle, E, 137760 {Glaucoma, normal tension, susceptibility to}, 606657
ORAI1	226.8	92	89	Immunodeficiency 9, 612782 Myopathy, tubular aggregate, 2, 615883
ORC1	106.7	99	98	Meier-Gorlin syndrome 1, 224690
ORC4	57.5	95	82	Meier-Gorlin syndrome 2, 613800
ORC6	126.6	100	100	Meier-Gorlin syndrome 3, 613803
OSBPL2	145.9	100	100	Deafness, autosomal dominant 67, 616340
OSMR	146	100	99	Amyloidosis, primary localized cutaneous, 1, 105250
OSTM1	80.8	90	88	Osteopetrosis, autosomal recessive 5, 259720
OTC	124.1	99	99	Ornithine transcarbamylase deficiency, 311250
OTOA	117.5	98	96	Deafness, autosomal recessive 22, 607039
OTOF	131.4	100	99	Auditory neuropathy, autosomal recessive, 1, 601071 Deafness, autosomal recessive 9, 601071

OTOG	146.3	99	98	Deafness, autosomal recessive 18B, 614945
OTOGL	113.3	98	93	Deafness, autosomal recessive 84B, 614944
OTX2	155.1	100	99	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125
OVOL2	119.1	97	90	Corneal dystrophy, posterior polymorphous, 1, 122000
OXCT1	121.2	99	97	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050
P2RX1	115.3	100	99	Bleeding disorder due to P2RX1 defect, somatic, 609821
P2RX2	130.5	98	95	Deafness, autosomal dominant 41, 608224
P2RY12	185.8	100	100	Bleeding disorder, platelet-type, 8, 609821
P4HB	105.3	94	94	Cole-Carpenter syndrome 1, 112240
PABPN1	66.6	62	60	Oculopharyngeal muscular dystrophy, 164300
PACS1	117.5	97	95	Schuss-Hoeijmakers-syndrome, 615009
PAFAH1B1	105	89	81	Lissencephaly 1, 607432 Subcortical laminar heterotopia, 607432
PAH	151.8	100	100	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600
PAK3	83.3	97	92	Mental retardation, X-linked 30/47, 300558
PALB2	152.6	100	99	Fanconi anemia, complementation group N, 610832 {Breast cancer, susceptibility to}, 114480 {Pancreatic cancer, susceptibility to, 3}, 613348
PAM16	50.8	65	64	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320
PANK2	146.6	99	93	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PAPSS2	108.4	99	98	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847
PARK2	123.7	99	98	Adenocarcinoma of lung, somatic, 211980 Adenocarcinoma, ovarian, somatic, 167000 Parkinson disease, juvenile, type 2, 600116 {Leprosy, susceptibility to}, 607572
PARK7	86.9	100	99	Parkinson disease 7, autosomal recessive early-onset, 606324
PARN	128.3	99	98	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
PAX2	168.6	99	99	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330

PAX3	118.5	100	99	Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220 Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820
PAX4	98.6	100	99	Diabetes mellitus, type 2, 125853 Maturity-onset diabetes of the young, type IX, 612225 {Diabetes mellitus, ketosis-prone, susceptibility to}, 612227
PAX6	121.9	100	100	Aniridia, 106210 Cataract with late-onset corneal dystrophy, 106210 Coloboma of optic nerve, 120430 Coloboma, ocular, 120200 Foveal hypoplasia 1, 136520 Keratitis, 148190 Optic nerve hypoplasia, 165550 Peters anomaly, 604229 ?Morning glory disc anomaly, 120430
PAX7	117.9	100	100	Rhabdomyosarcoma 2, alveolar, 268220
PAX8	94.3	100	99	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700
PAX9	239	99	99	Tooth agenesis, selective, 3, 604625
PC	149.6	97	94	Pyruvate carboxylase deficiency, 266150
PCBD1	113.7	99	99	Hyperphenylalaninemia, BH4-deficient, D, 264070
PCCA	103.1	96	89	Propionicacidemia, 606054
PCCB	129.9	98	96	Propionicacidemia, 606054
PCDH15	155	99	98	Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1F, 602083
PCDH19	225.8	100	99	Epileptic encephalopathy, early infantile, 9, 300088
PCNT	117.7	99	96	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PCSK1	147.1	100	99	Obesity with impaired prohormone processing, 600955 {Obesity, susceptibility to, BMIQ12}, 612362
PCSK9	99.3	94	91	Hypercholesterolemia, familial, 3, 603776 {Low density lipoprotein cholesterol level QTL 1}, 603776
PCYT1A	113.6	98	94	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDCD10	85.1	96	87	Cerebral cavernous malformations 3, 603285

PDE10A	147	99	99	Dyskinesia,limb and orofacial,infantile-onset,616921 Striatal degeneration,autosomal dominant,616922
PDE11A	169	99	98	Pigmented nodular adrenocortical disease, primary, 2, 610475
PDE3A	124.4	99	98	Hypertension and brachydactyly syndrome, 112410
PDE4D	106	96	92	Acrodysostosis 2, with or without hormone resistance, 614613 {Stroke, susceptibility to, 1}, 606799
PDE6A	126	100	99	Retinitis pigmentosa 43, 613810
PDE6B	148.1	100	100	Night blindness, congenital stationary, autosomal dominant 2, 163500 Retinitis pigmentosa-40, 613801
PDE6C	137	99	96	Cone dystrophy 4, 613093
PDE6G	95.6	99	96	Retinitis pigmentosa 57, 613582
PDE6H	68.4	97	77	Achromatopsia 6, 610024 Retinal cone dystrophy 3, 610024
PDE8B	111	99	98	Pigmented nodular adrenocortical disease, primary, 3, 614190 Striatal degeneration, autosomal dominant, 609161
PDGFB	95.2	100	100	Basal ganglia calcification, idiopathic, 5, 615483 Dermatofibrosarcoma protuberans, 607907 Meningioma, SIS-related, 607174
PDGFRA	148.4	100	100	Gastrointestinal stromal tumor, somatic, 606764 Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685
PDGFRB	147.2	99	96	Basal ganglia calcification, idiopathic, 4, 615007 Kosaki overgrowth syndrome, 616592 Myeloproliferative disorder with eosinophilia, 131440 Myofibromatosis, infantile, 1, 228550 Premature aging syndrome, Penttinen type, 601812
PDGFRL	158.7	100	100	Colorectal cancer, somatic, 114500 Hepatocellular cancer, somatic, 114550
PDHA1	110.5	98	92	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	133	99	96	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	132.5	98	94	Lacticacidemia due to PDX1 deficiency,245349
PDP1	209.6	100	100	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	117.2	88	78	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	126.9	96	93	Coenzyme Q10 deficiency, primary, 3, 614652

PDX1	35.3	89	72	MODY, type IV, 606392 Pancreatic agenesis 1, 260370 {Diabetes mellitus, type II, susceptibility to}, 125853
PDYN	107.1	100	99	Spinocerebellar ataxia 23, 610245
PDZD7	96.7	100	98	Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472 {Retinal disease in Usher syndrome type IIA, modifier of}, 276901
PEPD	116.3	99	98	Prolidase deficiency, 170100
PER2	95.3	100	99	Advanced sleep phase syndrome, familial, 1, 604348
PET100	94.5	88	74	Mitochondrial complex IV deficiency, 220110
PEX1	115.7	97	95	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	112.1	96	90	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX11B	106.1	99	98	Peroxisome biogenesis disorder 14B, 614920
PEX12	168.4	100	100	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	197.2	99	98	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	149.2	99	97	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	137.3	97	93	Peroxisome biogenesis disorder 8A, (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	93	99	99	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	147	100	100	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	76.4	100	99	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	98.1	99	94	Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	111.8	99	98	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	94.6	90	86	Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863

PEX7	113.5	89	82	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PFKM	150.7	100	99	Glycogen storage disease VII, 232800
PFN1	152	100	100	Amyotrophic lateral sclerosis 18, 614808
PGAM2	170.9	100	99	Glycogen storage disease X, 261670
PGAP1	98.1	94	88	Mental retardation, autosomal recessive 42, 615802
PGAP2	162.2	100	100	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGAP3	126.3	98	95	Hyperphosphatasia with mental retardation syndrome 4, 615716
PGK1	54.9	93	81	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	133.7	100	99	Congenital disorder of glycosylation, type It, 614921
PGM3	191.6	99	99	Immunodeficiency 23, 615816
PHEX	123.8	98	96	Hypophosphatemic rickets, X-linked dominant, 307800
PHF6	63.3	92	83	Borjeson-Forssman-Lehmann syndrome, 301900
PHF8	94.7	99	97	Mental retardation syndrome, X-linked, Siderius type, 300263
PHGDH	118.9	100	99	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHKA1	107.4	98	95	Muscle glycogenosis, 300559
PHKA2	109	100	99	Glycogen storage disease, type IXa1, 306000 Glycogen storage disease, type IXa2, 306000
PHKB	130.4	99	97	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750
PHKG2	159.3	100	100	Cirrhosis due to liver phosphorylase kinase deficiency Glycogen storage disease IXc, 613027
PHOX2A	29.8	59	32	Fibrosis of extraocular muscles, congenital, 2, 602078
PHOX2B	93	93	87	Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, 209880 Neuroblastoma with Hirschsprung disease, 613013 {Neuroblastoma, susceptibility to, 2}, 613013
PHYH	74.7	97	90	Refsum disease, 266500
PI4KA	112.9	93	89	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531
PICALM	106.1	99	96	Leukemia, acute myeloid, somatic, 601626
PIEZO1	140.3	99	97	Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380 Lymphedema, hereditary, III, 616843



PIEZO2	126.2	99	99	Arthrogryposis, distal, type 3, 114300 Arthrogryposis, distal, type 5, 108145 Arthrogryposis, distal, with impaired proprioception and touch, 617146 ?Marden-Walker syndrome, 248700
PIGA	90.8	90	81	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGG	167.5	100	99	Mental retardation,autosomal recessive 53,616917
PIGL	117.7	100	99	CHIME syndrome, 280000
PIGM	165.5	100	100	Glycosylphosphatidylinositol deficiency, 610293
PIGN	119.7	98	92	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	147.2	100	99	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGT	174.9	100	99	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 ?Paroxysmal nocturnal hemoglobinuria 2, 615399
PIGV	145.7	100	100	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIGY	121.5	100	99	Hyperphosphatasia with mental retardation syndrome 6, 616809
PIK3CA	120.5	99	99	Breast cancer, somatic, 114480 CLOVE syndrome, somatic, 612918 Colorectal cancer, somatic, 114500 Cowden syndrome 5, 615108 Gastric cancer, somatic, 613659 Hepatocellular carcinoma, somatic, 114550 Keratosis, seborrheic, somatic, 182000 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Nevus, epidermal, somatic, 162900 Nonsmall cell lung cancer, somatic, 211980 Ovarian cancer, somatic, 167000
PIK3CD	132.6	99	96	Immunodeficiency 14, 615513
PIK3R1	129.3	99	97	Immunodeficiency 36, 616005 SHORT syndrome, 269880 ?Agammaglobulinemia 7, autosomal recessive, 615214
PIK3R2	87.1	88	86	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PIK3R5	110.3	100	99	Ataxia-oculomotor apraxia 3, 615217
PIKFYVE	141.5	99	98	Corneal fleck dystrophy, 121850
PINK1	90.4	87	81	Parkinson disease 6, early onset, 605909

PIP5K1C	107.7	96	95	Lethal congenital contractural syndrome 3, 611369
PITPNM3	117.4	99	97	Cone-rod dystrophy 5, 600977
PITX1	145.1	91	86	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800 Liebenberg syndrome, 186550
PITX2	147.9	99	97	Axenfeld-Rieger syndrome, type 1, 180500 Iridogoniodysgenesis, type 2, 137600 Peters anomaly, 604229 Ring dermoid of cornea, 180550
PITX3	40.2	95	82	Anterior segment mesenchymal dysgenesis, 107250 Cataract 11, multiple types, 610623 Cataract 11, syndromic, 610623
PKD1	28.1	42	34	Polycystic kidney disease, adult type I, 173900
PKD2	110.7	89	84	Polycystic kidney disease 2, 613095
PKHD1	155	100	99	Polycystic kidney and hepatic disease, 263200
PKLR	179.1	100	100	Adenosine triphosphate, elevated, of erythrocytes, 102900 Pyruvate kinase deficiency, 266200
PKP1	122.1	99	98	Ectodermal dysplasia/skin fragility syndrome, 604536
PKP2	99.7	94	87	Arrhythmogenic right ventricular dysplasia 9, 609040
PLA2G4A	134.2	99	98	Phospholipase A2, group IV A, deficiency of
PLA2G6	117.6	99	98	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953
PLA2G7	124.4	99	97	Platelet-activating factor acetylhydrolase deficiency, 614278 {Asthma, susceptibility to}, 600807 {Atopy, susceptibility to}, 147050
PLAG1	219.2	100	100	Adenomas, salivary gland pleomorphic, somatic, 181030
PLAU	111.5	99	98	Quebec platelet disorder, 601709 {Alzheimer disease, late-onset, susceptibility to}, 104300
PLCB1	144.6	100	99	Epileptic encephalopathy, early infantile, 12, 613722
PLCB4	126.1	99	95	Auriculocondylar syndrome 2, 614669
PLCD1	117.1	99	97	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCE1	155.4	99	98	Nephrotic syndrome, type 3, 610725
PLCG2	118.9	100	99	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468

