MENDELIOME GENE PANEL DG 2.15 (3671 genes)

Releasedate: 31-01-2019

Gene	*Median Coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
A4GALT	140.8	100	100	NOR polyagglutination syndrome, 111400 [Blood group, P1Pk system, p phenotype], 111400 [Blood group, P1Pk system, P(2) phenotype], 111400
AAAS	106.4	100	99.7	Achalasia-addisonianism-alacrimia syndrome, 231550
AAGAB	151.8	100	98.3	Keratoderma, palmoplantar, punctate type IA, 148600
AARS	124.3	100	99.6	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 Epileptic encephalopathy, early infantile, 29, 616339
AARS2	126.2	100	99.3	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889
AASS	128.8	99.6	97.4	Hyperlysinemia, 238700 Saccharopinuria, 268700
ABAT	92.7	100	99.5	GABA-transaminase deficiency, 613163
ABCA1	123.6	100	99.5	HDL deficiency, type 2, 604091 Tangier disease, 205400 {Coronary artery disease in familial hypercholesterolemia, protection against}, 143890
ABCA12	140	99.6	97.8	Ichthyosis, congenital, autosomal recessive 4A, 601277 Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500
ABCA3	124	99.9	99.3	Surfactant metabolism dysfunction, pulmonary, 3, 610921
ABCA4	127.1	100	99.5	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
ABCA5	59	93.1	79.6	?Hypertrichosis, congenital generalized, with gingival hyperplasia, 135400

ABCB11	158.2	99.9	99.1	Cholestasis, benign recurrent intrahepatic, 2, 605479 Cholestasis, progressive familial intrahepatic 2, 601847
ABCB4	129.8	99.9	97.9	Cholestasis, intrahepatic, of pregnancy, 3, 614972 Cholestasis, progressive familial intrahepatic 3, 602347 Gallbladder disease 1, 600803
ABCB6	127.2	100	99.6	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	131.5	99.9	98.4	Anemia, sideroblastic, with ataxia, 301310
ABCC2	135.8	100	100	Dubin-Johnson syndrome, 237500
ABCC6	116.4	93.6	92.6	Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850
ABCC8	146.6	100	99.9	Diabetes mellitus, noninsulin-dependent, 125853 Diabetes mellitus, permanent neonatal, 606176 Diabetes mellitus, transient neonatal 2, 610374 Hyperinsulinemic hypoglycemia, familial, 1, 256450 Hypoglycemia of infancy, leucine-sensitive, 240800
ABCC9	157.9	99.9	99.2	Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia, 239850
ABCD1	76	74.7	68	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABCD3	93.7	95.2	89.5	?Bile acid synthesis defect, congenital, 5, 616278
ABCD4	143.6	99.9	98.3	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	145.2	100	99.2	Sitosterolemia, 210250
ABCG8	148.4	99.2	96.6	Sitosterolemia, 210250 {Gallbladder disease 4}, 611465
ABHD12	107	97.3	88	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ABHD5	209.6	100	99.9	Chanarin-Dorfman syndrome, 275630
ABL1	140.2	100	99.9	Congenital heart defects and skeletal malformations syndrome, 617602 Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 0

ACAD8	141.5	100	100	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	135.2	98.4	95.7	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADM	101.3	98.8	95.6	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	123.9	99.3	97.6	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	119.1	99.5	95.6	2-methylbutyrylglycinuria, 610006
ACADVL	118.8	98.7	95.1	VLCAD deficiency, 201475
ACAN	121.6	91.6	85	?Spondyloepimetaphyseal dysplasia, aggrecan type, 612813 ?Spondyloepiphyseal dysplasia, Kimberley type, 608361 Short stature and advanced bone age, with or without early-onset osteoarthritis and/or osteochondritis dissecans, 165800
ACAT1	123.7	99.2	94.6	Alpha-methylacetoacetic aciduria, 203750
ACD	135.2	100	98.2	?Dyskeratosis congenita, autosomal dominant 6, 616553 ?Dyskeratosis congenita, autosomal recessive 7, 616553
ACE	120.7	99.5	97.4	Renal tubular dysgenesis, 267430 [Angiotensin I-converting enzyme, benign serum increase], 0 {Microvascular complications of diabetes 3}, 612624 {Myocardial infarction, susceptibility to}, 0 {SARS, progression of}, 0 {Stroke, hemorrhagic}, 614519
ACER3	105.8	99.9	97.5	?Leukodystrophy, progressive, early childhood-onset, 617762
ACO2	129.3	95.8	91.8	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559
ACOX1	155.3	100	100	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACOX2	136.2	100	99.8	Bile acid synthesis defect, congenital, 6, 617308
ACP4	64.5	90.2	82.7	Amelogenesis imperfecta, type IJ, 617297
ACP5	196.2	100	99.9	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACSF3	128.8	99.9	99.3	Combined malonic and methylmalonic aciduria, 614265
ACSL4	104.7	97.5	91.8	Mental retardation, X-linked 63, 300387
ACSL6	118.5	99.8	98.4	Myelodysplastic syndrome, 0 Myelogenous leukemia, acute, 0

ACTA1	99.7	99.2	95.3	?Myopathy, scapulohumeroperoneal, 616852 Myopathy, actin, congenital, with cores, 161800 Myopathy, actin, congenital, with excess of thin myofilaments, 161800 Myopathy, congenital, with fiber-type disproportion 1, 255310 Nemaline myopathy 3, autosomal dominant or recessive, 161800
ACTA2	137.6	100	99.8	Aortic aneurysm, familial thoracic 6, 611788 Moyamoya disease 5, 614042 Multisystemic smooth muscle dysfunction syndrome, 613834
ACTB	129	99.1	94.2	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACTC1	164.1	100	99.6	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	133.3	99.7	97.6	Visceral myopathy, 155310
ACTN1	143.6	100	99.9	Bleeding disorder, platelet-type, 15, 615193
ACTN2	156.3	100	100	Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158 Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158
ACTN4	143.7	99.5	97.7	Glomerulosclerosis, focal segmental, 1, 603278
ACVR1	165.1	100	100	Fibrodysplasia ossificans progressiva, 135100
ACVR1B	160.3	96.3	95.2	Pancreatic cancer, somatic, 0
ACVR2B	140.5	97.1	94.7	Heterotaxy, visceral, 4, autosomal, 613751
ACVRL1	122.7	99.9	98	Telangiectasia, hereditary hemorrhagic, type 2, 600376
ACY1	132.8	99.9	98.3	Aminoacylase 1 deficiency, 609924
ADA	113	98.9	97.3	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADA2	101.4	99.9	99.1	?Sneddon syndrome, 182410 Polyarteritis nodosa, childhood-onset, 615688
ADAM10	123.6	94.6	92	Reticulate acropigmentation of Kitamura, 615537 {Alzheimer disease 18, susceptibility to}, 615590
ADAM17	139.4	97.6	93.8	?Inflammatory skin and bowel disease, neonatal, 1, 614328
ADAM22	140.5	99.9	98.6	?Epileptic encephalopathy, early infantile, 61, 617933

ADAM9	146.3	98.6	94.1	Cone-rod dystrophy 9, 612775
ADAMTS10	107.8	99.9	98.7	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS13	99.9	96.3	91.7	Thrombotic thrombocytopenic purpura, familial, 274150
ADAMTS17	117.1	88.9	86.7	Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTS18	147.5	99.9	98.9	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
ADAMTS2	117.4	98.5	96.6	Ehlers-Danlos syndrome, dermatosparaxis type, 225410
ADAMTS3	161	100	99.9	?Hennekam lymphangiectasia-lymphedema syndrome 3, 618154
ADAMTSL2	112.2	96.5	91	Geleophysic dysplasia 1, 231050
ADAMTSL4	90.6	99.9	98.8	Ectopia lentis et pupillae, 225200 Ectopia lentis, isolated, autosomal recessive, 225100
ADAR	125	100	99.8	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
ADAT3	82.1	99.5	97.3	Mental retardation, autosomal recessive 36, 615286
ADCY1	145	94.7	93.3	?Deafness, autosomal recessive 44, 610154
ADCY5	129.2	92.3	89.1	Dyskinesia, familial, with facial myokymia, 606703
ADCY6	172.1	100	100	?Lethal congenital contracture syndrome 8, 616287
ADD3	169.7	100	99.7	Cerebral palsy, spastic quadriplegic, 3, 617008
ADGRE2	160.5	96.7	96.1	Vibratory urticaria, 125630
ADGRG1	149.7	100	100	Polymicrogyria, bilateral frontoparietal, 606854 Polymicrogyria, bilateral perisylvian, 615752
ADGRG2	96.5	97	92.3	Congenital bilateral absence of vas deferens, X-linked, 300985
ADGRG6	147.6	99.8	98	Lethal congenital contracture syndrome 9, 616503
ADGRV1	140.3	99.5	97	?Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472
ADIPOQ	139.5	100	100	Adiponectin deficiency, 612556
ADK	100.4	99.5	96.1	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADNP	242.8	100	100	Helsmoortel-van der Aa syndrome, 615873
ADPRHL2	163.7	100	99.9	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
ADRA2B	199.9	100	100	Epilepsy, myoclonic, familial adult, 2, 607876
ADRB2	131.3	100	100	Beta-2-adrenoreceptor agonist, reduced response to, 0 {Asthma, nocturnal, susceptibility to}, 600807 {Obesity, susceptibility to}, 601665

ADSL	183.6	99.2	99.1	Adenylosuccinase deficiency, 103050
ADSSL1	113.6	89.8	85.9	Myopathy, distal, 5, 617030
AEBP1	134.7	99.8	97.9	Ehlers-Danlos syndrome, classic-like, 2, 618000
AFF2	122.8	99.9	98.9	Mental retardation, X-linked, FRAXE type, 309548
AFF4	110.7	99.4	97.3	CHOPS syndrome, 616368
AFG3L2	121	91.9	84.9	Spastic ataxia 5, autosomal recessive, 614487 Spinocerebellar ataxia 28, 610246
AFP	100.2	94.2	84	Alpha-fetoprotein deficiency, 615969 [Hereditary persistence of alpha-fetoprotein], 615970
AGA	130.2	100	100	Aspartylglucosaminuria, 208400
AGBL1	132.3	98.5	98.4	Corneal dystrophy, Fuchs endothelial, 8, 615523
AGBL5	111	100	99.8	Retinitis pigmentosa 75, 617023
AGK	112.1	99.3	96.4	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AGL	146.7	99.7	98	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGPAT2	109.5	99	95.1	Lipodystrophy, congenital generalized, type 1, 608594
AGPS	51.7	96.8	84.8	Rhizomelic chondrodysplasia punctata, type 3, 600121
AGRN	114.8	95.2	89.3	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120
AGT	214.2	100	100	Renal tubular dysgenesis, 267430 {Hypertension, essential, susceptibility to}, 145500 {Preeclampsia, susceptibility to}, 0
AGTR1	134.6	92	91.9	Renal tubular dysgenesis, 267430 {Hypertension, essential}, 145500
AGXT	139.5	99.9	99.2	Hyperoxaluria, primary, type 1, 259900
AHCY	124.5	100	99.8	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHDC1	120.9	98	96.3	Xia-Gibbs syndrome, 615829
AHI1	139.3	99.2	95.1	Joubert syndrome 3, 608629
AHSG	177	100	99.8	?Alopecia-mental retardation syndrome 1, 203650
AICDA	139	89.8	82.6	Immunodeficiency with hyper-IgM, type 2, 605258
AIFM1	106.2	100	99.7	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Deafness, X-linked 5, 300614

AIMP1	84.8	97.3	89.7	Leukodystrophy, hypomyelinating, 3, 260600
AIMP2	119.1	93.6	86.9	Leukodystrophy, hypomyelinating, 17, 618006
AIP	154.2	99.9	99	Pituitary adenoma 1, multiple types, 102200 Pituitary adenoma predisposition, 102000
AIPL1	116	100	99.5	Cone-rod dystrophy, 604393 Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393
AIRE	68.2	98.9	92	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AK1	119.8	100	99.2	Hemolytic anemia due to adenylate kinase deficiency, 612631
AK2	111.8	99.8	96.6	Reticular dysgenesis, 267500
AK7	125.9	99.3	94.5	?Spermatogenic failure 27, 617965
AKAP9	98	98.3	94.2	?Long QT syndrome-11, 611820
AKR1C2	179.5	96.3	89.6	46XY sex reversal 8, 614279
AKR1D1	106.1	98.5	94.3	Bile acid synthesis defect, congenital, 2, 235555
AKT1	156.5	99.9	99.5	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	148.8	100	99.5	Diabetes mellitus, type II, 125853 Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900
AKT3	79.6	97.8	88.6	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937
ALAD	100.6	99.8	97.4	Porphyria, acute hepatic, 612740 {Lead poisoning, susceptibility to}, 612740
ALAS2	89.7	99.6	97.1	Anemia, sideroblastic, 1, 300751 Protoporphyria, erythropoietic, X-linked, 300752
ALB	156	99.9	98.7	Analbuminemia, 616000 [Dysalbuminemic hyperthyroxinemia], 615999
ALDH18A1	131.1	100	99.9	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	104.7	93.4	89.6	Microphthalmia, isolated 8, 615113
ALDH2	124.7	100	99.7	Alcohol sensitivity, acute, 610251 {Esophageal cancer, alcohol-related, susceptibility to}, 0 {Hangover, susceptibility to}, 610251 {Sublingual nitroglycerin, susceptibility to poor response to}, 0
ALDH3A2	125.7	95.3	94.6	Sjogren-Larsson syndrome, 270200
ALDH4A1	116	100	98.6	Hyperprolinemia, type II, 239510
ALDH5A1	87.6	86.4	80.1	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	127.3	100	100	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	77.1	93.7	85.4	Epilepsy, pyridoxine-dependent, 266100
ALDOA	139.1	76.3	74.7	Glycogen storage disease XII, 611881
ALDOB	165.7	100	99.4	Fructose intolerance, hereditary, 229600
ALG1	50.9	53.6	48.8	Congenital disorder of glycosylation, type Ik, 608540
ALG11	139.6	96.7	96	Congenital disorder of glycosylation, type Ip, 613661
ALG12	156.2	100	100	Congenital disorder of glycosylation, type Ig, 607143
ALG13	86.7	98.7	94.1	?Congenital disorder of glycosylation, type Is, 300884 Epileptic encephalopathy, early infantile, 36, 300884
ALG14	233.8	100	100	?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227
ALG2	115.9	100	100	?Congenital disorder of glycosylation, type Ii, 607906 Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228
ALG3	132.9	100	100	Congenital disorder of glycosylation, type Id, 601110
ALG6	96.4	96	93.3	Congenital disorder of glycosylation, type Ic, 603147
ALG8	126	96.5	95.1	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	124.3	100	99.6	Congenital disorder of glycosylation, type Il, 608776 Gillessen-Kaesbach-Nishimura syndrome, 263210
ALMS1	179.8	99.9	99.7	Alstrom syndrome, 203800
ALOX12B	130.6	100	99.5	Ichthyosis, congenital, autosomal recessive 2, 242100

ALOXE3	122.2	100	100	Ichthyosis, congenital, autosomal recessive 3, 606545
ALPK3	98.7	94.6	92.5	Cardiomyopathy, familial hypertrophic 27, 618052
ALPL	156.4	100	100	Hypophosphatasia, adult, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500 Odontohypophosphatasia, 146300
ALS2	170.2	99.9	99.2	Amyotrophic lateral sclerosis 2, juvenile, 205100 Primary lateral sclerosis, juvenile, 606353 Spastic paralysis, infantile onset ascending, 607225
ALX1	153.2	99.9	98.4	?Frontonasal dysplasia 3, 613456
ALX3	102.7	73.3	70.9	Frontonasal dysplasia 1, 136760
ALX4	132.7	98.4	92.5	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597 {Craniosynostosis 5, susceptibility to}, 615529
AMACR	157.9	100	100	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
AMBN	175.1	94.6	88.5	Amelogenesis imperfecta, type IF, 616270
AMELX	98.2	99	95	Amelogenesis imperfecta, type 1E, 301200
AMER1	96.9	99.8	98.9	Osteopathia striata with cranial sclerosis, 300373
AMH	42.1	92.8	74.2	Persistent Mullerian duct syndrome, type I, 261550
AMHR2	158	100	99.5	Persistent Mullerian duct syndrome, type II, 261550
AMMECR1	72.2	99	94	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
AMN	66.8	83.5	71.6	Megaloblastic anemia-1, Norwegian type, 261100
AMPD1	126.7	100	99.9	Myopathy due to myoadenylate deaminase deficiency, 615511
AMPD2	135.5	99.9	99.2	?Spastic paraplegia 63, 615686 Pontocerebellar hypoplasia, type 9, 615809
AMT	173.1	100	100	Glycine encephalopathy, 605899
AMTN	125	98.8	95.4	?Amelogenesis imperfecta, type IIIB, 617607
ANG	178	100	99.9	Amyotrophic lateral sclerosis 9, 611895
ANGPTL3	89.9	97.5	92.2	Hypobetalipoproteinemia, familial, 2, 605019
ANGPTL4	111.9	99	94.2	Plasma triglyceride level QTL, low, 615881
ANK1	136.5	100	99.3	Spherocytosis, type 1, 182900
ANK2	160.3	100	99.9	Cardiac arrhythmia, ankyrin-B-related, 600919

				Long QT syndrome 4, 600919
ANK3	155.1	99.1	98.8	?Mental retardation, autosomal recessive, 37, 615493
ANKH	118.6	100	99.7	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000
ANKLE2	162.5	98	94.7	?Microcephaly 16, primary, autosomal recessive, 616681
ANKRD11	96.3	97.4	94.1	KBG syndrome, 148050
ANKRD26	81	88.7	76.8	Thrombocytopenia 2, 188000
ANKS6	91.8	92.8	88.6	Nephronophthisis 16, 615382
ANLN	146.2	97.2	93.3	Focal segmental glomerulosclerosis 8, 616032
ANO10	116.7	98.8	96.5	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANO3	138.8	99.7	98.2	Dystonia 24, 615034
ANO5	142.2	99.5	95.9	Gnathodiaphyseal dysplasia, 166260 Miyoshi muscular dystrophy 3, 613319 Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307
ANO6	137.4	98	92.9	Scott syndrome, 262890
ANOS1	90.3	89.4	87.6	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
ANTXR1	123	98.3	95.7	GAPO syndrome, 230740 {?Hemangioma, capillary infantile, susceptibility to}, 602089
ANTXR2	100	98.9	94.9	Hyaline fibromatosis syndrome, 228600
ANXA11	80.8	98.9	88.8	Amytrophic lateral sclerosis 23, 617839
AP1S1	111.3	99.9	99.5	MEDNIK syndrome, 609313
AP1S2	65.7	78.6	70.9	Mental retardation, X-linked syndromic 5, 304340
AP2S1	115.7	90.4	89.7	Hypocalciuric hypercalcemia, type III, 600740
AP3B1	95	97.8	90.2	Hermansky-Pudlak syndrome 2, 608233
AP3B2	135.1	97.5	94.2	Epileptic encephalopathy, early infantile, 48, 617276
AP3D1	121	98.1	97.8	?Hermansky-Pudlak syndrome 10, 617050
AP4B1	147.4	100	99.8	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	98.7	99.7	97.9	Spastic paraplegia 51, autosomal recessive, 613744 Stuttering, familial persistent, 1, 184450
AP4M1	127.2	99.1	96.4	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	65.8	71.8	69.3	Spastic paraplegia 52, autosomal recessive, 614067
AP5Z1	96.8	99.7	96.6	Spastic paraplegia 48, autosomal recessive, 613647

APC	159	99.9	98.9	Adenoma, periampullary, somatic, 0 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
APC2	63.5	93.3	85.3	?Sotos syndrome 3, 617169
APCDD1	179.6	100	99.3	Hypotrichosis 1, 605389
APOA1	99.3	100	100	Amyloidosis, 3 or more types, 105200 ApoA-I and apoC-III deficiency, combined, 0 Corneal clouding, autosomal recessive, 0 Hypoalphalipoproteinemia, 604091
APOA2	106	88.5	82.2	Apolipoprotein A-II deficiency, 0 {Hypercholesterolemia, familial, modifier of}, 143890
APOA5	152.5	100	100	Hyperchylomicronemia, late-onset, 144650 {Hypertriglyceridemia, susceptibility to}, 145750
APOB	181.7	99.6	99.3	Hypercholesterolemia, due to ligand-defective apo B, 144010 Hypobetalipoproteinemia, 615558
APOC2	99.4	100	100	Hyperlipoproteinemia, type Ib, 207750
APOC3	88.8	100	100	Apolipoprotein C-III deficiency, 614028
APOE	56.2	93.9	83.1	Alzheimer disease-2, 104310 Hyperlipoproteinemia, type III, 617347 Lipoprotein glomerulopathy, 611771 Sea-blue histiocyte disease, 269600 {?Macular degeneration, age-related}, 603075 {Coronary artery disease, severe, susceptibility to}, 617347
APOPT1	63.8	81.4	78.1	Mitochondrial complex IV deficiency, 220110
APP	138.9	100	100	Alzheimer disease 1, familial, 104300 Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants, 605714
APRT	68.2	100	98.7	Adenine phosphoribosyltransferase deficiency, 614723
APTX	118.9	94.2	91.1	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920

AQP2	117.4	99.5	95.6	Diabetes insipidus, nephrogenic, 125800
AQP5	110.9	99.8	97.4	Palmoplantar keratoderma, Bothnian type, 600231
AR	85.3	93.8	88.3	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
ARCN1	167.9	96.6	96.6	Short stature, rhizomelic, with microcephaly, micrognathia, and developmental delay, 617164
ARF1	179.9	100	100	Periventricular nodular heterotopia 8, 618185
ARFGEF2	154.9	100	99.3	Periventricular heterotopia with microcephaly, 608097
ARG1	167.8	100	100	Argininemia, 207800
ARHGAP26	153.2	100	99.9	Leukemia, juvenile myelomonocytic, somatic, 607785
ARHGAP31	133.4	99.8	98.3	Adams-Oliver syndrome 1, 100300
ARHGDIA	143.9	100	99.9	Nephrotic syndrome, type 8, 615244
ARHGEF10	132.8	99.8	98	?Slowed nerve conduction velocity, AD, 608236
ARHGEF18	114	97.7	93	Retinitis pigmentosa 78, 617433
ARHGEF2	107.6	100	99.7	?Neurodevelopmental disorder with midbrain and hindbrain malformations, 617523
ARHGEF9	60.2	76.4	74.4	Epileptic encephalopathy, early infantile, 8, 300607
ARID1A	150	92.2	89.7	Coffin-Siris syndrome 2, 614607
ARID1B	156.7	94.3	89.5	Coffin-Siris syndrome 1, 135900
ARID2	216.9	99.2	95.7	Coffin-Siris syndrome 6, 617808
ARL13B	97.3	98.9	92.8	Joubert syndrome 8, 612291
ARL2BP	66.3	88.3	79.3	Retinitis pigmentosa with or without situs inversus, 615434
ARL3	85	98.9	93.3	?Retinitis pigmentosa 83, 618173 Joubert syndrome 35, 618161
ARL6	85.2	99.8	95.3	?Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151 {Bardet-Biedl syndrome 1, modifier of}, 209900
ARL6IP1	66	98.8	85.8	?Spastic paraplegia 61, autosomal recessive, 615685
ARMC4	129.1	94.4	93.4	Ciliary dyskinesia, primary, 23, 615451
ARMC5	136.5	99.8	97.5	ACTH-independent macronodular adrenal hyperplasia 2, 615954
ARMC9	127.3	99.8	98.5	Joubert syndrome 30, 617622
ARNT2	153	99.6	99	?Webb-Dattani syndrome, 615926

ARPC1B	126.4	100	99.9	Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease, 617718
ARR3	98.1	100	99.9	Myopia 26, X-linked, female-limited, 301010
ARSA	97.8	100	99.7	Metachromatic leukodystrophy, 250100
ARSB	117.5	94.9	87.7	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARSE	102.1	99.2	93	Chondrodysplasia punctata, X-linked recessive, 302950
ARSG	141.1	100	99.4	Usher syndrome, type IV, 618144
ARV1	133.5	100	99.3	Epileptic encephalopathy, early infantile, 38, 617020
ARX	29.1	75.8	59.5	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	105.9	97.6	92.1	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASB10	101.6	99.6	96.8	Glaucoma 1, open angle, F, 603383
ASCC1	139.1	96.7	93.2	?Spinal muscular atrophy with congenital bone fractures 2, 616867 Barrett esophagus/esophageal adenocarcinoma, 614266
ASCL1	213.1	90.2	79.5	Central hypoventilation syndrome, congenital, 209880 Haddad syndrome, 209880
ASH1L	160.1	98.7	98.5	Mental retardation, autosomal dominant 52, 617796
ASL	114.4	99.9	98.8	Argininosuccinic aciduria, 207900
ASNS	105.8	97.8	90.5	Asparagine synthetase deficiency, 615574
ASPA	127.6	99.1	95.8	Canavan disease, 271900
ASPH	117.3	98.8	93.9	Traboulsi syndrome, 601552
ASPM	101.2	98	92.2	Microcephaly 5, primary, autosomal recessive, 608716
ASPSCR1	88.4	98.4	93.7	Alveolar soft-part sarcoma, 606243
ASS1	97.9	95.7	87.5	Citrullinemia, 215700
ASXL1	159.8	99.1	97.7	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ASXL2	152	98.1	98	Shashi-Pena syndrome, 617190
ASXL3	162.9	99.8	98.9	Bainbridge-Ropers syndrome, 615485
ATAD1	59.1	94.9	86.4	Hyperekplexia 4, 618011

ATAD3A	87.9	89	86.2	Harel-Yoon syndrome, 617183
ATCAY	146.1	100	99.7	Ataxia, cerebellar, Cayman type, 601238
ATF6	134.1	100	99.6	Achromatopsia 7, 616517
ATG5	130.5	98.6	90.5	?Spinocerebellar ataxia, autosomal recessive 25, 617584
ATIC	119.5	99.7	99	AICA-ribosiduria due to ATIC deficiency, 608688
ATL1	161	99.7	97.9	Neuropathy, hereditary sensory, type ID, 613708 Spastic paraplegia 3A, autosomal dominant, 182600
ATL3	125	98.1	93.8	Neuropathy, hereditary sensory, type IF, 615632
ATM	109.7	99	94	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic, 0 Lymphoma, mantle cell, somatic, 0 T-cell prolymphocytic leukemia, somatic, 0 {Breast cancer, susceptibility to}, 114480
ATN1	120.9	99.7	97.7	Dentatorubro-pallidoluysian atrophy, 125370
ATOH7	102.8	95.8	89.6	Persistent hyperplastic primary vitreous, autosomal recessive, 221900
ATP11C	71.9	95.9	86.9	?Hemolytic anemia, congenital, X-linked, 301015
ATP13A2	117.4	100	98.8	Kufor-Rakeb syndrome, 606693 Spastic paraplegia 78, autosomal recessive, 617225
ATP1A1	142.9	100	99.9	Charcot-Marie-Tooth disease, axonal, type 2DD, 618036
ATP1A2	190.8	100	99.6	Alternating hemiplegia of childhood 1, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481
ATP1A3	177.3	100	100	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235
ATP2A1	155.9	100	100	Brody myopathy, 601003
ATP2A2	175.2	100	99.9	Acrokeratosis verruciformis, 101900 Darier disease, 124200
ATP2B3	135.2	99.5	97.7	?Spinocerebellar ataxia, X-linked 1, 302500
ATP2C1	118.6	99.9	99	Hailey-Hailey disease, 169600

ATP5A1	85.3	94.8	85.8	?Combined oxidative phosphorylation deficiency 22, 616045 ?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4, 615228
ATP5D	66.1	98.8	90.3	Mitochondrial complex V (ATP synthase) deficiency, 618120
ATP5E	135.5	100	100	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053
ATP6AP1	112.8	99.1	94.7	Immunodeficiency 47, 300972
ATP6AP2	46.1	81.2	55.6	?Parkinsonism with spasticity, X-linked, 300911 Mental retardation, X-linked, syndromic, Hedera type, 300423
ATP6V0A2	130	100	99.3	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250
ATP6V0A4	116.4	99.9	98.6	Renal tubular acidosis, distal, autosomal recessive, 602722
ATP6V1A	144.8	98.2	94.9	Cutis laxa, autosomal recessive, type IID, 617403 Epileptic encephalopathy, infantile or early childhood, 3, 618012
ATP6V1B1	176.6	100	100	Renal tubular acidosis with deafness, 267300
ATP6V1B2	137	99.9	98.2	Deafness, congenital, with onychodystrophy, autosomal dominant, 124480 Zimmermann-Laband syndrome 2, 616455
ATP6V1E1	67.2	92.2	85.6	Cutis laxa, autosomal recessive, type IIC, 617402
ATP7A	133.2	99.7	97.8	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATP7B	168.9	100	99.8	Wilson disease, 277900
ATP8A2	133.5	100	99.5	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268
ATP8B1	139	96.7	94.4	Cholestasis, benign recurrent intrahepatic, 243300 Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, progressive familial intrahepatic 1, 211600
ATPAF2	101.4	100	100	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
ATR	138.3	99.4	96.9	?Cutaneous telangiectasia and cancer syndrome, familial, 614564 Seckel syndrome 1, 210600
ATRX	82.6	98.2	92.2	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Alpha-thalassemia/mental retardation syndrome, 301040 Mental retardation-hypotonic facies syndrome, X-linked, 309580
ATXN1	159.7	100	100	Spinocerebellar ataxia 1, 164400

ATXN10	137.7	99.8	97.9	Spinocerebellar ataxia 10, 603516
ATXN2	89.3	85.8	77.8	Spinocerebellar ataxia 2, 183090 {Amyotrophic lateral sclerosis, susceptibility to, 13}, 183090 {Parkinson disease, late-onset, susceptibility to}, 168600
ATXN3	98.3	91.1	86.2	Machado-Joseph disease, 109150
ATXN7	131	96.6	94.1	Spinocerebellar ataxia 7, 164500
ATXN8OS	NC	NC	NC	Spinocerebellar ataxia 8, 608768
AUH	90.9	99.9	97.6	3-methylglutaconic aciduria, type I, 250950
AURKC	79	99.8	97.5	Spermatogenic failure 5, 243060
AUTS2	110.3	96.9	95.5	Mental retardation, autosomal dominant 26, 615834
AVP	53.3	76.3	57.3	Diabetes insipidus, neurohypophyseal, 125700
AVPR2	127.5	99.3	97.1	Diabetes insipidus, nephrogenic, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539
AXIN1	141.2	98.6	96.8	?Caudal duplication anomaly, 607864 Hepatocellular carcinoma, somatic, 114550
AXIN2	114.5	99.7	98.9	Colorectal cancer, somatic, 114500 Oligodontia-colorectal cancer syndrome, 608615
B2M	252.1	100	99.9	?Amyloidosis, familial visceral, 105200 Immunodeficiency 43, 241600
B3GALNT2	115	92.4	89.7	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B3GALT6	47.5	76.4	71.7	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B3GAT3	93.6	99.4	95.9	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B3GLCT	101.2	97.4	93.4	Peters-plus syndrome, 261540
B4GALNT1	151	95.6	90.1	Spastic paraplegia 26, autosomal recessive, 609195
B4GALT1	105.4	99.9	99	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	104.3	96.1	95	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
B4GAT1	120.4	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
B9D1	115.3	92.1	91.4	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120

B9D2	110.9	100	100	?Meckel syndrome 10, 614175 Joubert syndrome 34, 614175
BAAT	121.4	98.3	95.3	Hypercholanemia, familial, 607748
BAG3	136.5	100	100	Cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954
BANF1	58.3	98	88.1	Nestor-Guillermo progeria syndrome, 614008
BAP1	111	85.1	82.7	Tumor predisposition syndrome, 614327
BAX	92.9	96.9	95.3	Colorectal cancer, somatic, 114500 T-cell acute lymphoblastic leukemia, somatic, 613065
BBIP1	132	99.4	94.8	?Bardet-Biedl syndrome 18, 615995
BBS1	148.9	100	100	Bardet-Biedl syndrome 1, 209900
BBS10	172.6	100	100	Bardet-Biedl syndrome 10, 615987
BBS12	208.6	100	100	Bardet-Biedl syndrome 12, 615989
BBS2	181.8	100	99.8	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	135.9	99.7	97.3	Bardet-Biedl syndrome 4, 615982
BBS5	81.1	95.8	84.1	Bardet-Biedl syndrome 5, 615983
BBS7	120.7	98.1	91.7	Bardet-Biedl syndrome 7, 615984
BBS9	112.9	96	93.8	Bardet-Biedl syndrome 9, 615986
BCAP31	70.6	93.1	82.5	Deafness, dystonia, and cerebral hypomyelination, 300475
BCHE	162.9	100	99.8	Butyrylcholinesterase deficiency, 617936 {Apnea, postanesthetic, susceptibility to, due to BCHE deficiency}, 617936
BCKDHA	171.5	100	99.5	Maple syrup urine disease, type Ia, 248600
BCKDHB	112.6	88.9	81.3	Maple syrup urine disease, type Ib, 248600
BCKDK	178.8	100	100	Branched-chain ketoacid dehydrogenase kinase deficiency, 614923
BCL10	97.9	100	99.8	?Immunodeficiency 37, 616098 Lymphoma, MALT, somatic, 137245 {Lymphoma, follicular, somatic}, 605027 {Male germ cell tumor, somatic},, 273300 {Mesothelioma, somatic}, 156240 {Sezary syndrome, somatic}, 0
BCL11A	139.3	98.2	97	Dias-Logan syndrome, 617101

BCL11B	79.7	96.6	88.6	Immunodeficiency 49, 617237 Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092
BCL2	156	99.9	98.4	Leukemia/lymphoma, B-cell, 2, 0
BCL7A	136	100	100	B-cell non-Hodgkin lymphoma, high-grade, 0
BCO1	164.9	100	100	?Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300
BCOR	109.7	99.3	96.8	Microphthalmia, syndromic 2, 300166
BCR	110.6	87.7	84.3	Leukemia, acute lymphocytic, somatic, 613065 Leukemia, chronic myeloid, somatic, 608232
BCS1L	182.3	100	100	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BDP1	125.5	95.6	90	?Deafness, autosomal recessive 112, 618257
BEAN1	146.2	99.1	95.3	Spinocerebellar ataxia 31, 117210
BEST1	144.6	99.6	97.5	Bestrophinopathy, autosomal recessive, 611809 Macular dystrophy, vitelliform, 2, 153700 Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma, 193220 Retinitis pigmentosa, concentric, 613194 Retinitis pigmentosa-50, 613194 Vitreoretinochoroidopathy, 193220
BFSP1	98	98.2	88.9	Cataract 33, multiple types, 611391
BFSP2	89.5	99.8	97.6	Cataract 12, multiple types, 611597
BGN	128.9	100	99.5	Meester-Loeys syndrome, 300989 Spondyloepimetaphyseal dysplasia, X-linked, 300106
BHLHA9	13.8	57.8	41.3	?Camptosynpolydactyly, complex, 607539 Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432
BICD2	158.6	100	99.9	Spinal muscular atrophy, lower extremity-predominant, 2, AD, 615290
BIN1	100.2	99.1	95.3	Centronuclear myopathy 2, 255200
BLK	115.7	100	100	Maturity-onset diabetes of the young, type 11, 613375
BLM	116.3	99.4	96.5	Bloom syndrome, 210900
BLNK	95.7	93.7	91.3	?Agammaglobulinemia 4, 613502

BLOC1S3	28.7	88.7	65.3	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	97.2	98.7	91.3	?Hermansky-pudlak syndrome 9, 614171
BLVRA	125.1	100	99.7	Hyperbiliverdinemia, 614156
BMP1	143.9	99.9	99.1	Osteogenesis imperfecta, type XIII, 614856
BMP15	120.5	100	99.4	Ovarian dysgenesis 2, 300510 Premature ovarian failure 4, 300510
BMP2	173.4	100	99.9	Brachydactyly, type A2, 112600 Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 617877 {HFE hemochromatosis, modifier of}, 235200
BMP4	151.7	100	99.9	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625
BMPER	159.9	99.9	99	Diaphanospondylodysostosis, 608022
BMPR1A	98	99.7	94.2	Juvenile polyposis syndrome, infantile form, 174900 Polyposis syndrome, hereditary mixed, 2, 610069 Polyposis, juvenile intestinal, 174900
BMPR1B	172.4	100	98.9	Acromesomelic dysplasia, Demirhan type, 609441 Brachydactyly, type A1, D, 616849 Brachydactyly, type A2, 112600
BMPR2	194.7	99.9	99.7	Pulmonary hypertension, familial primary, 1, with or without HHT, 178600 Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600 Pulmonary venoocclusive disease 1, 265450
BMS1	96.6	66.7	65.6	?Aplasia cutis congenita, nonsyndromic, 107600
BOLA3	50.1	92.3	81.7	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BPGM	130.8	100	100	Erythrocytosis due to bisphosphoglycerate mutase deficiency, 222800
BPTF	177.7	96.1	95.1	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, 617755
BRAF	74.4	87.6	77.2	Adenocarcinoma of lung, somatic, 211980 Cardiofaciocutaneous syndrome, 115150 Colorectal cancer, somatic, 0 LEOPARD syndrome 3, 613707 Melanoma, malignant, somatic, 0 Nonsmall cell lung cancer, somatic, 0

				Noonan syndrome 7, 613706
BRAT1	108.5	99.8	97.4	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056 Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BRCA1	177.5	98.9	96.9	Fanconi anemia, complementation group S, 617883 {Breast-ovarian cancer, familial, 1}, 604370 {Pancreatic cancer, susceptibility to, 4}, 614320
BRCA2	102.7	99	97.4	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
BRDT	100.3	92.6	86.5	?Spermatogenic failure 21, 617644
BRF1	100	96.6	92.9	Cerebellofaciodental syndrome, 616202
BRIP1	117.8	99.8	97.7	Breast cancer, early-onset, 114480 Fanconi anemia, complementation group J, 609054
BRPF1	179.1	100	99.5	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333
BRWD3	97.1	97	92.3	Mental retardation, X-linked 93, 300659
BSCL2	113.5	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	137.1	100	100	Bartter syndrome, type 4a, 602522 Sensorineural deafness with mild renal dysfunction, 602522
BTD	166.6	100	99.9	Biotinidase deficiency, 253260

BTK	116.2	100	99.6	Agammaglobulinemia, X-linked 1, 300755 Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200
BUB1	136.4	99.8	97.9	Colorectal cancer with chromosomal instability, somatic, 0
BUB1B	136.5	98.6	97.9	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430
BVES	115	99.8	98.2	?Cardiac arrhythmia with increased serum creatine kinase, 616812
C11orf70	60.7	92.3	82.2	Ciliary dyskinesia, primary, 38, 618063
C12orf4	131.3	99.1	94.7	Mental retardation, autosomal recessive 66, 618221
C12orf57	152	100	100	Temtamy syndrome, 218340
C12orf65	88.2	97.3	91.9	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035
C15orf41	124.9	99.9	97.9	Dyserythropoietic anemia, congenital, type Ib, 615631
C19orf12	93.9	100	99.7	?Spastic paraplegia 43, autosomal recessive, 615043 Neurodegeneration with brain iron accumulation 4, 614298
C1GALT1C1	147.2	99.8	98.7	Tn polyagglutination syndrome, somatic, 300622
C1QA	120.4	100	99	C1q deficiency, 613652
C1QB	183.4	100	99.9	C1q deficiency, 613652
C1QBP	80.7	81.7	71	Combined oxidative phosphorylation deficiency 33, 617713
C1QC	198.1	100	98.9	C1q deficiency, 613652
C1QTNF5	151.2	79.7	65.5	Retinal degeneration, late-onset, autosomal dominant, 605670
C1R	156.9	100	100	Ehlers-Danlos syndrome, periodontal type, 1, 130080
C1S	117.6	100	99.7	C1s deficiency, 613783 Ehlers-Danlos syndrome, periodontal type, 2, 617174
C2	129.9	100	100	C2 deficiency, 217000 {Macular degeneration, age-related, 14, reduced risk of}, 615489
C21orf2	104.4	99.9	98.7	Retinal dystrophy with macular staphyloma, 617547 Spondylometaphyseal dysplasia, axial, 602271
C21orf59	145.8	98.7	94.6	Ciliary dyskinesia, primary, 26, 615500
C2CD3	143.1	95.8	95.6	?Orofaciodigital syndrome XIV, 615948
C2orf71	124.6	99.7	98.8	Retinitis pigmentosa 54, 613428

C3	145.5	100	99.7	C3 deficiency, 613779 {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 {Macular degeneration, age-related, 9}, 611378
C4A	91.5	98.1	95.9	C4a deficiency, 614380 [Blood group, Rodgers], 614374
C4B	90.5	98.5	96.5	C4B deficiency, 614379
C4orf26	197.6	100	100	Amelogenesis imperfecta, type IIA4, 614832
C5	134.4	98.4	95.3	C5 deficiency, 609536 [Eculizumab, poor response to], 615749
C5orf42	122.8	98.6	95.5	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
C6	157.5	100	99.9	C6 deficiency, 612446 Combined C6/C7 deficiency, 0
C7	132.3	99.1	94.8	C7 deficiency, 610102
C8A	120.2	100	99.8	C8 deficiency, type I, 613790
C8B	135.8	99.9	99.5	C8 deficiency, type II, 613789
C8orf37	126.4	100	99	Bardet-Biedl syndrome 21, 617406 Cone-rod dystrophy 16, 614500 Retinitis pigmentosa 64, 614500
C9	133.7	100	98.5	C9 deficiency, 613825 {Macular degeneration, age-related, 15, susceptibility to}, 615591
C9orf72	108.5	99	94.2	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1, 105550
CA12	109.6	100	100	Hyperchlorhidrosis, isolated, 143860
CA2	140.7	100	99.3	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA4	142.4	100	99.9	Retinitis pigmentosa 17, 600852
CA5A	124.1	99.5	94.9	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CA8	114.6	96.8	93	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CABP2	67.1	74.6	65.5	Deafness, autosomal recessive 93, 614899
CABP4	98.4	99.7	97.7	Cone-rod synaptic disorder, congenital nonprogressive, 610427

CACNA1A	87.8	92.7	89.1	Epileptic encephalopathy, early infantile, 42, 617106 Episodic ataxia, type 2, 108500 Migraine, familial hemiplegic, 1, 141500 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Spinocerebellar ataxia 6, 183086
CACNA1B	137.5	94.1	91.5	?Dystonia 23, 614860
CACNA1C	154.6	99.9	99.2	Brugada syndrome 3, 611875 Timothy syndrome, 601005
CACNA1D	149.9	98	97.8	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896
CACNA1F	99.4	99.8	97.9	Aland Island eye disease, 300600 Cone-rod dystrophy, X-linked, 3, 300476 Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071
CACNA1G	132.8	99	97.5	Spinocerebellar ataxia 42, 616795 Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087
CACNA1H	103.4	97.6	94.3	Hyperaldosteronism, familial, type IV, 617027 {Epilepsy, childhood absence, susceptibility to, 6}, 611942 {Epilepsy, idiopathic generalized, susceptibility to, 6}, 611942
CACNA1S	135.5	100	99.7	Hypokalemic periodic paralysis, type 1, 170400 {Malignant hyperthermia susceptibility 5}, 601887 {Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580
CACNA2D4	112	99.2	97.7	Retinal cone dystrophy 4, 610478
CACNB2	150.9	99.5	96.9	Brugada syndrome 4, 611876
CACNB4	106.1	96.3	94.6	Episodic ataxia, type 5, 613855 {Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 {Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682
CACNG2	114.9	100	99.9	?Mental retardation, autosomal dominant 10, 614256
CAD	158.9	100	99.7	Epileptic encephalopathy, early infantile, 50, 616457
CALM1	114.3	100	99.7	Long QT syndrome 14, 616247 Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916
CALM2	54	67.8	65.8	Long QT syndrome 15, 616249
CALR	113.5	99.9	97.4	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950