PLEC	114.3	99	98	Epidermolysis bullosa simplex with muscular dystrophy, 226670 Epidermolysis bullosa simplex with pyloric atresia, 612138 Epidermolysis bullosa simplex, Ogna type, 131950 Muscular dystrophy, limb-girdle, type 2Q, 613723 ?Epidermolysis bullosa simplex with nail dystrophy, 616487
PLEKHG2	131.9	99	97	Leukodystrophy and acquired microcephaly with or without dystonia, 616763
PLEKHG5	87	96	88	Charcot-Marie-Tooth disease, recessive intermediate C, 615376 Spinal muscular atrophy, distal, autosomal recessive, 4, 611067
PLEKHM1	69.2	49	48	Osteopetrosis, autosomal recessive 6, 611497
PLG	115.4	87	86	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PLIN1	81.2	96	88	Lipodystrophy, familial partial, type 4, 613877
PLK4	145.6	99	96	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLN	209.3	100	100	Cardiomyopathy, dilated, 1P, 609909 Cardiomyopathy, hypertrophic, 18, 613874
PLOD1	134.3	99	97	Ehlers-Danlos syndrome, type VI, 225400
PLOD2	108.5	94	88	Bruck syndrome 2, 609220
PLOD3	100.1	98	96	Lysyl hydroxylase 3 deficiency, 612394
PLP1	130.4	100	99	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PLS3	136.5	99	98	Bone mineral density QTL18, osteoporosis, 300910
PML	124.6	100	99	Leukemia, acute promyelocytic, PML/RARA type
PMM2	141.3	99	99	Congenital disorder of glycosylation, type Ia, 212065
PMP22	111.4	96	91	Charcot-Marie-Tooth disease, type 1A, 118220 Charcot-Marie-Tooth disease, type 1E, 118300 Dejerine-Sottas disease, 145900 Neuropathy, inflammatory demyelinating, 139393 Neuropathy, recurrent, with pressure palsies, 162500 Roussy-Levy syndrome, 180800
PMPCA	120.7	99	96	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMS2	95.2	83	80	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 Mismatch repair cancer syndrome, 276300
PMVK	125.7	100	99	Porokeratosis 1, multiple types, 175800
PNKD	99.9	100	99	Paroxysmal nonkinesigenic dyskinesia, 118800

PNKP	93.1	99	97	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNP	151.2	100	99	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA1	171.9	100	99	Ichthyosis, congenital, autosomal recessive 10, 615024
PNPLA2	113.3	99	97	Neutral lipid storage disease with myopathy, 610717
PNPLA6	122.4	99	98	Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 Spastic paraplegia 39, autosomal recessive, 612020 ?Laurence-Moon syndrome, 245800
PNPO	66.5	100	98	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
PNPT1	53.6	93	80	Combined oxidative phosphorylation deficiency 13, 614932 Deafness, autosomal recessive 70, 614934
POC1A	134.1	100	100	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POC1B	77.6	96	92	Cone-rod dystrophy 20, 615973
POF1B	75.4	91	81	Premature ovarian failure 2B,300604
POFUT1	139.6	99	97	Dowling-Degos disease 2, 615327
POGLUT1	117.5	98	94	Dowling-Degos disease 4, 615696
POGZ	168.2	99	99	White-Sutton syndrome, 616364
POLA1	111.4	98	92	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220
POLD1	101.3	93	90	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381 {Colorectal cancer, susceptibility to, 10}, 612591
POLE	144.3	100	99	FILS syndrome, 615139 {Colorectal cancer, susceptibility to, 12}, 615083
POLG	114.4	100	99	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
POLG2	157.6	98	96	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131
POLH	140.7	100	99	Xeroderma pigmentosum, variant type, 278750
POLR1A	120.2	99	99	Acrofacial dysostosis, Cincinnati type, 616462
POLR1C	117.2	99	96	Leukodystrophy, hypomyelinating, 11, 616494 Treacher Collins syndrome 3, 248390
POLR1D	192	100	100	Treacher Collins syndrome 2, 613717

POLR3A	137.8	100	99	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	146.4	99	98	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMC	116.3	100	100	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734 {Obesity, early-onset, susceptibility to}, 601665
POMGNT1	126.8	99	97	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157
POMGNT2	259.9	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 8, 614830
POMK	205.3	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249 ?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094
POMP	114.4	95	87	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952
POMT1	156	99	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), type C, 1, 609308
POMT2	111.2	98	97	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158
POR	163	99	98	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571
PORCN	118.7	100	99	Focal dermal hypoplasia, 305600
POU1F1	106.2	98	94	Pituitary hormone deficiency, combined, 1, 613038
POU3F4	152.3	100	99	Deafness, X-linked 2, 304400
POU4F3	298.7	100	100	Deafness, autosomal dominant 15, 602459
PPARG	154	100	99	Carotid intimal medial thickness 1, 609338 Insulin resistance, severe, digenic, 604367 Lipodystrophy, familial partial, type 3, 604367 Obesity, severe, 601665 [Obesity, resistance to] {Diabetes, type 2}, 125853
PPIB	118.7	100	100	Osteogenesis imperfecta, type IX, 259440
PPM1D	168.4	100	99	Breast cancer, 114480
PPOX	96.2	99	98	Porphyria variegata, 176200

PPP1R15B	133.3	99	98	Microcephaly, short stature, and impaired glucose metabolism 2, 616817
PPP1R3A	146.4	99	98	Insulin resistance, severe, digenic, 604367
PPP2R1A	137	93	93	Mental retardation, autosomal dominant 36, 616362
PPP2R1B	155.3	100	99	Lung cancer, 211980
PPP2R2B	142	99	97	Spinocerebellar ataxia 12, 604326
PPP2R5D	143.4	100	99	Mental retardation, autosomal dominant 35, 616355
PPT1	172.9	100	100	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	181.9	97	96	Renpenning syndrome, 309500
PRCC	146.6	99	98	Renal cell carcinoma, papillary, 605074
PRCD	89.4	100	99	Retinitis pigmentosa 36, 610599
PRDM12	112.5	91	87	Neuropathy, hereditary sensory and autonomic, type VIII, 616488
PRDM16	161.5	99	99	Cardiomyopathy, dilated, 1LL, 615373 Left ventricular noncompaction 8, 615373
PRDM5	129.4	99	95	Brittle cornea syndrome 2, 614170
PRDM6	108.2	92	79	Patent ductus arteriosus 3, 617039
PRF1	134.2	100	99	Aplastic anemia, 609135 Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027
PRG4	144.4	97	88	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250
PRICKLE1	117.3	100	100	Epilepsy, progressive myoclonic 1B, 612437
PRIMPOL	110	95	90	Myopia 22, autosomal dominant, 615420
PRKAG2	125.8	98	91	Cardiomyopathy, hypertrophic 6, 600858 Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200
PRKAR1A	90.6	99	93	Acrodysostosis 1, with or without hormone resistance, 101800 Adrenocortical tumor, somatic, Carney complex, type 1, 160980 Myxoma, intracardiac, 255960 Pigmented nodular adrenocortical disease, primary, 1, 610489
PRKCA	150.3	100	100	Pituitary tumor, invasive
PRKCD	181.6	100	99	Autoimmune lymphoproliferative syndrome, type III, 615559
PRKCG	114.5	98	93	Spinocerebellar ataxia 14, 605361
PRKCSH	135.4	99	96	Polycystic liver disease, 174050
PRKDC	106.7	98	94	Immunodeficiency 26, with or without neurologic abnormalities, 615966

PRKG1	123.6	98	95	Aortic aneurysm, familial thoracic 8, 615436
PRKRA	178.7	99	98	Dystonia 16, 612067
PRLR	150	100	99	Multiple fibroadenomas of the breast, 615554 ?Hyperprolactinemia, 615555
PRNP	153.8	100	100	Cerebral amyloid angiopathy, PRNP-related, 137440 Creutzfeldt-Jakob disease, 123400 Gerstmann-Straussler disease, 137440 Huntington disease-like 1, 603218 Insomnia, fatal familial, 600072 Prion disease with protracted course, 606688 {Kuru, susceptibility to}, 245300
PROC	140.4	99	97	Thrombophilia due to protein C deficiency, autosomal dominant, 176860 Thrombophilia due to protein C deficiency, autosomal recessive, 612304
PRODH	84.1	84	82	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850
PROK2	105.6	98	91	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	332	100	100	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROM1	111.5	95	92	Cone-rod dystrophy 12, 612657 Macular dystrophy, retinal, 2, 608051 Retinitis pigmentosa 41, 612095 Stargardt disease 4, 603786
PROP1	76.6	91	84	Pituitary hormone deficiency, combined, 2, 262600
PROS1	101.1	96	91	Thrombophilia due to protein S deficiency, autosomal dominant, 612336 Thrombophilia due to protein S deficiency, autosomal recessive, 614514
PRPF3	86	98	96	Retinitis pigmentosa 18, 601414
PRPF31	116.9	97	91	Retinitis pigmentosa 11, 600138
PRPF4	149.6	100	99	Retinitis pigmentosa 70, 615922
PRPF6	130.9	100	100	Retinitis pigmentosa 60, 613983
PRPF8	139.2	99	99	Retinitis pigmentosa 13, 600059
PRPH2	244.1	100	100	Choroidal dystrophy, central areolar 2, 613105 Leber congenital amaurosis 18, 608133 Macular dystrophy, patterned, 1, 169150 Macular dystrophy, vitelliform, 3, 608161 Retinitis pigmentosa 7 and digenic, 608133

				Retinitis punctata albescens, 136880
PRPS1	150.5	100	100	Arts syndrome, 301835 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Deafness, X-linked 1, 304500 Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661
PRRT2	78.9	99	98	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751
PRRX1	100.2	100	99	Agnathia-otocephaly complex, 202650
PRSS1	191.1	100	99	Pancreatitis, hereditary, 167800 Trypsinogen deficiency, 614044
PRSS12	153.3	99	98	Mental retardation, autosomal recessive 1, 249500
PRSS56	51.5	96	84	Microphthalmia, isolated 6, 613517
PRX	115.8	99	98	Charcot-Marie-Tooth disease, type 4F, 614895 Dejerine-Sottas disease, 145900
PSAP	114.3	99	99	Combined SAP deficiency, 611721 Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PSAT1	53.3	91	75	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992
PSEN1	153.1	99	98	Acne inversa, familial, 3, 613737 Alzheimer disease, type 3, 607822 Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 Cardiomyopathy, dilated, 1U, 613694 Dementia, frontotemporal, 600274 Pick disease, 172700
PSEN2	124.1	100	100	Alzheimer disease-4, 606889



				Cardiomyopathy, dilated, 1V, 613697
PSEENEN	67.4	100	98	Acne inversa, familial, 2, 613736
PSMB8	14.8	12	12	Autoinflammation, lipodystrophy, and dermatosis syndrome, 256040
PSMC3IP	113.2	99	99	Ovarian dysgenesis 3, 614324
PSPH	128.9	98	95	Phosphoserine phosphatase deficiency, 614023
PSTPIP1	88.5	99	97	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416
PTCH1	114.8	98	95	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly-7, 610828
PTCH2	120.4	99	97	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Medulloblastoma, 155255
PTDSS1	127.2	100	100	Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	142.9	99	95	Bannayan-Riley-Ruvalcaba syndrome, 153480 Cowden syndrome 1, 158350 Endometrial carcinoma, somatic, 608089 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309 Malignant melanoma, somatic, 155600 PTEN hamartoma tumor syndrome Squamous cell carcinoma, head and neck, somatic, 275355 VATER association with macrocephaly and ventriculomegaly, 276950 {Glioma susceptibility 2}, 613028 {Meningioma}, 607174 {Prostate cancer, somatic}, 176807
PTF1A	73.8	88	78	Pancreatic agenesis 2, 615935 Pancreatic and cerebellar agenesis, 609069
PTGIS	127	96	94	Hypertension, essential, 145500
PTH	103.3	99	97	Hypoparathyroidism, autosomal dominant, 146200 Hypoparathyroidism, autosomal recessive, 146200
PTH1R	108.6	99	98	Chondrodysplasia, Blomstrand type, 215045 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Metaphyseal chondrodysplasia, Murk Jansen type, 156400