CAMK2A	123.2	100	99.9	?Mental retardation, autosomal recessive 63, 618095 Mental retardation, autosomal dominant 53, 617798
CAMK2B	103.4	98.7	93.7	Mental retardation, autosomal dominant 54, 617799
CAMTA1	185.9	99.6	98.8	Cerebellar ataxia, nonprogressive, with mental retardation, 614756
CANT1	142.1	100	99.8	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719
CAPN1	146	100	100	Spastic paraplegia 76, autosomal recessive, 616907
CAPN3	111.4	99	96.7	Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129 Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600
CAPN5	166.1	100	99.9	Vitreoretinopathy, neovascular inflammatory, 193235
CARD11	154.6	99.9	98.6	B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11A, 615206 Immunodeficiency 11B with atopic dermatitis, 617638
CARD14	116.2	99.7	97.7	Pityriasis rubra pilaris, 173200 Psoriasis 2, 602723
CARD9	119.7	98.3	96.4	Candidiasis, familial, 2, autosomal recessive, 212050
CARMIL2	121.1	94.9	92.7	Immunodeficiency 58, 618131
CARS2	121.1	100	99.8	Combined oxidative phosphorylation deficiency 27, 616672
CASK	92.3	98.7	93.7	FG syndrome 4, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 Mental retardation, with or without nystagmus, 300422
CASP10	117.4	99.5	98	Autoimmune lymphoproliferative syndrome, type II, 603909 Gastric cancer, somatic, 613659 Lymphoma, non-Hodgkin, somatic, 605027
CASP14	85.5	100	100	Ichthyosis, congenital, autosomal recessive 12, 617320
CASP8	144.8	95.6	95.5	?Autoimmune lymphoproliferative syndrome, type IIB, 607271 Hepatocellular carcinoma, somatic, 114550 {Breast cancer, protection against}, 114480 {Lung cancer, protection against}, 211980
CASQ1	122	100	99.6	Myopathy, vacuolar, with CASQ1 aggregates, 616231
CASQ2	143.3	99.9	99.2	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938

CASR	178	100	99.7	Hyperparathyroidism, neonatal, 239200 Hypocalcemia, autosomal dominant, 601198 Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 Hypocalciuric hypercalcemia, type I, 145980 {Epilepsy idiopathic generalized, susceptibility to, 8}, 612899
CAST	110.2	96.8	92.8	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295
CAT	148.5	100	100	Acatalasemia, 614097
CATSPER1	147.2	100	99.4	Spermatogenic failure 7, 612997
CAV1	265.4	100	100	?Lipodystrophy, congenital generalized, type 3, 612526 ?Partial lipodystrophy, congenital cataracts, and neurodegeneration syndrome, 606721 Pulmonary hypertension, primary, 3, 615343
CAV3	304.7	100	100	Cardiomyopathy, familial hypertrophic, 192600 Creatine phosphokinase, elevated serum, 123320 Long QT syndrome 9, 611818 Myopathy, distal, Tateyama type, 614321 Rippling muscle disease, 606072
CAVIN1	137	99.9	99.3	Lipodystrophy, congenital generalized, type 4, 613327
CBL	129.8	96.9	95.7	?Juvenile myelomonocytic leukemia, 607785 Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563
CBS	116.2	97.1	91.1	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CBX2	96.6	99.9	98.4	?46XY sex reversal 5, 613080
CC2D1A	119.4	99.8	98.3	Mental retardation, autosomal recessive 3, 608443
CC2D2A	127.4	99.5	97.1	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284
CCBE1	75.9	98.9	95.5	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CCDC103	116.8	100	99	Ciliary dyskinesia, primary, 17, 614679
CCDC114	120.7	100	99.6	Ciliary dyskinesia, primary, 20, 615067
CCDC115	59.3	88	85.3	Congenital disorder of glycosylation, type IIo, 616828
CCDC151	116.2	100	99.7	Ciliary dyskinesia, primary, 30, 616037
CCDC174	133.1	98.1	93.3	Hypotonia, infantile, with psychomotor retardation, 616816

CCDC22	93.3	97.3	89.4	Ritscher-Schinzel syndrome 2, 300963
CCDC39	74.3	96.6	88.9	Ciliary dyskinesia, primary, 14, 613807
CCDC40	126.5	98.9	97.8	Ciliary dyskinesia, primary, 15, 613808
CCDC50	135.1	99.9	98.9	?Deafness, autosomal dominant 44, 607453
CCDC65	105.9	99.7	97.6	Ciliary dyskinesia, primary, 27, 615504
CCDC78	114.9	100	100	?Centronuclear myopathy 4, 614807
CCDC8	111.9	100	100	3-M syndrome 3, 614205
CCDC88A	78.9	94.7	84.9	?PEHO syndrome-like, 617507
CCDC88C	101.4	99.8	97.4	?Spinocerebellar ataxia 40, 616053 Hydrocephalus, congenital, 1, 236600
CCM2	160	97.9	97.9	Cerebral cavernous malformations-2, 603284
CCND2	152.3	100	100	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938
CCNK	82.4	88	76.6	?Intellectual developmental disorder with hypertelorism and distinctive facies, 618147
CCNO	103.1	99	95.6	Ciliary dyskinesia, primary, 29, 615872
CCT5	164.5	99.9	99.1	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CD151	132.3	100	100	Nephropathy with pretibial epidermolysis bullosa and deafness, 609057 [Blood group, Raph], 179620
CD164	130.4	98	93.9	?Deafness, autosomal dominant 66, 616969
CD19	88.8	99.9	98.4	Immunodeficiency, common variable, 3, 613493
CD247	101.6	100	98.9	?Immunodeficiency 25, 610163
CD27	118.1	100	99.6	Lymphoproliferative syndrome 2, 615122
CD2AP	98.2	99.6	96	Glomerulosclerosis, focal segmental, 3, 607832
CD320	92	100	99.5	Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646
CD36	123.3	99.2	95.7	Platelet glycoprotein IV deficiency, 608404 [Macrothrombocytopenia], 0 {Coronary heart disease, susceptibility to, 7}, 610938 {Malaria, cerebral, reduced risk of}, 611162 {Malaria, cerebral, susceptibility to}, 611162
CD3D	193.8	100	100	Immunodeficiency 19, 615617
CD3E	152.1	100	99.9	Immunodeficiency 18, 615615 Immunodeficiency 18, SCID variant, 615615

CD3G	156.8	100	100	Immunodeficiency 17, CD3 gamma deficient, 615607
CD4	119.9	100	99.7	OKT4 epitope deficiency, 613949
CD40	165.4	100	99.9	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	126.6	95.9	86.8	Immunodeficiency, X-linked, with hyper-IgM, 308230
CD55	138.9	94.4	86.2	Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300 [Blood group Cromer], 613793
CD59	200.9	93.6	86.5	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD79A	128.3	99.8	97	Agammaglobulinemia 3, 613501
CD79B	210.7	100	100	Agammaglobulinemia 6, 612692
CD81	142.6	99.9	98.1	Immunodeficiency, common variable, 6, 613496
CD8A	110	99.9	99	CD8 deficiency, familial, 608957
CD96	156.8	100	99.9	C syndrome, 211750
CDAN1	97.6	97.6	95.2	Dyserythropoietic anemia, congenital, type Ia, 224120
CDC14A	161.4	98.3	93.5	Deafness, autosomal recessive 32, with or without immotile sperm, 608653
CDC42	97	96.7	89.5	Takenouchi-Kosaki syndrome, 616737
CDC45	160.7	99.4	97.5	Meier-Gorlin syndrome 7, 617063
CDC6	165.4	99.8	98.3	?Meier-Gorlin syndrome 5, 613805
CDC73	102.7	99.8	97.7	Hyperparathyroidism, familial primary, 145000 Hyperparathyroidism-jaw tumor syndrome, 145001 Parathyroid adenoma with cystic changes, 145001 Parathyroid carcinoma, 608266
CDCA7	109.2	100	99.3	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910
CDH1	125.3	99.1	98.4	Blepharocheilodontic syndrome 1, 119580 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
CDH11	154.2	100	100	Elsahy-Waters syndrome, 211380
CDH15	120.6	99.8	97.2	Mental retardation, autosomal dominant 3, 612580

CDH23	197.2	100	100	Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D, 601067 Usher syndrome, type 1D/F digenic, 601067 {Pituitary adenoma 5, multiple types}, 617540
CDH3	159.3	99.5	97.3	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 Hypotrichosis, congenital, with juvenile macular dystrophy, 601553
CDHR1	154.2	99.2	98	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660
CDK10	114.6	100	99.9	Al Kaissi syndrome, 617694
CDK13	136.6	95.4	88.1	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360
CDK5	110.4	99.9	99.1	?Lissencephaly 7 with cerebellar hypoplasia, 616342
CDK5RAP2	123.9	99.9	98.7	Microcephaly 3, primary, autosomal recessive, 604804
CDK6	105.9	99.4	95.7	?Microcephaly 12, primary, autosomal recessive, 616080
CDKL5	114.4	94.9	91.8	Epileptic encephalopathy, early infantile, 2, 300672
CDKN1B	93.2	100	99.5	Multiple endocrine neoplasia, type IV, 610755
CDKN1C	21.1	68.1	51.8	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732
CDKN2A	79.6	92.2	91.4	Melanoma and neural system tumor syndrome, 155755 Orolaryngeal cancer, multiple, 0 Pancreatic cancer/melanoma syndrome, 606719 {Melanoma, cutaneous malignant, 2}, 155601
CDON	143.8	100	99.6	Holoprosencephaly 11, 614226
CDSN	119.3	100	99.5	Hypotrichosis 2, 146520 Peeling skin syndrome 1, 270300
CDT1	99.3	96.8	93.8	Meier-Gorlin syndrome 4, 613804
CEACAM16	144.9	100	99.7	Deafness, autosomal dominant 4B, 614614
CEBPA	46.7	75.5	65.1	?Leukemia, acute myeloid, 601626 Leukemia, acute myeloid, somatic, 601626
CEBPE	71.1	99.3	95.8	Specific granule deficiency, 245480
CEL	130	85	82.7	Maturity-onset diabetes of the young, type VIII, 609812
CENPE	61.5	95	83.3	?Microcephaly 13, primary, autosomal recessive, 616051
CENPF	139.5	99.5	97.6	Stromme syndrome, 243605

CENPJ	141.7	99.7	97.8	?Seckel syndrome 4, 613676 Microcephaly 6, primary, autosomal recessive, 608393
CEP104	119.9	99	97.9	Joubert syndrome 25, 616781
CEP120	129.7	99.8	98.1	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP135	79.2	98.1	89.1	Microcephaly 8, primary, autosomal recessive, 614673
CEP152	162.5	97.2	94.5	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823
CEP164	94.2	99.9	98	Nephronophthisis 15, 614845
CEP19	202.7	100	100	Morbid obesity and spermatogenic failure, 615703
CEP290	66.1	88.4	76.7	?Bardet-Biedl syndrome 14, 615991 Joubert syndrome 5, 610188 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Senior-Loken syndrome 6, 610189
CEP41	83.5	97.7	89.6	Joubert syndrome 15, 614464
CEP55	129.5	100	99.9	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500
CEP57	92.6	99.4	93.3	Mosaic variegated aneuploidy syndrome 2, 614114
CEP63	119.4	96.7	89.9	?Seckel syndrome 6, 614728
CEP78	112.1	97	93.9	Cone-rod dystrophy and hearing loss, 617236
CEP83	96.7	98.3	89.1	Nephronophthisis 18, 615862
CERKL	100.4	98.6	92.8	Retinitis pigmentosa 26, 608380
CERS1	55.7	70.5	60.5	?Epilepsy, progressive myoclonic, 8, 616230
CERS3	106.8	100	98.8	Ichthyosis, congenital, autosomal recessive 9, 615023
CES1	155.1	99.8	98.7	Drug metabolism, altered, CES1-related, 618057
CETP	131.9	100	100	Hyperalphalipoproteinemia, 143470 [High density lipoprotein cholesterol level QTL 10], 143470
CFAP43	123.7	99.4	96.1	Spermatogenic failure 19, 617592
CFAP44	104	99.4	97.1	?Spermatogenic failure 20, 617593
CFAP53	146.6	97.6	94.2	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFAP69	58.4	96.2	86.6	Spermatogenic failure 24, 617959
CFB	147.1	100	100	?Complement factor B deficiency, 615561 {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924

				{Macular degeneration, age-related, 14, reduced risk of}, 615489
CFC1	74.5	82.7	71.3	Heterotaxy, visceral, 2, autosomal, 605376
CFD	80.6	89.7	81.6	Complement factor D deficiency, 613912
CFH	183.2	98.7	95.3	Basal laminar drusen, 126700 Complement factor H deficiency, 609814 {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 {Macular degeneration, age-related, 4}, 610698
CFHR5	97.3	98.7	93.4	Nephropathy due to CFHR5 deficiency, 614809
CFI	145.5	96.6	92.8	Complement factor I deficiency, 610984 {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 {Macular degeneration, age-related, 13, susceptibility to}, 615439
CFL2	117.3	94.3	86.7	Nemaline myopathy 7, autosomal recessive, 610687
CFP	95.9	98.4	93.4	Properdin deficiency, X-linked, 312060
CFTR	124	99.1	96.3	Congenital bilateral absence of vas deferens, 277180 Cystic fibrosis, 219700 Sweat chloride elevation without CF, 0 {Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400 {Hypertrypsinemia, neonatal}, 0 {Pancreatitis, hereditary}, 167800
CHAMP1	160.6	100	100	Mental retardation, autosomal dominant 40, 616579
CHAT	130.3	89.3	86.8	Myasthenic syndrome, congenital, 6, presynaptic, 254210
CHCHD10	20	43	35.2	?Myopathy, isolated mitochondrial, autosomal dominant, 616209 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 Spinal muscular atrophy, Jokela type, 615048
CHCHD2	95.4	99.5	91.9	Parkinson disease 22, autosomal dominant, 616710
CHD1	95.4	93.7	83.5	Pilarowski-Bjornsson syndrome, 617682
CHD2	137.7	99.3	98.5	Epileptic encephalopathy, childhood-onset, 615369
CHD3	106.1	94.7	92.1	Snijders Blok-Campeau syndrome, 618205
CHD4	131.3	100	99.8	Sifrim-Hitz-Weiss syndrome, 617159
CHD7	150.7	99.9	98.9	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370

CHEK2	100.8	82.4	78.7	Li-Fraumeni syndrome, 609265 Osteosarcoma, somatic, 259500 {Breast and colorectal cancer, susceptibility to}, 0 {Breast cancer, susceptibility to}, 114480 {Prostate cancer, familial, susceptibility to}, 176807
CHKB	98.5	100	99	Muscular dystrophy, congenital, megaconial type, 602541
CHM	102.6	96.5	87.3	Choroideremia, 303100
CHMP1A	133.7	100	100	Pontocerebellar hypoplasia, type 8, 614961
CHMP2B	92	98.6	91.7	Amyotrophic lateral sclerosis 17, 614696 Dementia, familial, nonspecific, 600795
CHMP4B	139.3	99.9	99	Cataract 31, multiple types, 605387
CHN1	141.8	99.2	97.7	Duane retraction syndrome 2, 604356
CHRDL1	109.7	100	99.4	Megalocornea 1, X-linked, 309300
CHRM3	160.2	100	100	?Prune belly syndrome, 100100
CHRNA1	121.8	94.7	94.6	Multiple pterygium syndrome, lethal type, 253290 Myasthenic syndrome, congenital, 1A, slow-channel, 601462 Myasthenic syndrome, congenital, 1B, fast-channel, 608930
CHRNA2	229.3	100	100	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA4	142.1	96.7	95.8	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction, susceptibility to}, 188890
CHRNB1	131.8	98.8	96.7	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314 Myasthenic syndrome, congenital, 2A, slow-channel, 616313
CHRNB2	247.5	98.4	94.6	Epilepsy, nocturnal frontal lobe, 3, 605375
CHRND	150.5	100	99	?Myasthenic syndrome, congenital, 3A, slow-channel, 616321 ?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323 Multiple pterygium syndrome, lethal type, 253290 Myasthenic syndrome, congenital, 3B, fast-channel, 616322
CHRNE	127.7	99.3	95.8	Myasthenic syndrome, congenital, 4A, slow-channel, 605809 Myasthenic syndrome, congenital, 4B, fast-channel, 616324 Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931
CHRNG	155.2	100	100	Escobar syndrome, 265000 Multiple pterygium syndrome, lethal type, 253290
CHST11	240.7	100	100	?Osteochondrodysplasia, brachydactyly, and overlapping malformed digits, 618167

CHST14	165.6	95.7	93.3	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	91.6	100	97.5	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHST6	334	100	100	Macular corneal dystrophy, 217800
CHST8	263.6	100	100	?Peeling skin syndrome 3, 616265
CHSY1	138.4	95.9	93.9	Temtamy preaxial brachydactyly syndrome, 605282
CHUK	131.6	100	98.4	Cocoon syndrome, 613630
CIB2	229.9	99.9	99.6	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CIC	54.2	63.7	60.9	Mental retardation, autosomal dominant 45, 617600
CIDEC	96.4	99.9	96.4	?Lipodystrophy, familial partial, type 5, 615238
CIITA	125	100	100	Bare lymphocyte syndrome, type II, complementation group A, 209920 {Rheumatoid arthritis, susceptibility to}, 180300
CISD2	127.9	83.4	83.4	Wolfram syndrome 2, 604928
CIT	108.6	99.9	98.2	Microcephaly 17, primary, autosomal recessive, 617090
CITED2	111.6	99.2	99	Atrial septal defect 8, 614433 Ventricular septal defect 2, 614431
CKAP2L	161.3	98.9	96.6	Filippi syndrome, 272440
CLCF1	76.2	98.7	97.7	Cold-induced sweating syndrome 2, 610313
CLCN1	137.4	100	99.5	Myotonia congenita, dominant, 160800 Myotonia congenita, recessive, 255700 Myotonia levior, recessive, 0
CLCN2	108.6	100	99.4	Hyperaldosteronism, familial, type II, 605635 Leukoencephalopathy with ataxia, 615651 {Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628 {Epilepsy, juvenile absence, susceptibility to, 2}, 607628 {Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628
CLCN4	123	100	99.8	Raynaud-Claes syndrome, 300114
CLCN5	134.6	99.6	98	Dent disease, 300009 Hypophosphatemic rickets, 300554 Nephrolithiasis, type I, 310468 Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990
CLCN7	129.7	99.5	98.2	Osteopetrosis, autosomal dominant 2, 166600 Osteopetrosis, autosomal recessive 4, 611490

CLCNKA	108.5	99.4	94.7	Bartter syndrome, type 4b, digenic, 613090
CLCNKB	100.7	98.5	90.5	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090
CLDN1	137.6	100	100	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626
CLDN10	148.9	100	100	HELIX syndrome, 617671
CLDN14	130.6	100	99.9	Deafness, autosomal recessive 29, 614035
CLDN16	136.3	100	99.9	Hypomagnesemia 3, renal, 248250
CLDN19	123.7	98.2	93.7	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLEC7A	150.7	100	99.9	Candidiasis, familial, 4, autosomal recessive, 613108 {Aspergillosis, susceptibility to}, 614079
CLIC2	72.5	99.3	95.2	?Mental retardation, X-linked, syndromic 32, 300886
CLIC5	118.3	100	99.9	?Deafness, autosomal recessive 103, 616042
CLMP	111	100	99.9	Congenital short bowel syndrome, 615237
CLN3	114.9	92.5	90.7	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	146.1	98.2	92.2	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	131.6	98.9	95.3	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	163.9	83.5	83.5	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLP1	182.4	100	99.8	Pontocerebellar hypoplasia, type 10, 615803
CLPB	140.2	100	99.5	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CLPP	115.4	99.8	96.9	Perrault syndrome 3, 614129
CLPX	151	99.7	97	?Protoporphyria, erythropoietic, 2, 618015
CLRN1	157.2	100	99.8	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902
CLTC	171.5	99.8	99.2	Mental retardation, autosomal dominant 56, 617854
CNBP	136.7	100	100	Myotonic dystrophy 2, 602668
CNGA1	127.2	89.4	84.6	Retinitis pigmentosa 49, 613756
CNGA3	167.7	100	99.9	Achromatopsia 2, 216900
CNGB1	102.5	98.4	94.8	Retinitis pigmentosa 45, 613767
CNGB3	101.4	97.7	93	Achromatopsia 3, 262300 Macular degeneration, juvenile, 248200
CNKSR2	98.1	96.5	89.7	Mental retardation, X-linked, syndromic, Houge type, 301008

CNNM2	188.4	100	99.2	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNNM4	190.5	98.7	97.8	Jalili syndrome, 217080
CNPY3	87.1	99.8	96.7	Epileptic encephalopathy, early infantile, 60, 617929
CNTN1	151.7	99.8	98.3	?Myopathy, congenital, Compton-North, 612540
CNTN2	125.4	92.7	92.6	?Epilepsy, myoclonic, familial adult, 5, 615400
CNTNAP1	161.2	99.2	97.5	Hypomyelinating neuropathy, congenital, 3, 618186 Lethal congenital contracture syndrome 7, 616286
CNTNAP2	148	100	99.9	Cortical dysplasia-focal epilepsy syndrome, 610042 Pitt-Hopkins like syndrome 1, 610042 {Autism susceptibility 15}, 612100
COA5	59.2	85.6	84	?Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 3, 616500
COA6	78.7	98.8	91.9	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 4, 616501
COA7	146	100	100	?Mitochondrial complex IV deficiency, 220110
COASY	168.5	100	100	Neurodegeneration with brain iron accumulation 6, 615643
COCH	194.4	99.9	99.6	?Deafness, autosomal recessive 110, 618094 Deafness, autosomal dominant 9, 601369
COG1	124.2	100	99.9	Congenital disorder of glycosylation, type IIg, 611209
COG2	122.6	97.2	94.7	?Congenital disorder of glycosylation, type IIq, 617395
COG4	123.8	100	99.9	Congenital disorder of glycosylation, type IIj, 613489 Saul-Wilson syndrome, 618150
COG5	107	97.4	93.8	Congenital disorder of glycosylation, type IIi, 613612
COG6	78.4	95	85.9	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328
COG7	125.1	100	100	Congenital disorder of glycosylation, type IIe, 608779
COG8	122.4	99.9	98.4	Congenital disorder of glycosylation, type IIh, 611182
COL10A1	88.4	98.6	93.9	Metaphyseal chondrodysplasia, Schmid type, 156500
COL11A1	90.8	94.9	89.6	Fibrochondrogenesis 1, 228520 Marshall syndrome, 154780 Stickler syndrome, type II, 604841 {Lumbar disc herniation, susceptibility to}, 603932

COL11A2	92.2	99.9	98.3	Deafness, autosomal dominant 13, 601868 Deafness, autosomal recessive 53, 609706 Fibrochondrogenesis 2, 614524 Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840 Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150
COL12A1	137.5	99.5	97.5	?Ullrich congenital muscular dystrophy 2, 616470 Bethlem myopathy 2, 616471
COL13A1	85.4	99.8	97.1	Myasthenic syndrome, congenital, 19, 616720
COL17A1	107.9	99.2	96.6	Epidermolysis bullosa, junctional, localisata variant, 226650 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epithelial recurrent erosion dystrophy, 122400
COL18A1	88.7	93.9	87.7	Knobloch syndrome, type 1, 267750
COL1A1	134.9	98.1	96.3	Caffey disease, 114000 Ehlers-Danlos syndrome, arthrochalasia type, 1, 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	101.8	96.7	93.6	Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821 Ehlers-Danlos syndrome, cardiac valvular type, 225320 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type III, 259420 Osteogenesis imperfecta, type IV, 166220 {Osteoporosis, postmenopausal}, 166710
COL25A1	126.8	98.4	95.8	Fibrosis of extraocular muscles, congenital, 5, 616219
COL27A1	116.1	99.5	97.4	Steel syndrome, 615155

COL2A1	103.4	99.9	99	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 Platyspondylic skeletal dysplasia, Torrance type, 151210 SED congenita, 183900 SMED Strudwick type, 184250 Spondyloepiphyseal dysplasia, Stanescu type, 616583 Spondyloperipheral dysplasia, 271700 Stickler sydrome, type I, nonsyndromic ocular, 609508 Stickler syndrome, type I, 108300 Vitreoretinopathy with phalangeal epiphyseal dysplasia, 0
COL3A1	104.3	97.8	92.3	Ehlers-Danlos syndrome, vascular type, 130050
COL4A1	92.8	97.9	94	?Retinal arteries, tortuosity of, 180000 Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Brain small vessel disease with or without ocular anomalies, 607595 Porencephaly 1, 175780 Schizencephaly, 269160 {Hemorrhage, intracerebral, susceptibility to}, 614519
COL4A2	96.8	98.5	93.9	Porencephaly 2, 614483 {Hemorrhage, intracerebral, susceptibility to}, 614519
COL4A3	89.6	97.8	95.5	Alport syndrome, autosomal dominant, 104200 Alport syndrome, autosomal recessive, 203780 Hematuria, benign familial, 141200
COL4A3BP	121.6	98.3	92.7	Mental retardation, autosomal dominant 34, 616351
COL4A4	85	97.6	93.5	Alport syndrome, autosomal recessive, 203780 Hematuria, familial benign, 0
COL4A5	52.3	92.1	77.5	Alport syndrome, 301050
COL4A6	81.2	96	89.4	?Deafness, X-linked 6, 300914
COL5A1	114.3	97.7	95	Ehlers-Danlos syndrome, classic type, 1, 130000
COL5A2	89.1	99.5	97.4	Ehlers-Danlos syndrome, classic type, 2, 130010

COL6A1	137.2	99.5	97.8	Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090
COL6A2	165.3	99.3	98.4	?Myosclerosis, congenital, 255600 Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090
COL6A3	174.7	100	99.9	Bethlem myopathy 1, 158810 Dystonia 27, 616411 Ullrich congenital muscular dystrophy 1, 254090
COL7A1	129.5	99.6	97.5	EBD inversa, 226600 EBD, Bart type, 132000 EBD, localisata variant, 0 Epidermolysis bullosa dystrophica, AD, 131750 Epidermolysis bullosa dystrophica, AR, 226600 Epidermolysis bullosa pruriginosa, 604129 Epidermolysis bullosa, pretibial, 131850 Toenail dystrophy, isolated, 607523 Transient bullous of the newborn, 131705
COL8A2	37.5	84.6	69.4	Corneal dystrophy, Fuchs endothelial, 1, 136800 Corneal dystrophy, posterior polymorphous 2, 609140
COL9A1	121.2	99.5	96.9	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	65.1	98.3	88.8	?Stickler syndrome, type V, 614284 Epiphyseal dysplasia, multiple, 2, 600204
COL9A3	65.5	95.6	86.7	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 {Intervertebral disc disease, susceptibility to}, 603932
COLEC10	162.6	100	99.1	3MC syndrome 3, 248340
COLEC11	203	100	100	3MC syndrome 2, 265050
COLQ	113.2	99.8	98.1	Myasthenic syndrome, congenital, 5, 603034
COMP	121.4	93.6	92.4	Epiphyseal dysplasia, multiple, 1, 132400 Pseudoachondroplasia, 177170
COPB2	149.7	99.9	99.1	?Microcephaly 19, primary, autosomal recessive, 617800
COQ2	89.3	96.1	93.2	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ4	89.8	88.4	84.9	Coenzyme Q10 deficiency, primary, 7, 616276

COQ6	143.9	99.3	96	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	158.5	99.7	98.9	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ8A	134.3	100	99.1	Coenzyme Q10 deficiency, primary, 4, 612016
COQ8B	90.5	100	99.1	Nephrotic syndrome, type 9, 615573
COQ9	91.4	99.9	96.6	Coenzyme Q10 deficiency, primary, 5, 614654
CORIN	179.1	99.9	99.4	Preeclampsia/eclampsia 5, 614595
CORO1A	154.4	99.8	96.9	Immunodeficiency 8, 615401
COX10	241.9	100	99.6	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 Mitochondrial complex IV deficiency, 220110
COX14	108.1	100	99.9	?Mitochondrial complex IV deficiency, 220110
COX15	98.6	100	99.7	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000
COX20	58.1	83	65.4	Mitochondrial complex IV deficiency, 220110
COX4I2	120.1	100	100	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX6A1	180.6	100	99.4	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
COX6B1	159.6	100	100	Mitochondrial complex IV deficiency, 220110
COX7B	47.9	73.3	42	Linear skin defects with multiple congenital anomalies 2, 300887
COX8A	98.1	100	100	?Mitochondrial complex IV deficiency, 220110
CP	120	93.9	89.6	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 [Hypoceruloplasminemia, hereditary], 604290
CPA6	118.3	99.8	98.5	Epilepsy, familial temporal lobe, 5, 614417 Febrile seizures, familial, 11, 614418
CPAMD8	102	94.7	91.4	Anterior segment dysgenesis 8, 617319
CPLX1	79.8	99.9	97.9	Epileptic encephalopathy, early infantile, 63, 617976
CPN1	118.3	100	98.8	Carboxypeptidase N deficiency, 212070
CPOX	116.8	95.2	88.1	Coproporphyria, 121300 Harderoporphyria, 121300
CPS1	143.8	100	99.8	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}, 0
CPT1A	169.3	100	98.7	CPT deficiency, hepatic, type IA, 255120

CPT1C	111.8	100	99.9	?Spastic paraplegia 73, autosomal dominant, 616282
CPT2	162.8	97.2	95.4	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	99.8	Immunodeficiency, common variable, 7, 614699 {Systemic lupus erythematosus, susceptibility to, 9}, 610927
CRADD	115.2	99.9	98.5	Mental retardation, autosomal recessive 34, with variant lissencephaly, 614499
CRAT	126.6	100	99.9	?Neurodegeneration with brain iron accumulation 8, 617917
CRB1	191.8	100	100	Leber congenital amaurosis 8, 613835 Pigmented paravenous chorioretinal atrophy, 172870 Retinitis pigmentosa-12, 600105
CRB2	112.4	99.4	94.7	Focal segmental glomerulosclerosis 9, 616220 Ventriculomegaly with cystic kidney disease, 219730
CRBN	130.7	87.8	83.8	Mental retardation, autosomal recessive 2, 607417
CREB1	147.3	98.6	93.7	Histiocytoma, angiomatoid fibrous, somatic, 612160
CREB3L1	113.8	99.4	97	Osteogenesis imperfecta, type XVI, 616229
CREBBP	123.5	99.4	96.7	Rubinstein-Taybi syndrome 1, 180849
CRELD1	114.4	99.9	97.8	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217 {Atrioventricular septal defect, susceptibility to, 2}, 606217
CRIPT	34.5	96.5	74.8	Short stature with microcephaly and distinctive facies, 615789
CRLF1	105.6	90.9	89.2	Cold-induced sweating syndrome 1, 272430
CRTAP	110.4	99.8	97.3	Osteogenesis imperfecta, type VII, 610682
CRTC1	135.2	99	95.7	Mucoepidermoid salivary gland carcinoma, 0
CRX	114.2	99.9	98.7	Cone-rod retinal dystrophy-2, 120970 Leber congenital amaurosis 7, 613829
CRYAA	135.3	92.7	86.2	Cataract 9, multiple types, 604219
CRYAB	125.7	99.9	98.7	Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, 2, 608810 Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869
CRYBA1	135.7	100	99.2	Cataract 10, multiple types, 600881

CRYBA2	150.6	100	100	?Cataract 42, 115900
CRYBA4	117.5	100	100	Cataract 23, 610425
CRYBB1	129.1	100	99.4	Cataract 17, multiple types, 611544
CRYBB2	150.3	100	100	Cataract 3, multiple types, 601547
CRYBB3	144.3	100	100	Cataract 22, 609741
CRYGB	97.4	99.8	97.4	Cataract 39, multiple types, autosomal dominant, 615188
CRYGC	128.5	100	99.3	Cataract 2, multiple types, 604307
CRYGD	100.4	100	99.8	Cataract 4, multiple types, 115700
CRYGS	105.4	96.4	88	Cataract 20, multiple types, 116100
CRYM	97.4	99.9	98.3	Deafness, autosomal dominant 40, 616357
CSF1R	139.5	99.5	98.4	Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
CSF2RA	66	89.9	88.2	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF2RB	94.8	99.6	97.8	Surfactant metabolism dysfunction, pulmonary, 5, 614370
CSF3R	94.4	99.3	96.5	Neutropenia, severe congenital, 7, autosomal recessive, 617014
CSNK1D	148.4	94.8	90.4	Advanced sleep-phase syndrome, familial, 2, 615224
CSNK2A1	126.4	94.1	86.2	Okur-Chung neurodevelopmental syndrome, 617062
CSPP1	112	99.8	97.8	Joubert syndrome 21, 615636
CSRP3	103	100	99.9	?Cardiomyopathy, dilated, 1M, 607482 Cardiomyopathy, hypertrophic, 12, 612124
CST3	106.4	92	74.3	Cerebral amyloid angiopathy, 105150 {Macular degeneration, age-related, 11}, 611953
CSTA	119	99.9	99	Peeling skin syndrome 4, 607936
CSTB	82.5	97.1	82.7	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTBP1	93.4	96.1	85	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915
CTC1	119	100	99.8	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTCF	158.4	98.6	96.7	Mental retardation, autosomal dominant 21, 615502
CTDP1	105	86.6	83.6	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTH	152.6	99.9	98.1	Cystathioninuria, 219500 Homocysteine, total plasma, elevated, 0
CTHRC1	90.2	91.9	84.5	Barrett esophagus/esophageal adenocarcinoma, 614266

CTLA4	193.9	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	125.6	99.9	99	Macular dystrophy, patterned, 2, 608970 Gastric cancer
CTNNA2	124.6	99.9	99.3	Cortical dysplasia, complex, with other brain malformations 9, 618174
CTNNA3	138.3	100	99.9	Arrhythmogenic right ventricular dysplasia, familial, 13, 615616
CTNNB1	163.8	100	99.9	Colorectal cancer, somatic, 114500 Exudative vitreoretinopathy 7, 617572 Hepatocellular carcinoma, somatic, 114550 Medulloblastoma, somatic, 155255 Mental retardation, autosomal dominant 19, 615075 Ovarian cancer, somatic, 167000 Pilomatricoma, somatic, 132600
CTNND1	153.8	100	99.9	Blepharocheilodontic syndrome 2, 617681
CTNS	120.1	100	99.9	Cystinosis, atypical nephropathic, 219800 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750
CTPS1	143.1	100	99.6	Immunodeficiency 24, 615897
CTSA	134.1	100	99.4	Galactosialidosis, 256540
CTSC	127.5	100	100	Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000 Periodontitis 1, juvenile, 170650
CTSD	163.7	98	95.3	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	112.8	84.2	80.2	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362
CTSK	105	100	99.9	Pycnodysostosis, 265800
CTU2	106.7	98.6	91.7	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142
CUBN	127.8	99.8	98.4	Megaloblastic anemia-1, Finnish type, 261100
CUL3	110.3	98.6	94.7	Pseudohypoaldosteronism, type IIE, 614496
CUL4B	72.8	98	88.5	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354

CUL7	149.6	99.8	97.9	3-M syndrome 1, 273750
CUX2	85.6	99.8	97.9	Epileptic encephalopathy, early infantile, 67, 618141
CWC27	74.7	97.2	89.7	Retinitis pigmentosa with or without skeletal anomalies, 250410
CWF19L1	119.1	99.5	96.7	Spinocerebellar ataxia, autosomal recessive 17, 616127
CXCR4	202.7	100	99.9	Myelokathexis, isolated, 0 WHIM syndrome, 193670
CXorf56	91.8	99.7	95.5	?Mental retardation, X-linked 107, 301013
CYB561	131	92.8	92.6	Orthostatic hypotension 2, 618182
CYB5A	133.5	100	100	Methemoglobinemia and ambiguous genitalia, 250790
CYB5R3	147.3	98	98	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYBA	97.3	77.9	71	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690
CYBB	110.8	99.9	99.2	Chronic granulomatous disease, X-linked, 306400 Immunodeficiency 34, mycobacteriosis, X-linked, 300645
CYC1	184.5	88.1	86.8	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYCS	72.1	99.6	95	Thrombocytopenia 4, 612004
CYFIP2	139.9	100	99.6	Epileptic encephalopathy, early infantile, 65, 618008
CYLD	119.9	98.1	93	Brooke-Spiegler syndrome, 605041 Cylindromatosis, familial, 132700 Trichoepithelioma, multiple familial, 1, 601606
CYP11A1	123.9	99.6	97.7	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	175.9	100	100	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 Aldosteronism, glucocorticoid-remediable, 103900
CYP11B2	173.4	100	100	Aldosterone to renin ratio raised, 0 Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 {Low renin hypertension, susceptibility to}, 0
CYP17A1	135	100	99.7	17,20-lyase deficiency, isolated, 202110 17-alpha-hydroxylase/17,20-lyase deficiency, 202110
CYP19A1	160.6	99.1	97.3	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300

CYP1B1	134.8	100	100	Anterior segment dysgenesis 6, multiple subtypes, 617315 Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300
CYP21A2	93.8	95.8	86.6	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, 1, 143880
CYP26B1	178.1	100	99.9	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
CYP26C1	87.2	99.5	95.6	Focal facial dermal dysplasia 4, 614974
CYP27A1	175.1	98.3	96.1	Cerebrotendinous xanthomatosis, 213700
CYP27B1	137.1	100	99.1	Vitamin D-dependent rickets, type I, 264700
CYP2A6	167.9	100	99.8	Coumarin resistance, 122700 {Lung cancer, resistance to}, 211980 {Nicotine addiction, protection from}, 188890
CYP2B6	120.3	100	98.9	Efavirenz, poor metabolism of, 614546 {Efavirenz central nervous system toxicity, susceptibility to}, 614546
CYP2C19	186.3	99.5	96.3	Clopidogrel, impaired responsiveness to, 609535 Mephenytoin poor metabolizer, 609535 Omeprazole poor metabolizer, 609535 Proguanil poor metabolizer, 609535
CYP2C9	186.9	99.3	94.4	Tolbutamide poor metabolizer, 0 Warfarin sensitivity, 122700
CYP2R1	138.5	96.4	86.6	Rickets due to defect in vitamin D 25-hydroxylation, 600081
CYP2U1	119.2	93.7	91.2	Spastic paraplegia 56, autosomal recessive, 615030
CYP4F22	127.7	100	99.7	Ichthyosis, congenital, autosomal recessive 5, 604777
CYP4V2	147.5	99.8	98.5	Bietti crystalline corneoretinal dystrophy, 210370
CYP7B1	93.2	94.7	87.7	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, autosomal recessive, 270800
D2HGDH	134.5	97.5	95.2	D-2-hydroxyglutaric aciduria, 600721
DAB1	123.9	100	100	Spinocerebellar ataxia 37, 615945
DACT1	127.1	92.5	89.4	?Townes-Brocks syndrome 2, 617466
DAG1	220.8	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	98.6	98.7	93.8	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2	122.3	100	99.6	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBH	140.7	100	99.8	Orthostatic hypotension 1, due to DBH deficiency, 223360
DBT	102.1	97.3	93.8	Maple syrup urine disease, type II, 248600
DCAF17	91.9	95.6	89.3	Woodhouse-Sakati syndrome, 241080
DCAF8	124.3	100	99.9	?Giant axonal neuropathy 2, autosomal dominant, 610100
DCC	138.5	100	99.9	Colorectal cancer, somatic, 114500 Esophageal carcinoma, somatic, 133239 Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 Mirror movements 1 and/or agenesis of the corpus callosum, 157600
DCDC2	150.5	99.9	99.6	?Deafness, autosomal recessive 66, 610212 Nephronophthisis 19, 616217 Sclerosing cholangitis, neonatal, 617394
DCHS1	160.1	99.8	99.2	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390
DCLRE1C	128.8	98.2	94.5	Omenn syndrome, 603554 Severe combined immunodeficiency, Athabascan type, 602450
DCN	140	95.7	94.9	Corneal dystrophy, congenital stromal, 610048
DCPS	143.8	100	99.8	Al-Raqad syndrome, 616459
DCTN1	131.6	99.7	98.3	Neuropathy, distal hereditary motor, type VIIB, 607641 Perry syndrome, 168605 {Amyotrophic lateral sclerosis, susceptibility to}, 105400
DCX	113.2	100	99.7	Lissencephaly, X-linked, 300067 Subcortical laminal heterotopia, X-linked, 300067
DDB2	162.4	100	99.7	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DDC	101	99.1	95	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	141.8	97.1	94.8	Spastic paraplegia 28, autosomal recessive, 609340
DDHD2	149.7	99.9	98	Spastic paraplegia 54, autosomal recessive, 615033
DDOST	120.5	99.9	99.1	?Congenital disorder of glycosylation, type Ir, 614507
DDR2	155	100	99.9	Spondylometaepiphyseal dysplasia, short limb-hand type, 271665
DDRGK1	87.2	100	99.9	Spondyloepimetaphyseal dysplasia, Shohat type, 602557

DDX11	113.9	86	81	Warsaw breakage syndrome, 613398
DDX3X	80.5	85.9	82.1	Mental retardation, X-linked 102, 300958
DDX58	123.3	98.6	95	Singleton-Merten syndrome 2, 616298
DDX59	151.7	99.7	97.6	Orofaciodigital syndrome V, 174300
DEAF1	125.9	88.3	83.7	?Dyskinesia, seizures, and intellectual developmental disorder, 617171 Mental retardation, autosomal dominant 24, 615828
DENND5A	123	99.8	97.9	Epileptic encephalopathy, early infantile, 49, 617281
DEPDC5	148.3	99.8	99.3	Epilepsy, familial focal, with variable foci 1, 604364
DES	120.8	99.9	98.1	Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400
DFNA5	112.9	99.9	99.4	Deafness, autosomal dominant 5, 600994
DFNB59	123.6	100	99.2	Deafness, autosomal recessive 59, 610220
DGAT1	156.2	88.5	86.3	?Diarrhea 7, protein-losing enteropathy type, 615863
DGKE	142.3	99.5	95.2	Nephrotic syndrome, type 7, 615008 {Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008
DGUOK	119.2	100	100	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880 Portal hypertension, noncirrhotic, 617068 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070
DHCR24	183	100	100	Desmosterolosis, 602398
DHCR7	158.3	100	100	Smith-Lemli-Opitz syndrome, 270400
DHDDS	93.5	97.8	94.8	?Congenital disorder of glycosylation, type 1bb, 613861 Developmental delay and seizures with or without movement abnormalities, 617836 Retinitis pigmentosa 59, 613861
DHFR	48.4	91.1	72	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	92.2	100	99.9	Miller syndrome, 263750
DHTKD1	141	99.6	98.2	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DHX30	160.6	99.9	99.2	Neurodevelopmental disorder with severe motor impairment and absent language, 617804

DHX38	130.3	99.9	99	Retinitis pigmentosa 84, 618220
DIABLO	206	100	99.5	Deafness, autosomal dominant 64, 614152
DIAPH1	120.7	99.3	97.8	Deafness, autosomal dominant 1, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632
DIAPH2	61.7	93.1	79.7	?Premature ovarian failure 2A, 300511
DIAPH3	73.9	97.9	90.1	Auditory neuropathy, autosomal dominant, 1, 609129
DICER1	145.4	99.5	98.1	Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800 Pleuropulmonary blastoma, 601200 Rhabdomyosarcoma, embryonal, 2, 180295
DIP2B	160.3	99.2	98.1	Mental retardation, FRA12A type, 136630
DIS3L2	158.5	99.8	99	Perlman syndrome, 267000
DKC1	111.9	99.6	98.1	Dyskeratosis congenita, X-linked, 305000
DLAT	91.6	99.1	96	Pyruvate dehydrogenase E2 deficiency, 245348
DLC1	179.2	99.9	99.4	Colorectal cancer, somatic, 114500
DLD	123.5	99.9	98.6	Dihydrolipoamide dehydrogenase deficiency, 246900
DLG3	90	99.3	94.3	Mental retardation, X-linked 90, 300850
DLL3	64.1	88.8	79.9	Spondylocostal dysostosis 1, autosomal recessive, 277300
DLL4	181.7	99.8	98.8	Adams-Oliver syndrome 6, 616589
DLX3	109.8	100	99.1	Amelogenesis imperfecta, type IV, 104510 Trichodontoosseous syndrome, 190320
DLX4	162.4	100	100	?Orofacial cleft 15, 616788
DLX5	123.6	99.9	97	?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600
DMD	112.4	99.4	97.4	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200
DMGDH	157	98.8	97.2	Dimethylglycine dehydrogenase deficiency, 605850
DMP1	159.5	99.9	99.1	Hypophosphatemic rickets, AR, 241520
DMPK	117.7	99.9	97.9	Myotonic dystrophy 1, 160900
DMXL2	181.4	98.6	96	?Deafness, autosomal dominant 71, 617605 ?Polyendocrine-polyneuropathy syndrome, 616113
DNA2	123.6	99.8	96.9	?Seckel syndrome 8, 615807 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156

DNAAF1	115.8	100	99.7	Ciliary dyskinesia, primary, 13, 613193
DNAAF2	105.1	99.7	96.9	Ciliary dyskinesia, primary, 10, 612518
DNAAF3	91.8	97.7	90.6	Ciliary dyskinesia, primary, 2, 606763
DNAAF4	79.6	96.3	84.1	Ciliary dyskinesia, primary, 25, 615482 {Dyslexia, susceptibility to, 1}, 127700
DNAAF5	107.9	84.5	78.2	Ciliary dyskinesia, primary, 18, 614874
DNAH1	183.3	100	99.7	?Ciliary dyskinesia, primary, 37, 617577 Spermatogenic failure 18, 617576
DNAH11	134	99.8	98.4	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH5	123.8	99.7	98.5	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAI1	135.3	100	100	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	156.6	98.4	95.5	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJB11	111.3	100	99.6	Polycystic kidney disease 6 with or without polycystic liver disease, 618061
DNAJB13	138.3	100	97.3	Ciliary dyskinesia, primary, 34, 617091
DNAJB2	102	100	100	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881
DNAJB6	59.8	91.7	79.3	Muscular dystrophy, limb-girdle, autosomal dominant 1, 603511
DNAJC12	129.3	87.4	87.3	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC19	97.9	98.5	90	3-methylglutaconic aciduria, type V, 610198
DNAJC21	125.7	99.8	98.5	Bone marrow failure syndrome 3, 617052
DNAJC3	116.3	99.9	98.1	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192
DNAJC5	200.2	100	99.9	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350
DNAJC6	160.8	99.9	98.9	Parkinson disease 19a, juvenile-onset, 615528 Parkinson disease 19b, early-onset, 615528
DNAL1	99	95.7	84.5	Ciliary dyskinesia, primary, 16, 614017
DNAL4	72.6	99.3	93.5	?Mirror movements 3, 616059
DNASE1L3	141.3	100	100	Systemic lupus erythematosus 16, 614420
DNM1	156.7	89.3	87.5	Epileptic encephalopathy, early infantile, 31, 616346
DNM1L	123.5	99.7	96.6	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 Optic atrophy 5, 610708

DNM2	127.4	97.5	94.4	Centronuclear myopathy 1, 160150 Charcot-Marie-Tooth disease, axonal type 2M, 606482 Charcot-Marie-Tooth disease, dominant intermediate B, 606482 Lethal congenital contracture syndrome 5, 615368
DNMT1	113.4	99.2	98.3	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 Neuropathy, hereditary sensory, type IE, 614116
DNMT3A	115.5	98.8	95.8	Acute myeloid leukemia, somatic, 601626 Tatton-Brown-Rahman syndrome, 615879
DNMT3B	124.8	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK2	143.8	100	99.9	Immunodeficiency 40, 616433
DOCK6	119.9	98.9	96.5	Adams-Oliver syndrome 2, 614219
DOCK7	114.4	97.9	95.6	Epileptic encephalopathy, early infantile, 23, 615859
DOCK8	129.1	100	99.8	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
DOK7	105.7	93.3	92.5	?Fetal akinesia deformation sequence, 208150 Myasthenic syndrome, congenital, 10, 254300
DOLK	202.9	100	99.9	Congenital disorder of glycosylation, type Im, 610768
DONSON	104.9	83.9	78.3	Microcephaly, short stature, and limb abnormalities, 617604 Microcephaly-micromelia syndrome, 251230
DPAGT1	110.7	100	100	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPF2	102.4	100	97.9	Coffin-Siris syndrome 7, 618027
DPH1	157.1	100	99.7	Developmental delay with short stature, dysmorphic features, and sparse hair, 616901
DPM1	131.1	91.7	86.7	Congenital disorder of glycosylation, type Ie, 608799
DPM2	102.1	100	99.4	Congenital disorder of glycosylation, type Iu, 615042
DPM3	183.9	100	100	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937
DPP6	145.5	96.5	94.5	Mental retardation, autosomal dominant 33, 616311 {Ventricular fibrillation, paroxysmal familial, 2}, 612956
DPY19L2	94.8	72.9	66.6	Spermatogenic failure 9, 613958
DPYD	158.3	95.6	93.7	5-fluorouracil toxicity, 274270 Dihydropyrimidine dehydrogenase deficiency, 274270

DPYS	133.5	100	99.5	Dihydropyrimidinuria, 222748
DRAM2	131.8	100	99.7	Cone-rod dystrophy 21, 616502
DRC1	97	99.9	98.6	Ciliary dyskinesia, primary, 21, 615294
DRD4	79	80.9	70.7	Autonomic nervous system dysfunction, 0 [Novelty seeking personality], 601696 {Attention deficit-hyperactivity disorder}, 143465
DSC2	128.5	99.4	96.2	Arrhythmogenic right ventricular dysplasia 11, 610476 Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476
DSC3	87.1	97.2	89.6	?Hypotrichosis and recurrent skin vesicles, 613102
DSE	124.3	99.8	98.5	Ehlers-Danlos syndrome, musculocontractural type 2, 615539
DSG1	175	98.4	96.1	Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508 Keratosis palmoplantaris striata I, AD, 148700
DSG2	140.6	99.9	98.7	Arrhythmogenic right ventricular dysplasia 10, 610193 Cardiomyopathy, dilated, 1BB, 612877
DSG4	198.4	98.5	95.7	Hypotrichosis 6, 607903
DSP	154	100	99.8	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	155.7	99.9	99.3	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	154.1	99.7	98.2	?Neuropathy, hereditary sensory and autonomic, type VI, 614653 Epidermolysis bullosa simplex, autosomal recessive 2, 615425
DSTYK	144.7	100	99.4	Congenital anomalies of kidney and urinary tract 1, 610805 Spastic paraplegia 23, 270750
DTNA	156.5	100	100	Left ventricular noncompaction 1, with or without congenital heart defects, 604169
DTNBP1	115.2	99.3	95.1	Hermansky-Pudlak syndrome 7, 614076

DUOX2	136.4	96.7	94.8	Thyroid dyshormonogenesis 6, 607200
DUOXA2	122.2	100	99.9	Thyroid dyshormonogenesis 5, 274900
DUSP6	175.9	100	99.9	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
DVL1	113.9	97.8	94.1	Robinow syndrome, autosomal dominant 2, 616331
DVL3	149.4	100	100	Robinow syndrome, autosomal dominant 3, 616894
DYM	101.3	97.2	94.8	Dyggve-Melchior-Clausen disease, 223800 Smith-McCort dysplasia, 607326
DYNC1H1	179.8	100	99.6	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600
DYNC2H1	90.5	96.6	87	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYNC2LI1	95.1	99.3	96	Short-rib thoracic dysplasia 15 with polydactyly, 617088
DYRK1A	159.6	100	100	Mental retardation, autosomal dominant 7, 614104
DYRK1B	105	93	85	Abdominal obesity-metabolic syndrome 3, 615812
DYSF	133.1	100	99.8	Miyoshi muscular dystrophy 1, 254130 Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601 Myopathy, distal, with anterior tibial onset, 606768
DZIP1L	98.3	99.4	95.5	Polycystic kidney disease 5, 617610
EARS2	103.4	99.7	98.3	Combined oxidative phosphorylation deficiency 12, 614924
EBF3	130.4	100	99.1	Hypotonia, ataxia, and delayed development syndrome, 617330
EBP	83.3	100	98	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
ECE1	144.6	97.9	97.8	?Hirschsprung disease, cardiac defects, and autonomic dysfunction, 613870 {Hypertension, essential, susceptibility to}, 145500
ECEL1	100.7	88.8	83.1	Arthrogryposis, distal, type 5D, 615065
ECHS1	112.8	99.8	97.8	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ECM1	170.8	100	99.7	Urbach-Wiethe disease, 247100
EDA	88.5	85.7	77.3	Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100 Tooth agenesis, selective, X-linked 1, 313500

EDAR	138.6	100	99.6	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	99.1	99.3	93.3	Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940 Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941
EDC3	144.7	100	99.6	?Mental retardation, autosomal recessive 50, 616460
EDN1	145.5	100	100	Auriculocondylar syndrome 3, 615706 Question mark ears, isolated, 612798 {High density lipoprotein cholesterol level QTL 7}, 0
EDN3	134.4	100	99.5	Central hypoventilation syndrome, congenital, 209880 Waardenburg syndrome, type 4B, 613265 {Hirschsprung disease, susceptibility to, 4}, 613712
EDNRA	218.9	100	99.7	Mandibulofacial dysostosis with alopecia, 616367 {Migraine, resistance to}, 157300
EDNRB	131	95.6	90.9	ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580 {Hirschsprung disease, susceptibility to, 2}, 600155
EED	94.2	98.4	91.6	Cohen-Gibson syndrome, 617561
EEF1A2	177.7	98.8	93.8	Epileptic encephalopathy, early infantile, 33, 616409 Mental retardation, autosomal dominant 38, 616393
EEF2	167.6	100	99.9	?Spinocerebellar ataxia 26, 609306
EFEMP1	167.9	100	99.6	Doyne honeycomb degeneration of retina, 126600
EFEMP2	120.9	100	99.9	Cutis laxa, autosomal recessive, type IB, 614437
EFL1	174.8	99.4	97.7	Shwachman-Diamond syndrome 2, 617941
EFNB1	118.5	100	99.9	Craniofrontonasal dysplasia, 304110
EFTUD2	124.2	100	99.4	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EGF	135.2	100	99.8	Hypomagnesemia 4, renal, 611718
EGFR	160.8	100	99.1	?Inflammatory skin and bowel disease, neonatal, 2, 616069 Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980 Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980 {Nonsmall cell lung cancer, susceptibility to}, 211980
EGLN1	39	79	66.3	Erythrocytosis, familial, 3, 609820 [Hemoglobin, high altitude adaptation], 609070

EGR2	124.4	100	100	Charcot-Marie-Tooth disease, type 1D, 607678 Dejerine-Sottas disease, 145900 Hypomyelinating neuropathy, congenital, 1, 605253
EHHADH	163.2	100	99.7	?Fanconi renotubular syndrome 3, 615605
EHMT1	141.4	94	92.9	Kleefstra syndrome 1, 610253
EIF2AK3	147.1	95.1	91.3	Wolcott-Rallison syndrome, 226980
EIF2AK4	146.3	99.8	98	Pulmonary venoocclusive disease 2, 234810
EIF2B1	149.9	100	100	Leukoencephalopathy with vanishing white matter, 603896
EIF2B2	131.9	100	99.5	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B3	163.8	100	100	Leukoencephalopathy with vanishing white matter, 603896
EIF2B4	146	100	99.5	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B5	128	99.6	97.9	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2S3	84.9	97	89.8	MEHMO syndrome, 300148
EIF4A3	106.8	100	99.9	Robin sequence with cleft mandible and limb anomalies, 268305
ELAC2	123.8	100	99.3	Combined oxidative phosphorylation deficiency 17, 615440 {Prostate cancer, hereditary, 2, susceptibility to}, 614731
ELANE	80.9	99.7	95.9	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
ELMO2	134.7	100	99.1	Vascular malformation, primary intraosseous, 606893
ELMOD3	156.2	100	99.7	?Deafness, autosomal recessive 88, 615429
ELN	91.1	99.4	97.4	Cutis laxa, autosomal dominant, 123700 Supravalvar aortic stenosis, 185500
ELOVL4	91.9	99.9	98	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110
ELOVL5	120.9	100	99.8	Spinocerebellar ataxia 38, 615957
ELP1	142.6	99.7	98.1	Dysautonomia, familial, 223900
ELP2	125.5	99.2	96.9	Mental retardation, autosomal recessive 58, 617270
ELP4	54.9	72.2	66.7	?Aniridia 2, 617141

EMC1	124.3	100	99.8	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EMD	100.3	99.8	97.2	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
EMG1	137.7	100	100	Bowen-Conradi syndrome, 211180
EML1	163.6	99	97.1	Band heterotopia, 600348
EMP2	101.1	99.7	96.9	Nephrotic syndrome, type 10, 615861
EMX2	118	100	100	Schizencephaly, 269160
ENAM	148.9	100	99.9	Amelogenesis imperfecta, type IB, 104500 Amelogenesis imperfecta, type IC, 204650
ENG	128.8	97.4	93.6	Telangiectasia, hereditary hemorrhagic, type 1, 187300
ENO3	179.3	100	100	?Glycogen storage disease XIII, 612932
ENPP1	134.8	92.4	83.2	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	165.1	100	99.4	Spastic paraplegia 64, autosomal recessive, 615683
EOGT	119.9	79.5	78.4	Adams-Oliver syndrome 4, 615297
EP300	199.7	99.6	97.9	Colorectal cancer, somatic, 114500 Rubinstein-Taybi syndrome 2, 613684
EPAS1	136.7	99.4	97.1	Erythrocytosis, familial, 4, 611783
EPB41	135.5	98.7	95.6	Elliptocytosis-1, 611804
EPB41L1	128.7	99.9	97.7	?Mental retardation, autosomal dominant 11, 614257
EPB42	160.7	100	99.7	Spherocytosis, type 5, 612690
EPCAM	64.5	93.3	79.8	Colorectal cancer, hereditary nonpolyposis, type 8, 613244 Diarrhea 5, with tufting enteropathy, congenital, 613217
EPG5	126	99.3	97.7	Vici syndrome, 242840
EPHA2	175	99.2	97.8	Cataract 6, multiple types, 116600
EPHB4	140.6	99.8	98.6	Capillary malformation-arteriovenous malformation 2, 618196 Lymphatic malformation 7, 617300
EPHX1	122.6	98.8	96.1	?Hypercholanemia, familial, 607748
EPM2A	110.1	86.2	84	Epilepsy, progressive myoclonic 2A (Lafora), 254780

EPO	90.2	98.1	96.4	?Diamond-Blackfan anemia-like, 617911 Erythrocytosis, familial, 5, 617907 {Microvascular complications of diabetes 2}, 612623
EPRS	126.3	100	99.2	Leukodystrophy, hypomyelinating, 15, 617951
EPS8	122.8	96.9	91.4	?Deafness, autosomal recessive 102, 615974
EPS8L2	115.1	94.4	90.9	Deafness autosomal recessive 106, 617637
ERAL1	181.5	100	100	Perrault syndrome 6, 617565
ERBB2	135	98.3	97.3	Adenocarcinoma of lung, somatic, 211980 Gastric cancer, somatic, 613659 Glioblastoma, somatic, 137800 Ovarian cancer, somatic, 0
ERBB3	139.2	100	99.9	?Lethal congenital contractural syndrome 2, 607598 {?Erythroleukemia, familial, susceptibility to}, 133180
ERBB4	144.5	99.9	99.1	Amyotrophic lateral sclerosis 19, 615515
ERCC1	76.6	100	97.3	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	123.7	100	99.7	?Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy 1, photosensitive, 601675 Xeroderma pigmentosum, group D, 278730
ERCC3	113.2	99.9	98.9	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651
ERCC4	139.2	100	99.5	?XFE progeroid syndrome, 610965 Fanconi anemia, complementation group Q, 615272 Xeroderma pigmentosum, group F, 278760 Xeroderma pigmentosum, type F/Cockayne syndrome, 278760
ERCC5	139.8	100	99.4	Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	191.3	100	99.9	Cerebrooculofacioskeletal syndrome 1, 214150 Cockayne syndrome, type B, 133540 De Sanctis-Cacchione syndrome, 278800 Premature ovarian failure 11, 616946 UV-sensitive syndrome 1, 600630 {Lung cancer, susceptibility to}, 211980 {Macular degeneration, age-related, susceptibility to, 5}, 613761
ERCC6L2	107.6	99.7	97.7	Bone marrow failure syndrome 2, 615715

ERCC8	89.5	92.9	78.4	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621
ERF	107.9	99.9	97.7	Chitayat syndrome, 617180 Craniosynostosis 4, 600775
ERGIC1	195.2	95.3	94.7	?Arthrogryposis multiplex congenita, neurogenic type, 208100
ERLIN1	146.9	100	100	Spastic paraplegia 62, 615681
ERLIN2	156	100	99.3	Spastic paraplegia 18, autosomal recessive, 611225
ERMARD	131	99.8	98.1	?Periventricular nodular heterotopia 6, 615544
ESCO2	105.2	97.3	90.4	Roberts syndrome, 268300 SC phocomelia syndrome, 269000
ESPN	28.6	44.2	35.3	Deafness, autosomal recessive 36, 609006 Deafness, neurosensory, without vestibular involvement, autosomal dominant, 0
ESR1	127.5	99.9	99.4	Estrogen resistance, 615363 {Atherosclerosis, susceptibility to}, 0 {Breast cancer}, 114480 {HDL response to hormone replacement, augmented}, 0 {Migraine, susceptibility to}, 157300 {Myocardial infarction, susceptibility to}, 608446
ESR2	133.7	100	99.5	?Ovarian dysgenesis 8, 618187 Medullary thyroid carcinoma (Smith (2016) Hum Mol Genet 25,1836) ?Primary amenorrhea (Asadi (2013) Clin Genet 83,497) ?Breast cancer, increased risk (Pylkas (2012) PLoS Genet 8,e1002734
ESRP1	106.5	99.9	98.5	?Deafness, autosomal recessive 109, 618013
ESRRB	119.7	100	99.2	Deafness, autosomal recessive 35, 608565
ETFA	143.3	100	99.4	Glutaric acidemia IIA, 231680
ETFB	126.6	100	100	Glutaric acidemia IIB, 231680
ETFDH	105.4	100	99.3	Glutaric acidemia IIC, 231680
ETHE1	85.5	99.3	95.8	Ethylmalonic encephalopathy, 602473
ETV6	140.1	100	99.9	Leukemia, acute myeloid, somatic, 601626 Thrombocytopenia 5, 616216
EVC	110.4	93.2	89.8	?Weyers acrofacial dysostosis, 193530 Ellis-van Creveld syndrome, 225500
EVC2	119.3	96.4	94.3	Ellis-van Creveld syndrome, 225500

				Weyers acrofacial dysostosis, 193530
EWSR1	76.6	91.1	80.9	Ewing sarcoma, 612219 Neuroepithelioma, 612219
EXOSC2	142	100	100	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EXOSC3	88.5	97.3	89.4	Pontocerebellar hypoplasia, type 1B, 614678
EXOSC8	80.1	91.8	76.9	Pontocerebellar hypoplasia, type 1C, 616081
EXOSC9	129.8	97.5	87.4	Pontocerebellar hypoplasia, type 1D, 618065
EXPH5	183.3	100	99.9	Epidermolysis bullosa, nonspecific, autosomal recessive, 615028
EXT1	105.4	99.9	98.5	Chondrosarcoma, 215300 Exostoses, multiple, type 1, 133700
EXT2	163.5	99.9	99.1	?Seizures, scoliosis, and macrocephaly syndrome, 616682 Exostoses, multiple, type 2, 133701
EXTL3	206.4	100	100	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
EYA1	144.2	100	99.7	?Otofaciocervical syndrome, 166780 Anterior segment anomalies with or without cataract, 602588 Branchiootic syndrome 1, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650
EYA4	160.6	100	99.5	?Cardiomyopathy, dilated, 1J, 605362 Deafness, autosomal dominant 10, 601316
EYS	135.8	98.9	94.9	Retinitis pigmentosa 25, 602772
EZH2	139.5	99.8	97.6	Weaver syndrome, 277590
F10	185.1	99	98.3	Factor X deficiency, 227600
F11	155	100	100	Factor XI deficiency, autosomal dominant, 612416 Factor XI deficiency, autosomal recessive, 612416
F12	111.4	100	99.5	Angioedema, hereditary, type III, 610618 Factor XII deficiency, 234000
F13A1	147.3	100	99.4	Factor XIIIA deficiency, 613225 {Myocardial infarction, protection against}, 608446 {Venous thrombosis, protection against}, 188050
F13B	113.5	96.6	87.6	Factor XIIIB deficiency, 613235

F2	124.2	99.8	98.1	Dysprothrombinemia, 613679 Hypoprothrombinemia, 613679 Thrombophilia due to thrombin defect, 188050 {Pregnancy loss, recurrent, susceptibility to, 2}, 614390 {Stroke, ischemic, susceptibility to}, 601367
F5	173.5	99	97.3	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	166.6	100	98.5	Factor VII deficiency, 227500 {Myocardial infarction, decreased susceptibility to}, 608446
F8	118	99.5	98.1	Hemophilia A, 306700
F9	144.5	99.2	95.9	Hemophilia B, 306900 Thrombophilia, X-linked, due to factor IX defect, 300807 {Deep venous thrombosis, protection against}, 300807 {Warfarin sensitivity}, 122700
FA2H	94.1	87.9	79.9	Spastic paraplegia 35, autosomal recessive, 612319
FADD	142.8	100	99.6	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations, 613759
FAH	151.3	100	99.9	Tyrosinemia, type I, 276700
FAM111A	292.2	100	99.9	Gracile bone dysplasia, 602361 Kenny-Caffey syndrome, type 2, 127000
FAM111B	152.9	100	99.8	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704
FAM126A	125.2	97.3	95.2	Leukodystrophy, hypomyelinating, 5, 610532
FAM161A	115.2	98.5	95	Retinitis pigmentosa 28, 606068
FAM20A	105.4	98.4	92.1	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM20C	101.3	100	98.9	Raine syndrome, 259775
FAM46A	146.1	99.9	99.7	Osteogenesis imperfecta, type XVIII, 617952
FAM58A	73.2	82.8	78.8	STAR syndrome, 300707
FAM83H	76.7	94.9	87.7	Amelogenesis imperfecta, type IIIA, 130900
FAM92A	90	84.9	70	?Polydactyly, postaxial, type A9, 618219

FAN1	143.9	100	99.9	Interstitial nephritis, karyomegalic, 614817 Colorectal cancer
FANCA	123.3	99.8	98.5	Fanconi anemia, complementation group A, 227650
FANCB	68.4	96.7	87.9	Fanconi anemia, complementation group B, 300514
FANCC	121.6	99.4	97.1	Fanconi anemia, complementation group C, 227645
FANCD2	127.6	98.7	95.5	Fanconi anemia, complementation group D2, 227646
FANCE	108	85.9	84.6	Fanconi anemia, complementation group E, 600901
FANCF	166.8	100	100	Fanconi anemia, complementation group F, 603467
FANCG	147.7	100	100	Fanconi anemia, complementation group G, 614082
FANCI	152.1	99.5	97.5	Fanconi anemia, complementation group I, 609053
FANCL	87.8	99.4	94.7	Fanconi anemia, complementation group L, 614083
FANCM	96.8	99.2	94.3	?Premature ovarian failure 15, 618096 Spermatogenic failure 28, 618086
FAR1	80.4	96.3	92.4	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154
FARS2	207.7	100	100	Combined oxidative phosphorylation deficiency 14, 614946 Spastic paraplegia 77, autosomal recessive, 617046
FARSB	76.6	96	93.4	?Neurodevelopmental disorder with brain, liver, and lung abnormalities, 618007
FAS	272.1	100	99.3	Autoimmune lymphoproliferative syndrome, type IA, 601859 Squamous cell carcinoma, burn scar-related, somatic, 0 {Autoimmune lymphoproliferative syndrome}, 601859
FASLG	86.2	100	98.5	Autoimmune lymphoproliferative syndrome, type IB, 601859 {Lung cancer, susceptibility to}, 211980
FASTKD2	118.9	99.5	96.8	?Mitochondrial complex IV deficiency, 220110
FAT2	166.3	100	99.9	Spinocerebellar ataxia 45, 617769
FAT4	224.5	100	99.9	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 Van Maldergem syndrome 2, 615546
FBLN5	119.6	91.8	91.1	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	99.9	99.5	Acromicric dysplasia, 102370 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	161.7	100	99.5	Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118
FBP1	127	100	98.8	Fructose-1,6-bisphosphatase deficiency, 229700
FBXL4	189.8	100	100	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FBXO11	75.9	93.7	84.9	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities, 618089
FBXO31	108.8	93.5	89	?Mental retardation, autosomal recessive 45, 615979
FBXO38	179.7	99.7	98.6	Neuronopathy, distal hereditary motor, type IID, 615575
FBXO7	189.4	98.5	93.3	Parkinson disease 15, autosomal recessive, 260300
FCGR3A	225	99.6	97.7	Immunodeficiency 20, 615707
FCGR3B	176.6	99.1	98.2	Neutropenia, alloimmune neonatal, 0
FCN3	127.8	100	99.4	Immunodeficiency due to ficolin 3 deficiency, 613860
FDFT1	153.6	98.1	95.7	Squalene synthase deficiency, 618156
FDPS	72.8	99.7	95	Porokeratosis 9, multiple types, 616631
FDXR	93.4	100	99.1	Auditory neuropathy and optic atrophy, 617717
FECH	121.9	99.9	99.4	Protoporphyria, erythropoietic, 1, 177000
FERMT1	104.9	98.9	96.3	Kindler syndrome, 173650
FERMT3	122.4	100	98.9	Leukocyte adhesion deficiency, type III, 612840
FEZF1	158.8	99.9	99.3	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FGA	157	99	96.6	Afibrinogenemia, congenital, 202400 Amyloidosis, familial visceral, 105200 Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, congenital, 616004
FGB	190.8	99.7	97.9	Afibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004

				Hypofibrinogenemia, congenital, 202400
FGD1	85.7	92.7	86.5	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400
FGD4	111.9	99.3	97.3	Charcot-Marie-Tooth disease, type 4H, 609311
FGF10	142.2	100	100	Aplasia of lacrimal and salivary glands, 180920 LADD syndrome, 149730
FGF12	95.5	99.6	96.3	Epileptic encephalopathy, early infantile, 47, 617166
FGF14	190.1	100	99.7	Spinocerebellar ataxia 27, 609307
FGF16	125.5	99.9	96.7	Metacarpal 4-5 fusion, 309630
FGF17	138.6	100	100	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
FGF20	99.2	96.9	84.4	?Renal hypodysplasia/aplasia 2, 615721
FGF23	106	99.9	97.8	Hypophosphatemic rickets, autosomal dominant, 193100 Osteomalacia, tumor-induced, 0 Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993
FGF3	73.9	92	75.7	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGF5	109.2	99.7	97.8	Trichomegaly, 190330
FGF8	111.4	90.2	79.7	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGF9	165	100	100	Multiple synostoses syndrome 3, 612961
FGFR1	148	99.7	98.3	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	140.1	97.4	96.4	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, 0 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, 0 Scaphocephaly, maxillary retrusion, and mental retardation, 609579
FGFR3	110.2	99.6	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	137	99.3	96.5	Afibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, 616004 Hypofibrinogenemia, congenital, 202400
FH	146.4	91.7	87.6	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800

FHL1	87.2	98.8	93	?Uruguay faciocardiomusculoskeletal syndrome, 300280 Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 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
FIBP	123.3	100	100	Thauvin-Robinet-Faivre syndrome, 617107
FIG4	154.9	99.8	98.4	?Polymicrogyria, bilateral temporooccipital, 612691 Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228 Yunis-Varon syndrome, 216340
FIGLA	81.8	94.3	89.4	Premature ovarian failure 6, 612310
FKBP10	158.6	96.9	92.8	Bruck syndrome 1, 259450 Osteogenesis imperfecta, type XI, 610968
FKBP14	74.3	100	99.4	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557
FKRP	94.5	100	99.7	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	120	99.2	94.2	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	191.6	100	98.9	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100
FLCN	160.5	100	99.5	Birt-Hogg-Dube syndrome, 135150 Colorectal cancer, somatic, 114500 Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700
FLG	234.1	100	99.9	Ichthyosis vulgaris, 146700 {Dermatitis, atopic, susceptibility to, 2}, 605803
FLG2	691.5	100	100	Peeling skin syndrome 6, 618084

FLI1	189.6	99.4	97.7	Bleeding disorder, platelet-type, 21, 617443
FLNA	138.1	100	99.5	?FG syndrome 2, 300321 Cardiac valvular dysplasia, X-linked, 314400 Congenital short bowel syndrome, 300048 Frontometaphyseal dysplasia 1, 305620 Heterotopia, periventricular, 1, 300049 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Terminal osseous dysplasia, 300244
FLNB	149.9	99.8	99.2	Atelosteogenesis, type I, 108720 Atelosteogenesis, type III, 108721 Boomerang dysplasia, 112310 Larsen syndrome, 150250 Spondylocarpotarsal synostosis syndrome, 272460
FLNC	165	100	99.7	Cardiomyopathy, familial hypertrophic, 26, 0 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Myopathy, myofibrillar, 5, 609524
FLRT3	225.7	100	100	Hypogonadotropic hypogonadism 21 with anosmia, 615271
FLT3	134.7	99.7	98.2	Leukemia, acute lymphoblastic, somatic, 613065 Leukemia, acute myeloid, reduced survival in, somatic, 601626 Leukemia, acute myeloid, somatic, 601626
FLT4	155.9	98.6	97.9	Hemangioma, capillary infantile, somatic, 602089 Lymphatic malformation 1, 153100
FLVCR1	139.5	99.2	95.8	Ataxia, posterior column, with retinitis pigmentosa, 609033
FLVCR2	159.7	100	100	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790
FMN2	86.6	83.1	77.7	Mental retardation, autosomal recessive 47, 616193
FMO3	153.5	99.9	99.2	Trimethylaminuria, 602079
FMR1	78.9	94	84.7	Fragile X syndrome, 300624 Fragile X tremor/ataxia syndrome, 300623 Premature ovarian failure 1, 311360

FN1	145.4	100	99.5	Glomerulopathy with fibronectin deposits 2, 601894 Plasma fibronectin deficiency, 614101 Spondylometaphyseal dysplasia, corner fracture type, 184255
FOLR1	150.4	100	100	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXC1	32.7	86	68.5	Anterior segment dysgenesis 3, multiple subtypes, 601631 Axenfeld-Rieger syndrome, type 3, 602482
FOXC2	44.3	95.2	78.8	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXE1	29.3	72.3	56.2	Bamforth-Lazarus syndrome, 241850 {Thyroid cancer, nonmedullary, 4}, 616534
FOXE3	20.6	69	47.8	Anterior segment dysgenesis 2, multiple subtypes, 610256 Cataract 34, multiple types, 612968 {Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349
FOXF1	93.5	98.9	93.7	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380
FOXG1	157.5	84.8	81	Rett syndrome, congenital variant, 613454
FOXI1	152.5	100	100	Enlarged vestibular aqueduct, 600791
FOXL2	39.5	92.8	71	Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 Premature ovarian failure 3, 608996
FOXN1	112.5	100	99.5	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXO1	139	92.8	85.7	Rhabdomyosarcoma, alveolar, 268220
FOXP1	129.6	100	99.9	Mental retardation with language impairment and with or without autistic features, 613670
FOXP2	160.2	98.9	96.9	Speech-language disorder-1, 602081
FOXP3	124.6	98.7	91.6	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790 {Diabetes mellitus, type I, susceptibility to}, 222100
FOXRED1	136.6	100	99.6	Mitochondrial complex I deficiency, nuclear type 19, 618241
FRAS1	147.8	100	99.7	Fraser syndrome 1, 219000
FREM1	138.4	99.9	99.1	Bifid nose with or without anorectal and renal anomalies, 608980 Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485

FREM2	182.4	100	99.5	Fraser syndrome 2, 617666
FRMD4A	116.7	91.4	90.3	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819
FRMD7	114.2	99.9	98.8	Nystagmus 1, congenital, X-linked, 310700 Nystagmus, infantile periodic alternating, X-linked, 310700
FRMPD4	114.1	99.7	98	Mental retardation, X-linked 104, 300983
FRRS1L	103.1	68.3	63.4	Epileptic encephalopathy, early infantile, 37, 616981
FSCN2	145.1	100	100	Retinitis pigmentosa 30, 607921
FSHB	149.1	100	100	Hypogonadotropic hypogonadism 24 without anosmia, 229070
FSHR	140.9	100	98.8	Ovarian dysgenesis 1, 233300 Ovarian hyperstimulation syndrome, 608115 Ovarian response to FSH stimulation, 276400
FSIP2	95.6	99.3	97	Spermatogenic failure 34, 618153
FTCD	89.8	94.6	89.8	Glutamate formiminotransferase deficiency, 229100
FTH1	96.4	95.4	84.2	?Hemochromatosis, type 5, 615517
FTL	147.7	99	93.2	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159
FTO	118.9	83.7	82.5	Growth retardation, developmental delay, facial dysmorphism, 612938 {Obesity, susceptibility to, BMIQ14}, 612460
FTSJ1	128.3	98.1	94.5	Mental retardation, X-linked 9/44, 309549
FUCA1	135	100	99.5	Fucosidosis, 230000
FUS	137.4	97.6	94.7	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 Essential tremor, hereditary, 4, 614782
FUT6	165.8	100	100	Fucosyltransferase 6 deficiency, 613852
FUT8	166.1	99.9	98.8	Congenital disorder of glycosylation with defective fucosylation, 618005
FUZ	118.8	100	100	Neural tube defects, 182940
FXN	75.2	85.7	75.9	Friedreich ataxia, 229300 Friedreich ataxia with retained reflexes, 229300
FXYD2	96.7	99.8	99.2	Hypomagnesemia 2, renal, 154020
FYB1	92.3	97.7	91.9	Thrombocytopenia 3, 273900
FYCO1	123.7	100	100	Cataract 18, autosomal recessive, 610019
FZD4	224.2	100	99.8	Exudative vitreoretinopathy 1, 133780

				Retinopathy of prematurity, 133780
FZD6	208.6	100	100	Nail disorder, nonsyndromic congenital, 10, (claw-shaped nails), 614157
G6PC	180.7	100	100	Glycogen storage disease Ia, 232200
G6PC3	123.7	100	100	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
G6PD	118.3	99.5	97.5	Hemolytic anemia, G6PD deficient (favism), 300908 {Resistance to malaria due to G6PD deficiency}, 611162
GAA	128.5	100	99.9	Glycogen storage disease II, 232300
GAB1	170	100	99.7	?Deafness, autosomal recessive 26, 605428
GABBR2	136	95.2	92.4	Epileptic encephalopathy, early infantile, 59, 617904 Neurodevelopmental disorder with poor language and loss of hand skills, 617903 {Nicotine dependence, protection against}, 188890 {Nicotine dependence, susceptibility to}, 188890
GABRA1	179.5	100	100	Epileptic encephalopathy, early infantile, 19, 615744 {Epilepsy, childhood absence, susceptibility to, 4}, 611136 {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136
GABRB1	174.1	100	99.9	Epileptic encephalopathy, early infantile, 45, 617153
GABRB2	148.9	100	100	Epileptic encephalopathy, infantile or early childhood, 2, 617829
GABRB3	140.5	98.1	93.7	Epileptic encephalopathy, early infantile, 43, 617113 {Epilepsy, childhood absence, susceptibility to, 5}, 612269
GABRG2	137.9	91.1	90.3	Epilepsy, generalized, with febrile seizures plus, type 3, 611277 Febrile seizures, familial, 8, 611277 {Epilepsy, childhood absence, susceptibility to, 2}, 607681
GAD1	128.7	99.9	98.4	?Cerebral palsy, spastic quadriplegic, 1, 603513
GAL	164.2	100	98.4	?Epilepsy, familial temporal lobe, 8, 616461
GALC	100.6	98.9	94.6	Krabbe disease, 245200
GALE	154.8	100	100	Galactose epimerase deficiency, 230350
GALK1	125.4	100	99.7	Galactokinase deficiency with cataracts, 230200
GALNS	93.2	99	95.6	Mucopolysaccharidosis IVA, 253000

GALNT3	128.2	99.2	96	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GALT	168.7	100	100	Galactosemia, 230400
GAMT	93.5	90.9	80.7	Cerebral creatine deficiency syndrome 2, 612736
GAN	190	100	99.9	Giant axonal neuropathy-1, 256850
GANAB	120.1	99.9	98.9	Polycystic kidney disease 3, 600666
GARS	125.7	99.9	98.5	Charcot-Marie-Tooth disease, type 2D, 601472 Neuropathy, distal hereditary motor, type VA, 600794
GAS8	150.7	99.8	99.4	Ciliary dyskinesia, primary, 33, 616726
GATA1	83.5	99.6	95.7	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	99.9	98.5	Emberger syndrome, 614038 Immunodeficiency 21, 614172 {Leukemia, acute myeloid, susceptibility to}, 601626 {Myelodysplastic syndrome, susceptibility to}, 614286
GATA3	186.8	100	100	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GATA4	87.4	68.6	60.7	?Testicular anomalies with or without congenital heart disease, 615542 Atrial septal defect 2, 607941 Atrioventricular septal defect 4, 614430 Tetralogy of Fallot, 187500 Ventricular septal defect 1, 614429
GATA5	44.2	98.3	84.4	Congenital heart defects, multiple types, 5, 617912
GATA6	61.7	83.7	72.1	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
GATAD1	126.6	97	92	?Cardiomyopathy, dilated, 2B, 614672
GATAD2B	129.8	100	99.8	Mental retardation, autosomal dominant 18, 615074
GATM	150.6	100	100	Cerebral creatine deficiency syndrome 3, 612718

GBA	240.3	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	176.2	99.9	99.3	Spastic paraplegia 46, autosomal recessive, 614409
GBE1	145.5	99.6	97.2	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GCDH	147.6	99.9	99.1	Glutaricaciduria, type I, 231670
GCH1	74.4	97	86.5	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GCK	141.4	100	100	Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, permanent neonatal, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485 MODY, type II, 125851
GCLC	133.2	99.9	98.9	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 {Myocardial infarction, susceptibility to}, 608446
GCM2	161.8	100	100	Hyperparathyroidism 4, 617343 Hypoparathyroidism, familial isolated, 146200
GCNT2	166.8	99.5	99.5	Adult i phenotype without cataract, 110800 Cataract 13 with adult i phenotype, 116700 [Blood group, Ii], 110800
GCSH	34.2	83.1	67.8	?Glycine encephalopathy, 605899
GDAP1	163.1	99.3	96.1	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	19.5	65	48.4	Congenital heart defects, multiple types, 6, 613854 Right atrial isomerism (Ivemark), 208530
GDF2	163.2	100	100	Telangiectasia, hereditary hemorrhagic, type 5, 615506

GDF3	134.9	100	100	Klippel-Feil syndrome 3, autosomal dominant, 613702 Microphthalmia with coloboma 6, 613703 Microphthalmia, isolated 7, 613704
GDF5	141.8	100	100	?Acromesomelic dysplasia, Hunter-Thompson type, 201250 Brachydactyly, type A1, C, 615072 Brachydactyly, type A2, 112600 Brachydactyly, type C, 113100 Chondrodysplasia, Grebe type, 200700 Du Pan syndrome, 228900 Multiple synostoses syndrome 2, 610017 Symphalangism, proximal, 1B, 615298 {Osteoarthritis-5}, 612400
GDF6	75.2	98.7	89	Klippel-Feil syndrome 1, autosomal dominant, 118100 Leber congenital amaurosis 17, 615360 Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094 Multiple synostoses syndrome 4, 617898
GDF9	169.5	100	100	?Premature ovarian failure 14, 618014
GDI1	152	98.9	97.3	Mental retardation, X-linked 41, 300849
GDNF	185.9	99.9	98.8	Central hypoventilation syndrome, 209880 {Hirschsprung disease, susceptibility to, 3}, 613711 {Pheochromocytoma, modifier of}, 171300
GEMIN4	167.1	100	99.9	Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities, 617913
GFAP	102.6	91.7	90.3	Alexander disease, 203450
GFER	76.1	92.9	75.4	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076
GFI1	83.1	99	92.9	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 ?Neutropenia, severe congenital 2, autosomal dominant, 613107
GFI1B	170.2	98.2	95.7	Bleeding disorder, platelet-type, 17, 187900
GFM1	100.3	99.2	95.3	Combined oxidative phosphorylation deficiency 1, 609060
GFPT1	144.4	99.9	97.6	Myasthenia, congenital, 12, with tubular aggregates, 610542

GGCX	115.3	100	99.7	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842 Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450
GGT1	12.9	19.8	17.2	?Glutathioninuria, 231950
GH1	175.2	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	212.3	99.8	99.5	Growth hormone insensitivity, partial, 604271 Increased responsiveness to growth hormone, 604271 Laron dwarfism, 262500 {Hypercholesterolemia, familial, modifier of}, 143890
GHRHR	116.4	95.3	94.7	Growth hormone deficiency, isolated, type IV, 618157
GHSR	206	99.9	98.4	Growth hormone deficiency, isolated partial, 615925
GIF	141.2	100	99.9	Intrinsic factor deficiency, 261000
GINS1	124.3	96.6	83.2	Immunodeficiency 55, 617827
GIPC3	103.3	91.8	85.5	Deafness, autosomal recessive 15, 601869
GJA1	246.4	100	100	Atrioventricular septal defect 3, 600309 Craniometaphyseal dysplasia, autosomal recessive, 218400 Erythrokeratodermia variabilis et progressiva 3, 617525 Hypoplastic left heart syndrome 1, 241550 Oculodentodigital dysplasia, 164200 Oculodentodigital dysplasia, autosomal recessive, 257850 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100
GJA3	164.7	100	99.5	Cataract 14, multiple types, 601885
GJA5	268.4	100	100	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic (GJA5/SCN5A), 108770
GJA8	153.6	100	100	Cataract 1, multiple types, 116200
GJB1	229.8	100	99.8	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800

GJB2	205.1	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	308.9	100	100	Deafness, autosomal dominant 2B, 612644 Deafness, autosomal dominant, with peripheral neuropathy, 0 Deafness, autosomal recessive, 0 Deafness, digenic, GJB2/GJB3, 220290 Erythrokeratodermia variabilis et progressiva 1, 133200
GJB4	369.9	100	100	Erythrokeratodermia variabilis et progressiva 2, 617524
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	41.9	68.9	58.6	Leukodystrophy, hypomyelinating, 2, 608804 Lymphatic malformation 3, 613480 Spastic paraplegia 44, autosomal recessive, 613206
GK	43.6	74	54.7	Glycerol kinase deficiency, 307030
GLA	81.3	99.7	97.6	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	94.3	99.6	97	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLDC	78.9	90.6	82.7	Glycine encephalopathy, 605899
GLDN	130.4	98.2	91.8	Lethal congenital contracture syndrome 11, 617194
GLE1	110.8	100	99.7	Arthrogryposis, lethal, with anterior horn cell disease, 611890 Lethal congenital contracture syndrome 1, 253310

GLI1	129.7	100	99.9	Polydactyly, postaxial, type A8, 618123
GLI2	138.5	99.4	97.4	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829
GLI3	154.2	100	99.7	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	99.9	98.2	Nephronophthisis 7, 611498
GLIS3	133.4	99.9	99.3	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GLMN	66.8	97.8	86.4	Glomuvenous malformations, 138000
GLRA1	123.3	100	100	Hyperekplexia 1, 149400
GLRB	98.6	96.6	88.8	Hyperekplexia 2, 614619
GLRX5	108.2	92.6	83.8	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GLUD1	74.5	94.4	84.3	Hyperinsulinism-hyperammonemia syndrome, 606762
GLUL	108.8	99.9	98.2	Glutamine deficiency, congenital, 610015
GLYCTK	202.6	100	99.5	D-glyceric aciduria, 220120
GM2A	139.6	100	100	GM2-gangliosidosis, AB variant, 272750
GMNN	101.2	92.6	83.8	Meier-Gorlin syndrome 6, 616835
GMPPA	136.8	100	99.9	Alacrima, achalasia, and mental retardation syndrome, 615510
GMPPB	228.5	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	149.5	99.5	96.4	Hypocalcemia, autosomal dominant 2, 615361 Hypocalciuric hypercalcemia, type II, 145981
GNAI2	138.5	100	99.9	Pituitary ACTH-secreting adenoma, 0 Ventricular tachycardia, idiopathic, 192605