PTHLH	120.4	99	93	Brachydactyly, type E2, 613382 Humoral hypercalcemia of malignancy
PTPN11	103.4	97	92	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
PTPN12	144.6	97	94	Colon cancer, somatic, 114500
PTPN14	176	99	96	Choanal atresia and lymphedema, 613611
PTPRC	101.6	93	86	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971 {Hepatitis C virus, susceptibility to}, 609532
PTPRJ	169.5	97	96	Colon cancer, somatic, 114500
PTPRO	140.8	99	99	Nephrotic syndrome, type 6, 614196
PTPRQ	104.6	93	89	Deafness, autosomal recessive 84A, 613391
PTRF	137.5	99	99	Lipodystrophy, congenital generalized, type 4, 613327
PTRH2	279.8	100	100	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PTS	107.3	99	93	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUF60	173.7	99	98	Verheij syndrome, 615583
PURA	121.9	94	87	Mental retardation, autosomal dominant 31, 616158
PUS1	127.4	98	94	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PVRL1	145.6	100	100	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060
PVRL4	136.1	100	100	Ectodermal dysplasia-syndactyly syndrome 1, 613573
PXDN	164	99	98	Corneal opacification and other ocular anomalies, 269400
PYCR1	86.4	99	94	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
PYCR2	127.8	100	97	Leukodystrophy, hypomyelinating, 10, 616420
PYGL	158.6	100	100	Glycogen storage disease VI, 232700
PYGM	127.3	100	99	McArdle disease, 232600
QARS	165.5	100	100	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	92.6	100	99	Hyperphenylalaninemia, BH4-deficient, C, 261630
RAB18	80.4	96	84	Warburg micro syndrome 3, 614222
RAB23	110.4	99	97	Carpenter syndrome, 201000
RAB27A	144.1	100	99	Griscelli syndrome, type 2, 607624
RAB28	52	96	87	Cone-rod dystrophy 18, 615374

RAB33B	233.7	100	100	Smith-McCort dysplasia 2, 615222
RAB39B	113.5	100	99	Mental retardation, X-linked 72, 300271 ?Waisman syndrome, 311510
RAB3GAP1	124.2	99	98	Warburg micro syndrome 1, 600118
RAB3GAP2	94.2	98	93	Martsolf syndrome, 212720 Warburg micro syndrome 2, 614225
RAB7A	158.2	100	100	Charcot-Marie-Tooth disease,type 2B, 600882
RAC2	104.2	100	99	Neutrophil immunodeficiency syndrome, 608203
RAD21	78.4	98	94	Cornelia de Lange syndrome 4, 614701
RAD50	99	92	86	Nijmegen breakage syndrome-like disorder, 613078
RAD51	123.3	89	89	Mirror movements 2, 614508 {Breast cancer, susceptibility to}, 114480 ?Fanconi anemia, complementation group R, 617244
RAD51C	143.5	100	98	Fanconi anemia, complementation group O, 613390 {Breast-ovarian cancer, familial, susceptibility to, 3}, 613399
RAD54B	104.5	98	93	Colon cancer, somatic, 114500 Lymphoma, non-Hodgkin, somatic, 605027
RAD54L	123.7	100	99	Adenocarcinoma, colonic, somatic Lymphoma, non-Hodgkin, somatic, 605027 {Breast cancer, invasive ductal}, 114480
RAF1	127.4	100	99	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553
RAG1	206.9	100	100	Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889 Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457
RAG2	221.3	100	100	Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457
RAI1	145.4	100	99	Smith-Magenis syndrome, 182290
RAP1GDS1	96.1	99	93	Lymphocytic leukemia, acute T-cell
RAPSN	129.5	99	96	Fetal akinesia deformation sequence, 208150

				Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326
RARB	138.7	100	100	Microphthalmia, syndromic 12, 615524
RARS	91.5	98	90	Leukodystrophy, hypomyelinating, 9, 616140
RARS2	107.1	100	99	Pontocerebellar hypoplasia, type 6, 611523
RASA1	96.9	95	84	Basal cell carcinoma, somatic, 605462 Capillary malformation-arteriovenous malformation, 608354 Parkes Weber syndrome, 608355
RAX	82.9	88	77	Microphthalmia, isolated 3, 611038
RAX2	52.4	90	67	?Macular degeneration, age-related, 6, 613757 Cone-rod dystrophy 11, 610381
RB1	88	90	76	Bladder cancer, somatic, 109800 Osteosarcoma, somatic, 259500 Retinoblastoma, 180200 Retinoblastoma, trilateral, 180200 Small cell cancer of the lung, somatic, 182280
RB1CC1	111.4	96	91	Breast cancer, somatic, 114480
RBBP8	110.8	99	96	Jawad syndrome, 251255 Pancreatic carcinoma, somatic Seckel syndrome 2, 606744
RBCK1	104.3	99	94	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RBM10	118.1	99	97	TARP syndrome, 311900
RBM20	181.2	99	96	Cardiomyopathy, dilated, 1DD, 613172
RBM8A	106.6	100	99	Thrombocytopenia-absent radius syndrome, 274000
RBP4	99.7	99	96	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RBPJ	89.2	94	86	Adams-Oliver syndrome 3, 614814
RD3	161.6	100	99	Leber congenital amaurosis 12, 610612
RDH12	94.5	100	98	Leber congenital amaurosis 13, 612712
RDH5	160.2	100	99	Fundus albipunctatus, 136880
RDX	43.1	84	64	Deafness, autosomal recessive 24, 611022
RECQL4	149.8	99	96	Baller-Gerold syndrome, 218600 RAPADILINO syndrome, 266280 Rothmund-Thomson syndrome, 268400

REEP1	99.6	96	95	Spastic paraplegia 31, autosomal dominant, 610250 ?Neuronopathy, distal hereditary motor, type VB, 614751
RELN	155.6	100	99	Lissencephaly 2 (Norman-Roberts type), 257320 {Epilepsy, familial temporal lobe, 7}, 616436
REN	149.1	100	100	Hyperuricemic nephropathy, familial juvenile 2, 613092 Renal tubular dysgenesis, 267430 [Hyperproreninemia]
RERE	71.4	94	88	Neurodevelopmental disorder with or without anomalies of the brain, eye or heart, 616975
RET	141.3	99	97	Central hypoventilation syndrome, congenital, 209880 Medullary thyroid carcinoma, 155240 Multiple endocrine neoplasia IIA, 171400 Multiple endocrine neoplasia IIB, 162300 Pheochromocytoma, 171300 {Hirschsprung disease, susceptibility to, 1}, 142623
RFT1	108.4	99	97	Congenital disorder of glycosylation, type In, 612015
RFX5	117.1	98	96	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFX6	154	100	99	Mitchell-Riley syndrome, 615710
RFXANK	106.1	100	100	MHC class II deficiency, complementation group B, 209920
RFXAP	84.8	94	91	Bare lymphocyte syndrome, type II, complementation group D, 209920
RGR	126.9	100	99	Retinitis pigmentosa 44, 613769
RGS9	104	100	99	Bradyopsia, 608415
RGS9BP	93.9	100	99	Bradyopsia, 608415
RHAG	152.1	100	99	Anemia, hemolytic, Rh-null, regulator type, 268150 Overhydrated hereditary stomatocytosis, 185000 Rh-mod syndrome
RHBDF2	98	99	97	Tylosis with esophageal cancer, 148500
RHCE	211.3	97	97	Rh-null disease, amorph type [Blood group, Rhesus], 111690
RHO	210.5	100	100	Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 Retinitis punctata albescens, 136880
RIMS1	125.7	98	95	Cone-rod dystrophy 7, 603649
RIN2	113.5	100	99	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075

RIPK4	163.5	100	99	Popliteal pterygium syndrome, Bartsocas-Papas type, 263650
RIT1	165.5	100	100	Noonan syndrome 8, 615355
RLBP1	144.9	100	100	Bothnia retinal dystrophy, 607475 Fundus albipunctatus, 136880 Newfoundland rod-cone dystrophy, 607476 Retinitis punctata albescens, 136880
RLIM	150.4	99	97	Mental Retardation, X-linked 61, 300978
RMND1	137	99	97	Combined oxidative phosphorylation deficiency 11, 614922
RMRP	NC	NC	NC	Anauxetic dysplasia, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RNASEH1	98.5	99	95	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479
RNASEH2A	142.2	100	99	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	103.8	93	87	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	209.6	100	99	Aicardi-Goutieres syndrome 3, 610329
RNASEL	147.9	100	99	Prostate cancer 1, 601518
RNASET2	87.3	90	85	Leukoencephalopathy, cystic, without megalencephaly, 612951
RNF125	178.1	100	99	Tenorio syndrome, 616260
RNF135	96	93	91	Macrocephaly, macrosomia, facial dysmorphism syndrome, 614192
RNF139	223.3	100	100	Renal cell carcinoma, 144700
RNF168	215.5	100	99	RIDDLE syndrome, 611943
RNF170	147.3	98	91	Ataxia, sensory, 1, autosomal dominant, 608984
RNF212	119	99	96	Recombination rate QTL 1, 612042
RNF216	137	99	98	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840
RNF6	195.6	100	99	Esophageal carcinoma, somatic, 133239
RNU4ATAC	NC	NC	NC	Microcephalic osteodysplastic primordial dwarfism, type I, 210710 Roifman syndrome, 616651
ROBO2	155.3	98	96	Vesicoureteral reflux 2, 610878
ROBO3	99.9	98	95	Gaze palsy, horizontal, with progressive scoliosis, 607313
ROGDI	112.1	97	95	Kohlschutter-Tonz syndrome, 226750
ROM1	115.3	100	99	Retinitis pigmentosa 7, digenic, 608133
ROR2	166.2	99	98	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RORC	129.9	100	99	Immunodeficiency 42, 616622

RP1L1	94.9	100	99	Occult macular dystrophy, 613587
RP2	180.9	100	98	Retinitis pigmentosa 2, 312600
RPE65	130.3	100	99	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794
RPGR	92	83	74	Cone-rod dystrophy, X-linked, 1, 304020 Macular degeneration, X-linked atrophic, 300834 Retinitis pigmentosa 3, 300029 Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455
RPGRIP1	153.8	100	99	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826
RPGRIP1L	126.2	96	93	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561
RPL11	100	100	99	Diamond-Blackfan anemia 7, 612562
RPL21	64.6	78	57	Hypotrichosis 12, 615885
RPL35A	84	99	91	Diamond-Blackfan anemia 5, 612528
RPL5	43.7	81	69	Diamond-Blackfan anemia 6, 612561
RPS10	140.1	99	97	Diamond-Blackfan anemia 9, 613308
RPS14	133.5	99	96	Macrocytic anemia, refractory, due to 5q deletion, somatic, 153550
RPS17	52.5	87	74	Diamond-Blackfan anemia 4, 612527
RPS19	82.6	99	95	Diamond-Blackfan anemia 1, 105650
RPS24	111.3	92	87	Diamond-blackfan anemia 3, 610629
RPS26	107	94	82	Diamond-Blackfan anemia 10, 613309
RPS28	47.7	100	92	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164
RPS29	70.6	97	95	Diamond-Blackfan anemia 13, 615909
RPS6KA3	79	94	83	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844
RPS7	93.5	76	63	Diamond-Blackfan anemia 8, 612563
RPSA	88.6	100	99	Asplenia, isolated congenital, 271400
RRAS2	71.7	85	68	Ovarian carcinoma
RRM2B	128.8	99	97	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077
RS1	60.4	97	88	Retinoschisis, 312700

RSPH1	146.2	100	100	Ciliary dyskinesia, primary, 24, 615481
RSPH3	132.6	99	97	Ciliary dyskinesia, primary, 32, 616481
RSPH4A	139.4	98	96	Ciliary dyskinesia, primary, 11, 612649
RSPH9	127.6	100	99	Ciliary dyskinesia, primary, 12, 612650
RSPO1	109.7	100	100	Palmoplantar hyperkeratosis and true hermaphroditism, 610644 Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644
RSPO4	107.5	100	100	Anonychia congenita, 206800
RSPRY1	168.9	100	99	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723
RTEL1	115.2	99	95	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373
RTN2	104.8	99	96	Spastic paraplegia 12, autosomal dominant, 604805
RTN4IP1	98.2	99	99	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
RTTN	130.7	97	95	Microcephaly, short stature, and polymicrogyria with seizures, 614833
RUNX1	91.6	94	86	Leukemia, acute myeloid, 601626 Platelet disorder, familial, with associated myeloid malignancy, 601399
RUNX2	107.1	74	74	Cleidocranial dysplasia, 119600 Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600 Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510
RYR1	120.9	96	93	Central core disease, 117000 King-Denborough syndrome, 145600 Minicore myopathy with external ophthalmoplegia, 255320 Neuromuscular disease, congenital, with uniform type 1 fiber, 117000 {Malignant hyperthermia susceptibility 1}, 145600
RYR2	142.3	99	98	Arrhythmogenic right ventricular dysplasia 2, 600996 Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772
S1PR2	278.5	97	92	Deafness, autosomal recessive 68, 610419
SACS	154.4	100	99	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAG	130.8	100	99	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758
SALL1	138.8	99	98	Townes-Brocks branchiootorenal-like syndrome, 107480 Townes-Brocks syndrome, 107480
SALL4	147.6	97	96	Duane-radial ray syndrome, 607323