GNAI3	110.6	99	92	Auriculocondylar syndrome 1, 602483
GNAL	134.9	94.5	91.6	Dystonia 25, 615073
GNAO1	167.8	93.8	93.8	Epileptic encephalopathy, early infantile, 17, 615473 Neurodevelopmental disorder with involuntary movements, 617493
GNAQ	82.6	81.1	69.5	Capillary malformations, congenital, 1, somatic, mosaic, 163000 Sturge-Weber syndrome, somatic, mosaic, 185300
GNAS	141	98.5	95.9	ACTH-independent macronodular adrenal hyperplasia, 219080 McCune-Albright syndrome, somatic, mosaic, 174800 Osseous heteroplasia, progressive, 166350 Pituitary adenoma 3, multiple types, somatic, 617686 Pseudohypoparathyroidism Ia, 103580 Pseudohypoparathyroidism Ib, 603233 Pseudohypoparathyroidism Ic, 612462 Pseudopseudohypoparathyroidism, 612463
GNAS-AS1	NC	NC	NC	Pseudohypoparathyroidism, type IB, 603233
GNAT1	153.9	100	100	Night blindness, congenital stationary, autosomal dominant 3, 610444 Night blindness, congenital stationary, type 1G, 616389
GNAT2	130.7	99.9	99.1	Achromatopsia 4, 613856
GNB1	189	100	100	Leukemia, acute lymphoblastic, somatic, 613065 Mental retardation, autosomal dominant 42, 616973
GNB3	179	100	100	Night blindness, congenital stationary, type 1H, 617024 {Hypertension, essential, susceptibility to}, 145500
GNB4	152.6	100	99.7	Charcot-Marie-Tooth disease, dominant intermediate F, 615185
GNB5	125.9	99.9	98.3	Intellectual developmental disorder with cardiac arrhythmia, 617173 Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182
GNE	153.7	100	99.8	Nonaka myopathy, 605820 Sialuria, 269921
GNMT	147.2	99.4	97	Glycine N-methyltransferase deficiency, 606664
GNPAT	133.6	99.4	96.4	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	167.7	98.3	97.4	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GNPTG	151.6	96.1	89.7	Mucolipidosis III gamma, 252605

GNRH1	86.7	99.5	91.5	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841
GNRHR	161.9	100	100	Hypogonadotropic hypogonadism 7 without anosmia, 146110
GNS	107.9	96.9	92	Mucopolysaccharidosis type IIID, 252940
GORAB	176.3	99.7	97.8	Geroderma osteodysplasticum, 231070
GOSR2	127.2	95.9	95	Epilepsy, progressive myoclonic 6, 614018
GOT1	127	100	99.5	Aspartate aminotransferase, serum level of, QTL1, 614419
GP1BA	153	97	94.3	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	74.2	64.3	Bernard-Soulier syndrome, type B, 231200 Giant platelet disorder, isolated, 231200
GP6	136.5	100	100	Bleeding disorder, platelet-type, 11, 614201
GP9	123.3	96.6	89.3	Bernard-Soulier syndrome, type C, 231200
GPAA1	130	96.4	95.2	Glycosylphosphatidylinositol biosynthesis defect 15, 617810
GPC3	85.1	98.6	92.6	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPC6	142	100	100	Omodysplasia 1, 258315
GPD1	93.5	99.9	99.1	Hypertriglyceridemia, transient infantile, 614480
GPD1L	138.3	100	98.5	Brugada syndrome 2, 611777
GPHN	167.2	98.4	96.9	Molybdenum cofactor deficiency C, 615501
GPI	142.6	100	99.5	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470
GPIHBP1	119.7	99.9	99.2	Hyperlipoproteinemia, type 1D, 615947
GPNMB	177.8	100	100	Amyloidosis, primary localized cutaneous, 3, 617920
GPR101	159	100	100	Pituitary adenoma 2, GH-secreting, 300943
GPR143	61.5	85.3	75.5	Nystagmus 6, congenital, X-linked, 300814 Ocular albinism, type I, Nettleship-Falls type, 300500
GPR179	133.6	100	99.7	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
GPR68	186.4	96.5	90.2	Amelogenesis imperfecta, hypomaturation type, IIA6, 617217
GPR88	89.9	98.8	94.1	?Chorea, childhood-onset, with psychomotor retardation, 616939

GPRASP2	87.3	100	99.5	?Deafness, X-linked 7, 301018
GPSM2	112.9	99.8	97	Chudley-McCullough syndrome, 604213
GPT2	138	98.2	90.9	Mental retardation, autosomal recessive 49, 616281
GPX4	119.2	85.2	76.6	Spondylometaphyseal dysplasia, Sedaghatian type, 250220
GREB1L	148.6	100	99.5	Renal hypodysplasia/aplasia 3, 617805
GREM2	133.7	100	100	Tooth agenesis, selective, 9, 617275
GRHL2	134.6	100	100	Corneal dystrophy, posterior polymorphous, 4, 618031 Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029
GRHL3	140.7	100	99.9	Van der Woude syndrome 2, 606713
GRHPR	112.5	85.1	78.2	Hyperoxaluria, primary, type II, 260000
GRIA3	98.2	99.3	94.7	Mental retardation, X-linked 94, 300699
GRIA4	148.3	99.8	98.1	Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864
GRID2	175.4	100	99.9	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRIK2	133.2	96	94.7	Mental retardation, autosomal recessive, 6, 611092
GRIN1	150.7	100	99.5	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254 Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820
GRIN2A	159.1	100	100	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570
GRIN2B	189.4	99.9	99.3	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation, autosomal dominant 6, 613970
GRIN2D	73.1	69.1	62.1	Epileptic encephalopathy, early infantile, 46, 617162
GRIP1	130.8	100	99.9	Fraser syndrome 3, 617667
GRK1	126.4	100	99.9	Oguchi disease-2, 613411
GRM1	185.8	100	99.9	Spinocerebellar ataxia 44, 617691 Spinocerebellar ataxia, autosomal recessive 13, 614831
GRM6	151.7	93.3	86.7	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270
GRN	184.5	100	100	Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
GRXCR1	183.7	100	99.8	Deafness, autosomal recessive 25, 613285

GRXCR2	114.9	100	100	?Deafness, autosomal recessive 101, 615837
GSC	85.4	86.9	74.5	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471
GSN	119.2	94.2	89	Amyloidosis, Finnish type, 105120
GSS	104	100	99.8	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900
GTF2E2	83.5	96.5	91	Trichothiodystrophy 6, nonphotosensitive, 616943
GTF2H5	113.6	100	99.1	Trichothiodystrophy 3, photosensitive, 616395
GTPBP2	156.8	96.5	94.7	Jaberi-Elahi syndrome, 617988
GTPBP3	137.4	100	99.7	Combined oxidative phosphorylation deficiency 23, 616198
GUCA1A	160.6	100	100	Cone dystrophy-3, 602093 Cone-rod dystrophy 14, 602093
GUCA1B	144.2	100	99.9	Retinitis pigmentosa 48, 613827
GUCY1A3	171.3	99.4	98	Moyamoya 6 with achalasia, 615750
GUCY2C	135.2	100	99.7	Diarrhea 6, 614616 Meconium ileus, 614665
GUCY2D	91.3	98.3	91.1	?Choroidal dystrophy, central areolar 1, 215500 Cone-rod dystrophy 6, 601777 Leber congenital amaurosis 1, 204000
GUF1	70.5	98.7	92.3	?Epileptic encephalopathy, early infantile, 40, 617065
GULOP	NC	NC	NC	Scurvy, 0
GUSB	116.1	92.2	89.4	Mucopolysaccharidosis VII, 253220
GYG1	157.8	100	99.6	?Glycogen storage disease XV, 613507 Polyglucosan body myopathy 2, 616199
GYS1	110.8	100	98.5	Glycogen storage disease 0, muscle, 611556
GYS2	150.2	98.5	93.9	Glycogen storage disease 0, liver, 240600
GZF1	229	100	100	Joint laxity, short stature, and myopia, 617662
H19	NC	NC	NC	Beckwith-Wiedemann syndrome, 130650 Silver-Russell syndrome, 180860 Wilms tumor 2, 194071
H6PD	169.5	99	99	Cortisone reductase deficiency 1, 604931
HAAO	100.8	100	99.9	Vertebral, cardiac, renal, and limb defects syndrome 1, 617660
HACE1	125.3	99.2	95	Spastic paraplegia and psychomotor retardation with or without seizures, 616756

HADH	110.8	98	95.1	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HADHA	84.4	96.5	90.3	Fatty liver, acute, of pregnancy, 609016 HELLP syndrome, maternal, of pregnancy, 609016 LCHAD deficiency, 609016 Trifunctional protein deficiency, 609015
HADHB	80.5	92.5	79.5	Trifunctional protein deficiency, 609015
HAMP	175.6	100	100	Hemochromatosis, type 2B, 613313
HARS	159.4	100	100	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504
HARS2	169.7	99.9	99.2	?Perrault syndrome 2, 614926
HAX1	136.5	100	100	Neutropenia, severe congenital 3, autosomal recessive, 610738
HBA1	141.9	100	100	Erythrocytosis, 7, 617981 Heinz body anemias, alpha-, 140700 Hemoglobin H disease, nondeletional, 613978 Methemoglobinemia, alpha type, 617973 Thalassemias, alpha-, 604131
HBA2	133.7	97.8	91.1	Erythrocytosis 7, 617981 Heinz body anemia, 140700 Hemoglobin H disease, deletional and nondeletional, 613978 Thalassemia, alpha-, 604131
HBB	176.7	100	100	Delta-beta thalassemia, 141749 Erythrocytosis 6, 617980 Heinz body anemia, 140700 Hereditary persistence of fetal hemoglobin, 141749 Methmoglobinemia, beta type, 617971 Sickle cell anemia, 603903 Thalassemia, beta, 613985 Thalassemia-beta, dominant inclusion-body, 603902 {Malaria, resistance to}, 611162
HBD	242.4	100	100	Thalassemia due to Hb Lepore, 0 Thalassemia, delta-, 0
HBG1	174.4	98.3	94.7	Fetal hemoglobin quantitative trait locus 1, 141749

HBG2	329.4	100	100	Cyanosis, transient neonatal, 613977 Fetal hemoglobin quantitative trait locus 1, 141749
HCCS	106.6	99.9	99.2	Linear skin defects with multiple congenital anomalies 1, 309801
HCFC1	105.8	99.3	96.1	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541
HCN1	122.4	99.9	97.8	Epileptic encephalopathy, early infantile, 24, 615871
HCN4	79.4	98.3	91.8	Brugada syndrome 8, 613123 Sick sinus syndrome 2, 163800
HCRT	54.2	77.4	66.8	?Narcolepsy 1, 161400
HDAC6	118.7	99.7	97	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863
HDAC8	131.9	100	99.8	Cornelia de Lange syndrome 5, 300882
HECW2	133.1	99.9	98.6	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268
HELLS	93.7	94.1	86.8	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911
HEPACAM	142.3	81.4	76.1	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926
HERC1	173.7	99.9	99.4	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
HERC2	114.4	80.9	77.9	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	29.1	64.9	42.6	Spondylocostal dysostosis 4, autosomal recessive, 613686
HESX1	57.6	99.2	92.6	Growth hormone deficiency with pituitary anomalies, 182230 Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230
HEXA	118.3	93.8	92.2	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800 [Hex A pseudodeficiency], 272800
HEXB	129.7	99.4	94	Sandhoff disease, infantile, juvenile, and adult forms, 268800

HFE	142	100	99.7	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	116.8	100	100	Hemochromatosis, type 2A, 602390
HFM1	42.8	90.3	76.7	Premature ovarian failure 9, 615724
HGD	127.8	100	99.8	Alkaptonuria, 203500
HGF	146.9	99.4	96.9	Deafness, autosomal recessive 39, 608265
HGSNAT	101	86.4	85.7	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544
HIBCH	67.7	92.7	69.5	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HIKESHI	61.7	90.9	79.7	Leukodystrophy, hypomyelinating, 13, 616881
HINT1	60	98.5	88	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200
HIST1H1E	60.9	99.9	94.7	Rahman syndrome, 617537
HIVEP2	191.2	100	100	Mental retardation, autosomal dominant 43, 616977
HK1	143.4	100	99.9	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285 Retinitis pigmentosa 79, 617460
HLCS	172.8	100	100	Holocarboxylase synthetase deficiency, 253270
HMBS	109	100	99.8	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000
HMGB3	38	88.1	68.6	?Microphthalmia, syndromic 13, 300915
HMGCL	143.3	100	99.9	HMG-CoA lyase deficiency, 246450
HMGCS2	131.5	100	100	HMG-CoA synthase-2 deficiency, 605911
HMOX1	128.7	95.8	89.5	Heme oxygenase-1 deficiency, 614034 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963
HMX1	23	56.8	42	Oculoauricular syndrome, 612109

HNF1A	156.7	100	99.4	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	123.7	99.9	98.9	Diabetes mellitus, noninsulin-dependent, 125853 Renal cysts and diabetes syndrome, 137920 {Renal cell carcinoma}, 144700
HNF4A	140.2	99.9	99.1	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026 MODY, type I, 125850 {Diabetes mellitus, noninsulin-dependent}, 125853
HNMT	135	100	99.7	Mental retardation, autosomal recessive 51, 616739 {Asthma, susceptibility to}, 600807
HNRNPA1	72.7	97.4	87.2	?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3, 615424 Amyotrophic lateral sclerosis 20, 615426
HNRNPA2B1	134.7	98.3	96.9	?Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2, 615422
HNRNPDL	59	90.6	79.8	Muscular dystrophy, limb-girdle, autosomal dominant 3, 609115
HNRNPH2	170.8	100	100	Mental retardation, X-linked, syndromic, Bain type, 300986
HNRNPK	71.7	86.9	78.4	Au-Kline syndrome, 616580
HNRNPU	126.6	99.3	97.9	Epileptic encephalopathy, early infantile, 54, 617391
HOGA1	147.5	99.8	98.1	Hyperoxaluria, primary, type III, 613616
HOMER2	133.5	99.5	99.4	?Deafness, autosomal dominant 68, 616707
HOXA1	165.3	100	100	Athabaskan brainstem dysgenesis syndrome, 601536 Bosley-Salih-Alorainy syndrome, 601536
HOXA11	86.3	88	78.5	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
HOXA13	49	69.2	61.7	Guttmacher syndrome, 176305 Hand-foot-uterus syndrome, 140000
HOXA2	74.1	99.5	95.6	?Microtia, hearing impairment, and cleft palate (AR), 612290 Microtia with or without hearing impairment (AD), 612290
HOXB1	104.5	100	100	Facial paresis, hereditary congenital, 3, 614744

HOXC13	104.9	97.5	91.1	Ectodermal dysplasia 9, hair/nail type, 614931
HOXD10	137.7	100	99.8	Charcot-Marie-Tooth disease, foot deformity of, 192950 Vertical talus, congenital, 192950
HOXD13	142.3	93.1	89.6	?Brachydactyly-syndactyly syndrome, 610713 Brachydactyly, type D, 113200 Brachydactyly, type E, 113300 Syndactyly, type V, 186300 Synpolydactyly 1, 186000
HPCA	283.4	100	100	Dystonia 2, torsion, autosomal recessive, 224500
HPD	137.8	100	100	Hawkinsinuria, 140350 Tyrosinemia, type III, 276710
HPGD	88	100	98.5	Cranioosteoarthropathy, 259100 Digital clubbing, isolated congenital, 119900 Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100
HPRT1	58.2	96	84.8	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322
HPS1	117.8	100	99.3	Hermansky-Pudlak syndrome 1, 203300
HPS3	135.2	99.6	96.4	Hermansky-Pudlak syndrome 3, 614072
HPS4	141.9	100	100	Hermansky-Pudlak syndrome 4, 614073
HPS5	133	99.9	98.7	Hermansky-Pudlak syndrome 5, 614074
HPS6	139.1	91	84.3	Hermansky-Pudlak syndrome 6, 614075
HPSE2	110.2	98.8	95.4	Urofacial syndrome 1, 236730
HR	94.9	97.3	94.2	Alopecia universalis, 203655 Atrichia with papular lesions, 209500 Hypotrichosis 4, 146550
HRAS	164.7	99.8	98.1	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	178.3	95	94.3	Thrombophilia due to elevated HRG, 613116 Thrombophilia due to HRG deficiency, 613116

HSD11B1	147.1	100	99.9	Cortisone reductase deficiency 2, 614662
HSD11B2	165.2	85.7	82.5	Apparent mineralocorticoid excess, 218030
HSD17B10	117.1	100	99.2	HSD10 mitochondrial disease, 300438
HSD17B3	156.4	100	100	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	95.1	93.9	90.8	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B2	189.2	100	100	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810
HSD3B7	136.3	98.2	91	Bile acid synthesis defect, congenital, 1, 607765
HSF4	103	97.6	94.9	Cataract 5, multiple types, 116800
HSPA9	91.6	91.1	85.9	Anemia, sideroblastic, 4, 182170 Even-plus syndrome, 616854
HSPB1	39.7	93.7	81.8	Charcot-Marie-Tooth disease, axonal, type 2F, 606595 Neuropathy, distal hereditary motor, type IIB, 608634
HSPB3	290.9	100	100	?Neuronopathy, distal hereditary motor, type IIC, 613376
HSPB8	156.4	100	100	Charcot-Marie-Tooth disease, axonal, type 2L, 608673 Neuropathy, distal hereditary motor, type IIA, 158590
HSPD1	96.5	98.3	93.2	Leukodystrophy, hypomyelinating, 4, 612233 Spastic paraplegia 13, autosomal dominant, 605280
HSPG2	121.3	99.4	98.2	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
HTR1A	220.4	100	100	Periodic fever, menstrual cycle dependent, 614674
HTRA1	98.2	84.5	81.4	CARASIL syndrome, 600142 Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779 {Macular degeneration, age-related, 7}, 610149 {Macular degeneration, age-related, neovascular type}, 610149
HTRA2	122.1	100	99.7	3-methylglutaconic aciduria, type VIII, 617248 {Parkinson disease 13}, 610297
HTT	143.7	98.5	97.1	Huntington disease, 143100 Lopes-Maciel-Rodan syndrome, 617435
HUWE1	98.4	99.2	97	Mental retardation, X-linked syndromic, Turner type, 300706
HYAL1	115.3	100	100	?Mucopolysaccharidosis type IX, 601492
HYDIN	133.6	99.9	99.5	Ciliary dyskinesia, primary, 5, 608647
HYLS1	171.1	100	100	Hydrolethalus syndrome, 236680

HYOU1	134	99.9	99.7	?Immunodeficiency 59 and hypoglycemia, 233600
IARS	148.8	99.8	98.6	Growth retardation, intellectual developmental disorder, hypotonia, and hepatopathy, 617093
IARS2	131.5	100	99.9	?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
IBA57	113.3	93.3	89.5	?Spastic paraplegia 74, autosomal recessive, 616451 Multiple mitochondrial dysfunctions syndrome 3, 615330
ICK	131.6	99.9	98.2	Endocrine-cerebroosteodysplasia, 612651 {Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924
ICOS	160.2	100	100	Immunodeficiency, common variable, 1, 607594
IDH2	103.5	99.6	96.9	D-2-hydroxyglutaric aciduria 2, 613657
IDH3B	165.5	95.9	95.4	Retinitis pigmentosa 46, 612572
IDS	111.3	99.6	98.3	Mucopolysaccharidosis II, 309900
IDUA	123	88.1	80	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016
IER3IP1	73	93.2	82.2	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	113.5	99.6	97.1	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFITM5	63.4	99.4	94.9	Osteogenesis imperfecta, type V, 610967
IFNAR2	138.8	98.5	95.4	?Immunodeficiency 45, 616669 {Hepatitis B virus, susceptibility to}, 610424
IFNGR1	138.5	99.2	97.3	Immunodeficiency 27A, mycobacteriosis, AR, 209950 Immunodeficiency 27B, mycobacteriosis, AD, 615978 {H. pylori infection, susceptibility to}, 600263 {Hepatitis B virus infection, susceptibility to}, 610424 {Tuberculosis infection, protection against}, 607948 {Tuberculosis, susceptibility to}, 607948
IFNGR2	142.3	93.2	93.1	Immunodeficiency 28, mycobacteriosis, 614889
IFT122	152	100	99.9	Cranioectodermal dysplasia 1, 218330
IFT140	114.7	99.9	99	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	116.5	100	99.6	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	131.7	100	99.6	?Bardet-Biedl syndrome 19, 615996

IFT43	114.8	100	100	?Cranioectodermal dysplasia 3, 614099 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866
IFT52	123.6	100	99.3	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102
IFT57	109.9	99	94	?Orofaciodigital syndrome XVIII, 617927
IFT74	80.1	97.8	88.2	?Bardet-Biedl syndrome 20, 617119
IFT80	57.8	87.6	70.7	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IFT81	92.9	88.3	81.2	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IGBP1	118.3	99.8	97.7	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472
IGF1	122.5	100	100	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IGF1R	144	100	99.8	Insulin-like growth factor I, resistance to, 270450
IGF2	100	100	100	?Growth restriction, severe, with distinctive facies, 616489
IGF2R	139.5	98.3	96.7	Hepatocellular carcinoma, somatic, 114550
IGFALS	79.7	99.9	96.8	Acid-labile subunit, deficiency of, 615961
IGFBP7	71.6	91.7	83.5	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis, 614224
IGHG2	34.5	79.6	60	IgG2 deficiency, selective, 0
IGHM	185.2	100	100	Agammaglobulinemia 1, 601495
IGHMBP2	107.8	99.3	96	Charcot-Marie-Tooth disease, axonal, type 2S, 616155 Neuronopathy, distal hereditary motor, type VI, 604320
IGKC	157.9	100	100	Kappa light chain deficiency, 614102
IGLL1	86.2	99.3	94.9	Agammaglobulinemia 2, 613500
IGSF1	84.8	99.5	96.6	Hypothyroidism, central, and testicular enlargement, 300888
IGSF3	115.9	96.7	95	?Lacrimal duct defect, 149700
IHH	129.3	100	100	Acrocapitofemoral dysplasia, 607778 Brachydactyly, type A1, 112500
IKBKB	123.5	98.5	94.2	Immunodeficiency 15A, 618204 Immunodeficiency 15B, 615592

IKBKG	52.5	84.6	73.2	Ectodermal dysplasia and immunodeficiency 1, 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, 13, 616873
IL10RA	141.9	100	99.9	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	168.8	98.7	96.1	Inflammatory bowel disease 25, early onset, autosomal recessive, 612567 {Hepatitis B virus, susceptibility to}, 610424
IL11RA	139.9	100	99.5	Craniosynostosis and dental anomalies, 614188
IL12B	121.1	100	99.9	Immunodeficiency 29, mycobacteriosis, 614890
IL12RB1	124.3	97	94.7	Immunodeficiency 30, 614891
IL17F	85.8	99.1	94.4	?Candidiasis, familial, 6, autosomal dominant, 613956
IL17RA	140.5	99.8	96.9	Immunodeficiency 51, 613953
IL17RC	96.1	99.8	99	Candidiasis, familial, 9, 616445
IL17RD	135.6	99.3	97.7	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
IL1RAPL1	116.8	99.7	98	Mental retardation, X-linked 21/34, 300143
IL1RN	162.8	100	100	Interleukin 1 receptor antagonist deficiency, 612852 {Gastric cancer risk after H. pylori infection}, 137215 {Microvascular complications of diabetes 4}, 612628
IL21	94.7	97.8	87.5	?Immunodeficiency, common variable, 11, 615767
IL21R	128.5	100	100	Immunodeficiency 56, 615207 [IgE, elevated level of], 147050
IL2RA	116.4	100	99.5	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367 {Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942
IL2RG	65.2	99.8	97.3	Combined immunodeficiency, X-linked, moderate, 312863 Severe combined immunodeficiency, X-linked, 300400
IL31RA	124.2	99.9	99.6	?Amyloidosis, primary localized cutaneous, 2, 613955
IL36RN	99	100	100	Psoriasis 14, pustular, 614204
IL7R	129.5	99.9	99.4	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
ILDR1	107.4	100	99.9	Deafness, autosomal recessive 42, 609646

IMPA1	71.4	97.1	85.8	Mental retardation, autosomal recessive 59, 617323
IMPAD1	147.2	99.9	99.4	Chondrodysplasia with joint dislocations, GPAPP type, 614078
IMPDH1	61.3	87.8	83.5	Leber congenital amaurosis 11, 613837 Retinitis pigmentosa 10, 180105
IMPG1	103.4	99.6	97.3	Macular dystrophy, vitelliform, 4, 616151
IMPG2	154.3	99.5	97.8	Macular dystrophy, vitelliform, 5, 616152 Retinitis pigmentosa 56, 613581
INF2	79.2	84.1	81.1	Charcot-Marie-Tooth disease, dominant intermediate E, 614455 Glomerulosclerosis, focal segmental, 5, 613237
ING1	127.8	99.5	95.4	Squamous cell carcinoma, head and neck, somatic, 275355
INPP5E	89.1	95.8	90	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INPP5K	108.3	100	99.6	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404
INPPL1	123.6	96.7	93.7	Opsismodysplasia, 258480
INS	87.7	100	99.7	Diabetes mellitus, insulin-dependent, 2, 125852 Diabetes mellitus, permanent neonatal, 606176 Hyperproinsulinemia, 616214 Maturity-onset diabetes of the young, type 10, 613370
INSL3	67.5	81	77.9	Cryptorchidism, 219050
INSR	141.1	97.1	94.5	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968 Leprechaunism, 246200 Rabson-Mendenhall syndrome, 262190
INTU	122	99.7	96.6	?Orofaciodigital syndrome XVII, 617926 ?Short-rib thoracic dysplasia 20 with polydactyly, 617925
INVS	160.5	100	100	Nephronophthisis 2, infantile, 602088
IQCB1	92.2	89.3	75.4	Senior-Loken syndrome 5, 609254
IQCE	132.2	98.8	96.6	?Polydactyly, postaxial, type A7, 617642
IQSEC2	61.2	92.1	82.5	Mental retardation, X-linked 1/78, 309530
IRAK4	95.4	98.3	90.1	Invasive pneumococcal disease, recurrent isolated, 1, 610799 IRAK4 deficiency, 607676

IRF1	185.4	100	100	Gastric cancer, somatic, 613659 Myelodysplastic syndrome, preleukemic, 0 Myelogenous leukemia, acute, 0 Nonsmall cell lung cancer, somatic, 211980
IRF2BP2	63.2	88.2	70.8	?Immunodeficiency, common variable, 14, 617765
IRF2BPL	112.1	95.8	87.4	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088
IRF6	113.7	99.9	97.9	Popliteal pterygium syndrome 1, 119500 van der Woude syndrome, 119300 {Orofacial cleft 6}, 608864
IRF7	89.8	99.9	99.1	?Immunodeficiency 39, 616345
IRF8	114.7	99.6	97.4	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893 Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990
IRX5	74.7	94.2	86.1	Hamamy syndrome, 611174
ISCA1	50.8	92.5	80.5	Multiple mitochondrial dysfunctions syndrome 5, 617613
ISCA2	92	99.7	96.9	Multiple mitochondrial dysfunctions syndrome 4, 616370
ISCU	111.2	100	99.7	Myopathy with lactic acidosis, hereditary, 255125
ISG15	160.1	100	100	Immunodeficiency 38, 616126
ISPD	104.4	95.2	84.8	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	95.4	94.8	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITGA2B	107.2	99.6	97.4	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Glanzmann thrombasthenia, 273800 Thrombocytopenia, neonatal alloimmune, BAK antigen related, 0
ITGA3	141.5	99.8	98.3	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748
ITGA6	146.5	99.8	99	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730
ITGA7	129.6	99.6	97.6	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
ITGA8	122.4	99.6	98.1	Renal hypodysplasia/aplasia 1, 191830
ITGB2	152.1	100	99.8	Leukocyte adhesion deficiency, 116920

ITGB3	142.2	99.3	97.4	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Glanzmann thrombasthenia, 273800 Purpura, posttransfusion, 0 Thrombocytopenia, neonatal alloimmune, 0 {Myocardial infarction, susceptibility to}, 608446
ITGB4	150.1	97.4	94.8	Epidermolysis bullosa of hands and feet, 131800 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa, junctional, with pyloric atresia, 226730
ITGB6	137	96.5	95.2	Amelogenesis imperfecta, type IH, 616221
ITK	125.2	100	99.6	Lymphoproliferative syndrome 1, 613011
ITM2B	110	99.7	97.9	?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079 Dementia, familial British, 176500 Dementia, familial Danish, 117300
ITPA	120.2	100	100	Epileptic encephalopathy, early infantile, 35, 616647 [Inosine triphosphatase deficiency], 613850
ITPR1	161.4	100	99.9	Gillespie syndrome, 206700 Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360
ITPR2	137.3	99	95.9	?Anhidrosis, isolated, with normal sweat glands, 106190
IVD	114.9	100	100	Isovaleric acidemia, 243500
IYD	117.3	99.7	97.8	Thyroid dyshormonogenesis 4, 274800
JAG1	148.4	98.1	97.5	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500
JAGN1	147.3	100	100	Neutropenia, severe congenital, 6, autosomal recessive, 616022
JAK2	90.6	95.9	94.1	Erythrocytosis, somatic, 133100 Leukemia, acute myeloid, somatic, 601626 Myelofibrosis, somatic, 254450 Polycythemia vera, somatic, 263300 Thrombocythemia 3, 614521 {Budd-Chiari syndrome, somatic}, 600880
JAK3	104.2	98.2	95.2	SCID, autosomal recessive, T-negative/B-positive type, 600802
JAM3	158.6	100	100	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730

JPH1	173.9	100	99.8	?Charcot-Marie-Tooth disease, axonal, autosomal dominant, type 2K, 607831
JPH2	87.5	90.4	75.1	Cardiomyopathy, hypertrophic, 17, 613873
JPH3	156.6	100	99.7	Huntington disease-like 2, 606438
JUP	145.1	100	99.6	Arrhythmogenic right ventricular dysplasia 12, 611528 Naxos disease, 601214
KANK1	166.2	100	100	Cerebral palsy, spastic quadriplegic, 2, 612900
KANK2	151.4	99.9	99.5	Nephrotic syndrome, type 16, 617783 Palmoplantar keratoderma and woolly hair, 616099
KANSL1	172.3	99.9	99.2	Koolen-De Vries syndrome, 610443
KARS	122.6	100	99.3	?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness, autosomal recessive 89, 613916
KAT6A	169.3	100	99.8	Mental retardation, autosomal dominant 32, 616268
KAT6B	192.3	99.6	98.5	Genitopatellar syndrome, 606170 SBBYSS syndrome, 603736
KATNB1	141.7	100	100	Lissencephaly 6, with microcephaly, 616212
KBTBD13	107.1	99.8	96.8	Nemaline myopathy 6, autosomal dominant, 609273
KCNA1	167	100	99.7	Episodic ataxia/myokymia syndrome, 160120
KCNA2	157.7	100	99.9	Epileptic encephalopathy, early infantile, 32, 616366
KCNA5	143.3	99.4	96	Atrial fibrillation, familial, 7, 612240
KCNB1	145	100	99.9	Epileptic encephalopathy, early infantile, 26, 616056
KCNC1	199.2	100	100	Epilepsy, progressive myoclonic 7, 616187
KCNC3	144	68.5	59	Spinocerebellar ataxia 13, 605259
KCND3	182.5	99.9	99.1	Brugada syndrome 9, 616399 Spinocerebellar ataxia 19, 607346
KCNE1	462.6	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	177.1	100	100	?Brugada syndrome 6, 613119
KCNH1	185.8	98.7	98.7	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500
KCNH2	102.6	92.3	84.8	Long QT syndrome 2, 613688 Short QT syndrome 1, 609620

				{Long QT syndrome 2, acquired, susceptibility to}, 613688
KCNJ1	233.9	100	100	Bartter syndrome, type 2, 241200
KCNJ10	213.4	89.3	89.1	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ11	299.5	100	100	Diabetes mellitus, transient neonatal, 3, 610582 Diabetes, permanent neonatal, with or without neurologic features, 606176 Hyperinsulinemic hypoglycemia, familial, 2, 601820 Maturity-onset diabetes of the young, type 13, 616329 {Diabetes mellitus, type 2, susceptibility to}, 125853
KCNJ13	210.4	100	99.9	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	193.9	100	99.8	Hyperaldosteronism, familial, type III, 613677 Long QT syndrome 13, 613485
KCNJ6	156.6	100	99.9	Keppen-Lubinsky syndrome, 614098
KCNK3	165.3	98.9	96.1	Pulmonary hypertension, primary, 4, 615344
KCNK9	193.7	100	100	Birk-Barel mental retardation dysmorphism syndrome, 612292
KCNMA1	120.4	94.4	93.2	?Cerebellar atrophy, developmental delay, and seizures, 617643 Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446
KCNN4	136.1	100	99.8	Dehydrated hereditary stomatocytosis 2, 616689
KCNQ1	114.7	93	90.3	Atrial fibrillation, familial, 3, 607554 Jervell and Lange-Nielsen syndrome, 220400 Long QT syndrome 1, 192500 Short QT syndrome 2, 609621 {Long QT syndrome 1, acquired, susceptibility to}, 192500
KCNQ1OT1	NC	NC	NC	Beckwith-Wiedemann syndrome, 130650
KCNQ2	85.4	90.1	86.5	Epileptic encephalopathy, early infantile, 7, 613720 Myokymia, 121200 Seizures, benign neonatal, 1, 121200
KCNQ3	110.7	98.9	95.5	Seizures, benign neonatal, 2, 121201
KCNQ4	135.2	93.7	89.1	Deafness, autosomal dominant 2A, 600101

KCNQ5	158	96.2	94.2	Mental retardation, autosomal dominant 46, 617601
KCNT1	112	95.3	92.3	Epilepsy, nocturnal frontal lobe, 5, 615005 Epileptic encephalopathy, early infantile, 14, 614959
KCNT2	105.1	97.6	89.9	?Epileptic encephalopathy, early infantile, 57, 617771
KCNV2	137.7	100	100	Retinal cone dystrophy 3B, 610356
KCTD1	140.5	100	100	Scalp-ear-nipple syndrome, 181270
KCTD17	93.7	95.3	88	Dystonia 26, myoclonic, 616398
KCTD7	166.7	95	95	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDF1	103.9	100	99.6	?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/nail type, 617337
KDM1A	129.7	96.2	93.4	Cleft palate, psychomotor retardation, and distinctive facial features, 616728
KDM5B	138	98.8	96.9	Mental retardation, autosomal recessive 65, 618109
KDM5C	112.6	97.9	95.1	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
KDM6A	109	93.2	84.3	Kabuki syndrome 2, 300867
KDR	137.9	100	99.4	Hemangioma, capillary infantile, somatic, 602089 {Hemangioma, capillary infantile, susceptibility to}, 602089
KDSR	173.7	100	100	Erythrokeratodermia variabilis et progressiva 4, 617526
KERA	191.6	100	100	Cornea plana 2, autosomal recessive, 217300
KHDC3L	110.3	99.9	99.2	Hydatidiform mole, recurrent, 2, 614293
KIAA0556	134.4	99.9	99.4	Joubert syndrome 26, 616784
KIAA0586	114.7	98.2	92.7	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546
KIAA0753	123.9	99.9	98.7	?Orofaciodigital syndrome XV, 617127
KIAA1109	144.1	99.1	97.2	Alkuraya-Kucinskas syndrome, 617822
KIDINS220	155.4	99.9	99.5	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296
KIF11	83.8	97.2	94.2	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF14	111.3	98.1	89.9	?Meckel syndrome 12, 616258 Microcephaly 20, primary, autosomal recessive, 617914
KIF1A	114	99.2	96.1	Mental retardation, autosomal dominant 9, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal recessive, 610357

KIF1B	154.8	100	99.5	?Charcot-Marie-Tooth disease, type 2A1, 118210 Pheochromocytoma, 171300 {Neuroblastoma, susceptibility to, 1}, 256700
KIF1BP	159.7	96.2	96.1	Goldberg-Shprintzen megacolon syndrome, 609460
KIF1C	121.3	99.9	99.1	Spastic ataxia 2, autosomal recessive, 611302
KIF21A	123.5	99.4	96.1	Fibrosis of extraocular muscles, congenital, 1, 135700 Fibrosis of extraocular muscles, congenital, 3B, 135700
KIF22	163.1	100	99.9	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546
KIF2A	105.2	97.7	88.7	Cortical dysplasia, complex, with other brain malformations 3, 615411
KIF4A	92.4	98.2	93.7	?Mental retardation, X-linked 100, 300923
KIF5A	136.1	100	99.9	Myoclonus, intractable, neonatal, 617235 Spastic paraplegia 10, autosomal dominant, 604187 {Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921
KIF5C	116.3	99.9	99.1	Cortical dysplasia, complex, with other brain malformations 2, 615282
KIF7	85.7	93.5	88.9	?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990
KISS1	41.4	98.5	91.2	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842
KISS1R	106.4	99.5	95.3	?Precocious puberty, central, 1, 176400 Hypogonadotropic hypogonadism 8 with or without anosmia, 614837
KIT	153	100	99.7	Gastrointestinal stromal tumor, familial, 606764 Germ cell tumors, somatic, 273300 Leukemia, acute myeloid, 601626 Mastocytosis, cutaneous, 154800 Mastocytosis, systemic, somatic, 154800 Piebaldism, 172800
KITLG	81.8	97	91.6	Deafness, autosomal dominant 69, unilateral or asymmetric, 616697 Hyperpigmentation with or without hypopigmentation, 145250 [Skin/hair/eye pigmentation 7, blond/brown hair], 611664

KIZ	174.2	98.3	96	Retinitis pigmentosa 69, 615780
KL	179.9	97.2	96	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994
KLC2	115.9	99.8	98.2	Spastic paraplegia, optic atrophy, and neuropathy, 609541
KLF1	52.1	90.8	81.7	Blood group--Lutheran inhibitor, 111150 Dyserythropoietic anemia, congenital, type IV, 613673 [Hereditary persistence of fetal hemoglobin], 613566
KLF11	181.3	99.9	99.4	Maturity-onset diabetes of the young, type VII, 610508
KLF6	154	100	100	Gastric cancer, somatic, 613659 Prostate cancer, somatic, 176807
KLHL10	209.7	100	100	Spermatogenic failure 11, 615081
KLHL15	178.1	100	99.9	Mental retardation, X-linked 103, 300982
KLHL24	192.9	100	100	Epidermolysis bullosa simplex, generalized, with scarring and hair loss, 617294
KLHL3	141.1	100	99.8	Pseudohypoaldosteronism, type IID, 614495
KLHL40	157.9	100	100	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	203.5	100	99.6	Nemaline myopathy 9, 615731
KLHL7	123.5	100	99.5	Cold-induced sweating syndrome 3, 617055 Retinitis pigmentosa 42, 612943
KLK4	185.1	100	98.8	Amelogenesis imperfecta, type IIA1, 204700
KLKB1	143.4	99.6	96.7	Fletcher factor (prekallikrein) deficiency, 612423
KLLN	117.1	100	100	Cowden syndrome 4, 615107
KMT2A	152.5	99.3	98.6	Leukemia, myeloid/lymphoid or mixed-lineage, 159555 Wiedemann-Steiner syndrome, 605130
KMT2B	120.3	94	91.2	Dystonia 28, childhood-onset, 617284
KMT2C	170.2	91	88.6	Kleefstra syndrome 2, 617768
KMT2D	142.1	99.9	99	Kabuki syndrome 1, 147920
KMT5B	202.3	100	99.6	Mental retardation, autosomal dominant 51, 617788
KNL1	113.7	98.3	95.2	Microcephaly 4, primary, autosomal recessive, 604321
KPTN	112.1	100	99.9	Mental retardation, autosomal recessive 41, 615637

KRAS	64.7	99.9	98.7	Arteriovenous malformation of the brain, somatic, 108010 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
KREMEN1	149.1	94.7	93.6	Ectodermal dysplasia 13, hair/tooth type, 617392
KRIT1	85.1	99.9	97.7	Cavernous malformations of CNS and retina, 116860 Cerebral cavernous malformations-1, 116860 Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations, 116860
KRT1	120.1	99.9	96.7	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	98.7	93.9	Epidermolytic hyperkeratosis, 113800 Ichthyosis with confetti, 609165 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602
KRT12	126.1	98.6	95.4	Meesmann corneal dystrophy, 122100
KRT13	140.4	99.9	98.9	White sponge nevus 2, 615785
KRT14	59.3	89	82.1	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	38.5	72.4	53.4	Pachyonychia congenita 1, 167200 Palmoplantar keratoderma, nonepidermolytic, focal, 613000

KRT17	21.5	47.2	31.9	Pachyonychia congenita 2, 167210 Steatocystoma multiplex, 184500
KRT18	40.2	84.8	67.8	Cirrhosis, cryptogenic, 215600 {Cirrhosis, noncryptogenic, susceptibility to}, 215600
KRT2	140.9	100	99.8	Ichthyosis bullosa of Siemens, 146800
KRT25	142.9	100	100	Woolly hair, autosomal recessive 3, 616760
KRT3	105.1	100	99.7	Meesmann corneal dystrophy, 122100
KRT4	130.7	100	99.9	White sponge nevus 1, 193900
KRT5	133.5	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-MCR, 609352 Epidermolysis bullosa simplex-MP, 131960
KRT6A	197.8	94.8	87.9	Pachyonychia congenita 3, 615726
KRT6B	194.4	95.9	89.3	Pachyonychia congenita 4, 615728
KRT6C	174.5	87.3	79.7	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735
KRT71	139.8	100	100	?Hypotrichosis 13, 615896
KRT74	150.2	99.9	98.9	?Ectodermal dysplasia 7, hair/nail type, 614929 ?Hypotrichosis 3, 613981 Woolly hair, autosomal dominant, 194300
KRT8	39.2	91.7	73.4	Cirrhosis, cryptogenic, 215600 {Cirrhosis, noncryptogenic, susceptibility to}, 215600
KRT81	93.4	99.7	96	Monilethrix, 158000
KRT83	81.3	98.4	89.5	Erythrokeratodermia variabilis et progressiva 5, 617756 Monilethrix, 158000
KRT85	108.3	98.8	95.3	Ectodermal dysplasia 4, hair/nail type, 602032
KRT86	101.4	100	98	Monilethrix, 158000
KRT9	84.8	98.2	96	Palmoplantar keratoderma, epidermolytic, 144200
KY	135.5	100	100	Myopathy, myofibrillar, 7, 617114
KYNU	116.9	98.5	92.2	?Hydroxykynureninuria, 236800 Vertebral, cardiac, renal, and limb defects syndrome 2, 617661

L1CAM	133.3	99.8	98.4	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	98.4	97	L-2-hydroxyglutaric aciduria, 236792
LAGE3	50.3	95	81.2	Galloway-Mowat syndrome 2, X-linked, 301006
LAMA1	137.5	100	99.6	Poretti-Boltshauser syndrome, 615960
LAMA2	143.5	99.9	99.5	Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855 Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138
LAMA3	147.6	99.7	99.2	Epidermolysis bullosa, generalized atrophic benign, 226650 Epidermolysis bullosa, junctional, Herlitz type, 226700 Laryngoonychocutaneous syndrome, 245660
LAMA4	132.6	100	99.9	Cardiomyopathy, dilated, 1JJ, 615235
LAMB1	169.8	100	99.6	Lissencephaly 5, 615191
LAMB2	201.7	100	99.8	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 Pierson syndrome, 609049
LAMB3	123.4	100	99.6	Amelogenesis imperfecta, type IA, 104530 Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LAMC2	117.6	99.9	98.8	Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LAMC3	130.5	98.3	96.1	Cortical malformations, occipital, 614115
LAMP2	106.1	92.7	91.2	Danon disease, 300257
LAMTOR2	167	100	99.9	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LARGE1	143	100	99.6	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	56.3	80.5	63.7	Alazami syndrome, 615071
LARS	133.5	99.5	97.1	?Infantile liver failure syndrome 1, 615438

LARS2	143	100	100	?Hydrops, lactic acidosis, and sideroblastic anemia, 617021 Perrault syndrome 4, 615300
LAS1L	90.2	99.7	97.7	Wilson-Turner syndrome, 309585
LAT	94.6	99.8	98.4	Immunodeficiency 52, 617514
LBR	87.8	93.3	83.9	?Reynolds syndrome, 613471 Greenberg skeletal dysplasia, 215140 Pelger-Huet anomaly, 169400 Pelger-Huet anomaly with mild skeletal anomalies, 618019
LCA5	127.8	97.3	95.7	Leber congenital amaurosis 5, 604537
LCAT	145.6	97.8	91.7	Fish-eye disease, 136120 Norum disease, 245900
LCK	161.4	98.2	95.5	?Immunodeficiency 22, 615758
LCT	142	99.9	99.1	Lactase deficiency, congenital, 223000
LDB3	127.3	95.5	93.7	Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 Cardiomyopathy, hypertrophic, 24, 601493 Left ventricular noncompaction 3, 601493 Myopathy, myofibrillar, 4, 609452
LDHA	59.8	94.1	87.1	Glycogen storage disease XI, 612933
LDLR	171	99.6	97.9	Hypercholesterolemia, familial, 143890 LDL cholesterol level QTL2, 143890
LDLRAP1	156.1	95.7	91.2	Hypercholesterolemia, familial, autosomal recessive, 603813
LEF1	115.9	100	99.9	Sebaceous tumors, somatic, 0
LEMD2	68.6	89.1	80.6	Cataract 46, juvenile-onset, 212500
LEMD3	96.7	95.4	88.8	Buschke-Ollendorff syndrome, 166700 Osteopoikilosis with or without melorheostosis, 166700
LEP	188.8	100	99.6	Obesity, morbid, due to leptin deficiency, 614962
LEPR	109.6	93.8	90.2	Obesity, morbid, due to leptin receptor deficiency, 614963
LFNG	91.6	85.4	83.3	?Spondylocostal dysostosis 3, autosomal recessive, 609813
LGI1	163.8	97.8	94.8	Epilepsy, familial temporal lobe, 1, 600512
LGI4	73.8	99	95.8	Arthrogryposis multiplex congenita, neurogenic, with myelin defect, 617468
LHB	29	97.2	73.2	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300

LHCGR	154.5	95.4	92.8	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	287.7	100	100	Deafness, autosomal recessive 67, 610265
LHX3	84.7	94	80.6	Pituitary hormone deficiency, combined, 3, 221750
LHX4	144.9	100	99.8	Pituitary hormone deficiency, combined, 4, 262700
LIAS	133.7	99.5	97.1	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIFR	123.4	97.2	92.1	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LIG4	165.6	100	99.6	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500
LIM2	103.2	100	98.6	Cataract 19, multiple types, 615277
LIMS2	110.8	93	92.3	?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827
LINGO1	230.8	100	99.8	Mental retardation, autosomal recessive 64, 618103
LINS1	147.8	99.9	98	Mental retardation, autosomal recessive 27, 614340
LIPA	110.9	98.8	95.8	Cholesteryl ester storage disease, 278000 Wolman disease, 278000
LIPC	115.2	100	99.8	Hepatic lipase deficiency, 614025 [High density lipoprotein cholesterol level QTL 12], 612797 {Diabetes mellitus, noninsulin-dependent}, 125853
LIPE	113.4	99.9	98.4	Lipodystrophy, familial partial, type 6, 615980
LIPH	140.6	100	100	Hypotrichosis 7, 604379 Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379
LIPN	125.2	99.2	95.8	Ichthyosis, congenital, autosomal recessive 8, 613943
LIPT1	227.4	100	100	Lipoyltransferase 1 deficiency, 616299
LIPT2	92.2	97.3	83.2	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668
LITAF	126.3	94.8	91.5	Charcot-Marie-Tooth disease, type 1C, 601098
LMAN1	129.8	99.3	94.4	Combined factor V and VIII deficiency, 227300
LMAN2L	127.9	100	99.8	?Mental retardation, autosomal recessive, 52, 616887

LMBR1	94	95.9	88.1	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	80.2	91.9	83	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LMF1	132.4	99.6	97.8	Lipase deficiency, combined, 246650
LMNA	89.2	97.9	91.3	Cardiomyopathy, dilated, 1A, 115200 Charcot-Marie-Tooth disease, type 2B1, 605588 Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 Heart-hand syndrome, Slovenian type, 610140 Hutchinson-Gilford progeria, 176670 Lipodystrophy, familial partial, type 2, 151660 Malouf syndrome, 212112 Mandibuloacral dysplasia, 248370 Muscular dystrophy, congenital, 613205 Restrictive dermopathy, lethal, 275210
LMNB1	123.3	99.9	99.1	Leukodystrophy, adult-onset, autosomal dominant, 169500
LMNB2	121.2	97.1	93.2	?Epilepsy, progressive myoclonic, 9, 616540 {Lipodystrophy, partial, acquired, susceptibility to}, 608709
LMOD3	141.5	99.9	98.5	Nemaline myopathy 10, 616165
LMX1B	111.4	97	92.3	Nail-patella syndrome, 161200
LNPK	84.7	92	84.1	Neurodevelopmental disorder with epilepsy and hypoplasia of the corpus callosum, 618090
LONP1	141.5	97.9	96.4	CODAS syndrome, 600373
LOR	13.5	62.6	33.1	Vohwinkel syndrome with ichthyosis, 604117
LOX	104.4	99.8	97.6	Aortic aneurysm, familial thoracic 10, 617168
LOXHD1	136.5	100	99.6	Deafness, autosomal recessive 77, 613079
LPAR6	104.2	99.7	98.2	Hypotrichosis 8, 278150 Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150
LPIN1	134.6	99.8	97.8	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	111.5	100	99.6	Majeed syndrome, 609628

LPL	147.2	100	100	Combined hyperlipidemia, familial, 144250 Lipoprotein lipase deficiency, 238600 [High density lipoprotein cholesterol level QTL 11], 0
LPP	132.1	100	100	Leukemia, acute myeloid, 601626 Lipoma, 0
LRAT	298.3	100	100	Leber congenital amaurosis 14, 613341 Retinal dystrophy, early-onset severe, 613341 Retinitis pigmentosa, juvenile, 613341
LRBA	134.4	99.3	97.8	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRIG2	148.5	99.4	97.5	Urofacial syndrome 2, 615112
LRIT3	142.4	94.4	94.1	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRMDA	142.1	97.2	95.6	Albinism, oculocutaneous, type VII, 615179
LRP1	196	99.7	99.1	?Keratosis pilaris atrophicans, 604093
LRP2	176.3	100	99.8	Donnai-Barrow syndrome, 222448
LRP4	166.6	99.1	98.9	?Myasthenic syndrome, congenital, 17, 616304 Cenani-Lenz syndactyly syndrome, 212780 Sclerosteosis 2, 614305
LRP5	189.8	98.2	97.9	Exudative vitreoretinopathy 4, 601813 Hyperostosis, endosteal, 144750 Osteopetrosis, autosomal dominant 1, 607634 Osteoporosis-pseudoglioma syndrome, 259770 Osteosclerosis, 144750 Polycystic liver disease 4 with or without kidney cysts, 617875 van Buchem disease, type 2, 607636 [Bone mineral density variability 1], 601884 {Osteoporosis}, 166710
LRP6	169.3	100	99.7	Tooth agenesis, selective, 7, 616724 {Coronary artery disease, autosomal dominant, 2}, 610947
LRPAP1	138.3	99.5	97.2	Myopia 23, autosomal recessive, 615431
LRPPRC	127.3	99.4	97.2	Leigh syndrome, French-Canadian type, 220111
LRRC56	106.6	99	95.8	Ciliary dyskinesia, primary, 39, 618254
LRRC6	137.9	94.7	91	Ciliary dyskinesia, primary, 19, 614935
LRRC8A	273	100	99.9	?Agammaglobulinemia 5, 613506

LRSAM1	130.4	100	99.7	Charcot-Marie-Tooth disease, axonal, type 2P, 614436
LRTOMT	125.9	99.8	96.6	Deafness, autosomal recessive 63, 611451
LSS	127.8	100	99.1	Cataract 44, 616509
LTBP2	104.6	99.6	97.1	?Weill-Marchesani syndrome 3, recessive, 614819 Glaucoma 3, primary congenital, D, 613086 Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750
LTBP3	113.5	98.7	94.7	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
LTBP4	117.1	98.6	95	Cutis laxa, autosomal recessive, type IC, 613177
LYRM4	60.1	63.2	54.3	?Combined oxidative phosphorylation deficiency 19, 615595
LYRM7	49	87.6	72.4	Mitochondrial complex III deficiency, nuclear type 8, 615838
LYST	134.6	97.8	93.9	Chediak-Higashi syndrome, 214500
LYZ	165.3	100	100	Amyloidosis, renal, 105200
LZTFL1	109.1	99.1	95.3	Bardet-Biedl syndrome 17, 615994
LZTR1	134	100	99.4	Noonan syndrome 10, 616564 Noonan syndrome 2, 605275 {Schwannomatosis-2, susceptibility to}, 615670
LZTS1	105.5	100	100	Esophageal squamous cell carcinoma, 133239
MAB21L2	245.6	100	100	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877
MAD1L1	97.8	99.8	97.1	Lymphoma, somatic, 0 Prostate cancer, somatic, 176807
MAD2L2	125.9	100	100	?Fanconi anemia, complementation group V, 617243
MAF	60.2	77.5	72.7	Ayme-Gripp syndrome, 601088 Cataract 21, multiple types, 610202
MAFA	28.6	79	59.6	Insulinomatosis and diabetes mellitus, 147630
MAFB	93.8	99.7	97.8	Duane retraction syndrome 3, 617041 Multicentric carpotarsal osteolysis syndrome, 166300
MAG	146.5	100	99.5	Spastic paraplegia 75, autosomal recessive, 616680
MAGED2	80	99.7	97.4	Bartter syndrome, type 5, antenatal, transient, 300971
MAGEL2	83.4	90.6	81.2	Schaaf-Yang syndrome, 615547
MAGI2	98	91.8	88.3	Nephrotic syndrome, type 15, 617609
MAGT1	101.8	98.4	95.8	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia,

				300853
MAK	139.1	95.6	94.1	Retinitis pigmentosa 62, 614181
MALT1	136.6	89.1	85.4	Immunodeficiency 12, 615468
MAML2	122.9	100	100	Mucoepidermoid salivary gland carcinoma, 0
MAMLD1	131.4	99.9	98.3	Hypospadias 2, X-linked, 300758
MAN1B1	128.9	100	99.7	Mental retardation, autosomal recessive 15, 614202
MAN2B1	122.3	99.1	96.2	Mannosidosis, alpha-, types I and II, 248500
MANBA	119.9	99.7	97.2	Mannosidosis, beta, 248510
MAOA	113.8	99.9	98.7	Brunner syndrome, 300615 {Antisocial behavior}, 300615
MAP2K1	92.3	99.8	95.6	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	107.9	97.6	89.2	Cardiofaciocutaneous syndrome 4, 615280
MAP3K1	166	92.9	89.2	46XY sex reversal 6, 613762
MAP3K20	125.3	99.8	98	Centronuclear myopathy 6 with fiber-type disproportion, 617760 Split-foot malformation with mesoaxial polydactyly, 616890
MAP3K7	114.6	99.7	98.1	Cardiospondylocarpofacial syndrome, 157800 Frontometaphyseal dysplasia 2, 617137
MAP3K8	143.2	100	99.8	Lung cancer, somatic, 211980
MAPKAPK3	92.8	98.9	96.3	?Macular dystrophy, patterned, 3, 617111
MAPKBP1	144.1	100	100	Nephronophthisis 20, 617271
MAPRE2	205.7	100	99.5	Symmetric circumferential skin creases, congenital, 2, 616734
MAPT	127.2	100	98.6	Dementia, frontotemporal, with or without parkinsonism, 600274 Pick disease, 172700 Supranuclear palsy, progressive, 601104 Supranuclear palsy, progressive atypical, 260540 {Parkinson disease, susceptibility to}, 168600
MARS	125.2	99.7	97.3	Charcot-Marie-Tooth disease, axonal, type 2U, 616280 Interstitial lung and liver disease, 615486
MARS2	173.2	100	100	?Combined oxidative phosphorylation deficiency 25, 616430 Spastic ataxia 3, autosomal recessive, 611390
MARVELD2	159.4	97.5	94	Deafness, autosomal recessive 49, 610153
MASP1	148.6	100	99.6	3MC syndrome 1, 257920
MASP2	139.8	100	99.3	MASP2 deficiency, 613791

MAT1A	185.4	99.7	97.5	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MATN3	116.6	84.7	84.7	?Spondyloepimetaphyseal dysplasia, 608728 Epiphyseal dysplasia, multiple, 5, 607078 {Osteoarthritis susceptibility 2}, 140600
MATR3	92.5	95.7	88.8	Amyotrophic lateral sclerosis 21, 606070
MBD5	196.2	99.9	99.6	Mental retardation, autosomal dominant 1, 156200
MBOAT7	91.8	99.3	94.7	Mental retardation, autosomal recessive 57, 617188
MBTPS2	113.4	99.6	97.6	?Olmsted syndrome, X-linked, 300918 IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800 Osteogenesis imperfecta, type XIX, 301014
MC2R	213.1	100	100	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MC4R	272	100	100	Obesity, autosomal dominant, 601665
MCC	139	100	99.6	Colorectal cancer, somatic, 114500
MCCC1	151.7	100	99.4	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	131.1	99.9	98.9	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	99.8	100	100	Methylmalonyl-CoA epimerase deficiency, 251120
MCFD2	101.7	99.9	98.8	Factor V and factor VIII, combined deficiency of, 613625
MCM2	176.8	100	100	?Deafness, autosomal dominant 70, 616968
MCM3AP	137.9	100	99.3	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124
MCM4	164.3	99.9	98.8	Immunodeficiency 54, 609981
MCM5	131.5	100	100	?Meier-Gorlin syndrome 8, 617564
MCM6	154.1	100	99.7	Lactase persistence/nonpersistence, 223100
MCM8	123.6	99.9	98.5	?Premature ovarian failure 10, 612885
MCM9	151.6	100	99.9	Ovarian dysgenesis 4, 616185
MCOLN1	150.2	98.8	97	Mucolipidosis IV, 252650
MCPH1	148.6	99.9	98.1	Microcephaly 1, primary, autosomal recessive, 251200
MDH2	123.3	98	97.9	Epileptic encephalopathy, early infantile, 51, 617339
MECOM	143.4	100	99.6	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738

MECP2	87.3	99.1	93.1	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
MECR	108.1	98.8	96.1	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MED12	105.7	98	94.8	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Opitz-Kaveggia syndrome, 305450
MED13L	134.6	100	99.6	Mental retardation and distinctive facial features with or without cardiac defects, 616789 Transposition of the great arteries, dextro-looped 1, 608808
MED17	118	95.2	91.7	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	131.2	98.7	96.9	Mental retardation, autosomal recessive 18, 614249
MED25	103.9	99.1	95.7	?Charcot-Marie-Tooth disease, type 2B2, 605589 Basel-Vanagait-Smirin-Yosef syndrome, 616449
MEF2C	137.7	97.9	93.5	Chromosome 5q14.3 deletion syndrome, 613443 Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
MEFV	108.8	94.9	91	Familial Mediterranean fever, AD, 134610 Familial Mediterranean fever, AR, 249100
MEGF10	154.3	100	99.8	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399
MEGF8	127.6	99.9	98.6	Carpenter syndrome 2, 614976
MEIOB	109.3	98.8	92.5	?Spermatogenic failure 22, 617706
MEIS2	138.2	100	99.9	Cleft palate, cardiac defects, and mental retardation, 600987
MEN1	123.2	99.5	96.3	Adrenal adenoma, somatic, 0 Angiofibroma, somatic, 0 Carcinoid tumor of lung, 0 Lipoma, somatic, 0 Multiple endocrine neoplasia 1, 131100

				Parathyroid adenoma, somatic, 0
MEOX1	76.8	96.6	91.2	Klippel-Feil syndrome 2, 214300
MERTK	161.4	99.4	97.7	Retinitis pigmentosa 38, 613862
MESP2	81.5	93.1	87.9	Spondylocostal dysostosis 2, autosomal recessive, 608681
MET	184.7	100	99.6	?Deafness, autosomal recessive 97, 616705 Hepatocellular carcinoma, childhood type, somatic, 114550 Renal cell carcinoma, papillary, 1, familial and somatic, 605074 {Osteofibrous dysplasia, susceptibility to}, 607278
METTL23	145	100	100	Mental retardation, autosomal recessive 44, 615942
MFAP5	126.8	100	99.5	Aortic aneurysm, familial thoracic 9, 616166
MFF	93.7	90.4	87.6	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFN2	150.6	100	99.9	Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260 Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 Hereditary motor and sensory neuropathy VIA, 601152
MFRP	121.3	100	100	Microphthalmia, isolated 5, 611040 Nanophthalmos 2, 609549
MFSD2A	115.1	98.8	95.9	Microcephaly 15, primary, autosomal recessive, 616486
MFSD8	125.1	99.9	98.4	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170
MGAT2	157.2	100	99.9	Congenital disorder of glycosylation, type IIa, 212066
MGME1	151.1	100	100	Mitochondrial DNA depletion syndrome 11, 615084
MGP	132	92.7	91.6	Keutel syndrome, 245150
MIB1	141.7	100	99.6	Left ventricular noncompaction 7, 615092
MICU1	134.2	96	88.8	Myopathy with extrapyramidal signs, 615673
MID1	164.6	99.8	98.4	Opitz GBBB syndrome, type I, 300000
MID2	141	99.6	97.3	?Mental retardation, X-linked 101, 300928
MINPP1	147.8	98.8	94.8	Thyroid carcinoma, follicular, 188470
MIP	132.2	99.8	96.1	Cataract 15, multiple types, 615274
MIPEP	102.1	95.3	88.6	Combined oxidative phosphorylation deficiency 31, 617228

MIR17HG	NC	NC	NC	Feingold syndrome 2, 614326
MIR184	NC	NC	NC	EDICT syndrome, 614303
MIR204	NC	NC	NC	?Retinal dystrophy and iris coloboma with or without cataract, 616722
MIR96	NC	NC	NC	Deafness, autosomal dominant 50, 613074
MITF	155.5	100	99.9	COMMAD syndrome, 617306 Tietz albinism-deafness syndrome, 103500 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456
MKKS	208.5	83.2	83.1	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKL1	101	96.2	91.1	Megakaryoblastic leukemia, acute, 0
MKRN3	162	100	99.9	Precocious puberty, central, 2, 615346
MKS1	114.5	99.9	98.5	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000
MLC1	103.4	100	99.8	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MLH1	162	100	99.7	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MLH3	151.7	100	100	Colorectal cancer, hereditary nonpolyposis, type 7, 614385 Colorectal cancer, somatic, 114500 {Endometrial cancer, susceptibility to}, 608089
MLLT10	142	94.8	91.4	Leukemia, acute myeloid, 601626
MLPH	99.1	99.6	95.9	Griscelli syndrome, type 3, 609227
MLYCD	75.6	91.3	86.9	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	183.2	100	99.6	Methylmalonic aciduria, vitamin B12-responsive, 251100
MMAB	101.2	100	99.9	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110
MMACHC	205.8	100	100	Methylmalonic aciduria and homocystinuria, cblC type, 277400

MMADHC	77	89.3	75	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410
MME	101.1	98.2	93.6	?Spinocerebellar ataxia 43, 617018 Charcot-Marie-Tooth disease, axonal, type 2T, 617017
MMP1	163.6	100	98.2	COPD, rate of decline of lung function in, 606963 {Epidermolysis bullosa dystrophica, autosomal recessive, modifier of}, 226600
MMP13	124.1	93.5	91.5	Metaphyseal anadysplasia 1, 602111 Metaphyseal dysplasia, Spahr type, 250400 Spondyloepimetaphyseal dysplasia, Missouri type, 602111
MMP14	152.1	100	99.5	?Winchester syndrome, 277950
MMP19	129.1	99.6	97.7	Cavitary optic disc anomalies, 611543
MMP2	164.4	100	100	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP20	100.5	100	98.6	Amelogenesis imperfecta, type IIA2, 612529
MMP21	93.3	90.2	84.6	Heterotaxy, visceral, 7, autosomal, 616749
MMP9	125.4	96.2	91.8	Metaphyseal anadysplasia 2, 613073
MN1	93.6	98.6	93	Meningioma, 607174
MNX1	29.3	63	51.9	Currarino syndrome, 176450
MOCOS	169	99.1	96.6	Xanthinuria, type II, 603592
MOCS1	87.4	98.4	93	Molybdenum cofactor deficiency A, 252150
MOCS2	139.5	99.6	99.6	Molybdenum cofactor deficiency B, 252160
MOG	105.3	100	99.9	?Narcolepsy 7, 614250
MOGS	121.6	99.8	99.1	Congenital disorder of glycosylation, type IIb, 606056
MORC2	135.9	100	99.7	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688
MPC1	121.8	100	99.5	Mitochondrial pyruvate carrier deficiency, 614741
MPDU1	111.8	100	99.7	Congenital disorder of glycosylation, type If, 609180
MPDZ	149	98.7	96.6	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219
MPI	146.2	100	100	Congenital disorder of glycosylation, type Ib, 602579
MPIG6B	85.4	100	99.3	?Thrombocytopenia, anemia, and myelofibrosis, 617441
MPL	136.7	99.6	97.5	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498
MPLKIP	72.5	97.1	79.3	Trichothiodystrophy 4, nonphotosensitive, 234050

MPO	155	100	99.9	Myeloperoxidase deficiency, 254600 {Alzheimer disease, susceptibility to}, 104300 {Lung cancer, protection against, in smokers}, 0
MPV17	108.5	100	99.4	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MPZ	123.4	100	99.3	Charcot-Marie-Tooth disease, dominant intermediate D, 607791 Charcot-Marie-Tooth disease, type 1B, 118200 Charcot-Marie-Tooth disease, type 2I, 607677 Charcot-Marie-Tooth disease, type 2J, 607736 Dejerine-Sottas disease, 145900 Hypomyelinating neuropathy, congenital, 2, 618184 Roussy-Levy syndrome, 180800
MPZL2	96.9	100	100	Deafness, autosomal recessive 111, 618145
MRAP	161.3	100	100	Glucocorticoid deficiency 2, 607398
MRE11	51.2	95.3	82.3	Ataxia-telangiectasia-like disorder 1, 604391
MRPL3	66.3	91.2	77.9	Combined oxidative phosphorylation deficiency 9, 614582
MRPL44	110.5	99.7	97.6	?Combined oxidative phosphorylation deficiency 16, 615395
MRPS16	161.1	100	99.1	Combined oxidative phosphorylation deficiency 2, 610498
MRPS2	166.4	99.7	97.9	Combined oxidative phosphorylation deficiency 36, 617950
MRPS22	138.8	95.3	91.8	Combined oxidative phosphorylation deficiency 5, 611719 Ovarian dysgenesis 7, 618117
MRPS34	132.7	99.9	98.3	Combined oxidative phosphorylation deficiency 32, 617664
MRPS7	173.5	100	100	?Combined oxidative phosphorylation deficiency 34, 617872
MS4A1	123.7	99.4	96.2	Immunodeficiency, common variable, 5, 613495
MSH2	113.4	98.6	93.1	Colorectal cancer, hereditary nonpolyposis, type 1, 120435 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MSH3	113.4	99	95	Endometrial carcinoma, somatic, 608089 Familial adenomatous polyposis 4, 617100
MSH5	116.5	99.5	96.6	?Premature ovarian failure 13, 617442
MSH6	171.1	100	99.5	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 Endometrial cancer, familial, 608089

				Mismatch repair cancer syndrome, 276300
MSMO1	45.8	92.6	78.5	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MSN	88.6	99	95	Immunodeficiency 50, 300988
MSR1	170.8	100	99.4	Barrett esophagus/esophageal adenocarcinoma, 614266
MSRB3	155.6	99.7	98.5	Deafness, autosomal recessive 74, 613718
MSTN	161.5	100	99.5	Muscle hypertrophy, 614160
MSTO1	140	99.8	97	Myopathy, mitochondrial, and ataxia, 617675
MSX1	75.2	95.4	87.5	Ectodermal dysplasia 3, Witkop type, 189500 Orofacial cleft 5, 608874 Tooth agenesis, selective, 1, with or without orofacial cleft, 106600
MSX2	94.2	98	85.5	Craniosynostosis 2, 604757 Parietal foramina 1, 168500 Parietal foramina with cleidocranial dysplasia, 168550
MTAP	109.2	93.6	86.5	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250
MTFMT	124.6	99.3	96.2	Combined oxidative phosphorylation deficiency 15, 614947 Mitochondrial complex I deficiency, nuclear type 27, 618248
MTHFD1	139.6	99.8	98.4	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTHFR	126.1	98.4	97.2	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}, 0
MTM1	93.7	99.2	93	Myotubular myopathy, X-linked, 310400
MTMR2	106.6	100	99.2	Charcot-Marie-Tooth disease, type 4B1, 601382
MTO1	173.7	89.5	87.3	Combined oxidative phosphorylation deficiency 10, 614702
MTOR	140	100	99.8	Focal cortical dysplasia, type II, somatic, 607341 Smith-Kingsmore syndrome, 616638
MTPAP	109.6	98.9	93.5	?Spastic ataxia 4, autosomal recessive, 613672

MTR	140.9	99.8	98.8	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTRR	139.1	100	99.2	Homocystinuria-megaloblastic anemia, cbl E type, 236270 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTTP	132.4	99.9	98.8	Abetalipoproteinemia, 200100 {Metabolic syndrome, protection against}, 605552
MUC1	55.6	77.1	66.4	Medullary cystic kidney disease 1, 174000
MUSK	159.4	100	99.9	Fetal akinesia deformation sequence, 208150 Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325
MUT	121.8	99.2	95.1	Methylmalonic aciduria, mut(0) type, 251000
MUTYH	165	100	99.9	Adenomas, multiple colorectal, 608456 Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600 Gastric cancer, somatic, 613659
MVD	101.2	100	99	Porokeratosis 7, multiple types, 614714
MVK	124.3	92.1	90.4	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900
MXI1	108.3	90.6	83.6	Neurofibrosarcoma, somatic, 0 {Prostate cancer, susceptibility to, somatic}, 176807
MYBPC1	150.7	99.9	99.4	Arthrogryposis, distal, type 1B, 614335 Lethal congenital contracture syndrome 4, 614915
MYBPC3	142.5	98.5	95.7	Cardiomyopathy, dilated, 1MM, 615396 Cardiomyopathy, hypertrophic, 4, 115197 Left ventricular noncompaction 10, 615396
MYC	114.3	64.8	63.1	Burkitt lymphoma, 113970
MYCN	94.1	94.9	84.8	Feingold syndrome 1, 164280
MYD88	186.5	100	99.9	Macroglobulinemia, Waldenstrom, somatic, 153600 Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260
MYF5	180.4	100	100	Ophthalmoplegia, external, with rib and vertebral anomalies, 618155
MYF6	121.6	100	100	Centronuclear myopathy 3, 614408
MYH11	132.6	100	99.3	Aortic aneurysm, familial thoracic 4, 132900

MYH14	102	97.7	91.5	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369 Deafness, autosomal dominant 4A, 600652
MYH2	129.4	99.9	98.8	Proximal myopathy and ophthalmoplegia, 605637
MYH3	110.4	99.9	98.6	Arthrogryposis, distal, type 2A, 193700 Arthrogryposis, distal, type 2B, 601680 Arthrogryposis, distal, type 8, 178110
MYH6	113.3	99	96.1	Atrial septal defect 3, 614089 Cardiomyopathy, dilated, 1EE, 613252 Cardiomyopathy, hypertrophic, 14, 613251 {Sick sinus syndrome 3}, 614090
MYH7	111.4	99.4	96.8	Cardiomyopathy, dilated, 1S, 613426 Cardiomyopathy, hypertrophic, 1, 192600 Laing distal myopathy, 160500 Left ventricular noncompaction 5, 613426 Myopathy, myosin storage, autosomal dominant, 608358 Myopathy, myosin storage, autosomal recessive, 255160 Scapuloperoneal syndrome, myopathic type, 181430
MYH8	134.9	100	99.4	Carney complex variant, 608837 Trismus-pseudocamptodactyly syndrome, 158300
MYH9	130.5	99.4	98.1	Deafness, autosomal dominant 17, 603622 Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100
MYL2	134.6	98.7	90.1	Cardiomyopathy, hypertrophic, 10, 608758
MYL3	103.1	100	100	Cardiomyopathy, hypertrophic, 8, 608751
MYL4	158.5	100	100	?Atrial fibrillation, familial, 18, 617280
MYLK	148.4	99.9	99.3	Aortic aneurysm, familial thoracic 7, 613780
MYLK2	120	100	100	Cardiomyopathy, hypertrophic, 1, digenic, 192600
MYMK	141	100	100	Carey-Fineman-Ziter syndrome, 254940
MYO15A	116.5	97.3	94	Deafness, autosomal recessive 3, 600316
MYO18B	132.4	100	99.1	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549
MYO1E	131.9	98.6	97	Glomerulosclerosis, focal segmental, 6, 614131
MYO3A	119.8	98.5	93	Deafness, autosomal recessive 30, 607101
MYO5A	125.3	99.5	97.4	Griscelli syndrome, type 1, 214450
MYO5B	137.6	98.2	95.5	Microvillus inclusion disease, 251850

MYO6	89.7	98.1	92.3	Deafness, autosomal dominant 22, 606346 Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346 Deafness, autosomal recessive 37, 607821
MYO7A	134.1	99.7	98.1	Deafness, autosomal dominant 11, 601317 Deafness, autosomal recessive 2, 600060 Usher syndrome, type 1B, 276900
MYO9A	142.5	99.5	97.8	Myasthenic syndrome, congenital, 24, presynaptic, 618198
MYOC	172.5	100	99.3	Glaucoma 1A, primary open angle, 137750
MYOT	139.4	99.3	95.5	Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
MYOZ2	145.9	100	100	Cardiomyopathy, hypertrophic, 16, 613838
MYPN	142.4	99.3	98.4	Cardiomyopathy, dilated, 1KK, 615248 Cardiomyopathy, familial restrictive, 4, 615248 Cardiomyopathy, hypertrophic, 22, 615248 Nemaline myopathy 11, autosomal recessive, 617336
MYRF	116.1	96.9	95.9	Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113
MYSM1	96	97.6	92.5	Bone marrow failure syndrome 4, 618116
MYT1L	178.3	100	99.7	Mental retardation, autosomal dominant 39, 616521
NAA10	102.4	98.7	96.7	?Microphthalmia, syndromic 1, 309800 Ogden syndrome, 300855
NAA15	86.6	96.5	90.9	Mental retardation, autosomal dominant 50, 617787
NACC1	167.7	100	99.9	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393
NADK2	143	96.2	89.3	?2,4-dienoyl-CoA reductase deficiency, 616034
NAGA	139.4	100	100	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241
NAGLU	108.7	92.4	90.4	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NAGS	81	98	91.6	N-acetylglutamate synthase deficiency, 237310
NALCN	139.5	99.8	97.5	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	87.1	64.7	Spermatogenic failure 12, 615413

NANS	106.1	100	99.9	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NARS2	120	97.4	97.1	Combined oxidative phosphorylation deficiency 24, 616239
NAT8L	70.1	89.3	79.9	?N-acetylaspartate deficiency, 614063
NAXE	81.1	99.7	95.9	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NBAS	145.3	99.5	97.6	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NBEAL2	172.7	99.5	99.3	Gray platelet syndrome, 139090
NBN	80.6	99.1	94.6	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260
NCAPD2	138.5	100	99.1	?Microcephaly 21, primary, autosomal recessive, 617983
NCAPD3	120.7	99.4	98	Microcephaly 22, primary, autosomal recessive, 617984
NCAPH	141.2	100	100	?Microcephaly 23, primary, autosomal recessive, 617985
NCF1	23.9	25.8	22.1	Chronic granulomatous disease due to deficiency of NCF-1, 233700
NCF2	124.3	100	99.4	Chronic granulomatous disease due to deficiency of NCF-2, 233710
NCF4	158.7	100	100	?Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III, 613960
NCSTN	111.9	100	99.8	Acne inversa, familial, 1, 142690
NDE1	100.9	100	99.5	?Microhydranencephaly, 605013 Lissencephaly 4 (with microcephaly), 614019
NDN	74.2	92.7	79.7	Prader-Willi syndrome, 176270
NDP	116.8	100	100	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600
NDRG1	128.4	99.9	98.8	Charcot-Marie-Tooth disease, type 4D, 601455
NDST1	201.4	100	100	Mental retardation, autosomal recessive 46, 616116
NDUFA1	166.8	100	99.6	Mitochondrial complex I deficiency, nuclear type 12, 301020
NDUFA10	136.7	98.9	96.8	Mitochondrial complex I deficiency, nuclear type 22, 618243
NDUFA11	86.9	99.5	95.8	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFA12	160.2	100	100	Mitochondrial complex I deficiency, nuclear type 23, 618244
NDUFA13	91.4	92.3	91.6	?Mitochondrial complex I deficiency, nuclear type 28, 618249 {Thyroid carcinoma, Hurthle cell}, 607464

NDUFA2	133.9	100	100	Mitochondrial complex I deficiency, nuclear type 13, 618235
NDUFA6	212.4	100	100	Mitochondrial complex I deficiency, nuclear type 33, 618253
NDUFA9	124.7	98.6	93.2	Mitochondrial complex I deficiency, nuclear type 26, 618247
NDUFAF1	115.6	100	100	Mitochondrial complex I deficiency, nuclear type 11, 618234
NDUFAF2	58.6	85.7	70.9	Mitochondrial complex I deficiency, nuclear type 10, 618233
NDUFAF3	120.8	100	100	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF4	79.4	98.9	91.8	Mitochondrial complex I deficiency, nuclear type 15, 618237
NDUFAF5	95.7	98.8	94.5	Mitochondrial complex I deficiency, nuclear type 16, 616238
NDUFAF6	79.3	97.1	85.6	Mitochondrial complex I deficiency, nuclear type 17, 612392
NDUFB11	109.6	94.4	88	?Mitochondrial complex I deficiency, nuclear type 30, 301021 Linear skin defects with multiple congenital anomalies 3, 300952
NDUFB3	22.6	91.9	59.2	Mitochondrial complex I deficiency, nuclear type 25, 618246
NDUFB8	116.6	100	100	Mitochondrial complex I deficiency, nuclear type 32, 618252
NDUFB9	120.1	99.8	97.4	?Mitochondrial complex I deficiency, nuclear type 24, 618245
NDUFS1	132.2	99.8	98.6	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	117.8	100	100	Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	142.4	90.7	90.6	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	147.3	100	99.1	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS6	119.1	99.9	99.4	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFS7	118.4	100	99.7	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFS8	141.4	100	99.9	Mitochondrial complex I deficiency, nuclear type 2, 618222
NDUFV1	136.7	99.7	97.8	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	69.5	78.7	53.9	Mitochondrial complex I deficiency, nuclear type 7, 618229
NEB	124	82.9	81.9	Nemaline myopathy 2, autosomal recessive, 256030
NECAP1	116.9	100	99.9	?Epileptic encephalopathy, early infantile, 21, 615833
NECTIN1	145.4	100	100	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060
NECTIN4	135.8	100	100	Ectodermal dysplasia-syndactyly syndrome 1, 613573
NEDD4L	105.2	71.7	70.5	Periventricular nodular heterotopia 7, 617201
NEFH	111.6	96.6	87.5	?{Amyotrophic lateral sclerosis, susceptibility to}, 105400 Charcot-Marie-Tooth disease, axonal, type 2CC, 616924

NEFL	164.6	99.7	98.1	Charcot-Marie-Tooth disease, dominant intermediate G, 617882 Charcot-Marie-Tooth disease, type 1F, 607734 Charcot-Marie-Tooth disease, type 2E, 607684
NEK1	103.2	98.1	93	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520 {Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892
NEK2	110.4	98.8	93.3	?Retinitis pigmentosa 67, 615565
NEK8	171.4	100	99.9	?Nephronophthisis 9, 613824 Renal-hepatic-pancreatic dysplasia 2, 615415
NEK9	136.8	99.7	98.5	?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262 Lethal congenital contracture syndrome 10, 617022 Nevus comedonicus, somatic, 617025
NEU1	148.1	99.4	97.1	Sialidosis, type I, 256550 Sialidosis, type II, 256550
NEUROD1	166.5	100	100	Maturity-onset diabetes of the young 6, 606394 {Diabetes mellitus, noninsulin-dependent}, 125853
NEUROG3	119.4	100	99.3	Diarrhea 4, malabsorptive, congenital, 610370
NEXMIF	139.2	99.9	99	Mental retardation, X-linked 98, 300912
NEXN	79.8	94.2	79.9	Cardiomyopathy, dilated, 1CC, 613122 Cardiomyopathy, hypertrophic, 20, 613876
NF1	125.9	92.3	89.3	Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321 Watson syndrome, 193520
NF2	100.2	100	99.9	Meningioma, NF2-related, somatic, 607174 Neurofibromatosis, type 2, 101000 Schwannomatosis, somatic, 162091
NFE2L2	180.4	100	99.6	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744
NFIA	145.1	100	99.3	Brain malformations with or without urinary tract defects, 613735
NFIX	165.3	97.7	94.9	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753
NFKB1	105.3	99.3	96.8	Immunodeficiency, common variable, 12, 616576

NFKB2	123.3	97.5	92.6	Immunodeficiency, common variable, 10, 615577
NFKBIA	116.3	98.5	93.8	Ectodermal dysplasia and immunodeficiency, 612132
NFU1	47.7	94.9	77.2	Multiple mitochondrial dysfunctions syndrome 1, 605711
NGF	257.6	100	100	Neuropathy, hereditary sensory and autonomic, type V, 608654
NGLY1	128	100	99.5	Congenital disorder of deglycosylation, 615273
NHEJ1	80.3	100	99.1	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
NHLRC1	174.2	100	100	Epilepsy, progressive myoclonic 2B (Lafora), 254780
NHP2	111	100	100	Dyskeratosis congenita, autosomal recessive 2, 613987
NHS	127.1	94.3	93.3	Cataract 40, X-linked, 302200 Nance-Horan syndrome, 302350
NIN	143.5	99.7	98.6	?Seckel syndrome 7, 614851
NIPA1	174.3	99.9	99.1	Spastic paraplegia 6, autosomal dominant, 600363
NIPAL4	157.8	99.4	93.2	Ichthyosis, congenital, autosomal recessive 6, 612281
NIPBL	116.1	96.5	94.5	Cornelia de Lange syndrome 1, 122470
NKX2-1	52	96.6	83.3	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 {Thyroid cancer, nonmedullary, 1}, 188550
NKX2-5	83.2	100	99.5	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 Tetralogy of Fallot, 187500 Ventricular septal defect 3, 614432
NKX2-6	104.4	100	99.7	Conotruncal heart malformations, 217095 Persistent truncus arteriosus, 217095
NKX3-2	55.8	92.4	73.9	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330
NKX6-2	52	79	74.5	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560
NLGN4X	193.6	99.4	97.1	Mental retardation, X-linked, 300495 {Asperger syndrome susceptibility, X-linked 2}, 300497 {Autism susceptibility, X-linked 2}, 300495
NLRC4	179.9	100	99.7	?Familial cold autoinflammatory syndrome 4, 616115 Autoinflammation with infantile enterocolitis, 616050

NLRP1	126.1	99	96.5	Autoinflammation with arthritis and dyskeratosis, 617388 Palmoplantar carcinoma, multiple self-healing, 615225 {Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579
NLRP12	165.9	100	99.9	Familial cold autoinflammatory syndrome 2, 611762
NLRP3	150.4	100	100	CINCA syndrome, 607115 Deafness, autosomal dominant 34, with or without inflammation, 617772 Familial cold inflammatory syndrome 1, 120100 Keratoendothelitis fugax hereditaria, 148200 Muckle-Wells syndrome, 191900
NLRP7	135.9	99.8	98.7	Hydatidiform mole, recurrent, 1, 231090
NME1	104.2	99.9	99.2	Neuroblastoma, 256700
NME8	105.6	97.5	91	Ciliary dyskinesia, primary, 6, 610852
NMNAT1	137.9	100	99.7	Leber congenital amaurosis 9, 608553
NNT	136.9	98.6	97.1	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736
NOBOX	79.9	99.5	97.4	Premature ovarian failure 5, 611548
NOD2	135.8	100	99.7	Blau syndrome, 186580 {Inflammatory bowel disease 1, Crohn disease}, 266600 {Psoriatic arthritis, susceptibility to}, 607507 {Yao syndrome}, 617321
NODAL	160.7	100	99.9	Heterotaxy, visceral, 5, 270100
NOG	191.9	100	100	Brachydactyly, type B2, 611377 Multiple synostoses syndrome 1, 186500 Stapes ankylosis with broad thumbs and toes, 184460 Symphalangism, proximal, 1A, 185800 Tarsal-carpal coalition syndrome, 186570
NOL3	76.3	93.5	83.9	Myoclonus, familial cortical, 614937
NONO	93.8	99.7	96.4	Mental retardation, X-linked, syndromic 34, 300967
NOP10	160.5	100	100	Dyskeratosis congenita, autosomal recessive 1, 224230
NOP56	137.3	99.9	98.6	Spinocerebellar ataxia 36, 614153
NOTCH1	137.5	99.1	98	Adams-Oliver syndrome 5, 616028 Aortic valve disease 1, 109730
NOTCH2	172.4	100	99.9	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500

NOTCH3	110.6	93	88.2	?Myofibromatosis, infantile 2, 615293 Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310 Lateral meningocele syndrome, 130720
NPC1	147.9	99.2	97.8	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220
NPC2	140.7	100	99.9	Niemann-pick disease, type C2, 607625
NPHP1	117.6	98.8	96.4	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NPHP3	115.6	99.4	96.1	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540
NPHP4	136.7	99.9	99.3	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
NPHS1	102	99.9	98.9	Nephrotic syndrome, type 1, 256300
NPHS2	116.9	99.8	95.9	Nephrotic syndrome, type 2, 600995
NPM1	70.3	88.7	77.9	Leukemia, acute myeloid, somatic, 601626
NPPA	115.4	100	100	Atrial fibrillation, familial, 6, 612201 Atrial standstill 2, 615745
NPR2	164.8	100	100	Acromesomelic dysplasia, Maroteaux type, 602875 Epiphyseal chondrodysplasia, Miura type, 615923 Short stature with nonspecific skeletal abnormalities, 616255
NPRL2	168.5	100	100	Epilepsy, familial focal, with variable foci 2, 617116
NPRL3	127.5	100	99.7	Epilepsy, familial focal, with variable foci 3, 617118
NR0B1	119.3	99.9	98.6	46XY sex reversal 2, dosage-sensitive, 300018 Adrenal hypoplasia, congenital, 300200
NR0B2	96.8	100	99.8	Obesity, mild, early-onset, 601665
NR1H4	141.9	96.6	92.2	Cholestasis, progressive familial intrahepatic, 5, 617049
NR2E3	93.4	99.9	98.9	Enhanced S-cone syndrome, 268100 Retinitis pigmentosa 37, 611131
NR2F1	201.6	99.9	98.4	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NR2F2	246.1	98.7	94.3	Congenital heart defects, multiple types, 4, 615779
NR3C1	137.4	100	99.8	Glucocorticoid resistance, 615962