				IVIC syndrome, 147750
SAMD9	158.7	99	99	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455
SAMD9L	165.3	100	99	Ataxia-pancytopenia syndrome, 159550
SAMHD1	127.8	99	96	Aicardi-Goutieres syndrome 5, 612952 ?Chilblain lupus 2, 614415
SAR1B	125	89	88	Chylomicron retention disease, 246700
SARS2	106.6	95	93	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SATB2	110.6	98	93	Glass syndrome, 612313
SBDS	212.4	100	99	Shwachman-Diamond syndrome, 260400 {Aplastic anemia, susceptibility to}, 609135
SBF1	108.1	98	96	Charcot-Marie-Tooth disease, type 4B3, 615284
SBF2	116.9	99	96	Charcot-Marie-Tooth disease, type 4B2, 604563
SC5D	198.5	100	99	Lathosterolosis, 607330
SCARB2	120.6	100	99	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCARF2	80.9	85	73	Van den Ende-Gupta syndrome, 600920
SCN10A	165.5	100	99	Episodic pain syndrome, familial, 2, 615551
SCN11A	138	99	97	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN1A	137	99	98	Dravet syndrome, 607208 Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Febrile seizures, familial, 3A, 604403 Migraine, familial hemiplegic, 3, 609634
SCN1B	168.8	97	96	Atrial fibrillation, familial, 13, 615377 Brugada syndrome 5, 612838 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233
SCN2A	156.7	99	96	Epileptic encephalopathy, early infantile, 11, 613721 Seizures, benign familial infantile, 3, 607745
SCN2B	186.2	100	100	Atrial fibrillation, familial, 14, 615378
SCN3B	147.5	100	100	Atrial fibrillation, familial, 16, 613120 Brugada syndrome 7, 613120

SCN4A	214.2	99	99	Hyperkalemic periodic paralysis, type 2, 170500 Hypokalemic periodic paralysis, type 2, 613345 Myasthenic syndrome, congenital, 16, 614198 Myotonia congenita, atypical, acetazolamide-responsive, 608390 Paramyotonia congenita, 168300
SCN4B	77.7	100	97	Atrial fibrillation, familial, 17, 611819 Long QT syndrome-10, 611819
SCN5A	171.4	100	100	Atrial fibrillation, familial, 10, 614022 Brugada syndrome 1, 601144 Cardiomyopathy, dilated, 1E, 601154 Heart block, nonprogressive, 113900 Heart block, progressive, type IA, 113900 Long QT syndrome-3, 603830 Sick sinus syndrome 1, 608567 Ventricular fibrillation, familial, 1, 603829 {Sudden infant death syndrome, susceptibility to}, 272120
SCN8A	198.5	100	99	Epileptic encephalopathy, early infantile, 13, 614558 ?Cognitive impairment with or without cerebellar ataxia, 614306
SCN9A	148.3	98	97	Epilepsy, generalized, with febrile seizures plus, type 7, 613863 Erythralgia, primary, 133020 Febrile seizures, familial, 3B, 613863 HSAN2D, autosomal recessive, 243000 Insensitivity to pain, congenital, 243000 Paroxysmal extreme pain disorder, 167400, Small fiber neuropathy, 133020 {Dravet syndrome, modifier of}, 607208
SCNN1A	130.9	96	93	Bronchiectasis with or without elevated sweat chloride 2, 613021 Pseudohypoaldosteronism, type I, 264350
SCNN1B	149	100	99	Bronchiectasis with or without elevated sweat chloride 1, 211400 Liddle syndrome, 177200 Pseudohypoaldosteronism, type I, 264350
SCNN1G	139.6	99	97	Bronchiectasis with or without elevated sweat chloride 3, 613071 Liddle syndrome, 177200 Pseudohypoaldosteronism, type I, 264350
SCO1	110	98	94	Mitochondrial complex IV deficiency, 220110

SCO2	113.3	100	100	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908
SCP2	106.9	99	96	Leukoencephalopathy with dystonia and motor neuropathy, 613724
SCYL1	143.6	98	96	Spinocerebellar ataxia, autosomal recessive 21, 616719
SDCCAG8	124.4	99	97	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SDHA	122.2	84	80	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Parangliomas 5, 614165
SDHAF1	42.6	100	96	Mitochondrial complex II deficiency, 252011
SDHAF2	144.1	94	94	Parangliomas 2, 601650
SDHB	120.5	100	99	Cowden syndrome 2, 612359 Gastrointestinal stromal tumor, 606764 Paranglioma and gastric stromal sarcoma, 606864 Parangliomas 4, 115310 Pheochromocytoma, 171300
SDHC	100	99	96	Gastrointestinal stromal tumor, 606764 Paranglioma and gastric stromal sarcoma, 606864 Parangliomas 3, 605373
SDHD	48.5	62	57	Carcinoid tumors, intestinal, 114900 Cowden syndrome 3, 615106 Merkel cell carcinoma, somatic Mitochondrial complex II deficiency, 252011 Paranglioma and gastric stromal sarcoma, 606864 Parangliomas 1, with or without deafness, 168000 Pheochromocytoma, 171300
SEC23A	121.5	98	94	Craniolenticulosutural dysplasia, 607812
SEC23B	161.2	97	96	Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SEC24D	136.8	99	98	Cole-Carpenter syndrome 2, 616294
SEC61A1	133.2	100	100	Hyperuricemic nephropathy, familial juvenile, 4,617056
SEC63	60.7	85	75	Polycystic liver disease, 174050
SECISBP2	122.6	98	95	Thyroid hormone metabolism, abnormal, 609698

SEMA4A	127.6	99	98	Cone-rod dystrophy 10, 610283 Retinitis pigmentosa 35, 610282
SEPN1	111.9	85	83	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310
SEPSECS	159.4	100	100	Pontocerebellar hypoplasia type 2D, 613811
SEPT12	105.2	97	93	Spermatogenic failure 10, 614822
SEPT9	118.8	99	96	Amyotrophy, hereditary neuralgic, 162100 Leukemia, acute myeloid, therapy-related Ovarian carcinoma
SERAC1	112.4	98	94	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPINA1	141.3	100	99	Emphysema due to AAT deficiency, 613490 Emphysema-cirrhosis, due to AAT deficiency, 613490 Hemorrhagic diathesis due to '\antithrombin\' Pittsburgh, 613490 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963
SERPINA3	152.8	100	100	Alpha-1-antichymotrypsin deficiency Cerebrovascular disease, occlusive
SERPINA6	191.3	100	100	Corticosteroid-binding globulin deficiency,611489
SERPINB7	127.3	100	99	Palmoplantar keratoderma, Nagashima type, 615598
SERPINC1	143.2	100	100	Thrombophilia due to antithrombin III deficiency, 613118
SERPIND1	181.9	100	100	Thrombophilia due to heparin cofactor II deficiency,612356
SERPINE1	155.2	100	100	Plasminogen activator inhibitor-1 deficiency, 613329 {Transcription of plasminogen activator inhibitor, modulator of}
SERPINF1	101.7	100	99	Osteogenesis imperfecta, type VI, 613982
SERPINF2	144.1	99	99	Alpha-2-plasmin inhibitor deficiency,262850
SERPING1	98	97	92	Angioedema, hereditary, types I and II, 106100 Complement component 4, partial deficiency of, 120790
SERPINI1	104.9	99	96	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218
SETBP1	151.8	97	96	Mental retardation, autosomal dominant 29, 616078 Schinzel-Giedion midface retraction syndrome, 269150
SETD2	148.1	100	99	Luscan-Lumish syndrome, 616831
SETD5	184.5	100	99	Mental retardation, autosomal dominant 23, 615761
SETX	162.7	99	99	Amyotrophic lateral sclerosis 4, juvenile, 602433 Spinocerebellar ataxia, autosomal recessive 1, 606002
SF3B1	128.7	99	97	Myelodysplastic syndrome, somatic, 614286

SF3B4	89.6	99	97	Acrofacial dysostosis 1, Nager type, 154400
SFRP4	164.4	100	98	Pyle disease, 265900
SFTPA2	174.3	100	100	Pulmonary fibrosis, idiopathic, 178500
SFTPB	99.6	99	98	Surfactant metabolism dysfunction, pulmonary, 1, 265120
SFTPC	102.8	99	95	Surfactant metabolism dysfunction, pulmonary, 2, 610913
SFXN4	131.8	100	99	Combined oxidative phosphorylation deficiency 18, 615578
SGCA	144.8	100	99	Muscular dystrophy, limb-girdle, type 2D, 608099
SGCB	154.1	96	94	Muscular dystrophy, limb-girdle, type 2E, 604286
SGCD	94.8	100	99	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, type 2F, 601287
SGCE	88.9	93	90	Dystonia-11, myoclonic, 159900
SGCG	138.9	100	100	Muscular dystrophy, limb-girdle, type 2C, 253700
SGOL1	94.7	97	94	Chronic atrial and intestinal dysrhythmia, 616201
SGSH	129.3	95	93	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SH2B3	97.5	90	79	Erythrocytosis, somatic, 133100 Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
SH2D1A	105.3	89	89	Lymphoproliferative syndrome, X-linked, 1, 308240
SH3BP2	108.8	91	91	Cherubism, 118400
SH3PXD2B	140.4	100	99	Frank-ter Haar syndrome, 249420
SH3TC2	122.8	100	99	Charcot-Marie-Tooth disease, type 4C, 601596 Mononeuropathy of the median nerve, mild, 613353
SHANK3	85	80	72	Phelan-McDermid syndrome, 606232 {Schizophrenia 15}, 613950
SHH	117.6	99	94	Holoprosencephaly-3, 142945 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250
SHOC2	140.4	100	99	Noonan-like syndrome with loose anagen hair, 607721
SHOX	29.2	73	61	Langer mesomelic dysplasia, 249700 Leri-Weill dyschondrosteosis, 127300 Short stature, idiopathic familial, 300582
SI	97.9	96	87	Sucrase-isomaltase deficiency, congenital, 222900
SIK1	85.7	97	92	Epileptic encephalopathy, early infantile, 30, 616341

SIL1	154.8	99	98	Marinesco-Sjogren syndrome, 248800
SIM1	151.4	100	99	Obesity, severe, 601665
SIX1	117.3	99	97	Brachiootoc syndrome 3, 608389 Deafness, autosomal dominant 23, 605192
SIX3	145.6	100	98	Holoprosencephaly-2, 157170 Schizencephaly, 269160
SIX5	43.8	88	76	Branchiootorenal syndrome 2, 610896
SIX6	229.1	100	100	Optic disc anomalies with retinal and/or macular dystrophy, 212550
SKI	85.5	96	90	Shprintzen-Goldberg syndrome, 182212
SKIV2L	24.9	16	16	Trichohepatoenteric syndrome 2, 614602
SLC10A2	150.9	100	100	Bile acid malabsorption, primary, 613291
SLC11A2	135.3	100	99	Anemia, hypochromic microcytic, with iron overload 1, 206100
SLC12A1	172.7	99	99	Bartter syndrome, type 1, 601678
SLC12A3	139.5	100	100	Gitelman syndrome, 263800
SLC12A5	149.9	99	97	Epileptic encephalopathy, early infantile, 34, 616645 {Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685
SLC12A6	141.8	100	99	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC13A5	164.5	100	100	Epileptic encephalopathy, early infantile, 25, 615905
SLC16A1	156	99	98	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia, familial, 7, 610021 Monocarboxylate transporter 1 deficiency, 616095
SLC16A12	164.4	100	99	Cataract, juvenile, with microcornea and glucosuria, 612018
SLC16A2	57.7	93	81	Allan-Herndon-Dudley syndrome, 300523
SLC17A5	119.7	96	92	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC17A8	132.8	100	100	Deafness, autosomal dominant 25, 605583
SLC17A9	111.6	95	95	Porokeratosis 8, disseminated superficial actinic type, 616063
SLC19A2	119.3	99	97	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	178	100	99	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A1	174.1	100	99	Dicarboxylic aminoaciduria, 222730 {?Schizophrenia susceptibility 18}, 615232
SLC1A3	122.2	100	99	Episodic ataxia, type 6, 612656
SLC1A4	156.6	98	94	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657

SLC20A2	119.1	99	97	Basal ganglia calcification, idiopathic, 1, 213600
SLC22A12	105.9	100	99	Hypouricemia, renal, 220150
SLC22A18	85.1	92	81	Breast cancer, somatic, 114480 Lung cancer, somatic, 211980 Rhabdomyosarcoma, somatic, 268210
SLC22A5	153.5	100	100	Carnitine deficiency, systemic primary, 212140
SLC24A1	218.5	100	100	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
SLC24A4	127	99	97	Amelogenesis imperfecta, type IIA5, 615887 [Skin/hair/eye pigmentation 6, blond/brown hair], 210750 [Skin/hair/eye pigmentation 6, blue/green eyes], 210750
SLC24A5	114.4	99	97	Albinism, oculocutaneous, type VI, 113750 [Skin/hair/eye pigmentation 4, fair/dark skin], 113750
SLC25A1	71.1	92	86	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A12	150.4	99	98	Epileptic encephalopathy, early infantile, 39, 612949
SLC25A13	110.6	95	92	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A15	192.8	98	95	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A19	88.7	99	98	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A20	110.6	100	99	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A22	108.8	99	96	Epileptic encephalopathy, early infantile, 3, 609304
SLC25A26	98	98	96	Combined oxidative phosphorylation deficiency 28, 616794
SLC25A3	139.1	99	97	Mitochondrial phosphate carrier deficiency, 610773
SLC25A38	111.7	99	98	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC25A4	134	100	100	Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184 Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283
SLC25A46	204.3	95	87	Neuropathy, hereditary motor and sensory, type VIB, 616505
SLC26A2	233	100	100	Achondrogenesis Ib, 600972 Atelosteogenesis II, 256050 De la Chapelle dysplasia, 256050 Diastrophic dysplasia, 222600 Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 Epiphyseal dysplasia, multiple, 4, 226900

SLC26A3	156.2	99	98	Diarrhea 1, secretory chloride, congenital, 214700
SLC26A4	122.9	99	99	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791 Pendred syndrome, 274600
SLC26A8	139	100	99	Spermatogenic failure 3, 606766
SLC27A4	156.1	99	98	Ichthyosis prematurity syndrome, 608649
SLC29A3	203.8	99	99	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC2A1	179.6	100	100	Dystonia 9, 601042 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 GLUT1 deficiency syndrome 2, childhood onset, 612126 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847
SLC2A10	170.6	100	99	Arterial tortuosity syndrome, 208050
SLC2A2	178.3	100	99	Fanconi-Bickel syndrome, 227810 {Diabetes mellitus, noninsulin-dependent}, 125853
SLC2A9	119	99	96	Hypouricemia, renal, 2, 612076 {Uric acid concentration, serum, QTL 2}, 612076
SLC30A10	161.7	100	99	Hypermanganesemia with dystonia 1, 613280
SLC30A2	146.4	100	99	Zinc deficiency, transient neonatal, 608118
SLC33A1	140.7	96	90	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539
SLC34A1	153.5	100	99	Fanconi renotubular syndrome 2, 613388 Hypercalcemia, infantile 2, 616963 Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286
SLC34A2	169.4	100	100	Pulmonary alveolar microlithiasis, 265100
SLC34A3	105.8	98	94	Hypophosphatemic rickets with hypercalciuria, 241530
SLC35A1	123.9	99	97	Congenital disorder of glycosylation, type II f, 603585
SLC35A2	111.4	99	97	Congenital disorder of glycosylation, type II m, 300896
SLC35C1	230.6	99	98	Congenital disorder of glycosylation, type II c, 266265
SLC35D1	113.1	96	90	Schneckenbecken dysplasia, 269250
SLC36A2	124.6	100	100	Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC37A4	140.4	100	99	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240
SLC38A8	76.5	99	95	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218



SLC39A13	114.9	99	98	Spondylocheiroadysplasia, Ehlers-Danlos syndrome-like, 612350
SLC39A14	107.9	99	98	Hypermanganesemia with dystonia 2, 617013
SLC39A4	81.9	99	96	Acrodermatitis enteropathica, 201100
SLC39A5	119.5	100	99	Myopia 24, autosomal dominant, 615946
SLC39A8	128.6	100	99	Congenital disorder of glycosylation, type II, 616721
SLC3A1	162.9	100	99	Cystinuria, 220100
SLC40A1	155.8	99	99	Hemochromatosis, type 4, 606069
SLC45A2	144.2	99	99	Albinism, oculocutaneous, type IV, 606574 [Skin/hair/eye pigmentation 5, black/nonblack hair], 227240 [Skin/hair/eye pigmentation 5, dark/fair skin], 227240 [Skin/hair/eye pigmentation 5, dark/light eyes], 227240
SLC46A1	106.1	99	96	Folate malabsorption, hereditary, 229050
SLC4A1	142.3	100	99	Cryohydrocytosis, 185020 Ovalocytosis, SA type, 166900 Renal tubular acidosis, distal, AD, 179800 Renal tubular acidosis, distal, AR, 611590 Spherocytosis, type 4, 612653 [Blood group, Diego], 110500 [Blood group, Froese], 601551 [Blood group, Swann], 601550 [Blood group, Waldner], 112010 [Blood group, Wright], 112050 [Malaria, resistance to], 611162
SLC4A11	153.4	100	99	Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy, autosomal recessive, 217700 Corneal endothelial dystrophy and perceptive deafness, 217400
SLC4A4	122.1	99	97	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC52A1	219.8	100	100	Riboflavin deficiency, 615026
SLC52A2	177.8	100	100	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	119.7	100	100	Brown-Vialetto-Van Laere syndrome 1, 211530 Fazio-Londe disease, 211500
SLC5A1	139.9	100	100	Glucose/galactose malabsorption, 606824
SLC5A2	118.9	100	100	Renal glucosuria, 233100
SLC5A5	94	99	96	Thyroid dysmorphogenesis 1, 274400

SLC5A7	117.2	100	99	Neuronopathy, distal hereditary motor, type VIIA, 158580
SLC6A1	143.8	100	100	Myoclonic-atonic epilepsy, 616421
SLC6A17	190.3	100	100	Mental retardation, autosomal recessive 48, 616269
SLC6A19	149.5	100	99	Hartnup disorder, 234500 Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC6A2	150.1	100	99	Orthostatic intolerance, 604715
SLC6A20	178.9	100	99	Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC6A3	146	100	99	Parkinsonism-dystonia, infantile, 613135 {Nicotine dependence, protection against}, 188890
SLC6A5	136.7	100	99	Hyperekplexia 3, 614618
SLC6A8	57	89	79	Cerebral creatine deficiency syndrome 1, 300352
SLC7A14	192.1	100	100	Retinitis pigmentosa 68, 615725
SLC7A7	124	100	99	Lysinuric protein intolerance, 222700
SLC7A9	125.5	99	99	Cystinuria, 220100
SLC9A3	147.5	98	96	Diarrhea 8, secretory sodium, congenital, 616868
SLC9A3R1	111.6	99	96	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
SLC9A6	105	97	91	Mental retardation, X-linked syndromic, Christianson type, 300243
SLCO1B1	45.9	92	84	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO1B3	45.2	91	74	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO2A1	110.8	100	99	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
SLFN14	193	100	100	Bleeding disorder, platelet-type, 20, 616913
SLITRK1	132.5	100	100	Tourette syndrome, 137580 ?Trichotillomania, 613229
SLITRK6	206.7	100	100	Deafness and myopia, 221200
SLURP1	97.3	99	96	Meleda disease, 248300
SLX4	114.4	100	99	Fanconi anemia, complementation group P, 613951
SMAD3	132	99	99	Loeys-Dietz syndrome 3, 613795
SMAD4	121.2	99	98	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Polyposis, juvenile intestinal, 174900
SMAD6	100.5	81	73	Aortic valve disease 2, 614823

SMAD9	132.7	100	100	Pulmonary hypertension,primary,615342 Polyposis & gastrointestinal ganglioneuromas (Ngeow (2015) Gastroenterology 149,886)
SMARCA2	115.8	97	95	Nicolaidis-Baraitser syndrome, 601358
SMARCA4	144.8	100	99	Coffin-Siris syndrome 4, 614609 {Rhabdoid tumor predisposition syndrome 2}, 613325
SMARCAD1	85.5	99	96	Adermatoglyphia, 136000
SMARCAL1	134.9	100	99	Schimke immunoosseous dysplasia, 242900
SMARCB1	214.6	100	100	Coffin-Siris syndrome 3, 614608 Rhabdoid tumors, somatic, 609322 {Rhabdoid predisposition syndrome 1}, 609322 {Schwannomatosis-1, susceptibility to}, 162091
SMARCE1	73.6	96	86	Coffin-Siris syndrome 5,616938 {Meningioma, familial, susceptibility to}, 607174
SMC1A	101.3	99	98	Cornelia de Lange syndrome 2,300590
SMC3	81.3	93	87	Cornelia de Lange syndrome 3, 610759
SMCHD1	90.8	98	91	Fascioscapulohumeral muscular dystrophy 2, digenic, 158901
SMN1	116.3	99	96	Spinal muscular atrophy-1, 253300 Spinal muscular atrophy-2, 253550 Spinal muscular atrophy-3, 253400 Spinal muscular atrophy-4, 271150
SMO	149.4	96	93	Basal cell carcinoma, somatic, 605462 Curry-Jones syndrome,somatic mosaic, 601707
SMOC1	129.8	99	97	Microphthalmia with limb anomalies, 206920
SMOC2	119	97	94	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SMPD1	127.5	99	98	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SMPX	68.7	99	95	Deafness, X-linked 4, 300066
SMS	65.5	85	70	Mental retardation, X-linked, Snyder-Robinson type, 309583
SNAI2	130.1	100	99	Piebaldism, 172800 Waardenburg syndrome, type 2D, 608890
SNAP29	153.8	100	100	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNCA	129.8	100	100	Dementia, Lewy body, 127750 Parkinson disease 1, 168601 Parkinson disease 4, 605543

SNCB	79.2	100	99	Dementia, Lewy body, 127750
SNIP1	139.7	99	96	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501
SNRNP200	161.3	100	99	Retinitis pigmentosa 33, 610359
SNRPB	76.1	99	97	Cerebrocostomandibular syndrome, 117650
SNRPE	78.9	98	88	Hypotrichosis 11, 615059
SNRPN	117.1	99	94	Prader-Willi syndrome, 176270
SNTA1	97.1	82	77	Long QT syndrome 12, 612955
SNX10	114.3	100	99	Osteopetrosis, autosomal recessive 8, 615085
SNX14	70.1	95	82	Spinocerebellar ataxia, autosomal recessive 20, 616354
SOBP	132.3	93	86	Mental retardation, anterior maxillary protrusion, and strabismus, 613671
SOD1	162.3	100	100	Amyotrophic lateral sclerosis 1, 105400
SOS1	95.5	96	90	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOS2	97	98	92	Noonan syndrome 9, 616559
SOST	112.6	100	99	Craniodiaphyseal dysplasia, autosomal dominant, 122860 Sclerosteosis 1, 269500 Van Buchem disease, 239100
SOX10	65.8	98	91	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266
SOX11	118.5	99	96	Mental retardation, autosomal dominant, 27, 615866
SOX17	70.9	99	94	Vesicoureteral reflux 3, 613674
SOX18	21.2	62	48	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823 Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940
SOX2	129.2	98	93	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SOX3	37.9	86	71	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000
SOX5	107.3	99	96	Lamb-Shaffer syndrome, 616803
SOX9	134.2	97	93	Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia with autosomal sex reversal, 114290 Campomelic dysplasia, 114290
SP110	121.7	100	99	Hepatic venoocclusive disease with immunodeficiency, 235550 {Mycobacterium tuberculosis, susceptibility to}, 607948

SPAG1	87.3	96	88	Ciliary dyskinesia, primary, 28, 615505
SPARC	161	100	100	Osteogenesis imperfecta, type XVII, 616507
SPAST	63.8	93	81	Spastic paraplegia 4, autosomal dominant, 182601
SPATA5	131.8	99	99	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPATA7	119.4	97	90	Leber congenital amaurosis 3, 604232  Retinitis pigmentosa, juvenile, autosomal recessive, 604232
SPECC1L	157.9	100	100	Opitz GBBB syndrome, type II, 145410 ?Facial clefting, oblique, 1, 600251
SPEG	91.4	92	85	Centronuclear myopathy 5, 615959
SPG11	129.2	99	96	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360
SPG20	132.3	99	98	Troyer syndrome, 275900
SPG21	121.5	98	94	Mast syndrome, 248900
SPG7	119.4	93	92	Spastic paraplegia 7, autosomal recessive, 607259
SPINK1	80.2	100	99	Pancreatitis, hereditary, 167800 Tropical calcific pancreatitis, 608189 {Fibrocalculous pancreatic diabetes, susceptibility to}, 608189
SPINK5	145	99	96	Atopy, 147050 Netherton syndrome, 256500
SPINT2	71.6	97	84	Diarrhea 3, secretory sodium, congenital, syndromic, 270420
SPR	166.9	98	89	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRED1	164.3	98	96	Legius syndrome, 611431
SPRTN	167.9	100	100	Ruijs-Aalfs syndrome, 616200
SPRY4	139.1	100	100	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
SPTA1	119.4	99	98	Elliptocytosis-2, 130600 Pyropoikilocytosis, 266140 Spherocytosis, type 3, 270970
SPTAN1	126.6	99	99	Epileptic encephalopathy, early infantile, 5, 613477
SPTB	148.3	100	99	Anemia, neonatal hemolytic, fatal and near-fatal Elliptocytosis-3 Spherocytosis, type 2, 616649
SPTBN2	118.4	99	99	Spinocerebellar ataxia 5, 600224