NR3C2	159.5	99.4	95.9	Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115 Pseudohypoaldosteronism type I, autosomal dominant, 177735
NR4A3	112.6	99.8	98	Chondrosarcoma, extraskeletal myxoid, 612237
NR5A1	79.9	100	98.3	46, XX sex reversal 4, 617480 46XY sex reversal 3, 612965 Adrenocortical insufficiency, 612964 Premature ovarian failure 7, 612964 Spermatogenic failure 8, 613957
NRAS	188.4	100	100	?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 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
NRL	68.7	99.7	95.8	Retinal degeneration, autosomal recessive, clumped pigment type, 0 Retinitis pigmentosa 27, 613750
NRXN1	160.9	96.8	95.7	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332
NSD1	155.2	100	99.9	Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550
NSDHL	169.2	99.9	98.2	CHILD syndrome, 308050 CK syndrome, 300831
NSMCE2	100.3	99	92.5	Seckel syndrome 10, 617253
NSMCE3	130	99.9	98.5	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241
NSMF	78.4	95.7	95.2	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
NSUN2	114.7	95.3	92.2	Mental retardation, autosomal recessive 5, 611091
NT5C2	125.3	97.1	92.7	Spastic paraplegia 45, autosomal recessive, 613162
NT5C3A	62	89.3	78.8	Anemia, hemolytic, due to UMPH1 deficiency, 266120
NT5E	166.3	100	99.8	Calcification of joints and arteries, 211800
NTF4	98.8	99.7	92.1	Glaucoma 1, open angle, 1O, 613100

NTHL1	98.1	99.1	95.7	Familial adenomatous polyposis 3, 616415
NTRK1	130.6	99.7	97.7	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma, familial, 155240
NTRK2	170.8	100	100	Epileptic encephalopathy, early infantile, 58, 617830 Obesity, hyperphagia, and developmental delay, 613886
NUBPL	89.8	92.9	85.9	Mitochondrial complex I deficiency, nuclear type 21, 618242
NUMA1	127.1	100	99.5	Leukemia, acute promyelocytic, somatic, 612376
NUP107	122.9	99.2	94.1	?Ovarian dysgenesis 6, 618078 Nephrotic syndrome, type 11, 616730
NUP133	127.1	96.9	94.3	Nephrotic syndrome, type 18, 618177
NUP155	115.1	97.6	92.3	?Atrial fibrillation 15, 615770
NUP160	161.1	100	99.8	?Nephrotic syndrome, type 19, 618178
NUP205	133.7	98.9	98	?Nephrotic syndrome, type 13, 616893
NUP214	167.3	99.8	99.2	Leukemia, acute myeloid, somatic, 601626 Leukemia, T-cell acute lymphoblastic, somatic, 613065
NUP37	168.6	98.5	93.5	?Microcephaly 24, primary, autosomal recessive, 618179
NUP62	111.6	100	99.9	Striatonigral degeneration, infantile, 271930
NUP85	135.3	100	100	Nephrotic syndrome, type 17, 618176
NUP93	140.7	97.9	94.9	Nephrotic syndrome, type 12, 616892
NUS1	69.6	62	40.7	?Congenital disorder of glycosylation, type 1aa, 617082 Mental retardation, autosomal dominant 55, with seizures, 617831
NYX	94.9	98.1	96	Night blindness, congenital stationary (complete), 1A, X-linked, 310500
OAT	89.2	77.7	70.5	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OBSL1	140.5	99.8	98.5	3-M syndrome 2, 612921
OCA2	139.9	99.5	97.9	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	220.7	100	100	Pseudo-TORCH syndrome 1, 251290
OCRL	122.2	98.8	96.3	Dent disease 2, 300555 Lowe syndrome, 309000

OFD1	51.5	84	67.8	?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209
OGG1	128.5	100	99.6	Renal cell carcinoma, clear cell, somatic, 144700
OGT	125.9	100	99.4	Mental retardation, X-linked 106, 300997
OPA1	122.5	99.1	94.1	?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896 Behr syndrome, 210000 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250 {Glaucoma, normal tension, susceptibility to}, 606657
OPA3	128	99.5	97.4	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPHN1	89	99.1	96.2	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
OPLAH	109.4	99.4	97.9	5-oxoprolinase deficiency, 260005
OPN1LW	76.6	67.4	61.9	Blue cone monochromacy, 303700 Colorblindness, protan, 303900
OPN1MW	67.2	68.2	60.4	Blue cone monochromacy, 303700 Colorblindness, deutan, 303800
OPN1SW	130.2	100	100	Colorblindness, tritan, 190900
OPTN	113.8	100	99.4	Amyotrophic lateral sclerosis 12, 613435 Glaucoma 1, open angle, E, 137760 {Glaucoma, normal tension, susceptibility to}, 606657
ORAI1	237.3	93.8	89.8	Immunodeficiency 9, 612782 Myopathy, tubular aggregate, 2, 615883
ORC1	106.7	99.9	98.9	Meier-Gorlin syndrome 1, 224690
ORC4	57.6	95.8	82	Meier-Gorlin syndrome 2, 613800
ORC6	126.8	100	100	Meier-Gorlin syndrome 3, 613803
OSBPL2	145.8	100	100	Deafness, autosomal dominant 67, 616340
OSGEP	120.5	100	99.6	Galloway-Mowat syndrome 3, 617729
OSMR	145.9	100	99.9	Amyloidosis, primary localized cutaneous, 1, 105250
OSTM1	80.7	90.8	88.4	Osteopetrosis, autosomal recessive 5, 259720
OTC	123.3	99.9	99.4	Ornithine transcarbamylase deficiency, 311250
OTOA	117.3	99	96.6	Deafness, autosomal recessive 22, 607039

OTOF	131.2	100	99.7	Auditory neuropathy, autosomal recessive, 1, 601071 Deafness, autosomal recessive 9, 601071
OTOG	145.1	99.4	98.2	Deafness, autosomal recessive 18B, 614945
OTOGL	113.3	98.4	93.9	Deafness, autosomal recessive 84B, 614944
OTUD6B	123.4	99.9	98.2	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452
OTULIN	149.5	90.5	86.3	Autoinflammation, panniculitis, and dermatosis syndrome, 617099
OTX2	154.8	100	99.8	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125
OVOL2	119.1	96.9	90.6	Corneal dystrophy, posterior polymorphous, 1, 122000
OXCT1	121.4	99.6	97.8	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050
P2RX2	132.8	99	95.5	Deafness, autosomal dominant 41, 608224
P2RY12	186.2	100	100	Bleeding disorder, platelet-type, 8, 609821
P3H1	137	100	99.9	Osteogenesis imperfecta, type VIII, 610915
P3H2	100.2	99.2	93.4	Myopia, high, with cataract and vitreoretinal degeneration, 614292
P4HA2	158.7	100	99.5	Myopia 25, autosomal dominant, 617238
P4HB	105.1	94.6	94.4	Cole-Carpenter syndrome 1, 112240
PABPN1	66.6	62.4	60.5	Oculopharyngeal muscular dystrophy, 164300
PACS1	117.4	97.5	95.7	Schuurs-Hoeijmakers syndrome, 615009
PACS2	150.7	98.4	95.9	Epileptic encephalopathy, early infantile, 66, 618067
PADI3	148.5	100	100	Uncombable hair syndrome, 191480
PADI6	110.5	100	99	Preimplantation embryonic lethality 2, 617234
PAFAH1B1	105.2	89.1	81.4	Lissencephaly 1, 607432 Subcortical laminar heterotopia, 607432
PAH	151.7	100	100	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600
PAK1	119.4	100	99.4	Intellectual developmental disorder with macrocephaly, seizures, and speech delay, 618158
PAK3	82.8	97.6	91.8	Mental retardation, X-linked 30/47, 300558
PALB2	152.6	100	99.7	Fanconi anemia, complementation group N, 610832 {Breast cancer, susceptibility to}, 114480 {Pancreatic cancer, susceptibility to, 3}, 613348

PAM16	50.7	65.2	64.7	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320
PANK2	146.6	99.3	93.1	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PAPSS2	108.5	99.7	98.5	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847
PARK7	86.9	100	99	Parkinson disease 7, autosomal recessive early-onset, 606324
PARN	128.4	99.9	98	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
PATL2	107.8	99.9	99.2	Oocyte maturation defect 4, 617743
PAX1	132.4	87.7	82.4	?Otofaciocervical syndrome 2, 615560
PAX2	168.5	99.9	99.3	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330
PAX3	118.5	100	100	Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220 Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820
PAX4	96.6	99.9	98.8	Diabetes mellitus, type 2, 125853 Maturity-onset diabetes of the young, type IX, 612225 {Diabetes mellitus, ketosis-prone, susceptibility to}, 612227
PAX6	119.9	100	99.9	?Coloboma of optic nerve, 120430 ?Coloboma, ocular, 120200 ?Morning glory disc anomaly, 120430 Aniridia, 106210 Anterior segment dysgenesis 5, multiple subtypes, 604229 Cataract with late-onset corneal dystrophy, 106210 Foveal hypoplasia 1, 136520 Keratitis, 148190 Optic nerve hypoplasia, 165550
PAX7	117.8	100	100	Rhabdomyosarcoma 2, alveolar, 268220
PAX8	94.1	100	99.9	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700
PAX9	238.8	99.6	99.3	Tooth agenesis, selective, 3, 604625

PBX1	111.8	99.3	95.2	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PC	149.3	97.7	94.6	Pyruvate carboxylase deficiency, 266150
PCBD1	113.3	99.5	99.1	Hyperphenylalaninemia, BH4-deficient, D, 264070
PCCA	103.1	96.4	89.2	Propionicacidemia, 606054
PCCB	129.8	98.7	96.4	Propionicacidemia, 606054
PCDH12	206.7	100	100	Microcephaly, seizures, spasticity, and brain calcification, 251280
PCDH15	153.9	99	98	Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1F, 602083
PCDH19	224.1	100	99.3	Epileptic encephalopathy, early infantile, 9, 300088
PCK1	143	100	100	?Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680
PCLO	165.2	99.7	98.5	?Pontocerebellar hypoplasia, type 3, 608027
PCNA	92.1	100	98.2	?Ataxia-telangiectasia-like disorder 2, 615919
PCNT	117.6	98.9	96	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PCSK1	147.2	100	99	Obesity with impaired prohormone processing, 600955 {Obesity, susceptibility to, BMIQ12}, 612362
PCSK9	99.3	94.3	91.3	Hypercholesterolemia, familial, 3, 603776 {Low density lipoprotein cholesterol level QTL 1}, 603776
PCYT1A	113.5	98.3	94.7	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDCD10	85.1	96.3	88	Cerebral cavernous malformations 3, 603285
PDE10A	119.8	81.2	80.8	Dyskinesia, limb and orofacial, infantile-onset, 616921 Striatal degeneration, autosomal dominant, 616922
PDE11A	168.9	99.9	98.9	Pigmented nodular adrenocortical disease, primary, 2, 610475
PDE1C	124	100	99.7	?Deafness, autosomal dominant 74, 618140
PDE3A	124.5	99.9	98.8	Hypertension and brachydactyly syndrome, 112410
PDE4D	101.2	92.8	88.3	Acrodysostosis 2, with or without hormone resistance, 614613 {Stroke, susceptibility to, 1}, 606799
PDE6A	125.8	100	99.8	Retinitis pigmentosa 43, 613810
PDE6B	147.9	100	100	Night blindness, congenital stationary, autosomal dominant 2, 163500 Retinitis pigmentosa-40, 613801

PDE6C	137	99	96.7	Cone dystrophy 4, 613093
PDE6D	106.1	100	99.9	?Joubert syndrome 22, 615665
PDE6G	95.3	99.5	96.3	Retinitis pigmentosa 57, 613582
PDE6H	68.4	97.6	77	Achromatopsia 6, 610024 Retinal cone dystrophy 3, 610024
PDE8B	111	99.9	98.9	Pigmented nodular adrenocortical disease, primary, 3, 614190 Striatal degeneration, autosomal dominant, 609161
PDGFB	95.1	100	100	Basal ganglia calcification, idiopathic, 5, 615483 Dermatofibrosarcoma protuberans, 607907 Meningioma, SIS-related, 607174
PDGFRA	148.3	100	100	Gastrointestinal stromal tumor, somatic, 606764 Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685
PDGFRB	147.1	99.1	96.5	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	109.8	98.1	92.1	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	133	99.3	96.8	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	132.5	98.9	94.6	Lacticacidemia due to PDX1 deficiency, 245349
PDK3	105.1	96.4	94.3	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905
PDP1	209.6	100	100	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	116.7	88.8	78.7	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	126.8	96.5	93.5	Coenzyme Q10 deficiency, primary, 3, 614652
PDX1	35.4	89	72.1	MODY, type IV, 606392 Pancreatic agenesis 1, 260370 {Diabetes mellitus, type II, susceptibility to}, 125853
PDYN	107.1	100	99.9	Spinocerebellar ataxia 23, 610245
PDZD7	80.9	98.4	93.9	Deafness, autosomal recessive 57, 618003 Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472 {Retinal disease in Usher syndrome type IIA, modifier of}, 276901
PEPD	116	99.6	98.5	Prolidase deficiency, 170100

PER2	95.7	100	99.6	Advanced sleep phase syndrome, familial, 1, 604348
PER3	170.7	99.2	95.8	?Advanced sleep phase syndrome, familial, 3, 616882
PET100	94.5	88.8	74.8	Mitochondrial complex IV deficiency, 220110
PEX1	115.8	97.7	95.4	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	111.8	96.1	90.1	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX11B	105.7	99.7	98.3	?Peroxisome biogenesis disorder 14B, 614920
PEX12	168.3	100	100	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	197.6	99.8	98.7	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	149	99.7	97.5	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	137	97.1	93.1	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	92.9	99.9	99.2	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	147.1	100	100	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	76.4	100	99.8	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	98.1	99.1	94.3	?Peroxisome biogenesis disorder 10B, 617370 Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	111.7	99.9	98.3	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	94.5	90.4	86.1	Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863
PEX7	113.5	89.6	82	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PFKM	150.4	100	99.8	Glycogen storage disease VII, 232800
PFN1	152	100	100	Amyotrophic lateral sclerosis 18, 614808
PGAM2	170.9	100	99.9	Glycogen storage disease X, 261670

PGAP1	98.1	94.9	88.6	Mental retardation, autosomal recessive 42, 615802
PGAP2	158.4	100	100	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGAP3	72.8	62.5	58	Hyperphosphatasia with mental retardation syndrome 4, 615716
PGK1	54.5	93.3	81.6	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	133.6	100	99.9	Congenital disorder of glycosylation, type It, 614921
PGM3	191.4	99.9	99.7	Immunodeficiency 23, 615816
PHC1	234.9	100	99.9	?Microcephaly 11, primary, autosomal recessive, 615414
PHEX	125	99.9	98	Hypophosphatemic rickets, X-linked dominant, 307800
PHF6	62.8	92.6	83.7	Borjeson-Forssman-Lehmann syndrome, 301900
PHF8	94	99.8	97.7	Mental retardation syndrome, X-linked, Siderius type, 300263
PHGDH	115.6	100	99.8	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHIP	124.5	95.9	91	Developmental delay, intellectual disability, obesity, and dysmorphic features, 617991
PHKA1	106.7	98.9	95.3	Muscle glycogenosis, 300559
PHKA2	108.2	100	99.5	Glycogen storage disease, type IXa1, 306000 Glycogen storage disease, type IXa2, 306000
PHKB	130.4	99.8	97.5	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750
PHKG2	159.1	100	100	Cirrhosis due to liver phosphorylase kinase deficiency, 0 Glycogen storage disease IXc, 613027
PHOX2A	29.9	59.9	32.6	Fibrosis of extraocular muscles, congenital, 2, 602078
PHOX2B	92.9	93	87.2	Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, 209880 Neuroblastoma with Hirschsprung disease, 613013 {Neuroblastoma, susceptibility to, 2}, 613013
PHYH	74.6	97.5	90.8	Refsum disease, 266500
PI4KA	112.8	93.6	89.4	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531
PIBF1	66	96.6	82.4	Joubert syndrome 33, 617767
PICALM	106.1	99.9	96.1	Leukemia, acute myeloid, somatic, 601626
PIEZO1	140.2	99.5	97.4	Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380 Lymphatic malformation 6, 616843

PIEZO2	126.1	99.9	99.2	?Marden-Walker syndrome, 248700 Arthrogryposis, distal, type 3, 114300 Arthrogryposis, distal, type 5, 108145 Arthrogryposis, distal, with impaired proprioception and touch, 617146
PIGA	90.5	90.4	81.3	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGC	129	99.7	96.4	Glycosylphosphatidylinositol biosynthesis defect 16, 617816
PIGG	167.4	100	99.7	Mental retardation, autosomal recessive 53, 616917
PIGH	96.5	77.8	68.5	Glycosylphosphatidylinositol biosynthesis defect 17, 618010
PIGL	121.8	99.9	99.3	CHIME syndrome, 280000
PIGM	165.4	100	100	Glycosylphosphatidylinositol deficiency, 610293
PIGN	111.3	92.6	87.1	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	147	100	99.9	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGP	101.4	91.6	83.8	?Epileptic encephalopathy, early infantile, 55, 617599
PIGS	108.8	100	99.9	Glycosylphosphatidylinositol biosynthesis defect 18, 618143
PIGT	171.3	98.1	98	?Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGV	145.5	100	100	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIGW	147.6	100	99.8	Glycosylphosphatidylinositol biosynthesis defect 11, 616025
PIGY	121.4	100	99.9	Hyperphosphatasia with mental retardation syndrome 6, 616809
PIH1D3	70.5	94.3	79.6	Ciliary dyskinesia, primary, 36, X-linked, 300991

PIK3CA	120.7	99.9	99.1	Breast cancer, somatic, 114480 CLAPO syndrome, somatic, 613089 CLOVE syndrome, somatic, 612918 Colorectal cancer, somatic, 114500 Cowden syndrome 5, 615108 Gastric cancer, somatic, 613659 Hepatocellular carcinoma, somatic, 114550 Keratosis, seborrheic, somatic, 182000 Macrodactyly, somatic, 155500 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Nevus, epidermal, somatic, 162900 Nonsmall cell lung cancer, somatic, 211980 Ovarian cancer, somatic, 167000
PIK3CD	132.7	99.2	96.8	Immunodeficiency 14, 615513
PIK3R1	129.3	99.7	97.3	?Agammaglobulinemia 7, autosomal recessive, 615214 Immunodeficiency 36, 616005 SHORT syndrome, 269880
PIK3R2	86.2	89.1	86.1	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PIK3R5	110.1	100	99.8	Ataxia-oculomotor apraxia 3, 615217
PIKFYVE	141.6	99.8	98.4	Corneal fleck dystrophy, 121850
PINK1	90.3	87.2	81.1	Parkinson disease 6, early onset, 605909
PIP5K1C	107.6	96.3	95.1	Lethal congenital contractural syndrome 3, 611369
PITPNM3	117.2	99	97.8	Cone-rod dystrophy 5, 600977
PITX1	144.6	91.4	86.8	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800 Liebenberg syndrome, 186550
PITX2	147.8	99.7	97.5	Anterior segment dysgenesis 4, 137600 Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550
PITX3	40.2	95.6	82.8	Anterior segment dysgenesis 1, multiple subtypes, 107250 Cataract 11, multiple types, 610623 Cataract 11, syndromic, autosomal recessive, 610623
PKD1	28.1	42.6	34.5	Polycystic kidney disease 1, 173900
PKD1L1	123.8	100	99.6	Heterotaxy, visceral, 8, autosomal, 617205

PKD2	110.6	89.3	84.2	Polycystic kidney disease 2, 613095
PKHD1	154.9	100	99.7	Polycystic kidney disease 4, with or without hepatic disease, 263200
PKLR	178.8	100	100	Adenosine triphosphate, elevated, of erythrocytes, 102900 Pyruvate kinase deficiency, 266200
PKP1	122	99.9	98.5	Ectodermal dysplasia/skin fragility syndrome, 604536
PKP2	99.6	94.6	87.7	Arrhythmogenic right ventricular dysplasia 9, 609040
PLA2G4A	134.3	99.9	98.8	Phospholipase A2, group IV A, deficiency of, 0
PLA2G6	117.5	99.9	98.4	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953
PLA2G7	125.6	99.9	97.2	Platelet-activating factor acetylhydrolase deficiency, 614278 {Asthma, susceptibility to}, 600807 {Atopy, susceptibility to}, 147050
PLAA	169.4	100	99.1	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527
PLAG1	219.1	100	100	Adenomas, salivary gland pleomorphic, somatic, 181030
PLAU	111.4	99.8	98.4	Quebec platelet disorder, 601709 {Alzheimer disease, late-onset, susceptibility to}, 104300
PLCB1	142.8	100	99.7	Epileptic encephalopathy, early infantile, 12, 613722
PLCB4	126.1	99.2	95.7	Auriculocondylar syndrome 2, 614669
PLCD1	116.9	99.5	97.1	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCE1	155.3	99.5	98.9	Nephrotic syndrome, type 3, 610725
PLCG2	118.9	100	99.8	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468
PLCZ1	67.1	96.8	85.1	?Spermatogenic failure 17, 617214
PLD1	125.2	99.9	99.1	Cardiac valvular defect, developmental, 212093
PLD3	157.2	100	99.3	?Spinocerebellar ataxia 46, 617770

PLEC	114.1	99.7	98.7	?Epidermolysis bullosa simplex with nail dystrophy, 616487 Epidermolysis bullosa simplex with muscular dystrophy, 226670 Epidermolysis bullosa simplex with pyloric atresia, 612138 Epidermolysis bullosa simplex, Ogna type, 131950 Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723
PLEKHG2	131.7	99.8	97.9	Leukodystrophy and acquired microcephaly with or without dystonia, 616763
PLEKHG5	86.9	96.2	89	Charcot-Marie-Tooth disease, recessive intermediate C, 615376 Spinal muscular atrophy, distal, autosomal recessive, 4, 611067
PLEKHM1	141.1	100	99.9	?Osteopetrosis, autosomal recessive 6, 611497 Osteopetrosis, autosomal dominant 3, 618107
PLG	115.4	87.8	87	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PLIN1	81	96.2	88.4	Lipodystrophy, familial partial, type 4, 613877
PLK4	145.5	99.5	96.3	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLN	209.7	100	100	Cardiomyopathy, dilated, 1P, 609909 Cardiomyopathy, hypertrophic, 18, 613874
PLOD1	137.9	99.8	97.5	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD2	108.6	94.7	88.6	Bruck syndrome 2, 609220
PLOD3	100.1	99	96.3	Lysyl hydroxylase 3 deficiency, 612394
PLP1	129.2	100	99.4	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PLPBP	112.6	99.1	92.6	Epilepsy, early-onset, vitamin B6-dependent, 617290
PLPP6	164.4	96.6	84.4	Phospholipid phosphatase 6, 611666
PLS3	131.5	96.8	95.2	Bone mineral density QTL18, osteoporosis, 300910
PLVAP	158.9	100	100	Diarrhea 10, protein-losing enteropathy type, 618183
PMFBP1	109.6	100	99.5	Spermatogenic failure 31, 618112
PML	124.4	100	99.9	Leukemia, acute promyelocytic, PML/RARA type, 0
PMM2	141.1	99.9	99.4	Congenital disorder of glycosylation, type Ia, 212065

PMP22	111.2	96.7	91.9	?Neuropathy, inflammatory demyelinating, 139393 Charcot-Marie-Tooth disease, type 1A, 118220 Charcot-Marie-Tooth disease, type 1E, 118300 Dejerine-Sottas disease, 145900 Neuropathy, recurrent, with pressure palsies, 162500 Roussy-Levy syndrome, 180800
PMPCA	120.8	99.4	96.8	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMPCB	121	99.7	97.8	Multiple mitochondrial dysfunctions syndrome 6, 617954
PMS2	95.1	83.5	80.7	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 Mismatch repair cancer syndrome, 276300
PMVK	125.3	100	99.9	Porokeratosis 1, multiple types, 175800
PNKD	99.8	100	99.2	Paroxysmal nonkinesigenic dyskinesia 1, 118800
PNKP	93	99.8	97.7	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNLIP	160.5	99.5	95.6	?Pancreatic lipase deficiency, 614338
PNP	151.4	100	99.5	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA1	192.6	100	100	Ichthyosis, congenital, autosomal recessive 10, 615024
PNPLA2	113.2	99.7	97.4	Neutral lipid storage disease with myopathy, 610717
PNPLA6	122.1	99.7	98.5	?Laurence-Moon syndrome, 245800 Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 Spastic paraplegia 39, autosomal recessive, 612020
PNPLA8	114.4	100	99.7	?Mitochondrial myopathy with lactic acidosis, 251950
PNPO	66.4	100	98.3	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
PNPT1	53.7	93.3	80.9	Combined oxidative phosphorylation deficiency 13, 614932 Deafness, autosomal recessive 70, 614934
POC1A	133.8	100	100	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POC1B	78.7	98	94.2	Cone-rod dystrophy 20, 615973
POF1B	74.9	90.9	81	?Premature ovarian failure 2B, 300604
POFUT1	139.4	99.9	97.5	Dowling-Degos disease 2, 615327
POGLUT1	117.4	98.2	93.8	?Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232 Dowling-Degos disease 4, 615696
POGZ	168.2	99.4	99.2	White-Sutton syndrome, 616364

POLA1	110.7	98.2	92.8	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220
POLD1	101.2	93.9	90.8	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381 {Colorectal cancer, susceptibility to, 10}, 612591
POLE	144.1	100	99.5	FILS syndrome, 615139 {Colorectal cancer, susceptibility to, 12}, 615083
POLG	114.4	100	99.5	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	157.5	98.8	96.8	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131
POLH	140.7	100	99.8	Xeroderma pigmentosum, variant type, 278750
POLR1A	120.1	99.9	99.1	Acrofacial dysostosis, Cincinnati type, 616462
POLR1C	117	99.7	96.1	Leukodystrophy, hypomyelinating, 11, 616494 Treacher Collins syndrome 3, 248390
POLR1D	176.2	91.6	91.6	Treacher Collins syndrome 2, 613717
POLR3A	137.4	100	99.9	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	146.4	99.9	98.5	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMC	116.2	100	100	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734 {Obesity, early-onset, susceptibility to}, 601665
POMGNT1	127.6	99.7	97.1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 Retinitis pigmentosa 76, 617123
POMGNT2	259.6	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830 Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135
POMK	205.1	100	100	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249

POMP	114.4	95.2	87.5	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952 Proteasome-associated autoinflammatory syndrome 2, 618048
POMT1	155.7	99.7	98.1	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	111.1	98.9	97.5	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158
POP1	114.2	100	99.7	Anauxetic dysplasia 2, 617396
POR	167.7	99.9	98.7	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571
PORCN	117.7	100	99.3	Focal dermal hypoplasia, 305600
POU1F1	106.3	98.2	94.7	Pituitary hormone deficiency, combined, 1, 613038
POU3F4	151	100	99.9	Deafness, X-linked 2, 304400
POU4F3	298.1	100	100	Deafness, autosomal dominant 15, 602459
PPA2	80.4	94.6	82.5	?Sudden cardiac failure, alcohol-induced, 617223 Sudden cardiac failure, infantile, 617222
PPARG	153.9	100	99.9	Carotid intimal medial thickness 1, 609338 Insulin resistance, severe, digenic, 604367 Lipodystrophy, familial partial, type 3, 604367 Obesity, severe, 601665 [Obesity, resistance to], 0 {Diabetes, type 2}, 125853
PPCS	108.7	99.9	98.4	Cardiomyopathy, dilated, 2C, 618189
PPIB	118.4	100	100	Osteogenesis imperfecta, type IX, 259440
PPM1D	166.7	100	99.8	Breast cancer, somatic, 114480 Intellectual developmental disorder with gastrointestinal difficulties and high pain threshold, 617450
PPM1K	180	100	100	?Maple syrup urine disease, mild variant, 615135
PPOX	96.1	99.7	98.2	Porphyria variegata, 176200

PPP1CB	96.6	99.6	98.4	Noonan syndrome-like disorder with loose anagen hair 2, 617506
PPP1R15B	133.4	99.4	98	Microcephaly, short stature, and impaired glucose metabolism 2, 616817
PPP1R3A	146.5	99.5	98	Insulin resistance, severe, digenic, 125853
PPP2R1A	134	91.7	91.6	Mental retardation, autosomal dominant 36, 616362
PPP2R1B	156	100	99.8	Lung cancer, 211980
PPP2R2B	141.9	99.7	97.3	Spinocerebellar ataxia 12, 604326
PPP2R5D	143.2	100	99.8	Mental retardation, autosomal dominant 35, 616355
PPP3CA	123.8	99	92.9	Epileptic encephalopathy, infantile or early childhood, 1, 617711
PPT1	144.5	90	87.3	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	186.1	100	100	Renpenning syndrome, 309500
PRCC	146.6	99.8	97.9	Renal cell carcinoma, papillary, 605074
PRCD	89.4	100	99.9	Retinitis pigmentosa 36, 610599
PRDM12	112.2	91	87.7	Neuropathy, hereditary sensory and autonomic, type VIII, 616488
PRDM16	161.5	100	99.1	Cardiomyopathy, dilated, 1LL, 615373 Left ventricular noncompaction 8, 615373
PRDM5	129.4	99.5	95.5	Brittle cornea syndrome 2, 614170
PRDM6	108.3	92.8	79	Patent ductus arteriosus 3, 617039
PRDM8	81.2	92	85.1	?Epilepsy, progressive myoclonic, 10, 616640
PRDX1	115	100	99.6	Methylmalonic aciduria and homocystinuria, cblC type, digenic, 277400
PREPL	107.6	99.5	95.9	Myasthenic syndrome, congenital, 22, 616224
PRF1	122.5	91.2	90.8	Aplastic anemia, 609135 Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027
PRG4	144.6	97.6	88.4	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250
PRICKLE1	117.3	100	100	Epilepsy, progressive myoclonic 1B, 612437
PRIMPOL	110.1	95.7	90.1	Myopia 22, autosomal dominant, 615420
PRKACA	97.4	78.9	76.9	Cushing syndrome, ACTH-independent adrenal, somatic, 615830
PRKACG	217.6	100	99.9	?Bleeding disorder, platelet-type, 19, 616176

PRKAG2	125.6	98.1	91.6	Cardiomyopathy, hypertrophic 6, 600858 Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200
PRKAR1A	90.7	99.1	93.9	Acrodysostosis 1, with or without hormone resistance, 101800 Adrenocortical tumor, somatic, 0 Carney complex, type 1, 160980 Myxoma, intracardiac, 255960 Pigmented nodular adrenocortical disease, primary, 1, 610489
PRKCA	150.1	100	100	Pituitary tumor, invasive, 0
PRKCD	181.2	100	99.9	Autoimmune lymphoproliferative syndrome, type III, 615559
PRKCG	116.1	99	94.5	Spinocerebellar ataxia 14, 605361
PRKCSH	135.2	99.7	96.3	Polycystic liver disease 1, 174050
PRKD1	145.3	97.4	94.8	Congenital heart defects and ectodermal dysplasia, 617364
PRKDC	106.7	98.4	94.8	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PRKG1	123.4	98.7	95.4	Aortic aneurysm, familial thoracic 8, 615436
PRKN	98.6	79.6	78.8	Adenocarcinoma of lung, somatic, 211980 Adenocarcinoma, ovarian, somatic, 167000 Parkinson disease, juvenile, type 2, 600116 {Leprosy, susceptibility to}, 607572
PRKRA	179.6	99.8	98.4	Dystonia 16, 612067
PRLR	151.2	100	99.9	?Hyperprolactinemia, 615555 Multiple fibroadenomas of the breast, 615554
PRMT7	138.7	100	99.8	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157
PRNP	153.4	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	138.5	99.7	97.2	Thrombophilia due to protein C deficiency, autosomal dominant, 176860 Thrombophilia due to protein C deficiency, autosomal recessive, 612304

PRODH	83.8	84.9	82.3	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850
PROK2	105.6	98.4	91.9	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	331.8	100	100	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROM1	112.3	95.4	92.8	Cone-rod dystrophy 12, 612657 Macular dystrophy, retinal, 2, 608051 Retinitis pigmentosa 41, 612095 Stargardt disease 4, 603786
PROP1	76.5	91.6	84.3	Pituitary hormone deficiency, combined, 2, 262600
PROS1	101.4	96.8	91.4	Thrombophilia due to protein S deficiency, autosomal dominant, 612336 Thrombophilia due to protein S deficiency, autosomal recessive, 614514
PRPF3	86	98.8	96	Retinitis pigmentosa 18, 601414
PRPF31	115.9	97.5	92	Retinitis pigmentosa 11, 600138
PRPF4	149.4	100	99.4	Retinitis pigmentosa 70, 615922
PRPF6	130.3	100	100	Retinitis pigmentosa 60, 613983
PRPF8	139	99.9	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 form, 608133 Retinitis punctata albescens, 136880
PRPS1	149.5	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	99.9	98.4	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751
PRRX1	100.2	100	99.9	Agnathia-otocephaly complex, 202650
PRSS1	190.8	100	99.9	Pancreatitis, hereditary, 167800

				Trypsinogen deficiency, 614044
PRSS12	153.3	99.9	98.5	Mental retardation, autosomal recessive 1, 249500
PRSS56	51.5	96.6	83.8	Microphthalmia, isolated 6, 613517
PRUNE1	136.1	100	100	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481
PRX	115.6	99.8	98.3	Charcot-Marie-Tooth disease, type 4F, 614895 Dejerine-Sottas disease, 145900
PSAP	114.4	99.9	99	Combined SAP deficiency, 611721 Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PSAT1	53.2	91.4	75.8	?Phosphoserine aminotransferase deficiency, 610992 Neu-Laxova syndrome 2, 616038
PSEN1	160.7	100	99.9	?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	123.9	100	100	Alzheimer disease-4, 606889 Cardiomyopathy, dilated, 1V, 613697
PSENEN	67.6	100	98.4	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736
PSMB4	122.5	100	99.2	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591
PSMB8	118.7	100	99.8	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040
PSMB9	85.6	99.5	95.7	?Proteasome-associated autoinflammatory syndrome 3, digenic, 617591
PSMC3IP	113.4	99.9	99.7	Ovarian dysgenesis 3, 614324
PSMD12	76.3	98.1	90.9	Stankiewicz-Isidor syndrome, 617516
PSPH	128.9	98.8	95.4	Phosphoserine phosphatase deficiency, 614023
PSTPIP1	88.2	99.7	97.7	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416

PTCH1	114.6	98.4	95.9	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly 7, 610828
PTCH2	120.1	99.4	97.5	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Medulloblastoma, somatic, 155255
PTDSS1	127.2	100	100	Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	143.2	99.6	96	Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309 VATER association with macrocephaly and ventriculomegaly, 276950 {Glioma susceptibility 2}, 613028 {Meningioma}, 607174 {Prostate cancer, somatic}, 176807
PTF1A	74	88.4	78.1	Pancreatic agenesis 2, 615935 Pancreatic and cerebellar agenesis, 609069
PTGIS	126.7	96.7	94.6	Hypertension, essential, 145500
PTH	103.5	99.9	97.6	Hypoparathyroidism, autosomal dominant, 146200 Hypoparathyroidism, autosomal recessive, 146200
PTH1R	108.5	99.9	98.8	Chondrodysplasia, Blomstrand type, 215045 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Metaphyseal chondrodysplasia, Murk Jansen type, 156400
PTHLH	120.5	99.4	93.2	Brachydactyly, type E2, 613382
PTPN11	103.1	97.9	92.5	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
PTPN12	144.8	97.7	94.8	Colon cancer, somatic, 114500
PTPN14	175.7	99.4	96.4	?Choanal atresia and lymphedema, 613611
PTPRC	101.6	93.9	86.3	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971 {Hepatitis C virus, susceptibility to}, 609532
PTPRF	170.9	100	99.9	?Breasts and/or nipples, aplasia or hypoplasia of, 2, 616001

PTPRJ	169	97.2	96.1	Colon cancer, somatic, 114500
PTPRO	140.8	99.9	99	Nephrotic syndrome, type 6, 614196
PTPRQ	104.7	93.3	89.1	Deafness, autosomal dominant 73, 617663 Deafness, autosomal recessive 84A, 613391
PTRH2	279.6	100	100	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PTS	107.2	99.6	94.1	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUF60	173.4	99.9	98.3	Verheij syndrome, 615583
PUM1	158	100	99.9	Spinocerebellar ataxia 47, 617931
PURA	121.8	94.5	87.2	Mental retardation, autosomal dominant 31, 616158
PUS1	127.2	98.6	93.9	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PUS3	192	100	100	?Mental retardation, autosomal recessive 55, 617051
PXDN	163.8	99.8	98.5	Anterior segment dysgenesis 7, with sclerocornea, 269400
PYCR1	86.3	99.4	94.3	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
PYCR2	127.6	100	97.6	Leukodystrophy, hypomyelinating, 10, 616420
PYGL	158.6	100	100	Glycogen storage disease VI, 232700
PYGM	127.1	100	99.9	McArdle disease, 232600
PYROXD1	48.4	85.5	70.6	Myopathy, myofibrillar, 8, 617258
QARS	166.6	100	100	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	92.3	100	99.5	Hyperphenylalaninemia, BH4-deficient, C, 261630
QRICH1	165.9	99.9	98.9	Ververi-Brady syndrome, 617982
RAB11B	249.8	100	100	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807
RAB18	82.7	97.1	86.4	Warburg micro syndrome 3, 614222
RAB23	110.3	99.7	98	Carpenter syndrome, 201000
RAB27A	143.9	100	99.9	Griscelli syndrome, type 2, 607624
RAB28	52.1	96.5	87.1	Cone-rod dystrophy 18, 615374
RAB33B	233.9	100	100	Smith-McCort dysplasia 2, 615222
RAB39B	113	100	99.7	?Waisman syndrome, 311510 Mental retardation, X-linked 72, 300271
RAB3GAP1	124.2	99.4	98.8	Warburg micro syndrome 1, 600118
RAB3GAP2	94.1	98.4	93.9	Martsolf syndrome, 212720 Warburg micro syndrome 2, 614225

RAB7A	157.9	100	100	Charcot-Marie-Tooth disease, type 2B, 600882
RAC1	117.9	97.6	92.8	Mental retardation, autosomal dominant 48, 617751
RAC2	104.1	100	99.4	Neutrophil immunodeficiency syndrome, 608203
RAD21	78.5	98.8	94.7	?Mungan syndrome, 611376 Cornelia de Lange syndrome 4, 614701
RAD50	99	92.6	86.2	Nijmegen breakage syndrome-like disorder, 613078
RAD51	123.2	89.4	89.4	?Fanconi anemia, complementation group R, 617244 Mirror movements 2, 614508 {Breast cancer, susceptibility to}, 114480
RAD51C	143.4	100	98.9	Fanconi anemia, complementation group O, 613390 {Breast-ovarian cancer, familial, susceptibility to, 3}, 613399
RAD54B	104.6	99	93.9	Colon cancer, somatic, 114500 Lymphoma, non-Hodgkin, somatic, 605027
RAD54L	123.6	100	99.2	Adenocarcinoma, colonic, somatic, 0 Lymphoma, non-Hodgkin, somatic, 605027 {Breast cancer, invasive ductal}, 114480
RAF1	127.3	100	99.7	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	100	100	Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457
RAI1	146.3	100	99.7	Smith-Magenis syndrome, 182290
RAP1GDS1	96.2	99.3	93.5	Lymphocytic leukemia, acute T-cell, 0
RAPGEF2	157.3	99	97.5	?Epilepsy, familial adult myoclonic, 7, 618075
RAPSN	140.5	99.6	96.3	Fetal akinesia deformation sequence, 208150 Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326
RARB	118.7	100	100	Microphthalmia, syndromic 12, 615524
RARS	86.4	92.7	85.9	Leukodystrophy, hypomyelinating, 9, 616140

RARS2	107.2	100	99.1	Pontocerebellar hypoplasia, type 6, 611523
RASA1	96.9	95.6	84.6	Basal cell carcinoma, somatic, 605462 Capillary malformation-arteriovenous malformation 1, 608354
RASGRP2	97.5	99.9	98.6	?Bleeding disorder, platelet-type, 18, 615888
RAX	82.5	88.7	77.3	Microphthalmia, isolated 3, 611038
RAX2	52.2	91	67.8	?Macular degeneration, age-related, 6, 613757 Cone-rod dystrophy 11, 610381
RB1	88	90.1	76.3	Bladder cancer, somatic, 109800 Osteosarcoma, somatic, 259500 Retinoblastoma, 180200 Retinoblastoma, trilateral, 180200 Small cell cancer of the lung, somatic, 182280
RB1CC1	111.6	96.8	91	Breast cancer, somatic, 114480
RBBP8	110.7	99.6	96.4	Jawad syndrome, 251255 Pancreatic carcinoma, somatic, 0 Seckel syndrome 2, 606744
RBCK1	104.1	99.2	94.9	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RBM10	112.1	99.4	95.4	TARP syndrome, 311900
RBM20	180.9	99.2	96.6	Cardiomyopathy, dilated, 1DD, 613172
RBM28	138.7	100	100	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RBM8A	106.6	100	99.4	Thrombocytopenia-absent radius syndrome, 274000
RBMX	52.9	94	82.1	?Mental retardation, X-linked, syndromic 11, Shashi type, 300238
RBP3	155.2	100	100	?Retinitis pigmentosa 66, 615233
RBP4	99.6	99.4	96.1	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RBPJ	89.2	94.1	86.4	Adams-Oliver syndrome 3, 614814
RCBTB1	123.7	100	99.7	Retinal dystrophy with or without extraocular anomalies, 617175
RD3	161.6	100	99.9	Leber congenital amaurosis 12, 610612
RDH11	119.2	100	99.9	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108

RDH12	94.4	100	98.4	Leber congenital amaurosis 13, 612712
RDH5	160.2	100	99.7	Fundus albipunctatus, 136880
RDX	43.2	84.7	64.8	Deafness, autosomal recessive 24, 611022
RECQL4	149.6	99.2	96.5	Baller-Gerold syndrome, 218600 RAPADILINO syndrome, 266280 Rothmund-Thomson syndrome, 268400
REEP1	78.3	76.3	75.7	?Neuronopathy, distal hereditary motor, type VB, 614751 Spastic paraplegia 31, autosomal dominant, 610250
REEP2	135.1	98.7	96.1	?Spastic paraplegia 72, autosomal dominant, 615625 ?Spastic paraplegia 72, autosomal recessive, 615625
REEP6	171.8	99.4	96.4	Retinitis pigmentosa 77, 617304
RELB	90.6	87.9	75.1	?Immunodeficiency 53, 617585
RELN	155.6	100	99.8	Lissencephaly 2 (Norman-Roberts type), 257320 {Epilepsy, familial temporal lobe, 7}, 616436
REN	148.9	100	100	Hyperuricemic nephropathy, familial juvenile 2, 613092 Renal tubular dysgenesis, 267430 [Hyperproreninemia], 0
REPS1	123	96	93.5	?Neurodegeneration with brain iron accumulation 7, 617916
RERE	71.4	94.9	88.2	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975
REST	128.6	98.5	98.4	Fibromatosis, gingival, 5, 617626 {Wilms tumor 6, susceptibility to}, 616806
RET	141	99.7	97.8	Central hypoventilation syndrome, congenital, 209880 Medullary thyroid carcinoma, 155240 Multiple endocrine neoplasia IIA, 171400 Multiple endocrine neoplasia IIB, 162300 Pheochromocytoma, 171300 {Hirschsprung disease, protection against}, 142623 {Hirschsprung disease, susceptibility to, 1}, 142623
RETREG1	126.1	95.6	90.1	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
RFT1	108.3	99.8	97.3	Congenital disorder of glycosylation, type In, 612015
RFWD3	139.2	99.8	98.9	?Fanconi anemia, complementation group W, 617784