				Spinocerebellar ataxia, autosomal recessive 14, 615386
SPTLC1	115.4	98	94	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	160.3	100	100	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SQSTM1	110.3	98	95	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 Paget disease of bone 3, 167250
SRC	105.5	99	97	Colon cancer, advanced, somatic ?Thrombocytopenia 6,616937
SRCAP	154.2	99	99	Floating-Harbor syndrome, 136140
SRD5A2	77.7	100	96	Pseudovaginal perineoscrotal hypospadias, 264600
SRD5A3	135.8	100	99	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713
SRP72	69.8	93	83	Bone marrow failure syndrome 1, 614675
SRY	45.2	49	49	46XX sex reversal 1, 400045 46XY sex reversal 1, 400046
SSTR5	155.1	99	99	Somatostatin analog, resistance to
ST14	154.1	98	97	Ichthyosis, congenital, autosomal recessive 11, 602400
ST3GAL3	144.9	100	99	Epileptic encephalopathy, early infantile, 15, 615006 Mental retardation, autosomal recessive 12, 611090
ST3GAL5	133.1	95	94	Amish infantile epilepsy syndrome, 609056
STAC3	120.1	100	99	Native American myopathy, 255995
STAMBP	112.3	99	96	Microcephaly-capillary malformation syndrome, 614261
STAR	124	100	100	Lipoid adrenal hyperplasia, 201710
STAT1	126.2	98	95	Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796 Immunodeficiency 31C, autosomal dominant, 614162
STAT2	116.1	100	99	Immunodeficiency 44, 616636
STAT3	119.6	99	99	Autoimmune disease, multisystem, infantile-onset, 615952 Hyper-IgE recurrent infection syndrome, 147060
STAT5B	130.7	99	97	Growth hormone insensitivity with immunodeficiency, 245590 Leukemia, acute promyelocytic, somatic, 102578
STIL	157.2	99	98	Microcephaly 7, primary, autosomal recessive, 612703
STIM1	127.2	100	99	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 1 160565 Stormorken syndrome, 185070

STK11	112.2	99	95	Melanoma, malignant, somatic Pancreatic cancer, 260350 Peutz-Jeghers syndrome, 175200 Testicular tumor, somatic, 273300
STK4	139	100	99	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STOX1	177.9	89	89	Preeclampsia/eclampsia 4, 609404
STRA6	116.6	100	99	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186
STRADA	134.3	100	98	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087
STRC	119	99	98	Deafness, autosomal recessive 16, 603720
STS	92.2	99	97	Ichthyosis, X-linked, 308100
STUB1	176.5	100	98	Spinocerebellar ataxia, autosomal recessive 16, 615768
STX11	312.1	100	100	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STX16	138.2	99	97	Pseudohypoparathyroidism, type 1B, 603233
STX1B	152.2	100	98	Generalized epilepsy with febrile seizures plus, type 9, 616172
STXBP1	125.8	100	100	Epileptic encephalopathy, early infantile, 4, 612164
STXBP2	125.3	98	97	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
SUCLA2	65.3	93	82	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	101.2	99	95	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUFU	122.7	100	99	Basal cell nevus syndrome, 109400 Medulloblastoma, desmoplastic, 155255 {Meningioma, familial, susceptibility to}, 607174
SUMF1	106.4	99	92	Multiple sulfatase deficiency, 272200
SUMO1	17.7	58	37	Orofacial cleft 10, 613705
SUOX	213.1	100	100	Sulfite oxidase deficiency, 272300
SYCP3	78.8	97	86	Pregnancy loss, recurrent, 4, 270960 Spermatogenic failure 4, 270960
SYN1	64.8	74	63	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNE1	139.1	99	99	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 Spinocerebellar ataxia, autosomal recessive 8, 610743
SYNE2	122.9	98	96	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999
SYNE4	73.9	98	91	Deafness, autosomal recessive 76, 615540

SYNGAP1	70.9	49	49	Mental retardation, autosomal dominant 5, 612621
SYNJ1	127.2	99	96	Parkinson disease 20, early-onset, 615530
SYP	72.5	99	94	Mental retardation, X-linked 96, 300802
SYT14	180.2	95	85	Spinocerebellar ataxia, autosomal recessive 11, 614229
SYT2	101.3	100	99	Myasthenic syndrome, congenital, 7, presynaptic, 616040
SZT2	147.4	99	99	Epileptic encephalopathy, early infantile, 18, 615476
T	141.4	98	93	Sacral agenesis with vertebral anomalies, 615709 {Neural tube defects, susceptibility to}, 182940
TAB2	209.6	99	97	Congenital heart defects, nonsyndromic, 2, 614980
TAC3	80.5	99	90	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACO1	91.7	97	92	Mitochondrial complex IV deficiency, 220110
TACR3	180.5	100	100	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
TACSTD2	224	99	96	Corneal dystrophy, gelatinous drop-like, 204870
TAF1	113.3	99	96	Dystonia-Parkinsonism, X-linked, 314250 Mental retardation, X-linked, syndromic 33, 300966
TAF2	112.7	98	94	Mental retardation, autosomal recessive 40, 615599
TAL1	44	74	63	Leukemia, T-cell acute lymphocytic, somatic, 613065
TAL2	112.6	100	100	Leukemia, T-cell acute lymphocytic, somatic, 613065
TALDO1	130.6	100	99	Transaldolase deficiency, 606003
TANGO2	145.6	100	100	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias and neurodegeneration, 616878
TAP1	12.9	12	12	Bare lymphocyte syndrome, type I, 604571
TAP2	15.2	15	15	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571 Wegener-like granulomatosis
TAPBP	20.2	19	18	Bare lymphocyte syndrome, type I, 604571
TAPT1	83.6	88	85	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897
TARDBP	171.7	100	100	Amyotrophic lateral sclerosis 10, with or without FTD, 612069 Frontotemporal lobar degeneration, TARDBP-related, 612069
TAT	143.3	100	100	Tyrosinemia, type II, 276600
TAZ	94.9	99	98	Barth syndrome, 302060
TBC1D20	146.1	94	94	Warburg micro syndrome 4, 615663



TBC1D24	179.4	100	100	Deafness , autosomal recessive 86, 614617 Deafness, autosomal dominant 65, 616044 DOOR syndrome, 220500 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021
TBC1D7	105.5	99	96	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000
TBCE	127.9	99	98	Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Kenny-Caffey syndrome, type 1, 244460
TBCK	86.4	95	89	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3,616900
TBK1	102.4	97	90	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439
TBL1XR1	79.4	91	72	Mental retardation, autosomal dominant 41,616944 Piermont syndrome,602342
TBP	129.7	100	98	Spinocerebellar ataxia 17, 607136 {Parkinson disease, susceptibility to}, 168600
TBX1	75.5	77	67	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Velocardiofacial syndrome, 192430
TBX15	131.9	100	99	Cousin syndrome, 260660
TBX18	94.4	98	95	Congenital anomalies of kidney and urinary tract 2, 143400
TBX19	174.7	100	100	Adrenocorticotrophic hormone deficiency, 201400
TBX20	143.2	99	99	Atrial septal defect 4, 611363
TBX21	81.8	89	83	Asthma and nasal polyps, 208550 {Asthma, aspirin-induced, susceptibility to}, 208550
TBX22	122.5	99	96	Cleft palate with ankyloglossia, 303400 ?Abruzzo-Erickson syndrome, 302905
TBX3	80.6	99	95	Ulnar-mammary syndrome, 181450
TBX4	170.9	94	92	Ischiocoxopodopatellar syndrome, 147891
TBX5	141.6	100	100	Holt-Oram syndrome, 142900
TBX6	118.6	91	78	Spondylocostal dysostosis 5, 122600
TBXAS1	140.3	100	100	Ghosal hematodiaphyseal syndrome, 231095 ?Thromboxane synthase deficiency, 614158
TCAP	89.3	100	99	Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, type 2G, 601954

TCF12	147.9	100	99	Craniosynostosis 3, 615314
TCF3	67.2	98	92	Agammaglobulinemia 8,autosomal dominant,616941
TCF4	128.7	99	99	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954
TCIRG1	113.7	95	89	Osteopetrosis, autosomal recessive 1, 259700
TCN2	175.9	100	100	Transcobalamin II deficiency, 275350
TCOF1	98.6	99	97	Treacher Collins syndrome 1, 154500
TCTN1	100.5	96	94	Joubert syndrome 13, 614173
TCTN2	144.2	99	97	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	127.5	100	99	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TDGF1	151.5	99	96	Forebrain defects
TDP1	123	98	95	Spinocerebellar ataxia, autosomal recessive with axonal neuropathy, 607250
TDP2	156.8	99	98	Spinocerebellar ataxia, autosomal recessive, 616949
TDRD7	168.4	98	97	Cataract 36, 613887
TEAD1	158.9	99	98	Sveinsson choreoretinal atrophy, 108985
TECPR2	161.3	100	99	Spastic paraplegia 49, autosomal recessive, 615031
TECR	93.8	99	98	Mental retardation, autosomal recessive 14, 614020
TECTA	208.2	100	100	Deafness, autosomal dominant 8/12, 601543 Deafness, autosomal recessive 21, 603629
TEK	185.6	100	100	Glaucoma 3,primary congenital, E, 617272 Venous malformations, multiple cutaneous and mucosal, 600195
TELO2	98.2	97	93	You-Hoover-Fong syndrome, 616954
TENM3	185.8	99	98	Microphthalmia, isolated, with coloboma 9, 615145
TENM4	161	99	99	Tremor, hereditary essential, 5, 616736
TERC	NC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550 {Aplastic anemia}, 614743 {Pulmonary fibrosis, idiopathic, susceptibility to}, 614743
TET2	212.5	100	99	Myelodysplastic syndrome, somatic, 614286
TEX11	77.8	91	83	Spermatogenic failure, X-linked, 2, 309120
TF	126	100	100	Atransferrinemia, 209300
TFAP2A	109.5	100	99	Branchiooculofacial syndrome, 113620

TFAP2B	155.6	98	96	Char syndrome, 169100 Patent ductus arteriosus 2, 617035
TFE3	74.2	99	93	Renal cell carcinoma, papillary, 1, 300854
TFG	125.3	96	93	Hereditary motor and sensory neuropathy, Okinawa type, 604484 ?Spastic paraplegia 57, autosomal recessive, 615658
TFR2	104.6	98	93	Hemochromatosis, type 3, 604250
TFRC	157.3	99	99	Immunodeficiency 46, 616740
TG	133.5	100	99	Thyroid dyshormonogenesis 3, 274700 {Autoimmune thyroid disease, susceptibility to, 3}, 608175
TGDS	82.3	98	88	Catel-Manzke syndrome, 616145
TGFB1	87.1	99	95	Camurati-Engelmann disease, 131300 {Cystic fibrosis lung disease, modifier of}, 219700
TGFB2	176.9	100	99	Loeys-Dietz syndrome 4, 614816
TGFB3	171.8	100	100	Arrhythmogenic right ventricular dysplasia 1, 107970 Loeys-Dietz syndrome 5, 615582
TGFBI	130.1	99	94	Corneal dystrophy, Avellino type, 607541 Corneal dystrophy, epithelial basement membrane, 121820 Corneal dystrophy, Groenouw type I, 121900 Corneal dystrophy, lattice type I, 122200 Corneal dystrophy, lattice type IIIA, 608471 Corneal dystrophy, Reis-Bucklers type, 608470 Corneal dystrophy, Thiel-Behnke type, 602082
TGFBR1	173.5	93	93	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800
TGFBR2	193.8	100	99	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome 2, 610168
TGIF1	138.4	100	100	Holoprosencephaly-4, 142946
TGM1	159.2	100	100	Ichthyosis, congenital, autosomal recessive 1, 242300
TGM5	174.3	100	100	Peeling skin syndrome 2, 609796
TGM6	149.9	99	98	Spinocerebellar ataxia 35, 613908
TH	68.4	97	88	Segawa syndrome, recessive, 605407
THAP1	121.7	100	100	Dystonia 6, torsion, 602629
THBD	108.4	99	97	Thrombophilia due to thrombomodulin defect, 614486

				{Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
THOC2	78.4	96	86	Mental retardation, X-linked 12/35, 300957
THOC6	248.6	100	99	Beaulieu-Boycott-Innes syndrome, 613680
THPO	88.1	100	100	Thrombocytopenia 1, 187950
THRA	173.1	100	99	Hypothyroidism, congenital, nongoitrous, 6, 614450
THRB	167.3	100	99	Thyroid hormone resistance, 188570 Thyroid hormone resistance, autosomal recessive, 274300 Thyroid hormone resistance, selective pituitary, 145650
TIA1	128.4	98	89	Welander distal myopathy, 604454
TIMM8A	46.2	94	78	Jensen syndrome, 311150 Mohr-Tranebjaerg syndrome, 304700
TIMP3	147.4	100	100	Sorsby fundus dystrophy, 136900
TINF2	183.9	100	100	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TJP2	119.5	99	98	Cholestasis, progressive familial intrahepatic 4, 615878 Hypercholanemia, familial, 607748
TK2	105.4	93	89	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560
TKT	114.1	98	97	Short stature, developmental delay, and congenital heart defects, 617044
TLE6	110.1	99	98	Preimplantation embryonic lethality, 616814
TLL1	140	100	99	Atrial septal defect 6, 613087
TMC1	122.7	98	93	Deafness, autosomal dominant 36, 606705 Deafness, autosomal recessive 7, 600974
TMC6	83.8	99	99	Epidermodysplasia verruciformis, 226400
TMC8	108.1	97	91	Epidermodysplasia verruciformis, 226400
TMCO1	89.3	100	98	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980
TMEM126A	120.1	98	86	Optic atrophy 7, 612989
TMEM126B	79.1	99	97	Mitochondrial complex I deficiency, 252010
TMEM138	100.3	100	99	Joubert syndrome 16, 614465
TMEM165	112.2	99	97	Congenital disorder of glycosylation, type IIk, 614727
TMEM173	100.8	98	93	STING-associated vasculopathy, infantile-onset, 615934
TMEM199	120.8	100	99	Congenital disorder of glycosylation, type IIp, 616829
TMEM216	112	100	98	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194

TMEM231	111.8	100	99	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	100.6	99	98	Joubert syndrome 14, 614424
TMEM240	112.7	99	97	Spinocerebellar ataxia 21, 607454
TMEM38B	114.3	100	99	Osteogenesis imperfecta, type XIV, 615066
TMEM43	125	100	99	Arrhythmogenic right ventricular dysplasia 5, 604400 Emery-Dreifuss muscular dystrophy 7, AD, 614302
TMEM5	120.5	96	92	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
TMEM67	72.9	93	83	COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991
TMEM70	138.8	94	90	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMEM98	136.9	99	98	Nanophthalmos 4, 615972
TMIE	109.8	98	92	Deafness, autosomal recessive 6, 600971
TMPRSS15	116.9	95	89	Enterokinase deficiency, 226200
TMPRSS3	119.3	99	99	Deafness, autosomal recessive 8/10, 601072
TMPRSS6	102	100	99	Iron-refractory iron deficiency anemia, 206200
TNC	187.7	100	99	Deafness, autosomal dominant 56, 615629
TNFAIP3	136.1	100	99	Autoinflammatory syndrome, familial, Behcet-like, 616744
TNFRSF10B	114.3	100	100	Squamous cell carcinoma, head and neck, 275355
TNFRSF11A	147	93	91	Osteolysis, familial expansile, 174810 Osteopetrosis, autosomal recessive 7, 612301 {Paget disease of bone 2, early-onset}, 602080
TNFRSF11B	224.9	100	100	Paget disease of bone 5, juvenile-onset, 239000
TNFRSF13B	111.2	100	99	Immunodeficiency, common variable, 2, 240500 Immunoglobulin A deficiency 2, 609529
TNFRSF13C	55.9	76	66	Immunodeficiency, common variable, 4, 613494
TNFRSF1A	93.2	90	87	Periodic fever, familial, 142680 {Multiple sclerosis, susceptibility to, 5}, 614810
TNFSF11	150.3	99	93	Osteopetrosis, autosomal recessive 2, 259710
TNIK	110.7	99	99	Mental retardation, autosomal recessive 54, 617028

TNNC1	175.2	100	100	Cardiomyopathy, dilated, 1Z, 611879 Cardiomyopathy, hypertrophic, 13, 613243
TNNI2	122.5	100	99	Arthrogryposis multiplex congenita, distal, type 2B, 601680
TNNI3	86.8	98	86	Cardiomyopathy, dilated, 1FF, 613286 Cardiomyopathy, familial restrictive, 1, 115210 Cardiomyopathy, hypertrophic, 7, 613690 ?Cardiomyopathy, dilated, 2A, 611880
TNNT1	87.1	96	94	Nemaline myopathy 5, Amish type, 605355
TNNT2	105.8	100	99	Cardiomyopathy, dilated, 1D, 601494 Cardiomyopathy, familial restrictive, 3, 612422 Cardiomyopathy, hypertrophic, 2, 115195 Left ventricular noncompaction 6, 601494
TNNT3	118.4	99	97	Arthrogryposis, distal, type 2B, 601680
TNPO3	139.7	100	99	Muscular dystrophy, limb-girdle, type 1F, 608423
TNXB	19.7	19	18	Ehlers-Danlos syndrome due to tenascin X deficiency, 606408 Vesicoureteral reflux 8, 615963
TOPORS	210.9	100	100	Retinitis pigmentosa 31, 609923
TOR1A	185.2	100	99	Dystonia-1, torsion, 128100 {Dystonia-1, modifier of}
TP53	92.1	99	98	Adrenal cortical carcinoma, 202300 Breast cancer, 114480 Choroid plexus papilloma, 260500 Colorectal cancer, 114500 Hepatocellular carcinoma, 114550 Li-Fraumeni syndrome, 151623 Nasopharyngeal carcinoma, 607107 Osteosarcoma, 259500 Pancreatic cancer, 260350 {Basal cell carcinoma 7}, 614740 {Glioma susceptibility 1}, 137800

TP63	206.5	100	100	ADULT syndrome, 103285 Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 Hay-Wells syndrome, 106260 Limb-mammary syndrome, 603543 Orofacial cleft 8, 129400 Rapp-Hodgkin syndrome, 129400 Split-hand/foot malformation 4, 605289
TPI1	103.2	99	96	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512
TPK1	112.7	99	97	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TPM1	137.4	99	97	Cardiomyopathy, dilated, 1Y, 611878 Cardiomyopathy, hypertrophic, 3, 115196 Left ventricular noncompaction 9, 611878
TPM2	109.4	100	99	Arthrogryposis multiplex congenita, distal, type 1, 108120 Arthrogryposis, distal, type 2B, 601680 CAP myopathy 2, 609285 Nemaline myopathy 4, autosomal dominant, 609285
TPM3	98.9	89	89	CAP myopathy 1, 609284 Myopathy, congenital, with fiber-type disproportion, 255310 Nemaline myopathy 1, autosomal dominant or recessive, 609284
TPO	139.7	99	98	Thyroid dyshormonogenesis 2A, 274500
TPP1	146.4	100	100	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TPRN	66.2	75	66	Deafness, autosomal recessive 79, 613307
TRAC	170.7	100	100	Immunodeficiency 7, TCR-alpha/beta deficient, 615387
TRAF3IP1	90.2	96	92	Senior-Loken syndrome 9, 616629
TRAIP	141.8	100	100	Seckel syndrome 9, 616777
TRAPPC11	126.2	99	96	Muscular dystrophy, limb-girdle, type 2S, 615356
TRAPPC2	89	87	68	Spondyloepiphyseal dysplasia tarda, 313400
TRAPPC9	135.2	100	99	Mental retardation, autosomal recessive 13, 613192
TRDN	68.2	83	69	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441
TREM2	148.9	99	99	Nasu-Hakola disease, 221770
TREX1	242.7	100	100	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315

				{Systemic lupus erythematosus, susceptibility to}, 152700
TRIM2	168.7	99	97	Charcot-Marie-Tooth disease, type 2R, 615490
TRIM32	141.3	100	100	Muscular dystrophy, limb-girdle, type 2H, 254110 ?Bardet-Biedl syndrome 11, 615988
TRIM37	111.5	99	98	Mulibrey nanism, 253250
TRIO	135.3	98	96	Mental retardation, autosomal dominant 44,617061
TRIOBP	135.9	97	94	Deafness, autosomal recessive 28, 609823
TRIP11	84.2	95	87	Achondrogenesis, type IA, 200600
TRIP4	113.5	100	98	Spinal muscular atrophy with congenital bone fractures 1,616866 ?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066
TRMT10A	135.1	100	99	Microcephaly, short stature, and impaired glucose metabolism 1, 616033
TRMT10C	131.5	99	98	Combined oxidative phosphorylation deficiency 30, 616974
TRMT5	208.4	99	93	Combined oxidative phosphorylation deficiency 26, 616539
TRMU	99.1	100	99	Liver failure, transient infantile, 613070 {Deafness, mitochondrial, modifier of}, 580000
TRNT1	104.6	97	92	Retinitis pigmentosa and erythrocytic microcytosis, 616959 Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084
TRPA1	92.2	91	85	Episodic pain syndrome, familial, 615040
TRPC6	103.7	99	96	Glomerulosclerosis, focal segmental, 2, 603965
TRPM1	161	100	99	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TRPM4	109.3	99	98	Progressive familial heart block, type IB, 604559
TRPM6	150.6	99	98	Hypomagnesemia 1, intestinal, 602014
TRPS1	174.7	100	99	Trichorhinophalangeal syndrome, type I, 190350 Trichorhinophalangeal syndrome, type III, 190351
TRPV3	148.3	100	99	Olmsted syndrome, 614594 ?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400



TRPV4	172.7	99	98	Brachyolmia type 3, 113500 Digital arthropathy-brachydactyly, familial, 606835 Hereditary motor and sensory neuropathy, type IIc, 606071 Metatropic dysplasia, 156530 Parastremmatic dwarfism, 168400 Scapuloperoneal spinal muscular atrophy, 181405 SED, Maroteaux type, 184095 Spinal muscular atrophy, distal, congenital nonprogressive, 600175 Spondylometaphyseal dysplasia, Kozlowski type, 184252 [Sodium serum level QTL 1], 613508
TSC1	128.9	99	98	Lymphangiomyomatosis, 606690 Tuberous sclerosis-1, 191100
TSC2	131.3	100	99	Lymphangiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254
TSEN15	74.1	99	93	Pontocerebellar hypoplasia, type 2F, 617026
TSEN2	123.8	100	99	Pontocerebellar hypoplasia type 2B, 612389
TSEN34	53.7	90	86	Pontocerebellar hypoplasia type 2C, 612390
TSEN54	83.1	95	92	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753 ?Pontocerebellar hypoplasia type 5, 610204
TSFM	123.2	100	100	Combined oxidative phosphorylation deficiency 3, 610505
TSG101	103.3	100	98	Breast cancer, somatic, 114480
TSHB	271.1	100	100	Hypothyroidism, congenital, nongoitrous 4, 275100
TSHR	216.7	99	96	Hyperthyroidism, familial gestational, 603373 Hyperthyroidism, nonautoimmune, 609152 Hypothyroidism, congenital, nongoitrous, 1 275200 Thyroid adenoma, hyperfunctioning, somatic Thyroid carcinoma with thyrotoxicosis
TSHZ1	167	98	98	Aural atresia, congenital, 607842
TSPAN12	129.3	100	99	Exudative vitreoretinopathy 5, 613310
TSPAN7	122.9	99	98	Mental retardation, X-linked 58, 300210
TSPEAR	141.8	100	99	Deafness, autosomal recessive 98, 614861 Ectodermal dysplasia (Peled et al. (2016) PLOS Genetics online)
TSPYL1	141.4	100	99	Sudden infant death with dysgenesis of the testes syndrome, 608800

TTBK2	148.5	100	99	Spinocerebellar ataxia 11, 604432
TTC19	93.2	85	79	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTC21B	100.6	99	97	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
TTC25	103.6	100	99	Ciliary dyskinesia, primary, 35, 617092
TTC37	123.9	99	98	Trichohepatoenteric syndrome 1, 222470
TTC7A	121.4	99	98	Gastrointestinal defects and immunodeficiency syndrome, 243150
TTC8	106.8	97	92	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
TTI2	104.5	100	99	Mental retardation, autosomal recessive 39, 615541
TTLL5	152.6	99	98	Cone-rod dystrophy 19, 615860
TTN	187.7	98	97	Cardiomyopathy, dilated, 1G, 604145 Cardiomyopathy, familial hypertrophic, 9, 613765 Muscular dystrophy, limb-girdle, type 2J, 608807 Myopathy, early-onset, with fatal cardiomyopathy, 611705 Myopathy, proximal, with early respiratory muscle involvement, 603689 Tibial muscular dystrophy, tardive, 600334
TTPA	101.2	83	76	Ataxia with isolated vitamin E deficiency, 277460
TTR	159.7	100	100	Amyloidosis, hereditary, transthyretin-related, 105210 Carpal tunnel syndrome, familial, 115430 [Dystransthyretinemic hyperthyroxinemia], 145680
TUBA1A	113.3	99	97	Lissencephaly 3, 611603
TUBA4A	220.9	100	100	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia, 616208
TUBA8	177.3	99	99	Polymicrogyria with optic nerve hypoplasia, 613180
TUBB	19.7	12	12	Cortical dysplasia, complex, with other brain malformations 6, 615771 Symmetric circumferential skin creases, congenital, 1, 156610
TUBB1	186.5	100	100	Macrothrombocytopenia, autosomal dominant, TUBB1-related, 613112
TUBB2A	109.8	96	95	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB2B	100.2	100	100	Polymicrogyria, symmetric or asymmetric, 610031
TUBB3	205.5	98	98	Cortical dysplasia, complex, with other brain malformations 1, 614039 Fibrosis of extraocular muscles, congenital, 3A, 600638
TUBB4A	121.5	96	95	Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438
TUBB8	31.1	95	69	Oocyte maturation defect 2, 616780

TUBG1	164.6	100	100	Cortical dysplasia, complex, with other brain malformations 4, 615412
TUBGCP4	130.6	99	96	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TUBGCP6	152.4	99	98	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
TUFM	135.7	100	99	Combined oxidative phosphorylation deficiency 4, 610678
TULP1	98	96	91	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132
TUSC3	136.1	100	98	Mental retardation, autosomal recessive 7, 611093
TWIST1	134.5	96	87	Craniosynostosis, type 1, 123100 Robinow-Sorauf syndrome, 180750 Saethre-Chotzen syndrome with eyelid anomalies, 101400 Saethre-Chotzen syndrome, 101400
TWIST2	131.7	100	99	Ablepharon-macrostomia syndrome, 200110 Barber-Say syndrome, 209885 Focal facial dermal dysplasia 3, Setleis type, 227260
TXNL4A	109.3	100	99	Burn-McKeown syndrome, 608572
TYK2	119.5	99	98	Immunodeficiency 35, 611521
TYMP	95.4	98	84	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYR	185.4	100	100	Albinism, oculocutaneous, type IA, 203100 Albinism, oculocutaneous, type IB, 606952 Waardenburg syndrome/albinism, digenic, 103470 [Skin/hair/eye pigmentation 3, blue/green eyes], 601800 [Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 {Melanoma, cutaneous malignant, susceptibility to, 8}, 601800
TYROBP	95.2	100	99	Nasu-Hakola disease, 221770
TYRP1	176.6	100	99	Albinism, oculocutaneous, type III, 203290 [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271
UBA1	163.3	99	98	Spinal muscular atrophy, X-linked 2, infantile, 301830
UBE2A	94	99	96	Mental retardation, X-linked syndromic, Nascimento-type, 300860
UBE2T	107.1	100	99	Fanconi anemia, complementation group T, 616435
UBE3A	90.1	97	91	Angelman syndrome, 105830
UBE3B	126.7	100	99	Kaufman oculocerebrofacial syndrome, 244450
UBIAD1	249.3	98	95	Corneal dystrophy, Schnyder type, 121800
UBQLN2	137.5	99	98	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857
UBR1	129.8	99	96	Johanson-Blizzard syndrome, 243800

UGT1A1	222.5	100	100	Crigler-Najjar syndrome, type I, 218800 Crigler-Najjar syndrome, type II, 606785 Hyperbilirubinemia, familial transient neonatal, 237900 [Bilirubin, serum level of, QTL1], 601816 [Gilbert syndrome], 143500
UMOD	127.4	97	97	Glomerulocystic kidney disease with hyperuricemia and isosthenuria, 609886 Hyperuricemic nephropathy, familial juvenile 1, 162000 Medullary cystic kidney disease 2, 603860
UMPS	178.4	100	99	Orotic aciduria, 258900
UNC13D	97.1	99	97	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC80	133.4	99	99	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801
UNG	78.6	99	94	Immunodeficiency with hyper IgM, type 5, 608106
UPB1	157.5	100	100	Beta-ureidopropionase deficiency, 613161
UPF3B	47.7	91	77	Mental retardation, X-linked, syndromic 14, 300676
UQCRB	107.4	99	96	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC2	126.7	100	99	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRQ	131.4	100	99	Mitochondrial complex III deficiency, nuclear type 4, 615159
UROD	163.1	99	97	Porphyria cutanea tarda, 176100 Porphyria, hepatoerythropoietic, 176100
UROS	108.2	100	99	Porphyria, congenital erythropoietic, 263700
USB1	125	99	98	Poikiloderma with neutropenia, 604173
USH1C	97.7	100	99	Deafness, autosomal recessive 18A, 602092 Usher syndrome, type 1C, 276904
USH1G	195.6	98	96	Usher syndrome, type 1G, 606943
USH2A	149.1	100	99	Retinitis pigmentosa 39, 613809 Usher syndrome, type 2A, 276901
USP9X	109	97	91	Mental retardation, X-linked 99, 300919 Mental retardation, X-linked 99, syndromic, female-restricted, 300968
USP9Y	30.1	46	39	Spermatogenic failure, Y-linked, 2, 415000
UVSSA	115.3	99	99	UV-sensitive syndrome 3, 614640
VAC14	108.6	99	98	Striatonigral degeneration, childhood-onset, 617054
VAMP1	131.6	100	100	Spastic ataxia 1, autosomal dominant, 108600
VANGL1	165.4	100	100	Caudal regression syndrome, 600145 {Neural tube defects, susceptibility to}, 182940

VANGL2	184.9	100	99	Neural tube defects, 182940
VAPB	107.8	99	95	Amyotrophic lateral sclerosis 8, 608627 Spinal muscular atrophy, late-onset, Finkel type, 182980
VARS2	27.7	25	24	Combined oxidative phosphorylation deficiency 20, 615917
VCAN	186.4	100	100	Wagner syndrome 1, 143200
VCL	115.8	100	99	Cardiomyopathy, dilated, 1W, 611407 Cardiomyopathy, hypertrophic, 15, 613255
VCP	144.9	99	99	Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954 Charcot-Marie-Tooth disease, type 2Y, 616687 Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320
VDR	121.1	98	95	Rickets, vitamin D-resistant, type IIA, 277440 ?Osteoporosis, involutinal, 166710
VEGFC	164.7	100	99	Lymphedema, hereditary, ID, 615907
VHL	119.6	92	85	Erythrocytosis, familial, 2, 263400 Hemangioblastoma, cerebellar, somatic Pheochromocytoma, 171300 Renal cell carcinoma, somatic, 144700 von Hippel-Lindau syndrome, 193300
VIPAS39	144.7	100	100	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VKORC1	162.4	100	100	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473 Warfarin resistance, 122700
VLDLR	201.2	99	99	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VMA21	42.4	95	81	Myopathy, X-linked, with excessive autophagy, 310440
VPS11	167.6	100	99	Leukodystrophy, hypomyelinating, 12, 616683
VPS13A	69.4	95	85	Choreoacanthocytosis, 200150
VPS13B	143.7	98	96	Cohen syndrome, 216550
VPS13C	106.5	96	90	Parkinson disease 23, autosomal recessive, early onset, 616840
VPS33B	138.4	100	100	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
VPS37A	72.6	87	68	Spastic paraplegia 53, autosomal recessive, 614898
VPS45	130.7	96	94	Neutropenia, severe congenital, 5, autosomal recessive, 615285
VPS53	129.2	91	90	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	124.8	97	94	Pontocerebellar hypoplasia type 1A, 607596
VSX1	55.7	85	76	Keratoconus 1, 148300 ?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195

VSX2	77.9	99	97	Microphthalmia with coloboma 3, 610092 Microphthalmia, isolated 2, 610093
VWF	121.2	100	99	von Willebrand disease, type 1, 193400 von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 von Willibrand disease, type 3, 277480
WAC	163.2	99	96	Desanto-Shinawi syndrome, 616708
WAS	66.6	88	78	Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, 313900 Thrombocytopenia, X-linked, intermittent, 313900 Wiskott-Aldrich syndrome, 301000
WDR11	130.7	96	96	Hypogonadotropic hypogonadism 14 with or without anosmia, 614858
WDR19	132	99	98	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WDR34	106.7	99	96	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
WDR35	145.1	99	97	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091
WDR36	112.2	97	90	Glaucoma 1, open angle, G, 609887
WDR45	75.6	97	90	Neurodegeneration with brain iron acculation 5, 300894
WDR60	114.2	99	96	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WDR62	161	100	99	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
WDR72	136.2	99	98	Amelogenesis imperfecta, type IIA3, 613211
WDR73	139.2	100	100	Galloway-Mowat syndrome, 251300
WDR81	163.7	99	99	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185
WFS1	251.6	100	99	Deafness, autosomal dominant 6/14/38, 600965 Wolfram syndrome, 222300 Wolfram-like syndrome, autosomal dominant, 614296 ?Cataract 41, 116400 {Diabetes mellitus, noninsulin-dependent, association with}, 125853
WHSC1L1	125.2	100	99	Leukemia, acute myeloid, 601626
WISP3	115.5	100	100	Arthropathy, progressive pseudorheumatoid, of childhood, 208230 Spondyloepiphyseal dysplasia tarda with progressive arthropathy, 208230
WNK1	167.2	99	99	Neuropathy, hereditary sensory and autonomic, type II, 201300

				Pseudohypoaldosteronism, type IIC, 614492
WNK4	123.2	99	98	Pseudohypoaldosteronism, type IIB, 614491
WNT1	189.2	100	99	Osteogenesis imperfecta, type XV, 615220 {Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221
WNT10A	114.2	100	99	Odontoonychodermal dysplasia, 257980 Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400
WNT10B	145	100	99	Split-hand/foot malformation 6, 225300
WNT4	263.6	93	92	Mullerian aplasia and hyperandrogenism, 158330 SERKAL syndrome, 611812
WNT5A	156.1	100	100	Robinow syndrome, autosomal dominant 1, 180700
WNT7A	217.4	100	100	Fuhrmann syndrome, 228930 Ulna and fibula, absence of, with severe limb deficiency, 276820
WRAP53	154.5	100	100	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	123.6	98	94	Werner syndrome, 277700
WT1	85.3	93	86	Denys-Drash syndrome, 194080 Frasier syndrome, 136680 Meacham syndrome, 608978 Mesothelioma, somatic, 156240 Nephrotic syndrome, type 4, 256370 Wilms tumor, type 1, 194070
WWOX	131.1	100	99	Epileptic encephalopathy, early infantile, 28, 616211 Esophageal squamous cell carcinoma, somatic, 133239 Spinocerebellar ataxia, autosomal recessive 12, 614322
XDH	109.2	100	99	Xanthinuria, type I, 278300
XIAP	107.8	91	86	Lymphoproliferative syndrome, X-linked, 2, 300635
XIST	NC	NC	NC	X-inactivation,familial skewed,300087
XK	97.4	99	99	McLeod syndrome with or without chronic granulomatous disease, 300842
XPA	52.8	98	88	Xeroderma pigmentosum, group A, 278700
XPC	140.8	100	99	Xeroderma pigmentosum, group C, 278720
XPNPEP3	134.8	98	97	Nephronophthisis-like nephropathy 1, 613159
XPR1	131.6	100	99	Basal ganglia calcification, idiopathic, 6, 616413
XRCC4	102.9	99	97	Short stature, microcephaly, and endocrine dysfunction, 616541

XYLT1	132.7	90	87	Desbuquois dysplasia 2, 615777 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
XYLT2	136.5	98	94	Spondyloocular syndrome, 605822 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
YAP1	95.7	87	81	Coloboma, ocular, 120433 Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433
YARS	122.6	100	100	Charcot-Marie-Tooth disease, dominant intermediate C, 608323
YARS2	173.3	99	98	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
ZAP70	186.2	99	99	Autoimmune disease,multisystem,infantile-onset,2,617006 Immunodeficiency 48,269840
ZBTB16	151.4	100	100	Leukemia, acute promyelocytic, PL2F/RARA type Skeletal defects, genital hypoplasia, and mental retardation, 612447
ZBTB20	217.4	100	100	Primrose syndrome, 259050
ZBTB24	177.8	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome-2, 614069
ZC4H2	76.5	99	97	Wieacker-Wolff syndrome, 314580
ZDHC9	55.9	98	89	Mental retardation, X-linked syndromic, Raymond type, 300799
ZEB1	192.5	100	99	Corneal dystrophy, Fuchs endothelial, 6, 613270 Corneal dystrophy, posterior polymorphous, 3, 609141
ZEB2	163.4	100	99	Mowat-Wilson syndrome, 235730
ZFP57	14.2	13	13	Diabetes mellitus, transient neonatal, 1, 601410
ZFPM2	196.4	100	99	46XY sex reversal 9, 616067 Diaphragmatic hernia 3, 610187 Tetralogy of Fallot, 187500
ZFYVE26	120.3	100	99	Spastic paraplegia 15, autosomal recessive, 270700
ZFYVE27	118.7	100	100	Spastic paraplegia 33, autosomal dominant, 610244
ZIC1	231.4	100	100	Craniosynostosis 6, 616602
ZIC2	122.8	90	78	Holoprosencephaly-5, 609637
ZIC3	114.5	100	99	Congenital heart defects, nonsyndromic, 1, X-linked, 306955 Heterotaxy, visceral, 1, X-linked 306955 VACTERL association, X-linked, 314390
ZMPSTE24	113.3	100	99	Mandibuloacral dysplasia with type B lipodystrophy, 608612 Restrictive dermopathy, lethal, 275210
ZMYND10	136.7	100	100	Ciliary dyskinesia, primary, 22, 615444
ZMYND11	136.9	100	99	Mental retardation, autosomal dominant 30, 616083



ZNF408	135.9	100	100	Retinitis pigmentosa 72, 616469 ?Exudative vitreoretinopathy 6, 616468
ZNF41	104.4	100	99	Mental retardation, X-linked 89, 300848
ZNF423	251.1	100	100	Joubert syndrome 19, 614844 Nephronophthisis 14, 614844
ZNF469	93.2	98	96	Brittle cornea syndrome 1, 229200
ZNF513	110.9	100	99	Retinitis pigmentosa 58, 613617
ZNF644	156	100	99	Myopia 21, autosomal dominant, 614167
ZNF674	115.5	100	100	Mental retardation, X-linked 92, 300851
ZNF687	174.6	100	100	Paget disease of bone 6, 616833
ZNF711	138.7	98	95	Mental retardation, X-linked 97, 300803
ZNF750	150.4	100	99	Seborrhea-like dermatitis with psoriasiform elements, 610227
ZNF81	90.9	98	95	Mental retardation, X-linked 45, 300498
ZP1	196.4	100	100	Oocyte maturation defect 1, 615774
ZSWIM6	151	93	89	Acromelic frontonasal dysostosis, 603671

Gene symbols used follow HGNC guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. *Nucleic Acids Res.* 2015 Jan;43(Database issue):D1079-85.

Median Coverage describes the average number of reads seen across 50 exomes.

% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.

% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.

Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.

OMIM release used for OMIM disease identifiers and descriptions : April 14th, 2017.

This list is accurate for panel version DG 2.12

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

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