RFX5	116.9	98.7	96.3	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFX6	153.9	100	99.6	Mitchell-Riley syndrome, 615710
RFXANK	105.9	100	100	MHC class II deficiency, complementation group B, 209920
RFXAP	84.8	94.4	91.6	Bare lymphocyte syndrome, type II, complementation group D, 209920
RGR	126.8	100	99.4	Retinitis pigmentosa 44, 613769
RGS9	101.4	98.5	97.2	Bradyopsia, 608415
RGS9BP	93.9	100	99.6	Bradyopsia, 608415
RHAG	151.4	100	99.2	Anemia, hemolytic, Rh-null, regulator type, 268150 Overhydrated hereditary stomatocytosis, 185000
RHBDF2	97.7	99.5	97	Tylosis with esophageal cancer, 148500
RHCE	216.6	97.7	97.2	Rh-null disease, amorph type, 617970 [Blood group, Rhesus], 0
RHO	210.1	100	100	Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 Retinitis punctata albescens, 136880
RHOBTB2	227.5	100	100	Epileptic encephalopathy, early infantile, 64, 618004
RIMS1	126.6	98.8	96	Cone-rod dystrophy 7, 603649
RIN2	113.4	100	99.9	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075
RIPK1	128.9	100	98.7	Immunodeficiency 57, 618108
RIPK4	163.3	100	99.6	CHAND syndrome, 214350 Popliteal pterygium syndrome, Bartsocas-Papas type, 263650
RIPOR2	121.4	100	99.9	?Deafness, autosomal recessive 104, 616515
RIPPLY2	63.5	99	83.8	?Spondylocostal dysostosis 6, 616566
RIT1	165.6	100	100	Noonan syndrome 8, 615355
RLBP1	144.8	100	100	Bothnia retinal dystrophy, 607475 Fundus albipunctatus, 136880 Newfoundland rod-cone dystrophy, 607476 Retinitis punctata albescens, 136880
RLIM	149.6	99.6	97.8	Tonne-Kalscheuer syndrome, 300978
RMND1	137.2	99.8	97.3	Combined oxidative phosphorylation deficiency 11, 614922

RMRP	NC	NC	NC	Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RNASEH1	98.6	99.1	95.6	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479
RNASEH2A	142.1	100	99.9	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	103.8	93.2	87.5	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	209.2	100	99.9	Aicardi-Goutieres syndrome 3, 610329
RNASEL	147.9	100	99.6	Prostate cancer 1, 601518
RNASET2	96.4	91.9	88.3	Leukoencephalopathy, cystic, without megalencephaly, 612951
RNF113A	144.8	100	100	?Trichothiodystrophy 5, nonphotosensitive, 300953
RNF125	177.8	100	99.1	Tenorio syndrome, 616260
RNF135	96	94	91.6	Macrocephaly, macrosomia, facial dysmorphism syndrome, 614192
RNF139	223.5	100	100	Renal cell carcinoma, 144700
RNF168	215.3	100	99.1	RIDDLE syndrome, 611943
RNF170	147.1	98.3	91.2	Ataxia, sensory, 1, autosomal dominant, 608984
RNF212	118.7	99.1	96.9	Recombination rate QTL 1, 612042
RNF216	137.1	99.8	98.6	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840
RNF43	126.5	100	99.6	Sessile serrated polyposis cancer syndrome, 617108
RNF6	195.4	100	99.4	Esophageal carcinoma, somatic, 133239
RNPC3	41.3	85.1	61.1	?Growth hormone deficiency, isolated, type V, 618160
RNU4ATAC	NC	NC	NC	Microcephalic osteodysplastic primordial dwarfism, type I, 210710 Roifman syndrome, 616651
ROBO2	155.2	97.9	96.7	Vesicoureteral reflux 2, 610878
ROBO3	99.7	98.7	95.6	Gaze palsy, familial horizontal, with progressive scoliosis, 1, 607313
ROGDI	112.2	97.9	95.3	Kohlschutter-Tonz syndrome, 226750
ROM1	115	100	99.4	Retinitis pigmentosa 7, digenic form, 608133
ROR1	177.5	96.8	96.8	?Deafness, autosomal recessive 108, 617654
ROR2	165.9	99.4	98	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310

RORA	129.8	92.1	87.7	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060
RORC	132.3	100	100	Immunodeficiency 42, 616622
RP1	121.8	91.4	90.3	Retinitis pigmentosa 1, 180100
RP1L1	94.8	100	99.8	Occult macular dystrophy, 613587
RP2	180	100	98.9	Retinitis pigmentosa 2, 312600
RP9	62.9	77.8	76	?Retinitis pigmentosa 9, 180104
RPE65	130.3	100	99.3	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794
RPGR	91.8	83.1	73.8	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	154.2	100	99.9	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826
RPGRIP1L	126.2	96.4	93.9	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561
RPIA	113.1	94.3	90.9	?Ribose 5-phosphate isomerase deficiency, 608611
RPL10	85.9	99.1	93.1	Mental retardation, X-linked, syndromic, 35, 300998 {Autism, susceptibility to, X-linked 5}, 300847
RPL11	99.8	100	99.5	Diamond-Blackfan anemia 7, 612562
RPL15	41.6	88.3	81.2	?Diamond-Blackfan anemia 12, 615550
RPL21	64.7	79	57.4	Hypotrichosis 12, 615885
RPL26	46.8	94.8	83.7	?Diamond-Blackfan anemia 11, 614900
RPL27	39.3	80.4	58.3	?Diamond-Blackfan anemia 16, 617408
RPL35A	83.9	99.2	91	Diamond-Blackfan anemia 5, 612528
RPL5	43.8	81.8	69.3	Diamond-Blackfan anemia 6, 612561
RPS10	140.1	99.8	97.1	Diamond-Blackfan anemia 9, 613308
RPS14	133.3	99.7	96.2	Macrocytic anemia, refractory, due to 5q deletion, somatic, 153550
RPS17	52.4	85	73.7	Diamond-Blackfan anemia 4, 612527
RPS19	82.5	99.7	95.5	Diamond-Blackfan anemia 1, 105650
RPS23	63.2	90.3	82.2	Brachycephaly, trichomegaly, and developmental delay, 617412
RPS24	110.4	92.4	87.2	Diamond-blackfan anemia 3, 610629

RPS26	106.8	94.8	82.1	Diamond-Blackfan anemia 10, 613309
RPS27	39.7	86.7	60.3	?Diamond-Blackfan anemia 17, 617409
RPS28	47.7	100	92	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164
RPS29	106.2	97.9	96.6	Diamond-Blackfan anemia 13, 615909
RPS6KA3	79.3	94.2	83.3	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844
RPS7	93.7	76.9	63.4	Diamond-Blackfan anemia 8, 612563
RPSA	88.8	100	99.7	Asplenia, isolated congenital, 271400
RRAS2	73.4	86.9	70.3	Ovarian carcinoma, 0
RRM2B	128.6	99.7	97.5	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	97.8	88.3	Retinoschisis, 312700
RSPH1	146.1	100	100	Ciliary dyskinesia, primary, 24, 615481
RSPH3	132.5	99.7	97.5	Ciliary dyskinesia, primary, 32, 616481
RSPH4A	139	98.3	96.5	Ciliary dyskinesia, primary, 11, 612649
RSPH9	127.4	100	99.6	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
RSPO2	127.1	98.9	95.2	?Humerofemoral hypoplasia with radiotibial ray deficiency, 618022 Tetraamelia syndrome 2, 618021
RSPO4	107.2	100	100	Anonychia congenita, 206800
RSPRY1	168.9	100	99.9	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723
RTEL1	110.9	99.2	95.1	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	99.2	96.7	Spastic paraplegia 12, autosomal dominant, 604805
RTN4IP1	98.1	99.9	99.1	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732

RTTN	129.5	97.2	94.7	Microcephaly, short stature, and polymicrogyria with seizures, 614833
RUBCN	104.1	98	97.5	?Spinocerebellar ataxia, autosomal recessive 15, 615705
RUNX1	92	97.2	89.7	Leukemia, acute myeloid, 601626 Platelet disorder, familial, with associated myeloid malignancy, 601399
RUNX2	106.4	72.3	72.2	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
RUSC2	182.3	100	99.9	Mental retardation, autosomal recessive 61, 617773
RYR1	120.7	96.8	93.7	Central core disease, 117000 King-Denborough syndrome, 145600 Minicore myopathy with external ophthalmoplegia, 255320 Neuromuscular disease, congenital, with uniform type 1 fiber, 117000 {Malignant hyperthermia susceptibility 1}, 145600
RYR2	142.2	99.7	98.4	Arrhythmogenic right ventricular dysplasia 2, 600996 Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772
S1PR2	278	97.4	92.7	Deafness, autosomal recessive 68, 610419
SACS	154.5	100	99.7	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAG	131.3	100	99.9	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758
SALL1	138.5	99.3	98.4	Townes-Brocks branchiootorenal-like syndrome, 107480 Townes-Brocks syndrome 1, 107480
SALL2	138.9	100	100	?Coloboma, ocular, autosomal recessive, 216820
SALL4	147.5	97.6	96.3	Duane-radial ray syndrome, 607323 IVIC syndrome, 147750
SAMD12	161.6	100	99.9	Epilepsy, familial adult myoclonic, 1, 601068
SAMD9	159.1	99.9	99.3	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455
SAMD9L	165.7	100	99.9	Ataxia-pancytopenia syndrome, 159550
SAMHD1	127.9	99.6	96.6	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SAR1B	125	89.8	88.9	Chylomicron retention disease, 246700

SARS	113.7	100	99.7	?Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709
SARS2	104.8	94.8	92.7	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SASS6	62.9	97.6	84.1	?Microcephaly 14, primary, autosomal recessive, 616402
SATB2	110.5	98.5	93.4	Glass syndrome, 612313
SBDS	212.3	100	99.9	Shwachman-Diamond syndrome, 260400 {Aplastic anemia, susceptibility to}, 609135
SBF1	107.9	98.5	96.5	Charcot-Marie-Tooth disease, type 4B3, 615284
SBF2	117	99.6	96.8	Charcot-Marie-Tooth disease, type 4B2, 604563
SC5D	198.4	100	99.2	Lathosterolosis, 607330
SCAPER	135.9	96	93.6	Intellectual developmental disorder and retinitis pigmentosa, 618195
SCARB2	121.2	100	99.9	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCARF2	82.5	85.2	73.6	Van den Ende-Gupta syndrome, 600920
SCN10A	165.3	100	99.5	Episodic pain syndrome, familial, 2, 615551
SCN11A	138.1	99.2	97.6	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN1A	135.2	99.6	98	Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Epileptic encephalopathy, early infantile, 6 (Dravet syndrome), 607208 Febrile seizures, familial, 3A, 604403 Migraine, familial hemiplegic, 3, 609634
SCN1B	168.3	97.1	96.1	Atrial fibrillation, familial, 13, 615377 Brugada syndrome 5, 612838 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233 Epileptic encephalopathy, early infantile, 52, 617350
SCN2A	156.7	99	96.4	Epileptic encephalopathy, early infantile, 11, 613721 Seizures, benign familial infantile, 3, 607745
SCN2B	185.8	100	100	Atrial fibrillation, familial, 14, 615378
SCN3A	166	99.3	97.5	Epilepsy, familial focal, with variable foci 4, 617935 Epileptic encephalopathy, early infantile, 62, 617938
SCN3B	147.3	100	100	Atrial fibrillation, familial, 16, 613120 Brugada syndrome 7, 613120

SCN4A	214	99.9	99.5	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	77.5	100	97.9	Atrial fibrillation, familial, 17, 611819 Long QT syndrome-10, 611819
SCN5A	169.4	99	99	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	198.3	100	99.7	?Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy, early infantile, 13, 614558 Seizures, benign familial infantile, 5, 617080
SCN9A	146.5	98.5	97	Epilepsy, generalized, with febrile seizures plus, type 7, 613863 Erythermalgia, primary, 133020 Febrile seizures, familial, 3B, 613863 HSAN2D, autosomal recessive, 243000 Insensitivity to pain, congenital, 243000 Paroxysmal extreme pain disorder,, 167400 Small fiber neuropathy, 133020 {Dravet syndrome, modifier of}, 607208
SCNN1A	134.3	99	96.3	?Liddle syndrome 3, 618126 Bronchiectasis with or without elevated sweat chloride 2, 613021 Pseudohypoaldosteronism, type I, 264350
SCNN1B	148.9	100	99.8	Bronchiectasis with or without elevated sweat chloride 1, 211400 Liddle syndrome 1, 177200 Pseudohypoaldosteronism, type I, 264350

SCNN1G	139.4	99.7	97.1	Bronchiectasis with or without elevated sweat chloride 3, 613071 Liddle syndrome, 618114 Pseudohypoaldosteronism, type I, 264350
SCO1	109.6	97.9	94.3	Mitochondrial complex IV deficiency, 220110
SCO2	113.1	100	100	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908
SCP2	106.9	99.6	96.5	?Leukoencephalopathy with dystonia and motor neuropathy, 613724
SCYL1	143.2	98.6	96.3	Spinocerebellar ataxia, autosomal recessive 21, 616719
SDCCAG8	123.9	99.8	97.4	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SDHA	122.2	84.8	80.8	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165
SDHAF1	42.5	100	96.2	Mitochondrial complex II deficiency, 252011
SDHAF2	144.3	94.7	94.3	Paragangliomas 2, 601650
SDHB	120.3	100	99.3	Gastrointestinal stromal tumor, 606444 Paragangliolas 4, 115310 Paraganglioma and gastric stromal sarcoma, 606864 Pheochromocytoma, 171300
SDHC	100.1	99.8	96.8	Gastrointestinal stromal tumor, 606764 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 3, 605373
SDHD	48.4	55.2	50.4	Mitochondrial complex II deficiency, 252011 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 1, with or without deafness, 168000 Pheochromocytoma, 171300
SDR9C7	198.3	100	100	Ichthyosis, congenital, autosomal recessive 13, 617574
SEC23A	121.8	98	94.6	Craniolenticulosutural dysplasia, 607812
SEC23B	161.1	97.5	96.4	?Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SEC24D	136.9	99.9	98.6	Cole-Carpenter syndrome 2, 616294
SEC61A1	132.9	100	100	Hyperuricemic nephropathy, familial juvenile, 4, 617056

SEC63	60.8	85.4	75.7	Polycystic liver disease 2, 617004
SECISBP2	122.5	98.3	95.3	Thyroid hormone metabolism, abnormal, 609698
SELENBP1	140.5	100	100	Extraoral halitosis due to MTO deficiency, 618148
SELENON	111.7	85.2	83.3	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310
SEMA3E	142.6	99.9	99	?CHARGE syndrome, 214800
SEMA4A	127.5	99.9	98.9	Cone-rod dystrophy 10, 610283 Retinitis pigmentosa 35, 610282
SEPSECS	159.3	100	100	Pontocerebellar hypoplasia type 2D, 613811
SEPT12	105	97.4	93.8	Spermatogenic failure 10, 614822
SEPT9	118.7	99.7	96.8	Amyotrophy, hereditary neuralgic, 162100 Leukemia, acute myeloid, therapy-related, 0 Ovarian carcinoma, 0
SERAC1	112.5	98.8	94.6	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPINA1	141.1	100	99.9	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, 0 Cerebrovascular disease, occlusive, 0
SERPINA6	190.9	100	100	Corticosteroid-binding globulin deficiency, 611489
SERPINB6	164.1	95.9	95.9	?Deafness, autosomal recessive 91, 613453
SERPINB7	127.4	100	99.5	Palmoplantar keratoderma, Nagashima type, 615598
SERPINB8	151.6	95	95	Peeling skin syndrome 5, 617115
SERPINC1	143.1	100	100	Thrombophilia due to antithrombin III deficiency, 613118
SERPIND1	181.8	100	100	Thrombophilia due to heparin cofactor II deficiency, 612356
SERPINE1	155	100	100	Plasminogen activator inhibitor-1 deficiency, 613329 {Transcription of plasminogen activator inhibitor, modulator of}, 0
SERPINF1	101.8	100	99.4	Osteogenesis imperfecta, type VI, 613982
SERPINF2	143.8	99.9	99.3	Alpha-2-plasmin inhibitor deficiency, 262850
SERPING1	97.9	97.3	92.6	Angioedema, hereditary, types I and II, 106100 Complement component 4, partial deficiency of, 120790

SERPINH1	183.7	100	99.9	Osteogenesis imperfecta, type X, 613848 {Preterm premature rupture of the membranes, susceptibility to}, 610504
SERPINI1	105.2	99.9	96.9	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218
SET	55.4	89.9	79.4	Mental retardation, autosomal dominant 58, 618106
SETBP1	151.7	97.6	96.1	Mental retardation, autosomal dominant 29, 616078 Schinzel-Giedion midface retraction syndrome, 269150
SETD2	147.8	100	99.6	Luscan-Lumish syndrome, 616831
SETD5	184.5	100	99.7	Mental retardation, autosomal dominant 23, 615761
SETX	163.2	99.9	99.1	Amyotrophic lateral sclerosis 4, juvenile, 602433 Spinocerebellar ataxia, autosomal recessive 1, 606002
SF3B1	128.7	99.3	97.5	Myelodysplastic syndrome, somatic, 614286
SF3B4	89.5	99.8	97.7	Acrofacial dysostosis 1, Nager type, 154400
SFRP4	164.1	100	98.5	Pyle disease, 265900
SFTPA2	167.9	100	99.1	Pulmonary fibrosis, idiopathic, 178500
SFTPB	99.4	99.9	98.7	Surfactant metabolism dysfunction, pulmonary, 1, 265120
SFTPC	102.6	99.8	95.6	Surfactant metabolism dysfunction, pulmonary, 2, 610913
SFXN4	131.7	100	99.1	Combined oxidative phosphorylation deficiency 18, 615578
SGCA	147.3	100	99.7	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099
SGCB	154.2	96.6	94.2	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286
SGCD	94.8	100	99.4	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287
SGCE	88.9	93.7	90	Dystonia-11, myoclonic, 159900
SGCG	138.7	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700
SGO1	94.8	98	94.3	Chronic atrial and intestinal dysrhythmia, 616201
SGPL1	164.1	100	100	Nephrotic syndrome, type 14, 617575
SGSH	129	95.1	93.6	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SH2B3	97.5	90.7	79	Erythrocytosis, somatic, 133100 Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
SH2D1A	104.7	89.9	89.4	Lymphoproliferative syndrome, X-linked, 1, 308240
SH3BP2	110.9	91.4	91.4	Cherubism, 118400

SH3PXD2B	140.2	100	99.8	Frank-ter Haar syndrome, 249420
SH3TC2	121.3	100	99.7	Charcot-Marie-Tooth disease, type 4C, 601596 Mononeuropathy of the median nerve, mild, 613353
SHANK3	84.8	81.2	73.5	Phelan-McDermid syndrome, 606232 {Schizophrenia 15}, 613950
SHH	117.5	99	94	Holoprosencephaly 3, 142945 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250
SHOC2	140.4	100	99.4	Noonan-like syndrome with loose anagen hair, 607721
SHOX	29.1	73.5	61.4	Langer mesomelic dysplasia, 249700 Leri-Weill dyschondrosteosis, 127300 Short stature, idiopathic familial, 300582
SHROOM4	100.8	99.8	98.2	Stocco dos Santos X-linked mental retardation syndrome, 300434
SI	98.1	96.2	87.1	Sucrase-isomaltase deficiency, congenital, 222900
SIGMAR1	148.5	100	100	?Amyotrophic lateral sclerosis 16, juvenile, 614373 ?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726
SIK1	85.6	97	92.4	Epileptic encephalopathy, early infantile, 30, 616341
SIK3	117.2	98	96.8	?Spondyloepimetaphyseal dysplasia, Krakow type, 618162
SIL1	154.4	99.8	98	Marinesco-Sjogren syndrome, 248800
SIM1	151.4	100	99.8	Obesity, severe, 601665
SIN3A	137.9	100	99.4	Witteveen-Kolk syndrome, 613406
SIPA1L3	140.8	99.7	98.6	?Cataract 45, 616851
SIX1	117.3	99.7	97.6	Branchiootic syndrome 3, 608389 Deafness, autosomal dominant 23, 605192
SIX3	145.3	100	98.9	Holoprosencephaly 2, 157170 Schizencephaly, 269160
SIX5	43.8	88.3	76.1	Branchiootorenal syndrome 2, 610896
SIX6	228.6	100	100	Optic disc anomalies with retinal and/or macular dystrophy, 212550
SKI	85.3	96.4	90.8	Shprintzen-Goldberg syndrome, 182212
SKIV2L	149.1	100	99.8	Trichohepatoenteric syndrome 2, 614602
SLC10A2	151	100	100	Bile acid malabsorption, primary, 613291
SLC11A2	135.2	100	99.3	Anemia, hypochromic microcytic, with iron overload 1, 206100

SLC12A1	172.7	99.8	99.1	Bartter syndrome, type 1, 601678
SLC12A3	139.3	100	100	Gitelman syndrome, 263800
SLC12A5	125.9	85.4	81.6	Epileptic encephalopathy, early infantile, 34, 616645 {Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685
SLC12A6	141.8	100	99.9	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC13A5	164.1	100	100	Epileptic encephalopathy, early infantile, 25, 615905
SLC16A1	157.1	99.9	98.8	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia, familial, 7, 610021 Monocarboxylate transporter 1 deficiency, 616095
SLC16A12	164.4	100	99.9	Cataract 47, juvenile, with microcornea, 612018
SLC16A2	60.3	92.8	82.1	Allan-Herndon-Dudley syndrome, 300523
SLC17A5	119.6	96.8	92.9	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC17A8	132.7	100	100	Deafness, autosomal dominant 25, 605583
SLC17A9	111.5	95.6	95.6	Porokeratosis 8, disseminated superficial actinic type, 616063
SLC18A2	126.5	100	99.2	?Parkinsonism-dystonia, infantile, 2, 618049
SLC18A3	276.6	100	100	Myasthenic syndrome, congenital, 21, presynaptic, 617239
SLC19A2	119.5	99.8	97.8	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	186.4	100	99.9	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A1	174.2	100	99.9	Dicarboxylic aminoaciduria, 222730 {?Schizophrenia susceptibility 18}, 615232
SLC1A2	128	99.9	99.4	Epileptic encephalopathy, early infantile, 41, 617105
SLC1A3	121.9	100	99.8	Episodic ataxia, type 6, 612656
SLC1A4	156.5	98.9	94.9	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC20A2	119	99.7	97.3	Basal ganglia calcification, idiopathic, 1, 213600
SLC22A12	105.7	100	99.7	Hypouricemia, renal, 220150

SLC22A18	85.1	92.8	81.7	Breast cancer, somatic, 114480 Lung cancer, somatic, 211980 Rhabdomyosarcoma, somatic, 268210
SLC22A5	153.3	100	100	Carnitine deficiency, systemic primary, 212140
SLC24A1	218.5	100	100	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
SLC24A4	126.8	99.6	97.3	Amelogenesis imperfecta, type IIA5, 615887 [Skin/hair/eye pigmentation 6, blond/brown hair], 210750 [Skin/hair/eye pigmentation 6, blue/green eyes], 210750
SLC24A5	114.5	99.6	97.7	Albinism, oculocutaneous, type VI, 113750 [Skin/hair/eye pigmentation 4, fair/dark skin], 113750
SLC25A1	71	92.2	87	?Myasthenic syndrome, congenital, 23, presynaptic, 618197 Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A12	150.5	99.8	98.4	Epileptic encephalopathy, early infantile, 39, 612949
SLC25A13	110.7	95.7	92.3	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A15	192.5	98.8	95	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A19	88.6	99.9	98.3	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A20	110.3	100	99.7	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A22	108.7	99.5	96.9	Epileptic encephalopathy, early infantile, 3, 609304
SLC25A24	115.8	98.6	96.3	Fontaine progeroid syndrome, 612289
SLC25A26	98	98.9	96	Combined oxidative phosphorylation deficiency 28, 616794
SLC25A3	139	99.8	97.6	Mitochondrial phosphate carrier deficiency, 610773
SLC25A32	117	100	100	?Exercise intolerance, riboflavin-responsive, 616839
SLC25A38	111.4	99.8	98.1	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC25A4	134.1	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	205.7	95.9	87.3	Neuropathy, hereditary motor and sensory, type VIB, 616505
SLC26A1	139.8	100	99.9	?Nephrolithiasis, calcium oxalate, 167030

SLC26A2	233.2	100	100	Achondrogenesis Ib, 600972 Atelosteogenesis, type II, 256050 De la Chapelle dysplasia, 256050 Diastrophic dysplasia, 222600 Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 Epiphyseal dysplasia, multiple, 4, 226900
SLC26A3	156.1	99.9	98.9	Diarrhea 1, secretory chloride, congenital, 214700
SLC26A4	123.3	99.9	99.1	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791 Pendred syndrome, 274600
SLC26A5	150.9	98.7	95.8	?Deafness, autosomal recessive 61, 613865
SLC26A8	138.9	100	99.3	Spermatogenic failure 3, 606766
SLC27A4	155.8	99.6	97.9	Ichthyosis prematurity syndrome, 608649
SLC29A3	203.6	99.9	99.5	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC2A1	190.1	92.9	92.8	Dystonia 9, 601042 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 GLUT1 deficiency syndrome 2, childhood onset, 612126 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847
SLC2A10	166.4	97.7	97.6	Arterial tortuosity syndrome, 208050
SLC2A2	178.4	100	99.9	Fanconi-Bickel syndrome, 227810 {Diabetes mellitus, noninsulin-dependent}, 125853
SLC2A9	119	99.2	96.2	Hypouricemia, renal, 2, 612076 {Uric acid concentration, serum, QTL 2}, 612076
SLC30A10	164.4	100	100	Hypermanganesemia with dystonia 1, 613280
SLC30A2	146.1	100	99.6	Zinc deficiency, transient neonatal, 608118
SLC30A9	81.2	96	88.3	?Birk-Landau-Perez syndrome, 617595
SLC33A1	140.9	96.8	90.1	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539
SLC34A1	153.2	100	99.5	?Fanconi renotubular syndrome 2, 613388 Hypercalcemia, infantile, 2, 616963 Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286
SLC34A2	169.1	100	100	Pulmonary alveolar microlithiasis, 265100
SLC34A3	105.6	98.9	94.5	Hypophosphatemic rickets with hypercalciuria, 241530
SLC35A1	124	99.9	97.7	Congenital disorder of glycosylation, type IIf, 603585

SLC35A2	108.8	99.7	96.8	Congenital disorder of glycosylation, type IIm, 300896
SLC35A3	50.2	78.1	71.9	?Arthrogryposis, mental retardation, and seizures, 615553
SLC35C1	230.2	99.9	98.4	Congenital disorder of glycosylation, type IIc, 266265
SLC35D1	115.4	95.7	90.4	Schneckenbecken dysplasia, 269250
SLC36A2	123.9	100	100	Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC37A4	140.2	100	99.9	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240
SLC38A8	76.4	99.4	95.8	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218
SLC39A13	114.8	99.8	98	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350
SLC39A14	107.7	99.8	97.9	?Hyperostosis cranalis interna, 144755 Hypermanganesemia with dystonia 2, 617013
SLC39A4	81.8	99.2	96	Acrodermatitis enteropathica, 201100
SLC39A5	119.3	100	99.2	Myopia 24, autosomal dominant, 615946
SLC39A8	128.5	100	99.7	Congenital disorder of glycosylation, type IIn, 616721
SLC3A1	162.8	100	99.5	Cystinuria, 220100
SLC40A1	155.7	99.9	99.4	Hemochromatosis, type 4, 606069
SLC44A4	122.9	100	99.9	?Deafness, autosomal dominant 72, 617606
SLC45A1	120.7	100	99.2	Intellectual developmental disorder with neuropsychiatric features, 617532
SLC45A2	148.1	100	99.9	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	99.4	96.4	Folate malabsorption, hereditary, 229050
SLC4A1	140	100	99.9	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	100	99.7	Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy and perceptive deafness, 217400 Corneal endothelial dystrophy, autosomal recessive, 217700
SLC4A4	122.3	99	97.1	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC52A1	219.3	100	100	Riboflavin deficiency, 615026
SLC52A2	177.6	100	100	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	119.6	100	100	?Fazio-Londe disease, 211500 Brown-Vialetto-Van Laere syndrome 1, 211530
SLC5A1	140	100	100	Glucose/galactose malabsorption, 606824
SLC5A2	118.7	100	100	Renal glucosuria, 233100
SLC5A5	93.9	99.7	96.8	Thyroid dyshormonogenesis 1, 274400
SLC5A7	117.1	100	99.9	Myasthenic syndrome, congenital, 20, presynaptic, 617143 Neuronopathy, distal hereditary motor, type VIIA, 158580
SLC6A1	143.6	100	100	Myoclonic-atonic epilepsy, 616421
SLC6A17	189.8	100	100	Mental retardation, autosomal recessive 48, 616269
SLC6A19	149.1	100	99.3	Hartnup disorder, 234500 Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC6A2	149.9	100	99.8	?Orthostatic intolerance, 604715
SLC6A20	178.6	100	99.9	Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC6A3	145.7	100	99.8	Parkinsonism-dystonia, infantile, 1, 613135 {Nicotine dependence, protection against}, 188890

SLC6A5	136.5	100	99.7	Hyperekplexia 3, 614618
SLC6A8	56.5	89.8	79.1	Cerebral creatine deficiency syndrome 1, 300352
SLC6A9	161.2	100	99.4	Glycine encephalopathy with normal serum glycine, 617301
SLC7A14	191.9	100	100	Retinitis pigmentosa 68, 615725
SLC7A7	123.9	100	99.9	Lysinuric protein intolerance, 222700
SLC7A9	125.5	99.9	99	Cystinuria, 220100
SLC9A1	160.9	100	100	?Lichtenstein-Knorr syndrome, 616291
SLC9A3	147.1	98.7	96.9	Diarrhea 8, secretory sodium, congenital, 616868
SLC9A3R1	111.5	99.5	96	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
SLC9A6	104.2	97.6	91.3	Mental retardation, X-linked syndromic, Christianson type, 300243
SLCO1B1	46	92.8	84.4	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO1B3	48.9	94.9	79.5	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO2A1	110.5	100	99.6	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
SLFN14	193.1	100	100	Bleeding disorder, platelet-type, 20, 616913
SLITRK1	132.6	100	100	?Trichotillomania, 613229 Tourette syndrome, 137580
SLITRK6	206.9	100	100	Deafness and myopia, 221200
SLURP1	97.1	99.8	96.2	Meleda disease, 248300
SLX4	114.2	100	99.8	Fanconi anemia, complementation group P, 613951
SMAD3	131.7	99.9	99.2	Loeys-Dietz syndrome 3, 613795
SMAD4	125.5	100	100	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Polyposis, juvenile intestinal, 174900
SMAD6	100.5	80	72	Aortic valve disease 2, 614823 {Craniosynostosis 7, susceptibility to}, 617439
SMAD9	132.8	100	100	Pulmonary hypertension, primary, 2, 615342
SMARCA2	113.8	95.7	93.7	Nicolaides-Baraitser syndrome, 601358
SMARCA4	143.8	100	99.5	Coffin-Siris syndrome 4, 614609 {Rhabdoid tumor predisposition syndrome 2}, 613325

SMARCAD1	85.5	99.6	96.1	Adermatoglyphia, 136000 Basan syndrome, 129200
SMARCAL1	134.6	100	99.9	Schimke immunoosseous dysplasia, 242900
SMARCB1	214.3	100	100	Coffin-Siris syndrome 3, 614608 Rhabdoid tumors, somatic, 609322 {Rhabdoid tumor predisposition syndrome 1}, 609322 {Schwannomatosis-1, susceptibility to}, 162091
SMARCD2	92.9	87	85.7	Specific granule deficiency 2, 617475
SMARCE1	73.6	96.5	86.8	Coffin-Siris syndrome 5, 616938 {Meningioma, familial, susceptibility to}, 607174
SMC1A	99.4	99.9	98.8	Cornelia de Lange syndrome 2, 300590
SMC3	81.4	93.8	87.6	Cornelia de Lange syndrome 3, 610759
SMCHD1	91.3	98.1	92.3	Bosma arhinia microphthalmia syndrome, 603457 Fascioscapulohumeral muscular dystrophy 2, digenic, 158901
SMG9	101.8	100	99.9	Heart and brain malformation syndrome, 616920
SMN1	112.7	99.8	96.5	Spinal muscular atrophy-1, 253300 Spinal muscular atrophy-2, 253550 Spinal muscular atrophy-3, 253400 Spinal muscular atrophy-4, 271150
SMO	149.2	96.5	93.4	Basal cell carcinoma, somatic, 605462 Curry-Jones syndrome, somatic mosaic, 601707
SMOC1	129.8	99.5	97.3	Microphthalmia with limb anomalies, 206920
SMOC2	91.5	75.4	72.6	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SMPD1	123.5	99.6	97.9	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SMPX	68.5	99.9	95.5	Deafness, X-linked 4, 300066
SMS	67.8	88.3	73.9	Mental retardation, X-linked, Snyder-Robinson type, 309583
SNAI2	129.8	100	99.8	Piebaldism, 172800 Waardenburg syndrome, type 2D, 608890
SNAP25	133.7	100	99.9	?Myasthenic syndrome, congenital, 18, 616330
SNAP29	153.5	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	79.3	100	99.9	Dementia, Lewy body, 127750
SNIP1	139.3	99.2	96.7	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501
SNORD118	NC	NC	NC	Leukoencephalopathy, brain calcifications, and cysts, 614561
SNRNP200	161.1	100	99.6	Retinitis pigmentosa 33, 610359
SNRPB	75.2	99.8	97.4	Cerebrocostomandibular syndrome, 117650
SNRPE	79	98.5	89.1	Hypotrichosis 11, 615059
SNRPN	116.8	99.4	95	Prader-Willi syndrome, 176270
SNTA1	97	82.3	77.3	Long QT syndrome 12, 612955
SNX10	118.9	96.2	96.1	Osteopetrosis, autosomal recessive 8, 615085
SNX14	70.1	95.2	82.9	Spinocerebellar ataxia, autosomal recessive 20, 616354
SOBP	130.7	92.9	85.4	Mental retardation, anterior maxillary protrusion, and strabismus, 613671
SOD1	161.9	100	100	Amyotrophic lateral sclerosis 1, 105400
SOHLH1	87	97.9	90	Ovarian dysgenesis 5, 617690 Spermatogenic failure 32, 618115
SON	159.7	98.4	94.4	ZTTK syndrome, 617140
SOS1	94.3	96.7	90.3	?Fibromatosis, gingival, 1, 135300 Noonan syndrome 4, 610733
SOS2	97.1	98.5	92.8	Noonan syndrome 9, 616559
SOST	112.6	100	99.6	Craniodiaphyseal dysplasia, autosomal dominant, 122860 Sclerosteosis 1, 269500 Van Buchem disease, 239100
SOX10	65.8	98.2	91.3	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266
SOX11	118.2	99.7	96.8	Mental retardation, autosomal dominant 27, 615866
SOX17	70.8	99.6	94.6	Vesicoureteral reflux 3, 613674
SOX18	21.2	62.6	48.8	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823 Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940

SOX2	128.8	98.3	93.1	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SOX3	37.7	86.4	71.5	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000
SOX5	107.3	99.1	96.2	Lamb-Shaffer syndrome, 616803
SOX9	134	97.8	93.8	Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290 Campomelic dysplasia with autosomal sex reversal, 114290
SP110	121.6	100	99.5	Hepatic venoocclusive disease with immunodeficiency, 235550 {Mycobacterium tuberculosis, susceptibility to}, 607948
SP7	159.1	99.9	99.3	Osteogenesis imperfecta, type XII, 613849
SPAG1	87.3	96.3	88.2	Ciliary dyskinesia, primary, 28, 615505
SPARC	161	100	100	Osteogenesis imperfecta, type XVII, 616507
SPART	132.4	99.8	98.2	Troyer syndrome, 275900
SPAST	63.8	93.1	81.9	Spastic paraplegia 4, autosomal dominant, 182601
SPATA16	142.2	99.7	96.6	?Spermatogenic failure 6, 102530
SPATA5	132	99.9	99.2	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPATA7	119.6	97.8	90.8	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, autosomal recessive, 604232
SPECC1L	157.9	100	100	?Facial clefting, oblique, 1, 600251 Hypertelorism, Teebi type, 145420 Opitz GBBB syndrome, type II, 145410
SPEG	100.9	93.3	86.5	Centronuclear myopathy 5, 615959
SPG11	129.2	99.2	96.9	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360
SPG21	121.4	98.6	94.8	Mast syndrome, 248900
SPG7	119.2	93.3	92.4	Spastic paraplegia 7, autosomal recessive, 607259
SPINK1	80.2	100	99.4	Pancreatitis, hereditary, 167800 Tropical calcific pancreatitis, 608189 {Fibrocalculous pancreatic diabetes, susceptibility to}, 608189
SPINK2	83.4	99.1	96.8	?Spermatogenic failure 29, 618091
SPINK5	145	99.4	96.5	Netherton syndrome, 256500

SPINT2	71.5	97.8	84.2	Diarrhea 3, secretory sodium, congenital, syndromic, 270420
SPR	166.5	98.9	90	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRED1	164.3	98.7	96.7	Legius syndrome, 611431
SPRTN	167.8	100	100	Ruijs-Aalfs syndrome, 616200
SPRY4	138.7	100	100	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
SPTA1	119.4	99.8	98.9	Elliptocytosis-2, 130600 Pyropoikilocytosis, 266140 Spherocytosis, type 3, 270970
SPTAN1	125.5	99.1	98.6	Epileptic encephalopathy, early infantile, 5, 613477
SPTB	148	100	99.9	Anemia, neonatal hemolytic, fatal or near-fatal, 617948 Elliptocytosis-3, 617948 Spherocytosis, type 2, 616649
SPTBN2	118	99.9	99.3	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia, autosomal recessive 14, 615386
SPTBN4	83.9	96.6	89.1	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519
SPTLC1	115.5	99	93.9	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	160.2	100	100	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SQSTM1	109.1	98.6	94.5	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 Myopathy, distal, with rimmed vacuoles, 617158 Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 Paget disease of bone 3, 167250
SRC	105.2	99.8	97.3	?Thrombocytopenia 6, 616937 Colon cancer, advanced, somatic, 114500
SRCAP	153.9	99.8	99.1	Floating-Harbor syndrome, 136140
SRD5A2	77.6	100	96.4	Pseudovaginal perineoscrotal hypospadias, 264600
SRD5A3	135.9	100	99.7	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713
SRP72	70	93.1	84	Bone marrow failure syndrome 1, 614675
SRPX2	81.8	100	98.5	?Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643
SRY	46.1	50	50	46XX sex reversal 1, 400045 46XY sex reversal 1, 400044

SSR4	89.8	100	98.8	Congenital disorder of glycosylation, type Iy, 300934
SSTR5	155.8	100	99.2	Somatostatin analog, resistance to, 0
SSX1	103.4	80.8	76.5	?Sarcoma, synovial, 300813
SSX2	69.8	63	57.8	?Sarcoma, synovial, 300813
ST14	154	98	97	Ichthyosis, congenital, autosomal recessive 11, 602400
ST3GAL3	144.5	100	99.9	?Epileptic encephalopathy, early infantile, 15, 615006 Mental retardation, autosomal recessive 12, 611090
ST3GAL5	121.9	84.4	84.2	Salt and pepper developmental regression syndrome, 609056
STAC3	120	100	99.9	Myopathy, congenital, Baily-Bloch, 255995
STAG1	104.4	98.8	95.5	Mental retardation, autosomal dominant 47, 617635
STAG3	112.3	93.5	92.8	Premature ovarian failure 8, 615723
STAMBP	112.3	99.3	96.5	Microcephaly-capillary malformation syndrome, 614261
STAR	124	100	100	Lipoid adrenal hyperplasia, 201710
STAT1	126.2	98	95.8	Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796 Immunodeficiency 31C, autosomal dominant, 614162
STAT2	116	100	99.9	Immunodeficiency 44, 616636
STAT3	119.5	99.9	99	Autoimmune disease, multisystem, infantile-onset, 1, 615952 Hyper-IgE recurrent infection syndrome, 147060
STAT5B	130.6	99.7	97.2	Growth hormone insensitivity with immunodeficiency, 245590 Leukemia, acute promyelocytic, somatic, 102578
STEAP3	199	100	99.8	?Anemia, hypochromic microcytic, with iron overload 2, 615234
STIL	157.2	99.8	98.6	Microcephaly 7, primary, autosomal recessive, 612703
STIM1	145.3	100	99.2	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070
STK11	111.9	99.7	95.8	Melanoma, malignant, somatic, 0 Pancreatic cancer, 260350 Peutz-Jeghers syndrome, 175200 Testicular tumor, somatic, 273300
STK4	138.9	100	99.3	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STN1	94.4	99.9	99.5	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341

STOX1	177.7	89.6	89.5	Preeclampsia/eclampsia 4, 609404
STRA6	116.5	100	99.9	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186
STRADA	134.1	100	98.9	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087
STRC	119	99.9	98.4	Deafness, autosomal recessive 16, 603720
STS	91.6	99.7	97.8	Ichthyosis, X-linked, 308100
STT3A	156.2	100	100	?Congenital disorder of glycosylation, type Iw, 615596
STT3B	125.1	99.2	96	?Congenital disorder of glycosylation, type Ix, 615597
STUB1	176.1	100	98.9	?Spinocerebellar ataxia 48, 618093 Spinocerebellar ataxia, autosomal recessive 16, 615768
STX11	311.4	100	100	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STX16	140.6	99.8	97.8	Pseudohypoparathyroidism, type IB, 603233
STX1B	152.2	100	98.4	Generalized epilepsy with febrile seizures plus, type 9, 616172
STXBP1	124.5	96.8	96.8	Epileptic encephalopathy, early infantile, 4, 612164
STXBP2	102.3	88.9	83.8	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
SUCLA2	64.9	93.3	82.8	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	101.3	99.6	95.4	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUFU	122.6	99.9	99	Basal cell nevus syndrome, 109400 Joubert syndrome 32, 617757 Medulloblastoma, desmoplastic, 155255 {Meningioma, familial, susceptibility to}, 607174
SUGCT	129.1	94.1	87.6	Glutaric aciduria III, 231690
SULT2B1	111.4	100	100	Ichthyosis, congenital, autosomal recessive 14, 617571
SUMF1	103.3	98.6	91.1	Multiple sulfatase deficiency, 272200
SUMO1	17.6	58.8	37	?Orofacial cleft 10, 613705
SUN5	115	99.9	99.1	Spermatogenic failure 16, 617187
SUOX	212.6	100	100	Sulfite oxidase deficiency, 272300
SURF1	96.2	88.3	88.3	Charcot-Marie-Tooth disease, type 4K, 616684 Leigh syndrome, due to COX IV deficiency, 256000
SYCE1	92.9	98.7	93.6	?Premature ovarian failure 12, 616947

				?Spermatogenic failure 15, 616950
SYCP3	79	97.5	86.8	Pregnancy loss, recurrent, 4, 270960 Spermatogenic failure 4, 270960
SYN1	64.2	74	63.2	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNE1	136.6	98.2	97.6	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 Spinocerebellar ataxia, autosomal recessive 8, 610743
SYNE2	123.1	98.6	96	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999
SYNE4	73.9	98.3	91.6	Deafness, autosomal recessive 76, 615540
SYNGAP1	141.3	98.4	98	Mental retardation, autosomal dominant 5, 612621
SYNJ1	127.2	99.3	96.1	Epileptic encephalopathy, early infantile, 53, 617389 Parkinson disease 20, early-onset, 615530
SYP	72	99.8	94	Mental retardation, X-linked 96, 300802
SYT1	171.3	99.8	98.6	Baker-Gordon syndrome, 618218
SYT14	113.5	59.9	53.8	?Spinocerebellar ataxia, autosomal recessive 11, 614229
SYT2	101.3	100	99	Myasthenic syndrome, congenital, 7, presynaptic, 616040
SZT2	149.5	99.5	99.2	Epileptic encephalopathy, early infantile, 18, 615476
T	141.2	98.8	93.6	Sacral agenesis with vertebral anomalies, 615709 {Neural tube defects, susceptibility to}, 182940
TAB2	210.5	99.7	97.6	Congenital heart defects, nonsyndromic, 2, 614980
TAC3	80.5	99.1	91.1	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACO1	91.7	97	92.6	Mitochondrial complex IV deficiency, 220110
TACR3	180.3	100	100	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
TACSTD2	223.6	99.1	96.6	Corneal dystrophy, gelatinous drop-like, 204870
TAF1	112.4	99.4	96.6	Dystonia-Parkinsonism, X-linked, 314250 Mental retardation, X-linked, syndromic 33, 300966
TAF13	81.8	100	99.9	Mental retardation, autosomal recessive 60, 617432
TAF2	112.8	98.8	94.7	Mental retardation, autosomal recessive 40, 615599
TAF4B	146.1	96.5	90.9	?Spermatogenic failure 13, 615841
TAF6	130	99.9	98.6	Alazami-Yuan syndrome, 617126
TAL1	43.7	73.9	63.3	Leukemia, T-cell acute lymphocytic, somatic, 613065

TAL2	112.5	100	100	Leukemia, T-cell acute lymphocytic, somatic, 613065
TALDO1	130.5	100	99.9	Transaldolase deficiency, 606003
TANGO2	145.3	100	100	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TAP1	103.3	100	99.1	Bare lymphocyte syndrome, type I, 604571
TAP2	95.2	99.6	98.6	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	100.7	96.1	94.3	Bare lymphocyte syndrome, type I, 604571
TAPT1	83.5	88.5	85.9	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897
TARDBP	175.2	100	100	Amyotrophic lateral sclerosis 10, with or without FTD, 612069 Frontotemporal lobar degeneration, TARDBP-related, 612069
TARS2	98.8	99.9	98.5	?Combined oxidative phosphorylation deficiency 21, 615918
TAT	143.1	100	100	Tyrosinemia, type II, 276600
TAZ	94	99.9	98.8	Barth syndrome, 302060
TBC1D20	145.7	94.2	94.1	Warburg micro syndrome 4, 615663
TBC1D23	86	95.7	91.5	Pontocerebellar hypoplasia, type 11, 617695
TBC1D24	179.2	100	100	Deafness , autosomal recessive 86, 614617 Deafness, autosomal dominant 65, 616044 DOORS syndrome, 220500 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021
TBC1D7	105.5	99.6	96.6	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000
TBCD	152.9	95.5	92.3	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TBCE	128	99.9	98.2	Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Kenny-Caffey syndrome, type 1, 244460
TBCK	86.5	95.7	89.3	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900
TBK1	102.5	97.8	90.7	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439 {Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 8}, 617900
TBL1XR1	79.4	91.5	73	Mental retardation, autosomal dominant 41, 616944

				Pierpont syndrome, 602342
TBP	129.5	100	98.1	Spinocerebellar ataxia 17, 607136 {Parkinson disease, susceptibility to}, 168600
TBR1	120.8	100	99.1	Intellectual developmental disorder with autism and speech delay, 606053
TBX1	75.3	77.1	67.4	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Velocardiofacial syndrome, 192430
TBX15	131.7	100	99.4	Cousin syndrome, 260660
TBX18	94.4	98	95.2	Congenital anomalies of kidney and urinary tract 2, 143400
TBX19	174.4	100	100	Adrenocorticotropic hormone deficiency, 201400
TBX2	107.6	99.1	92.7	Vertebral anomalies and variable endocrine and T-cell dysfunction, 618223
TBX20	142.8	99.9	99.3	Atrial septal defect 4, 611363
TBX21	81.7	90	83.1	Asthma and nasal polyps, 208550 {Asthma, aspirin-induced, susceptibility to}, 208550
TBX22	121.8	99.2	96.3	?Abruzzo-Erickson syndrome, 302905 Cleft palate with ankyloglossia, 303400
TBX3	80.5	99.6	95.3	Ulnar-mammary syndrome, 181450
TBX4	170.7	94.9	92.8	Ischiocoxopodopatellar syndrome, 147891
TBX5	141.3	100	100	Holt-Oram syndrome, 142900
TBX6	122.1	91.5	79.7	Spondylocostal dysostosis 5, 122600
TBXAS1	140.3	100	100	?Thromboxane synthase deficiency, 614158 Ghosal hematodiaphyseal syndrome, 231095
TCAP	89	100	99.2	Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954
TCF12	150.3	100	99.8	Craniosynostosis 3, 615314
TCF3	67.8	98.9	92.3	Agammaglobulinemia 8, autosomal dominant, 616941
TCF4	128	99.9	99.5	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954
TCHH	148.1	100	100	?Uncombable hair syndrome 3, 617252
TCIRG1	113.5	95.4	89.4	Osteopetrosis, autosomal recessive 1, 259700
TCN2	175.6	100	100	Transcobalamin II deficiency, 275350
TCOF1	98.6	99.5	97.3	Treacher Collins syndrome 1, 154500

TCTEX1D2	126.1	100	99.3	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405
TCTN1	98.8	95.7	92.8	Joubert syndrome 13, 614173
TCTN2	144.2	99.5	97	?Meckel syndrome 8, 613885 Joubert syndrome 24, 616654
TCTN3	127.6	100	99.8	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TDGF1	151.4	99.8	96.4	Forebrain defects, 0
TDP1	122.9	98.7	95.3	Spinocerebellar ataxia, autosomal recessive with axonal neuropathy, 607250
TDP2	165.1	99.9	98.8	Spinocerebellar ataxia, autosomal recessive 23, 616949
TDRD7	168.3	99	97.7	Cataract 36, 613887
TDRD9	121.2	98.5	96.5	?Spermatogenic failure 30, 618110
TEAD1	158.6	99.8	98.2	Sveinsson chorioretinal atrophy, 108985
TECPR2	161.1	100	99.9	Spastic paraplegia 49, autosomal recessive, 615031
TECR	94.6	99.9	97.9	Mental retardation, autosomal recessive 14, 614020
TECRL	59.3	89.9	77.1	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021
TECTA	208	100	100	Deafness, autosomal dominant 8/12, 601543 Deafness, autosomal recessive 21, 603629
TEK	184.1	100	100	Glaucoma 3, primary congenital, E, 617272 Venous malformations, multiple cutaneous and mucosal, 600195
TELO2	98.1	97.4	93.7	You-Hoover-Fong syndrome, 616954
TENM3	185.7	99.5	98.7	Microphthalmia, isolated, with coloboma 9, 615145
TENM4	160.8	99.9	99.2	Essential tremor, hereditary, 5, 616736
TERC	NC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550 {Aplastic anemia}, 614743 {Pulmonary fibrosis, idiopathic, susceptibility to}, 614743
TET2	212.4	100	99.9	Myelodysplastic syndrome, somatic, 614286
TEX11	77.3	91.6	83.4	Spermatogenic failure, X-linked, 2, 309120
TEX14	111.6	99.7	98	?Spermatogenic failure 23, 617707
TEX15	114.6	99.8	99	Spermatogenic failure 25, 617960
TF	125.9	100	100	Atransferrinemia, 209300
TFAM	65	88.7	66.2	?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156
TFAP2A	109.3	100	99.3	Branchiooculofacial syndrome, 113620

TFAP2B	153.5	98.8	96.3	Char syndrome, 169100 Patent ductus arteriosus 2, 617035
TFE3	73.5	99.5	93.1	Renal cell carcinoma, papillary, 1, 300854
TFG	121.5	93.9	90.7	?Spastic paraplegia 57, autosomal recessive, 615658 Hereditary motor and sensory neuropathy, Okinawa type, 604484
TFR2	104.4	98.2	93.5	Hemochromatosis, type 3, 604250
TFRC	157.2	99.9	99.1	Immunodeficiency 46, 616740
TG	133.4	100	99.7	Thyroid dyshormonogenesis 3, 274700 {Autoimmune thyroid disease, susceptibility to, 3}, 608175
TGDS	82.4	98.1	88.8	Catel-Manzke syndrome, 616145
TGFB1	86.9	99.7	95.1	Camurati-Engelmann disease, 131300 Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213 {Cystic fibrosis lung disease, modifier of}, 219700
TGFB2	176.9	100	99.9	Loeys-Dietz syndrome 4, 614816
TGFB3	171.5	100	100	Arrhythmogenic right ventricular dysplasia 1, 107970 Loeys-Dietz syndrome 5, 615582
TGFBI	130.9	99	94.5	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	173.4	93.7	93.6	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800
TGFBR2	193.5	100	99.9	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome 2, 610168
TGIF1	138.3	100	100	Holoprosencephaly 4, 142946
TGM1	158.8	100	100	Ichthyosis, congenital, autosomal recessive 1, 242300
TGM3	187.1	100	99.8	?Uncombable hair syndrome 2, 617251
TGM5	173.9	100	100	Peeling skin syndrome 2, 609796
TGM6	149.7	99.7	98	Spinocerebellar ataxia 35, 613908
TH	68.2	97.6	88.7	Segawa syndrome, recessive, 605407

THAP1	122	100	100	Dystonia 6, torsion, 602629
THBD	108.2	99.8	97.8	Thrombophilia due to thrombomodulin defect, 614486 {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
THOC2	77.9	96.2	86.6	Mental retardation, X-linked 12/35, 300957
THOC6	248.6	100	99.9	Beaulieu-Boycott-Innes syndrome, 613680
THPO	88.2	100	100	Thrombocythemia 1, 187950
THRA	172.8	100	99.8	Hypothyroidism, congenital, nongoitrous, 6, 614450
THRB	167.4	100	99.5	Thyroid hormone resistance, 188570 Thyroid hormone resistance, autosomal recessive, 274300 Thyroid hormone resistance, selective pituitary, 145650
TIA1	128.5	98.6	89.6	Welander distal myopathy, 604454
TIMM50	108.2	98.8	95.3	3-methylglutaconic aciduria, type IX, 617698
TIMM8A	46	94.5	78.8	Mohr-Tranebjaerg syndrome, 304700
TIMMDC1	152.2	100	100	Mitochondrial complex I deficiency, nuclear type 31, 618251
TIMP3	147	100	100	Sorsby fundus dystrophy, 136900
TINF2	184	100	100	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TJP2	111.1	93.8	92.2	Cholestasis, progressive familial intrahepatic 4, 615878 Hypercholanemia, familial, 607748
TK2	105.7	93.4	89.4	?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069 Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560
TKT	114.1	98.7	97.7	Short stature, developmental delay, and congenital heart defects, 617044
TLE6	109.9	99.9	98.9	Preimplantation embryonic lethality, 616814
TLK2	113	98.8	94.7	Mental retardation, autosomal dominant 57, 618050
TLL1	140.1	100	99.9	Atrial septal defect 6, 613087
TMC1	122.8	98.2	93.8	Deafness, autosomal dominant 36, 606705 Deafness, autosomal recessive 7, 600974
TMC6	83.7	99.9	99	Epidermodysplasia verruciformis, 226400
TMC8	108.1	97.6	91.6	Epidermodysplasia verruciformis 2, 618231
TMCO1	78.7	88	86.5	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980

TMEM106B	120.2	99.8	96.4	Leukodystrophy, hypomyelinating, 16, 617964
TMEM107	163.8	100	100	?Joubert syndrome 29, 617562 Meckel syndrome 13, 617562 Orofaciodigital syndrome XVI, 617563
TMEM126A	120.3	98.4	86.2	Optic atrophy 7, 612989
TMEM126B	79.2	99.8	97.7	Mitochondrial complex I deficiency, nuclear type 29, 618250
TMEM138	100.2	100	99.5	Joubert syndrome 16, 614465
TMEM165	113.9	99.8	98.1	Congenital disorder of glycosylation, type IIk, 614727
TMEM173	100.8	98.7	93.4	STING-associated vasculopathy, infantile-onset, 615934
TMEM199	105.1	100	99.9	Congenital disorder of glycosylation, type IIp, 616829
TMEM216	111.9	100	98.7	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	111.5	100	99.9	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	100.7	99.8	98.3	Joubert syndrome 14, 614424
TMEM240	112.2	99.8	97.4	Spinocerebellar ataxia 21, 607454
TMEM260	116.7	96.8	90	Structural heart defects and renal anomalies syndrome, 617478
TMEM38B	114.3	100	99.4	Osteogenesis imperfecta, type XIV, 615066
TMEM43	124.9	100	99.5	Arrhythmogenic right ventricular dysplasia 5, 604400 Emery-Dreifuss muscular dystrophy 7, AD, 614302
TMEM5	120.5	96.8	92.9	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
TMEM67	72.9	93.3	83.4	?RHYNS syndrome, 602152 COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991
TMEM70	138.7	94.6	90.3	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMEM98	136.3	99.4	98.7	Nanophthalmos 4, 615972
TMIE	109.6	98.8	92.1	Deafness, autosomal recessive 6, 600971
TMPRSS15	116.9	95.5	89.1	Enterokinase deficiency, 226200
TMPRSS3	125.5	100	99.9	Deafness, autosomal recessive 8/10, 601072

TMPRSS6	101.7	100	99.1	Iron-refractory iron deficiency anemia, 206200
TMTC3	83.2	98.9	93	Lissencephaly 8, 617255
TNC	187.5	100	99.7	Deafness, autosomal dominant 56, 615629
TNFAIP3	135.9	100	99.9	Autoinflammatory syndrome, familial, Behcet-like, 616744
TNFRSF10B	114.1	100	100	Squamous cell carcinoma, head and neck, 275355
TNFRSF11A	146.3	93.3	91.4	Osteolysis, familial expansile, 174810 Osteopetrosis, autosomal recessive 7, 612301 {Paget disease of bone 2, early-onset}, 602080
TNFRSF11B	224.8	100	100	Paget disease of bone 5, juvenile-onset, 239000
TNFRSF13B	102.1	100	99.7	Immunodeficiency, common variable, 2, 240500 Immunoglobulin A deficiency 2, 609529
TNFRSF13C	55.8	76.5	66.8	Immunodeficiency, common variable, 4, 613494
TNFRSF1A	93.2	90.8	87.9	Periodic fever, familial, 142680 {Multiple sclerosis, susceptibility to, 5}, 614810
TNFRSF4	51.6	97.3	85.4	?Immunodeficiency 16, 615593
TNFSF11	150.4	99.3	93.2	Osteopetrosis, autosomal recessive 2, 259710
TNIK	111.1	99.9	99.3	Mental retardation, autosomal recessive 54, 617028
TNNC1	174.5	100	100	Cardiomyopathy, dilated, 1Z, 611879 Cardiomyopathy, hypertrophic, 13, 613243
TNNI2	121.2	100	99.6	Arthrogryposis multiplex congenita, distal, type 2B, 601680
TNNI3	86.7	98.1	86.5	?Cardiomyopathy, dilated, 2A, 611880 Cardiomyopathy, dilated, 1FF, 613286 Cardiomyopathy, familial restrictive, 1, 115210 Cardiomyopathy, hypertrophic, 7, 613690
TNNI3K	118.8	98.8	96	Cardiac conduction disease with or without dilated cardiomyopathy, 616117
TNNT1	86.9	96.3	94	Nemaline myopathy 5, Amish type, 605355
TNNT2	106.3	100	99.9	Cardiomyopathy, dilated, 1D, 601494 Cardiomyopathy, familial restrictive, 3, 612422 Cardiomyopathy, hypertrophic, 2, 115195 Left ventricular noncompaction 6, 601494
TNNT3	121	99.9	97.8	Arthrogryposis, distal, type 2B, 601680
TNPO3	139.6	100	99.7	Muscular dystrophy, limb-girdle, autosomal dominant 2, 608423
TNRC6A	162.9	99.9	99.4	?Epilepsy, familial adult myoclonic, 6, 618074

TNXB	96.4	98.4	91.4	Ehlers-Danlos syndrome, classic-like, 1, 606408 Vesicoureteral reflux 8, 615963
TOE1	165.1	100	100	Pontocerebellar hypoplasia, type 7, 614969
TOP1	97.9	99.5	97.3	DNA topoisomerase I, camptothecin-resistant, 0
TOP2A	123.8	99.6	97.2	DNA topoisomerase II, resistance to inhibition of, by amsacrine, 0
TOP3A	129.8	98.9	96.5	?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098 Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097
TOPORS	210.6	100	100	Retinitis pigmentosa 31, 609923
TOR1A	185	100	99.8	Dystonia-1, torsion, 128100 {Dystonia-1, modifier of}, 0
TOR1AIP1	143.8	97.6	95.9	?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072
TP53	92	99.9	98.1	Adrenocortical carcinoma, pediatric, 202300 Bone marrow failure syndrome 5, 618165 Breast cancer, somatic, 114480 Choroid plexus papilloma, 260500 Colorectal cancer, 114500 Hepatocellular carcinoma, somatic, 114550 Li-Fraumeni syndrome, 151623 Nasopharyngeal carcinoma, somatic, 607107 Osteosarcoma, 259500 Pancreatic cancer, somatic, 260350 {Basal cell carcinoma 7}, 614740 {Glioma susceptibility 1}, 137800
TP53RK	37.4	91.3	76.5	Galloway-Mowat syndrome 4, 617730
TP63	206.3	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, 618149 Rapp-Hodgkin syndrome, 129400 Split-hand/foot malformation 4, 605289
TPI1	103	99.2	96.7	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512

TPK1	112.7	99.8	97.3	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TPM1	132.9	99.7	97.9	Cardiomyopathy, dilated, 1Y, 611878 Cardiomyopathy, hypertrophic, 3, 115196 Left ventricular noncompaction 9, 611878
TPM2	109.1	100	99.6	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	89.4	89.1	CAP myopathy 1, 609284 Myopathy, congenital, with fiber-type disproportion, 255310 Nemaline myopathy 1, autosomal dominant or recessive, 609284
TPO	134.8	99.9	98.5	Thyroid dyshormonogenesis 2A, 274500
TPP1	146.3	100	100	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TPRKB	56.7	79.3	67.1	Galloway-Mowat syndrome 5, 617731
TPRN	62.7	74.7	65.4	Deafness, autosomal recessive 79, 613307
TRAC	170.9	100	100	Immunodeficiency 7, TCR-alpha/beta deficient, 615387
TRAF3IP1	90.3	96.3	92.8	Senior-Loken syndrome 9, 616629
TRAF3IP2	116.6	99.9	97.7	?Candidiasis, familial, 8, 615527 {Psoriasis susceptibility 13}, 614070
TRAF7	147.2	98.3	95	Cardiac, facial, and digital anomalies with developmental delay, 618164
TRAIP	141.6	100	100	Seckel syndrome 9, 616777
TRAK1	152.3	99	98.1	Epileptic encephalopathy, early infantile, 68, 618201
TRAPPC11	126.2	99.4	96.4	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356
TRAPPC12	141.6	99.9	98.5	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669
TRAPPC2	85.3	87	66.7	Spondyloepiphyseal dysplasia tarda, 313400
TRAPPC6B	61.8	99.4	94.4	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862
TRAPPC9	135	100	99.9	Mental retardation, autosomal recessive 13, 613192
TRDN	71.9	83.6	70.8	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441
TREH	143.1	98	93.5	Trehalase deficiency, 612119

TREM2	149	99.9	99.6	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193
TREX1	242.4	100	100	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700
TRHR	232	100	99.7	Thyrotropin-releasing hormone resistance, generalized, 0
TRIM2	157.7	93.6	91.4	Charcot-Marie-Tooth disease, type 2R, 615490
TRIM32	141.2	100	100	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRIM36	156.8	99.1	96.8	?Anencephaly, 206500
TRIM37	110.2	98.2	97.2	Mulibrey nanism, 253250
TRIM44	141.7	100	100	?Aniridia 3, 617142
TRIO	134.3	97.9	95.4	Mental retardation, autosomal dominant 44, 617061
TRIOBP	135.6	97	94.9	Deafness, autosomal recessive 28, 609823
TRIP11	84.3	95.2	87.4	Achondrogenesis, type IA, 200600
TRIP12	139.8	99.5	98.8	Mental retardation, autosomal dominant 49, 617752
TRIP13	141.2	100	100	Mosaic variegated aneuploidy syndrome 3, 617598
TRIP4	113.5	100	98.8	?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066 Spinal muscular atrophy with congenital bone fractures 1, 616866
TRIT1	119.4	100	99.8	Combined oxidative phosphorylation deficiency 35, 617873
TRMT10A	135.2	100	99.4	Microcephaly, short stature, and impaired glucose metabolism 1, 616033
TRMT10C	131.4	99.8	98.8	Combined oxidative phosphorylation deficiency 30, 616974
TRMT5	208.7	99.2	93.9	Combined oxidative phosphorylation deficiency 26, 616539
TRMU	99	100	99.6	Liver failure, transient infantile, 613070 {Deafness, mitochondrial, modifier of}, 580000
TRNT1	104.6	97.8	92.3	Retinitis pigmentosa and erythrocytic microcytosis, 616959 Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084
TRPA1	92.4	91.9	85.3	?Episodic pain syndrome, familial, 1, 615040
TRPC3	180.8	98.7	95.6	?Spinocerebellar ataxia 41, 616410
TRPC6	103.8	99	96.1	Glomerulosclerosis, focal segmental, 2, 603965
TRPM1	161	100	99.6	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216

TRPM4	109.2	99.8	98.5	Progressive familial heart block, type IB, 604559
TRPM6	151.1	99.8	98.7	Hypomagnesemia 1, intestinal, 602014
TRPS1	175	100	99.8	Trichorhinophalangeal syndrome, type I, 190350 Trichorhinophalangeal syndrome, type III, 190351
TRPV3	144.9	100	99.4	?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400 Olmsted syndrome, 614594
TRPV4	172.4	99.5	98.7	?Avascular necrosis of femoral head, primary, 2, 617383 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
TRPV6	167.4	99.5	98.5	Hyperparathyroidism, transient neonatal, 618188
TSC1	128.8	99.8	98.8	Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100
TSC2	131.2	100	99	?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254
TSEN15	74.2	99	93.6	Pontocerebellar hypoplasia, type 2F, 617026
TSEN2	123.8	100	99.8	Pontocerebellar hypoplasia type 2B, 612389
TSEN34	53.5	90.5	85.7	?Pontocerebellar hypoplasia type 2C, 612390
TSEN54	82.9	95.9	92.9	?Pontocerebellar hypoplasia type 5, 610204 Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753
TSFM	127.2	100	100	Combined oxidative phosphorylation deficiency 3, 610505
TSGA10	102.5	98.5	93.1	?Spermatogenic failure 26, 617961
TSHB	271.7	100	100	Hypothyroidism, congenital, nongoitrous 4, 275100

TSHR	216.5	99.2	96.8	Hyperthyroidism, familial gestational, 603373 Hyperthyroidism, nonautoimmune, 609152 Hypothyroidism, congenital, nongoitrous, 1, 275200 Thyroid adenoma, hyperfunctioning, somatic, 0 Thyroid carcinoma with thyrotoxicosis, 0
TSHZ1	166.6	98.8	98.5	Aural atresia, congenital, 607842
TSPAN12	129.4	100	99.5	Exudative vitreoretinopathy 5, 613310
TSPAN7	120.7	99.9	98.6	Mental retardation, X-linked 58, 300210
TSPEAR	141.5	100	99	?Deafness, autosomal recessive 98, 614861 Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180
TSPYL1	141.5	100	99.4	Sudden infant death with dysgenesis of the testes syndrome, 608800
TSR2	81.6	100	99.1	?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946
TTBK2	123.2	100	98.9	Spinocerebellar ataxia 11, 604432
TTC19	92.1	80.6	72.5	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTC21B	100.7	99.7	97.6	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
TTC25	103.4	100	99.5	Ciliary dyskinesia, primary, 35, 617092
TTC37	124	99.6	98.1	Trichohepatoenteric syndrome 1, 222470
TTC7A	123	99.9	98.3	Gastrointestinal defects and immunodeficiency syndrome, 243150
TTC8	106.9	97.9	92	?Retinitis pigmentosa 51, 613464 Bardet-Biedl syndrome 8, 615985
TTI2	104.5	100	99.7	Mental retardation, autosomal recessive 39, 615541
TTLL5	152.7	99.9	98.7	Cone-rod dystrophy 19, 615860
TTN	187.8	98.2	97.2	Cardiomyopathy, dilated, 1G, 604145 Cardiomyopathy, familial hypertrophic, 9, 613765 Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807 Myopathy, proximal, with early respiratory muscle involvement, 603689 Salih myopathy, 611705 Tibial muscular dystrophy, tardive, 600334
TTPA	101.5	83.6	76.6	Ataxia with isolated vitamin E deficiency, 277460

TTR	152.3	94.6	94.6	Amyloidosis, hereditary, transthyretin-related, 105210 Carpal tunnel syndrome, familial, 115430 [Dystransthyretinemic hyperthyroxinemia], 145680
TUB	103	97.3	95.2	?Retinal dystrophy and obesity, 616188
TUBA1A	113.2	99.9	97.8	Lissencephaly 3, 611603
TUBA3D	144.5	100	99.6	Keratoconus 9, 617928
TUBA4A	220.6	100	100	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia, 616208
TUBA8	177.1	99.9	99.7	Cortical dysplasia, complex, with other brain malformations 8, 613180
TUBB	158.6	99.3	97.4	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	109.9	96.7	95.6	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB2B	100	100	100	Cortical dysplasia, complex, with other brain malformations 7, 610031
TUBB3	136.1	98.1	96.9	Cortical dysplasia, complex, with other brain malformations 1, 614039 Fibrosis of extraocular muscles, congenital, 3A, 600638
TUBB4A	121.2	96	95.3	Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438
TUBB4B	109.8	98.4	96	Leber congenital amaurosis with early-onset deafness, 617879
TUBB6	111.2	90.9	90.5	?Facial palsy, congenitla, with ptosis and velopharyngeal dysfunction, 617732
TUBB8	31.3	95.5	71	Oocyte maturation defect 2, 616780
TUBG1	164.2	100	100	Cortical dysplasia, complex, with other brain malformations 4, 615412
TUBGCP4	130.8	99.1	96.2	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TUBGCP6	152.2	99.9	98.9	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
TUFM	135.4	100	99.7	Combined oxidative phosphorylation deficiency 4, 610678
TULP1	97.8	96.8	91.7	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132
TUSC3	136.4	100	98.3	Mental retardation, autosomal recessive 7, 611093

TWIST1	134.4	96.6	87.2	Craniosynostosis 1, 123100 Robinow-Sorauf syndrome, 180750 Saethre-Chotzen syndrome with or without eyelid anomalies, 101400 Sweeney-Cox syndrome, 617746
TWIST2	131.3	100	99.3	Ablepharon-macrostomia syndrome, 200110 Barber-Say syndrome, 209885 Focal facial dermal dysplasia 3, Setleis type, 227260
TWNK	178.8	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
TXN2	81.2	100	100	?Combined oxidative phosphorylation deficiency 29, 616811
TXNL4A	109	100	99.5	Burn-McKeown syndrome, 608572
TXNRD2	119.3	93.3	91.2	?Glucocorticoid deficiency 5, 617825
TYK2	119.2	99.9	98.8	Immunodeficiency 35, 611521
TYMP	95.2	98.3	85	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYR	185.3	100	100	Albinism, oculocutaneous, type IA, 203100 Albinism, oculocutaneous, type IB, 606952 Waardenburg syndrome/albinism, digenic, 103470 [Skin/hair/eye pigmentation 3, blue/green eyes], 601800 [Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 {Melanoma, cutaneous malignant, susceptibility to, 8}, 601800
TYROBP	95.2	100	99.9	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770
TYRP1	176.9	100	99.9	Albinism, oculocutaneous, type III, 203290 [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271
UBA1	162	99.8	98.9	Spinal muscular atrophy, X-linked 2, infantile, 301830
UBA5	75.4	94.1	77.1	?Spinocerebellar ataxia, autosomal recessive 24, 617133 Epileptic encephalopathy, early infantile, 44, 617132
UBE2A	100.5	99.9	96.9	Mental retardation, X-linked syndromic, Nascimento-type, 300860
UBE2T	107	100	99.3	Fanconi anemia, complementation group T, 616435
UBE3A	89.8	97.8	91.4	Angelman syndrome, 105830
UBE3B	127.8	100	99.9	Kaufman oculocerebrofacial syndrome, 244450

UBIAD1	248.8	98.9	95.2	Corneal dystrophy, Schnyder type, 121800
UBQLN2	136.3	99.7	98	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857
UBR1	128.2	99.2	96	Johanson-Blizzard syndrome, 243800
UBTF	123.6	99.9	99	Neurodegeneration, childhood-onset, with brain atrophy, 617672
UCHL1	109.1	97.9	89.1	Spastic paraplegia 79, autosomal recessive, 615491 {?Parkinson disease 5, susceptibility to}, 613643
UFC1	150.3	100	100	Neurodevelopmental disorder with spasticity and poor growth, 618076
UFM1	105.8	71	70.1	Leukodystrophy, hypomyelinating, 14, 617899
UFSP2	128.4	99.7	96.5	?Hip dysplasia, Beukes type, 142669 ?Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974
UGT1A1	240.6	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	97.8	97.2	Glomerulocystic kidney disease with hyperuricemia and isosthenuria, 609886 Hyperuricemic nephropathy, familial juvenile 1, 162000 Medullary cystic kidney disease 2, 603860
UMPS	173.6	99.3	97.2	Orotic aciduria, 258900
UNC119	92.9	97.8	90.4	?Cone-rod dystrophy, 0 ?Immunodeficiency 13, 615518
UNC13D	97	99.6	97.7	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC45B	143	100	99.5	?Cataract 43, 616279
UNC80	133.4	99.9	99.1	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801
UNG	78.4	99.5	94.2	Immunodeficiency with hyper IgM, type 5, 608106
UPB1	157.4	100	100	Beta-ureidopropionase deficiency, 613161
UPF3B	47.4	91.2	76.6	Mental retardation, X-linked, syndromic 14, 300676
UQCC2	96.6	100	99.1	Mitochondrial complex III deficiency, nuclear type 7, 615824
UQCC3	95	100	99.2	?Mitochondrial complex III deficiency, nuclear type 9, 616111
UQCRB	107.6	99.6	96.8	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC2	122.6	99.9	99.1	Mitochondrial complex III deficiency, nuclear type 5, 615160

UQCRQ	131.3	100	99.9	Mitochondrial complex III deficiency, nuclear type 4, 615159
UROC1	132	99.9	99	?Urocanase deficiency, 276880
UROD	163.1	99.8	97.9	Porphyria cutanea tarda, 176100 Porphyria, hepatoerythropoietic, 176100
UROS	108.3	100	99.9	Porphyria, congenital erythropoietic, 263700
USB1	125	99.9	98.2	Poikiloderma with neutropenia, 604173
USH1C	97.5	100	99.4	Deafness, autosomal recessive 18A, 602092 Usher syndrome, type 1C, 276904
USH1G	195.3	98.4	96.3	Usher syndrome, type 1G, 606943
USH2A	148.5	100	99.7	Retinitis pigmentosa 39, 613809 Usher syndrome, type 2A, 276901
USP18	201.4	95.9	95.9	Pseudo-TORCH syndrome 2, 617397
USP27X	248.7	100	100	Mental retardation, X-linked 105, 300984
USP8	71.5	97.3	87.8	Pituitary adenoma 4, ACTH-secreting, somatic, 219090
USP9X	108.2	97.2	91.1	Mental retardation, X-linked 99, 300919 Mental retardation, X-linked 99, syndromic, female-restricted, 300968
USP9Y	30.8	47.2	39.9	Spermatogenic failure, Y-linked, 2, 415000
UVSSA	149.4	99.1	98.4	UV-sensitive syndrome 3, 614640
VAC14	108.4	99.8	98.5	Striatonigral degeneration, childhood-onset, 617054
VAMP1	131.5	100	100	Spastic ataxia 1, autosomal dominant, 108600
VANGL1	165.3	100	100	Caudal regression syndrome, 600145 {Neural tube defects, susceptibility to}, 182940
VANGL2	184.6	100	99.6	Neural tube defects, 182940
VAPB	107.8	99	95.9	Amyotrophic lateral sclerosis 8, 608627 Spinal muscular atrophy, late-onset, Finkel type, 182980
VARS	126.1	99.9	98	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802
VARS2	110.9	99.9	98.9	Combined oxidative phosphorylation deficiency 20, 615917
VAX1	52.2	88.4	78	?Microphthalmia, syndromic 11, 614402
VCAN	186.5	100	100	Wagner syndrome 1, 143200
VCL	115.8	100	99.8	Cardiomyopathy, dilated, 1W, 611407 Cardiomyopathy, hypertrophic, 15, 613255
VCP	144.8	99.9	99.5	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	123.3	98	95.2	?Osteoporosis, involutional, 166710 Rickets, vitamin D-resistant, type IIA, 277440
VEGFC	164.5	100	99.5	Lymphatic malformation 4, 615907
VHL	119.7	92.6	85.3	Erythrocytosis, familial, 2, 263400 Hemangioblastoma, cerebellar, somatic, 0 Pheochromocytoma, 171300 Renal cell carcinoma, somatic, 144700 von Hippel-Lindau syndrome, 193300
VIM	126.5	99.1	97.2	Cataract 30, pulverulent, 116300
VIPAS39	144.6	100	100	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VKORC1	162.1	100	100	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473 Warfarin resistance, 122700
VLDLR	200.9	99.9	99.4	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VMA21	42.3	95.2	81.3	Myopathy, X-linked, with excessive autophagy, 310440
VPS11	144.9	95.3	93.2	Leukodystrophy, hypomyelinating, 12, 616683
VPS13A	69.5	95.3	85.3	Choreoacanthocytosis, 200150
VPS13B	143.8	98.6	96.8	Cohen syndrome, 216550
VPS13C	106.6	96.6	90	Parkinson disease 23, autosomal recessive, early onset, 616840
VPS13D	158.6	99.9	99.4	Spinocerebellar ataxia, autosomal recessive 4, 607317
VPS33A	106.3	96.2	95.1	Mucopolysaccharidosis-plus syndrome, 617303
VPS33B	138.3	100	100	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
VPS37A	73.6	86.6	66.4	Spastic paraplegia 53, autosomal recessive, 614898
VPS45	131.5	96.2	94.9	Neutropenia, severe congenital, 5, autosomal recessive, 615285
VPS53	129.2	91.4	90.4	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	124.8	97.5	94.2	Pontocerebellar hypoplasia type 1A, 607596
VSX1	52.2	85.5	76.1	?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195 Keratoconus 1, 148300
VSX2	77.7	99.8	97.3	Microphthalmia with coloboma 3, 610092 Microphthalmia, isolated 2, 610093
VWA3B	141.5	99.8	98.4	?Spinocerebellar ataxia, autosomal recessive 22, 616948

VWF	120.9	100	99.6	von Willebrand disease, type 1, 193400 von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 von Willibrand disease, type 3, 277480
WAC	165.5	99.7	96.7	Desanto-Shinawi syndrome, 616708
WARS	123.8	99.7	98	Neuronopathy, distal hereditary motor, type IX, 617721
WARS2	140.7	100	99.5	Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710
WAS	66.1	88.2	78.7	Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, 313900 Thrombocytopenia, X-linked, intermittent, 313900 Wiskott-Aldrich syndrome, 301000
WASHC4	91.8	95.3	89.6	?Mental retardation, autosomal recessive 43, 615817
WASHC5	146.6	99.6	98.1	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, autosomal dominant, 603563
WBP2	93.7	100	100	Deafness, autosomal recessive 107, 617639
WDFY3	133.6	99.7	98.1	?Microcephaly 18, primary, autosomal dominant, 617520
WDPCP	107.3	93.9	88.9	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR11	130.6	96.9	96.4	Hypogonadotropic hypogonadism 14 with or without anosmia, 614858
WDR19	132.1	99.8	98.1	?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307
WDR26	97.5	98.3	94.6	Skraban-Deardorff syndrome, 617616
WDR34	106.6	99.5	96.2	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
WDR35	145.1	99.3	97.7	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091
WDR36	112.3	97.7	90.1	Glaucoma 1, open angle, G, 609887
WDR45	75	97.4	90.1	Neurodegeneration with brain iron accumulation 5, 300894
WDR45B	85.5	95.8	85.6	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977
WDR60	114.2	99.1	96.3	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503

WDR62	161.5	100	99.7	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
WDR66	141	100	99.9	Spermatogenic failure 33, 618152
WDR72	132.2	96.5	95.4	Amelogenesis imperfecta, type IIA3, 613211
WDR73	138.9	100	100	Galloway-Mowat syndrome 1, 251300
WDR81	163.3	99.9	99.4	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185 Hydrocephalus, congenital, 3, with brain anomalies, 617967
WEE2	102.2	99.6	95.7	Oocyte maturation defect 5, 617996
WFS1	251.4	100	99.7	?Cataract 41, 116400 Deafness, autosomal dominant 6/14/38, 600965 Wolfram syndrome 1, 222300 Wolfram-like syndrome, autosomal dominant, 614296 {Diabetes mellitus, noninsulin-dependent, association with}, 125853
WHRN	114	99.8	98.8	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
WIPF1	77.5	100	99.2	?Wiskott-Aldrich syndrome 2, 614493
WISP3	118.4	100	100	Arthropathy, progressive pseudorheumatoid, of childhood, 208230 Spondyloepiphyseal dysplasia tarda with progressive arthropathy, 208230
WNK1	167.7	99.9	99.5	Neuropathy, hereditary sensory and autonomic, type II, 201300 Pseudohypoaldosteronism, type IIC, 614492
WNK4	123.1	99.7	98.7	Pseudohypoaldosteronism, type IIB, 614491
WNT1	188.8	100	99.9	Osteogenesis imperfecta, type XV, 615220 {Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221
WNT10A	114	100	99.1	Odontoonychodermal dysplasia, 257980 Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400
WNT10B	144.7	100	99.9	Split-hand/foot malformation 6, 225300 Tooth agenesis, selective, 8, 617073
WNT2B	170.5	95.9	90	Diarrhea 9, 618168
WNT3	166.8	100	99.6	?Tetra-amelia syndrome 1, 273395
WNT4	263.1	93.4	92.7	?SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330
WNT5A	155.7	100	100	Robinow syndrome, autosomal dominant 1, 180700

WNT7A	216.8	100	100	Fuhrmann syndrome, 228930 Ulna and fibula, absence of, with severe limb deficiency, 276820
WRAP53	154.4	100	100	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	123.6	98.3	94.6	Werner syndrome, 277700
WT1	76.5	91.8	81.4	Denys-Drash syndrome, 194080 Frasier syndrome, 136680 Meacham syndrome, 608978 Mesothelioma, somatic, 156240 Nephrotic syndrome, type 4, 256370 Wilms tumor, type 1, 194070
WWOX	130.9	100	99.7	Epileptic encephalopathy, early infantile, 28, 616211 Esophageal squamous cell carcinoma, somatic, 133239 Spinocerebellar ataxia, autosomal recessive 12, 614322
XDH	109.1	100	99.9	Xanthinuria, type I, 278300
XIAP	107.1	91.9	86.6	Lymphoproliferative syndrome, X-linked, 2, 300635
XIST	NC	NC	NC	X-inactivation, familial skewed, 300087
XK	96.8	99.9	99.1	McLeod syndrome with or without chronic granulomatous disease, 300842
XPA	52.9	98.5	88.9	Xeroderma pigmentosum, group A, 278700
XPC	140.7	100	99.7	Xeroderma pigmentosum, group C, 278720
XPNPEP3	134	100	99.2	Nephronophthisis-like nephropathy 1, 613159
XPR1	131.7	100	99.8	Basal ganglia calcification, idiopathic, 6, 616413
XRCC1	106.6	99.7	97.5	?Spinocerebellar ataxia, autosomal recessive 26, 617633
XRCC2	165.6	93	89.4	?Fanconi anemia, complementation group U, 617247
XRCC4	103.2	99.7	97.3	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	132.5	90.4	87.1	Desbuquois dysplasia 2, 615777 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
XYLT2	136.3	98.9	94.9	Spondyloocular syndrome, 605822 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
YAP1	95.6	87.8	81.6	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433
YARS	122.4	100	100	Charcot-Marie-Tooth disease, dominant intermediate C, 608323
YARS2	173.2	99.8	98.9	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561

YME1L1	105.3	97.7	91.9	?Optic atrophy 11, 617302
YWHAG	226.6	100	100	Epileptic encephalopathy, early infantile, 56, 617665
YY1	134.8	100	98.6	Gabriele-de Vries syndrome, 617557
YY1AP1	159.2	98.3	97	Grange syndrome, 602531
ZAP70	185.6	99.9	99.5	Autoimmune disease, multisystem, infantile-onset, 2, 617006 Immunodeficiency 48, 269840
ZBTB16	151.4	100	100	Leukemia, acute promyelocytic, PL2F/RARA type, 0 Skeletal defects, genital hypoplasia, and mental retardation, 612447
ZBTB18	222.7	99.7	99	Mental retardation, autosomal dominant 22, 612337
ZBTB20	216.9	100	100	Primrose syndrome, 259050
ZBTB24	178.1	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069
ZBTB42	125.6	100	100	?Lethal congenital contracture syndrome 6, 616248
ZC3H14	184.1	99.6	97.2	Mental retardation, autosomal recessive 56, 617125
ZC4H2	78.6	99.8	98.1	Wieacker-Wolff syndrome, 314580
ZDHHC15	89.8	97.9	92.6	?Mental retardation, X-linked 91, 300577
ZDHHC9	55.5	98.4	89.4	Mental retardation, X-linked syndromic, Raymond type, 300799
ZEB1	192.7	100	99.3	Corneal dystrophy, Fuchs endothelial, 6, 613270 Corneal dystrophy, posterior polymorphous, 3, 609141
ZEB2	157	99.8	98.8	Mowat-Wilson syndrome, 235730
ZFHX2	119	99.9	99.2	?Marsili syndrome, 147430
ZFP57	104.4	99.8	98.7	Diabetes mellitus, transient neonatal, 1, 601410
ZFPM2	196.3	100	99.6	46XY sex reversal 9, 616067 Diaphragmatic hernia 3, 610187 Tetralogy of Fallot, 187500
ZFYVE26	120.3	99.9	99.4	Spastic paraplegia 15, autosomal recessive, 270700
ZFYVE27	118.3	100	100	Spastic paraplegia 33, autosomal dominant, 610244
ZIC1	231.1	100	100	Craniosynostosis 6, 616602
ZIC2	122.5	90.5	78.9	Holoprosencephaly 5, 609637
ZIC3	113.7	100	99.8	Congenital heart defects, nonsyndromic, 1, X-linked, 306955 Heterotaxy, visceral, 1, X-linked, 306955 VACTERL association, X-linked, 314390
ZMPSTE24	113.3	100	99.1	Mandibuloacral dysplasia with type B lipodystrophy, 608612 Restrictive dermopathy, lethal, 275210

ZMYND10	136.5	100	100	Ciliary dyskinesia, primary, 22, 615444
ZMYND11	137	100	99.8	Mental retardation, autosomal dominant 30, 616083
ZMYND15	129.6	99.5	96	?Spermatogenic failure 14, 615842
ZNF141	160.6	100	99.9	?Polydactyly, postaxial, type A6, 615226
ZNF148	190.8	99.8	98.8	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260
ZNF335	139.9	99.6	98.9	?Microcephaly 10, primary, autosomal recessive, 615095
ZNF408	135.7	100	100	?Exudative vitreoretinopathy 6, 616468 Retinitis pigmentosa 72, 616469
ZNF423	250.8	100	100	Joubert syndrome 19, 614844 Nephronophthisis 14, 614844
ZNF469	93.1	98.7	96.3	Brittle cornea syndrome 1, 229200
ZNF513	110.7	100	99.7	?Retinitis pigmentosa 58, 613617
ZNF644	156.3	100	99.8	Myopia 21, autosomal dominant, 614167
ZNF687	164.7	100	100	Paget disease of bone 6, 616833
ZNF711	137.7	98.7	95.5	Mental retardation, X-linked 97, 300803
ZNF750	150.3	100	99.9	Seborrhea-like dermatitis with psoriasiform elements, 610227
ZNHIT3	121	74.4	74.4	PEHO syndrome, 260565
ZP1	196	100	100	Oocyte maturation defect 1, 615774
ZP3	181.7	100	100	Oocyte maturation defect 3, 617712
ZSWIM6	150.9	93.1	89.1	Acromelic frontonasal dysostosis, 603671 Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan;43(Database issue):D1079-85.
Median Coverage describes the average number of reads seen across 50 exomes.
% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : December 31st, 2018.
This list is accurate for panel version DG 2.15
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors