MENDELIOME GENE PANEL DG 2.17 (3839 genes)

Releasedate: 06-12-2019

Gene	*Median Coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
A4GALT	194.8	100.0%	100.0%	NOR polyagglutination syndrome, 111400
AAAS	109.1	100.0%	99.8%	Achalasia-addisonianism-alacrimia syndrome, 231550
AAGAB	132.8	100.0%	100.0%	Keratoderma, palmoplantar, punctate type IA, 148600
AARS	109.2	100.0%	99.7%	Epileptic encephalopathy, early infantile, 29, 616339 Charcot-Marie-Tooth disease, axonal, type 2N, 613287
AARS2	135.3	100.0%	100.0%	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889
AASS	131.3	100.0%	99.6%	Hyperlysinemia, 238700
ABAT	86.1	99.9%	98.4%	GABA-transaminase deficiency, 613163
ABCA1	99.2	99.9%	98.7%	Tangier disease, 205400 HDL deficiency, familial, 1, 604091
ABCA12	129.0	99.6%	98.3%	Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500 Ichthyosis, congenital, autosomal recessive 4A, 601277
ABCA3	130.9	100.0%	99.7%	Surfactant metabolism dysfunction, pulmonary, 3, 610921
ABCA4	109.9	99.9%	99.1%	Retinal dystrophy, early-onset severe, 248200 Stargardt disease 1, 248200 Fundus flavimaculatus, 248200 Cone-rod dystrophy 3, 604116 Retinitis pigmentosa 19, 601718
ABCA5	69.0	97.7%	89.1%	?Hypertrichosis, congenital generalized, with gingival hyperplasia, 135400
ABCB11	135.9	100.0%	99.4%	Cholestasis, progressive familial intrahepatic 2, 601847 Cholestasis, benign recurrent intrahepatic, 2, 605479
ABCB4	125.0	100.0%	99.1%	Gallbladder disease 1, 600803 Cholestasis, intrahepatic, of pregnancy, 3, 614972 Cholestasis, progressive familial intrahepatic 3, 602347
ABCB6	136.7	100.0%	99.9%	Dyschromatosis universalis hereditaria 3, 615402 Microphthalmia, isolated, with coloboma 7, 614497 Pseudohyperkalemia, familial, 2, due to red cell leak, 609153
ABCB7	125.6	99.9%	98.8%	Anemia, sideroblastic, with ataxia, 301310
ABCC2	113.1	100.0%	99.8%	Dubin-Johnson syndrome, 237500

ABCC6	116.6	93.7%	93.1%	Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850 Arterial calcification, generalized, of infancy, 2, 614473
ABCC8	134.7	100.0%	99.9%	Diabetes mellitus, permanent neonatal, 606176 Diabetes mellitus, noninsulin-dependent, 125853 Diabetes mellitus, transient neonatal 2, 610374 Hyperinsulinemic hypoglycemia, familial, 1, 256450 Hypoglycemia of infancy, leucine-sensitive, 240800
ABCC9	140.4	100.0%	99.9%	Hypertrichotic osteochondrodysplasia, 239850 Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569
ABCD1	95.7	77.7%	74.9%	Adrenomyeloneuropathy, adult, 300100 Adrenoleukodystrophy, 300100
ABCD3	106.9	99.6%	96.7%	?Bile acid synthesis defect, congenital, 5, 616278
ABCD4	139.9	99.9%	98.5%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	147.1	100.0%	99.9%	Sitosterolemia 2, 618666
ABCG8	146.0	99.9%	98.9%	Sitosterolemia 1, 210250
ABHD12	96.9	100.0%	99.5%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ABHD5	183.6	100.0%	100.0%	Chanarin-Dorfman syndrome, 275630
ABL1	170.5	100.0%	100.0%	Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 608232 Congenital heart defects and skeletal malformations syndrome, 617602
ACAD8	131.8	100.0%	99.9%	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	130.9	100.0%	98.8%	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADM	124.9	99.9%	99.0%	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	164.6	100.0%	100.0%	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	112.2	100.0%	99.1%	2-methylbutyrylglycinuria, 610006
ACADVL	125.2	99.9%	98.7%	VLCAD deficiency, 201475
ACAN	132.5	96.0%	90.1%	Short stature and advanced bone age, with or without early-onset osteoarthritis and/or osteochondritis dissecans, 165800 Spondyloepimetaphyseal dysplasia, aggrecan type, 612813 ?Spondyloepiphyseal dysplasia, Kimberley type, 608361
ACAT1	110.0	99.9%	97.1%	Alpha-methylacetoacetic aciduria, 203750
ACD	180.3	100.0%	100.0%	?Dyskeratosis congenita, autosomal dominant 6, 616553 ?Dyskeratosis congenita, autosomal recessive 7, 616553
ACE	129.3	100.0%	99.8%	Renal tubular dysgenesis, 267430
ACER3	112.4	100.0%	99.0%	?Leukodystrophy, progressive, early childhood-onset, 617762

ACO2	125.5	95.6%	90.3%	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559
ACOX1	129.5	100.0%	100.0%	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACOX2	117.5	100.0%	99.6%	Bile acid synthesis defect, congenital, 6, 617308
ACP4	102.5	98.0%	90.4%	Amelogenesis imperfecta, type IJ, 617297
ACP5	189.0	100.0%	99.9%	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACSF3	158.9	99.9%	99.4%	Combined malonic and methylmalonic aciduria, 614265
ACSL4	100.3	98.2%	92.7%	Mental retardation, X-linked 63, 300387
ACSL6	110.7	99.5%	97.9%	Myelodysplastic syndrome, 0 Myelogenous leukemia, acute, 0
ACTA1	106.8	99.9%	98.5%	Myopathy, actin, congenital, with cores, 161800 Nemaline myopathy 3, autosomal dominant or recessive, 161800 Myopathy, congenital, with fiber-type disproportion 1, 255310 Myopathy, actin, congenital, with excess of thin myofilaments, 161800 ?Myopathy, scapulohumeroperoneal, 616852
ACTA2	89.6	99.9%	98.6%	Aortic aneurysm, familial thoracic 6, 611788 Multisystemic smooth muscle dysfunction syndrome, 613834 Moyamoya disease 5, 614042
ACTB	92.6	100.0%	99.9%	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACTC1	118.4	100.0%	99.3%	Left ventricular noncompaction 4, 613424 Atrial septal defect 5, 612794 Cardiomyopathy, dilated, 1R, 613424 Cardiomyopathy, hypertrophic, 11, 612098
ACTG1	131.2	100.0%	100.0%	Baraitser-Winter syndrome 2, 614583 Deafness, autosomal dominant 20/26, 604717
ACTG2	102.5	99.4%	96.5%	Visceral myopathy, 155310
ACTL6B	144.9	100.0%	100.0%	Epileptic encephalopathy, early infantile, 76, 618468 Intellectual developmental disorder with severe speech and ambulation defects, 618470
ACTN1	140.2	100.0%	100.0%	Bleeding disorder, platelet-type, 15, 615193
ACTN2	134.8	100.0%	99.9%	Myopathy, distal, 6, adult onset, 618655 Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158 Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158 Myopathy, congenital with structured cores and Z-line abnormalities, 618654
ACTN4	143.2	100.0%	99.9%	Glomerulosclerosis, focal segmental, 1, 603278
ACVR1	139.8	100.0%	100.0%	Fibrodysplasia ossificans progressiva, 135100
ACVR1B	143.0	100.0%	99.8%	Pancreatic cancer, somatic, 0
ACVR2B	124.2	99.9%	98.3%	Heterotaxy, visceral, 4, autosomal, 613751

ACVRL1	125.9	100.0%	98.8%	Telangiectasia, hereditary hemorrhagic, type 2, 600376
ACY1	128.5	99.9%	99.1%	Aminoacylase 1 deficiency, 609924
ADA	111.3	100.0%	99.6%	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADA2	88.7	99.8%	97.9%	?Sneddon syndrome, 182410 Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688
ADAM10	117.5	94.7%	92.7%	Reticulate acropigmentation of Kitamura, 615537
ADAM17	117.8	99.7%	98.5%	?Inflammatory skin and bowel disease, neonatal, 1, 614328
ADAM22	133.4	100.0%	99.4%	?Epileptic encephalopathy, early infantile, 61, 617933
ADAM9	140.7	99.9%	98.1%	Cone-rod dystrophy 9, 612775
ADAMTS10	135.4	100.0%	99.9%	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS13	115.6	98.0%	96.0%	Thrombotic thrombocytopenic purpura, familial, 274150
ADAMTS17	118.3	98.7%	93.9%	Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTS18	134.6	100.0%	99.8%	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
ADAMTS2	136.7	100.0%	99.9%	Ehlers-Danlos syndrome, dermatosparaxis type, 225410
ADAMTS3	137.9	100.0%	99.9%	Hennekam lymphangiectasia-lymphedema syndrome 3, 618154
ADAMTSL2	126.3	99.1%	96.8%	Geleophysic dysplasia 1, 231050
ADAMTSL4	137.7	100.0%	99.8%	Ectopia lentis et pupillae, 225200 Ectopia lentis, isolated, autosomal recessive, 225100
ADAR	117.2	99.9%	99.4%	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
ADAT3	153.0	100.0%	100.0%	Mental retardation, autosomal recessive 36, 615286
ADCY1	146.1	97.7%	96.3%	?Deafness, autosomal recessive 44, 610154
ADCY5	144.1	98.7%	96.4%	Dyskinesia, familial, with facial myokymia, 606703
ADCY6	180.3	100.0%	100.0%	?Lethal congenital contracture syndrome 8, 616287
ADD3	145.1	100.0%	99.6%	Cerebral palsy, spastic quadriplegic, 3, 617008
ADGRE2	148.0	96.6%	96.1%	Vibratory urticaria, 125630
ADGRG1	159.1	100.0%	100.0%	Polymicrogyria, bilateral perisylvian, 615752 Polymicrogyria, bilateral frontoparietal, 606854
ADGRG2	81.9	97.4%	91.3%	Congenital bilateral absence of vas deferens, X-linked, 300985
ADGRG6	134.6	99.8%	98.8%	Lethal congenital contracture syndrome 9, 616503
ADGRV1	126.0	99.8%	98.3%	Usher syndrome, type 2C, 605472 ?Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472
ADIPOQ	127.9	100.0%	100.0%	Adiponectin deficiency, 612556
ADK	101.3	99.9%	97.3%	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADNP	198.1	100.0%	100.0%	Helsmoortel-van der Aa syndrome, 615873

ADPRHL2	177.4	100.0%	100.0%	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
ADRB2	110.3	100.0%	100.0%	Beta-2-adrenoreceptor agonist, reduced response to, 0
ADSL	147.2	99.2%	98.9%	Adenylosuccinase deficiency, 103050
ADSSL1	117.5	96.8%	89.5%	Myopathy, distal, 5, 617030
AEBP1	163.7	100.0%	100.0%	Ehlers-Danlos syndrome, classic-like, 2, 618000
AFF2	109.6	99.9%	98.8%	Mental retardation, X-linked, FRAXE type, 309548
AFF4	102.7	99.9%	98.7%	CHOPS syndrome, 616368
AFG3L2	100.8	95.7%	85.1%	Spastic ataxia 5, autosomal recessive, 614487 Spinocerebellar ataxia 28, 610246
AFP	105.4	97.0%	89.3%	Alpha-fetoprotein deficiency, 615969
AGA	144.3	100.0%	100.0%	Aspartylglucosaminuria, 208400
AGBL1	109.9	98.5%	98.3%	Corneal dystrophy, Fuchs endothelial, 8, 615523
AGBL5	111.4	100.0%	99.4%	Retinitis pigmentosa 75, 617023
AGK	109.6	99.6%	95.5%	Sengers syndrome, 212350 Cataract 38, autosomal recessive, 614691
AGL	141.9	100.0%	99.7%	Glycogen storage disease IIIb, 232400 Glycogen storage disease IIIa, 232400
AGPAT2	180.5	99.7%	97.0%	Lipodystrophy, congenital generalized, type 1, 608594
AGPS	74.8	100.0%	97.8%	Rhizomelic chondrodysplasia punctata, type 3, 600121
AGRN	174.0	99.0%	95.9%	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120
AGT	211.6	100.0%	100.0%	Renal tubular dysgenesis, 267430
AGTPBP1	114.3	99.4%	95.6%	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276
AGTR1	147.7	92.0%	91.9%	Renal tubular dysgenesis, 267430
AGXT	176.8	100.0%	100.0%	Hyperoxaluria, primary, type 1, 259900
AHCY	120.8	100.0%	98.5%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHDC1	172.9	99.9%	98.8%	Xia-Gibbs syndrome, 615829
AHI1	125.5	99.9%	97.6%	Joubert syndrome 3, 608629
AHR	181.2	100.0%	99.5%	?Retinitis pigmentosa 85, 618345
AHSG	173.3	100.0%	99.9%	?Alopecia-mental retardation syndrome 1, 203650
AICDA	141.1	100.0%	99.6%	Immunodeficiency with hyper-IgM, type 2, 605258
AIFM1	92.9	99.7%	96.5%	Cowchock syndrome, 310490 Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 Combined oxidative phosphorylation deficiency 6, 300816 Deafness, X-linked 5, 300614
AIMP1	80.4	99.1%	91.4%	Leukodystrophy, hypomyelinating, 3, 260600
AIMP2	126.1	97.0%	89.6%	Leukodystrophy, hypomyelinating, 17, 618006

AIP	150.5	100.0%	99.9%	Pituitary adenoma 1, multiple types, 102200 Pituitary adenoma predisposition, 102200
AIPL1	124.2	100.0%	100.0%	Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393 Cone-rod dystrophy, 604393
AIRE	113.3	100.0%	100.0%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AK1	149.9	100.0%	100.0%	Hemolytic anemia due to adenylate kinase deficiency, 612631
AK2	100.4	98.7%	94.5%	Reticular dysgenesis, 267500
AK7	117.6	99.9%	97.2%	?Spermatogenic failure 27, 617965
AKAP9	96.2	99.0%	96.3%	?Long QT syndrome 11, 611820
AKR1C2	137.7	95.9%	90.1%	46XY sex reversal 8, 614279
AKR1D1	92.8	99.2%	96.1%	Bile acid synthesis defect, congenital, 2, 235555
AKT1	168.9	100.0%	99.8%	Breast cancer, somatic, 114480 Cowden syndrome 6, 615109 Proteus syndrome, somatic, 176920 Ovarian cancer, somatic, 167000 Colorectal cancer, somatic, 114500
AKT2	176.9	100.0%	99.8%	Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900 Diabetes mellitus, type II, 125853
AKT3	79.2	98.6%	94.0%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937
AL032819.3	4.6	18.4%	1.9%	Diarrhea 11, malabsorptive, congenital, 618662
ALAD	101.8	99.4%	95.4%	Porphyria, acute hepatic, 612740
ALAS2	77.2	99.1%	95.5%	Protoporphyria, erythropoietic, X-linked, 300752 Anemia, sideroblastic, 1, 300751
ALB	157.8	100.0%	99.1%	Analbuminemia, 616000
ALDH18A1	116.9	100.0%	99.8%	Cutis laxa, autosomal recessive, type IIIA, 219150 Cutis laxa, autosomal dominant 3, 616603 Spastic paraplegia 9B, autosomal recessive, 616586 Spastic paraplegia 9A, autosomal dominant, 601162
ALDH1A3	110.3	100.0%	98.4%	Microphthalmia, isolated 8, 615113
ALDH2	136.6	100.0%	100.0%	Alcohol sensitivity, acute, 610251
ALDH3A2	116.9	95.3%	94.2%	Sjogren-Larsson syndrome, 270200
ALDH4A1	136.8	100.0%	99.8%	Hyperprolinemia, type II, 239510
ALDH5A1	95.5	99.6%	95.6%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	106.5	100.0%	99.4%	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	68.0	94.2%	86.7%	Epilepsy, pyridoxine-dependent, 266100
ALDOA	130.1	76.6%	75.2%	Glycogen storage disease XII, 611881

ALDOB	140.0	100.0%	99.1%	Fructose intolerance, hereditary, 229600
ALG1	51.3	53.6%	52.1%	Congenital disorder of glycosylation, type Ik, 608540
ALG11	132.1	96.8%	96.5%	Congenital disorder of glycosylation, type Ip, 613661
ALG12	169.5	100.0%	100.0%	Congenital disorder of glycosylation, type Ig, 607143
ALG13	77.7	98.6%	92.4%	Epileptic encephalopathy, early infantile, 36, 300884 ?Congenital disorder of glycosylation, type Is, 300884
ALG14	204.6	100.0%	100.0%	?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227
ALG2	112.6	100.0%	100.0%	Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 ?Congenital disorder of glycosylation, type Ii, 607906
ALG3	117.9	100.0%	100.0%	Congenital disorder of glycosylation, type Id, 601110
ALG6	98.1	98.9%	94.9%	Congenital disorder of glycosylation, type Ic, 603147
ALG8	118.5	96.8%	95.7%	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	114.8	100.0%	99.8%	Gillessen-Kaesbach-Nishimura syndrome, 263210 Congenital disorder of glycosylation, type Il, 608776
ALKBH8	109.3	100.0%	98.1%	Intellectual developmental disorder, autosomal recessive 71, 618504
ALMS1	178.0	100.0%	99.8%	Alstrom syndrome, 203800
ALOX12B	137.8	100.0%	100.0%	Ichthyosis, congenital, autosomal recessive 2, 242100
ALOXE3	137.6	100.0%	99.6%	Ichthyosis, congenital, autosomal recessive 3, 606545
ALPK3	126.5	99.7%	97.9%	Cardiomyopathy, familial hypertrophic 27, 618052
ALPL	168.4	99.9%	99.5%	Hypophosphatasia, adult, 146300 Odontohypophosphatasia, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500
ALS2	145.2	100.0%	99.9%	Primary lateral sclerosis, juvenile, 606353 Amyotrophic lateral sclerosis 2, juvenile, 205100 Spastic paralysis, infantile onset ascending, 607225
ALX1	139.5	100.0%	99.2%	?Frontonasal dysplasia 3, 613456
ALX3	148.9	91.7%	80.3%	Frontonasal dysplasia 1, 136760
ALX4	175.4	100.0%	100.0%	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597
AMACR	168.4	100.0%	100.0%	Bile acid synthesis defect, congenital, 4, 214950 Alpha-methylacyl-CoA racemase deficiency, 614307
AMBN	181.1	99.3%	95.8%	Amelogenesis imperfecta, type IF, 616270
AMELX	90.7	99.0%	93.8%	Amelogenesis imperfecta, type 1E, 301200
AMER1	106.2	99.8%	99.1%	Osteopathia striata with cranial sclerosis, 300373
AMH	97.3	100.0%	99.3%	Persistent Mullerian duct syndrome, type I, 261550

AMHR2	152.1	100.0%	99.4%	Persistent Mullerian duct syndrome, type II, 261550
AMMECR1	102.3	100.0%	99.6%	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
AMN	118.2	99.1%	93.0%	Megaloblastic anemia-1, Norwegian type, 261100
AMPD1	115.8	99.9%	98.7%	Myopathy due to myoadenylate deaminase deficiency, 615511
AMPD2	146.3	100.0%	100.0%	?Spastic paraplegia 63, 615686 Pontocerebellar hypoplasia, type 9, 615809
AMT	151.3	100.0%	100.0%	Glycine encephalopathy, 605899
AMTN	120.4	99.7%	98.1%	?Amelogenesis imperfecta, type IIIB, 617607
ANAPC1	74.5	59.5%	56.7%	Rothmund-Thomson syndrome, type 1, 618625
ANG	161.8	100.0%	100.0%	Amyotrophic lateral sclerosis 9, 611895
ANGPTL3	88.9	99.0%	95.0%	Hypobetalipoproteinemia, familial, 2, 605019
ANGPTL4	135.2	99.9%	98.0%	Plasma triglyceride level QTL, low, 615881
ANK1	143.8	100.0%	99.6%	Spherocytosis, type 1, 182900
ANK2	143.6	100.0%	100.0%	Cardiac arrhythmia, ankyrin-B-related, 600919 Long QT syndrome 4, 600919
ANK3	144.2	99.4%	99.2%	?Mental retardation, autosomal recessive, 37, 615493
ANKH	116.6	100.0%	100.0%	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000
ANKLE2	155.0	100.0%	99.9%	Microcephaly 16, primary, autosomal recessive, 616681
ANKRD11	131.8	99.6%	97.6%	KBG syndrome, 148050
ANKRD26	81.5	94.9%	89.1%	Thrombocytopenia 2, 188000
ANKS6	101.2	99.3%	96.5%	Nephronophthisis 16, 615382
ANLN	139.3	98.6%	97.0%	Focal segmental glomerulosclerosis 8, 616032
ANO10	106.0	98.6%	96.5%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANO3	119.0	99.6%	97.5%	Dystonia 24, 615034
ANO5	126.7	99.7%	97.1%	Miyoshi muscular dystrophy 3, 613319 Gnathodiaphyseal dysplasia, 166260 Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307
ANO6	132.9	99.8%	98.5%	Scott syndrome, 262890
ANOS1	78.3	91.4%	87.3%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
ANTXR1	112.9	99.3%	97.3%	GAPO syndrome, 230740
ANTXR2	117.5	99.8%	97.7%	Hyaline fibromatosis syndrome, 228600
ANXA11	93.3	99.9%	98.9%	Amytrophic lateral sclerosis 23, 617839
AP1S1	105.8	100.0%	99.9%	MEDNIK syndrome, 609313
AP1S2	50.8	75.6%	68.1%	Mental retardation, X-linked syndromic 5, 304340
AP2M1	115.2	100.0%	99.9%	Intellectual developmental disorder 60 with seizures, 618587
AP2S1	120.3	90.4%	89.6%	Hypocalciuric hypercalcemia, type III, 600740

AP3B1	108.2	99.4%	95.7%	Hermansky-Pudlak syndrome 2, 608233
AP3B2	135.0	99.8%	97.9%	Epileptic encephalopathy, early infantile, 48, 617276
AP3D1	135.1	98.5%	97.9%	?Hermansky-Pudlak syndrome 10, 617050
AP4B1	124.8	99.9%	98.7%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	103.1	100.0%	99.0%	Stuttering, familial persistent, 1, 184450 Spastic paraplegia 51, autosomal recessive, 613744
AP4M1	140.7	99.9%	98.6%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	66.4	77.7%	70.3%	Spastic paraplegia 52, autosomal recessive, 614067
AP5Z1	136.8	100.0%	100.0%	Spastic paraplegia 48, autosomal recessive, 613647
APC	143.0	100.0%	99.6%	Desmoid disease, hereditary, 135290 Adenomatous polyposis coli, 175100 Gardner syndrome, 175100 Hepatoblastoma, somatic, 114550 Colorectal cancer, somatic, 114500 Brain tumor-polyposis syndrome 2, 175100 Gastric cancer, somatic, 613659 Adenoma, periampullary, somatic, 0
APC2	142.2	100.0%	99.6%	?Sotos syndrome 3, 617169 Cortical dysplasia, complex, with other brain malformations 10, 618677
APCDD1	182.1	100.0%	99.5%	Hypotrichosis 1, 605389
APOA1	164.8	100.0%	100.0%	Hypoalphalipoproteinemia, primary, 2, with or without corneal clouding, 618463 Amyloidosis, 3 or more types, 105200 ApoA-I and apoC-III deficiency, combined, 618463
APOA2	92.3	89.8%	82.2%	Apolipoprotein A-II deficiency, 0
APOA5	212.1	100.0%	100.0%	Hyperchylomicronemia, late-onset, 144650
APOB	158.4	100.0%	99.8%	Hypobetalipoproteinemia, 615558 Hypercholesterolemia, familial, 2, 144010
APOC2	112.2	100.0%	100.0%	Hyperlipoproteinemia, type Ib, 207750
APOC3	106.5	100.0%	100.0%	Apolipoprotein C-III deficiency, 614028
APOE	95.2	100.0%	100.0%	Hyperlipoproteinemia, type III, 617347 Lipoprotein glomerulopathy, 611771 Sea-blue histiocyte disease, 269600 Alzheimer disease 2, 104310
APOPT1	80.4	82.1%	82.1%	Mitochondrial complex IV deficiency, 220110
APP	109.6	100.0%	99.8%	Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants, 605714 Alzheimer disease 1, familial, 104300
APRT	105.5	100.0%	100.0%	Adenine phosphoribosyltransferase deficiency, 614723

APTX	99.2	94.5%	91.6%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
AQP2	142.2	100.0%	100.0%	Diabetes insipidus, nephrogenic, 125800
AQP5	136.8	100.0%	99.9%	Palmoplantar keratoderma, Bothnian type, 600231
AR	97.8	98.9%	95.3%	Hypospadias 1, X-linked, 300633 Androgen insensitivity, 300068 Androgen insensitivity, partial, with or without breast cancer, 312300 Spinal and bulbar muscular atrophy of Kennedy, 313200
ARCN1	142.5	96.8%	96.6%	Short stature, rhizomelic, with microcephaly, micrognathia, and developmental delay, 617164
ARF1	180.4	100.0%	99.9%	Periventricular nodular heterotopia 8, 618185
ARFGEF2	129.2	99.9%	98.9%	Periventricular heterotopia with microcephaly, 608097
ARG1	158.2	100.0%	100.0%	Argininemia, 207800
ARHGAP26	128.6	100.0%	99.9%	Leukemia, juvenile myelomonocytic, somatic, 607785
ARHGAP31	151.5	99.9%	99.1%	Adams-Oliver syndrome 1, 100300
ARHGDIA	226.3	100.0%	99.9%	Nephrotic syndrome, type 8, 615244
ARHGEF1	117.1	100.0%	99.6%	?Immunodeficiency 62, 618459
ARHGEF10	126.8	99.8%	98.4%	?Slowed nerve conduction velocity, AD, 608236
ARHGEF18	153.5	99.9%	98.7%	Retinitis pigmentosa 78, 617433
ARHGEF2	122.3	100.0%	99.9%	?Neurodevelopmental disorder with midbrain and hindbrain malformations, 617523
ARHGEF9	52.4	76.3%	72.8%	Epileptic encephalopathy, early infantile, 8, 300607
ARID1A	145.9	99.7%	98.9%	Coffin-Siris syndrome 2, 614607
ARID1B	150.6	99.5%	99.3%	Coffin-Siris syndrome 1, 135900
ARID2	158.2	99.9%	98.5%	Coffin-Siris syndrome 6, 617808
ARL13B	98.7	100.0%	99.7%	Joubert syndrome 8, 612291
ARL2BP	66.5	92.9%	83.8%	Retinitis pigmentosa with or without situs inversus, 615434
ARL3	76.1	99.9%	97.6%	Joubert syndrome 35, 618161 Retinitis pigmentosa 83, 618173
ARL6	91.8	99.9%	97.7%	?Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151
ARL6IP1	64.2	96.1%	77.0%	?Spastic paraplegia 61, autosomal recessive, 615685
ARMC2	132.2	99.8%	98.3%	Spermatogenic failure 38, 618433
ARMC4	107.7	94.4%	93.5%	Ciliary dyskinesia, primary, 23, 615451
ARMC5	194.5	100.0%	99.9%	ACTH-independent macronodular adrenal hyperplasia 2, 615954
ARMC9	129.0	100.0%	99.4%	Joubert syndrome 30, 617622
ARNT2	127.7	100.0%	100.0%	?Webb-Dattani syndrome, 615926
ARPC1B	150.6	100.0%	100.0%	Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease, 617718
ARR3	87.8	99.9%	99.6%	Myopia 26, X-linked, female-limited, 301010
ARSA	154.9	100.0%	100.0%	Metachromatic leukodystrophy, 250100

ARSB	111.3	100.0%	99.4%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARSE	83.5	98.5%	91.1%	Chondrodysplasia punctata, X-linked recessive, 302950
ARSG	120.4	100.0%	98.7%	Usher syndrome, type IV, 618144
ARV1	108.7	100.0%	99.7%	Epileptic encephalopathy, early infantile, 38, 617020
ARX	58.2	90.9%	83.3%	Proud syndrome, 300004 Lissencephaly, X-linked 2, 300215 Partington syndrome, 309510 Epileptic encephalopathy, early infantile, 1, 308350 Mental retardation, X-linked 29 and others, 300419 Hydranencephaly with abnormal genitalia, 300215
ASAH1	124.7	99.6%	96.8%	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASB10	123.4	99.9%	98.6%	Glaucoma 1, open angle, F, 603383
ASCC1	122.4	95.7%	92.1%	Barrett esophagus/esophageal adenocarcinoma, 614266 ?Spinal muscular atrophy with congenital bone fractures 2, 616867
ASCL1	334.2	100.0%	100.0%	Haddad syndrome, 209880 Central hypoventilation syndrome, congenital, 209880
ASH1L	146.8	98.7%	98.4%	Mental retardation, autosomal dominant 52, 617796
ASL	135.7	99.9%	99.2%	Argininosuccinic aciduria, 207900
ASNS	81.9	97.9%	91.0%	Asparagine synthetase deficiency, 615574
ASPA	118.0	99.9%	96.9%	Canavan disease, 271900
ASPH	109.3	100.0%	98.9%	Traboulsi syndrome, 601552
ASPM	111.3	99.7%	97.8%	Microcephaly 5, primary, autosomal recessive, 608716
ASPSCR1	137.0	100.0%	99.9%	Alveolar soft-part sarcoma, 606243
ASS1	106.1	95.4%	88.7%	Citrullinemia, 215700
ASXL1	141.0	100.0%	99.6%	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ASXL2	150.0	99.4%	98.2%	Shashi-Pena syndrome, 617190
ASXL3	141.2	99.8%	99.1%	Bainbridge-Ropers syndrome, 615485
ATAD1	63.3	98.8%	91.0%	Hyperekplexia 4, 618011
ATAD3A	100.8	93.8%	88.8%	Harel-Yoon syndrome, 617183
ATCAY	164.9	100.0%	99.7%	Ataxia, cerebellar, Cayman type, 601238
ATF6	125.0	100.0%	99.4%	Achromatopsia 7, 616517
ATG5	119.6	99.1%	94.3%	?Spinocerebellar ataxia, autosomal recessive 25, 617584
ATIC	114.9	100.0%	99.9%	AICA-ribosiduria due to ATIC deficiency, 608688
ATL1	136.1	100.0%	99.4%	Spastic paraplegia 3A, autosomal dominant, 182600 Neuropathy, hereditary sensory, type ID, 613708

ATL3	115.9	99.7%	97.4%	Neuropathy, hereditary sensory, type IF, 615632
ATM	108.4	99.6%	96.8%	Ataxia-telangiectasia, 208900 Lymphoma, mantle cell, somatic, 0 Lymphoma, B-cell non-Hodgkin, somatic, 0 T-cell prolymphocytic leukemia, somatic, 0
ATN1	175.9	99.9%	99.2%	Dentatorubral-pallidoluysian atrophy, 125370 Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494
ATOH7	210.8	99.8%	99.2%	Persistent hyperplastic primary vitreous, autosomal recessive, 221900
ATP11C	75.7	98.7%	91.5%	?Hemolytic anemia, congenital, X-linked, 301015
ATP13A2	149.6	100.0%	99.8%	Kufor-Rakeb syndrome, 606693 Spastic paraplegia 78, autosomal recessive, 617225
ATP1A1	114.1	100.0%	99.7%	Charcot-Marie-Tooth disease, axonal, type 2DD, 618036 Hypomagnesemia, seizures, and mental retardation 2, 618314
ATP1A2	173.7	100.0%	99.8%	Migraine, familial hemiplegic, 2, 602481 Migraine, familial basilar, 602481 Alternating hemiplegia of childhood 1, 104290
ATP1A3	173.9	100.0%	100.0%	CAPOS syndrome, 601338 Alternating hemiplegia of childhood 2, 614820 Dystonia-12, 128235
ATP2A1	157.9	100.0%	100.0%	Brody myopathy, 601003
ATP2A2	150.4	100.0%	99.9%	Acrokeratosis verruciformis, 101900 Darier disease, 124200
ATP2B3	133.9	99.8%	98.5%	?Spinocerebellar ataxia, X-linked 1, 302500
ATP2C1	109.2	100.0%	99.5%	Hailey-Hailey disease, 169600
ATP5A1	73.8	93.7%	85.2%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4, 615228 ?Combined oxidative phosphorylation deficiency 22, 616045
ATP5D	118.5	99.2%	93.8%	Mitochondrial complex V (ATP synthase) deficiency, 618120
ATP5E	146.3	100.0%	100.0%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053
ATP6AP1	113.6	99.8%	97.7%	Immunodeficiency 47, 300972
ATP6AP2	45.5	89.4%	67.3%	Mental retardation, X-linked, syndromic, Hedera type, 300423 ?Parkinsonism with spasticity, X-linked, 300911
ATP6V0A2	120.5	100.0%	99.6%	Wrinkly skin syndrome, 278250 Cutis laxa, autosomal recessive, type IIA, 219200
ATP6V0A4	108.1	100.0%	99.2%	Renal tubular acidosis, distal, autosomal recessive, 602722
ATP6V1A	133.1	99.8%	97.3%	Epileptic encephalopathy, infantile or early childhood, 3, 618012 Cutis laxa, autosomal recessive, type IID, 617403
ATP6V1B1	184.8	100.0%	100.0%	Renal tubular acidosis with deafness, 267300

ATP6V1B2	125.1	99.9%	99.1%	Zimmermann-Laband syndrome 2, 616455 Deafness, congenital, with onychodystrophy, autosomal dominant, 124480
ATP6V1E1	67.0	91.8%	86.3%	Cutis laxa, autosomal recessive, type IIC, 617402
ATP7A	109.4	99.7%	97.2%	Occipital horn syndrome, 304150 Menkes disease, 309400 Spinal muscular atrophy, distal, X-linked 3, 300489
ATP7B	137.1	99.9%	99.3%	Wilson disease, 277900
ATP8A2	118.4	100.0%	99.7%	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268
ATP8B1	115.1	98.0%	95.0%	Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, progressive familial intrahepatic 1, 211600 Cholestasis, benign recurrent intrahepatic, 243300
ATPAF2	109.2	100.0%	100.0%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
ATR	142.1	99.9%	98.9%	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
ATRX	86.2	99.1%	95.1%	Alpha-thalassemia/mental retardation syndrome, 301040 Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Mental retardation-hypotonic facies syndrome, X-linked, 309580
ATXN1	190.1	100.0%	100.0%	Spinocerebellar ataxia 1, 164400
ATXN10	136.2	100.0%	99.6%	Spinocerebellar ataxia 10, 603516
ATXN2	84.8	94.8%	90.4%	Spinocerebellar ataxia 2, 183090
ATXN3	92.1	91.7%	88.1%	Machado-Joseph disease, 109150
ATXN7	117.4	99.8%	98.5%	Spinocerebellar ataxia 7, 164500
ATXN8OS	NC	NC	NC	Spinocerebellar ataxia 8, 608768
AUH	135.7	100.0%	99.8%	3-methylglutaconic aciduria, type I, 250950
AURKC	72.7	99.5%	93.6%	Spermatogenic failure 5, 243060
AUTS2	143.1	99.8%	98.7%	Mental retardation, autosomal dominant 26, 615834
AVIL	119.0	100.0%	99.7%	Nephrotic syndrome, type 21, 618594
AVP	73.9	99.9%	90.5%	Diabetes insipidus, neurohypophyseal, 125700
AVPR2	150.2	100.0%	99.8%	Nephrogenic syndrome of inappropriate antidiuresis, 300539 Diabetes insipidus, nephrogenic, 304800
AXIN1	149.4	99.7%	98.3%	Hepatocellular carcinoma, somatic, 114550 ?Caudal duplication anomaly, 607864
AXIN2	134.4	100.0%	99.9%	Colorectal cancer, somatic, 114500 Oligodontia-colorectal cancer syndrome, 608615
B2M	194.6	100.0%	100.0%	Immunodeficiency 43, 241600 ?Amyloidosis, familial visceral, 105200
B3GALNT2	94.8	93.1%	91.1%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181

B3GALT6	96.5	87.5%	80.2%	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B3GAT3	134.2	99.9%	97.0%	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B3GLCT	97.3	100.0%	99.7%	Peters-plus syndrome, 261540
B4GALNT1	164.9	99.9%	98.3%	Spastic paraplegia 26, autosomal recessive, 609195
B4GALT1	121.2	99.7%	97.4%	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	138.9	100.0%	99.1%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
B4GAT1	153.5	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
B9D1	111.4	92.2%	92.2%	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120
B9D2	115.7	100.0%	100.0%	Joubert syndrome 34, 614175 ?Meckel syndrome 10, 614175
BAAT	110.3	99.7%	97.9%	Hypercholanemia, familial, 607748
BACH2	173.7	100.0%	99.8%	Immunodeficiency 60, 618394
BAG3	189.4	100.0%	99.9%	Cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954
BANF1	52.9	97.3%	86.5%	Nestor-Guillermo progeria syndrome, 614008
BAP1	113.0	85.4%	83.2%	Tumor predisposition syndrome, 614327
BAX	115.4	99.9%	98.6%	T-cell acute lymphoblastic leukemia, somatic, 613065 Colorectal cancer, somatic, 114500
BBIP1	116.1	98.5%	91.4%	?Bardet-Biedl syndrome 18, 615995
BBS1	156.1	100.0%	100.0%	Bardet-Biedl syndrome 1, 209900
BBS10	156.7	100.0%	100.0%	Bardet-Biedl syndrome 10, 615987
BBS12	193.6	100.0%	100.0%	Bardet-Biedl syndrome 12, 615989
BBS2	153.3	100.0%	99.7%	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	113.2	100.0%	98.4%	Bardet-Biedl syndrome 4, 615982
BBS5	94.9	98.4%	92.3%	Bardet-Biedl syndrome 5, 615983
BBS7	136.8	99.0%	95.3%	Bardet-Biedl syndrome 7, 615984
BBS9	113.2	98.8%	94.8%	Bardet-Biedl syndrome 9, 615986
BCAP31	78.0	93.9%	81.2%	Deafness, dystonia, and cerebral hypomyelination, 300475
BCHE	179.3	100.0%	100.0%	Butyrylcholinesterase deficiency, 617936
BCKDHA	193.6	100.0%	99.8%	Maple syrup urine disease, type Ia, 248600
BCKDHB	122.4	97.8%	90.2%	Maple syrup urine disease, type Ib, 248600
BCKDK	223.0	100.0%	100.0%	Branched-chain ketoacid dehydrogenase kinase deficiency, 614923

BCL10	127.1	100.0%	100.0%	?Immunodeficiency 37, 616098 Lymphoma, MALT, somatic, 137245
BCL11A	160.4	99.6%	98.0%	Dias-Logan syndrome, 617101
BCL11B	147.9	100.0%	99.3%	Immunodeficiency 49, 617237 Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092
BCL2	224.2	100.0%	100.0%	Leukemia/lymphoma, B-cell, 2, 0
BCL7A	163.1	100.0%	100.0%	B-cell non-Hodgkin lymphoma, high-grade, 0
BCO1	135.0	100.0%	100.0%	?Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300
BCOR	109.0	99.2%	96.2%	Microphthalmia, syndromic 2, 300166
BCORL1	162.3	99.9%	98.5%	Shukla-Vernon syndrome, 301029
BCS1L	160.0	100.0%	100.0%	Leigh syndrome, 256000 GRACILE syndrome, 603358 Bjornstad syndrome, 262000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BDP1	136.2	98.4%	94.0%	?Deafness, autosomal recessive 112, 618257
BEAN1	158.6	99.9%	98.2%	Spinocerebellar ataxia 31, 117210
BEST1	137.3	99.9%	98.2%	Retinitis pigmentosa-50, 613194 Bestrophinopathy, autosomal recessive, 611809 Retinitis pigmentosa, concentric, 613194 Vitreoretinochoroidopathy, 193220 Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma, 193220 Macular dystrophy, vitelliform, 2, 153700
BFSP1	105.8	100.0%	99.3%	Cataract 33, multiple types, 611391
BFSP2	106.9	100.0%	99.2%	Cataract 12, multiple types, 611597
BGN	147.7	100.0%	100.0%	Meester-Loeys syndrome, 300989 Spondyloepimetaphyseal dysplasia, X-linked, 300106
BHLHA9	41.5	97.8%	83.4%	Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432 ?Camptosynpolydactyly, complex, 607539
BICD2	166.3	100.0%	99.9%	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291 Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290
BIN1	125.8	100.0%	99.5%	Centronuclear myopathy 2, 255200
BLK	137.8	100.0%	100.0%	Maturity-onset diabetes of the young, type 11, 613375
BLM	111.3	99.9%	98.1%	Bloom syndrome, 210900
BLNK	93.3	96.9%	92.4%	?Agammaglobulinemia 4, 613502
BLOC1S3	79.6	100.0%	100.0%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	103.0	99.3%	92.1%	?Hermansky-pudlak syndrome 9, 614171
BLVRA	117.6	100.0%	99.9%	Hyperbiliverdinemia, 614156

BMP1	167.3	100.0%	100.0%	Osteogenesis imperfecta, type XIII, 614856
BMP15	98.8	99.9%	98.7%	Ovarian dysgenesis 2, 300510 Premature ovarian failure 4, 300510
BMP2	180.6	100.0%	100.0%	Brachydactyly, type A2, 112600 Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 617877
BMP4	192.0	100.0%	100.0%	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625
BMPER	132.4	100.0%	99.6%	Diaphanospondylodysostosis, 608022
BMPR1A	81.3	99.8%	94.3%	Polyposis syndrome, hereditary mixed, 2, 610069 Polyposis, juvenile intestinal, 174900 Juvenile polyposis syndrome, infantile form, 174900
BMPR1B	141.6	100.0%	100.0%	Brachydactyly, type A2, 112600 Brachydactyly, type A1, D, 616849 Acromesomelic dysplasia, Demirhan type, 609441
BMPR2	155.6	99.9%	99.9%	Pulmonary venoocclusive disease 1, 265450 Pulmonary hypertension, familial primary, 1, with or without HHT, 178600 Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600
BMS1	79.8	66.8%	65.9%	?Aplasia cutis congenita, nonsyndromic, 107600
BNC2	136.3	99.1%	99.1%	Lower urinary tract obstruction, congenital, 618612
BOLA3	50.9	99.8%	94.6%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BPGM	105.8	100.0%	100.0%	Erythrocytosis, familial, 8, 222800
BPTF	144.9	96.6%	94.9%	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, 617755
BRAF	71.0	91.7%	79.4%	Noonan syndrome 7, 613706 Cardiofaciocutaneous syndrome, 115150 Adenocarcinoma of lung, somatic, 211980 LEOPARD syndrome 3, 613707 Nonsmall cell lung cancer, somatic, 0 Melanoma, malignant, somatic, 0 Colorectal cancer, somatic, 0
BRAT1	155.4	100.0%	99.6%	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498 Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056
BRCA1	164.4	99.2%	98.2%	Fanconi anemia, complementation group S, 617883
BRCA2	103.2	99.7%	98.8%	Wilms tumor, 194070 Fanconi anemia, complementation group D1, 605724
BRDT	105.2	96.4%	90.9%	?Spermatogenic failure 21, 617644
BRF1	120.2	99.9%	98.8%	Cerebellofaciodental syndrome, 616202
BRIP1	122.2	99.9%	98.6%	Fanconi anemia, complementation group J, 609054
BRPF1	174.8	100.0%	100.0%	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333

BRWD3	103.5	98.9%	95.1%	Mental retardation, X-linked 93, 300659
BSCL2	112.9	100.0%	99.9%	Lipodystrophy, congenital generalized, type 2, 269700 Silver spastic paraplegia syndrome, 270685 Neuropathy, distal hereditary motor, type VA, 600794 Encephalopathy, progressive, with or without lipodystrophy, 615924
BSND	150.8	100.0%	100.0%	Sensorineural deafness with mild renal dysfunction, 602522 Bartter syndrome, type 4a, 602522
BTD	135.6	100.0%	99.8%	Biotinidase deficiency, 253260
BTK	98.6	100.0%	99.1%	Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200 Agammaglobulinemia, X-linked 1, 300755
BUB1	127.7	99.9%	98.2%	Colorectal cancer with chromosomal instability, somatic, 114500
BUB1B	121.6	99.9%	99.0%	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300
BVES	109.4	100.0%	98.4%	Muscular dystrophy, limb-girdle, autosomal recessive 25, 616812
C11orf70	83.8	99.8%	95.3%	Ciliary dyskinesia, primary, 38, 618063
C11orf80	91.9	99.1%	96.8%	Hydatidiform mole, recurrent, 4, 618432
C12orf4	126.3	100.0%	99.7%	Mental retardation, autosomal recessive 66, 618221
C12orf57	159.8	100.0%	100.0%	Temtamy syndrome, 218340
C12orf65	112.4	100.0%	99.8%	Spastic paraplegia 55, autosomal recessive, 615035 Combined oxidative phosphorylation deficiency 7, 613559
C15orf41	121.9	100.0%	99.4%	Dyserythropoietic anemia, congenital, type Ib, 615631
C19orf12	117.5	100.0%	99.9%	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
C19orf70	84.4	99.9%	99.2%	Combined oxidative phosphorylation deficiency 37, 618329
C1GALT1C1	132.5	99.9%	99.0%	Tn polyagglutination syndrome, somatic, 300622
C1QA	222.4	100.0%	100.0%	C1q deficiency, 613652
C1QB	178.7	100.0%	99.9%	C1q deficiency, 613652
C1QBP	68.9	92.8%	81.2%	Combined oxidative phosphorylation deficiency 33, 617713
C1QC	209.9	100.0%	100.0%	C1q deficiency, 613652
C1QTNF5	173.0	97.8%	91.6%	Retinal degeneration, late-onset, autosomal dominant, 605670
C1R	161.6	100.0%	100.0%	Ehlers-Danlos syndrome, periodontal type, 1, 130080
C1S	101.7	99.9%	98.4%	Ehlers-Danlos syndrome, periodontal type, 2, 617174 C1s deficiency, 613783
C2	134.5	100.0%	100.0%	C2 deficiency, 217000
C21orf2	146.9	100.0%	99.4%	Spondylometaphyseal dysplasia, axial, 602271 Retinal dystrophy with macular staphyloma, 617547
C21orf59	144.1	99.3%	96.3%	Ciliary dyskinesia, primary, 26, 615500

C2CD3	121.4	95.8%	95.3%	Orofaciodigital syndrome XIV, 615948
C2orf71	128.5	100.0%	99.3%	Retinitis pigmentosa 54, 613428
C3	153.6	100.0%	99.7%	C3 deficiency, 613779
C4A	97.5	99.0%	97.4%	C4a deficiency, 614380
C4B	94.6	99.2%	97.8%	C4B deficiency, 614379
C4orf26	207.0	100.0%	100.0%	Amelogenesis imperfecta, type IIA4, 614832
C5	118.6	99.6%	97.6%	C5 deficiency, 609536
C5orf42	122.3	99.7%	97.4%	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
C6	139.6	100.0%	99.7%	C6 deficiency, 612446 Combined C6/C7 deficiency, 0
C7	113.0	99.9%	98.0%	C7 deficiency, 610102
C7orf43	161.4	100.0%	100.0%	?Microcephaly 25, primary, autosomal recessive, 618351
C8A	108.7	100.0%	99.5%	C8 deficiency, type I, 613790
C8B	108.4	99.9%	98.7%	C8 deficiency, type II, 613789
C8orf37	146.4	100.0%	99.9%	Retinitis pigmentosa 64, 614500 Bardet-Biedl syndrome 21, 617406 Cone-rod dystrophy 16, 614500
C9	120.8	100.0%	99.5%	C9 deficiency, 613825
C9orf72	95.8	99.7%	96.7%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1, 105550
CA12	107.7	100.0%	100.0%	Hyperchlorhidrosis, isolated, 143860
CA2	141.8	100.0%	99.9%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA4	175.5	100.0%	100.0%	Retinitis pigmentosa 17, 600852
CA5A	99.0	99.9%	97.2%	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CA8	112.7	99.3%	96.2%	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CABP2	85.6	80.9%	74.6%	Deafness, autosomal recessive 93, 614899
CABP4	168.9	100.0%	100.0%	Cone-rod synaptic disorder, congenital nonprogressive, 610427
CACNA1A	101.2	98.2%	96.2%	Spinocerebellar ataxia 6, 183086 Epileptic encephalopathy, early infantile, 42, 617106 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Episodic ataxia, type 2, 108500 Migraine, familial hemiplegic, 1, 141500
CACNA1B	147.4	99.8%	98.6%	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497
CACNA1C	151.7	99.9%	99.4%	Timothy syndrome, 601005 Long QT syndrome 8, 618447 Brugada syndrome 3, 611875

CACNA1D	135.3	98.0%	97.8%	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896
CACNA1E	129.3	100.0%	99.5%	Epileptic encephalopathy, early infantile, 69, 618285
CACNA1F	91.4	99.8%	97.7%	Cone-rod dystrophy, X-linked, 3, 300476 Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071 Aland Island eye disease, 300600
CACNA1G	165.6	100.0%	99.9%	Spinocerebellar ataxia 42, 616795 Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087
CACNA1H	157.1	99.5%	98.5%	Hyperaldosteronism, familial, type IV, 617027
CACNA1S	130.1	100.0%	99.7%	Hypokalemic periodic paralysis, type 1, 170400
CACNA2D2	139.2	96.6%	94.3%	Cerebellar atrophy with seizures and variable developmental delay, 618501
CACNA2D4	103.4	99.2%	97.4%	Retinal cone dystrophy 4, 610478
CACNB2	135.8	100.0%	99.4%	Brugada syndrome 4, 611876
CACNB4	98.5	97.3%	96.2%	Episodic ataxia, type 5, 613855
CACNG2	128.6	100.0%	99.5%	?Mental retardation, autosomal dominant 10, 614256
CAD	147.4	100.0%	99.6%	Epileptic encephalopathy, early infantile, 50, 616457
CALM1	95.2	99.8%	98.1%	Long QT syndrome 14, 616247 Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916
CALM2	43.0	66.5%	61.9%	Long QT syndrome 15, 616249
CALR	118.8	98.9%	92.5%	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
CAMK2A	125.8	100.0%	99.9%	?Mental retardation, autosomal recessive 63, 618095 Mental retardation, autosomal dominant 53, 617798
CAMK2B	122.1	100.0%	100.0%	Mental retardation, autosomal dominant 54, 617799
CAMK2G	114.9	99.9%	99.1%	Mental retardation, autosomal dominant 59, 618522
CAMTA1	197.4	100.0%	99.9%	Cerebellar ataxia, nonprogressive, with mental retardation, 614756
CANT1	158.4	100.0%	100.0%	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719
CAPN1	177.4	100.0%	100.0%	Spastic paraplegia 76, autosomal recessive, 616907
CAPN3	103.8	99.4%	97.3%	Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600 Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129
CAPN5	167.1	100.0%	100.0%	Vitreoretinopathy, neovascular inflammatory, 193235
CARD11	148.2	100.0%	99.8%	Immunodeficiency 11B with atopic dermatitis, 617638 B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11A, 615206
CARD14	136.0	100.0%	99.5%	Pityriasis rubra pilaris, 173200 Psoriasis 2, 602723

CARD9	152.2	100.0%	99.9%	Candidiasis, familial, 2, autosomal recessive, 212050
CARMIL2	153.2	99.5%	98.0%	Immunodeficiency 58, 618131
CARS2	138.8	100.0%	100.0%	Combined oxidative phosphorylation deficiency 27, 616672
CASK	84.5	99.4%	94.0%	Mental retardation, with or without nystagmus, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 FG syndrome 4, 300422
CASP10	111.4	99.9%	98.9%	Lymphoma, non-Hodgkin, somatic, 605027 Gastric cancer, somatic, 613659 Autoimmune lymphoproliferative syndrome, type II, 603909
CASP14	89.0	100.0%	99.9%	Ichthyosis, congenital, autosomal recessive 12, 617320
CASP8	133.1	95.6%	95.5%	?Autoimmune lymphoproliferative syndrome, type IIB, 607271 Hepatocellular carcinoma, somatic, 114550
CASQ1	98.5	100.0%	98.7%	Myopathy, vacuolar, with CASQ1 aggregates, 616231
CASQ2	113.9	100.0%	99.3%	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CASR	167.3	100.0%	99.8%	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 Hypocalciuric hypercalcemia, type I, 145980 Hypocalcemia, autosomal dominant, 601198 Hyperparathyroidism, neonatal, 239200
CAST	112.2	99.5%	97.0%	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295
CAT	141.9	100.0%	100.0%	Acatalasemia, 614097
CATSPER1	126.7	100.0%	99.7%	Spermatogenic failure 7, 612997
CAV1	200.0	100.0%	100.0%	Pulmonary hypertension, primary, 3, 615343 Lipodystrophy, familial partial, type 7, 606721 ?Lipodystrophy, congenital generalized, type 3, 612526
CAV3	233.9	100.0%	100.0%	Creatine phosphokinase, elevated serum, 123320 Long QT syndrome 9, 611818 Myopathy, distal, Tateyama type, 614321 Rippling muscle disease 2, 606072 Cardiomyopathy, familial hypertrophic, 192600
CAVIN1	200.3	100.0%	100.0%	Lipodystrophy, congenital generalized, type 4, 613327
CBL	131.1	97.4%	97.1%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
CBS	136.4	100.0%	99.3%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CBX2	168.8	100.0%	100.0%	?46XY sex reversal 5, 613080
CC2D1A	147.9	100.0%	99.8%	Mental retardation, autosomal recessive 3, 608443

CC2D2A	112.6	99.0%	97.0%	Meckel syndrome 6, 612284 Joubert syndrome 9, 612285 COACH syndrome, 216360
CCBE1	80.9	99.8%	98.6%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CCDC103	126.7	100.0%	100.0%	Ciliary dyskinesia, primary, 17, 614679
CCDC114	148.7	100.0%	99.9%	Ciliary dyskinesia, primary, 20, 615067
CCDC115	89.9	88.5%	87.0%	Congenital disorder of glycosylation, type IIo, 616828
CCDC151	141.6	100.0%	100.0%	Ciliary dyskinesia, primary, 30, 616037
CCDC174	126.5	99.3%	96.6%	Hypotonia, infantile, with psychomotor retardation, 616816
CCDC22	105.7	99.0%	95.5%	Ritscher-Schinzel syndrome 2, 300963
CCDC39	82.4	99.6%	96.0%	Ciliary dyskinesia, primary, 14, 613807
CCDC40	120.1	99.5%	98.5%	Ciliary dyskinesia, primary, 15, 613808
CCDC47	144.5	99.0%	96.4%	Trichohepatoneurodevelopmental syndrome, 618268
CCDC50	124.0	100.0%	99.8%	?Deafness, autosomal dominant 44, 607453
CCDC65	84.8	99.8%	97.8%	Ciliary dyskinesia, primary, 27, 615504
CCDC78	156.1	100.0%	100.0%	?Centronuclear myopathy 4, 614807
CCDC8	211.7	100.0%	100.0%	3-M syndrome 3, 614205
CCDC88A	90.3	99.3%	96.4%	?PEHO syndrome-like, 617507
CCDC88C	119.2	100.0%	99.7%	?Spinocerebellar ataxia 40, 616053 Hydrocephalus, congenital, 1, 236600
CCM2	143.5	99.2%	98.1%	Cerebral cavernous malformations-2, 603284
CCND2	147.6	100.0%	100.0%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938
CCNK	92.2	91.0%	87.3%	?Intellectual developmental disorder with hypertelorism and distinctive facies, 618147
CCNO	151.7	100.0%	99.9%	Ciliary dyskinesia, primary, 29, 615872
CCT5	123.2	100.0%	99.1%	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CD151	135.4	100.0%	100.0%	Nephropathy with pretibial epidermolysis bullosa and deafness, 609057
CD164	124.5	98.6%	93.9%	?Deafness, autosomal dominant 66, 616969
CD19	116.9	100.0%	100.0%	Immunodeficiency, common variable, 3, 613493
CD247	99.1	99.9%	99.2%	?Immunodeficiency 25, 610163
CD27	115.3	100.0%	99.9%	Lymphoproliferative syndrome 2, 615122
CD2AP	118.7	99.9%	98.7%	Glomerulosclerosis, focal segmental, 3, 607832
CD320	126.1	100.0%	100.0%	Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646
CD36	115.0	99.9%	99.0%	Platelet glycoprotein IV deficiency, 608404
CD3D	146.3	100.0%	99.9%	Immunodeficiency 19, 615617
CD3E	126.6	100.0%	99.6%	Immunodeficiency 18, SCID variant, 615615 Immunodeficiency 18, 615615
CD3G	137.2	100.0%	100.0%	Immunodeficiency 17, CD3 gamma deficient, 615607

CD4	128.9	100.0%	99.8%	OKT4 epitope deficiency, 613949
CD40	157.9	100.0%	100.0%	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	109.8	97.1%	88.0%	Immunodeficiency, X-linked, with hyper-IgM, 308230
CD55	133.3	95.6%	91.0%	Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300
CD59	153.6	94.1%	86.4%	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD70	117.0	100.0%	99.7%	Lymphoproliferative syndrome 3, 618261
CD79A	144.2	100.0%	99.1%	Agammaglobulinemia 3, 613501
CD79B	212.9	100.0%	100.0%	Agammaglobulinemia 6, 612692
CD81	174.0	100.0%	100.0%	Immunodeficiency, common variable, 6, 613496
CD8A	166.1	100.0%	100.0%	CD8 deficiency, familial, 608957
CD96	140.9	100.0%	99.4%	C syndrome, 211750
CDAN1	123.7	100.0%	99.8%	Dyserythropoietic anemia, congenital, type Ia, 224120
CDC14A	155.4	99.8%	97.3%	Deafness, autosomal recessive 32, with or without immotile sperm, 608653
CDC42	89.8	98.1%	90.1%	Takenouchi-Kosaki syndrome, 616737
CDC45	148.0	99.6%	98.4%	Meier-Gorlin syndrome 7, 617063
CDC6	142.2	100.0%	99.7%	?Meier-Gorlin syndrome 5, 613805
CDC73	111.0	100.0%	98.9%	Parathyroid adenoma with cystic changes, 145001 Hyperparathyroidism-jaw tumor syndrome, 145001 Parathyroid carcinoma, 608266 Hyperparathyroidism, familial primary, 145000
CDCA7	117.7	100.0%	99.7%	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910
CDH1	109.4	99.2%	99.1%	Endometrial carcinoma, somatic, 608089 Blepharocheilodontic syndrome 1, 119580 Gastric cancer, hereditary diffuse, with or without cleft lip and/or palate, 137215 Ovarian cancer, somatic, 167000
CDH11	131.7	100.0%	100.0%	Elsahy-Waters syndrome, 211380
CDH15	173.0	100.0%	99.9%	Mental retardation, autosomal dominant 3, 612580
CDH23	186.7	100.0%	100.0%	Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1D, 601067
CDH3	148.2	100.0%	99.9%	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 Hypotrichosis, congenital, with juvenile macular dystrophy, 601553
CDHR1	153.8	99.9%	99.2%	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660
CDK10	141.9	100.0%	100.0%	Al Kaissi syndrome, 617694
CDK13	133.2	100.0%	99.2%	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360
CDK5	118.7	100.0%	100.0%	?Lissencephaly 7 with cerebellar hypoplasia, 616342

CDK5RAP2	109.3	99.8%	99.0%	Microcephaly 3, primary, autosomal recessive, 604804
CDK6	115.0	99.4%	96.8%	?Microcephaly 12, primary, autosomal recessive, 616080
CDKL5	102.7	95.0%	92.9%	Epileptic encephalopathy, early infantile, 2, 300672
CDKN1B	164.2	100.0%	99.9%	Multiple endocrine neoplasia, type IV, 610755
CDKN1C	116.5	93.6%	84.7%	IMAGE syndrome, 614732 Beckwith-Wiedemann syndrome, 130650
CDKN2A	139.4	92.3%	92.3%	No OMIM disease ID
CDON	110.7	99.9%	99.0%	Holoprosencephaly 11, 614226
CDSN	141.6	100.0%	100.0%	Hypotrichosis 2, 146520 Peeling skin syndrome 1, 270300
CDT1	150.4	100.0%	100.0%	Meier-Gorlin syndrome 4, 613804
CEACAM16	139.8	100.0%	100.0%	Deafness, autosomal recessive 113, 618410 Deafness, autosomal dominant 4B, 614614
CEBPA	168.8	100.0%	100.0%	Leukemia, acute myeloid, somatic, 601626 ?Leukemia, acute myeloid, 601626
CEBPE	113.9	100.0%	100.0%	Specific granule deficiency, 245480
CEL	158.1	96.1%	91.6%	Maturity-onset diabetes of the young, type VIII, 609812
CELA2A	156.0	99.0%	95.4%	Abdominal obesity-metabolic syndrome 4, 618620
CENPE	72.2	98.5%	91.7%	?Microcephaly 13, primary, autosomal recessive, 616051
CENPF	143.9	99.8%	98.7%	Stromme syndrome, 243605
CENPJ	135.8	100.0%	99.4%	Microcephaly 6, primary, autosomal recessive, 608393 ?Seckel syndrome 4, 613676
CEP104	108.8	99.3%	97.8%	Joubert syndrome 25, 616781
CEP120	131.3	100.0%	99.6%	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP135	88.3	99.1%	92.0%	Microcephaly 8, primary, autosomal recessive, 614673
CEP152	145.3	99.6%	97.8%	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823
CEP164	96.7	99.9%	98.4%	Nephronophthisis 15, 614845
CEP19	188.4	100.0%	100.0%	Morbid obesity and spermatogenic failure, 615703
CEP250	108.0	99.9%	99.2%	Cone-rod dystrophy and hearing loss 2, 618358
CEP290	77.6	96.9%	88.7%	?Bardet-Biedl syndrome 14, 615991 Leber congenital amaurosis 10, 611755 Senior-Loken syndrome 6, 610189 Meckel syndrome 4, 611134 Joubert syndrome 5, 610188
CEP41	79.1	98.7%	94.4%	Joubert syndrome 15, 614464

CEP55	123.2	100.0%	100.0%	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500
CEP57	84.9	99.3%	92.8%	Mosaic variegated aneuploidy syndrome 2, 614114
CEP63	120.6	98.4%	93.8%	?Seckel syndrome 6, 614728
CEP78	123.1	99.8%	96.9%	Cone-rod dystrophy and hearing loss, 617236
CEP83	103.3	99.8%	96.2%	Nephronophthisis 18, 615862
CERKL	115.2	99.5%	96.8%	Retinitis pigmentosa 26, 608380
CERS1	80.3	95.6%	85.4%	?Epilepsy, progressive myoclonic, 8, 616230
CERS3	93.3	100.0%	98.5%	Ichthyosis, congenital, autosomal recessive 9, 615023
CES1	135.3	99.6%	97.6%	Drug metabolism, altered, CES1-related, 618057
CETP	129.3	100.0%	99.9%	Hyperalphalipoproteinemia, 143470
CFAP43	116.1	99.9%	97.5%	Spermatogenic failure 19, 617592
CFAP44	106.5	99.7%	98.5%	?Spermatogenic failure 20, 617593
CFAP53	135.5	99.2%	97.0%	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFAP69	65.3	98.1%	91.0%	Spermatogenic failure 24, 617959
CFB	125.5	100.0%	100.0%	?Complement factor B deficiency, 615561
CFC1	136.1	91.1%	82.3%	Heterotaxy, visceral, 2, autosomal, 605376
CFD	128.9	97.9%	92.1%	Complement factor D deficiency, 613912
CFH	148.8	99.4%	97.4%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814
CFHR5	91.5	99.9%	97.2%	Nephropathy due to CFHR5 deficiency, 614809
CFI	137.6	99.2%	96.8%	Complement factor I deficiency, 610984
CFL2	120.4	100.0%	99.5%	Nemaline myopathy 7, autosomal recessive, 610687
CFP	107.2	99.8%	98.3%	Properdin deficiency, X-linked, 312060
CFTR	112.9	99.4%	97.3%	Cystic fibrosis, 219700 Congenital bilateral absence of vas deferens, 277180 Sweat chloride elevation without CF, 0
CHAMP1	181.4	100.0%	100.0%	Mental retardation, autosomal dominant 40, 616579
CHAT	125.5	97.1%	89.5%	Myasthenic syndrome, congenital, 6, presynaptic, 254210
CHCHD10	28.6	67.7%	41.5%	Spinal muscular atrophy, Jokela type, 615048 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 ?Myopathy, isolated mitochondrial, autosomal dominant, 616209
CHCHD2	75.2	100.0%	96.6%	Parkinson disease 22, autosomal dominant, 616710
CHD1	104.2	97.7%	90.7%	Pilarowski-Bjornsson syndrome, 617682
CHD2	126.1	99.4%	99.1%	Epileptic encephalopathy, childhood-onset, 615369
CHD3	101.6	98.4%	95.1%	Snijders Blok-Campeau syndrome, 618205
CHD4	117.3	100.0%	99.9%	Sifrim-Hitz-Weiss syndrome, 617159

CHD7	143.6	100.0%	99.5%	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CHEK2	91.6	83.8%	79.3%	Li-Fraumeni syndrome, 609265 Osteosarcoma, somatic, 259500
CHKB	126.8	100.0%	100.0%	Muscular dystrophy, congenital, megaconial type, 602541
CHM	98.7	98.3%	91.4%	Choroideremia, 303100
CHMP1A	133.0	100.0%	99.8%	Pontocerebellar hypoplasia, type 8, 614961
CHMP2B	87.0	100.0%	96.8%	Amyotrophic lateral sclerosis 17, 614696 Dementia, familial, nonspecific, 600795
CHMP4B	150.6	100.0%	98.5%	Cataract 31, multiple types, 605387
CHN1	149.3	99.8%	98.7%	Duane retraction syndrome 2, 604356
CHP1	74.2	97.1%	87.4%	?Spastic ataxia 9, autosomal recessive, 618438
CHRDL1	89.5	100.0%	99.1%	Megalocornea 1, X-linked, 309300
CHRM3	140.5	100.0%	100.0%	?Prune belly syndrome, 100100
CHRNA1	96.7	94.5%	93.4%	Myasthenic syndrome, congenital, 1A, slow-channel, 601462 Myasthenic syndrome, congenital, 1B, fast-channel, 608930 Multiple pterygium syndrome, lethal type, 253290
CHRNA2	192.3	100.0%	100.0%	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA4	125.3	100.0%	99.3%	Epilepsy, nocturnal frontal lobe, 1, 600513
CHRNB1	134.8	100.0%	99.8%	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314 Myasthenic syndrome, congenital, 2A, slow-channel, 616313
CHRNB2	176.5	100.0%	98.9%	Epilepsy, nocturnal frontal lobe, 3, 605375
CHRND	154.8	99.9%	98.3%	?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323 ?Myasthenic syndrome, congenital, 3A, slow-channel, 616321 Myasthenic syndrome, congenital, 3B, fast-channel, 616322 Multiple pterygium syndrome, lethal type, 253290
CHRNE	186.2	100.0%	100.0%	Myasthenic syndrome, congenital, 4A, slow-channel, 605809 Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931 Myasthenic syndrome, congenital, 4B, fast-channel, 616324
CHRNG	153.9	100.0%	100.0%	Escobar syndrome, 265000 Multiple pterygium syndrome, lethal type, 253290
CHST11	202.4	100.0%	100.0%	?Osteochondrodysplasia, brachydactyly, and overlapping malformed digits, 618167
CHST14	180.8	100.0%	99.5%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	146.9	100.0%	100.0%	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHST6	322.9	100.0%	100.0%	Macular corneal dystrophy, 217800
CHST8	295.2	100.0%	100.0%	?Peeling skin syndrome 3, 616265
CHSY1	134.8	99.8%	99.0%	Temtamy preaxial brachydactyly syndrome, 605282

CHUK	124.5	99.7%	98.1%	Cocoon syndrome, 613630
CIB1	134.0	99.8%	97.7%	Epidermodysplasia verruciformis 3, 618267
CIB2	218.3	100.0%	99.5%	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CIC	83.4	64.8%	63.4%	Mental retardation, autosomal dominant 45, 617600
CIDEC	90.2	99.9%	97.5%	?Lipodystrophy, familial partial, type 5, 615238
CIITA	166.3	100.0%	100.0%	Bare lymphocyte syndrome, type II, complementation group A, 209920
CISD2	116.7	83.4%	83.4%	Wolfram syndrome 2, 604928
CIT	106.3	100.0%	98.9%	Microcephaly 17, primary, autosomal recessive, 617090
CITED2	174.9	99.2%	99.2%	Atrial septal defect 8, 614433 Ventricular septal defect 2, 614431
CKAP2L	154.2	99.9%	98.9%	Filippi syndrome, 272440
CLCF1	95.0	100.0%	99.6%	Cold-induced sweating syndrome 2, 610313
CLCN1	134.2	100.0%	99.9%	Myotonia congenita, dominant, 160800 Myotonia congenita, recessive, 255700 Myotonia levior, recessive, 0
CLCN2	126.9	100.0%	99.8%	Leukoencephalopathy with ataxia, 615651 Hyperaldosteronism, familial, type II, 605635
CLCN4	111.7	99.9%	98.8%	Raynaud-Claes syndrome, 300114
CLCN5	105.8	99.7%	96.8%	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990 Dent disease, 300009 Hypophosphatemic rickets, 300554 Nephrolithiasis, type I, 310468
CLCN7	162.0	99.9%	98.9%	Osteopetrosis, autosomal recessive 4, 611490 Osteopetrosis, autosomal dominant 2, 166600 Hypopigmentation, organomegaly, and delayed myelination and development, 618541
CLCNKA	121.7	99.9%	98.4%	Bartter syndrome, type 4b, digenic, 613090
CLCNKB	109.1	100.0%	98.3%	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090
CLDN1	129.7	100.0%	100.0%	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626
CLDN10	144.8	100.0%	99.9%	HELIX syndrome, 617671
CLDN14	121.5	100.0%	100.0%	Deafness, autosomal recessive 29, 614035
CLDN16	132.1	100.0%	100.0%	Hypomagnesemia 3, renal, 248250
CLDN19	141.2	99.8%	97.2%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLEC7A	146.7	100.0%	100.0%	Candidiasis, familial, 4, autosomal recessive, 613108
CLIC2	72.0	100.0%	98.2%	?Mental retardation, X-linked, syndromic 32, 300886
CLIC5	103.2	100.0%	99.9%	?Deafness, autosomal recessive 103, 616042

CLMP	89.1	100.0%	99.8%	Congenital short bowel syndrome, 615237
CLN3	123.4	92.5%	92.2%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	139.4	100.0%	99.5%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	141.7	100.0%	100.0%	Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300 Ceroid lipofuscinosis, neuronal, 6, 601780
CLN8	156.2	83.5%	83.5%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLP1	146.0	100.0%	100.0%	Pontocerebellar hypoplasia, type 10, 615803
CLPB	135.3	99.7%	97.4%	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CLPP	152.0	100.0%	99.6%	Perrault syndrome 3, 614129
CLPX	149.0	99.9%	98.8%	?Protoporphyria, erythropoietic, 2, 618015
CLRN1	140.6	100.0%	99.5%	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902
CLTC	154.5	100.0%	99.8%	Mental retardation, autosomal dominant 56, 617854
CNBP	119.7	100.0%	100.0%	Myotonic dystrophy 2, 602668
CNGA1	110.6	93.0%	86.9%	Retinitis pigmentosa 49, 613756
CNGA3	160.9	100.0%	99.8%	Achromatopsia 2, 216900
CNGB1	116.9	99.7%	98.3%	Retinitis pigmentosa 45, 613767
CNGB3	101.6	98.4%	93.7%	Macular degeneration, juvenile, 248200 Achromatopsia 3, 262300
CNKSR2	88.5	98.6%	92.0%	Mental retardation, X-linked, syndromic, Houge type, 301008
CNNM2	222.5	100.0%	100.0%	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNNM4	177.3	100.0%	99.9%	Jalili syndrome, 217080
CNOT1	127.6	100.0%	99.8%	Holoprosencephaly 12, with or without pancreatic agenesis, 618500
CNOT2	130.4	100.0%	99.8%	Intellectual developmental disorder with nasal speech, dysmorphic facies, and variable skeletal anomalies, 618608
CNPY3	83.9	100.0%	100.0%	Epileptic encephalopathy, early infantile, 60, 617929
CNTN1	128.8	99.9%	99.1%	?Myopathy, congenital, Compton-North, 612540
CNTN2	132.4	92.7%	92.7%	?Epilepsy, myoclonic, familial adult, 5, 615400
CNTNAP1	170.0	100.0%	99.4%	Lethal congenital contracture syndrome 7, 616286 Hypomyelinating neuropathy, congenital, 3, 618186
CNTNAP2	129.8	100.0%	99.8%	Pitt-Hopkins like syndrome 1, 610042 Cortical dysplasia-focal epilepsy syndrome, 610042
COA5	81.0	87.4%	83.4%	?Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 3, 616500
COA6	125.2	100.0%	97.4%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 4, 616501
COA7	132.3	100.0%	100.0%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387

COASY	190.6	100.0%	100.0%	Pontocerebellar hypoplasia, type 12, 618266 Neurodegeneration with brain iron accumulation 6, 615643
COCH	163.5	100.0%	100.0%	?Deafness, autosomal recessive 110, 618094 Deafness, autosomal dominant 9, 601369
COG1	117.9	100.0%	99.9%	Congenital disorder of glycosylation, type IIg, 611209
COG2	126.1	99.5%	97.2%	?Congenital disorder of glycosylation, type IIq, 617395
COG4	99.0	100.0%	99.7%	Saul-Wilson syndrome, 618150 Congenital disorder of glycosylation, type IIj, 613489
COG5	123.1	99.8%	97.9%	Congenital disorder of glycosylation, type IIi, 613612
COG6	87.3	98.6%	95.6%	Shaheen syndrome, 615328 Congenital disorder of glycosylation, type III, 614576
COG7	111.8	100.0%	99.9%	Congenital disorder of glycosylation, type IIe, 608779
COG8	160.1	100.0%	98.6%	Congenital disorder of glycosylation, type IIh, 611182
COL10A1	116.2	100.0%	100.0%	Metaphyseal chondrodysplasia, Schmid type, 156500
COL11A1	94.6	98.0%	93.6%	Stickler syndrome, type II, 604841 Marshall syndrome, 154780 ?Deafness, autosomal dominant 37, 618533 Fibrochondrogenesis 1, 228520
COL11A2	122.3	100.0%	99.5%	Deafness, autosomal dominant 13, 601868 Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150 Fibrochondrogenesis 2, 614524 Deafness, autosomal recessive 53, 609706 Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840
COL12A1	123.5	99.9%	99.1%	Bethlem myopathy 2, 616471 ?Ullrich congenital muscular dystrophy 2, 616470
COL13A1	96.7	100.0%	99.4%	Myasthenic syndrome, congenital, 19, 616720
COL17A1	110.6	99.6%	97.7%	Epithelial recurrent erosion dystrophy, 122400 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa, junctional, localisata variant, 226650
COL18A1	153.2	99.7%	97.9%	Knobloch syndrome, type 1, 267750
COL1A1	154.6	99.9%	99.1%	Osteogenesis imperfecta, type I, 166200 Osteogenesis imperfecta, type IV, 166220 Osteogenesis imperfecta, type II, 166210 Caffey disease, 114000 Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060 Osteogenesis imperfecta, type III, 259420
COL1A2	96.3	98.6%	94.6%	Ehlers-Danlos syndrome, cardiac valvular type, 225320 Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821

				Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type IV, 166220 Osteogenesis imperfecta, type III, 259420
COL25A1	134.2	99.3%	98.7%	Fibrosis of extraocular muscles, congenital, 5, 616219
COL27A1	154.1	99.9%	99.3%	Steel syndrome, 615155
COL2A1	121.1	100.0%	99.8%	Achondrogenesis, type II or hypochondrogenesis, 200610 Spondyloperipheral dysplasia, 271700 Kniest dysplasia, 156550 Stickler syndrome, type I, 108300 Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 Osteoarthritis with mild chondrodysplasia, 604864 Platyspondylic skeletal dysplasia, Torrance type, 151210 Avascular necrosis of the femoral head, 608805 SED congenita, 183900 Legg-Calve-Perthes disease, 150600 SMED Strudwick type, 184250 Czech dysplasia, 609162 Stickler sydrome, type I, nonsyndromic ocular, 609508 Spondyloepiphyseal dysplasia, Stanescu type, 616583 Vitreoretinopathy with phalangeal epiphyseal dysplasia, 0
COL3A1	99.6	99.4%	97.0%	Ehlers-Danlos syndrome, vascular type, 130050 Polymicrogyria with or without vascular-type EDS, 618343
COL4A1	100.3	99.8%	98.0%	?Retinal arteries, tortuosity of, 180000 Brain small vessel disease with or without ocular anomalies, 175780 Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564
COL4A2	118.1	100.0%	99.4%	Brain small vessel disease 2, 614483
COL4A3	93.8	99.6%	97.9%	Hematuria, benign familial, 141200 Alport syndrome 2, autosomal recessive, 203780 Alport syndrome 3, autosomal dominant, 104200
COL4A3BP	132.6	99.6%	97.3%	Mental retardation, autosomal dominant 34, 616351
COL4A4	96.5	99.7%	98.0%	Alport syndrome 2, autosomal recessive, 203780 Hematuria, familial benign, 141200
COL4A5	57.3	97.4%	87.3%	Alport syndrome 1, X-linked, 301050
COL4A6	85.2	97.2%	92.1%	?Deafness, X-linked 6, 300914
COL5A1	146.8	100.0%	99.4%	Ehlers-Danlos syndrome, classic type, 1, 130000
COL5A2	100.3	99.9%	99.3%	Ehlers-Danlos syndrome, classic type, 2, 130010

COL6A1	178.1	100.0%	100.0%	Ullrich congenital muscular dystrophy 1, 254090 Bethlem myopathy 1, 158810
COL6A2	195.3	100.0%	99.9%	Bethlem myopathy 1, 158810 ?Myosclerosis, congenital, 255600 Ullrich congenital muscular dystrophy 1, 254090
COL6A3	163.9	100.0%	99.8%	Bethlem myopathy 1, 158810 Dystonia 27, 616411 Ullrich congenital muscular dystrophy 1, 254090
COL7A1	153.9	99.9%	99.2%	EBD inversa, 226600 Epidermolysis bullosa dystrophica, AR, 226600 Toenail dystrophy, isolated, 607523 EBD, Bart type, 132000 Transient bullous of the newborn, 131705 Epidermolysis bullosa dystrophica, AD, 131750 Epidermolysis bullosa pruriginosa, 604129 Epidermolysis bullosa, pretibial, 131850 EBD, localisata variant, 0
COL8A2	140.0	100.0%	100.0%	Corneal dystrophy, posterior polymorphous 2, 609140 Corneal dystrophy, Fuchs endothelial, 1, 136800
COL9A1	132.9	100.0%	99.5%	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	104.9	100.0%	99.6%	?Stickler syndrome, type V, 614284 Epiphyseal dysplasia, multiple, 2, 600204
COL9A3	120.1	99.9%	98.3%	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969
COLEC10	119.9	100.0%	100.0%	3MC syndrome 3, 248340
COLEC11	197.8	100.0%	100.0%	3MC syndrome 2, 265050
COLGALT1	157.8	98.6%	93.8%	Brain small vessel disease 3, 618360
COLQ	106.4	99.9%	98.2%	Myasthenic syndrome, congenital, 5, 603034
COMP	147.0	97.4%	93.9%	Epiphyseal dysplasia, multiple, 1, 132400 Pseudoachondroplasia, 177170
COPB2	137.4	99.9%	98.9%	?Microcephaly 19, primary, autosomal recessive, 617800
COQ2	107.7	97.7%	97.0%	Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	116.2	91.7%	90.8%	Coenzyme Q10 deficiency, primary, 7, 616276
COQ6	136.6	99.6%	97.4%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	142.8	99.9%	99.6%	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ8A	177.7	100.0%	100.0%	Coenzyme Q10 deficiency, primary, 4, 612016
COQ8B	109.0	100.0%	99.9%	Nephrotic syndrome, type 9, 615573
COQ9	78.6	99.9%	98.5%	Coenzyme Q10 deficiency, primary, 5, 614654

CORIN	143.8	100.0%	99.9%	Preeclampsia/eclampsia 5, 614595
CORO1A	166.6	100.0%	99.2%	Immunodeficiency 8, 615401
COX10	232.8	100.0%	100.0%	Mitochondrial complex IV deficiency, 220110 Leigh syndrome due to mitochondrial COX4 deficiency, 256000
COX14	103.1	100.0%	100.0%	?Mitochondrial complex IV deficiency, 220110
COX15	90.4	99.9%	98.7%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000
COX20	66.5	93.9%	81.6%	Mitochondrial complex IV deficiency, 220110
COX4I2	125.6	100.0%	99.9%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX6A1	160.6	100.0%	100.0%	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
COX6A2	64.2	99.6%	95.6%	Mitochondrial complex IV deficiency, 220110
COX6B1	143.0	100.0%	100.0%	Mitochondrial complex IV deficiency, 220110
COX7B	39.4	62.3%	31.6%	Linear skin defects with multiple congenital anomalies 2, 300887
COX8A	120.1	100.0%	100.0%	?Mitochondrial complex IV deficiency, 220110
CP	99.2	92.9%	87.3%	Hemosiderosis, systemic, due to aceruloplasminemia, 604290 Cerebellar ataxia, 604290
CPA6	110.2	99.7%	97.2%	Febrile seizures, familial, 11, 614418 Epilepsy, familial temporal lobe, 5, 614417
CPAMD8	109.5	98.4%	95.6%	Anterior segment dysgenesis 8, 617319
CPLX1	119.2	100.0%	100.0%	Epileptic encephalopathy, early infantile, 63, 617976
CPN1	102.5	99.8%	98.4%	Carboxypeptidase N deficiency, 212070
CPOX	141.1	99.7%	97.8%	Harderoporphyria, 121300 Coproporphyria, 121300
CPS1	133.4	100.0%	99.9%	Carbamoylphosphate synthetase I deficiency, 237300
CPT1A	135.4	99.9%	98.6%	CPT deficiency, hepatic, type IA, 255120
CPT1C	131.6	100.0%	100.0%	?Spastic paraplegia 73, autosomal dominant, 616282
CPT2	152.7	98.3%	98.3%	CPT II deficiency, myopathic, stress-induced, 255110 CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836
CR2	135.0	100.0%	100.0%	Immunodeficiency, common variable, 7, 614699
CRADD	120.5	100.0%	99.0%	Mental retardation, autosomal recessive 34, with variant lissencephaly, 614499
CRAT	127.2	100.0%	100.0%	?Neurodegeneration with brain iron accumulation 8, 617917
CRB1	158.9	100.0%	100.0%	Pigmented paravenous chorioretinal atrophy, 172870 Retinitis pigmentosa-12, 600105 Leber congenital amaurosis 8, 613835
CRB2	132.1	99.9%	98.7%	Ventriculomegaly with cystic kidney disease, 219730 Focal segmental glomerulosclerosis 9, 616220

CRBN	122.9	88.2%	87.4%	Mental retardation, autosomal recessive 2, 607417
CREB1	120.9	99.6%	96.1%	Histiocytoma, angiomatoid fibrous, somatic, 612160
CREB3L1	145.4	100.0%	99.9%	Osteogenesis imperfecta, type XVI, 616229
CREBBP	120.9	99.6%	97.3%	Rubinstein-Taybi syndrome 1, 180849 Menke-Hennekam syndrome 1, 618332
CRELD1	105.6	99.9%	96.0%	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217
CRIPT	40.5	99.0%	89.7%	Short stature with microcephaly and distinctive facies, 615789
CRLF1	140.3	93.9%	91.4%	Cold-induced sweating syndrome 1, 272430
CRTAP	129.9	100.0%	99.3%	Osteogenesis imperfecta, type VII, 610682
CRTC1	184.2	99.8%	99.8%	Mucoepidermoid salivary gland carcinoma, 0
CRX	216.9	100.0%	100.0%	Cone-rod retinal dystrophy-2, 120970 Leber congenital amaurosis 7, 613829
CRYAA	145.4	96.7%	91.1%	Cataract 9, multiple types, 604219
CRYAB	96.3	99.9%	97.9%	Myopathy, myofibrillar, 2, 608810 Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869
CRYBA1	108.4	100.0%	99.8%	Cataract 10, multiple types, 600881
CRYBA2	183.6	100.0%	100.0%	?Cataract 42, 115900
CRYBA4	126.6	100.0%	100.0%	Cataract 23, 610425
CRYBB1	132.7	100.0%	99.9%	Cataract 17, multiple types, 611544
CRYBB2	148.3	100.0%	100.0%	Cataract 3, multiple types, 601547
CRYBB3	149.6	100.0%	100.0%	Cataract 22, 609741
CRYGB	111.5	100.0%	98.3%	Cataract 39, multiple types, autosomal dominant, 615188
CRYGC	138.2	100.0%	99.7%	Cataract 2, multiple types, 604307
CRYGD	112.7	100.0%	99.7%	Cataract 4, multiple types, 115700
CRYGS	84.2	94.4%	85.4%	Cataract 20, multiple types, 116100
CRYM	87.0	99.8%	97.5%	Deafness, autosomal dominant 40, 616357
CSF1R	121.9	100.0%	99.4%	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
CSF2RA	56.6	90.0%	88.2%	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF2RB	136.0	99.9%	98.7%	Surfactant metabolism dysfunction, pulmonary, 5, 614370
CSF3R	116.5	100.0%	99.2%	Neutropenia, severe congenital, 7, autosomal recessive, 617014
CSNK1D	128.3	99.5%	96.3%	Advanced sleep-phase syndrome, familial, 2, 615224
CSNK2A1	106.2	94.0%	89.7%	Okur-Chung neurodevelopmental syndrome, 617062
CSPP1	117.4	100.0%	99.4%	Joubert syndrome 21, 615636

CSRP3	94.3	99.9%	97.8%	Cardiomyopathy, hypertrophic, 12, 612124 ?Cardiomyopathy, dilated, 1M, 607482
CST3	85.9	100.0%	97.3%	Cerebral amyloid angiopathy, 105150
CST6	126.0	100.0%	98.8%	?Ectodermal dysplasia 15, hypohidrotic/hair type, 618535
CSTA	113.5	100.0%	99.4%	Peeling skin syndrome 4, 607936
CSTB	74.8	99.1%	93.0%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTBP1	111.3	95.7%	88.6%	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915
CTC1	113.5	100.0%	99.6%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTCF	131.3	100.0%	99.1%	Mental retardation, autosomal dominant 21, 615502
CTDP1	141.7	96.2%	88.2%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTH	140.6	100.0%	99.7%	Cystathioninuria, 219500 Homocysteine, total plasma, elevated, 0
CTHRC1	103.6	99.6%	97.3%	Barrett esophagus/esophageal adenocarcinoma, 614266
CTLA4	146.7	100.0%	100.0%	Autoimmune lymphoproliferative syndrome, type V, 616100
CTNNA1	113.7	99.4%	97.7%	Macular dystrophy, patterned, 2, 608970
CTNNA2	110.0	100.0%	99.7%	Cortical dysplasia, complex, with other brain malformations 9, 618174
CTNNA3	128.9	100.0%	99.9%	Arrhythmogenic right ventricular dysplasia, familial, 13, 615616
CTNNB1	129.5	100.0%	100.0%	Ovarian cancer, somatic, 167000 Colorectal cancer, somatic, 114500 Medulloblastoma, somatic, 155255 Hepatocellular carcinoma, somatic, 114550 Pilomatricoma, somatic, 132600 Neurodevelopmental disorder with spastic diplegia and visual defects, 615075 Exudative vitreoretinopathy 7, 617572
CTNND1	130.4	100.0%	100.0%	Blepharocheilodontic syndrome 2, 617681
CTNS	118.7	100.0%	99.6%	Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, atypical nephropathic, 219800
CTPS1	112.2	100.0%	99.9%	Immunodeficiency 24, 615897
CTSA	146.1	100.0%	100.0%	Galactosialidosis, 256540
CTSC	119.4	100.0%	100.0%	Periodontitis 1, juvenile, 170650 Papillon-Lefevre syndrome, 245000 Haim-Munk syndrome, 245010
CTSD	187.3	100.0%	99.0%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	116.2	94.6%	83.7%	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362
CTSK	90.0	100.0%	100.0%	Pycnodysostosis, 265800

CTU2	152.3	100.0%	99.9%	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142
CUBN	103.2	99.6%	98.0%	Megaloblastic anemia-1, Finnish type, 261100
CUL3	114.0	99.9%	98.7%	Pseudohypoaldosteronism, type IIE, 614496
CUL4B	75.8	97.7%	88.2%	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
CUL7	139.6	100.0%	100.0%	3-M syndrome 1, 273750
CUX1	128.7	99.0%	96.5%	Global developmental delay with or without impaired intellectual development, 618330
CUX2	134.5	100.0%	99.4%	Epileptic encephalopathy, early infantile, 67, 618141
CWC27	82.5	99.8%	97.3%	Retinitis pigmentosa with or without skeletal anomalies, 250410
CWF19L1	103.5	100.0%	99.1%	Spinocerebellar ataxia, autosomal recessive 17, 616127
CXCR4	127.9	100.0%	99.9%	WHIM syndrome, 193670 Myelokathexis, isolated, 0
CXorf56	74.7	99.5%	93.2%	?Mental retardation, X-linked 107, 301013
CYB561	160.2	92.8%	92.8%	Orthostatic hypotension 2, 618182
CYB5A	137.4	100.0%	100.0%	Methemoglobinemia and ambiguous genitalia, 250790
CYB5R3	163.2	99.6%	98.5%	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYBA	119.0	97.4%	89.2%	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690
CYBB	98.6	99.9%	99.1%	Immunodeficiency 34, mycobacteriosis, X-linked, 300645 Chronic granulomatous disease, X-linked, 306400
CYC1	167.9	99.7%	97.3%	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYCS	60.4	99.5%	93.7%	Thrombocytopenia 4, 612004
CYFIP2	122.2	99.9%	98.7%	Epileptic encephalopathy, early infantile, 65, 618008
CYLD	110.8	99.8%	97.6%	Cylindromatosis, familial, 132700 Brooke-Spiegler syndrome, 605041 Trichoepithelioma, multiple familial, 1, 601606
CYP11A1	130.1	99.2%	94.8%	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	171.1	100.0%	100.0%	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 Aldosteronism, glucocorticoid-remediable, 103900
CYP11B2	170.2	100.0%	100.0%	Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 Aldosterone to renin ratio raised, 0
CYP17A1	116.2	100.0%	99.8%	17-alpha-hydroxylase/17,20-lyase deficiency, 202110 17,20-lyase deficiency, isolated, 202110
CYP19A1	128.2	99.7%	97.7%	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300
CYP1B1	153.6	100.0%	100.0%	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 Anterior segment dysgenesis 6, multiple subtypes, 617315

CYP21A2	102.4	99.9%	97.2%	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910
CYP24A1	178.7	100.0%	100.0%	Hypercalcemia, infantile, 1, 143880
CYP26B1	188.5	100.0%	100.0%	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
CYP26C1	155.8	100.0%	100.0%	Focal facial dermal dysplasia 4, 614974
CYP27A1	184.4	100.0%	99.8%	Cerebrotendinous xanthomatosis, 213700
CYP27B1	164.1	100.0%	99.8%	Vitamin D-dependent rickets, type I, 264700
CYP2A6	153.0	100.0%	99.7%	Coumarin resistance, 122700
CYP2B6	107.9	100.0%	98.1%	Efavirenz, poor metabolism of, 614546
CYP2C19	150.5	100.0%	97.5%	Clopidogrel, impaired responsiveness to, 609535 Mephenytoin poor metabolizer, 609535 Proguanil poor metabolizer, 609535 Omeprazole poor metabolizer, 609535
CYP2C9	156.8	99.5%	95.8%	Warfarin sensitivity, 122700 Tolbutamide poor metabolizer, 0
CYP2R1	133.4	99.9%	97.5%	Rickets due to defect in vitamin D 25-hydroxylation, 600081
CYP2U1	139.8	99.1%	96.8%	Spastic paraplegia 56, autosomal recessive, 615030
CYP4F22	124.4	100.0%	99.2%	Ichthyosis, congenital, autosomal recessive 5, 604777
CYP4V2	140.6	99.8%	98.4%	Bietti crystalline corneoretinal dystrophy, 210370
CYP7B1	103.2	99.7%	97.2%	Spastic paraplegia 5A, autosomal recessive, 270800 Bile acid synthesis defect, congenital, 3, 613812
D2HGDH	157.7	100.0%	99.8%	D-2-hydroxyglutaric aciduria, 600721
DAB1	118.6	100.0%	100.0%	Spinocerebellar ataxia 37, 615945
DACT1	137.7	98.9%	95.6%	?Townes-Brocks syndrome 2, 617466
DAG1	205.3	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538
DARS	121.4	99.9%	99.0%	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2	125.4	100.0%	98.6%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBH	161.3	100.0%	99.9%	Orthostatic hypotension 1, due to DBH deficiency, 223360
DBT	109.6	99.7%	96.9%	Maple syrup urine disease, type II, 248600
DCAF17	87.5	100.0%	99.2%	Woodhouse-Sakati syndrome, 241080
DCAF8	118.2	100.0%	99.5%	?Giant axonal neuropathy 2, autosomal dominant, 610100
DCC	119.4	100.0%	99.9%	Esophageal carcinoma, somatic, 133239 Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 Mirror movements 1 and/or agenesis of the corpus callosum, 157600 Colorectal cancer, somatic, 114500

DCDC2	158.0	100.0%	99.9%	Sclerosing cholangitis, neonatal, 617394 Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212
DCHS1	164.5	100.0%	100.0%	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390
DCLRE1C	139.2	99.9%	98.0%	Omenn syndrome, 603554 Severe combined immunodeficiency, Athabascan type, 602450
DCN	131.5	95.7%	95.3%	Corneal dystrophy, congenital stromal, 610048
DCPS	140.6	100.0%	99.9%	Al-Raqad syndrome, 616459
DCTN1	121.1	100.0%	99.5%	Perry syndrome, 168605 Neuropathy, distal hereditary motor, type VIIB, 607641
DCX	93.7	99.8%	98.9%	Subcortical laminal heterotopia, X-linked, 300067 Lissencephaly, X-linked, 300067
DDB2	154.2	100.0%	98.9%	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DDC	100.3	99.4%	96.2%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	166.9	100.0%	99.1%	Spastic paraplegia 28, autosomal recessive, 609340
DDHD2	130.8	100.0%	99.7%	Spastic paraplegia 54, autosomal recessive, 615033
DDOST	123.1	100.0%	99.9%	?Congenital disorder of glycosylation, type Ir, 614507
DDR2	119.0	100.0%	99.7%	Warburg-Cinotti syndrome, 618175 Spondylometaepiphyseal dysplasia, short limb-hand type, 271665
DDRGK1	108.7	100.0%	99.9%	Spondyloepimetaphyseal dysplasia, Shohat type, 602557
DDX11	108.3	88.3%	82.2%	Warsaw breakage syndrome, 613398
DDX3X	74.5	86.2%	82.8%	Mental retardation, X-linked 102, 300958
DDX58	111.6	99.9%	98.4%	Singleton-Merten syndrome 2, 616298
DDX59	143.3	100.0%	99.7%	Orofaciodigital syndrome V, 174300
DDX6	58.0	95.7%	81.6%	Intellectual developmental disorder with impaired language and dysmorphic facies, 618653
DEAF1	124.1	100.0%	99.1%	Mental retardation, autosomal dominant 24, 615828 ?Dyskinesia, seizures, and intellectual developmental disorder, 617171
DEGS1	150.2	100.0%	100.0%	Leukodystrophy, hypomyelinating, 18, 618404
DENND5A	101.5	99.8%	98.9%	Epileptic encephalopathy, early infantile, 49, 617281
DEPDC5	129.9	100.0%	99.8%	Epilepsy, familial focal, with variable foci 1, 604364
DES	138.0	100.0%	100.0%	Cardiomyopathy, dilated, 1I, 604765 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 Myopathy, myofibrillar, 1, 601419
DFNA5	95.9	100.0%	99.4%	Deafness, autosomal dominant 5, 600994
DFNB59	116.0	100.0%	99.2%	Deafness, autosomal recessive 59, 610220
DGAT1	167.3	97.0%	92.8%	?Diarrhea 7, protein-losing enteropathy type, 615863

DGKE	132.2	100.0%	98.3%	Nephrotic syndrome, type 7, 615008
DGUOK	127.0	100.0%	98.8%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 Portal hypertension, noncirrhotic, 617068 Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHCR24	170.7	100.0%	99.9%	Desmosterolosis, 602398
DHCR7	158.7	100.0%	100.0%	Smith-Lemli-Opitz syndrome, 270400
DHDDS	84.5	97.3%	94.0%	Retinitis pigmentosa 59, 613861 ?Congenital disorder of glycosylation, type 1bb, 613861 Developmental delay and seizures with or without movement abnormalities, 617836
DHFR	48.6	92.6%	80.9%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHH	181.5	100.0%	100.0%	46XY sex reversal 7, 233420 46XY partial gonadal dysgenesis, with minifascicular neuropathy, 607080
DHODH	107.2	100.0%	100.0%	Miller syndrome, 263750
DHPS	126.4	100.0%	100.0%	Neurodevelopmental disorder with seizures and speech and walking impairment, 618480
DHTKD1	127.4	99.9%	99.0%	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DHX30	178.6	100.0%	100.0%	Neurodevelopmental disorder with severe motor impairment and absent language, 617804
DHX38	112.7	100.0%	99.5%	Retinitis pigmentosa 84, 618220
DIABLO	176.3	100.0%	99.8%	Deafness, autosomal dominant 64, 614152
DIAPH1	104.4	100.0%	99.8%	Seizures, cortical blindness, microcephaly syndrome, 616632 Deafness, autosomal dominant 1, 124900
DIAPH2	64.5	95.8%	86.4%	?Premature ovarian failure 2A, 300511
DIAPH3	80.8	99.8%	96.0%	Auditory neuropathy, autosomal dominant, 1, 609129
DICER1	138.9	99.9%	98.7%	GLOW syndrome, somatic mosaic, 618272 Rhabdomyosarcoma, embryonal, 2, 180295 Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800 Pleuropulmonary blastoma, 601200
DIP2B	131.4	100.0%	100.0%	Mental retardation, FRA12A type, 136630
DIS3L2	151.0	100.0%	99.9%	Perlman syndrome, 267000
DKC1	93.9	99.7%	98.0%	Dyskeratosis congenita, X-linked, 305000
DLAT	104.9	100.0%	99.4%	Pyruvate dehydrogenase E2 deficiency, 245348
DLC1	168.1	100.0%	99.8%	Colorectal cancer, somatic, 114500
DLD	117.2	100.0%	99.9%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLG3	84.8	99.3%	94.6%	Mental retardation, X-linked 90, 300850
DLL3	122.2	97.9%	93.8%	Spondylocostal dysostosis 1, autosomal recessive, 277300
DLL4	194.6	100.0%	100.0%	Adams-Oliver syndrome 6, 616589
DLST	83.5	94.5%	87.3%	Paragangliomas 7, 618475

DLX3	165.9	100.0%	99.8%	Trichodontoosseous syndrome, 190320 Amelogenesis imperfecta, type IV, 104510
DLX4	253.0	100.0%	100.0%	?Orofacial cleft 15, 616788
DLX5	159.7	100.0%	99.8%	?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600
DMD	107.5	99.6%	97.9%	Cardiomyopathy, dilated, 3B, 302045 Becker muscular dystrophy, 300376 Duchenne muscular dystrophy, 310200
DMGDH	135.4	100.0%	99.8%	Dimethylglycine dehydrogenase deficiency, 605850
DMP1	135.8	100.0%	99.9%	Hypophosphatemic rickets, AR, 241520
DMPK	166.2	100.0%	99.9%	Myotonic dystrophy 1, 160900
DMXL2	151.5	99.8%	98.6%	?Deafness, autosomal dominant 71, 617605 ?Polyendocrine-polyneuropathy syndrome, 616113 Epileptic encephalopathy, early infantile, 81, 618663
DNA2	121.9	99.9%	97.8%	?Seckel syndrome 8, 615807 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156
DNAAF1	118.6	100.0%	99.7%	Ciliary dyskinesia, primary, 13, 613193
DNAAF2	169.3	99.9%	98.9%	Ciliary dyskinesia, primary, 10, 612518
DNAAF3	128.5	100.0%	99.2%	Ciliary dyskinesia, primary, 2, 606763
DNAAF4	92.2	99.9%	98.2%	Ciliary dyskinesia, primary, 25, 615482
DNAAF5	119.1	98.6%	90.6%	Ciliary dyskinesia, primary, 18, 614874
DNAH1	170.9	100.0%	99.8%	?Ciliary dyskinesia, primary, 37, 617577 Spermatogenic failure 18, 617576
DNAH11	132.3	99.9%	98.9%	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH17	134.9	100.0%	99.7%	Spermatogenic failure 39, 618643
DNAH5	115.4	100.0%	99.3%	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAH9	116.1	99.9%	99.0%	Ciliary dyskinesia, primary, 40, 618300
DNAI1	120.5	100.0%	100.0%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	163.8	99.7%	98.0%	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJB11	107.4	99.9%	99.4%	Polycystic kidney disease 6 with or without polycystic liver disease, 618061
DNAJB13	124.0	100.0%	99.7%	Ciliary dyskinesia, primary, 34, 617091
DNAJB2	129.2	100.0%	100.0%	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881
DNAJB6	65.0	97.5%	87.0%	Muscular dystrophy, limb-girdle, autosomal dominant 1, 603511
DNAJC12	144.7	87.4%	87.4%	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC19	92.5	99.1%	90.4%	3-methylglutaconic aciduria, type V, 610198
DNAJC21	130.6	100.0%	99.2%	Bone marrow failure syndrome 3, 617052
DNAJC3	136.0	100.0%	99.9%	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192
DNAJC5	203.8	100.0%	100.0%	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350

DNAJC6	131.2	100.0%	99.4%	Parkinson disease 19b, early-onset, 615528 Parkinson disease 19a, juvenile-onset, 615528
DNAL1	104.2	99.8%	94.5%	Ciliary dyskinesia, primary, 16, 614017
DNAL4	73.0	100.0%	97.3%	?Mirror movements 3, 616059
DNASE1L3	118.0	99.9%	99.7%	Systemic lupus erythematosus 16, 614420
DNM1	151.2	94.9%	93.3%	Epileptic encephalopathy, early infantile, 31, 616346
DNM1L	120.8	99.9%	97.7%	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 Optic atrophy 5, 610708
DNM2	134.2	99.8%	97.7%	Lethal congenital contracture syndrome 5, 615368 Charcot-Marie-Tooth disease, axonal type 2M, 606482 Centronuclear myopathy 1, 160150 Charcot-Marie-Tooth disease, dominant intermediate B, 606482
DNMBP	132.7	99.9%	99.1%	Cataract 48, 618415
DNMT1	122.3	99.4%	98.7%	Neuropathy, hereditary sensory, type IE, 614116 Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121
DNMT3A	132.4	99.9%	98.7%	Acute myeloid leukemia, somatic, 601626 Tatton-Brown-Rahman syndrome, 615879
DNMT3B	125.5	100.0%	99.9%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK2	123.0	100.0%	99.5%	Immunodeficiency 40, 616433
DOCK3	118.4	100.0%	99.5%	Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292
DOCK6	132.3	99.6%	98.9%	Adams-Oliver syndrome 2, 614219
DOCK7	118.3	99.6%	97.8%	Epileptic encephalopathy, early infantile, 23, 615859
DOCK8	115.2	100.0%	99.7%	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
DOK7	156.2	94.4%	93.5%	Myasthenic syndrome, congenital, 10, 254300 ?Fetal akinesia deformation sequence 3, 618389
DOLK	171.4	100.0%	100.0%	Congenital disorder of glycosylation, type Im, 610768
DONSON	92.4	99.6%	94.7%	Microcephaly-micromelia syndrome, 251230 Microcephaly, short stature, and limb abnormalities, 617604
DPAGT1	93.2	100.0%	100.0%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPF2	105.3	99.9%	97.4%	Coffin-Siris syndrome 7, 618027
DPH1	177.3	100.0%	100.0%	Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901
DPM1	134.2	95.5%	87.7%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	95.4	100.0%	99.1%	Congenital disorder of glycosylation, type Iu, 615042
DPM3	218.6	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937
DPP6	129.8	100.0%	99.8%	Mental retardation, autosomal dominant 33, 616311
DPY19L2	85.7	74.2%	69.6%	Spermatogenic failure 9, 613958

DPYD	140.7	99.4%	96.2%	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270
DPYS	121.4	100.0%	99.9%	Dihydropyrimidinuria, 222748
DRAM2	127.1	100.0%	100.0%	Cone-rod dystrophy 21, 616502
DRC1	96.8	100.0%	99.5%	Ciliary dyskinesia, primary, 21, 615294
DRD4	129.0	99.2%	92.3%	Autonomic nervous system dysfunction, 0
DSC2	120.1	99.7%	97.3%	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 Arrhythmogenic right ventricular dysplasia 11, 610476
DSC3	93.8	99.2%	97.2%	?Hypotrichosis and recurrent skin vesicles, 613102
DSE	95.2	99.8%	98.0%	Ehlers-Danlos syndrome, musculocontractural type 2, 615539
DSG1	131.8	99.3%	97.4%	Keratosis palmoplantaris striata I, AD, 148700 Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508
DSG2	131.5	100.0%	99.1%	Arrhythmogenic right ventricular dysplasia 10, 610193 Cardiomyopathy, dilated, 1BB, 612877
DSG4	161.1	99.9%	99.0%	Hypotrichosis 6, 607903
DSP	148.0	100.0%	99.6%	Keratosis palmoplantaris striata II, 612908 Epidermolysis bullosa, lethal acantholytic, 609638 Skin fragility-woolly hair syndrome, 607655 Arrhythmogenic right ventricular dysplasia 8, 607450 Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821
DSPP	83.9	98.8%	95.5%	Dentin dysplasia, type II, 125420 Deafness, autosomal dominant 39, with dentinogenesis, 605594 Dentinogenesis imperfecta, Shields type II, 125490 Dentinogenesis imperfecta, Shields type III, 125500
DST	145.3	99.9%	99.1%	?Neuropathy, hereditary sensory and autonomic, type VI, 614653 Epidermolysis bullosa simplex, autosomal recessive 2, 615425
DSTYK	127.7	99.8%	98.0%	Spastic paraplegia 23, 270750 Congenital anomalies of kidney and urinary tract 1, 610805
DTNA	127.7	100.0%	99.9%	Left ventricular noncompaction 1, with or without congenital heart defects, 604169
DTNBP1	118.8	99.8%	97.5%	Hermansky-Pudlak syndrome 7, 614076
DUOX2	138.9	99.6%	97.4%	Thyroid dyshormonogenesis 6, 607200
DUOXA2	152.4	100.0%	100.0%	Thyroid dyshormonogenesis 5, 274900
DUSP6	184.5	100.0%	100.0%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
DVL1	159.7	99.3%	97.2%	Robinow syndrome, autosomal dominant 2, 616331
DVL3	209.9	100.0%	100.0%	Robinow syndrome, autosomal dominant 3, 616894

DYM	102.4	97.4%	95.7%	Smith-McCort dysplasia, 607326 Dyggve-Melchior-Clausen disease, 223800
DYNC1H1	149.0	100.0%	99.8%	Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 Charcot-Marie-Tooth disease, axonal, type 20, 614228
DYNC1I2	49.4	83.2%	67.5%	Neurodevelopmental disorder with microcephaly and structural brain anomalies, 618492
DYNC2H1	98.0	98.9%	94.3%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYNC2LI1	94.8	99.6%	97.1%	Short-rib thoracic dysplasia 15 with polydactyly, 617088
DYRK1A	134.0	100.0%	100.0%	Mental retardation, autosomal dominant 7, 614104
DYRK1B	120.1	99.0%	96.2%	Abdominal obesity-metabolic syndrome 3, 615812
DYSF	143.5	100.0%	99.9%	Miyoshi muscular dystrophy 1, 254130 Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601 Myopathy, distal, with anterior tibial onset, 606768
DZIP1L	103.1	100.0%	98.4%	Polycystic kidney disease 5, 617610
EARS2	106.7	99.7%	98.4%	Combined oxidative phosphorylation deficiency 12, 614924
EBF3	153.4	100.0%	99.9%	Hypotonia, ataxia, and delayed development syndrome, 617330
EBP	68.9	99.8%	96.3%	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
ECE1	157.4	98.2%	97.8%	?Hirschsprung disease, cardiac defects, and autonomic dysfunction, 613870
ECEL1	121.5	100.0%	99.0%	Arthrogryposis, distal, type 5D, 615065
ECHS1	111.6	100.0%	100.0%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ECM1	169.4	100.0%	99.4%	Urbach-Wiethe disease, 247100
EDA	108.8	96.6%	88.1%	Tooth agenesis, selective, X-linked 1, 313500 Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100
EDAR	135.7	100.0%	100.0%	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490 Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900
EDARADD	93.4	99.8%	97.5%	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941 Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940
EDC3	120.5	100.0%	99.2%	?Mental retardation, autosomal recessive 50, 616460
EDN1	165.0	100.0%	99.9%	Auriculocondylar syndrome 3, 615706 Question mark ears, isolated, 612798
EDN3	147.0	100.0%	100.0%	Waardenburg syndrome, type 4B, 613265 Central hypoventilation syndrome, congenital, 209880
EDNRA	153.6	100.0%	99.9%	Mandibulofacial dysostosis with alopecia, 616367
EDNRB	126.2	96.4%	91.5%	ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580
EED	83.9	98.8%	93.6%	Cohen-Gibson syndrome, 617561

EEF1A2	209.4	100.0%	100.0%	Epileptic encephalopathy, early infantile, 33, 616409 Mental retardation, autosomal dominant 38, 616393
EEF2	168.8	100.0%	100.0%	?Spinocerebellar ataxia 26, 609306
EFEMP1	152.0	100.0%	99.5%	Doyne honeycomb degeneration of retina, 126600
EFEMP2	141.3	100.0%	100.0%	Cutis laxa, autosomal recessive, type IB, 614437
EFL1	152.7	99.4%	98.0%	Shwachman-Diamond syndrome 2, 617941
EFNB1	122.4	100.0%	100.0%	Craniofrontonasal dysplasia, 304110
EFTUD2	107.1	100.0%	99.5%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EGF	111.5	100.0%	99.8%	Hypomagnesemia 4, renal, 611718
EGFR	141.3	100.0%	100.0%	?Inflammatory skin and bowel disease, neonatal, 2, 616069 Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980 Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980
EGLN1	118.9	99.2%	89.3%	Erythrocytosis, familial, 3, 609820
EGR2	140.2	100.0%	100.0%	Dejerine-Sottas disease, 145900 Hypomyelinating neuropathy, congenital, 1, 605253 Charcot-Marie-Tooth disease, type 1D, 607678
EHHADH	138.8	100.0%	99.9%	?Fanconi renotubular syndrome 3, 615605
EHMT1	138.4	94.7%	94.5%	Kleefstra syndrome 1, 610253
EIF2AK3	134.0	99.5%	96.7%	Wolcott-Rallison syndrome, 226980
EIF2AK4	134.2	99.8%	98.2%	Pulmonary venoocclusive disease 2, 234810
EIF2B1	126.0	100.0%	100.0%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B2	116.2	99.7%	95.1%	Ovarioleukodystrophy, 603896 Leukoencephalopathy with vanishing white matter, 603896
EIF2B3	135.2	100.0%	100.0%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B4	127.9	100.0%	99.8%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B5	106.9	100.0%	99.8%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2S3	82.9	96.7%	88.2%	MEHMO syndrome, 300148
EIF3F	67.7	99.3%	92.9%	Mental retardation, autosomal recessive 67, 618295
EIF4A3	89.6	100.0%	98.9%	Robin sequence with cleft mandible and limb anomalies, 268305
ELAC2	117.1	100.0%	99.5%	Combined oxidative phosphorylation deficiency 17, 615440
ELANE	156.3	100.0%	99.8%	Neutropenia, severe congenital 1, autosomal dominant, 202700 Neutropenia, cyclic, 162800
ELMO2	109.0	100.0%	99.3%	Vascular malformation, primary intraosseous, 606893
ELMOD3	144.7	100.0%	99.8%	?Deafness, autosomal recessive 88, 615429

ELN	113.2	100.0%	99.6%	Cutis laxa, autosomal dominant, 123700 Supravalvar aortic stenosis, 185500
ELOVL1	90.9	99.7%	96.6%	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527
ELOVL4	103.3	100.0%	99.6%	Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
ELOVL5	106.1	100.0%	99.4%	Spinocerebellar ataxia 38, 615957
ELP1	118.4	99.9%	98.3%	Dysautonomia, familial, 223900
ELP2	118.6	99.8%	97.6%	Mental retardation, autosomal recessive 58, 617270
ELP4	57.1	73.3%	68.7%	?Aniridia 2, 617141
EMC1	111.0	100.0%	99.2%	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EMD	147.1	99.9%	99.1%	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
EMG1	128.0	100.0%	100.0%	Bowen-Conradi syndrome, 211180
EML1	134.8	100.0%	99.9%	Band heterotopia, 600348
EMP2	79.3	99.8%	96.9%	Nephrotic syndrome, type 10, 615861
EMX2	175.9	100.0%	100.0%	Schizencephaly, 269160
ENAM	144.6	100.0%	100.0%	Amelogenesis imperfecta, type IC, 204650 Amelogenesis imperfecta, type IB, 104500
ENG	137.2	100.0%	99.4%	Telangiectasia, hereditary hemorrhagic, type 1, 187300
ENO3	186.3	100.0%	100.0%	?Glycogen storage disease XIII, 612932
ENPP1	128.6	97.9%	92.4%	Hypophosphatemic rickets, autosomal recessive, 2, 613312 Cole disease, 615522 Arterial calcification, generalized, of infancy, 1, 208000
ENTPD1	125.9	100.0%	100.0%	Spastic paraplegia 64, autosomal recessive, 615683
EOGT	103.3	79.5%	78.1%	Adams-Oliver syndrome 4, 615297
EP300	173.3	99.7%	98.7%	Rubinstein-Taybi syndrome 2, 613684 Menke-Hennekam syndrome 2, 618333 Colorectal cancer, somatic, 114500
EPAS1	142.5	99.9%	98.8%	Erythrocytosis, familial, 4, 611783
EPB41	122.1	99.6%	97.5%	Elliptocytosis-1, 611804
EPB41L1	130.8	99.5%	95.7%	?Mental retardation, autosomal dominant 11, 614257
EPB42	137.3	99.9%	99.3%	Spherocytosis, type 5, 612690
EPCAM	79.6	99.8%	95.9%	Colorectal cancer, hereditary nonpolyposis, type 8, 613244 Diarrhea 5, with tufting enteropathy, congenital, 613217
EPG5	111.4	99.5%	98.3%	Vici syndrome, 242840
EPHA2	173.3	100.0%	99.9%	Cataract 6, multiple types, 116600
EPHB2	205.6	98.1%	98.1%	?Bleeding disorder, platelet-type, 22, 618462

EPHB4	170.7	100.0%	99.9%	Capillary malformation-arteriovenous malformation 2, 618196 Lymphatic malformation 7, 617300
EPHX1	124.0	99.2%	96.3%	?Hypercholanemia, familial, 607748
EPM2A	125.2	94.8%	90.2%	Epilepsy, progressive myoclonic 2A (Lafora), 254780
EPO	106.3	100.0%	99.8%	Erythrocytosis, familial, 5, 617907 ?Diamond-Blackfan anemia-like, 617911
EPRS	137.8	100.0%	99.1%	Leukodystrophy, hypomyelinating, 15, 617951
EPS8	114.5	99.7%	96.2%	?Deafness, autosomal recessive 102, 615974
EPS8L2	180.6	99.8%	97.0%	Deafness autosomal recessive 106, 617637
ERAL1	168.0	100.0%	100.0%	Perrault syndrome 6, 617565
ERBB2	152.0	99.8%	98.6%	Glioblastoma, somatic, 137800 Adenocarcinoma of lung, somatic, 211980 Gastric cancer, somatic, 613659 Ovarian cancer, somatic, 0
ERBB3	120.0	100.0%	99.3%	?Lethal congenital contractural syndrome 2, 607598
ERBB4	127.4	100.0%	99.3%	Amyotrophic lateral sclerosis 19, 615515
ERCC1	91.5	100.0%	98.8%	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	139.5	100.0%	99.9%	Trichothiodystrophy 1, photosensitive, 601675 ?Cerebrooculofacioskeletal syndrome 2, 610756 Xeroderma pigmentosum, group D, 278730
ERCC3	95.9	99.9%	98.7%	Xeroderma pigmentosum, group B, 610651 Trichothiodystrophy 2, photosensitive, 616390
ERCC4	136.9	100.0%	99.6%	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 Fanconi anemia, complementation group Q, 615272 XFE progeroid syndrome, 610965 Xeroderma pigmentosum, group F, 278760
ERCC5	130.9	100.0%	99.4%	Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 Xeroderma pigmentosum, group G, 278780 Cerebrooculofacioskeletal syndrome 3, 616570
ERCC6	161.8	100.0%	100.0%	Cerebrooculofacioskeletal syndrome 1, 214150 Cockayne syndrome, type B, 133540 Premature ovarian failure 11, 616946 UV-sensitive syndrome 1, 600630 De Sanctis-Cacchione syndrome, 278800
ERCC6L2	119.8	100.0%	99.1%	Bone marrow failure syndrome 2, 615715
ERCC8	79.9	99.0%	89.3%	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621

ERF	166.3	100.0%	99.6%	Craniosynostosis 4, 600775 Chitayat syndrome, 617180
ERGIC1	192.2	95.2%	94.4%	?Arthrogryposis multiplex congenita, neurogenic type, 208100
ERLIN1	140.5	100.0%	100.0%	Spastic paraplegia 62, 615681
ERLIN2	119.3	100.0%	99.4%	Spastic paraplegia 18, autosomal recessive, 611225
ERMARD	120.9	99.8%	98.3%	?Periventricular nodular heterotopia 6, 615544
ESCO2	112.4	99.5%	96.2%	Roberts syndrome, 268300 SC phocomelia syndrome, 269000
ESPN	35.1	55.8%	45.1%	?Usher syndrome, type 1M, 618632 Deafness, autosomal recessive 36, 609006 Deafness, neurosensory, without vestibular involvement, autosomal dominant, 609006
ESR1	150.0	100.0%	100.0%	Estrogen resistance, 615363 Breast cancer, somatic, 114480
ESR2	118.2	100.0%	99.8%	?Ovarian dysgenesis 8, 618187
ESRP1	96.9	99.9%	98.8%	?Deafness, autosomal recessive 109, 618013
ESRRB	131.0	100.0%	99.6%	Deafness, autosomal recessive 35, 608565
ETFA	132.0	100.0%	99.8%	Glutaric acidemia IIA, 231680
ETFB	127.7	100.0%	100.0%	Glutaric acidemia IIB, 231680
ETFDH	112.7	100.0%	99.7%	Glutaric acidemia IIC, 231680
ETHE1	105.9	99.9%	97.9%	Ethylmalonic encephalopathy, 602473
ETV6	157.6	99.9%	99.4%	Leukemia, acute myeloid, somatic, 601626 Thrombocytopenia 5, 616216
EVC	113.0	96.8%	92.1%	Ellis-van Creveld syndrome, 225500 ?Weyers acrofacial dysostosis, 193530
EVC2	115.9	99.6%	97.1%	Weyers acrofacial dysostosis, 193530 Ellis-van Creveld syndrome, 225500
EWSR1	71.0	92.6%	83.9%	Neuroepithelioma, 612219 Ewing sarcoma, 612219
EXOC6B	107.7	98.5%	97.1%	Spondyloepimetaphyseal dysplasia with joint laxity, type 3, 618395
EXOSC2	114.1	100.0%	99.9%	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EXOSC3	135.7	96.5%	87.0%	Pontocerebellar hypoplasia, type 1B, 614678
EXOSC8	86.9	98.9%	88.8%	Pontocerebellar hypoplasia, type 1C, 616081
EXOSC9	133.4	99.2%	95.0%	Pontocerebellar hypoplasia, type 1D, 618065
EXPH5	167.7	100.0%	100.0%	Epidermolysis bullosa, nonspecific, autosomal recessive, 615028
EXT1	91.1	99.9%	98.4%	Exostoses, multiple, type 1, 133700 Chondrosarcoma, 215300

EXT2	120.9	99.9%	99.0%	Exostoses, multiple, type 2, 133701 Seizures, scoliosis, and macrocephaly syndrome, 616682
EXTL3	200.7	100.0%	100.0%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
EYA1	121.6	100.0%	99.9%	?Otofaciocervical syndrome, 166780 Anterior segment anomalies with or without cataract, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650 Branchiootic syndrome 1, 602588
EYA4	134.5	100.0%	100.0%	Deafness, autosomal dominant 10, 601316 ?Cardiomyopathy, dilated, 1J, 605362
EYS	130.0	99.8%	97.3%	Retinitis pigmentosa 25, 602772
EZH2	130.3	99.5%	98.0%	Weaver syndrome, 277590
F10	191.3	99.9%	99.2%	Factor X deficiency, 227600
F11	129.2	100.0%	99.9%	Factor XI deficiency, autosomal dominant, 612416 Factor XI deficiency, autosomal recessive, 612416
F12	171.1	100.0%	99.7%	Factor XII deficiency, 234000 Angioedema, hereditary, type III, 610618
F13A1	116.5	100.0%	99.8%	Factor XIIIA deficiency, 613225
F13B	100.3	98.3%	90.9%	Factor XIIIB deficiency, 613235
F2	137.0	99.8%	97.7%	Hypoprothrombinemia, 613679 Dysprothrombinemia, 613679 Thrombophilia due to thrombin defect, 188050
F5	148.2	99.4%	97.6%	Factor V deficiency, 227400 Thrombophilia due to activated protein C resistance, 188055
F7	179.1	100.0%	100.0%	Factor VII deficiency, 227500
F8	105.6	99.4%	97.3%	Hemophilia A, 306700
F9	110.5	99.9%	97.9%	Thrombophilia, X-linked, due to factor IX defect, 300807 Hemophilia B, 306900
FA2H	101.5	99.3%	95.1%	Spastic paraplegia 35, autosomal recessive, 612319
FADD	201.3	100.0%	100.0%	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations, 613759
FAH	136.7	100.0%	99.8%	Tyrosinemia, type I, 276700
FAM111A	241.7	100.0%	99.6%	Gracile bone dysplasia, 602361 Kenny-Caffey syndrome, type 2, 127000
FAM111B	155.6	99.9%	99.5%	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704
FAM126A	124.1	100.0%	98.9%	Leukodystrophy, hypomyelinating, 5, 610532
FAM161A	142.0	99.9%	99.3%	Retinitis pigmentosa 28, 606068
FAM20A	122.6	99.9%	99.2%	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM20C	165.1	100.0%	100.0%	Raine syndrome, 259775

FAM46A	177.0	100.0%	100.0%	Osteogenesis imperfecta, type XVIII, 617952
FAM58A	57.8	83.7%	78.2%	STAR syndrome, 300707
FAM83H	139.0	100.0%	100.0%	Amelogenesis imperfecta, type IIIA, 130900
FAM92A	80.7	88.2%	77.3%	?Polydactyly, postaxial, type A9, 618219
FAN1	136.8	100.0%	99.9%	Interstitial nephritis, karyomegalic, 614817
FANCA	118.3	100.0%	99.2%	Fanconi anemia, complementation group A, 227650
FANCB	72.8	98.6%	93.0%	Fanconi anemia, complementation group B, 300514
FANCC	104.4	100.0%	99.3%	Fanconi anemia, complementation group C, 227645
FANCD2	116.2	99.2%	96.5%	Fanconi anemia, complementation group D2, 227646
FANCE	127.9	98.0%	91.8%	Fanconi anemia, complementation group E, 600901
FANCF	269.1	100.0%	100.0%	Fanconi anemia, complementation group F, 603467
FANCG	149.5	100.0%	100.0%	Fanconi anemia, complementation group G, 614082
FANCI	136.0	100.0%	98.8%	Fanconi anemia, complementation group I, 609053
FANCL	102.9	99.8%	97.9%	Fanconi anemia, complementation group L, 614083
FANCM	99.4	99.5%	96.6%	Spermatogenic failure 28, 618086 ?Premature ovarian failure 15, 618096
FAR1	72.4	97.6%	91.9%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154
FARS2	169.5	100.0%	100.0%	Spastic paraplegia 77, autosomal recessive, 617046 Combined oxidative phosphorylation deficiency 14, 614946
FARSB	77.5	96.9%	93.0%	Rajab interstitial lung disease with brain calcifications, 613658
FAS	228.0	100.0%	99.9%	Autoimmune lymphoproliferative syndrome, type IA, 601859 Squamous cell carcinoma, burn scar-related, somatic, 0
FASLG	84.8	100.0%	99.1%	Autoimmune lymphoproliferative syndrome, type IB, 601859
FASTKD2	116.5	99.9%	98.0%	?Mitochondrial complex IV deficiency, 220110
FAT2	139.8	100.0%	99.9%	Spinocerebellar ataxia 45, 617769
FAT4	195.5	100.0%	100.0%	Van Maldergem syndrome 2, 615546 Hennekam lymphangiectasia-lymphedema syndrome 2, 616006
FBLN5	103.0	91.8%	91.8%	Macular degeneration, age-related, 3, 608895 ?Cutis laxa, autosomal dominant 2, 614434 Neuropathy, hereditary, with or without age-related macular degeneration, 608895 Cutis laxa, autosomal recessive, type IA, 219100
FBN1	138.3	100.0%	99.7%	Marfan lipodystrophy syndrome, 616914 Marfan syndrome, 154700 MASS syndrome, 604308 Ectopia lentis, familial, 129600 Acromicric dysplasia, 102370 Weill-Marchesani syndrome 2, dominant, 608328

				Geleophysic dysplasia 2, 614185 Stiff skin syndrome, 184900
FBN2	144.5	100.0%	99.9%	Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118
FBP1	114.0	100.0%	99.2%	Fructose-1,6-bisphosphatase deficiency, 229700
FBXL3	187.9	100.0%	100.0%	Intellectual developmental disorder with short stature, facial anomalies, and speech defects, 606220
FBXL4	165.9	100.0%	100.0%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FBXO11	82.9	98.5%	94.3%	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities, 618089
FBXO31	123.6	100.0%	98.7%	?Mental retardation, autosomal recessive 45, 615979
FBXO38	162.9	99.6%	98.3%	Neuronopathy, distal hereditary motor, type IID, 615575
FBXO7	157.7	100.0%	99.5%	Parkinson disease 15, autosomal recessive, 260300
FCGR3A	184.5	99.7%	98.1%	Immunodeficiency 20, 615707
FCGR3B	147.4	99.1%	98.5%	Neutropenia, alloimmune neonatal, 0
FCN3	135.3	100.0%	100.0%	Immunodeficiency due to ficolin 3 deficiency, 613860
FDFT1	150.3	100.0%	99.8%	Squalene synthase deficiency, 618156
FDPS	64.3	98.1%	92.2%	Porokeratosis 9, multiple types, 616631
FDX2	164.5	100.0%	100.0%	Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900
FDXR	134.5	100.0%	99.4%	Auditory neuropathy and optic atrophy, 617717
FECH	107.9	100.0%	99.6%	Protoporphyria, erythropoietic, 1, 177000
FERMT1	93.0	99.6%	96.7%	Kindler syndrome, 173650
FERMT3	161.2	100.0%	99.9%	Leukocyte adhesion deficiency, type III, 612840
FEZF1	197.9	100.0%	100.0%	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FGA	142.0	99.4%	97.5%	Dysfibrinogenemia, congenital, 616004 Amyloidosis, familial visceral, 105200 Hypodysfibrinogenemia, congenital, 616004 Afibrinogenemia, congenital, 202400
FGB	138.9	99.9%	98.4%	Afibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Hypofibrinogenemia, congenital, 202400
FGD1	93.2	98.7%	94.4%	Mental retardation, X-linked syndromic 16, 305400 Aarskog-Scott syndrome, 305400
FGD4	105.3	99.8%	98.3%	Charcot-Marie-Tooth disease, type 4H, 609311
FGF10	118.7	100.0%	99.7%	Aplasia of lacrimal and salivary glands, 180920 LADD syndrome, 149730
FGF12	105.7	100.0%	100.0%	Epileptic encephalopathy, early infantile, 47, 617166
FGF14	225.9	100.0%	100.0%	Spinocerebellar ataxia 27, 609307
FGF16	95.4	99.4%	95.5%	Metacarpal 4-5 fusion, 309630

FGF17	181.7	100.0%	100.0%	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
FGF20	130.9	100.0%	99.0%	?Renal hypodysplasia/aplasia 2, 615721
FGF23	130.1	99.9%	98.7%	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993 Hypophosphatemic rickets, autosomal dominant, 193100
FGF3	160.1	100.0%	100.0%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGF5	167.5	100.0%	99.7%	Trichomegaly, 190330
FGF8	141.1	98.8%	90.5%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGF9	156.9	100.0%	100.0%	Multiple synostoses syndrome 3, 612961
FGFR1	131.6	100.0%	99.7%	Pfeiffer syndrome, 101600 Jackson-Weiss syndrome, 123150 Trigonocephaly 1, 190440 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Hartsfield syndrome, 615465 Osteoglophonic dysplasia, 166250 Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001
FGFR2	118.0	97.7%	97.1%	Apert syndrome, 101200 Jackson-Weiss syndrome, 123150 Saethre-Chotzen syndrome, 101400 Gastric cancer, somatic, 613659 Scaphocephaly, maxillary retrusion, and mental retardation, 609579 Bent bone dysplasia syndrome, 614592 LADD syndrome, 149730 Craniofacial-skeletal-dermatologic dysplasia, 101600 Pfeiffer syndrome, 101600 Crouzon syndrome, 123500 Beare-Stevenson cutis gyrata syndrome, 123790 Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Craniosynostosis, nonspecific, 0 Scaphocephaly and Axenfeld-Rieger anomaly, 0
FGFR3	157.1	100.0%	99.9%	Muenke syndrome, 602849 Nevus, epidermal, somatic, 162900 Thanatophoric dysplasia, type II, 187601 Bladder cancer, somatic, 109800 CATSHL syndrome, 610474 Crouzon syndrome with acanthosis nigricans, 612247 Hypochondroplasia, 146000 LADD syndrome, 149730 Achondroplasia, 100800

				Thanatophoric dysplasia, type I, 187600 Colorectal cancer, somatic, 114500 Spermatocytic seminoma, somatic, 273300 Cervical cancer, somatic, 603956 SADDAN, 616482
FGG	124.2	99.9%	98.3%	Hypofibrinogenemia, congenital, 202400 Hypodysfibrinogenemia, 616004 Dysfibrinogenemia, congenital, 616004 Afibrinogenemia, congenital, 202400
FH	126.0	95.9%	89.5%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FHL1	68.2	99.2%	92.8%	Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717 Scapuloperoneal myopathy, X-linked dominant, 300695 Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718 ?Uruguay faciocardiomusculoskeletal syndrome, 300280 Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 Myopathy, X-linked, with postural muscle atrophy, 300696
FIBP	135.9	100.0%	100.0%	Thauvin-Robinet-Faivre syndrome, 617107
FIG4	155.0	100.0%	99.7%	Yunis-Varon syndrome, 216340 ?Polymicrogyria, bilateral temporooccipital, 612691 Charcot-Marie-Tooth disease, type 4J, 611228 Amyotrophic lateral sclerosis 11, 612577
FIGLA	97.0	100.0%	99.5%	Premature ovarian failure 6, 612310
FITM2	160.3	100.0%	100.0%	Siddiqi syndrome, 618635
FKBP10	170.1	99.8%	98.3%	Bruck syndrome 1, 259450 Osteogenesis imperfecta, type XI, 610968
FKBP14	77.6	100.0%	99.2%	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557
FKRP	178.0	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153
FKTN	108.0	99.9%	96.4%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800
FLAD1	184.5	100.0%	99.8%	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100
FLCN	165.1	100.0%	100.0%	Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700

				Birt-Hogg-Dube syndrome, 135150 Colorectal cancer, somatic, 114500
FLG	158.7	100.0%	99.9%	Ichthyosis vulgaris, 146700
FLG2	374.4	100.0%	100.0%	Peeling skin syndrome 6, 618084
FLI1	163.7	99.4%	98.0%	Bleeding disorder, platelet-type, 21, 617443
FLNA	156.4	100.0%	99.9%	Otopalatodigital syndrome, type I, 311300 Congenital short bowel syndrome, 300048 Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Cardiac valvular dysplasia, X-linked, 314400 ?FG syndrome 2, 300321 Heterotopia, periventricular, 1, 300049 Terminal osseous dysplasia, 300244 Frontometaphyseal dysplasia 1, 305620
FLNB	131.8	99.8%	99.1%	Larsen syndrome, 150250 Atelosteogenesis, type I, 108720 Boomerang dysplasia, 112310 Spondylocarpotarsal synostosis syndrome, 272460 Atelosteogenesis, type III, 108721
FLNC	169.4	100.0%	99.9%	Myopathy, myofibrillar, 5, 609524 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Cardiomyopathy, familial hypertrophic, 26, 0
FLRT3	173.0	100.0%	100.0%	Hypogonadotropic hypogonadism 21 with anosmia, 615271
FLT3	109.8	99.9%	99.0%	Leukemia, acute myeloid, somatic, 601626 Leukemia, acute lymphoblastic, somatic, 613065 Leukemia, acute myeloid, reduced survival in, somatic, 601626
FLT4	177.8	99.2%	99.2%	Hemangioma, capillary infantile, somatic, 602089 Lymphatic malformation 1, 153100
FLVCR1	154.8	100.0%	99.4%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FLVCR2	131.7	100.0%	100.0%	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790
FMN2	120.8	87.1%	84.7%	Mental retardation, autosomal recessive 47, 616193
FMO3	133.3	99.9%	98.5%	Trimethylaminuria, 602079
FMR1	78.1	96.3%	90.1%	Premature ovarian failure 1, 311360 Fragile X tremor/ataxia syndrome, 300623 Fragile X syndrome, 300624

FN1	109.6	100.0%	99.3%	Glomerulopathy with fibronectin deposits 2, 601894 Spondylometaphyseal dysplasia, corner fracture type, 184255
FOLR1	115.7	100.0%	100.0%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXC1	96.1	99.9%	99.2%	Axenfeld-Rieger syndrome, type 3, 602482 Anterior segment dysgenesis 3, multiple subtypes, 601631
FOXC2	144.2	100.0%	100.0%	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXE1	110.9	100.0%	100.0%	Bamforth-Lazarus syndrome, 241850
FOXE3	111.7	93.8%	87.3%	Cataract 34, multiple types, 612968 Anterior segment dysgenesis 2, multiple subtypes, 610256
FOXF1	180.1	100.0%	100.0%	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380
FOXG1	162.5	99.7%	96.6%	Rett syndrome, congenital variant, 613454
FOXI1	209.3	100.0%	100.0%	Enlarged vestibular aqueduct, 600791
FOXL2	144.6	100.0%	99.9%	Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 Premature ovarian failure 3, 608996 Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100
FOXN1	149.7	100.0%	99.8%	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXO1	118.2	100.0%	100.0%	Rhabdomyosarcoma, alveolar, 268220
FOXP1	117.6	100.0%	99.9%	Mental retardation with language impairment and with or without autistic features, 613670
FOXP2	130.1	99.5%	98.4%	Speech-language disorder-1, 602081
FOXP3	126.8	99.3%	96.0%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790
FOXRED1	129.1	99.9%	99.0%	Mitochondrial complex I deficiency, nuclear type 19, 618241
FRAS1	123.1	99.9%	99.3%	Fraser syndrome 1, 219000
FREM1	112.5	99.8%	98.8%	Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485 Bifid nose with or without anorectal and renal anomalies, 608980
FREM2	160.8	100.0%	99.6%	Fraser syndrome 2, 617666 Cryptophthalmos, unilateral or bilateral, isolated, 123570
FRMD4A	124.0	91.5%	91.0%	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819
FRMD7	101.2	99.9%	97.9%	Nystagmus 1, congenital, X-linked, 310700 Nystagmus, infantile periodic alternating, X-linked, 310700
FRMPD4	115.6	99.7%	97.7%	Mental retardation, X-linked 104, 300983
FRRS1L	100.0	89.3%	81.8%	Epileptic encephalopathy, early infantile, 37, 616981
FSCN2	181.9	100.0%	100.0%	Retinitis pigmentosa 30, 607921
FSHB	111.9	100.0%	100.0%	Hypogonadotropic hypogonadism 24 without anosmia, 229070

FSHR	102.7	99.8%	97.6%	Ovarian hyperstimulation syndrome, 608115 Ovarian dysgenesis 1, 233300 Ovarian response to FSH stimulation, 276400
FSIP2	92.8	99.9%	99.1%	Spermatogenic failure 34, 618153
FTCD	129.8	99.1%	96.1%	Glutamate formiminotransferase deficiency, 229100
FTH1	70.5	99.2%	91.7%	?Hemochromatosis, type 5, 615517
FTL	164.3	100.0%	98.4%	Hyperferritinemia-cataract syndrome, 600886 Neurodegeneration with brain iron accumulation 3, 606159 L-ferritin deficiency, dominant and recessive, 615604
FTO	99.1	83.8%	83.7%	Growth retardation, developmental delay, facial dysmorphism, 612938
FTSJ1	134.4	99.6%	96.5%	Mental retardation, X-linked 9/44, 309549
FUCA1	135.9	100.0%	100.0%	Fucosidosis, 230000
FUK	116.4	99.6%	98.4%	Congenital disorder of glycosylation with defective fucosylation 2, 618324
FUS	126.1	99.8%	97.6%	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 Essential tremor, hereditary, 4, 614782
FUT6	159.8	100.0%	100.0%	Fucosyltransferase 6 deficiency, 613852
FUT8	130.4	100.0%	99.4%	Congenital disorder of glycosylation with defective fucosylation 1, 618005
FXN	67.4	100.0%	98.3%	Friedreich ataxia with retained reflexes, 229300 Friedreich ataxia, 229300
FXYD2	118.4	100.0%	100.0%	Hypomagnesemia 2, renal, 154020
FYB1	96.5	99.5%	96.0%	Thrombocytopenia 3, 273900
FYCO1	135.2	100.0%	100.0%	Cataract 18, autosomal recessive, 610019
FZD2	200.1	99.9%	98.5%	Omodysplasia 2, 164745
FZD4	192.1	100.0%	100.0%	Exudative vitreoretinopathy 1, 133780 Retinopathy of prematurity, 133780
FZD6	190.3	100.0%	100.0%	Nail disorder, nonsyndromic congenital, 10, (claw-shaped nails), 614157
G6PC	149.8	100.0%	100.0%	Glycogen storage disease Ia, 232200
G6PC3	126.1	100.0%	100.0%	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
G6PD	126.2	99.8%	98.4%	Hemolytic anemia, G6PD deficient (favism), 300908
GAA	180.3	100.0%	100.0%	Glycogen storage disease II, 232300
GAB1	154.4	100.0%	99.8%	?Deafness, autosomal recessive 26, 605428
GABBR2	115.6	99.1%	95.6%	Neurodevelopmental disorder with poor language and loss of hand skills, 617903 Epileptic encephalopathy, early infantile, 59, 617904
GABRA1	164.8	100.0%	100.0%	Epileptic encephalopathy, early infantile, 19, 615744
GABRA2	147.7	99.7%	96.5%	Epileptic encephalopathy, early infantile, 78, 618557
GABRA5	140.8	100.0%	100.0%	Epileptic encephalopathy, early infantile, 79, 618559

GABRB1	174.8	100.0%	100.0%	Epileptic encephalopathy, early infantile, 45, 617153
GABRB2	134.8	100.0%	100.0%	Epileptic encephalopathy, infantile or early childhood, 2, 617829
GABRB3	139.5	99.7%	98.5%	Epileptic encephalopathy, early infantile, 43, 617113
GABRG2	126.9	91.0%	90.0%	Epileptic encephalopathy, early infantile, 74, 618396 Febrile seizures, familial, 8, 607681 Epilepsy, generalized, with febrile seizures plus, type 3, 607681
GAD1	114.6	100.0%	99.8%	?Cerebral palsy, spastic quadriplegic, 1, 603513
GAL	176.0	100.0%	100.0%	?Epilepsy, familial temporal lobe, 8, 616461
GALC	103.0	99.8%	98.1%	Krabbe disease, 245200
GALE	153.0	100.0%	100.0%	Galactose epimerase deficiency, 230350
GALK1	186.1	100.0%	99.9%	Galactokinase deficiency with cataracts, 230200
GALNS	118.1	100.0%	99.4%	Mucopolysaccharidosis IVA, 253000
GALNT3	126.0	99.9%	98.8%	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GALT	165.3	100.0%	100.0%	Galactosemia, 230400
GAMT	125.7	99.7%	94.3%	Cerebral creatine deficiency syndrome 2, 612736
GAN	147.5	100.0%	99.6%	Giant axonal neuropathy-1, 256850
GANAB	113.2	100.0%	98.9%	Polycystic kidney disease 3, 600666
GARS	128.2	100.0%	99.6%	Charcot-Marie-Tooth disease, type 2D, 601472 Neuropathy, distal hereditary motor, type VA, 600794
GAS2L2	183.2	100.0%	99.8%	?Ciliary dyskinesia, primary, 41, 618449
GAS8	134.7	100.0%	99.7%	Ciliary dyskinesia, primary, 33, 616726
GATA1	102.0	99.9%	99.0%	Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835 Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367 Thrombocytopenia with beta-thalassemia, X-linked, 314050
GATA2	128.7	100.0%	99.7%	Emberger syndrome, 614038 Immunodeficiency 21, 614172
GATA3	247.4	100.0%	100.0%	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GATA4	95.7	98.9%	90.6%	?Testicular anomalies with or without congenital heart disease, 615542 Tetralogy of Fallot, 187500 Atrioventricular septal defect 4, 614430 Atrial septal defect 2, 607941 Ventricular septal defect 1, 614429
GATA5	84.0	100.0%	99.5%	Congenital heart defects, multiple types, 5, 617912
GATA6	128.0	99.6%	95.4%	Pancreatic agenesis and congenital heart defects, 600001 Atrial septal defect 9, 614475 Atrioventricular septal defect 5, 614474

				Persistent truncus arteriosus, 217095 Tetralogy of Fallot, 187500
GATAD1	143.2	100.0%	100.0%	?Cardiomyopathy, dilated, 2B, 614672
GATAD2B	101.2	100.0%	99.6%	Mental retardation, autosomal dominant 18, 615074
GATM	139.0	100.0%	100.0%	Cerebral creatine deficiency syndrome 3, 612718
GBA	180.2	100.0%	100.0%	Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 Gaucher disease, type I, 230800 Gaucher disease, perinatal lethal, 608013 Gaucher disease, type II, 230900
GBA2	151.6	100.0%	99.9%	Spastic paraplegia 46, autosomal recessive, 614409
GBE1	152.5	100.0%	99.5%	Polyglucosan body disease, adult form, 263570 Glycogen storage disease IV, 232500
GCDH	158.7	100.0%	99.7%	Glutaricaciduria, type I, 231670
GCH1	91.0	99.9%	99.4%	Hyperphenylalaninemia, BH4-deficient, B, 233910 Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230
GCK	152.9	100.0%	100.0%	Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, permanent neonatal, 606176 MODY, type II, 125851 Hyperinsulinemic hypoglycemia, familial, 3, 602485
GCLC	148.2	99.6%	97.0%	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450
GCM2	142.3	100.0%	100.0%	Hyperparathyroidism 4, 617343 Hypoparathyroidism, familial isolated, 146200
GCNT2	158.5	99.5%	99.5%	Adult i phenotype without cataract, 110800 Cataract 13 with adult i phenotype, 116700
GCSH	32.0	94.3%	74.1%	?Glycine encephalopathy, 605899
GDAP1	151.2	99.9%	98.7%	Charcot-Marie-Tooth disease, recessive intermediate, A, 608340 Charcot-Marie-Tooth disease, type 4A, 214400 Charcot-Marie-Tooth disease, axonal, type 2K, 607831 Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706
GDAP2	116.9	100.0%	99.3%	Spinocerebellar ataxia, autosomal recessive 27, 618369
GDF1	62.1	99.4%	91.2%	Right atrial isomerism (Ivemark), 208530 Congenital heart defects, multiple types, 6, 613854
GDF2	157.3	100.0%	100.0%	Telangiectasia, hereditary hemorrhagic, type 5, 615506
GDF3	132.5	100.0%	100.0%	Microphthalmia, isolated 7, 613704 Microphthalmia with coloboma 6, 613703 Klippel-Feil syndrome 3, autosomal dominant, 613702

GDF5	190.2	100.0%	100.0%	?Acromesomelic dysplasia, Hunter-Thompson type, 201250 Symphalangism, proximal, 1B, 615298 Brachydactyly, type A1, C, 615072 Chondrodysplasia, Grebe type, 200700 Brachydactyly, type A2, 112600 Du Pan syndrome, 228900 Brachydactyly, type C, 113100 Multiple synostoses syndrome 2, 610017
GDF6	180.6	100.0%	100.0%	Leber congenital amaurosis 17, 615360 Klippel-Feil syndrome 1, autosomal dominant, 118100 Multiple synostoses syndrome 4, 617898 Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094
GDF9	148.4	100.0%	100.0%	?Premature ovarian failure 14, 618014
GDI1	145.2	99.4%	97.9%	Mental retardation, X-linked 41, 300849
GDNF	192.6	100.0%	100.0%	Central hypoventilation syndrome, 209880
GEMIN4	159.5	100.0%	99.8%	Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities, 617913
GFAP	111.6	91.9%	91.7%	Alexander disease, 203450
GFER	103.0	100.0%	99.9%	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076
GFI1	118.9	100.0%	99.9%	?Neutropenia, severe congenital 2, autosomal dominant, 613107 ?Neutropenia, nonimmune chronic idiopathic, of adults, 607847
GFI1B	187.0	100.0%	99.1%	Bleeding disorder, platelet-type, 17, 187900
GFM1	104.4	100.0%	98.9%	Combined oxidative phosphorylation deficiency 1, 609060
GFM2	117.8	99.0%	95.2%	Combined oxidative phosphorylation deficiency 39, 618397
GFPT1	142.7	99.9%	99.3%	Myasthenia, congenital, 12, with tubular aggregates, 610542
GGCX	105.2	100.0%	99.6%	Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450 Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842
GGT1	13.5	21.0%	18.9%	?Glutathioninuria, 231950
GH1	167.8	100.0%	100.0%	Kowarski syndrome, 262650 Growth hormone deficiency, isolated, type IA, 262400 Growth hormone deficiency, isolated, type IB, 612781 Growth hormone deficiency, isolated, type II, 173100
GHR	152.0	99.8%	99.8%	Increased responsiveness to growth hormone, 604271 Laron dwarfism, 262500 Growth hormone insensitivity, partial, 604271
GHRHR	115.5	96.3%	95.4%	Growth hormone deficiency, isolated, type IV, 618157
GHSR	194.4	98.7%	95.5%	Growth hormone deficiency, isolated partial, 615925
GIF	110.0	100.0%	99.4%	Intrinsic factor deficiency, 261000

GINS1	126.5	98.9%	89.5%	Immunodeficiency 55, 617827
GIPC3	141.7	99.4%	97.0%	Deafness, autosomal recessive 15, 601869
GJA1	162.4	100.0%	100.0%	Erythrokeratodermia variabilis et progressiva 3, 617525 Craniometaphyseal dysplasia, autosomal recessive, 218400 Atrioventricular septal defect 3, 600309 Oculodentodigital dysplasia, 164200 Syndactyly, type III, 186100 Oculodentodigital dysplasia, autosomal recessive, 257850 Hypoplastic left heart syndrome 1, 241550 Palmoplantar keratoderma with congenital alopecia, 104100
GJA3	185.2	100.0%	100.0%	Cataract 14, multiple types, 601885
GJA5	225.2	100.0%	100.0%	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic (GJA5/SCN5A), 108770
GJA8	173.2	100.0%	100.0%	Cataract 1, multiple types, 116200
GJB1	161.4	100.0%	100.0%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GJB2	151.0	100.0%	100.0%	Deafness, autosomal dominant 3A, 601544 Deafness, autosomal recessive 1A, 220290 Bart-Pumphrey syndrome, 149200 Vohwinkel syndrome, 124500 Keratoderma, palmoplantar, with deafness, 148350 Keratitis-ichthyosis-deafness syndrome, 148210 Hystrix-like ichthyosis with deafness, 602540
GJB3	245.5	100.0%	100.0%	Deafness, autosomal dominant 2B, 612644 Deafness, digenic, GJB2/GJB3, 220290 Erythrokeratodermia variabilis et progressiva 1, 133200 Deafness, autosomal recessive, 0 Deafness, autosomal dominant, with peripheral neuropathy, 0
GJB4	270.0	100.0%	100.0%	Erythrokeratodermia variabilis et progressiva 2, 617524
GJB6	146.2	100.0%	100.0%	Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290 Ectodermal dysplasia 2, Clouston type, 129500
GJC2	59.7	97.7%	86.5%	Spastic paraplegia 44, autosomal recessive, 613206 Lymphatic malformation 3, 613480 Leukodystrophy, hypomyelinating, 2, 608804
GK	43.3	82.3%	61.8%	Glycerol kinase deficiency, 307030
GLA	74.4	99.4%	95.8%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500

GLB1	87.4	99.5%	95.2%	GM1-gangliosidosis, type III, 230650 GM1-gangliosidosis, type I, 230500 Mucopolysaccharidosis type IVB (Morquio), 253010 GM1-gangliosidosis, type II, 230600
GLDC	60.8	91.8%	80.4%	Glycine encephalopathy, 605899
GLDN	112.6	99.8%	97.6%	Lethal congenital contracture syndrome 11, 617194
GLE1	100.8	100.0%	99.9%	Lethal congenital contracture syndrome 1, 253310 Congenital arthrogryposis with anterior horn cell disease, 611890
GLI1	132.1	100.0%	99.9%	Polydactyly, postaxial, type A8, 618123 Polydactyly, preaxial I, 174400
GLI2	177.4	100.0%	100.0%	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829
GLI3	151.8	100.0%	99.5%	Polydactyly, postaxial, types A1 and B, 174200 Greig cephalopolysyndactyly syndrome, 175700 Polydactyly, preaxial, type IV, 174700 Pallister-Hall syndrome, 146510
GLIS2	148.5	100.0%	100.0%	Nephronophthisis 7, 611498
GLIS3	133.1	100.0%	99.6%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GLMN	66.0	99.5%	93.5%	Glomuvenous malformations, 138000
GLRA1	103.2	100.0%	99.8%	Hyperekplexia 1, 149400
GLRB	103.4	99.5%	94.9%	Hyperekplexia 2, 614619
GLRX5	149.3	99.8%	97.8%	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GLS	82.4	99.5%	95.4%	?Infantile cataract, skin abnormalities, glutamate excess, and impaired intellectual development, 618339 Global developmental delay, progressive ataxia, and elevated glutamine, 618412 Epileptic encephalopathy, early infantile, 71, 618328
GLUD1	66.3	98.0%	88.9%	Hyperinsulinism-hyperammonemia syndrome, 606762
GLUL	80.0	99.8%	97.3%	Glutamine deficiency, congenital, 610015
GLYCTK	175.0	100.0%	99.8%	D-glyceric aciduria, 220120
GM2A	129.1	100.0%	100.0%	GM2-gangliosidosis, AB variant, 272750
GMNN	123.0	99.7%	94.8%	Meier-Gorlin syndrome 6, 616835
GMPPA	158.4	100.0%	100.0%	Alacrima, achalasia, and mental retardation syndrome, 615510
GMPPB	233.1	100.0%	100.0%	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	176.2	100.0%	99.8%	Hypocalciuric hypercalcemia, type II, 145981 Hypocalcemia, autosomal dominant 2, 615361

GNAI2	134.4	100.0%	100.0%	Ventricular tachycardia, idiopathic, 192605 Pituitary ACTH-secreting adenoma, 0
GNAI3	88.5	98.9%	93.5%	Auriculocondylar syndrome 1, 602483
GNAL	137.1	99.9%	97.6%	Dystonia 25, 615073
GNAO1	160.9	93.8%	93.8%	Epileptic encephalopathy, early infantile, 17, 615473 Neurodevelopmental disorder with involuntary movements, 617493
GNAQ	53.7	82.9%	66.0%	Sturge-Weber syndrome, somatic, mosaic, 185300 Capillary malformations, congenital, 1, somatic, mosaic, 163000
GNAS	241.4	100.0%	100.0%	ACTH-independent macronodular adrenal hyperplasia, 219080 Pseudohypoparathyroidism Ic, 612462 Pseudohypoparathyroidism Ib, 603233 Pseudopseudohypoparathyroidism, 612463 McCune-Albright syndrome, somatic, mosaic, 174800 Osseous heteroplasia, progressive, 166350 Pituitary adenoma 3, multiple types, somatic, 617686 Pseudohypoparathyroidism Ia, 103580
GNAS-AS1	NC	NC	NC	Pseudohypoparathyroidism, type IB, 603233
GNAT1	197.4	100.0%	100.0%	Night blindness, congenital stationary, type 1G, 616389 Night blindness, congenital stationary, autosomal dominant 3, 610444
GNAT2	106.3	100.0%	98.5%	Achromatopsia 4, 613856
GNB1	148.9	100.0%	100.0%	Mental retardation, autosomal dominant 42, 616973 Leukemia, acute lymphoblastic, somatic, 613065
GNB3	159.3	100.0%	100.0%	Night blindness, congenital stationary, type 1H, 617024
GNB4	148.6	100.0%	100.0%	Charcot-Marie-Tooth disease, dominant intermediate F, 615185
GNB5	119.4	99.9%	98.0%	Intellectual developmental disorder with cardiac arrhythmia, 617173 Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182
GNE	115.7	100.0%	99.3%	Sialuria, 269921 Nonaka myopathy, 605820
GNMT	135.6	99.9%	98.8%	Glycine N-methyltransferase deficiency, 606664
GNPAT	128.8	99.7%	96.6%	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	149.5	100.0%	99.4%	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GNPTG	199.0	99.9%	99.4%	Mucolipidosis III gamma, 252605
GNRH1	82.8	98.1%	82.0%	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841
GNRHR	148.9	100.0%	100.0%	Hypogonadotropic hypogonadism 7 without anosmia, 146110
GNS	93.6	99.9%	97.2%	Mucopolysaccharidosis type IIID, 252940
GORAB	168.3	100.0%	99.1%	Geroderma osteodysplasticum, 231070

GOSR2	108.1	95.9%	94.1%	Epilepsy, progressive myoclonic 6, 614018
GOT1	111.8	100.0%	99.5%	Aspartate aminotransferase, serum level of, QTL1, 614419
GP1BA	144.7	99.1%	96.5%	Bernard-Soulier syndrome, type A1 (recessive), 231200 von Willebrand disease, platelet-type, 177820 Bernard-Soulier syndrome, type A2 (dominant), 153670
GP1BB	85.6	96.1%	86.6%	Giant platelet disorder, isolated, 231200 Bernard-Soulier syndrome, type B, 231200
GP6	135.4	100.0%	100.0%	Bleeding disorder, platelet-type, 11, 614201
GP9	160.5	100.0%	99.5%	Bernard-Soulier syndrome, type C, 231200
GPAA1	137.4	99.9%	98.9%	Glycosylphosphatidylinositol biosynthesis defect 15, 617810
GPC3	76.8	98.9%	93.5%	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPC4	107.1	99.9%	97.7%	Keipert syndrome, 301026
GPC6	132.5	100.0%	100.0%	Omodysplasia 1, 258315
GPD1	94.7	100.0%	99.6%	Hypertriglyceridemia, transient infantile, 614480
GPD1L	132.4	100.0%	99.9%	Brugada syndrome 2, 611777
GPHN	147.8	99.8%	98.8%	Molybdenum cofactor deficiency C, 615501
GPI	152.9	100.0%	100.0%	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470
GPIHBP1	170.2	100.0%	100.0%	Hyperlipoproteinemia, type 1D, 615947
GPNMB	154.7	100.0%	100.0%	Amyloidosis, primary localized cutaneous, 3, 617920
GPR101	118.8	100.0%	100.0%	Pituitary adenoma 2, GH-secreting, 300943
GPR143	60.8	91.8%	81.6%	Ocular albinism, type I, Nettleship-Falls type, 300500 Nystagmus 6, congenital, X-linked, 300814
GPR179	163.2	100.0%	100.0%	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
GPR68	184.6	100.0%	99.6%	Amelogenesis imperfecta, hypomaturation type, IIA6, 617217
GPR88	172.4	100.0%	100.0%	?Chorea, childhood-onset, with psychomotor retardation, 616939
GPRASP2	85.2	100.0%	99.9%	?Deafness, X-linked 7, 301018
GPSM2	121.0	100.0%	99.4%	Chudley-McCullough syndrome, 604213
GPT2	130.4	100.0%	99.6%	Mental retardation, autosomal recessive 49, 616281
GPX4	185.8	95.2%	91.9%	Spondylometaphyseal dysplasia, Sedaghatian type, 250220
GRAP	89.5	87.5%	81.2%	Deafness, autosomal recessive 114, 618456
GREB1L	133.2	100.0%	99.4%	Renal hypodysplasia/aplasia 3, 617805
GREM2	169.2	100.0%	100.0%	Tooth agenesis, selective, 9, 617275
GRHL2	119.8	100.0%	100.0%	Deafness, autosomal dominant 28, 608641 Corneal dystrophy, posterior polymorphous, 4, 618031 Ectodermal dysplasia/short stature syndrome, 616029
GRHL3	141.9	100.0%	99.9%	Van der Woude syndrome 2, 606713

GRHPR	106.8	85.1%	82.3%	Hyperoxaluria, primary, type II, 260000
GRIA3	83.0	98.9%	93.2%	Mental retardation, X-linked 94, 300699
GRIA4	123.6	99.6%	97.9%	Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864
GRID2	148.3	100.0%	99.6%	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRIK2	121.6	96.2%	95.3%	Mental retardation, autosomal recessive, 6, 611092
GRIN1	186.6	100.0%	100.0%	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820 Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254
GRIN2A	139.8	100.0%	100.0%	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570
GRIN2B	168.9	99.9%	99.2%	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation, autosomal dominant 6, 613970
GRIN2D	92.8	96.7%	85.3%	Epileptic encephalopathy, early infantile, 46, 617162
GRIP1	114.2	100.0%	99.4%	Fraser syndrome 3, 617667
GRK1	149.0	100.0%	100.0%	Oguchi disease-2, 613411
GRM1	167.3	100.0%	100.0%	Spinocerebellar ataxia 44, 617691 Spinocerebellar ataxia, autosomal recessive 13, 614831
GRM6	152.8	99.2%	94.6%	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270
GRN	193.7	100.0%	100.0%	Ceroid lipofuscinosis, neuronal, 11, 614706 Aphasia, primary progressive, 607485 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
GRXCR1	158.6	100.0%	99.9%	Deafness, autosomal recessive 25, 613285
GRXCR2	119.1	100.0%	100.0%	?Deafness, autosomal recessive 101, 615837
GSC	149.2	100.0%	99.6%	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471
GSN	123.5	95.6%	93.8%	Amyloidosis, Finnish type, 105120
GSS	98.9	100.0%	99.6%	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900
GSX2	224.1	100.0%	100.0%	Diencephalic-mesencephalic junction dysplasia syndrome 2, 618646
GTF2E2	81.7	100.0%	99.0%	Trichothiodystrophy 6, nonphotosensitive, 616943
GTF2H5	82.0	99.8%	97.4%	Trichothiodystrophy 3, photosensitive, 616395
GTPBP2	138.7	99.9%	99.2%	Jaberi-Elahi syndrome, 617988
GTPBP3	189.1	100.0%	100.0%	Combined oxidative phosphorylation deficiency 23, 616198
GUCA1A	189.2	100.0%	100.0%	Cone-rod dystrophy 14, 602093 Cone dystrophy-3, 602093
GUCA1B	135.9	100.0%	100.0%	Retinitis pigmentosa 48, 613827
GUCY1A3	150.4	100.0%	99.4%	Moyamoya 6 with achalasia, 615750

GUCY2C	116.9	100.0%	99.7%	Diarrhea 6, 614616 Meconium ileus, 614665
GUCY2D	120.6	100.0%	100.0%	Cone-rod dystrophy 6, 601777 Leber congenital amaurosis 1, 204000 Night blindness, congenital stationary, type 1I, 618555 ?Choroidal dystrophy, central areolar 1, 215500
GUF1	91.4	99.9%	97.4%	?Epileptic encephalopathy, early infantile, 40, 617065
GULOP	NC	NC	NC	Scurvy, 0
GUSB	106.6	92.6%	91.1%	Mucopolysaccharidosis VII, 253220
GYG1	126.6	100.0%	99.8%	?Glycogen storage disease XV, 613507 Polyglucosan body myopathy 2, 616199
GYS1	116.9	100.0%	99.2%	Glycogen storage disease 0, muscle, 611556
GYS2	119.6	99.8%	97.5%	Glycogen storage disease 0, liver, 240600
GZF1	205.4	100.0%	99.6%	Joint laxity, short stature, and myopia, 617662
H19	NC	NC	NC	Wilms tumor 2, 194071 Silver-Russell syndrome, 180860 Beckwith-Wiedemann syndrome, 130650
H6PD	219.1	99.0%	99.0%	Cortisone reductase deficiency 1, 604931
HAAO	113.2	100.0%	100.0%	Vertebral, cardiac, renal, and limb defects syndrome 1, 617660
HACE1	135.7	100.0%	99.4%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADH	118.2	99.3%	99.2%	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HADHA	74.6	96.1%	89.6%	LCHAD deficiency, 609016 HELLP syndrome, maternal, of pregnancy, 609016 Fatty liver, acute, of pregnancy, 609016 Trifunctional protein deficiency, 609015
HADHB	76.9	96.0%	83.7%	Trifunctional protein deficiency, 609015
HAMP	185.1	100.0%	100.0%	Hemochromatosis, type 2B, 613313
HARS	142.4	100.0%	100.0%	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504
HARS2	141.0	100.0%	99.9%	?Perrault syndrome 2, 614926
HAVCR2	122.6	100.0%	100.0%	T-cell lymphoma, subcutaneous panniculitis-like, 618398
HAX1	146.3	100.0%	100.0%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HBA1	149.4	100.0%	100.0%	Hemoglobin H disease, nondeletional, 613978 Thalassemias, alpha-, 604131 Erythrocytosis, 7, 617981

				Methemoglobinemia, alpha type, 617973 Heinz body anemias, alpha-, 140700
HBA2	140.4	99.4%	95.2%	Thalassemia, alpha-, 604131 Erythrocytosis 7, 617981 Heinz body anemia, 140700 Hemoglobin H disease, deletional and nondeletional, 613978
HBB	137.7	100.0%	100.0%	Thalassemia, beta, 613985 Methmoglobinemia, beta type, 617971 Erythrocytosis 6, 617980 Heinz body anemia, 140700 Delta-beta thalassemia, 141749 Thalassemia-beta, dominant inclusion-body, 603902 Hereditary persistence of fetal hemoglobin, 141749 Sickle cell anemia, 603903
HBD	189.5	100.0%	100.0%	Thalassemia, delta-, 0 Thalassemia due to Hb Lepore, 0
HBG1	139.5	98.5%	94.3%	Fetal hemoglobin quantitative trait locus 1, 141749
HBG2	257.7	100.0%	100.0%	Fetal hemoglobin quantitative trait locus 1, 141749 Cyanosis, transient neonatal, 613977
HCCS	90.4	99.6%	96.6%	Linear skin defects with multiple congenital anomalies 1, 309801
HCFC1	114.5	99.5%	96.5%	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541
HCN1	144.1	100.0%	99.9%	Generalized epilepsy with febrile seizures plus, type 10, 618482 Epileptic encephalopathy, early infantile, 24, 615871
HCN4	109.8	100.0%	99.9%	Brugada syndrome 8, 613123 Sick sinus syndrome 2, 163800
HCRT	144.8	99.9%	95.6%	?Narcolepsy 1, 161400
HDAC6	122.8	99.8%	98.0%	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863
HDAC8	110.1	100.0%	99.6%	Cornelia de Lange syndrome 5, 300882
HECW2	112.4	99.9%	98.8%	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268
HELLS	102.2	98.1%	91.0%	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911
HEPACAM	127.8	95.0%	89.6%	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926
HEPHL1	109.1	100.0%	99.8%	?Abnormal hair, joint laxity, and developmental delay, 261990
HERC1	145.0	100.0%	99.7%	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
HERC2	99.7	80.6%	76.7%	Mental retardation, autosomal recessive 38, 615516
HES7	61.9	93.1%	78.9%	Spondylocostal dysostosis 4, autosomal recessive, 613686

HESX1	65.7	99.9%	97.5%	Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230 Growth hormone deficiency with pituitary anomalies, 182230
HEXA	112.3	93.8%	92.6%	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800
HEXB	173.2	99.8%	97.3%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HFE	114.7	100.0%	99.1%	Hemochromatosis, 235200
HFE2	162.3	100.0%	100.0%	Hemochromatosis, type 2A, 602390
HFM1	50.4	96.4%	88.2%	Premature ovarian failure 9, 615724
HGD	101.4	100.0%	99.7%	Alkaptonuria, 203500
HGF	134.3	99.9%	99.3%	Deafness, autosomal recessive 39, 608265
HGSNAT	99.9	88.2%	86.3%	Retinitis pigmentosa 73, 616544 Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930
HIBCH	69.7	95.5%	75.9%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HIKESHI	53.5	94.8%	82.6%	Leukodystrophy, hypomyelinating, 13, 616881
HINT1	65.5	92.7%	79.4%	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200
HIST1H1E	125.1	100.0%	100.0%	Rahman syndrome, 617537
HIVEP2	171.1	100.0%	100.0%	Mental retardation, autosomal dominant 43, 616977
HK1	123.7	100.0%	99.6%	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285 Neurodevelopmental disorder with visual defects and brain anomalies, 618547 Retinitis pigmentosa 79, 617460
HLCS	148.0	100.0%	100.0%	Holocarboxylase synthetase deficiency, 253270
HMBS	102.8	100.0%	99.0%	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000
HMGB3	40.2	84.0%	66.6%	?Microphthalmia, syndromic 13, 300915
HMGCL	124.9	100.0%	99.5%	HMG-CoA lyase deficiency, 246450
HMGCS2	107.6	100.0%	99.6%	HMG-CoA synthase-2 deficiency, 605911
HMOX1	153.8	98.0%	91.0%	Heme oxygenase-1 deficiency, 614034
HMX1	47.2	89.3%	70.6%	Oculoauricular syndrome, 612109
HNF1A	179.0	100.0%	100.0%	MODY, type III, 600496 Hepatic adenoma, somatic, 142330 Renal cell carcinoma, 144700 Diabetes mellitus, insulin-dependent, 20, 612520
HNF1B	130.8	99.8%	97.9%	Renal cysts and diabetes syndrome, 137920 Diabetes mellitus, noninsulin-dependent, 125853

HNF4A	140.7	100.0%	99.3%	MODY, type I, 125850 Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026
HNMT	132.7	100.0%	99.7%	Mental retardation, autosomal recessive 51, 616739
HNRNPA1	63.5	99.0%	87.2%	Amyotrophic lateral sclerosis 20, 615426 ?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3, 615424
HNRNPA2B1	138.0	99.8%	98.0%	?Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2, 615422
HNRNPDL	93.2	99.4%	94.9%	Muscular dystrophy, limb-girdle, autosomal dominant 3, 609115
HNRNPH2	131.4	100.0%	100.0%	Mental retardation, X-linked, syndromic, Bain type, 300986
HNRNPK	61.0	87.9%	78.3%	Au-Kline syndrome, 616580
HNRNPU	154.7	100.0%	99.3%	Epileptic encephalopathy, early infantile, 54, 617391
HOGA1	163.0	100.0%	99.5%	Hyperoxaluria, primary, type III, 613616
HOMER2	120.6	99.8%	98.5%	?Deafness, autosomal dominant 68, 616707
HOXA1	184.7	100.0%	100.0%	Athabaskan brainstem dysgenesis syndrome, 601536 Bosley-Salih-Alorainy syndrome, 601536
HOXA11	96.7	99.8%	97.7%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
HOXA13	90.2	95.8%	84.5%	?Guttmacher syndrome, 176305 Hand-foot-uterus syndrome, 140000
HOXA2	90.9	100.0%	100.0%	Microtia with or without hearing impairment (AD), 612290 ?Microtia, hearing impairment, and cleft palate (AR), 612290
HOXB1	165.3	100.0%	100.0%	Facial paresis, hereditary congenital, 3, 614744
HOXC13	197.1	100.0%	100.0%	Ectodermal dysplasia 9, hair/nail type, 614931
HOXD10	168.0	100.0%	100.0%	Charcot-Marie-Tooth disease, foot deformity of, 192950 Vertical talus, congenital, 192950
HOXD13	222.6	100.0%	100.0%	Brachydactyly, type D, 113200 Brachydactyly, type E, 113300 ?Brachydactyly-syndactyly syndrome, 610713 Syndactyly, type V, 186300 Synpolydactyly 1, 186000
HPCA	279.8	100.0%	100.0%	Dystonia 2, torsion, autosomal recessive, 224500
HPD	159.7	100.0%	99.9%	Tyrosinemia, type III, 276710 Hawkinsinuria, 140350
HPGD	87.8	100.0%	99.7%	Digital clubbing, isolated congenital, 119900 Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100 Cranioosteoarthropathy, 259100
HPRT1	56.9	97.8%	87.8%	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322
HPS1	125.7	100.0%	100.0%	Hermansky-Pudlak syndrome 1, 203300

HPS3	133.9	99.9%	98.2%	Hermansky-Pudlak syndrome 3, 614072
HPS4	135.2	100.0%	100.0%	Hermansky-Pudlak syndrome 4, 614073
HPS5	122.5	99.9%	98.9%	Hermansky-Pudlak syndrome 5, 614074
HPS6	183.5	100.0%	99.2%	Hermansky-Pudlak syndrome 6, 614075
HPSE2	105.3	100.0%	99.9%	Urofacial syndrome 1, 236730
HR	133.4	99.7%	98.0%	Hypotrichosis 4, 146550 Alopecia universalis, 203655 Atrichia with papular lesions, 209500
HRAS	196.0	100.0%	100.0%	Nevus sebaceous or woolly hair nevus, somatic, 162900 Congenital myopathy with excess of muscle spindles, 218040 Bladder cancer, somatic, 109800 Thyroid carcinoma, follicular, somatic, 188470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Spitz nevus or nevus spilus, somatic, 137550 Costello syndrome, 218040
HRG	130.2	94.7%	94.2%	Thrombophilia due to HRG deficiency, 613116
HS6ST2	145.1	97.4%	96.9%	?Paganini-Miozzo syndrome, 301025
HSD11B1	116.2	100.0%	99.3%	Cortisone reductase deficiency 2, 614662
HSD11B2	183.2	95.8%	89.4%	Apparent mineralocorticoid excess, 218030
HSD17B10	98.0	100.0%	99.5%	HSD10 mitochondrial disease, 300438
HSD17B3	119.0	100.0%	100.0%	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	106.4	95.5%	93.1%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B2	137.1	100.0%	99.9%	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810
HSD3B7	157.0	99.8%	97.4%	Bile acid synthesis defect, congenital, 1, 607765
HSF4	166.2	99.9%	99.3%	Cataract 5, multiple types, 116800
HSPA9	83.8	88.2%	84.2%	Even-plus syndrome, 616854 Anemia, sideroblastic, 4, 182170
HSPB1	77.7	98.0%	92.7%	Charcot-Marie-Tooth disease, axonal, type 2F, 606595 Neuropathy, distal hereditary motor, type IIB, 608634
HSPB3	238.7	100.0%	100.0%	?Neuronopathy, distal hereditary motor, type IIC, 613376
HSPB8	235.6	100.0%	100.0%	Neuropathy, distal hereditary motor, type IIA, 158590 Charcot-Marie-Tooth disease, axonal, type 2L, 608673
HSPD1	73.7	97.9%	92.1%	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233
HSPG2	132.7	99.5%	99.2%	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800

HTR1A	208.2	100.0%	100.0%	Periodic fever, menstrual cycle dependent, 614674
HTRA1	95.8	98.9%	91.1%	Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779 CARASIL syndrome, 600142
HTRA2	145.3	100.0%	99.7%	3-methylglutaconic aciduria, type VIII, 617248
HTT	130.5	98.9%	97.7%	Huntington disease, 143100 Lopes-Maciel-Rodan syndrome, 617435
HUWE1	82.1	99.1%	95.0%	Mental retardation, X-linked syndromic, Turner type, 309590
HYAL1	121.3	100.0%	100.0%	?Mucopolysaccharidosis type IX, 601492
HYDIN	111.4	99.9%	99.2%	Ciliary dyskinesia, primary, 5, 608647
HYLS1	160.4	100.0%	100.0%	Hydrolethalus syndrome, 236680
HYOU1	140.5	100.0%	99.7%	?Immunodeficiency 59 and hypoglycemia, 233600
IARS	124.2	99.9%	99.2%	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093
IARS2	145.7	100.0%	100.0%	?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
IBA57	162.1	99.8%	98.5%	?Spastic paraplegia 74, autosomal recessive, 616451 Multiple mitochondrial dysfunctions syndrome 3, 615330
ICK	111.0	100.0%	98.8%	Endocrine-cerebroosteodysplasia, 612651
ICOS	154.5	100.0%	99.9%	Immunodeficiency, common variable, 1, 607594
IDH2	107.4	100.0%	99.6%	D-2-hydroxyglutaric aciduria 2, 613657
IDH3B	136.5	95.8%	95.4%	Retinitis pigmentosa 46, 612572
IDS	105.1	99.8%	97.2%	Mucopolysaccharidosis II, 309900
IDUA	169.2	99.3%	96.4%	Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Is, 607016
IER3IP1	108.7	88.3%	80.0%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	110.9	99.8%	98.2%	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFITM5	104.5	100.0%	99.6%	Osteogenesis imperfecta, type V, 610967
IFNAR2	133.4	99.6%	97.5%	?Immunodeficiency 45, 616669
IFNGR1	145.4	100.0%	99.2%	Immunodeficiency 27A, mycobacteriosis, AR, 209950 Immunodeficiency 27B, mycobacteriosis, AD, 615978
IFNGR2	129.9	97.9%	94.1%	Immunodeficiency 28, mycobacteriosis, 614889
IFT122	126.6	100.0%	99.6%	Cranioectodermal dysplasia 1, 218330
IFT140	127.6	100.0%	99.6%	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	98.4	100.0%	99.5%	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630

IFT27	122.9	100.0%	99.9%	?Bardet-Biedl syndrome 19, 615996
IFT43	119.5	100.0%	100.0%	?Cranioectodermal dysplasia 3, 614099 Short-rib thoracic dysplasia 18 with polydactyly, 617866 ?Retinitis pigmentosa 81, 617871
IFT52	120.0	100.0%	99.7%	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102
IFT57	119.3	99.9%	99.2%	?Orofaciodigital syndrome XVIII, 617927
IFT74	81.5	99.4%	93.7%	?Bardet-Biedl syndrome 20, 617119
IFT80	61.7	95.6%	81.0%	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IFT81	90.5	93.0%	88.0%	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IGBP1	106.4	98.8%	93.9%	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472
IGF1	100.6	100.0%	100.0%	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IGF1R	123.1	100.0%	99.7%	Insulin-like growth factor I, resistance to, 270450
IGF2	131.1	100.0%	100.0%	?Growth restriction, severe, with distinctive facies, 616489
IGF2R	118.7	99.8%	98.4%	Hepatocellular carcinoma, somatic, 114550
IGFALS	126.7	100.0%	100.0%	Acid-labile subunit, deficiency of, 615961
IGFBP7	91.3	99.8%	96.5%	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis, 614224
IGHG2	29.1	76.6%	56.2%	IgG2 deficiency, selective, 0
IGHM	190.2	100.0%	100.0%	Agammaglobulinemia 1, 601495
IGHMBP2	117.7	99.9%	98.2%	Neuronopathy, distal hereditary motor, type VI, 604320 Charcot-Marie-Tooth disease, axonal, type 2S, 616155
IGKC	119.8	100.0%	100.0%	Kappa light chain deficiency, 614102
IGLL1	99.7	100.0%	99.8%	Agammaglobulinemia 2, 613500
IGSF1	73.1	99.5%	95.2%	Hypothyroidism, central, and testicular enlargement, 300888
IGSF3	105.0	96.6%	94.0%	?Lacrimal duct defect, 149700
IHH	198.7	100.0%	100.0%	Acrocapitofemoral dysplasia, 607778 Brachydactyly, type A1, 112500
IKBKB	116.2	99.4%	97.1%	Immunodeficiency 15A, 618204 Immunodeficiency 15B, 615592
IKBKG	64.7	90.1%	80.2%	Immunodeficiency 33, 300636 Incontinentia pigmenti, 308300 Immunodeficiency, isolated, 300584 Ectodermal dysplasia and immunodeficiency 1, 300291 Invasive pneumococcal disease, recurrent isolated, 2, 300640 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301
IKZF1	196.9	100.0%	100.0%	Immunodeficiency, common variable, 13, 616873
IL10RA	158.7	100.0%	99.9%	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	132.0	100.0%	99.6%	Inflammatory bowel disease 25, early onset, autosomal recessive, 612567

IL11RA	141.0	100.0%	99.8%	Craniosynostosis and dental anomalies, 614188
IL12B	99.2	99.9%	97.5%	Immunodeficiency 29, mycobacteriosis, 614890
IL12RB1	119.9	98.2%	95.7%	Immunodeficiency 30, 614891
IL17F	76.7	98.8%	93.0%	?Candidiasis, familial, 6, autosomal dominant, 613956
IL17RA	167.0	100.0%	100.0%	Immunodeficiency 51, 613953
IL17RC	139.8	100.0%	100.0%	Candidiasis, familial, 9, 616445
IL17RD	143.4	99.9%	99.3%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
IL1RAPL1	100.0	99.5%	97.6%	Mental retardation, X-linked 21/34, 300143
IL1RN	145.3	100.0%	99.9%	Interleukin 1 receptor antagonist deficiency, 612852
IL21	74.7	99.9%	94.7%	?Immunodeficiency, common variable, 11, 615767
IL21R	160.2	100.0%	100.0%	Immunodeficiency 56, 615207
IL2RA	106.8	100.0%	99.2%	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367
IL2RB	120.0	100.0%	99.7%	Immunodeficiency 63 with lymphoproliferation and autoimmunity, 618495
IL2RG	60.2	99.7%	94.3%	Severe combined immunodeficiency, X-linked, 300400 Combined immunodeficiency, X-linked, moderate, 312863
IL31RA	113.8	99.9%	99.5%	?Amyloidosis, primary localized cutaneous, 2, 613955
IL36RN	102.7	100.0%	99.9%	Psoriasis 14, pustular, 614204
IL6ST	83.7	94.9%	88.0%	Hyper-IgE recurrent infection syndrome 4, autosomal recessive, 618523
IL7R	114.7	100.0%	99.7%	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
ILDR1	122.9	99.9%	98.6%	Deafness, autosomal recessive 42, 609646
IMPA1	73.2	96.0%	84.6%	Mental retardation, autosomal recessive 59, 617323
IMPAD1	181.6	100.0%	100.0%	Chondrodysplasia with joint dislocations, GPAPP type, 614078
IMPDH1	57.4	97.6%	87.3%	Leber congenital amaurosis 11, 613837 Retinitis pigmentosa 10, 180105
IMPG1	92.5	99.8%	98.2%	Macular dystrophy, vitelliform, 4, 616151
IMPG2	127.1	99.5%	98.0%	Macular dystrophy, vitelliform, 5, 616152 Retinitis pigmentosa 56, 613581
INF2	112.4	86.1%	84.3%	Glomerulosclerosis, focal segmental, 5, 613237 Charcot-Marie-Tooth disease, dominant intermediate E, 614455
ING1	175.4	100.0%	100.0%	Squamous cell carcinoma, head and neck, somatic, 275355
INPP5E	131.1	100.0%	99.3%	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300
INPP5K	94.7	100.0%	99.5%	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404
INPPL1	139.7	99.9%	98.9%	Opsismodysplasia, 258480
INS	130.5	100.0%	99.8%	Maturity-onset diabetes of the young, type 10, 613370 Hyperproinsulinemia, 616214

				Diabetes mellitus, insulin-dependent, 2, 125852 Diabetes mellitus, permanent neonatal, 606176
INSL3	75.2	81.1%	79.9%	Cryptorchidism, 219050
INSR	123.5	99.4%	96.1%	Hyperinsulinemic hypoglycemia, familial, 5, 609968 Rabson-Mendenhall syndrome, 262190 Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Leprechaunism, 246200
INTS1	140.4	99.8%	99.2%	Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies, 618571
INTS8	110.4	99.8%	98.2%	?Neurodevelopmental disorder with cerebellar hypoplasia and spasticity, 618572
INTU	112.9	99.8%	98.0%	?Short-rib thoracic dysplasia 20 with polydactyly, 617925 ?Orofaciodigital syndrome XVII, 617926
INVS	147.7	100.0%	100.0%	Nephronophthisis 2, infantile, 602088
IQCB1	90.6	91.0%	79.0%	Senior-Loken syndrome 5, 609254
IQCE	141.3	100.0%	99.4%	?Polydactyly, postaxial, type A7, 617642
IQSEC2	81.5	96.6%	90.5%	Mental retardation, X-linked 1/78, 309530
IRAK4	100.7	99.7%	94.9%	IRAK4 deficiency, 607676 Invasive pneumococcal disease, recurrent isolated, 1, 610799
IREB2	129.0	100.0%	99.8%	Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451
IRF1	148.6	100.0%	100.0%	Gastric cancer, somatic, 613659 Nonsmall cell lung cancer, somatic, 211980 Myelogenous leukemia, acute, 0 Myelodysplastic syndrome, preleukemic, 0
IRF2BP2	98.4	100.0%	99.7%	?Immunodeficiency, common variable, 14, 617765
IRF2BPL	197.8	99.6%	97.8%	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088
IRF6	94.5	99.4%	95.8%	Popliteal pterygium syndrome 1, 119500 van der Woude syndrome, 119300
IRF7	174.6	100.0%	99.9%	?Immunodeficiency 39, 616345
IRF8	123.3	100.0%	99.1%	Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990 Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893
IRF9	143.0	100.0%	99.5%	Immunodeficiency 65, susceptibility to viral infections, 618648
IRS4	164.0	100.0%	100.0%	Hypothyroidism, congenital, nongoitrous, 9, 301035
IRX5	156.2	100.0%	99.9%	Hamamy syndrome, 611174
ISCA1	71.9	94.0%	82.9%	Multiple mitochondrial dysfunctions syndrome 5, 617613
ISCA2	112.2	99.8%	97.4%	Multiple mitochondrial dysfunctions syndrome 4, 616370
ISCU	121.3	100.0%	99.9%	Myopathy with lactic acidosis, hereditary, 255125
ISG15	207.0	100.0%	100.0%	Immunodeficiency 38, 616126

ISPD	112.0	99.7%	97.8%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
ITCH	115.3	95.5%	94.5%	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITGA2B	137.3	100.0%	99.4%	Glanzmann thrombasthenia, 273800 Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Thrombocytopenia, neonatal alloimmune, BAK antigen related, 0
ITGA3	162.5	99.4%	98.1%	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748
ITGA6	144.0	99.9%	99.3%	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730
ITGA7	142.3	99.8%	98.4%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
ITGA8	117.1	100.0%	99.8%	Renal hypodysplasia/aplasia 1, 191830
ITGB2	171.3	100.0%	100.0%	Leukocyte adhesion deficiency, 116920
ITGB3	119.9	100.0%	100.0%	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Glanzmann thrombasthenia, 273800 Purpura, posttransfusion, 0 Thrombocytopenia, neonatal alloimmune, 0
ITGB4	166.0	99.4%	98.0%	Epidermolysis bullosa of hands and feet, 131800 Epidermolysis bullosa, junctional, with pyloric atresia, 226730 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
ITGB6	129.5	96.3%	95.1%	Amelogenesis imperfecta, type IH, 616221
ITK	105.3	99.9%	99.3%	Lymphoproliferative syndrome 1, 613011
ITM2B	134.8	100.0%	100.0%	Dementia, familial British, 176500 ?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079 Dementia, familial Danish, 117300
ITPA	142.5	100.0%	100.0%	Epileptic encephalopathy, early infantile, 35, 616647
ITPR1	136.4	100.0%	99.8%	Spinocerebellar ataxia 29, congenital nonprogressive, 117360 Spinocerebellar ataxia 15, 606658 Gillespie syndrome, 206700
ITPR2	127.9	99.8%	98.1%	?Anhidrosis, isolated, with normal sweat glands, 106190
IVD	106.7	100.0%	100.0%	Isovaleric acidemia, 243500
IYD	108.7	99.3%	94.8%	Thyroid dyshormonogenesis 4, 274800
JAG1	143.4	99.4%	97.6%	Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500 ?Deafness, congenital heart defects, and posterior embryotoxon, 617992
JAGN1	129.4	100.0%	100.0%	Neutropenia, severe congenital, 6, autosomal recessive, 616022
JAK2	100.6	97.1%	95.0%	Myelofibrosis, somatic, 254450 Thrombocythemia 3, 614521 Polycythemia vera, somatic, 263300

				Leukemia, acute myeloid, somatic, 601626 Erythrocytosis, somatic, 133100
JAK3	134.3	98.7%	97.3%	SCID, autosomal recessive, T-negative/B-positive type, 600802
JAM3	132.0	100.0%	100.0%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
JPH1	187.8	100.0%	100.0%	?Charcot-Marie-Tooth disease, axonal, autosomal dominant, type 2K, 607831
JPH2	139.7	99.7%	97.8%	Cardiomyopathy, hypertrophic, 17, 613873
JPH3	225.4	100.0%	99.9%	Huntington disease-like 2, 606438
JUP	137.0	100.0%	99.9%	Arrhythmogenic right ventricular dysplasia 12, 611528 Naxos disease, 601214
KANK1	129.0	100.0%	99.7%	Cerebral palsy, spastic quadriplegic, 2, 612900
KANK2	180.1	100.0%	100.0%	Nephrotic syndrome, type 16, 617783 Palmoplantar keratoderma and woolly hair, 616099
KANSL1	156.7	99.8%	99.1%	Koolen-De Vries syndrome, 610443
KARS	109.9	100.0%	99.3%	Deafness, autosomal recessive 89, 613916 ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641
KAT6A	155.1	100.0%	99.9%	Mental retardation, autosomal dominant 32, 616268
KAT6B	162.3	99.8%	99.2%	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KATNB1	170.5	100.0%	100.0%	Lissencephaly 6, with microcephaly, 616212
KBTBD13	208.9	100.0%	100.0%	Nemaline myopathy 6, autosomal dominant, 609273
KCNA1	164.5	100.0%	100.0%	Episodic ataxia/myokymia syndrome, 160120
KCNA2	132.6	100.0%	99.4%	Epileptic encephalopathy, early infantile, 32, 616366
KCNA4	128.0	100.0%	100.0%	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284
KCNA5	173.9	100.0%	100.0%	Atrial fibrillation, familial, 7, 612240
KCNB1	141.6	100.0%	99.8%	Epileptic encephalopathy, early infantile, 26, 616056
KCNC1	189.4	100.0%	100.0%	Epilepsy, progressive myoclonic 7, 616187
KCNC3	126.1	94.7%	80.0%	Spinocerebellar ataxia 13, 605259
KCND3	180.1	99.9%	99.3%	Brugada syndrome 9, 616399 Spinocerebellar ataxia 19, 607346
KCNE1	398.8	100.0%	100.0%	Long QT syndrome 5, 613695 Jervell and Lange-Nielsen syndrome 2, 612347
KCNE2	127.8	100.0%	98.4%	Atrial fibrillation, familial, 4, 611493 Long QT syndrome 6, 613693
KCNE3	149.5	100.0%	100.0%	?Brugada syndrome 6, 613119
KCNH1	159.4	98.7%	98.4%	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500

KCNH2	120.8	99.0%	96.3%	Long QT syndrome 2, 613688 Short QT syndrome 1, 609620
KCNJ1	159.7	100.0%	100.0%	Bartter syndrome, type 2, 241200
KCNJ10	157.5	89.3%	88.6%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ11	222.1	100.0%	100.0%	Maturity-onset diabetes of the young, type 13, 616329 Diabetes, permanent neonatal, with or without neurologic features, 606176 Diabetes mellitus, transient neonatal, 3, 610582 Hyperinsulinemic hypoglycemia, familial, 2, 601820
KCNJ13	142.5	100.0%	100.0%	Leber congenital amaurosis 16, 614186 Snowflake vitreoretinal degeneration, 193230
KCNJ2	164.2	100.0%	100.0%	Short QT syndrome 3, 609622 Atrial fibrillation, familial, 9, 613980 Andersen syndrome, 170390
KCNJ5	171.5	100.0%	100.0%	Long QT syndrome 13, 613485 Hyperaldosteronism, familial, type III, 613677
KCNJ6	165.7	100.0%	100.0%	Keppen-Lubinsky syndrome, 614098
KCNK3	184.4	100.0%	99.7%	Pulmonary hypertension, primary, 4, 615344
KCNK4	221.2	100.0%	100.0%	Facial dysmorphism, hypertrichosis, epilepsy, intellectual/developmental delay, and gingival overgrowth syndrome, 618381
KCNK9	189.3	100.0%	100.0%	Birk-Barel mental retardation dysmorphism syndrome, 612292
KCNMA1	107.9	94.9%	93.6%	Cerebellar atrophy, developmental delay, and seizures, 617643 Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446
KCNN3	130.4	100.0%	99.9%	Zimmermann-Laband syndrome 3, 618658
KCNN4	169.8	100.0%	99.8%	Dehydrated hereditary stomatocytosis 2, 616689
KCNQ1	150.5	98.9%	96.5%	Long QT syndrome 1, 192500 Jervell and Lange-Nielsen syndrome, 220400 Short QT syndrome 2, 609621 Atrial fibrillation, familial, 3, 607554
KCNQ1OT1	NC	NC	NC	Beckwith-Wiedemann syndrome, 130650
KCNQ2	133.0	91.5%	90.4%	Epileptic encephalopathy, early infantile, 7, 613720 Seizures, benign neonatal, 1, 121200 Myokymia, 121200
KCNQ3	116.5	100.0%	98.7%	Seizures, benign neonatal, 2, 121201
KCNQ4	182.4	99.8%	99.0%	Deafness, autosomal dominant 2A, 600101
KCNQ5	140.1	99.7%	98.5%	Mental retardation, autosomal dominant 46, 617601
KCNT1	145.3	96.3%	95.2%	Epilepsy, nocturnal frontal lobe, 5, 615005 Epileptic encephalopathy, early infantile, 14, 614959

KCNT2	101.7	99.7%	96.1%	?Epileptic encephalopathy, early infantile, 57, 617771
KCNV2	154.7	100.0%	100.0%	Retinal cone dystrophy 3B, 610356
KCTD1	100.6	100.0%	100.0%	Scalp-ear-nipple syndrome, 181270
KCTD17	126.9	100.0%	99.7%	Dystonia 26, myoclonic, 616398
KCTD7	171.1	95.0%	95.0%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDF1	120.4	100.0%	100.0%	?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/nail type, 617337
KDM1A	134.2	100.0%	99.4%	Cleft palate, psychomotor retardation, and distinctive facial features, 616728
KDM5B	123.2	99.1%	97.2%	Mental retardation, autosomal recessive 65, 618109
KDM5C	111.1	99.6%	97.5%	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
KDM6A	98.1	95.6%	87.7%	Kabuki syndrome 2, 300867
KDM6B	160.4	99.9%	98.1%	Neurodevelopmental disorder with coarse facies and mild distal skeletal abnormalities, 618505
KDR	118.5	100.0%	99.7%	Hemangioma, capillary infantile, somatic, 602089
KDSR	165.5	100.0%	99.5%	Erythrokeratodermia variabilis et progressiva 4, 617526
KERA	174.8	100.0%	100.0%	Cornea plana 2, autosomal recessive, 217300
KHDC3L	174.6	100.0%	100.0%	Hydatidiform mole, recurrent, 2, 614293
KIAA0556	135.2	100.0%	99.8%	Joubert syndrome 26, 616784
KIAA0586	115.1	97.3%	92.6%	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546
KIAA0753	117.1	99.9%	98.9%	?Orofaciodigital syndrome XV, 617127
KIAA0825	109.6	99.7%	98.2%	Polydactyly, postaxial, type A10, 618498
KIAA1109	137.8	99.8%	98.9%	Alkuraya-Kucinskas syndrome, 617822
KIAA1161	284.1	100.0%	100.0%	Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317
KIAA1549	125.5	98.7%	97.6%	Retinitis pigmentosa 86, 618613
KIDINS220	138.5	100.0%	99.9%	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296
KIF11	89.7	97.6%	94.9%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF14	112.4	99.8%	97.7%	?Meckel syndrome 12, 616258 Microcephaly 20, primary, autosomal recessive, 617914
KIF1A	125.3	99.8%	98.2%	Neuropathy, hereditary sensory, type IIC, 614213 Mental retardation, autosomal dominant 9, 614255 Spastic paraplegia 30, autosomal recessive, 610357
KIF1B	144.0	100.0%	99.7%	Pheochromocytoma, 171300 ?Charcot-Marie-Tooth disease, type 2A1, 118210
KIF1BP	168.4	96.1%	96.1%	Goldberg-Shprintzen megacolon syndrome, 609460
KIF1C	163.4	100.0%	99.5%	Spastic ataxia 2, autosomal recessive, 611302
KIF21A	118.6	99.9%	98.8%	Fibrosis of extraocular muscles, congenital, 1, 135700 Fibrosis of extraocular muscles, congenital, 3B, 135700
KIF22	188.7	100.0%	99.9%	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546

KIF2A	103.8	99.5%	95.9%	Cortical dysplasia, complex, with other brain malformations 3, 615411
KIF4A	78.5	98.5%	93.1%	?Mental retardation, X-linked 100, 300923
KIF5A	121.9	100.0%	100.0%	Myoclonus, intractable, neonatal, 617235 Spastic paraplegia 10, autosomal dominant, 604187
KIF5C	113.8	100.0%	99.0%	Cortical dysplasia, complex, with other brain malformations 2, 615282
KIF7	120.4	99.3%	96.6%	?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131
KISS1	90.8	100.0%	99.1%	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842
KISS1R	180.8	100.0%	100.0%	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 ?Precocious puberty, central, 1, 176400
KIT	137.7	100.0%	99.7%	Gastrointestinal stromal tumor, familial, 606764 Mastocytosis, cutaneous, 154800 Germ cell tumors, somatic, 273300 Leukemia, acute myeloid, somatic, 601626 Mastocytosis, systemic, somatic, 154800 Piebaldism, 172800
KITLG	82.5	99.8%	96.5%	Hyperpigmentation with or without hypopigmentation, 145250 Deafness, autosomal dominant 69, unilateral or asymmetric, 616697
KIZ	151.3	99.8%	97.7%	Retinitis pigmentosa 69, 615780
KL	185.9	99.8%	98.9%	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994
KLC2	143.0	100.0%	99.5%	Spastic paraplegia, optic atrophy, and neuropathy, 609541
KLF1	132.7	100.0%	100.0%	Blood group--Lutheran inhibitor, 111150 Dyserythropoietic anemia, congenital, type IV, 613673
KLF11	176.8	100.0%	99.8%	Maturity-onset diabetes of the young, type VII, 610508
KLF6	164.3	100.0%	100.0%	Gastric cancer, somatic, 613659 Prostate cancer, somatic, 176807
KLHL10	146.2	100.0%	100.0%	Spermatogenic failure 11, 615081
KLHL15	143.1	100.0%	99.9%	Mental retardation, X-linked 103, 300982
KLHL24	174.7	100.0%	100.0%	Epidermolysis bullosa simplex, generalized, with scarring and hair loss, 617294
KLHL3	110.5	99.9%	98.3%	Pseudohypoaldosteronism, type IID, 614495
KLHL40	144.2	100.0%	100.0%	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	174.8	100.0%	99.6%	Nemaline myopathy 9, 615731
KLHL7	116.8	100.0%	99.9%	Retinitis pigmentosa 42, 612943 Cold-induced sweating syndrome 3, 617055
KLK4	176.5	100.0%	99.8%	Amelogenesis imperfecta, type IIA1, 204700

KLKB1	132.4	99.9%	99.2%	Fletcher factor (prekallikrein) deficiency, 612423
KLLN	172.4	100.0%	100.0%	Cowden syndrome 4, 615107
KMT2A	138.5	100.0%	99.9%	Wiedemann-Steiner syndrome, 605130
KMT2B	156.7	98.1%	94.7%	Dystonia 28, childhood-onset, 617284
KMT2C	142.7	91.8%	90.3%	Kleefstra syndrome 2, 617768
KMT2D	150.7	100.0%	99.9%	Kabuki syndrome 1, 147920
KMT2E	156.1	99.9%	98.4%	O'Donnell-Luria-Rodan syndrome, 618512
KMT5B	170.8	100.0%	99.9%	Mental retardation, autosomal dominant 51, 617788
KNL1	103.6	99.0%	97.3%	Microcephaly 4, primary, autosomal recessive, 604321
KPTN	163.9	100.0%	100.0%	Mental retardation, autosomal recessive 41, 615637
KRAS	64.0	99.8%	96.8%	Leukemia, acute myeloid, 601626 Oculoectodermal syndrome, somatic, 600268 Breast cancer, somatic, 114480 RAS-associated autoimmune leukoproliferative disorder, 614470 Cardiofaciocutaneous syndrome 2, 615278 Arteriovenous malformation of the brain, somatic, 108010 Bladder cancer, somatic, 109800 Pancreatic carcinoma, somatic, 260350 Lung cancer, somatic, 211980 Gastric cancer, somatic, 137215 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Noonan syndrome 3, 609942
KREMEN1	152.0	99.8%	98.7%	Ectodermal dysplasia 13, hair/tooth type, 617392
KRIT1	92.4	100.0%	98.9%	Cavernous malformations of CNS and retina, 116860 Cerebral cavernous malformations-1, 116860 Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations, 116860
KRT1	104.0	100.0%	99.0%	Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602 Keratosis palmoplantaris striata III, 607654 Palmoplantar keratoderma, epidermolytic, 144200 Palmoplantar keratoderma, nonepidermolytic, 600962 Ichthyosis histrix, Curth-Macklin type, 146590 Epidermolytic hyperkeratosis, 113800
KRT10	137.0	100.0%	99.0%	Epidermolytic hyperkeratosis, 113800 Ichthyosis with confetti, 609165 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602
KRT12	142.5	99.8%	97.9%	Meesmann corneal dystrophy, 122100
KRT13	132.5	100.0%	98.9%	White sponge nevus 2, 615785

KRT14	48.3	91.4%	83.3%	Naegeli-Franceschetti-Jadassohn syndrome, 161000 Epidermolysis bullosa simplex, Dowling-Meara type, 131760 Dermatopathia pigmentosa reticularis, 125595 Epidermolysis bullosa simplex, Koebner type, 131900 Epidermolysis bullosa simplex, recessive 1, 601001 Epidermolysis bullosa simplex, Weber-Cockayne type, 131800
KRT16	38.7	78.4%	57.3%	Palmoplantar keratoderma, nonepidermolytic, focal, 613000 Pachyonychia congenita 1, 167200
KRT17	19.4	49.9%	31.2%	Pachyonychia congenita 2, 167210 Steatocystoma multiplex, 184500
KRT18	38.7	91.9%	74.0%	Cirrhosis, cryptogenic, 215600
KRT2	146.2	100.0%	99.7%	Ichthyosis bullosa of Siemens, 146800
KRT25	131.6	100.0%	100.0%	Woolly hair, autosomal recessive 3, 616760
KRT3	125.0	100.0%	99.9%	Meesmann corneal dystrophy, 122100
KRT4	130.8	100.0%	99.7%	White sponge nevus 1, 193900
KRT5	121.9	100.0%	99.9%	Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 Epidermolysis bullosa simplex-MCR, 609352 Epidermolysis bullosa simplex-MP, 131960 Dowling-Degos disease 1, 179850 Epidermolysis bullosa simplex, Koebner type, 131900 Epidermolysis bullosa simplex, Dowling-Meara type, 131760 Epidermolysis bullosa simplex, recessive 1, 601001
KRT6A	132.6	98.6%	92.0%	Pachyonychia congenita 3, 615726
KRT6B	132.9	99.7%	95.2%	Pachyonychia congenita 4, 615728
KRT6C	115.2	90.0%	81.1%	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735
KRT71	157.5	100.0%	100.0%	?Hypotrichosis 13, 615896
KRT74	154.9	100.0%	99.7%	?Ectodermal dysplasia 7, hair/nail type, 614929 Woolly hair, autosomal dominant, 194300 ?Hypotrichosis 3, 613981
KRT8	40.9	91.3%	73.9%	Cirrhosis, cryptogenic, 215600
KRT81	91.4	100.0%	99.3%	Monilethrix, 158000
KRT83	74.3	99.3%	93.0%	Erythrokeratodermia variabilis et progressiva 5, 617756 Monilethrix, 158000
KRT85	112.7	99.1%	96.1%	Ectodermal dysplasia 4, hair/nail type, 602032
KRT86	95.7	100.0%	99.4%	Monilethrix, 158000
KRT9	71.8	99.7%	97.3%	Palmoplantar keratoderma, epidermolytic, 144200
KY	118.9	100.0%	99.9%	Myopathy, myofibrillar, 7, 617114

KYNU	104.5	99.2%	94.2%	Vertebral, cardiac, renal, and limb defects syndrome 2, 617661 ?Hydroxykynureninuria, 236800
L1CAM	138.3	99.9%	98.6%	MASA syndrome, 303350 Hydrocephalus with Hirschsprung disease, 307000 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 Corpus callosum, partial agenesis of, 304100 CRASH syndrome, 303350 Hydrocephalus due to aqueductal stenosis, 307000
L2HGDH	123.6	99.2%	97.2%	L-2-hydroxyglutaric aciduria, 236792
LAGE3	73.3	99.2%	94.3%	Galloway-Mowat syndrome 2, X-linked, 301006
LAMA1	119.9	100.0%	99.6%	Poretti-Boltshauser syndrome, 615960
LAMA2	131.6	100.0%	99.4%	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855
LAMA3	129.8	100.0%	99.8%	Epidermolysis bullosa, junctional, Herlitz type, 226700 Laryngoonychocutaneous syndrome, 245660 Epidermolysis bullosa, generalized atrophic benign, 226650
LAMA4	118.6	100.0%	99.8%	Cardiomyopathy, dilated, 1JJ, 615235
LAMB1	147.7	100.0%	99.7%	Lissencephaly 5, 615191
LAMB2	182.1	100.0%	99.7%	Pierson syndrome, 609049 Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199
LAMB3	125.6	100.0%	99.5%	Amelogenesis imperfecta, type IA, 104530 Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LAMC2	105.4	99.8%	98.4%	Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa, junctional, Herlitz type, 226700
LAMC3	163.5	99.9%	99.2%	Cortical malformations, occipital, 614115
LAMP2	89.8	97.8%	92.3%	Danon disease, 300257
LAMTOR2	186.6	100.0%	100.0%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LARGE1	122.8	100.0%	99.8%	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154
LARP7	74.1	89.4%	74.5%	Alazami syndrome, 615071
LARS	128.4	99.8%	98.0%	?Infantile liver failure syndrome 1, 615438
LARS2	128.3	100.0%	100.0%	Perrault syndrome 4, 615300 ?Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LAS1L	83.2	99.5%	96.4%	Wilson-Turner syndrome, 309585
LAT	127.2	100.0%	99.8%	Immunodeficiency 52, 617514

LBR	104.4	97.4%	90.4%	Pelger-Huet anomaly, 169400 Greenberg skeletal dysplasia, 215140 ?Reynolds syndrome, 613471 Pelger-Huet anomaly with mild skeletal anomalies, 618019
LCA5	137.1	99.9%	98.9%	Leber congenital amaurosis 5, 604537
LCAT	156.5	99.6%	96.1%	Norum disease, 245900 Fish-eye disease, 136120
LCK	163.1	99.7%	98.3%	?Immunodeficiency 22, 615758
LCT	128.1	99.9%	98.2%	Lactase deficiency, congenital, 223000
LDB3	161.1	96.3%	95.0%	Cardiomyopathy, hypertrophic, 24, 601493 Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 Myopathy, myofibrillar, 4, 609452 Left ventricular noncompaction 3, 601493
LDHA	57.3	97.7%	89.0%	Glycogen storage disease XI, 612933
LDHD	158.1	100.0%	100.0%	D-lactic aciduria, 245450
LDLR	160.3	100.0%	99.3%	Hypercholesterolemia, familial, 1, 143890 LDL cholesterol level QTL2, 143890
LDLRAP1	162.0	100.0%	99.9%	Hypercholesterolemia, familial, 4, 603813
LEF1	108.5	100.0%	99.7%	Sebaceous tumors, somatic, 0
LEMD2	117.7	100.0%	99.5%	Cataract 46, juvenile-onset, 212500
LEMD3	127.5	99.8%	97.8%	Osteopoikilosis with or without melorheostosis, 166700 Buschke-Ollendorff syndrome, 166700
LEP	184.9	100.0%	100.0%	Obesity, morbid, due to leptin deficiency, 614962
LEPR	104.4	94.3%	92.0%	Obesity, morbid, due to leptin receptor deficiency, 614963
LFNG	132.2	95.1%	89.4%	Spondylocostal dysostosis 3, autosomal recessive, 609813
LGI1	132.9	98.5%	97.4%	Epilepsy, familial temporal lobe, 1, 600512
LGI4	114.5	99.5%	97.2%	Arthrogryposis multiplex congenita, neurogenic, with myelin defect, 617468
LHB	25.3	95.0%	61.7%	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300
LHCGR	141.5	98.5%	94.3%	Leydig cell adenoma, somatic, with precocious puberty, 176410 Precocious puberty, male, 176410 Luteinizing hormone resistance, female, 238320 Leydig cell hypoplasia with pseudohermaphroditism, 238320 Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320
LHFPL5	228.8	100.0%	100.0%	Deafness, autosomal recessive 67, 610265
LHX3	135.1	96.6%	96.6%	Pituitary hormone deficiency, combined, 3, 221750
LHX4	141.9	100.0%	100.0%	Pituitary hormone deficiency, combined, 4, 262700
LIAS	124.4	100.0%	98.7%	Hyperglycinemia, lactic acidosis, and seizures, 614462

LIFR	108.8	99.6%	96.7%	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LIG4	170.5	100.0%	99.9%	LIG4 syndrome, 606593
LIM2	122.5	100.0%	99.9%	Cataract 19, multiple types, 615277
LIMS2	131.0	96.5%	93.4%	?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827
LINGO1	231.7	100.0%	100.0%	Mental retardation, autosomal recessive 64, 618103
LINS1	135.6	100.0%	99.5%	Mental retardation, autosomal recessive 27, 614340
LIPA	107.3	96.6%	94.9%	Wolman disease, 278000 Cholesteryl ester storage disease, 278000
LIPC	104.4	100.0%	99.8%	Hepatic lipase deficiency, 614025
LIPE	136.4	100.0%	99.7%	Lipodystrophy, familial partial, type 6, 615980
LIPH	120.7	100.0%	99.7%	Hypotrichosis 7, 604379 Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379
LIPN	111.3	100.0%	98.8%	Ichthyosis, congenital, autosomal recessive 8, 613943
LIPT1	199.3	100.0%	99.8%	Lipoyltransferase 1 deficiency, 616299
LIPT2	107.4	100.0%	99.9%	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668
LITAF	109.3	95.2%	91.2%	Charcot-Marie-Tooth disease, type 1C, 601098
LMAN1	147.0	99.9%	98.5%	Combined factor V and VIII deficiency, 227300
LMAN2L	113.2	100.0%	99.4%	?Mental retardation, autosomal recessive, 52, 616887
LMBR1	121.1	98.5%	95.0%	Triphalangeal thumb-polysyndactyly syndrome, 174500 Syndactyly, type IV, 186200 Triphalangeal thumb, type I, 174500 Acheiropody, 200500 Laurin-Sandrow syndrome, 135750 Hypoplastic or aplastic tibia with polydactyly, 188740 Polydactyly, preaxial type II, 174500
LMBRD1	95.5	97.1%	91.2%	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LMF1	150.5	100.0%	99.9%	Lipase deficiency, combined, 246650
LMNA	118.2	98.3%	93.2%	Muscular dystrophy, congenital, 613205 Lipodystrophy, familial partial, type 2, 151660 Charcot-Marie-Tooth disease, type 2B1, 605588 Cardiomyopathy, dilated, 1A, 115200 Heart-hand syndrome, Slovenian type, 610140 Hutchinson-Gilford progeria, 176670 Restrictive dermopathy, lethal, 275210 Mandibuloacral dysplasia, 248370 Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 Malouf syndrome, 212112

LMNB1	122.6	100.0%	100.0%	Leukodystrophy, adult-onset, autosomal dominant, 169500
LMNB2	157.9	99.6%	98.3%	?Epilepsy, progressive myoclonic, 9, 616540
LMOD3	134.2	100.0%	99.9%	Nemaline myopathy 10, 616165
LMX1B	163.9	100.0%	99.3%	Nail-patella syndrome, 161200
LNPK	86.8	97.7%	90.4%	Neurodevelopmental disorder with epilepsy and hypoplasia of the corpus callosum, 618090
LONP1	164.8	100.0%	100.0%	CODAS syndrome, 600373
LOR	46.5	100.0%	97.7%	Vohwinkel syndrome with ichthyosis, 604117
LOX	176.0	100.0%	99.1%	Aortic aneurysm, familial thoracic 10, 617168
LOXHD1	120.2	100.0%	99.6%	Deafness, autosomal recessive 77, 613079
LPAR6	101.0	99.8%	97.7%	Hypotrichosis 8, 278150 Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150
LPIN1	128.5	99.5%	97.0%	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	101.1	100.0%	99.7%	Majeed syndrome, 609628
LPL	133.6	100.0%	100.0%	Lipoprotein lipase deficiency, 238600 Combined hyperlipidemia, familial, 144250
LPP	108.4	100.0%	100.0%	Leukemia, acute myeloid, 601626 Lipoma, 0
LRAT	252.8	100.0%	100.0%	Retinal dystrophy, early-onset severe, 613341 Leber congenital amaurosis 14, 613341 Retinitis pigmentosa, juvenile, 613341
LRBA	127.6	100.0%	99.5%	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRIG2	131.7	99.9%	99.0%	Urofacial syndrome 2, 615112
LRIT3	111.2	94.4%	93.5%	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRMDA	119.8	99.4%	98.0%	Albinism, oculocutaneous, type VII, 615179
LRP1	188.4	99.8%	99.3%	?Keratosis pilaris atrophicans, 604093
LRP12	168.6	100.0%	99.9%	Oculopharyngodistal myopathy 1, 164310
LRP2	140.5	100.0%	99.9%	Donnai-Barrow syndrome, 222448
LRP4	136.5	99.8%	99.1%	?Myasthenic syndrome, congenital, 17, 616304 Sclerosteosis 2, 614305 Cenani-Lenz syndactyly syndrome, 212780
LRP5	183.1	99.9%	99.4%	van Buchem disease, type 2, 607636 Exudative vitreoretinopathy 4, 601813 Hyperostosis, endosteal, 144750 Osteosclerosis, 144750 Polycystic liver disease 4 with or without kidney cysts, 617875 Osteoporosis-pseudoglioma syndrome, 259770 Osteopetrosis, autosomal dominant 1, 607634

LRP6	137.9	99.9%	99.3%	Tooth agenesis, selective, 7, 616724
LRPAP1	153.8	100.0%	99.6%	Myopia 23, autosomal recessive, 615431
LRPPRC	126.3	100.0%	99.7%	Leigh syndrome, French-Canadian type, 220111
LRRC56	148.8	100.0%	99.8%	Ciliary dyskinesia, primary, 39, 618254
LRRC6	138.2	99.5%	96.2%	Ciliary dyskinesia, primary, 19, 614935
LRRC8A	241.2	100.0%	100.0%	?Agammaglobulinemia 5, 613506
LRSAM1	145.8	100.0%	99.9%	Charcot-Marie-Tooth disease, axonal, type 2P, 614436
LRTOMT	123.2	99.8%	96.2%	Deafness, autosomal recessive 63, 611451
LSS	138.5	100.0%	99.9%	Cataract 44, 616509 Hypotrichosis 14, 618275
LTBP2	124.2	100.0%	99.7%	Glaucoma 3, primary congenital, D, 613086 ?Weill-Marchesani syndrome 3, recessive, 614819 Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750
LTBP3	166.1	100.0%	100.0%	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
LTBP4	167.8	100.0%	99.7%	Cutis laxa, autosomal recessive, type IC, 613177
LYRM4	78.4	68.0%	63.4%	?Combined oxidative phosphorylation deficiency 19, 615595
LYRM7	58.4	98.2%	87.5%	Mitochondrial complex III deficiency, nuclear type 8, 615838
LYST	135.6	99.3%	97.1%	Chediak-Higashi syndrome, 214500
LYZ	142.4	100.0%	100.0%	Amyloidosis, renal, 105200
LZTFL1	116.5	99.9%	99.2%	Bardet-Biedl syndrome 17, 615994
LZTR1	157.2	100.0%	99.9%	Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
LZTS1	158.2	100.0%	100.0%	Esophageal squamous cell carcinoma, somatic, 133239
MAB21L1	187.7	100.0%	100.0%	Cerebellar, ocular, craniofacial, and genital syndrome, 618479
MAB21L2	265.2	100.0%	100.0%	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877
MACF1	133.2	100.0%	99.5%	Lissencephaly 9 with complex brainstem malformation, 618325
MAD1L1	116.0	99.9%	98.3%	Prostate cancer, somatic, 176807 Lymphoma, somatic, 0
MAD2L2	150.7	100.0%	99.9%	?Fanconi anemia, complementation group V, 617243
MAF	103.8	89.3%	84.7%	Ayme-Gripp syndrome, 601088 Cataract 21, multiple types, 610202
MAFA	71.4	99.9%	97.8%	Insulinomatosis and diabetes mellitus, 147630
MAFB	140.5	100.0%	100.0%	Duane retraction syndrome 3, 617041 Multicentric carpotarsal osteolysis syndrome, 166300
MAG	175.3	100.0%	99.9%	Spastic paraplegia 75, autosomal recessive, 616680
MAGED2	95.1	99.8%	98.5%	Bartter syndrome, type 5, antenatal, transient, 300971

MAGEL2	137.7	98.8%	94.4%	Schaaf-Yang syndrome, 615547
MAGI2	93.7	95.1%	92.1%	Nephrotic syndrome, type 15, 617609
MAGT1	95.1	98.5%	95.0%	Congenital disorder of glycosylation, type Icc, 301031 Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853
MAK	134.1	99.2%	96.5%	Retinitis pigmentosa 62, 614181
MALT1	128.7	93.6%	89.3%	Immunodeficiency 12, 615468
MAML2	118.9	100.0%	100.0%	Mucoepidermoid salivary gland carcinoma, 0
MAMLD1	134.7	99.8%	98.4%	Hypospadias 2, X-linked, 300758
MAN1B1	137.5	100.0%	99.9%	Mental retardation, autosomal recessive 15, 614202
MAN2B1	139.1	99.9%	99.1%	Mannosidosis, alpha-, types I and II, 248500
MANBA	117.1	99.7%	98.1%	Mannosidosis, beta, 248510
MAOA	100.1	100.0%	99.2%	Brunner syndrome, 300615
MAP2K1	96.7	99.6%	97.1%	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	139.8	99.3%	95.6%	Cardiofaciocutaneous syndrome 4, 615280
MAP3K1	147.4	99.8%	98.2%	46XY sex reversal 6, 613762
MAP3K20	110.7	99.9%	98.8%	Split-foot malformation with mesoaxial polydactyly, 616890 Centronuclear myopathy 6 with fiber-type disproportion, 617760
MAP3K7	117.5	100.0%	99.7%	Cardiospondylocarpofacial syndrome, 157800 Frontometaphyseal dysplasia 2, 617137
MAP3K8	136.3	100.0%	99.9%	Lung cancer, somatic, 211980
MAPK8IP3	181.0	100.0%	100.0%	Neurodevelopmental disorder with or without variable brain abnormalities, 618443
MAPKAPK3	94.7	100.0%	99.6%	?Macular dystrophy, patterned, 3, 617111
MAPKBP1	144.1	100.0%	100.0%	Nephronophthisis 20, 617271
MAPRE2	163.4	99.8%	98.4%	Symmetric circumferential skin creases, congenital, 2, 616734
MAPT	165.9	100.0%	99.6%	Pick disease, 172700 Dementia, frontotemporal, with or without parkinsonism, 600274 Supranuclear palsy, progressive, 601104 Supranuclear palsy, progressive atypical, 260540
MARCH6	108.6	99.8%	97.7%	Epilepsy, familial adult myoclonic, 3, 613608
MARK3	126.9	99.9%	97.8%	?Visual impairment and progressive phthisis bulbi, 618283
MARS	106.2	99.9%	98.8%	Interstitial lung and liver disease, 615486 Charcot-Marie-Tooth disease, axonal, type 2U, 616280
MARS2	195.2	100.0%	100.0%	Spastic ataxia 3, autosomal recessive, 611390 ?Combined oxidative phosphorylation deficiency 25, 616430
MARVELD2	143.1	98.7%	95.2%	Deafness, autosomal recessive 49, 610153
MASP1	140.7	100.0%	99.5%	3MC syndrome 1, 257920
MASP2	129.9	100.0%	99.4%	MASP2 deficiency, 613791

MAST1	187.1	100.0%	99.9%	Mega-corpus-callosum syndrome with cerebellar hypoplasia and cortical malformations, 618273
MAT1A	154.2	99.7%	98.2%	Methionine adenosyltransferase deficiency, autosomal recessive, 250850 Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850
MATN3	103.0	87.1%	84.5%	?Spondyloepimetaphyseal dysplasia, 608728 Epiphyseal dysplasia, multiple, 5, 607078
MATR3	87.4	96.9%	92.7%	Amyotrophic lateral sclerosis 21, 606070
MBD5	151.8	99.9%	99.8%	Mental retardation, autosomal dominant 1, 156200
MBOAT7	121.9	100.0%	99.9%	Mental retardation, autosomal recessive 57, 617188
MBTPS1	116.8	99.8%	98.2%	?Spondyloepiphyseal dysplasia, Kondo-Fu type, 618392
MBTPS2	109.0	99.9%	98.4%	Osteogenesis imperfecta, type XIX, 301014 ?Olmsted syndrome, X-linked, 300918 IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800
MC2R	160.2	100.0%	99.4%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MC4R	193.7	100.0%	100.0%	Obesity (BMIQ20), 618406
MCC	128.4	100.0%	99.5%	Colorectal cancer, somatic, 114500
MCCC1	138.0	100.0%	99.6%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	121.8	100.0%	99.9%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	124.7	100.0%	100.0%	Methylmalonyl-CoA epimerase deficiency, 251120
MCFD2	97.5	97.7%	91.3%	Factor V and factor VIII, combined deficiency of, 613625
MCM2	164.5	100.0%	100.0%	?Deafness, autosomal dominant 70, 616968
MCM3AP	136.4	100.0%	99.4%	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124
MCM4	141.3	100.0%	99.5%	Immunodeficiency 54, 609981
MCM5	131.9	100.0%	100.0%	?Meier-Gorlin syndrome 8, 617564
MCM6	128.8	100.0%	100.0%	Lactase persistence/nonpersistence, 223100
MCM8	121.8	99.9%	98.9%	?Premature ovarian failure 10, 612885
MCM9	131.6	100.0%	99.5%	Ovarian dysgenesis 4, 616185
MCOLN1	170.5	100.0%	99.4%	Mucolipidosis IV, 252650
MCPH1	138.1	100.0%	98.7%	Microcephaly 1, primary, autosomal recessive, 251200
MDH2	116.5	98.0%	98.0%	Epileptic encephalopathy, early infantile, 51, 617339
MECOM	133.4	100.0%	99.8%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MECP2	135.2	100.0%	99.5%	Mental retardation, X-linked syndromic, Lubs type, 300260 Encephalopathy, neonatal severe, 300673 Mental retardation, X-linked, syndromic 13, 300055 Rett syndrome, atypical, 312750

				Rett syndrome, 312750 Rett syndrome, preserved speech variant, 312750
MECR	114.2	100.0%	99.7%	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MED12	89.4	99.6%	96.5%	Ohdo syndrome, X-linked, 300895 Lujan-Fryns syndrome, 309520 Opitz-Kaveggia syndrome, 305450
MED13L	112.2	100.0%	99.8%	Transposition of the great arteries, dextro-looped 1, 608808 Mental retardation and distinctive facial features with or without cardiac defects, 616789
MED17	134.7	97.8%	94.7%	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	131.6	99.7%	98.5%	Mental retardation, autosomal recessive 18, 614249
MED25	148.0	100.0%	99.9%	?Charcot-Marie-Tooth disease, type 2B2, 605589 Basel-Vanagait-Smirin-Yosef syndrome, 616449
MEF2C	131.2	99.5%	95.7%	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
MEFV	136.9	99.0%	97.0%	Familial Mediterranean fever, AR, 249100 Familial Mediterranean fever, AD, 134610
MEGF10	129.9	100.0%	99.9%	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399
MEGF8	158.7	100.0%	99.8%	Carpenter syndrome 2, 614976
MEI1	101.9	100.0%	99.5%	Hydatidiform mole, recurrent, 3, 618431
MEIOB	101.6	99.9%	97.9%	?Spermatogenic failure 22, 617706
MEIS2	128.4	100.0%	99.9%	Cleft palate, cardiac defects, and mental retardation, 600987
MEN1	145.4	100.0%	99.8%	Multiple endocrine neoplasia 1, 131100 Angiofibroma, somatic, 0 Adrenal adenoma, somatic, 0 Parathyroid adenoma, somatic, 0 Lipoma, somatic, 0 Carcinoid tumor of lung, 0
MEOX1	114.4	100.0%	98.8%	Klippel-Feil syndrome 2, 214300
MERTK	133.5	99.5%	99.0%	Retinitis pigmentosa 38, 613862
MESD	122.0	100.0%	100.0%	Osteogenesis imperfecta, type XX, 618644
MESP2	148.8	97.4%	96.2%	Spondylocostal dysostosis 2, autosomal recessive, 608681
MET	153.7	100.0%	99.6%	Hepatocellular carcinoma, childhood type, somatic, 114550 ?Deafness, autosomal recessive 97, 616705 Renal cell carcinoma, papillary, 1, familial and somatic, 605074
METTL13	125.5	100.0%	99.4%	No OMIM Disease ID
METTL23	117.0	100.0%	100.0%	Mental retardation, autosomal recessive 44, 615942
MFAP5	109.5	99.8%	95.3%	Aortic aneurysm, familial thoracic 9, 616166

MFF	86.8	93.5%	88.5%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFN2	130.9	100.0%	99.9%	Hereditary motor and sensory neuropathy VIA, 601152 Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260
MFRP	140.5	100.0%	100.0%	Nanophthalmos 2, 609549 Microphthalmia, isolated 5, 611040
MFSD2A	121.3	100.0%	99.6%	Microcephaly 15, primary, autosomal recessive, 616486
MFSD8	117.4	100.0%	99.6%	Macular dystrophy with central cone involvement, 616170 Ceroid lipofuscinosis, neuronal, 7, 610951
MGAT2	155.6	100.0%	100.0%	Congenital disorder of glycosylation, type IIa, 212066
MGME1	145.7	100.0%	99.9%	Mitochondrial DNA depletion syndrome 11, 615084
MGP	134.2	98.6%	93.2%	Keutel syndrome, 245150
MIB1	129.1	100.0%	100.0%	Left ventricular noncompaction 7, 615092
MICU1	103.4	98.7%	96.2%	Myopathy with extrapyramidal signs, 615673
MID1	131.5	99.9%	98.4%	Opitz GBBB syndrome, type I, 300000
MID2	111.8	99.7%	98.0%	?Mental retardation, X-linked 101, 300928
MIP	126.1	99.9%	97.4%	Cataract 15, multiple types, 615274
MIPEP	100.6	99.8%	96.1%	Combined oxidative phosphorylation deficiency 31, 617228
MIR140	NC	NC	NC	Spondyloepiphyseal dysplasia, Nishimura type, 618618
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	145.6	100.0%	100.0%	COMMAD syndrome, 617306 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 Tietz albinism-deafness syndrome, 103500
MKKS	161.5	83.2%	83.2%	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKL1	131.1	98.0%	96.3%	Megakaryoblastic leukemia, acute, 0
MKRN3	149.9	100.0%	100.0%	Precocious puberty, central, 2, 615346
MKS1	98.8	99.9%	98.5%	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000
MLC1	102.4	100.0%	99.9%	Megalencephalic leukoencephalopathy with subcortical cysts, 604004

MLH1	142.9	100.0%	99.6%	Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320 Colorectal cancer, hereditary nonpolyposis, type 2, 609310
MLH3	142.7	100.0%	100.0%	Colorectal cancer, somatic, 114500 Colorectal cancer, hereditary nonpolyposis, type 7, 614385
MLLT10	125.2	96.2%	94.5%	Leukemia, acute myeloid, 601626
MLPH	104.1	99.8%	97.9%	Griscelli syndrome, type 3, 609227
MLYCD	105.5	99.7%	97.3%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	168.7	100.0%	100.0%	Methylmalonic aciduria, vitamin B12-responsive, 251100
MMAB	101.3	100.0%	100.0%	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110
MMACHC	214.4	100.0%	100.0%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	76.8	93.0%	77.2%	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410
MME	111.2	99.8%	97.8%	Charcot-Marie-Tooth disease, axonal, type 2T, 617017 ?Spinocerebellar ataxia 43, 617018
MMP1	136.7	100.0%	99.0%	COPD, rate of decline of lung function in, 606963
MMP13	113.0	92.9%	92.3%	Metaphyseal anadysplasia 1, 602111 Metaphyseal dysplasia, Spahr type, 250400 Spondyloepimetaphyseal dysplasia, Missouri type, 602111
MMP14	158.2	100.0%	99.9%	?Winchester syndrome, 277950
MMP19	126.8	100.0%	99.0%	Cavitary optic disc anomalies, 611543
MMP2	162.9	100.0%	100.0%	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP20	91.0	99.9%	97.9%	Amelogenesis imperfecta, type IIA2, 612529
MMP21	103.7	100.0%	99.5%	Heterotaxy, visceral, 7, autosomal, 616749
MMP9	161.0	100.0%	99.1%	Metaphyseal anadysplasia 2, 613073
MN1	165.3	100.0%	100.0%	Meningioma, 607174
MNX1	65.4	81.8%	74.0%	Currarino syndrome, 176450
MOCOS	156.5	100.0%	99.2%	Xanthinuria, type II, 603592
MOCS1	101.3	99.3%	96.6%	Molybdenum cofactor deficiency A, 252150
MOCS2	134.2	99.6%	99.6%	Molybdenum cofactor deficiency B, 252160
MOG	101.8	100.0%	99.2%	?Narcolepsy 7, 614250
MOGS	157.9	100.0%	100.0%	Congenital disorder of glycosylation, type IIb, 606056
MORC2	132.4	100.0%	99.7%	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688
MPC1	161.4	100.0%	99.6%	Mitochondrial pyruvate carrier deficiency, 614741
MPDU1	110.1	100.0%	99.8%	Congenital disorder of glycosylation, type If, 609180

MPDZ	127.3	99.6%	98.1%	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219
MPI	115.8	100.0%	99.9%	Congenital disorder of glycosylation, type Ib, 602579
MPIG6B	130.3	100.0%	100.0%	?Thrombocytopenia, anemia, and myelofibrosis, 617441
MPL	134.4	100.0%	99.9%	Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498 Myelofibrosis with myeloid metaplasia, somatic, 254450
MPLKIP	106.6	100.0%	99.9%	Trichothiodystrophy 4, nonphotosensitive, 234050
MPO	169.8	100.0%	99.7%	Myeloperoxidase deficiency, 254600
MPV17	93.2	100.0%	98.5%	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MPZ	135.9	100.0%	99.0%	Charcot-Marie-Tooth disease, type 2J, 607736 Charcot-Marie-Tooth disease, type 1B, 118200 Dejerine-Sottas disease, 145900 Hypomyelinating neuropathy, congenital, 2, 618184 Charcot-Marie-Tooth disease, dominant intermediate D, 607791 Roussy-Levy syndrome, 180800 Charcot-Marie-Tooth disease, type 2I, 607677
MPZL2	92.7	100.0%	99.8%	Deafness, autosomal recessive 111, 618145
MRAP	173.5	100.0%	100.0%	Glucocorticoid deficiency 2, 607398
MRAS	109.3	99.7%	97.6%	Noonan syndrome 11, 618499
MRE11	48.1	97.7%	83.5%	Ataxia-telangiectasia-like disorder 1, 604391
MRM2	120.4	100.0%	99.4%	?Mitochondrial DNA depletion syndrome 17, 618567
MRPL3	66.2	93.4%	81.6%	Combined oxidative phosphorylation deficiency 9, 614582
MRPL44	130.4	100.0%	99.9%	?Combined oxidative phosphorylation deficiency 16, 615395
MRPS14	172.5	100.0%	100.0%	?Combined oxidative phosphorylation deficiency 38, 618378
MRPS16	136.8	100.0%	99.1%	Combined oxidative phosphorylation deficiency 2, 610498
MRPS2	175.4	100.0%	100.0%	Combined oxidative phosphorylation deficiency 36, 617950
MRPS22	138.5	100.0%	97.3%	Combined oxidative phosphorylation deficiency 5, 611719 Ovarian dysgenesis 7, 618117
MRPS34	184.8	100.0%	100.0%	Combined oxidative phosphorylation deficiency 32, 617664
MRPS7	161.3	100.0%	100.0%	?Combined oxidative phosphorylation deficiency 34, 617872
MS4A1	128.7	100.0%	99.1%	Immunodeficiency, common variable, 5, 613495
MSH2	110.0	99.5%	95.6%	Muir-Torre syndrome, 158320 Mismatch repair cancer syndrome, 276300 Colorectal cancer, hereditary nonpolyposis, type 1, 120435
MSH3	140.2	100.0%	99.1%	Familial adenomatous polyposis 4, 617100 Endometrial carcinoma, somatic, 608089

MSH5	108.4	100.0%	99.9%	?Premature ovarian failure 13, 617442
MSH6	171.5	100.0%	99.9%	Mismatch repair cancer syndrome, 276300 Colorectal cancer, hereditary nonpolyposis, type 5, 614350
MSL3	77.3	95.2%	84.1%	Mental retardation, X-linked, syndromic, 36, 301032
MSMO1	47.0	96.2%	87.7%	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MSN	69.6	98.2%	92.3%	Immunodeficiency 50, 300988
MSR1	143.5	99.9%	99.4%	Barrett esophagus/esophageal adenocarcinoma, 614266
MSRB3	138.7	99.9%	99.3%	Deafness, autosomal recessive 74, 613718
MSTN	156.1	100.0%	100.0%	Muscle hypertrophy, 614160
MSTO1	107.9	99.5%	97.1%	Myopathy, mitochondrial, and ataxia, 617675
MSX1	164.1	100.0%	99.4%	Orofacial cleft 5, 608874 Ectodermal dysplasia 3, Witkop type, 189500 Tooth agenesis, selective, 1, with or without orofacial cleft, 106600
MSX2	111.9	100.0%	100.0%	Parietal foramina 1, 168500 Craniosynostosis 2, 604757 Parietal foramina with cleidocranial dysplasia, 168550
MTAP	100.1	99.2%	94.8%	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250
MTFMT	131.5	100.0%	99.7%	Combined oxidative phosphorylation deficiency 15, 614947 Mitochondrial complex I deficiency, nuclear type 27, 618248
MTHFD1	119.4	99.9%	98.3%	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780
MTHFR	124.2	98.5%	96.7%	Homocystinuria due to MTHFR deficiency, 236250
MTHFS	94.4	76.7%	75.0%	Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367
MTM1	78.4	98.4%	91.8%	Myotubular myopathy, X-linked, 310400
MTMR2	99.0	99.9%	98.2%	Charcot-Marie-Tooth disease, type 4B1, 601382
MTO1	149.2	91.9%	89.5%	Combined oxidative phosphorylation deficiency 10, 614702
MTOR	116.6	100.0%	99.2%	Smith-Kingsmore syndrome, 616638 Focal cortical dysplasia, type II, somatic, 607341
MTPAP	124.6	99.3%	93.9%	?Spastic ataxia 4, autosomal recessive, 613672
MTR	134.7	100.0%	99.6%	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940
MTRR	135.6	100.0%	99.2%	Homocystinuria-megaloblastic anemia, cbl E type, 236270
MTTP	114.7	100.0%	99.5%	Abetalipoproteinemia, 200100
MUC1	77.4	94.3%	87.1%	Medullary cystic kidney disease 1, 174000
MUSK	135.2	100.0%	100.0%	Fetal akinesia deformation sequence 1, 208150 Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325
MUT	128.8	100.0%	99.0%	Methylmalonic aciduria, mut(0) type, 251000

MUTYH	165.8	100.0%	100.0%	Gastric cancer, somatic, 613659 Adenomas, multiple colorectal, 608456 Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600
MVD	126.6	99.9%	99.0%	Porokeratosis 7, multiple types, 614714
MVK	130.3	90.5%	90.4%	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377
MXI1	108.4	98.7%	93.9%	Prostate cancer, somatic, 176807 Neurofibrosarcoma, somatic, 0
MYBPC1	129.2	100.0%	99.4%	Arthrogryposis, distal, type 1B, 614335 Myopathy, congenital, with tremor, 618524 Lethal congenital contracture syndrome 4, 614915
MYBPC3	154.3	100.0%	98.9%	Cardiomyopathy, hypertrophic, 4, 115197 Cardiomyopathy, dilated, 1MM, 615396 Left ventricular noncompaction 10, 615396
MYC	145.3	66.5%	64.5%	Burkitt lymphoma, somatic, 113970
MYCN	200.6	100.0%	100.0%	Feingold syndrome 1, 164280
MYD88	219.1	100.0%	99.9%	Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260 Macroglobulinemia, Waldenstrom, somatic, 153600
MYF5	184.3	100.0%	100.0%	Ophthalmoplegia, external, with rib and vertebral anomalies, 618155
MYH11	131.1	100.0%	99.7%	Aortic aneurysm, familial thoracic 4, 132900
MYH14	120.4	99.4%	96.3%	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369 Deafness, autosomal dominant 4A, 600652
MYH2	113.6	100.0%	99.7%	Proximal myopathy and ophthalmoplegia, 605637
MYH3	100.2	100.0%	99.1%	Contractures, pterygia, and variable skeletal fusions syndrome 1B, 618469 Contractures, pterygia, and variable skeletal fusions syndrome 1A, 178110 Arthrogryposis, distal, type 2B3 (Sheldon-Hall), 618436 Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700
MYH6	104.2	99.3%	96.3%	Atrial septal defect 3, 614089 Cardiomyopathy, hypertrophic, 14, 613251 Cardiomyopathy, dilated, 1EE, 613252
MYH7	101.7	99.7%	97.2%	Myopathy, myosin storage, autosomal recessive, 255160 Left ventricular noncompaction 5, 613426 Laing distal myopathy, 160500 Myopathy, myosin storage, autosomal dominant, 608358 Cardiomyopathy, dilated, 1S, 613426 Scapuloperoneal syndrome, myopathic type, 181430 Cardiomyopathy, hypertrophic, 1, 192600

MYH8	119.4	100.0%	99.6%	Carney complex variant, 608837 Trismus-pseudocamptodactyly syndrome, 158300
MYH9	140.9	99.7%	99.0%	Deafness, autosomal dominant 17, 603622 Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100
MYL1	94.4	99.9%	99.5%	Myopathy, congenital, with fast-twitch (type II) fiber atrophy, 618414
MYL2	139.8	99.9%	97.0%	Cardiomyopathy, hypertrophic, 10, 608758
MYL3	106.8	100.0%	100.0%	Cardiomyopathy, hypertrophic, 8, 608751
MYL4	139.8	100.0%	100.0%	?Atrial fibrillation, familial, 18, 617280
MYLK	132.8	100.0%	99.6%	Aortic aneurysm, familial thoracic 7, 613780 Megacystis-microcolon-intestinal hypoperistalsis syndrome, 249210
MYLK2	141.9	100.0%	100.0%	Cardiomyopathy, hypertrophic, 1, digenic, 192600
MYMK	156.4	100.0%	100.0%	Carey-Fineman-Ziter syndrome, 254940
MYO15A	159.8	100.0%	99.6%	Deafness, autosomal recessive 3, 600316
MYO18B	132.4	100.0%	99.4%	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549
MYO1E	119.1	99.9%	98.5%	Glomerulosclerosis, focal segmental, 6, 614131
MYO3A	112.5	99.4%	95.4%	Deafness, autosomal recessive 30, 607101
MYO5A	109.9	99.7%	98.5%	Griscelli syndrome, type 1, 214450
MYO5B	115.0	98.1%	94.9%	Microvillus inclusion disease, 251850
MYO6	98.8	99.5%	95.6%	Deafness, autosomal recessive 37, 607821 Deafness, autosomal dominant 22, 606346 Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346
MYO7A	134.7	99.9%	99.1%	Deafness, autosomal recessive 2, 600060 Deafness, autosomal dominant 11, 601317 Usher syndrome, type 1B, 276900
MYO9A	140.5	100.0%	99.0%	Myasthenic syndrome, congenital, 24, presynaptic, 618198
MYOC	166.0	100.0%	99.1%	Glaucoma 1A, primary open angle, 137750
MYOT	142.2	100.0%	99.1%	Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
MYOZ2	142.3	100.0%	100.0%	Cardiomyopathy, hypertrophic, 16, 613838
MYPN	128.9	99.9%	99.0%	Cardiomyopathy, dilated, 1KK, 615248 Cardiomyopathy, familial restrictive, 4, 615248 Nemaline myopathy 11, autosomal recessive, 617336 Cardiomyopathy, hypertrophic, 22, 615248
MYRF	157.3	97.8%	96.6%	Cardiac-urogenital syndrome, 618280 Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113
MYSM1	110.1	99.8%	98.6%	Bone marrow failure syndrome 4, 618116

MYT1L	154.1	100.0%	99.9%	Mental retardation, autosomal dominant 39, 616521
NAA10	112.8	100.0%	99.4%	Ogden syndrome, 300855 ?Microphthalmia, syndromic 1, 309800
NAA15	92.0	97.6%	94.4%	Mental retardation, autosomal dominant 50, 617787
NACC1	188.0	100.0%	100.0%	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393
NADK2	163.3	100.0%	99.8%	?2,4-dienoyl-CoA reductase deficiency, 616034
NAGA	131.4	100.0%	100.0%	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241
NAGLU	130.5	98.5%	95.6%	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491
NAGS	118.8	100.0%	100.0%	N-acetylglutamate synthase deficiency, 237310
NALCN	117.7	99.7%	98.5%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419 Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266
NANOS1	131.0	100.0%	99.9%	Spermatogenic failure 12, 615413
NANS	105.0	100.0%	99.3%	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NARS2	121.5	97.6%	97.4%	?Deafness, autosomal recessive 94, 618434 Combined oxidative phosphorylation deficiency 24, 616239
NAT8L	106.1	99.9%	98.4%	?N-acetylaspartate deficiency, 614063
NAXD	144.4	100.0%	100.0%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321
NAXE	90.7	99.8%	97.2%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NBAS	138.4	99.9%	99.2%	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NBEAL2	182.9	100.0%	99.7%	Gray platelet syndrome, 139090
NBN	90.6	100.0%	98.2%	Nijmegen breakage syndrome, 251260 Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065
NCAPD2	123.8	100.0%	99.5%	?Microcephaly 21, primary, autosomal recessive, 617983
NCAPD3	103.3	99.8%	98.4%	Microcephaly 22, primary, autosomal recessive, 617984
NCAPG2	121.6	99.6%	97.6%	Khan-Khan-Katsanis syndrome, 618460
NCAPH	124.1	100.0%	100.0%	?Microcephaly 23, primary, autosomal recessive, 617985
NCF1	24.7	28.6%	22.6%	Chronic granulomatous disease due to deficiency of NCF-1, 233700
NCF2	112.5	100.0%	98.6%	Chronic granulomatous disease due to deficiency of NCF-2, 233710
NCF4	160.6	100.0%	100.0%	?Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III, 613960
NCSTN	97.9	100.0%	99.7%	Acne inversa, familial, 1, 142690

NDE1	95.8	100.0%	99.8%	Lissencephaly 4 (with microcephaly), 614019 ?Microhydranencephaly, 605013
NDN	138.5	99.7%	97.5%	Prader-Willi syndrome, 176270
NDP	96.2	100.0%	99.6%	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600
NDRG1	122.5	100.0%	100.0%	Charcot-Marie-Tooth disease, type 4D, 601455
NDST1	204.8	100.0%	100.0%	Mental retardation, autosomal recessive 46, 616116
NDUFA1	195.4	99.9%	98.7%	Mitochondrial complex I deficiency, nuclear type 12, 301020
NDUFA10	118.3	99.9%	99.2%	Mitochondrial complex I deficiency, nuclear type 22, 618243
NDUFA11	129.7	100.0%	98.5%	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFA12	162.1	100.0%	99.9%	?Mitochondrial complex I deficiency, nuclear type 23, 618244
NDUFA13	134.6	92.3%	92.0%	?Mitochondrial complex I deficiency, nuclear type 28, 618249
NDUFA2	179.9	100.0%	100.0%	?Mitochondrial complex I deficiency, nuclear type 13, 618235
NDUFA6	215.7	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 33, 618253
NDUFA9	105.5	99.7%	96.1%	Mitochondrial complex I deficiency, nuclear type 26, 618247
NDUFAF1	100.2	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 11, 618234
NDUFAF2	56.3	94.0%	79.5%	Mitochondrial complex I deficiency, nuclear type 10, 618233
NDUFAF3	159.3	100.0%	99.9%	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF4	98.6	99.4%	93.8%	Mitochondrial complex I deficiency, nuclear type 15, 618237
NDUFAF5	128.0	99.8%	99.3%	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFAF6	92.3	100.0%	98.9%	Mitochondrial complex I deficiency, nuclear type 17, 618239
NDUFB11	110.0	99.2%	96.2%	Linear skin defects with multiple congenital anomalies 3, 300952 ?Mitochondrial complex I deficiency, nuclear type 30, 301021
NDUFB3	22.8	89.3%	61.0%	Mitochondrial complex I deficiency, nuclear type 25, 618246
NDUFB8	112.6	100.0%	99.8%	Mitochondrial complex I deficiency, nuclear type 32, 618252
NDUFB9	114.2	98.3%	94.1%	?Mitochondrial complex I deficiency, nuclear type 24, 618245
NDUFS1	140.7	100.0%	99.7%	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	102.9	100.0%	99.9%	Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	132.3	90.7%	90.6%	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	148.6	100.0%	99.6%	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS6	120.0	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFS7	157.0	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFS8	171.3	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 2, 618222
NDUFV1	154.4	100.0%	99.5%	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	71.5	91.8%	77.9%	Mitochondrial complex I deficiency, nuclear type 7, 618229
NEB	101.5	83.0%	82.5%	Nemaline myopathy 2, autosomal recessive, 256030
NECAP1	108.4	100.0%	100.0%	?Epileptic encephalopathy, early infantile, 21, 615833

NECTIN1	146.4	100.0%	100.0%	Orofacial cleft 7, 225060 Cleft lip/palate-ectodermal dysplasia syndrome, 225060
NECTIN4	132.0	100.0%	100.0%	Ectodermal dysplasia-syndactyly syndrome 1, 613573
NEDD4L	96.3	72.4%	71.5%	Periventricular nodular heterotopia 7, 617201
NEFH	121.3	99.5%	98.0%	Charcot-Marie-Tooth disease, axonal, type 2CC, 616924
NEFL	205.5	99.9%	97.9%	Charcot-Marie-Tooth disease, type 1F, 607734 Charcot-Marie-Tooth disease, dominant intermediate G, 617882 Charcot-Marie-Tooth disease, type 2E, 607684
NEK1	111.2	99.9%	98.0%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NEK2	86.4	98.8%	92.9%	?Retinitis pigmentosa 67, 615565
NEK8	153.7	100.0%	99.9%	?Nephronophthisis 9, 613824 Renal-hepatic-pancreatic dysplasia 2, 615415
NEK9	123.4	99.9%	98.9%	Lethal congenital contracture syndrome 10, 617022 Nevus comedonicus, somatic, 617025 ?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262
NEU1	150.1	99.5%	96.5%	Sialidosis, type II, 256550 Sialidosis, type I, 256550
NEUROD1	172.7	100.0%	99.9%	Maturity-onset diabetes of the young 6, 606394
NEUROD2	172.1	100.0%	100.0%	Epileptic encephalopathy, early infantile, 72, 618374
NEUROG3	205.9	100.0%	100.0%	Diarrhea 4, malabsorptive, congenital, 610370
NEXMIF	135.1	99.9%	99.4%	Mental retardation, X-linked 98, 300912
NEXN	87.7	96.0%	85.4%	Cardiomyopathy, hypertrophic, 20, 613876 Cardiomyopathy, dilated, 1CC, 613122
NF1	105.8	92.5%	89.3%	Watson syndrome, 193520 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321 Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210
NF2	99.4	100.0%	99.9%	Meningioma, NF2-related, somatic, 607174 Schwannomatosis, somatic, 162091 Neurofibromatosis, type 2, 101000
NFASC	128.3	100.0%	99.8%	Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356
NFE2L2	168.9	100.0%	100.0%	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744
NFIA	158.0	99.9%	99.0%	Brain malformations with or without urinary tract defects, 613735
NFIB	109.8	97.5%	96.8%	Macrocephaly, acquired, with impaired intellectual development, 618286
NFIX	195.4	100.0%	99.8%	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753

NFKB1	93.3	100.0%	98.6%	Immunodeficiency, common variable, 12, 616576
NFKB2	149.9	99.3%	97.1%	Immunodeficiency, common variable, 10, 615577
NFKBIA	148.5	96.7%	90.7%	Ectodermal dysplasia and immunodeficiency 2, 612132
NFU1	61.6	96.2%	77.8%	Multiple mitochondrial dysfunctions syndrome 1, 605711
NGF	211.6	100.0%	100.0%	Neuropathy, hereditary sensory and autonomic, type V, 608654
NGLY1	134.1	100.0%	99.9%	Congenital disorder of deglycosylation, 615273
NHEJ1	60.2	99.4%	94.3%	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
NHLRC1	184.6	100.0%	100.0%	Epilepsy, progressive myoclonic 2B (Lafora), 254780
NHLRC2	116.9	99.8%	97.6%	FINCA syndrome, 618278
NHP2	135.0	100.0%	99.8%	Dyskeratosis congenita, autosomal recessive 2, 613987
NHS	114.3	98.6%	96.5%	Nance-Horan syndrome, 302350 Cataract 40, X-linked, 302200
NIN	130.6	99.9%	99.2%	?Seckel syndrome 7, 614851
NIPA1	177.0	100.0%	100.0%	Spastic paraplegia 6, autosomal dominant, 600363
NIPAL4	137.3	100.0%	99.7%	Ichthyosis, congenital, autosomal recessive 6, 612281
NIPBL	123.0	98.9%	96.7%	Cornelia de Lange syndrome 1, 122470
NKX2-1	102.8	100.0%	99.9%	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978
NKX2-5	141.9	100.0%	100.0%	Ventricular septal defect 3, 614432 Hypoplastic left heart syndrome 2, 614435 Conotruncal heart malformations, variable, 217095 Tetralogy of Fallot, 187500 Hypothyroidism, congenital nongoitrous, 5, 225250 Atrial septal defect 7, with or without AV conduction defects, 108900
NKX2-6	159.7	100.0%	100.0%	Persistent truncus arteriosus, 217095 Conotruncal heart malformations, 217095
NKX3-2	161.2	100.0%	99.9%	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330
NKX6-2	144.9	99.6%	94.8%	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560
NLGN4X	154.6	99.5%	97.8%	Mental retardation, X-linked, 300495
NLRC4	168.2	100.0%	99.9%	Autoinflammation with infantile enterocolitis, 616050 ?Familial cold autoinflammatory syndrome 4, 616115
NLRP1	126.8	99.5%	97.9%	Palmoplantar carcinoma, multiple self-healing, 615225 Autoinflammation with arthritis and dyskeratosis, 617388
NLRP12	176.4	100.0%	100.0%	Familial cold autoinflammatory syndrome 2, 611762
NLRP3	146.0	100.0%	99.9%	Familial cold inflammatory syndrome 1, 120100 Muckle-Wells syndrome, 191900

				CINCA syndrome, 607115 Deafness, autosomal dominant 34, with or without inflammation, 617772 Keratoendothelitis fugax hereditaria, 148200
NLRP7	131.5	99.9%	98.9%	Hydatidiform mole, recurrent, 1, 231090
NME8	103.2	98.9%	92.4%	Ciliary dyskinesia, primary, 6, 610852
NMNAT1	118.9	99.9%	98.3%	Leber congenital amaurosis 9, 608553
NNT	127.0	99.9%	98.1%	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736
NOBOX	107.2	99.9%	98.6%	Premature ovarian failure 5, 611548
NOD2	136.5	100.0%	100.0%	Blau syndrome, 186580
NODAL	156.2	100.0%	100.0%	Heterotaxy, visceral, 5, 270100
NOG	270.7	100.0%	100.0%	Tarsal-carpal coalition syndrome, 186570 Symphalangism, proximal, 1A, 185800 Stapes ankylosis with broad thumbs and toes, 184460 Multiple synostoses syndrome 1, 186500 Brachydactyly, type B2, 611377
NOL3	132.1	99.9%	98.6%	?Myoclonus, familial, 1, 614937
NONO	83.1	99.7%	96.8%	Mental retardation, X-linked, syndromic 34, 300967
NOP10	124.6	100.0%	100.0%	Dyskeratosis congenita, autosomal recessive 1, 224230
NOP56	122.1	100.0%	99.2%	Spinocerebellar ataxia 36, 614153
NOTCH1	158.4	99.8%	99.2%	Aortic valve disease 1, 109730 Adams-Oliver syndrome 5, 616028
NOTCH2	130.5	100.0%	99.8%	Hajdu-Cheney syndrome, 102500 Alagille syndrome 2, 610205
NOTCH3	117.6	99.0%	95.0%	?Myofibromatosis, infantile 2, 615293 Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310 Lateral meningocele syndrome, 130720
NPC1	120.3	100.0%	99.4%	Niemann-Pick disease, type D, 257220 Niemann-Pick disease, type C1, 257220
NPC2	130.7	100.0%	99.9%	Niemann-pick disease, type C2, 607625
NPHP1	119.7	99.8%	97.8%	Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900 Joubert syndrome 4, 609583
NPHP3	121.6	99.7%	98.3%	Meckel syndrome 7, 267010 Renal-hepatic-pancreatic dysplasia 1, 208540 Nephronophthisis 3, 604387
NPHP4	134.2	100.0%	99.8%	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996

NPHS1	116.5	100.0%	99.8%	Nephrotic syndrome, type 1, 256300
NPHS2	121.7	100.0%	99.5%	Nephrotic syndrome, type 2, 600995
NPM1	65.4	95.9%	83.8%	Leukemia, acute myeloid, somatic, 601626
NPPA	172.3	100.0%	100.0%	Atrial standstill 2, 615745 Atrial fibrillation, familial, 6, 612201
NPR2	154.2	100.0%	99.5%	Short stature with nonspecific skeletal abnormalities, 616255 Epiphyseal chondrodysplasia, Miura type, 615923 Acromesomelic dysplasia, Maroteaux type, 602875
NPRL2	150.6	100.0%	100.0%	Epilepsy, familial focal, with variable foci 2, 617116
NPRL3	129.2	100.0%	99.9%	Epilepsy, familial focal, with variable foci 3, 617118
NR0B1	155.1	100.0%	99.3%	Adrenal hypoplasia, congenital, 300200 46XY sex reversal 2, dosage-sensitive, 300018
NR0B2	113.0	100.0%	99.9%	Obesity, mild, early-onset, 601665
NR1H4	124.6	99.9%	97.5%	Cholestasis, progressive familial intrahepatic, 5, 617049
NR2E3	117.8	99.9%	98.9%	Enhanced S-cone syndrome, 268100 Retinitis pigmentosa 37, 611131
NR2F1	261.9	100.0%	100.0%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NR2F2	262.6	100.0%	100.0%	Congenital heart defects, multiple types, 4, 615779
NR3C1	131.2	100.0%	100.0%	Glucocorticoid resistance, 615962
NR3C2	129.7	99.8%	98.5%	Pseudohypoaldosteronism type I, autosomal dominant, 177735 Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115
NR4A3	124.3	100.0%	100.0%	Chondrosarcoma, extraskeletal myxoid, 612237
NR5A1	126.3	100.0%	100.0%	Adrenocortical insufficiency, 612964 46, XX sex reversal 4, 617480 Premature ovarian failure 7, 612964 Spermatogenic failure 8, 613957 46XY sex reversal 3, 612965
NRAS	145.3	100.0%	100.0%	Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Colorectal cancer, somatic, 114500 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Thyroid carcinoma, follicular, somatic, 188470 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224
NRIP1	187.9	100.0%	100.0%	?Congenital anomalies of kidney and urinary tract 3, 618270
NRL	129.8	100.0%	99.0%	Retinitis pigmentosa 27, 613750 Retinal degeneration, autosomal recessive, clumped pigment type, 0

NRXN1	147.9	97.6%	97.3%	Pitt-Hopkins-like syndrome 2, 614325
NSD1	152.6	100.0%	99.8%	Sotos syndrome 1, 117550
NSDHL	133.7	99.9%	98.2%	CHILD syndrome, 308050 CK syndrome, 300831
NSMCE2	82.1	100.0%	98.5%	Seckel syndrome 10, 617253
NSMCE3	213.7	100.0%	100.0%	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241
NSMF	123.0	99.6%	98.1%	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
NSUN2	96.2	98.5%	94.8%	Mental retardation, autosomal recessive 5, 611091
NT5C2	119.4	97.9%	95.8%	Spastic paraplegia 45, autosomal recessive, 613162
NT5C3A	62.1	97.2%	83.5%	Anemia, hemolytic, due to UMPH1 deficiency, 266120
NT5E	157.1	100.0%	99.9%	Calcification of joints and arteries, 211800
NTF4	176.4	100.0%	99.3%	Glaucoma 1, open angle, 1O, 613100
NTHL1	134.4	100.0%	100.0%	Familial adenomatous polyposis 3, 616415
NTN1	217.9	100.0%	100.0%	Mirror movements 4, 618264
NTRK1	144.9	100.0%	99.7%	Medullary thyroid carcinoma, familial, 155240 Insensitivity to pain, congenital, with anhidrosis, 256800
NTRK2	142.9	100.0%	99.9%	Obesity, hyperphagia, and developmental delay, 613886 Epileptic encephalopathy, early infantile, 58, 617830
NUBPL	102.4	98.9%	94.2%	Mitochondrial complex I deficiency, nuclear type 21, 618242
NUMA1	146.9	100.0%	99.6%	Leukemia, acute promyelocytic, somatic, 612376
NUP107	126.3	99.9%	98.0%	Galloway-Mowat syndrome 7, 618348 ?Ovarian dysgenesis 6, 618078 Nephrotic syndrome, type 11, 616730
NUP133	119.3	99.9%	98.3%	Nephrotic syndrome, type 18, 618177 ?Galloway-Mowat syndrome 8, 618349
NUP155	117.6	98.5%	96.6%	?Atrial fibrillation 15, 615770
NUP160	139.2	100.0%	99.8%	?Nephrotic syndrome, type 19, 618178
NUP205	134.3	99.5%	98.7%	?Nephrotic syndrome, type 13, 616893
NUP214	158.2	99.9%	99.5%	Leukemia, acute myeloid, somatic, 601626 Leukemia, T-cell acute lymphoblastic, somatic, 613065
NUP37	154.9	100.0%	99.9%	?Microcephaly 24, primary, autosomal recessive, 618179
NUP62	124.5	100.0%	100.0%	Striatonigral degeneration, infantile, 271930
NUP85	132.5	100.0%	100.0%	Nephrotic syndrome, type 17, 618176
NUP88	143.4	100.0%	100.0%	Fetal akinesia deformation sequence 4, 618393
NUP93	122.3	97.1%	94.3%	Nephrotic syndrome, type 12, 616892
NUS1	51.9	72.8%	44.9%	Mental retardation, autosomal dominant 55, with seizures, 617831 ?Congenital disorder of glycosylation, type 1aa, 617082

NUTM2B-AS1	NC	NC	NC	?Oculopharyngeal myopathy with leukoencephalopathy 1, 618637
NXN	140.2	100.0%	100.0%	Robinow syndrome, autosomal recessive 2, 618529
NYX	155.4	99.9%	99.1%	Night blindness, congenital stationary (complete), 1A, X-linked, 310500
OAT	69.1	80.2%	69.8%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OBSL1	166.0	100.0%	99.9%	3-M syndrome 2, 612921
OCA2	123.6	99.8%	97.4%	Albinism, oculocutaneous, type II, 203200 Albinism, brown oculocutaneous, 203200
OCLN	179.8	100.0%	100.0%	Pseudo-TORCH syndrome 1, 251290
OCRL	106.2	99.9%	98.6%	Lowe syndrome, 309000 Dent disease 2, 300555
OFD1	52.3	85.5%	70.0%	Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Simpson-Golabi-Behmel syndrome, type 2, 300209
OGG1	127.5	100.0%	99.8%	Renal cell carcinoma, clear cell, somatic, 144700
OGT	108.0	99.9%	98.7%	Mental retardation, X-linked 106, 300997
OPA1	121.4	99.7%	97.5%	Behr syndrome, 210000 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896
OPA3	171.9	100.0%	99.9%	Optic atrophy 3 with cataract, 165300 3-methylglutaconic aciduria, type III, 258501
OPCML	149.2	99.6%	99.6%	Ovarian cancer, somatic, 167000
OPHN1	80.7	99.0%	95.5%	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
OPLAH	158.6	100.0%	100.0%	5-oxoprolinase deficiency, 260005
OPN1LW	61.2	67.9%	61.0%	Blue cone monochromacy, 303700 Colorblindness, protan, 303900
OPN1MW	61.9	68.7%	60.9%	Colorblindness, deutan, 303800 Blue cone monochromacy, 303700
OPN1SW	100.0	100.0%	100.0%	Colorblindness, tritan, 190900
OPTN	107.5	100.0%	99.8%	Glaucoma 1, open angle, E, 137760 Amyotrophic lateral sclerosis 12, 613435
ORAI1	226.3	100.0%	99.0%	Myopathy, tubular aggregate, 2, 615883 Immunodeficiency 9, 612782
ORC1	93.8	99.9%	98.9%	Meier-Gorlin syndrome 1, 224690
ORC4	68.8	97.5%	90.4%	Meier-Gorlin syndrome 2, 613800
ORC6	130.5	100.0%	99.9%	Meier-Gorlin syndrome 3, 613803

OSBPL2	146.5	100.0%	100.0%	Deafness, autosomal dominant 67, 616340
OSGEP	104.4	100.0%	97.8%	Galloway-Mowat syndrome 3, 617729
OSMR	131.5	100.0%	99.6%	Amyloidosis, primary localized cutaneous, 1, 105250
OSTM1	113.8	97.8%	92.1%	Osteopetrosis, autosomal recessive 5, 259720
OTC	111.3	100.0%	99.7%	Ornithine transcarbamylase deficiency, 311250
OTOA	101.3	99.7%	98.4%	Deafness, autosomal recessive 22, 607039
OTOF	148.9	100.0%	99.8%	Auditory neuropathy, autosomal recessive, 1, 601071 Deafness, autosomal recessive 9, 601071
OTOG	147.3	99.7%	99.1%	Deafness, autosomal recessive 18B, 614945
OTOGL	101.2	99.4%	96.7%	Deafness, autosomal recessive 84B, 614944
OTUD6B	118.1	99.9%	97.9%	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452
OTULIN	137.5	99.1%	96.1%	Autoinflammation, panniculitis, and dermatosis syndrome, 617099
OTX2	135.4	100.0%	99.6%	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125
OVOL2	130.8	99.9%	98.7%	Corneal dystrophy, posterior polymorphous, 1, 122000
OXCT1	123.6	99.7%	98.2%	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050
P2RX2	193.5	100.0%	100.0%	Deafness, autosomal dominant 41, 608224
P2RY12	187.4	100.0%	100.0%	Bleeding disorder, platelet-type, 8, 609821
P3H1	140.0	100.0%	100.0%	Osteogenesis imperfecta, type VIII, 610915
P3H2	102.6	100.0%	99.1%	Myopia, high, with cataract and vitreoretinal degeneration, 614292
P4HA2	119.3	99.9%	98.7%	Myopia 25, autosomal dominant, 617238
P4HB	117.0	94.6%	93.7%	Cole-Carpenter syndrome 1, 112240
P4HTM	177.7	100.0%	99.4%	Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493
PABPN1	82.7	82.5%	63.4%	Oculopharyngeal muscular dystrophy, 164300
PACS1	115.1	100.0%	99.8%	Schuurs-Hoeijmakers syndrome, 615009
PACS2	170.7	100.0%	99.6%	Epileptic encephalopathy, early infantile, 66, 618067
PADI3	147.3	100.0%	100.0%	Uncombable hair syndrome, 191480
PADI6	109.6	100.0%	99.2%	Preimplantation embryonic lethality 2, 617234
PAFAH1B1	78.7	91.7%	82.7%	Subcortical laminar heterotopia, 607432 Lissencephaly 1, 607432
PAH	128.9	100.0%	100.0%	Phenylketonuria, 261600
PAK1	102.7	100.0%	99.3%	Intellectual developmental disorder with macrocephaly, seizures, and speech delay, 618158
PAK3	85.7	99.0%	94.4%	Mental retardation, X-linked 30/47, 300558
PALB2	146.3	100.0%	99.9%	Fanconi anemia, complementation group N, 610832
PAM16	69.6	65.7%	65.3%	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320

PANK2	161.5	100.0%	100.0%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PANX1	137.7	100.0%	99.9%	Oocyte maturation defect 7, 618550
PAPSS2	107.4	99.8%	97.8%	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847
PARK7	88.7	100.0%	100.0%	Parkinson disease 7, autosomal recessive early-onset, 606324
PARN	127.9	100.0%	99.6%	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371 Dyskeratosis congenita, autosomal recessive 6, 616353
PARS2	193.2	100.0%	100.0%	Epileptic encephalopathy, early infantile, 75, 618437
PATL2	100.1	99.8%	96.3%	Oocyte maturation defect 4, 617743
PAX1	212.4	98.6%	94.1%	?Otofaciocervical syndrome 2, 615560
PAX2	198.0	100.0%	100.0%	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330
PAX3	116.1	100.0%	99.9%	Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220 Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820
PAX4	89.3	100.0%	99.7%	Maturity-onset diabetes of the young, type IX, 612225 Diabetes mellitus, type 2, 125853
PAX6	122.8	100.0%	99.9%	Optic nerve hypoplasia, 165550 ?Coloboma, ocular, 120200 Foveal hypoplasia 1, 136520 Aniridia, 106210 Keratitis, 148190 Cataract with late-onset corneal dystrophy, 106210 ?Coloboma of optic nerve, 120430 ?Morning glory disc anomaly, 120430 Anterior segment dysgenesis 5, multiple subtypes, 604229
PAX7	147.9	100.0%	100.0%	Myopathy, congenital, progressive, with scoliosis, 618578 Rhabdomyosarcoma 2, alveolar, 268220
PAX8	103.2	100.0%	100.0%	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700
PAX9	262.1	99.8%	99.6%	Tooth agenesis, selective, 3, 604625
PBX1	115.4	100.0%	98.3%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PC	170.2	99.9%	98.8%	Pyruvate carboxylase deficiency, 266150
PCBD1	109.5	100.0%	99.8%	Hyperphenylalaninemia, BH4-deficient, D, 264070
PCCA	97.7	99.1%	95.4%	Propionicacidemia, 606054
PCCB	114.9	99.5%	97.1%	Propionicacidemia, 606054
PCDH12	195.1	100.0%	100.0%	Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280

PCDH15	139.4	99.2%	98.9%	Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1F, 602083 Deafness, autosomal recessive 23, 609533
PCDH19	192.4	99.9%	98.9%	Epileptic encephalopathy, early infantile, 9, 300088
PCGF2	103.7	99.8%	97.3%	Turnpenny-Fry syndrome, 618371
PCK1	128.7	100.0%	100.0%	?Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680
PCLO	147.8	99.9%	99.0%	?Pontocerebellar hypoplasia, type 3, 608027
PCNA	93.9	100.0%	98.7%	?Ataxia-telangiectasia-like disorder 2, 615919
PCNT	124.1	99.8%	98.2%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PCSK1	144.7	100.0%	99.1%	Obesity with impaired prohormone processing, 600955
PCSK9	113.0	97.1%	93.3%	Hypercholesterolemia, familial, 3, 603776
PCYT1A	97.1	99.1%	95.7%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDCD10	94.5	100.0%	98.3%	Cerebral cavernous malformations 3, 603285
PDE10A	107.4	81.5%	80.3%	Dyskinesia, limb and orofacial, infantile-onset, 616921 Striatal degeneration, autosomal dominant, 616922
PDE11A	156.9	100.0%	99.9%	Pigmented nodular adrenocortical disease, primary, 2, 610475
PDE1C	110.4	99.9%	99.4%	?Deafness, autosomal dominant 74, 618140
PDE3A	127.5	100.0%	99.2%	Hypertension and brachydactyly syndrome, 112410
PDE4D	104.9	95.8%	95.0%	Acrodysostosis 2, with or without hormone resistance, 614613
PDE6A	105.5	100.0%	99.7%	Retinitis pigmentosa 43, 613810
PDE6B	171.1	100.0%	100.0%	Night blindness, congenital stationary, autosomal dominant 2, 163500 Retinitis pigmentosa-40, 613801
PDE6C	116.7	99.4%	97.2%	Cone dystrophy 4, 613093
PDE6D	121.7	100.0%	100.0%	?Joubert syndrome 22, 615665
PDE6G	134.0	100.0%	99.7%	Retinitis pigmentosa 57, 613582
PDE6H	60.3	96.8%	76.1%	Retinal cone dystrophy 3, 610024 Achromatopsia 6, 610024
PDE8B	104.0	99.9%	99.0%	Striatal degeneration, autosomal dominant, 609161 Pigmented nodular adrenocortical disease, primary, 3, 614190
PDGFB	127.3	100.0%	100.0%	Dermatofibrosarcoma protuberans, 607907 Basal ganglia calcification, idiopathic, 5, 615483 Meningioma, SIS-related, 607174
PDGFRA	127.7	100.0%	100.0%	Gastrointestinal stromal tumor/GIST-plus syndrome, somatic or familial, 175510 Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685
PDGFRB	138.0	99.8%	98.2%	Kosaki overgrowth syndrome, 616592 Basal ganglia calcification, idiopathic, 4, 615007

				Premature aging syndrome, Penttinen type, 601812 Myofibromatosis, infantile, 1, 228550
PDGFRL	137.4	100.0%	99.7%	Hepatocellular cancer, somatic, 114550 Colorectal cancer, somatic, 114500
PDHA1	88.0	98.6%	95.4%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	113.7	99.6%	97.7%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	132.4	100.0%	99.5%	Lacticacidemia due to PDX1 deficiency, 245349
PDK3	109.5	97.4%	94.3%	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905
PDP1	134.9	100.0%	100.0%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	106.9	97.6%	88.2%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	115.4	99.3%	95.2%	Coenzyme Q10 deficiency, primary, 3, 614652
PDX1	83.7	99.8%	96.8%	Pancreatic agenesis 1, 260370 MODY, type IV, 606392
PDXK	109.5	82.3%	78.6%	Neuropathy, hereditary motor and sensory, type VIC, with optic atrophy, 618511
PDYN	131.7	100.0%	100.0%	Spinocerebellar ataxia 23, 610245
PDZD7	103.9	99.7%	98.4%	Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472 Deafness, autosomal recessive 57, 618003
PEPD	126.5	100.0%	99.9%	Prolidase deficiency, 170100
PER2	97.9	100.0%	99.3%	?Advanced sleep phase syndrome, familial, 1, 604348
PER3	155.4	99.7%	98.0%	?Advanced sleep phase syndrome, familial, 3, 616882
PET100	95.2	99.7%	90.6%	Mitochondrial complex IV deficiency, 220110
PEX1	126.3	100.0%	99.1%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	123.8	100.0%	98.4%	Peroxisome biogenesis disorder 6B, 614871 Peroxisome biogenesis disorder 6A (Zellweger), 614870
PEX11B	93.3	100.0%	99.9%	?Peroxisome biogenesis disorder 14B, 614920
PEX12	125.4	100.0%	100.0%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	189.6	100.0%	100.0%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	144.7	99.8%	98.8%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	157.0	98.9%	95.7%	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	85.8	100.0%	98.9%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	137.4	100.0%	100.0%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867

PEX26	105.1	100.0%	100.0%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	108.6	100.0%	99.6%	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370
PEX5	115.8	100.0%	99.4%	Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716 Peroxisome biogenesis disorder 2A (Zellweger), 214110
PEX6	117.6	99.1%	93.9%	Peroxisome biogenesis disorder 4B, 614863 Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862
PEX7	108.8	91.3%	91.0%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PFKM	119.4	99.9%	99.3%	Glycogen storage disease VII, 232800
PFN1	166.1	100.0%	100.0%	Amyotrophic lateral sclerosis 18, 614808
PGAM2	180.7	100.0%	100.0%	Glycogen storage disease X, 261670
PGAP1	106.2	98.8%	94.3%	Mental retardation, autosomal recessive 42, 615802
PGAP2	145.1	100.0%	99.8%	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGAP3	74.5	63.7%	60.6%	Hyperphosphatasia with mental retardation syndrome 4, 615716
PGK1	47.0	92.1%	78.7%	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	134.8	100.0%	99.8%	Congenital disorder of glycosylation, type It, 614921
PGM3	148.4	100.0%	99.9%	Immunodeficiency 23, 615816
PHACTR1	111.1	100.0%	99.8%	Epileptic encephalopathy, early infantile, 70, 618298
PHC1	191.7	100.0%	99.4%	?Microcephaly 11, primary, autosomal recessive, 615414
PHEX	107.9	99.7%	98.2%	Hypophosphatemic rickets, X-linked dominant, 307800
PHF6	57.7	96.9%	84.1%	Borjeson-Forssman-Lehmann syndrome, 301900
PHF8	78.0	99.5%	96.1%	Mental retardation syndrome, X-linked, Siderius type, 300263
PHGDH	116.2	100.0%	99.6%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHIP	126.6	99.4%	96.6%	Chung-Jansen syndrome, 617991
PHKA1	90.2	97.8%	91.7%	Muscle glycogenosis, 300559
PHKA2	97.2	100.0%	99.2%	Glycogen storage disease, type IXa2, 306000 Glycogen storage disease, type IXa1, 306000
PHKB	124.0	100.0%	99.2%	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750
PHKG2	171.3	100.0%	100.0%	Glycogen storage disease IXc, 613027 Cirrhosis due to liver phosphorylase kinase deficiency, 0
PHOX2A	63.1	99.8%	94.3%	Fibrosis of extraocular muscles, congenital, 2, 602078

PHOX2B	166.3	100.0%	100.0%	Neuroblastoma with Hirschsprung disease, 613013 Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, 209880
PHYH	75.9	100.0%	97.9%	Refsum disease, 266500
PI4KA	95.6	94.4%	90.4%	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531
PIBF1	71.9	99.3%	92.7%	Joubert syndrome 33, 617767
PICALM	98.7	99.8%	95.8%	Leukemia, acute myeloid, somatic, 601626
PIEZO1	162.6	100.0%	99.7%	Lymphatic malformation 6, 616843 Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380
PIEZO2	106.9	99.9%	99.4%	Arthrogryposis, distal, with impaired proprioception and touch, 617146 Arthrogryposis, distal, type 5, 108145 ?Marden-Walker syndrome, 248700 Arthrogryposis, distal, type 3, 114300
PIGA	72.9	93.0%	83.4%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGB	94.8	99.1%	93.8%	Epileptic encephalopathy, early infantile, 80, 618580
PIGC	91.5	99.9%	95.9%	Glycosylphosphatidylinositol biosynthesis defect 16, 617816
PIGG	149.1	100.0%	99.8%	Mental retardation, autosomal recessive 53, 616917
PIGH	97.2	77.7%	68.2%	Glycosylphosphatidylinositol biosynthesis defect 17, 618010
PIGL	132.2	100.0%	99.1%	CHIME syndrome, 280000
PIGM	157.7	100.0%	100.0%	Glycosylphosphatidylinositol deficiency, 610293
PIGN	103.7	93.6%	90.1%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	157.3	100.0%	100.0%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGP	88.3	95.6%	86.3%	?Epileptic encephalopathy, early infantile, 55, 617599
PIGQ	151.8	94.8%	92.6%	Epileptic encephalopathy, early infantile, 77, 618548
PIGS	96.8	100.0%	99.7%	Glycosylphosphatidylinositol biosynthesis defect 18, 618143
PIGT	169.4	98.1%	98.1%	?Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGU	93.2	99.8%	98.2%	Glycosylphosphatidylinositol biosynthesis defect 21, 618590
PIGV	129.3	100.0%	100.0%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIGW	144.9	100.0%	99.8%	Glycosylphosphatidylinositol biosynthesis defect 11, 616025
PIGY	89.9	100.0%	100.0%	Hyperphosphatasia with mental retardation syndrome 6, 616809
PIH1D3	71.3	97.6%	85.8%	Ciliary dyskinesia, primary, 36, X-linked, 300991
PIK3C2A	126.2	99.1%	96.6%	Oculoskeletodental syndrome, 618440
PIK3CA	122.5	100.0%	99.8%	Ovarian cancer, somatic, 167000 Colorectal cancer, somatic, 114500 CLAPO syndrome, somatic, 613089 Cowden syndrome 5, 615108

				Hepatocellular carcinoma, somatic, 114550 Breast cancer, somatic, 114480 Macrodactyly, somatic, 155500 Keratosis, seborrheic, somatic, 182000 Gastric cancer, somatic, 613659 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Nevus, epidermal, somatic, 162900 CLOVE syndrome, somatic, 612918 Nonsmall cell lung cancer, somatic, 211980
PIK3CD	173.9	99.8%	98.1%	Immunodeficiency 14, 615513
PIK3R1	125.7	99.9%	98.8%	SHORT syndrome, 269880 Immunodeficiency 36, 616005 ?Agammaglobulinemia 7, autosomal recessive, 615214
PIK3R2	115.8	95.4%	91.8%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PIK3R5	130.3	100.0%	100.0%	Ataxia-oculomotor apraxia 3, 615217
PIKFYVE	136.7	99.9%	99.3%	Corneal fleck dystrophy, 121850
PINK1	96.5	97.0%	92.1%	Parkinson disease 6, early onset, 605909
PIP5K1C	151.5	99.9%	98.4%	Lethal congenital contractural syndrome 3, 611369
PITPNM3	134.7	99.9%	99.2%	Cone-rod dystrophy 5, 600977
PITX1	202.5	99.8%	97.6%	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800
PITX2	186.2	100.0%	99.6%	Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550 Anterior segment dysgenesis 4, 137600
PITX3	103.0	100.0%	99.8%	Anterior segment dysgenesis 1, multiple subtypes, 107250 Cataract 11, syndromic, autosomal recessive, 610623 Cataract 11, multiple types, 610623
PKD1	41.1	44.3%	36.9%	Polycystic kidney disease 1, 173900
PKD1L1	113.1	100.0%	99.5%	Heterotaxy, visceral, 8, autosomal, 617205
PKD2	105.1	99.2%	96.8%	Polycystic kidney disease 2, 613095
PKHD1	132.5	100.0%	99.5%	Polycystic kidney disease 4, with or without hepatic disease, 263200
PKLR	189.0	100.0%	99.9%	Pyruvate kinase deficiency, 266200 Adenosine triphosphate, elevated, of erythrocytes, 102900
PKP1	130.1	99.9%	98.7%	Ectodermal dysplasia/skin fragility syndrome, 604536
PKP2	97.3	97.4%	91.6%	Arrhythmogenic right ventricular dysplasia 9, 609040
PLA2G4A	121.8	100.0%	99.4%	Gastrointestinal ulceration, recurrent, with dysfunctional platelets, 618372
PLA2G6	121.0	99.9%	98.6%	Infantile neuroaxonal dystrophy 1, 256600 Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217

PLA2G7	117.7	100.0%	99.1%	Platelet-activating factor acetylhydrolase deficiency, 614278
PLAA	162.9	99.9%	98.4%	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527
PLAG1	172.3	100.0%	100.0%	Adenomas, salivary gland pleomorphic, somatic, 181030
PLAU	105.2	100.0%	99.3%	Quebec platelet disorder, 601709
PLCB1	134.5	100.0%	99.8%	Epileptic encephalopathy, early infantile, 12, 613722
PLCB4	102.7	99.8%	97.9%	Auriculocondylar syndrome 2, 614669
PLCD1	127.9	100.0%	99.6%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCE1	129.1	99.9%	99.2%	Nephrotic syndrome, type 3, 610725
PLCG2	110.7	100.0%	99.5%	Familial cold autoinflammatory syndrome 3, 614468 Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878
PLCZ1	72.0	99.7%	95.9%	?Spermatogenic failure 17, 617214
PLD1	116.6	100.0%	99.6%	Cardiac valvular defect, developmental, 212093
PLD3	184.9	100.0%	100.0%	?Spinocerebellar ataxia 46, 617770
PLEC	165.0	100.0%	100.0%	Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723 Epidermolysis bullosa simplex with pyloric atresia, 612138 Epidermolysis bullosa simplex with muscular dystrophy, 226670 ?Epidermolysis bullosa simplex with nail dystrophy, 616487 Epidermolysis bullosa simplex, Ogna type, 131950
PLEKHG2	166.7	100.0%	99.6%	Leukodystrophy and acquired microcephaly with or without dystonia, 616763
PLEKHG5	113.7	99.9%	99.0%	Spinal muscular atrophy, distal, autosomal recessive, 4, 611067 Charcot-Marie-Tooth disease, recessive intermediate C, 615376
PLEKHM1	139.5	100.0%	100.0%	Osteopetrosis, autosomal dominant 3, 618107 ?Osteopetrosis, autosomal recessive 6, 611497
PLG	95.7	87.8%	86.7%	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PLIN1	107.2	100.0%	99.8%	Lipodystrophy, familial partial, type 4, 613877
PLK4	145.8	99.9%	98.1%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLN	150.9	100.0%	100.0%	Cardiomyopathy, hypertrophic, 18, 613874 Cardiomyopathy, dilated, 1P, 609909
PLOD1	141.5	99.9%	97.9%	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD2	115.6	99.4%	96.2%	Bruck syndrome 2, 609220
PLOD3	120.2	100.0%	99.8%	Lysyl hydroxylase 3 deficiency, 612394
PLP1	113.6	99.9%	98.2%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PLPBP	97.2	99.9%	97.2%	Epilepsy, early-onset, vitamin B6-dependent, 617290
PLPP6	206.0	100.0%	99.5%	No OMIM Disease ID
PLS3	115.0	97.0%	95.5%	Bone mineral density QTL18, osteoporosis, 300910

PLVAP	178.8	100.0%	100.0%	Diarrhea 10, protein-losing enteropathy type, 618183
PMFBP1	111.9	99.9%	99.2%	Spermatogenic failure 31, 618112
PML	171.9	100.0%	100.0%	Leukemia, acute promyelocytic, PML/RARA type, 0
PMM2	130.3	100.0%	99.7%	Congenital disorder of glycosylation, type Ia, 212065
PMP2	124.4	100.0%	100.0%	Charcot-Marie-Tooth disease, demyelinating, type 1G, 618279
PMP22	100.0	99.0%	95.4%	Dejerine-Sottas disease, 145900 ?Neuropathy, inflammatory demyelinating, 139393 Charcot-Marie-Tooth disease, type 1E, 118300 Roussy-Levy syndrome, 180800 Neuropathy, recurrent, with pressure palsies, 162500 Charcot-Marie-Tooth disease, type 1A, 118220
PMPCA	113.3	99.6%	97.1%	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMPCB	121.6	100.0%	99.3%	Multiple mitochondrial dysfunctions syndrome 6, 617954
PMS2	96.2	83.5%	81.4%	Mismatch repair cancer syndrome, 276300 Colorectal cancer, hereditary nonpolyposis, type 4, 614337
PMVK	130.2	100.0%	99.9%	Porokeratosis 1, multiple types, 175800
PNKD	139.6	100.0%	99.9%	Paroxysmal nonkinesigenic dyskinesia 1, 118800
PNKP	123.1	100.0%	100.0%	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNLIP	135.2	99.9%	97.6%	?Pancreatic lipase deficiency, 614338
PNP	113.1	100.0%	99.8%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA1	176.0	100.0%	100.0%	Ichthyosis, congenital, autosomal recessive 10, 615024
PNPLA2	159.9	100.0%	99.9%	Neutral lipid storage disease with myopathy, 610717
PNPLA6	153.1	100.0%	99.6%	Spastic paraplegia 39, autosomal recessive, 612020 Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 ?Laurence-Moon syndrome, 245800
PNPLA8	118.2	100.0%	100.0%	?Mitochondrial myopathy with lactic acidosis, 251950
PNPO	78.3	100.0%	99.2%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
PNPT1	54.5	96.4%	83.0%	Deafness, autosomal recessive 70, 614934 Combined oxidative phosphorylation deficiency 13, 614932
POC1A	120.3	100.0%	100.0%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POC1B	77.8	99.7%	97.8%	Cone-rod dystrophy 20, 615973
POF1B	68.0	93.4%	82.3%	?Premature ovarian failure 2B, 300604
POFUT1	146.2	100.0%	99.2%	Dowling-Degos disease 2, 615327
POGLUT1	100.2	100.0%	99.1%	?Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232 Dowling-Degos disease 4, 615696

POGZ	129.7	99.5%	99.2%	White-Sutton syndrome, 616364
POLA1	104.7	99.1%	94.7%	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 Van Esch-O'Driscoll syndrome, 301030
POLD1	137.8	98.3%	94.9%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381
POLE	136.9	99.9%	99.6%	FILS syndrome, 615139 IMAGE-I syndrome, 618336
POLG	124.4	100.0%	99.8%	Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
POLG2	190.7	99.8%	98.1%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131 Mitochondrial DNA depletion syndrome 16 (hepatic type), 618528
POLH	121.6	100.0%	99.2%	Xeroderma pigmentosum, variant type, 278750
POLR1A	108.5	99.9%	98.8%	Acrofacial dysostosis, Cincinnati type, 616462
POLR1C	103.3	99.3%	95.4%	Treacher Collins syndrome 3, 248390 Leukodystrophy, hypomyelinating, 11, 616494
POLR1D	186.7	91.6%	91.6%	Treacher Collins syndrome 2, 613717
POLR2A	176.1	100.0%	100.0%	Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603
POLR3A	119.8	100.0%	99.9%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 Wiedemann-Rautenstrauch syndrome, 264090
POLR3B	132.0	99.9%	98.3%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMC	165.9	100.0%	100.0%	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734
POMGNT1	123.6	100.0%	99.8%	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 Retinitis pigmentosa 76, 617123 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280
POMGNT2	225.4	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830 Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135
POMK	144.2	100.0%	100.0%	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249
POMP	122.5	99.9%	95.8%	Proteasome-associated autoinflammatory syndrome 2, 618048 Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952
POMT1	137.5	99.6%	97.8%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155

POMT2	109.7	100.0%	99.1%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156
POP1	123.0	100.0%	99.2%	Anauxetic dysplasia 2, 617396
POR	195.6	99.4%	97.5%	Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571 Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750
PORCN	119.6	99.9%	98.9%	Focal dermal hypoplasia, 305600
POU1F1	109.5	99.9%	98.2%	Pituitary hormone deficiency, combined, 1, 613038
POU3F3	65.0	90.8%	76.5%	Snijders Blok-Fisher syndrome, 618604
POU3F4	148.1	100.0%	100.0%	Deafness, X-linked 2, 304400
POU4F3	297.4	100.0%	100.0%	Deafness, autosomal dominant 15, 602459
PPA2	92.7	98.0%	90.2%	?Sudden cardiac failure, alcohol-induced, 617223 Sudden cardiac failure, infantile, 617222
PPARG	136.8	100.0%	99.9%	Carotid intimal medial thickness 1, 609338 Insulin resistance, severe, digenic, 604367 Obesity, severe, 601665 Lipodystrophy, familial partial, type 3, 604367
PPCS	166.9	100.0%	99.2%	Cardiomyopathy, dilated, 2C, 618189
PPIB	118.3	100.0%	100.0%	Osteogenesis imperfecta, type IX, 259440
PPIP5K2	84.4	99.0%	93.7%	Deafness, autosomal recessive 100, 618422
PPM1D	177.2	100.0%	99.8%	Breast cancer, somatic, 114480 Jansen de Vries syndrome, 617450
PPM1K	139.2	100.0%	100.0%	?Maple syrup urine disease, mild variant, 615135
PPOX	101.7	99.9%	97.6%	Porphyria variegata, 176200
PPP1CB	110.7	100.0%	99.6%	Noonan syndrome-like disorder with loose anagen hair 2, 617506
PPP1R15B	130.2	100.0%	100.0%	Microcephaly, short stature, and impaired glucose metabolism 2, 616817
PPP1R3A	145.8	100.0%	99.4%	Insulin resistance, severe, digenic, 125853
PPP2CA	166.7	100.0%	100.0%	Neurodevelopmental disorder and language delay with or without structural brain abnormalities, 618354
PPP2R1A	138.5	91.6%	91.6%	Mental retardation, autosomal dominant 36, 616362
PPP2R1B	137.3	100.0%	99.9%	Lung cancer, somatic, 211980
PPP2R2B	129.0	99.8%	98.2%	Spinocerebellar ataxia 12, 604326
PPP2R3C	89.4	98.7%	90.3%	Gonadal dysgenesis, dysmorphic facies, retinal dystrophy, and myopathy, 618419 Spermatogenic failure 36, 618420
PPP2R5D	145.7	100.0%	100.0%	Mental retardation, autosomal dominant 35, 616355
PPP3CA	120.5	99.8%	97.5%	Epileptic encephalopathy, infantile or early childhood, 1, 617711 Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265
PPT1	140.2	90.3%	89.2%	Ceroid lipofuscinosis, neuronal, 1, 256730

PQBP1	173.3	100.0%	100.0%	Renpenning syndrome, 309500
PRCC	162.4	100.0%	99.9%	Renal cell carcinoma, papillary, 605074
PRCD	107.5	100.0%	100.0%	Retinitis pigmentosa 36, 610599
PRDM12	153.1	93.7%	91.1%	Neuropathy, hereditary sensory and autonomic, type VIII, 616488
PRDM16	230.2	100.0%	99.3%	Left ventricular noncompaction 8, 615373 Cardiomyopathy, dilated, 1LL, 615373
PRDM5	136.5	99.9%	98.2%	Brittle cornea syndrome 2, 614170
PRDM6	122.6	100.0%	99.6%	Patent ductus arteriosus 3, 617039
PRDM8	132.7	100.0%	98.1%	?Epilepsy, progressive myoclonic, 10, 616640
PRDX1	95.6	100.0%	99.8%	Methylmalonic aciduria and homocystinuria, cblC type, digenic, 277400
PREPL	102.2	99.8%	97.9%	Myasthenic syndrome, congenital, 22, 616224
PRF1	154.3	91.2%	90.7%	Aplastic anemia, 609135 Lymphoma, non-Hodgkin, 605027 Hemophagocytic lymphohistiocytosis, familial, 2, 603553
PRG4	131.9	99.7%	95.4%	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250
PRICKLE1	104.3	100.0%	99.8%	Epilepsy, progressive myoclonic 1B, 612437
PRIMPOL	117.5	97.4%	93.8%	Myopia 22, autosomal dominant, 615420
PRKACA	109.2	80.3%	79.8%	Cushing syndrome, ACTH-independent adrenal, somatic, 615830
PRKACG	226.7	100.0%	100.0%	?Bleeding disorder, platelet-type, 19, 616176
PRKAG2	135.6	99.1%	96.5%	Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200 Cardiomyopathy, hypertrophic 6, 600858
PRKAR1A	80.5	98.1%	92.8%	Myxoma, intracardiac, 255960 Carney complex, type 1, 160980 Pigmented nodular adrenocortical disease, primary, 1, 610489 Acrodysostosis 1, with or without hormone resistance, 101800 Adrenocortical tumor, somatic, 0
PRKCA	132.5	100.0%	100.0%	Pituitary tumor, invasive, 0
PRKCD	177.4	100.0%	100.0%	Autoimmune lymphoproliferative syndrome, type III, 615559
PRKCG	141.8	100.0%	99.5%	Spinocerebellar ataxia 14, 605361
PRKCSH	165.4	99.8%	96.3%	Polycystic liver disease 1, 174050
PRKD1	142.8	99.7%	99.5%	Congenital heart defects and ectodermal dysplasia, 617364
PRKDC	98.5	99.4%	96.4%	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PRKG1	127.2	99.8%	98.8%	Aortic aneurysm, familial thoracic 8, 615436
PRKN	86.2	80.1%	78.6%	Parkinson disease, juvenile, type 2, 600116 Ovarian cancer, somatic, 167000 Adenocarcinoma of lung, somatic, 211980

PRKRA	191.8	100.0%	99.9%	Dystonia 16, 612067
PRLR	134.5	100.0%	99.7%	Hyperprolactinemia, 615555 Multiple fibroadenomas of the breast, 615554
PRMT7	128.8	100.0%	99.9%	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157
PRNP	125.5	100.0%	100.0%	Insomnia, fatal familial, 600072 Huntington disease-like 1, 603218 Prion disease with protracted course, 606688 Cerebral amyloid angiopathy, PRNP-related, 137440 Creutzfeldt-Jakob disease, 123400 Gerstmann-Straussler disease, 137440
PROC	158.3	100.0%	100.0%	Thrombophilia due to protein C deficiency, autosomal dominant, 176860 Thrombophilia due to protein C deficiency, autosomal recessive, 612304
PRODH	88.9	91.8%	83.0%	Hyperprolinemia, type I, 239500
PROK2	117.9	100.0%	100.0%	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	245.3	100.0%	100.0%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROM1	106.6	97.6%	95.2%	Retinitis pigmentosa 41, 612095 Stargardt disease 4, 603786 Cone-rod dystrophy 12, 612657 Macular dystrophy, retinal, 2, 608051
PROP1	107.5	93.9%	86.2%	Pituitary hormone deficiency, combined, 2, 262600
PROS1	92.6	97.3%	92.4%	Thrombophilia due to protein S deficiency, autosomal recessive, 614514 Thrombophilia due to protein S deficiency, autosomal dominant, 612336
PRPF3	74.9	98.9%	95.5%	Retinitis pigmentosa 18, 601414
PRPF31	127.5	99.8%	97.9%	Retinitis pigmentosa 11, 600138
PRPF4	128.6	100.0%	99.8%	Retinitis pigmentosa 70, 615922
PRPF6	120.0	100.0%	99.8%	Retinitis pigmentosa 60, 613983
PRPF8	109.8	100.0%	99.3%	Retinitis pigmentosa 13, 600059
PRPH2	217.4	100.0%	100.0%	Macular dystrophy, patterned, 1, 169150 Retinitis punctata albescens, 136880 Choroidal dystrophy, central areolar 2, 613105 Retinitis pigmentosa 7 and digenic form, 608133 Leber congenital amaurosis 18, 608133 Macular dystrophy, vitelliform, 3, 608161
PRPS1	113.2	100.0%	99.9%	Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Phosphoribosylpyrophosphate synthetase superactivity, 300661 Deafness, X-linked 1, 304500 Arts syndrome, 301835 Gout, PRPS-related, 300661

PRRT2	124.1	100.0%	99.7%	Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751 Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066
PRRX1	106.7	100.0%	99.9%	Agnathia-otocephaly complex, 202650
PRSS1	147.1	100.0%	99.9%	Pancreatitis, hereditary, 167800
PRSS12	144.0	100.0%	99.8%	Mental retardation, autosomal recessive 1, 249500
PRSS56	111.2	99.9%	99.3%	Microphthalmia, isolated 6, 613517
PRUNE1	120.3	100.0%	99.6%	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481
PRX	177.3	100.0%	99.9%	Charcot-Marie-Tooth disease, type 4F, 614895 Dejerine-Sottas disease, 145900
PSAP	103.3	100.0%	99.5%	Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Combined SAP deficiency, 611721 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PSAT1	46.2	91.6%	74.2%	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992
PSEN1	132.4	100.0%	100.0%	Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 ?Acne inversa, familial, 3, 613737 Alzheimer disease, type 3, 607822 Dementia, frontotemporal, 600274 Pick disease, 172700 Cardiomyopathy, dilated, 1U, 613694
PSEN2	117.1	100.0%	99.9%	Alzheimer disease-4, 606889 Cardiomyopathy, dilated, 1V, 613697
PSENEN	98.3	100.0%	100.0%	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736
PSMB4	124.8	100.0%	99.8%	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591
PSMB8	119.3	100.0%	99.3%	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040
PSMB9	85.2	100.0%	99.1%	?Proteasome-associated autoinflammatory syndrome 3, digenic, 617591
PSMC3IP	111.0	100.0%	100.0%	Ovarian dysgenesis 3, 614324
PSMD12	87.5	98.2%	90.2%	Stankiewicz-Isidor syndrome, 617516
PSPH	122.1	100.0%	99.8%	Phosphoserine phosphatase deficiency, 614023
PSTPIP1	114.3	99.9%	99.3%	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416
PTCH1	117.3	100.0%	99.2%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly 7, 610828

PTCH2	131.5	99.9%	98.8%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Medulloblastoma, somatic, 155255
PTDSS1	116.2	100.0%	100.0%	Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	125.3	99.7%	95.5%	Prostate cancer, somatic, 176807 Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309
PTF1A	138.4	100.0%	99.7%	Pancreatic and cerebellar agenesis, 609069 Pancreatic agenesis 2, 615935
PTGIS	123.3	99.8%	97.4%	Hypertension, essential, 145500
PTH	92.6	99.9%	95.9%	Hypoparathyroidism, autosomal dominant, 146200 Hypoparathyroidism, autosomal recessive, 146200
PTH1R	121.2	100.0%	99.5%	Metaphyseal chondrodysplasia, Murk Jansen type, 156400 Failure of tooth eruption, primary, 125350 Eiken syndrome, 600002 Chondrodysplasia, Blomstrand type, 215045
PTHLH	138.1	99.1%	91.2%	Brachydactyly, type E2, 613382
PTPN11	80.5	98.8%	91.3%	LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Noonan syndrome 1, 163950 Leukemia, juvenile myelomonocytic, somatic, 607785
PTPN12	141.4	99.2%	96.3%	Colon cancer, somatic, 114500
PTPN14	170.9	99.3%	96.4%	Choanal atresia and lymphedema, 613611
PTPRC	98.4	98.3%	93.6%	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971
PTPRF	170.4	100.0%	99.9%	?Breasts and/or nipples, aplasia or hypoplasia of, 2, 616001
PTPRJ	139.8	98.2%	96.5%	Colon cancer, somatic, 114500
PTPRO	127.1	100.0%	99.3%	Nephrotic syndrome, type 6, 614196
PTPRQ	97.7	94.6%	91.6%	Deafness, autosomal dominant 73, 617663 Deafness, autosomal recessive 84A, 613391
PTRH2	209.0	100.0%	100.0%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PTS	103.1	100.0%	98.3%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUF60	178.6	100.0%	99.6%	Verheij syndrome, 615583
PUM1	132.0	100.0%	99.7%	Spinocerebellar ataxia 47, 617931
PURA	233.4	99.9%	98.8%	Mental retardation, autosomal dominant 31, 616158
PUS1	125.7	99.8%	98.1%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PUS3	163.9	100.0%	100.0%	Mental retardation, autosomal recessive 55, 617051

PUS7	134.7	100.0%	99.5%	Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342
PXDN	150.2	100.0%	99.8%	Anterior segment dysgenesis 7, with sclerocornea, 269400
PYCR1	105.0	100.0%	99.0%	Cutis laxa, autosomal recessive, type IIIB, 614438 Cutis laxa, autosomal recessive, type IIB, 612940
PYCR2	129.0	99.7%	97.6%	Leukodystrophy, hypomyelinating, 10, 616420
PYGL	147.0	100.0%	99.9%	Glycogen storage disease VI, 232700
PYGM	130.4	100.0%	100.0%	McArdle disease, 232600
PYROXD1	46.8	90.5%	76.6%	Myopathy, myofibrillar, 8, 617258
QARS	137.7	100.0%	100.0%	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	103.7	99.9%	99.1%	Hyperphenylalaninemia, BH4-deficient, C, 261630
QRICH1	142.0	100.0%	99.6%	Ververi-Brady syndrome, 617982
QRICH2	123.2	95.5%	94.2%	Spermatogenic failure 35, 618341
RAB11B	229.2	100.0%	100.0%	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807
RAB18	80.5	99.7%	95.3%	Warburg micro syndrome 3, 614222
RAB23	102.4	100.0%	99.8%	Carpenter syndrome, 201000
RAB27A	123.3	100.0%	99.6%	Griscelli syndrome, type 2, 607624
RAB28	64.3	99.1%	91.0%	Cone-rod dystrophy 18, 615374
RAB33B	199.5	100.0%	100.0%	Smith-McCort dysplasia 2, 615222
RAB39B	108.8	100.0%	100.0%	Waisman syndrome, 311510 Mental retardation, X-linked 72, 300271
RAB3GAP1	123.3	99.4%	98.8%	Warburg micro syndrome 1, 600118
RAB3GAP2	89.9	99.7%	96.1%	Warburg micro syndrome 2, 614225 Martsolf syndrome, 212720
RAB7A	117.9	100.0%	100.0%	Charcot-Marie-Tooth disease, type 2B, 600882
RAC1	108.4	99.5%	95.4%	Mental retardation, autosomal dominant 48, 617751
RAC2	109.4	100.0%	99.4%	Neutrophil immunodeficiency syndrome, 608203
RAC3	132.6	98.1%	95.4%	Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577
RAD21	80.8	98.0%	93.5%	?Mungan syndrome, 611376 Cornelia de Lange syndrome 4, 614701
RAD50	100.6	96.4%	89.5%	Nijmegen breakage syndrome-like disorder, 613078
RAD51	104.8	89.4%	89.4%	?Fanconi anemia, complementation group R, 617244 Mirror movements 2, 614508
RAD51C	141.9	100.0%	99.7%	Fanconi anemia, complementation group O, 613390
RAD54B	111.6	99.4%	95.9%	Colon cancer, somatic, 114500 Lymphoma, non-Hodgkin, somatic, 605027
RAD54L	104.0	100.0%	99.3%	Lymphoma, non-Hodgkin, somatic, 605027 Adenocarcinoma, colonic, somatic, 0

RAF1	111.1	100.0%	99.9%	LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553 Cardiomyopathy, dilated, 1NN, 615916
RAG1	158.7	100.0%	100.0%	Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457 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
RAG2	188.7	100.0%	100.0%	Severe combined immunodeficiency, B cell-negative, 601457 Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554
RAI1	216.5	100.0%	100.0%	Smith-Magenis syndrome, 182290
RAP1GDS1	96.5	99.5%	92.9%	Lymphocytic leukemia, acute T-cell, 0
RAPGEF2	144.3	99.8%	98.8%	?Epilepsy, familial adult myoclonic, 7, 618075
RAPSN	162.2	99.9%	99.2%	Fetal akinesia deformation sequence 2, 618388 Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326
RARB	93.5	100.0%	100.0%	Microphthalmia, syndromic 12, 615524
RARS	93.8	93.3%	88.0%	Leukodystrophy, hypomyelinating, 9, 616140
RARS2	102.7	100.0%	99.3%	Pontocerebellar hypoplasia, type 6, 611523
RASA1	109.9	98.7%	95.6%	Capillary malformation-arteriovenous malformation 1, 608354 Basal cell carcinoma, somatic, 605462
RASGRP1	112.5	100.0%	99.7%	Immunodeficiency 64, 618534
RASGRP2	111.5	100.0%	99.8%	?Bleeding disorder, platelet-type, 18, 615888
RAX	156.7	100.0%	99.9%	Microphthalmia, isolated 3, 611038
RAX2	98.8	100.0%	100.0%	?Macular degeneration, age-related, 6, 613757 Cone-rod dystrophy 11, 610381
RB1	87.3	97.8%	91.8%	Small cell cancer of the lung, somatic, 182280 Bladder cancer, somatic, 109800 Retinoblastoma, trilateral, 180200 Osteosarcoma, somatic, 259500 Retinoblastoma, 180200
RB1CC1	105.8	99.4%	95.7%	Breast cancer, somatic, 114480
RBBP8	117.9	100.0%	99.3%	Jawad syndrome, 251255 Seckel syndrome 2, 606744 Pancreatic carcinoma, somatic, 0
RBCK1	118.4	100.0%	99.4%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RBM10	122.0	99.9%	98.5%	TARP syndrome, 311900
RBM20	193.1	100.0%	99.8%	Cardiomyopathy, dilated, 1DD, 613172

RBM28	132.1	100.0%	100.0%	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RBM8A	89.0	100.0%	98.2%	Thrombocytopenia-absent radius syndrome, 274000
RBMX	41.2	91.9%	74.8%	?Mental retardation, X-linked, syndromic 11, Shashi type, 300238
RBP3	168.5	100.0%	100.0%	?Retinitis pigmentosa 66, 615233
RBP4	149.6	99.8%	96.8%	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RBPJ	72.1	95.8%	87.2%	Adams-Oliver syndrome 3, 614814
RCBTB1	98.4	99.9%	99.0%	Retinal dystrophy with or without extraocular anomalies, 617175
RD3	190.9	100.0%	100.0%	Leber congenital amaurosis 12, 610612
RDH11	94.8	99.9%	98.8%	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108
RDH12	86.4	99.8%	98.1%	Leber congenital amaurosis 13, 612712
RDH5	182.9	100.0%	100.0%	Fundus albipunctatus, 136880
RDX	37.2	87.1%	66.8%	Deafness, autosomal recessive 24, 611022
RECQL4	181.4	100.0%	100.0%	RAPADILINO syndrome, 266280 Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2,, 268400
REEP1	74.4	78.8%	76.7%	Spastic paraplegia 31, autosomal dominant, 610250 ?Neuronopathy, distal hereditary motor, type VB, 614751
REEP2	189.2	100.0%	99.8%	?Spastic paraplegia 72, autosomal dominant, 615625 ?Spastic paraplegia 72, autosomal recessive, 615625
REEP6	226.6	100.0%	99.9%	Retinitis pigmentosa 77, 617304
RELA	105.2	99.8%	99.3%	?Mucocutaneous ulceration, chronic, 618287
RELB	120.3	99.7%	96.5%	?Immunodeficiency 53, 617585
RELN	131.8	100.0%	99.6%	Lissencephaly 2 (Norman-Roberts type), 257320
RELT	156.5	100.0%	100.0%	Amelogenesis imperfecta, type IIIC, 618386
REN	135.2	100.0%	99.9%	Renal tubular dysgenesis, 267430 Hyperuricemic nephropathy, familial juvenile 2, 613092
REPS1	120.7	99.3%	97.0%	?Neurodegeneration with brain iron accumulation 7, 617916
RERE	84.5	96.9%	94.3%	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975
REST	123.0	98.5%	98.4%	Fibromatosis, gingival, 5, 617626 ?Deafness, autosomal dominant 27, 612431
RET	149.1	100.0%	99.3%	Multiple endocrine neoplasia IIB, 162300 Pheochromocytoma, 171300 Multiple endocrine neoplasia IIA, 171400 Medullary thyroid carcinoma, 155240 Central hypoventilation syndrome, congenital, 209880
RETREG1	132.5	100.0%	98.6%	Neuropathy, hereditary sensory and autonomic, type IIB, 613115

RFC1	124.1	99.9%	98.3%	Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome, 614575
RFT1	106.6	100.0%	99.2%	Congenital disorder of glycosylation, type In, 612015
RFWD3	106.8	100.0%	99.6%	?Fanconi anemia, complementation group W, 617784
RFX5	117.8	99.9%	98.2%	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFX6	150.2	100.0%	99.7%	Mitchell-Riley syndrome, 615710
RFXANK	133.2	100.0%	99.9%	MHC class II deficiency, complementation group B, 209920
RFXAP	127.3	100.0%	100.0%	Bare lymphocyte syndrome, type II, complementation group D, 209920
RGR	131.3	100.0%	99.6%	Retinitis pigmentosa 44, 613769
RGS9	119.4	98.9%	96.9%	Bradyopsia, 608415
RGS9BP	170.1	100.0%	100.0%	Bradyopsia, 608415
RHAG	122.3	100.0%	99.0%	Overhydrated hereditary stomatocytosis, 185000 Anemia, hemolytic, Rh-null, regulator type, 268150
RHBDF2	117.3	99.9%	99.0%	Tylosis with esophageal cancer, 148500
RHCE	159.1	97.5%	97.3%	Rh-null disease, amorph type, 617970
RHO	180.4	100.0%	100.0%	Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis punctata albescens, 136880 Retinitis pigmentosa 4, autosomal dominant or recessive, 613731
RHOBTB2	206.4	100.0%	100.0%	Epileptic encephalopathy, early infantile, 64, 618004
RIMS1	129.8	99.6%	98.1%	Cone-rod dystrophy 7, 603649
RIN2	129.3	100.0%	99.7%	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075
RINT1	158.2	99.8%	98.2%	Infantile liver failure syndrome 3, 618641
RIPK1	106.9	99.9%	99.1%	Immunodeficiency 57, 618108
RIPK4	189.1	100.0%	100.0%	Popliteal pterygium syndrome, Bartsocas-Papas type, 263650 CHAND syndrome, 214350
RIPOR2	114.9	100.0%	99.9%	?Deafness, autosomal recessive 104, 616515
RIPPLY2	83.9	99.9%	97.4%	?Spondylocostal dysostosis 6, 616566
RIT1	142.5	100.0%	100.0%	Noonan syndrome 8, 615355
RLBP1	129.0	100.0%	99.9%	Fundus albipunctatus, 136880 Bothnia retinal dystrophy, 607475 Retinitis punctata albescens, 136880 Newfoundland rod-cone dystrophy, 607476
RLIM	99.8	99.7%	97.7%	Tonne-Kalscheuer syndrome, 300978
RMND1	130.7	100.0%	99.0%	Combined oxidative phosphorylation deficiency 11, 614922
RMRP	NC	NC	NC	Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460

RNASEH1	105.2	98.0%	92.6%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479
RNASEH2A	143.0	100.0%	100.0%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	98.0	99.8%	96.3%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	314.2	100.0%	100.0%	Aicardi-Goutieres syndrome 3, 610329
RNASEL	126.3	100.0%	99.9%	Prostate cancer 1, 601518
RNASET2	109.5	95.4%	89.8%	Leukoencephalopathy, cystic, without megalencephaly, 612951
RNF113A	149.1	100.0%	100.0%	?Trichothiodystrophy 5, nonphotosensitive, 300953
RNF125	178.2	99.9%	98.6%	Tenorio syndrome, 616260
RNF13	80.5	91.4%	71.6%	Epileptic encephalopathy, early infantile, 73, 618379
RNF139	179.3	100.0%	100.0%	Renal cell carcinoma, 144700
RNF168	187.1	100.0%	99.4%	RIDDLE syndrome, 611943
RNF170	124.9	99.9%	98.0%	Ataxia, sensory, 1, autosomal dominant, 608984
RNF212	116.9	99.9%	98.5%	Recombination rate QTL 1, 612042
RNF216	128.1	100.0%	98.3%	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840
RNF43	155.9	100.0%	99.0%	Sessile serrated polyposis cancer syndrome, 617108
RNF6	140.5	100.0%	99.2%	Esophageal carcinoma, somatic, 133239
RNPC3	40.8	86.2%	65.9%	?Growth hormone deficiency, isolated, type V, 618160
RNU4ATAC	NC	NC	NC	Microcephalic osteodysplastic primordial dwarfism, type I, 210710 Roifman syndrome, 616651
ROBO2	129.6	98.8%	97.4%	Vesicoureteral reflux 2, 610878
ROBO3	114.9	99.7%	98.0%	Gaze palsy, familial horizontal, with progressive scoliosis, 1, 607313
ROBO4	116.8	100.0%	99.9%	Aortic valve disease 8, 618496
ROGDI	141.6	100.0%	99.9%	Kohlschutter-Tonz syndrome, 226750
ROM1	139.5	100.0%	99.9%	Retinitis pigmentosa 7, digenic form, 608133
ROR1	154.0	98.9%	97.3%	?Deafness, autosomal recessive 108, 617654
ROR2	176.7	100.0%	99.9%	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RORA	105.6	97.1%	91.6%	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060
RORC	132.1	100.0%	100.0%	Immunodeficiency 42, 616622
RP1	112.2	91.5%	91.0%	Retinitis pigmentosa 1, 180100
RP1L1	153.1	100.0%	100.0%	Occult macular dystrophy, 613587
RP2	159.7	100.0%	99.2%	Retinitis pigmentosa 2, 312600
RP9	64.9	95.2%	81.1%	?Retinitis pigmentosa 9, 180104
RPE65	133.0	100.0%	99.8%	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794
RPGR	80.2	83.1%	74.6%	Retinitis pigmentosa 3, 300029 Cone-rod dystrophy, X-linked, 1, 304020

				Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 Macular degeneration, X-linked atrophic, 300834
RPGRIP1	132.7	100.0%	99.9%	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826
RPGRIP1L	124.2	96.8%	95.8%	COACH syndrome, 216360 Meckel syndrome 5, 611561 Joubert syndrome 7, 611560
RPIA	119.9	100.0%	99.4%	Ribose 5-phosphate isomerase deficiency, 608611
RPL10	68.6	98.5%	89.7%	Mental retardation, X-linked, syndromic, 35, 300998
RPL11	88.1	100.0%	99.3%	Diamond-Blackfan anemia 7, 612562
RPL15	34.3	87.5%	74.6%	?Diamond-Blackfan anemia 12, 615550
RPL18	97.9	100.0%	99.5%	?Diamond-Blackfan anemia 18, 618310
RPL21	54.3	84.1%	62.5%	Hypotrichosis 12, 615885
RPL26	33.4	93.3%	73.1%	?Diamond-Blackfan anemia 11, 614900
RPL27	35.3	76.1%	57.7%	?Diamond-Blackfan anemia 16, 617408
RPL35	68.4	89.9%	77.3%	?Diamond-Blackfan anemia 19, 618312
RPL35A	75.9	97.4%	84.8%	Diamond-Blackfan anemia 5, 612528
RPL5	36.3	86.2%	68.8%	Diamond-Blackfan anemia 6, 612561
RPS10	96.0	99.4%	93.2%	Diamond-Blackfan anemia 9, 613308
RPS14	111.4	99.5%	94.4%	Macrocytic anemia, refractory, due to 5q deletion, somatic, 153550
RPS15A	58.2	97.8%	87.3%	?Diamond-Blackfan anemia 20, 618313
RPS17	41.4	90.1%	73.3%	Diamond-Blackfan anemia 4, 612527
RPS19	81.5	100.0%	98.3%	Diamond-Blackfan anemia 1, 105650
RPS23	62.0	90.4%	80.4%	Brachycephaly, trichomegaly, and developmental delay, 617412
RPS24	91.0	94.6%	89.4%	Diamond-blackfan anemia 3, 610629
RPS26	84.1	91.3%	76.0%	Diamond-Blackfan anemia 10, 613309
RPS27	34.2	91.2%	59.7%	?Diamond-Blackfan anemia 17, 617409
RPS28	59.6	100.0%	97.9%	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164
RPS29	100.0	98.7%	96.1%	Diamond-Blackfan anemia 13, 615909
RPS6KA3	84.8	98.2%	91.5%	Mental retardation, X-linked 19, 300844 Coffin-Lowry syndrome, 303600
RPS7	79.0	86.9%	69.3%	Diamond-Blackfan anemia 8, 612563
RPSA	68.7	100.0%	99.6%	Asplenia, isolated congenital, 271400
RRAS2	84.3	92.3%	80.0%	Noonan syndrome 12, 618624 Ovarian carcinoma, 0

RRM2B	142.6	100.0%	99.4%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075
RS1	54.6	99.2%	89.0%	Retinoschisis, 312700
RSPH1	127.7	100.0%	100.0%	Ciliary dyskinesia, primary, 24, 615481
RSPH3	148.8	100.0%	99.3%	Ciliary dyskinesia, primary, 32, 616481
RSPH4A	155.2	98.7%	96.4%	Ciliary dyskinesia, primary, 11, 612649
RSPH9	143.1	99.9%	98.0%	Ciliary dyskinesia, primary, 12, 612650
RSPO1	115.6	100.0%	100.0%	Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644 Palmoplantar hyperkeratosis and true hermaphroditism, 610644
RSPO2	134.3	96.8%	89.6%	Tetraamelia syndrome 2, 618021 ?Humerofemoral hypoplasia with radiotibial ray deficiency, 618022
RSPO4	161.1	100.0%	100.0%	Anonychia congenita, 206800
RSPRY1	143.3	100.0%	100.0%	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723
RSRC1	74.3	99.1%	94.6%	Intellectual developmental disorder, autosomal recessive 70, 618402
RTEL1	145.6	99.8%	98.2%	Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 Dyskeratosis congenita, autosomal dominant 4, 615190
RTN2	156.1	99.9%	98.8%	Spastic paraplegia 12, autosomal dominant, 604805
RTN4IP1	80.2	99.6%	98.2%	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
RTTN	117.3	98.6%	97.3%	Microcephaly, short stature, and polymicrogyria with seizures, 614833
RUBCN	105.3	100.0%	99.5%	?Spinocerebellar ataxia, autosomal recessive 15, 615705
RUNX1	92.9	99.9%	97.4%	Platelet disorder, familial, with associated myeloid malignancy, 601399 Leukemia, acute myeloid, 601626
RUNX2	109.5	73.6%	72.3%	Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510 Cleidocranial dysplasia, 119600 Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600
RUSC2	208.1	100.0%	100.0%	Mental retardation, autosomal recessive 61, 617773
RYR1	128.2	99.3%	96.8%	Central core disease, 117000 King-Denborough syndrome, 145600 Minicore myopathy with external ophthalmoplegia, 255320 Neuromuscular disease, congenital, with uniform type 1 fiber, 117000
RYR2	127.4	99.9%	98.7%	Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772 Arrhythmogenic right ventricular dysplasia 2, 600996
S1PR2	225.1	99.7%	97.7%	Deafness, autosomal recessive 68, 610419
SACS	151.1	100.0%	100.0%	Spastic ataxia, Charlevoix-Saguenay type, 270550

SAG	131.3	100.0%	99.9%	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758
SALL1	127.8	99.9%	99.3%	Townes-Brocks syndrome 1, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480
SALL2	143.2	100.0%	100.0%	?Coloboma, ocular, autosomal recessive, 216820
SALL4	147.5	100.0%	98.7%	Duane-radial ray syndrome, 607323 IVIC syndrome, 147750
SAMD12	140.0	100.0%	100.0%	Epilepsy, familial adult myoclonic, 1, 601068
SAMD9	161.7	100.0%	100.0%	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455
SAMD9L	170.6	100.0%	100.0%	Ataxia-pancytopenia syndrome, 159550
SAMHD1	135.4	100.0%	98.7%	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SAR1B	116.2	96.5%	90.4%	Chylomicron retention disease, 246700
SARS	112.1	100.0%	99.2%	?Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709
SARS2	129.0	95.4%	93.8%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SASH1	163.9	99.4%	98.1%	?Cancer, alopecia, pigment dyscrasia, onychodystrophy, and keratoderma, 618373 Dyschromatosis universalis hereditaria 1, 127500
SASS6	73.5	99.9%	98.1%	?Microcephaly 14, primary, autosomal recessive, 616402
SATB2	115.6	99.9%	98.3%	Glass syndrome, 612313
SBDS	167.5	100.0%	100.0%	Shwachman-Diamond syndrome, 260400
SBF1	136.9	99.6%	98.4%	Charcot-Marie-Tooth disease, type 4B3, 615284
SBF2	107.8	99.9%	99.0%	Charcot-Marie-Tooth disease, type 4B2, 604563
SC5D	149.4	100.0%	99.6%	Lathosterolosis, 607330
SCAPER	137.7	97.8%	96.0%	Intellectual developmental disorder and retinitis pigmentosa, 618195
SCARB2	106.4	99.9%	99.1%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCARF2	120.2	99.6%	97.7%	Van den Ende-Gupta syndrome, 600920
SCN10A	141.5	100.0%	99.5%	Episodic pain syndrome, familial, 2, 615551
SCN11A	125.5	99.3%	97.2%	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN1A	120.1	99.9%	99.0%	Febrile seizures, familial, 3A, 604403 Migraine, familial hemiplegic, 3, 609634 Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Epileptic encephalopathy, early infantile, 6 (Dravet syndrome), 607208
SCN1B	186.5	100.0%	99.3%	Epileptic encephalopathy, early infantile, 52, 617350 Atrial fibrillation, familial, 13, 615377 Cardiac conduction defect, nonspecific, 612838

				Epilepsy, generalized, with febrile seizures plus, type 1, 604233 Brugada syndrome 5, 612838
SCN2A	135.0	99.5%	97.4%	Epileptic encephalopathy, early infantile, 11, 613721 Seizures, benign familial infantile, 3, 607745
SCN2B	190.3	100.0%	100.0%	Atrial fibrillation, familial, 14, 615378
SCN3A	139.1	99.9%	98.8%	Epilepsy, familial focal, with variable foci 4, 617935 Epileptic encephalopathy, early infantile, 62, 617938
SCN3B	145.5	100.0%	100.0%	Brugada syndrome 7, 613120 Atrial fibrillation, familial, 16, 613120
SCN4A	182.1	99.9%	99.4%	Hyperkalemic periodic paralysis, type 2, 170500 Paramyotonia congenita, 168300 Myotonia congenita, atypical, acetazolamide-responsive, 608390 Myasthenic syndrome, congenital, 16, 614198 Hypokalemic periodic paralysis, type 2, 613345
SCN4B	68.2	99.9%	98.0%	Atrial fibrillation, familial, 17, 611819 Long QT syndrome 10, 611819
SCN5A	154.0	99.0%	99.0%	Atrial fibrillation, familial, 10, 614022 Sick sinus syndrome 1, 608567 Ventricular fibrillation, familial, 1, 603829 Long QT syndrome 3, 603830 Heart block, nonprogressive, 113900 Cardiomyopathy, dilated, 1E, 601154 Brugada syndrome 1, 601144 Heart block, progressive, type IA, 113900
SCN8A	162.9	100.0%	99.9%	Seizures, benign familial infantile, 5, 617080 Cognitive impairment with or without cerebellar ataxia, 614306 ?Myoclonus, familial, 2, 618364 Epileptic encephalopathy, early infantile, 13, 614558
SCN9A	128.2	99.0%	97.6%	Small fiber neuropathy, 133020 HSAN2D, autosomal recessive, 243000 Paroxysmal extreme pain disorder, 167400 Epilepsy, generalized, with febrile seizures plus, type 7, 613863 Insensitivity to pain, congenital, 243000 Erythermalgia, primary, 133020 Febrile seizures, familial, 3B, 613863
SCNN1A	140.2	99.9%	98.9%	Pseudohypoaldosteronism, type I, 264350 ?Liddle syndrome 3, 618126 Bronchiectasis with or without elevated sweat chloride 2, 613021

SCNN1B	138.7	100.0%	100.0%	Pseudohypoaldosteronism, type I, 264350 Liddle syndrome 1, 177200 Bronchiectasis with or without elevated sweat chloride 1, 211400
SCNN1G	152.3	99.7%	97.5%	Liddle syndrome 2, 618114 Pseudohypoaldosteronism, type I, 264350 Bronchiectasis with or without elevated sweat chloride 3, 613071
SCO1	105.2	100.0%	99.6%	Mitochondrial complex IV deficiency, 220110
SCO2	134.9	100.0%	100.0%	Myopia 6, 608908 Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377
SCP2	107.1	99.8%	96.8%	?Leukoencephalopathy with dystonia and motor neuropathy, 613724
SCYL1	161.4	100.0%	100.0%	Spinocerebellar ataxia, autosomal recessive 21, 616719
SDCCAG8	123.5	100.0%	99.7%	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SDHA	94.1	85.1%	78.0%	Leigh syndrome, 256000 Paragangliomas 5, 614165 Cardiomyopathy, dilated, 1GG, 613642 Mitochondrial respiratory chain complex II deficiency, 252011
SDHAF1	104.7	100.0%	100.0%	Mitochondrial complex II deficiency, 252011
SDHAF2	134.6	96.1%	94.6%	Paragangliomas 2, 601650
SDHB	116.1	100.0%	100.0%	Gastrointestinal stromal tumor, 606764 Pheochromocytoma, 171300 Paragangliomas 4, 115310 Paraganglioma and gastric stromal sarcoma, 606864
SDHC	87.8	99.8%	96.1%	Paragangliomas 3, 605373 Paraganglioma and gastric stromal sarcoma, 606864 Gastrointestinal stromal tumor, 606764
SDHD	45.8	53.1%	50.6%	Paragangliomas 1, with or without deafness, 168000 Mitochondrial complex II deficiency, 252011 Paraganglioma and gastric stromal sarcoma, 606864 Pheochromocytoma, 171300
SDR9C7	183.1	100.0%	100.0%	Ichthyosis, congenital, autosomal recessive 13, 617574
SEC23A	119.7	99.7%	97.6%	Craniolenticulosutural dysplasia, 607812
SEC23B	132.1	99.7%	98.2%	?Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SEC24D	125.7	100.0%	99.6%	Cole-Carpenter syndrome 2, 616294
SEC31A	116.6	98.9%	97.0%	?Neurodevelopmental disorder with spastic quadriplegia, optic atrophy, seizures, and structural brain anomalies, 618651
SEC61A1	125.6	100.0%	99.9%	Hyperuricemic nephropathy, familial juvenile, 4, 617056

SEC63	76.2	87.1%	79.3%	Polycystic liver disease 2, 617004
SECISBP2	109.6	99.5%	96.6%	Thyroid hormone metabolism, abnormal, 609698
SELENBP1	132.1	100.0%	99.9%	Extraoral halitosis due to MTO deficiency, 618148
SELENON	143.9	85.3%	84.0%	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310
SEMA3E	131.0	100.0%	99.7%	?CHARGE syndrome, 214800
SEMA4A	133.6	100.0%	99.5%	Cone-rod dystrophy 10, 610283 Retinitis pigmentosa 35, 610282
SEPSECS	160.6	100.0%	100.0%	Pontocerebellar hypoplasia type 2D, 613811
SEPT12	111.9	99.9%	98.6%	Spermatogenic failure 10, 614822
SEPT9	159.1	100.0%	99.9%	Amyotrophy, hereditary neuralgic, 162100
SERAC1	110.4	100.0%	99.0%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPINA1	113.2	100.0%	99.8%	Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490 Emphysema-cirrhosis, due to AAT deficiency, 613490 Emphysema due to AAT deficiency, 613490
SERPINA3	122.2	100.0%	99.9%	Cerebrovascular disease, occlusive, 0 Alpha-1-antichymotrypsin deficiency, 0
SERPINA6	146.0	100.0%	100.0%	Corticosteroid-binding globulin deficiency, 611489
SERPINB6	142.3	95.9%	95.9%	?Deafness, autosomal recessive 91, 613453
SERPINB7	125.1	100.0%	99.9%	Palmoplantar keratoderma, Nagashima type, 615598
SERPINB8	129.3	95.0%	95.0%	Peeling skin syndrome 5, 617115
SERPINC1	122.9	100.0%	100.0%	Thrombophilia due to antithrombin III deficiency, 613118
SERPIND1	139.0	100.0%	100.0%	Thrombophilia due to heparin cofactor II deficiency, 612356
SERPINE1	142.1	100.0%	99.9%	Plasminogen activator inhibitor-1 deficiency, 613329
SERPINF1	109.4	100.0%	99.8%	Osteogenesis imperfecta, type VI, 613982
SERPINF2	167.5	100.0%	100.0%	Alpha-2-plasmin inhibitor deficiency, 262850
SERPING1	101.0	99.6%	97.5%	Angioedema, hereditary, types I and II, 106100 Complement component 4, partial deficiency of, 120790
SERPINH1	216.7	100.0%	99.9%	Osteogenesis imperfecta, type X, 613848
SERPINI1	99.0	99.8%	97.6%	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218
SET	59.0	96.8%	89.5%	Mental retardation, autosomal dominant 58, 618106
SETBP1	129.6	99.1%	97.9%	Mental retardation, autosomal dominant 29, 616078 Schinzel-Giedion midface retraction syndrome, 269150
SETD2	139.1	100.0%	99.7%	Luscan-Lumish syndrome, 616831
SETD5	151.2	100.0%	99.7%	Mental retardation, autosomal dominant 23, 615761
SETX	153.0	100.0%	99.7%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 Amyotrophic lateral sclerosis 4, juvenile, 602433

SF3B1	126.8	99.6%	98.5%	Myelodysplastic syndrome, somatic, 614286
SF3B4	82.6	99.9%	98.5%	Acrofacial dysostosis 1, Nager type, 154400
SFRP4	136.0	100.0%	99.1%	Pyle disease, 265900
SFTPA2	151.4	100.0%	100.0%	Pulmonary fibrosis, idiopathic, 178500
SFTPB	101.4	100.0%	99.7%	Surfactant metabolism dysfunction, pulmonary, 1, 265120
SFTPC	104.8	100.0%	99.3%	Surfactant metabolism dysfunction, pulmonary, 2, 610913
SFXN4	126.1	100.0%	99.7%	Combined oxidative phosphorylation deficiency 18, 615578
SGCA	170.3	100.0%	100.0%	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099
SGCB	135.1	99.8%	97.9%	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286
SGCD	78.2	100.0%	98.2%	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287
SGCE	96.9	95.8%	91.7%	Dystonia-11, myoclonic, 159900
SGCG	117.7	100.0%	100.0%	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700
SGMS2	153.7	100.0%	100.0%	Calvarial doughnut lesions with bone fragility with or without spondylometaphyseal dysplasia, 126550
SGO1	107.1	99.9%	98.7%	Chronic atrial and intestinal dysrhythmia, 616201
SGPL1	133.4	100.0%	100.0%	Nephrotic syndrome, type 14, 617575
SGSH	152.5	98.1%	94.9%	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SH2B3	122.5	99.8%	98.8%	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950 Erythrocytosis, somatic, 133100
SH2D1A	108.5	95.7%	90.7%	Lymphoproliferative syndrome, X-linked, 1, 308240
SH3BP2	154.6	92.4%	91.4%	Cherubism, 118400
SH3KBP1	93.4	99.4%	94.3%	?Immunodeficiency 61, 300310
SH3PXD2B	175.9	100.0%	99.8%	Frank-ter Haar syndrome, 249420
SH3TC2	110.2	100.0%	99.6%	Charcot-Marie-Tooth disease, type 4C, 601596 Mononeuropathy of the median nerve, mild, 613353
SHANK3	143.0	98.1%	93.8%	Phelan-McDermid syndrome, 606232
SHH	165.7	100.0%	100.0%	Schizencephaly, 269160 Microphthalmia with coloboma 5, 611638 Single median maxillary central incisor, 147250 Holoprosencephaly 3, 142945
SHOC2	136.8	100.0%	99.4%	Noonan syndrome-like with loose anagen hair, 607721
SHOX	40.6	85.4%	69.6%	Langer mesomelic dysplasia, 249700 Short stature, idiopathic familial, 300582 Leri-Weill dyschondrosteosis, 127300
SHROOM4	104.6	99.9%	98.9%	Stocco dos Santos X-linked mental retardation syndrome, 300434
SI	112.1	99.4%	95.3%	Sucrase-isomaltase deficiency, congenital, 222900

SIGMAR1	162.1	100.0%	100.0%	?Amyotrophic lateral sclerosis 16, juvenile, 614373 ?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726
SIK1	131.1	99.8%	98.1%	Epileptic encephalopathy, early infantile, 30, 616341
SIK3	104.5	100.0%	99.1%	?Spondyloepimetaphyseal dysplasia, Krakow type, 618162
SIL1	138.5	99.4%	96.7%	Marinesco-Sjogren syndrome, 248800
SIN3A	111.6	99.9%	98.7%	Witteveen-Kolk syndrome, 613406
SIPA1L3	192.1	100.0%	99.8%	?Cataract 45, 616851
SIX1	150.4	99.9%	99.2%	Deafness, autosomal dominant 23, 605192 Branchiootic syndrome 3, 608389
SIX3	240.1	100.0%	100.0%	Holoprosencephaly 2, 157170 Schizencephaly, 269160
SIX5	87.2	100.0%	98.9%	Branchiootorenal syndrome 2, 610896
SIX6	303.9	100.0%	100.0%	Optic disc anomalies with retinal and/or macular dystrophy, 212550
SKI	149.5	100.0%	99.7%	Shprintzen-Goldberg syndrome, 182212
SKIV2L	150.1	100.0%	99.9%	Trichohepatoenteric syndrome 2, 614602
SLC10A2	124.2	100.0%	99.9%	Bile acid malabsorption, primary, 613291
SLC10A7	110.2	99.9%	99.1%	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363
SLC11A2	97.4	99.9%	98.5%	Anemia, hypochromic microcytic, with iron overload 1, 206100
SLC12A1	144.9	100.0%	99.7%	Bartter syndrome, type 1, 601678
SLC12A3	151.1	100.0%	100.0%	Gitelman syndrome, 263800
SLC12A5	121.0	86.3%	84.2%	Epileptic encephalopathy, early infantile, 34, 616645
SLC12A6	120.5	100.0%	100.0%	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC13A3	94.6	100.0%	99.8%	Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate, 618384
SLC13A5	155.1	100.0%	99.9%	Epileptic encephalopathy, early infantile, 25, 615905
SLC16A1	140.0	100.0%	99.3%	Monocarboxylate transporter 1 deficiency, 616095 Hyperinsulinemic hypoglycemia, familial, 7, 610021 Erythrocyte lactate transporter defect, 245340
SLC16A12	134.4	100.0%	99.9%	Cataract 47, juvenile, with microcornea, 612018
SLC16A2	67.9	99.1%	93.5%	Allan-Herndon-Dudley syndrome, 300523
SLC17A5	136.7	98.7%	95.1%	Sialic acid storage disorder, infantile, 269920 Salla disease, 604369
SLC17A8	123.8	100.0%	100.0%	Deafness, autosomal dominant 25, 605583
SLC17A9	155.8	95.8%	95.4%	Porokeratosis 8, disseminated superficial actinic type, 616063
SLC18A2	112.3	100.0%	99.9%	?Parkinsonism-dystonia, infantile, 2, 618049
SLC18A3	301.6	100.0%	100.0%	Myasthenic syndrome, congenital, 21, presynaptic, 617239
SLC19A2	103.2	100.0%	99.7%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	139.2	100.0%	99.9%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483

SLC1A1	149.2	100.0%	99.7%	Dicarboxylic aminoaciduria, 222730
SLC1A2	100.8	99.5%	97.2%	Epileptic encephalopathy, early infantile, 41, 617105
SLC1A3	103.0	100.0%	99.8%	Episodic ataxia, type 6, 612656
SLC1A4	159.5	100.0%	99.8%	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC20A2	117.5	99.9%	98.5%	Basal ganglia calcification, idiopathic, 1, 213600
SLC22A12	131.0	100.0%	99.9%	Hypouricemia, renal, 220150
SLC22A18	125.0	100.0%	99.9%	Lung cancer, somatic, 211980 Breast cancer, somatic, 114480 Rhabdomyosarcoma, somatic, 268210
SLC22A5	144.8	100.0%	100.0%	Carnitine deficiency, systemic primary, 212140
SLC24A1	175.4	100.0%	100.0%	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
SLC24A4	109.6	100.0%	99.9%	Amelogenesis imperfecta, type IIA5, 615887
SLC24A5	102.2	100.0%	99.6%	Albinism, oculocutaneous, type VI, 113750
SLC25A1	114.2	99.8%	97.0%	?Myasthenic syndrome, congenital, 23, presynaptic, 618197 Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A11	189.1	100.0%	100.0%	Paragangliomas 6, 618464
SLC25A12	151.7	100.0%	99.7%	Epileptic encephalopathy, early infantile, 39, 612949
SLC25A13	118.2	99.9%	98.1%	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A15	152.1	98.4%	94.4%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A19	82.0	100.0%	98.5%	Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710 Microcephaly, Amish type, 607196
SLC25A20	96.7	100.0%	99.9%	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A22	138.5	100.0%	99.7%	Epileptic encephalopathy, early infantile, 3, 609304
SLC25A24	133.1	99.7%	99.5%	Fontaine progeroid syndrome, 612289
SLC25A26	102.0	99.8%	98.9%	Combined oxidative phosphorylation deficiency 28, 616794
SLC25A3	140.1	99.8%	97.0%	Mitochondrial phosphate carrier deficiency, 610773
SLC25A32	128.5	100.0%	100.0%	?Exercise intolerance, riboflavin-responsive, 616839
SLC25A38	98.5	99.0%	95.5%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC25A4	141.9	100.0%	99.9%	Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283
SLC25A42	143.7	100.0%	99.2%	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416
SLC25A46	175.2	99.8%	97.2%	Neuropathy, hereditary motor and sensory, type VIB, 616505
SLC26A1	171.5	100.0%	100.0%	?Nephrolithiasis, calcium oxalate, 167030
SLC26A2	203.7	100.0%	99.9%	De la Chapelle dysplasia, 256050 Atelosteogenesis, type II, 256050

				Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 Diastrophic dysplasia, 222600 Achondrogenesis Ib, 600972 Epiphyseal dysplasia, multiple, 4, 226900
SLC26A3	133.1	100.0%	99.1%	Diarrhea 1, secretory chloride, congenital, 214700
SLC26A4	115.6	100.0%	99.7%	Pendred syndrome, 274600 Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791
SLC26A5	132.5	99.2%	96.0%	?Deafness, autosomal recessive 61, 613865
SLC26A8	115.3	100.0%	99.6%	Spermatogenic failure 3, 606766
SLC27A4	167.0	100.0%	100.0%	Ichthyosis prematurity syndrome, 608649
SLC29A3	190.1	100.0%	99.7%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC2A1	160.0	92.8%	92.8%	Dystonia 9, 601042 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 GLUT1 deficiency syndrome 2, childhood onset, 612126
SLC2A10	167.7	98.0%	97.6%	Arterial tortuosity syndrome, 208050
SLC2A2	159.5	100.0%	99.9%	Fanconi-Bickel syndrome, 227810
SLC2A9	108.3	100.0%	98.9%	Hypouricemia, renal, 2, 612076
SLC30A10	200.7	100.0%	100.0%	Hypermanganesemia with dystonia 1, 613280
SLC30A2	131.1	100.0%	99.5%	Zinc deficiency, transient neonatal, 608118
SLC30A9	86.6	98.0%	91.6%	?Birk-Landau-Perez syndrome, 617595
SLC33A1	135.7	99.8%	97.0%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC34A1	166.0	100.0%	99.9%	Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286 Hypercalcemia, infantile, 2, 616963 ?Fanconi renotubular syndrome 2, 613388
SLC34A2	150.2	100.0%	99.9%	Pulmonary alveolar microlithiasis, 265100
SLC34A3	165.6	100.0%	99.5%	Hypophosphatemic rickets with hypercalciuria, 241530
SLC35A1	123.5	100.0%	99.8%	Congenital disorder of glycosylation, type IIf, 603585
SLC35A2	114.2	100.0%	99.1%	Congenital disorder of glycosylation, type IIm, 300896
SLC35A3	63.5	80.4%	77.0%	?Arthrogryposis, mental retardation, and seizures, 615553
SLC35C1	209.1	100.0%	99.9%	Congenital disorder of glycosylation, type IIc, 266265
SLC35D1	126.1	99.6%	95.7%	Schneckenbecken dysplasia, 269250
SLC36A2	104.3	100.0%	99.9%	Iminoglycinuria, digenic, 242600 Hyperglycinuria, 138500
SLC37A4	122.0	100.0%	99.7%	Glycogen storage disease Ic, 232240 Glycogen storage disease Ib, 232220

SLC38A8	77.1	99.3%	95.7%	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218
SLC39A13	158.7	100.0%	99.9%	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350
SLC39A14	101.7	99.9%	98.8%	?Hyperostosis cranalis interna, 144755 Hypermanganesemia with dystonia 2, 617013
SLC39A4	130.4	100.0%	99.7%	Acrodermatitis enteropathica, 201100
SLC39A5	143.6	100.0%	99.7%	Myopia 24, autosomal dominant, 615946
SLC39A8	144.7	100.0%	99.8%	Congenital disorder of glycosylation, type IIn, 616721
SLC3A1	147.7	100.0%	99.7%	Cystinuria, 220100
SLC40A1	121.3	100.0%	99.9%	Hemochromatosis, type 4, 606069
SLC44A4	122.2	100.0%	99.5%	?Deafness, autosomal dominant 72, 617606
SLC45A1	154.0	100.0%	100.0%	Intellectual developmental disorder with neuropsychiatric features, 617532
SLC45A2	119.4	100.0%	99.8%	Albinism, oculocutaneous, type IV, 606574
SLC46A1	121.7	100.0%	98.0%	Folate malabsorption, hereditary, 229050
SLC4A1	151.7	100.0%	100.0%	Cryohydrocytosis, 185020 Spherocytosis, type 4, 612653 Ovalocytosis, SA type, 166900 Renal tubular acidosis, distal, AD, 179800 Renal tubular acidosis, distal, AR, 611590
SLC4A11	173.6	100.0%	100.0%	Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy, autosomal recessive, 217700 Corneal endothelial dystrophy and perceptive deafness, 217400
SLC4A4	114.4	99.8%	97.9%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC52A1	219.5	100.0%	100.0%	Riboflavin deficiency, 615026
SLC52A2	213.2	100.0%	100.0%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	128.9	100.0%	99.9%	Brown-Vialetto-Van Laere syndrome 1, 211530 ?Fazio-Londe disease, 211500
SLC5A1	116.9	100.0%	99.5%	Glucose/galactose malabsorption, 606824
SLC5A2	150.5	100.0%	100.0%	Renal glucosuria, 233100
SLC5A5	118.2	100.0%	100.0%	Thyroid dyshormonogenesis 1, 274400
SLC5A7	102.5	100.0%	99.9%	Neuronopathy, distal hereditary motor, type VIIA, 158580 Myasthenic syndrome, congenital, 20, presynaptic, 617143
SLC6A1	136.0	100.0%	100.0%	Myoclonic-atonic epilepsy, 616421
SLC6A17	162.2	100.0%	100.0%	Mental retardation, autosomal recessive 48, 616269
SLC6A19	139.1	100.0%	100.0%	Iminoglycinuria, digenic, 242600 Hartnup disorder, 234500 Hyperglycinuria, 138500
SLC6A2	131.1	100.0%	99.9%	?Orthostatic intolerance, 604715

SLC6A20	164.1	100.0%	99.9%	Iminoglycinuria, digenic, 242600 Hyperglycinuria, 138500
SLC6A3	142.7	100.0%	99.9%	Parkinsonism-dystonia, infantile, 1, 613135
SLC6A5	137.1	100.0%	99.9%	Hyperekplexia 3, 614618
SLC6A8	58.5	97.6%	87.8%	Cerebral creatine deficiency syndrome 1, 300352
SLC6A9	164.3	100.0%	100.0%	Glycine encephalopathy with normal serum glycine, 617301
SLC7A14	155.0	100.0%	100.0%	Retinitis pigmentosa 68, 615725
SLC7A7	110.7	100.0%	99.8%	Lysinuric protein intolerance, 222700
SLC7A9	126.6	100.0%	99.3%	Cystinuria, 220100
SLC9A1	157.0	100.0%	100.0%	?Lichtenstein-Knorr syndrome, 616291
SLC9A3	181.6	100.0%	99.8%	Diarrhea 8, secretory sodium, congenital, 616868
SLC9A3R1	159.7	100.0%	100.0%	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
SLC9A6	104.0	98.5%	93.3%	Mental retardation, X-linked syndromic, Christianson type, 300243
SLC9A7	87.8	98.1%	92.1%	Intellectual developmental disorder, X-linked 108, 301024
SLCO1B1	47.9	97.4%	88.7%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO1B3	47.0	96.7%	86.6%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO2A1	104.4	100.0%	98.7%	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
SLFN14	174.8	100.0%	100.0%	Bleeding disorder, platelet-type, 20, 616913
SLITRK1	140.8	100.0%	100.0%	?Trichotillomania, 613229 Tourette syndrome, 137580
SLITRK6	170.3	100.0%	100.0%	Deafness and myopia, 221200
SLURP1	109.0	100.0%	99.6%	Meleda disease, 248300
SLX4	136.8	100.0%	99.9%	Fanconi anemia, complementation group P, 613951
SMAD3	138.0	100.0%	100.0%	Loeys-Dietz syndrome 3, 613795
SMAD4	109.9	100.0%	99.9%	Polyposis, juvenile intestinal, 174900 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050
SMAD6	209.5	99.9%	95.6%	Aortic valve disease 2, 614823
SMAD9	117.2	100.0%	99.8%	Pulmonary hypertension, primary, 2, 615342
SMARCA2	109.3	97.3%	96.3%	Nicolaides-Baraitser syndrome, 601358
SMARCA4	163.9	100.0%	99.6%	Coffin-Siris syndrome 4, 614609
SMARCAD1	89.5	99.6%	95.8%	Huriez syndrome, 181600 Basan syndrome, 129200 Adermatoglyphia, 136000
SMARCAL1	119.6	100.0%	99.8%	Schimke immunoosseous dysplasia, 242900

SMARCB1	192.9	100.0%	100.0%	Rhabdoid tumors, somatic, 609322 Coffin-Siris syndrome 3, 614608
SMARCC2	104.2	99.9%	98.7%	Coffin-Siris syndrome 8, 618362
SMARCD2	99.5	87.5%	86.1%	Specific granule deficiency 2, 617475
SMARCE1	66.9	94.6%	85.4%	Coffin-Siris syndrome 5, 616938
SMC1A	93.2	99.9%	98.3%	Cornelia de Lange syndrome 2, 300590
SMC3	82.8	96.4%	89.7%	Cornelia de Lange syndrome 3, 610759
SMCHD1	95.2	99.6%	97.4%	Fascioscapulohumeral muscular dystrophy 2, digenic, 158901 Bosma arhinia microphthalmia syndrome, 603457
SMG9	100.1	100.0%	100.0%	Heart and brain malformation syndrome, 616920
SMN1	89.1	99.7%	97.7%	Spinal muscular atrophy-2, 253550 Spinal muscular atrophy-3, 253400 Spinal muscular atrophy-1, 253300 Spinal muscular atrophy-4, 271150
SMO	154.2	100.0%	99.5%	Curry-Jones syndrome, somatic mosaic, 601707 Basal cell carcinoma, somatic, 605462
SMOC1	121.2	99.9%	98.4%	Microphthalmia with limb anomalies, 206920
SMOC2	93.7	76.9%	75.9%	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SMPD1	161.8	100.0%	99.6%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SMPD4	108.3	99.9%	96.9%	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622
SMPX	64.6	100.0%	96.6%	Deafness, X-linked 4, 300066
SMS	63.5	88.9%	74.1%	Mental retardation, X-linked, Snyder-Robinson type, 309583
SNAI2	106.3	100.0%	99.1%	Waardenburg syndrome, type 2D, 608890 Piebaldism, 172800
SNAP25	121.8	100.0%	99.7%	?Myasthenic syndrome, congenital, 18, 616330
SNAP29	182.5	100.0%	100.0%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNCA	102.9	100.0%	100.0%	Dementia, Lewy body, 127750 Parkinson disease 1, 168601 Parkinson disease 4, 605543
SNCB	109.5	100.0%	99.9%	Dementia, Lewy body, 127750
SNIP1	140.1	100.0%	99.6%	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501
SNORD118	NC	NC	NC	Leukoencephalopathy, brain calcifications, and cysts, 614561
SNRNP200	124.8	99.9%	98.7%	Retinitis pigmentosa 33, 610359
SNRPB	83.6	99.9%	98.8%	Cerebrocostomandibular syndrome, 117650
SNRPE	74.7	99.0%	87.8%	Hypotrichosis 11, 615059
SNRPN	98.3	100.0%	98.4%	Prader-Willi syndrome, 176270

SNTA1	103.0	99.0%	92.1%	Long QT syndrome 12, 612955
SNX10	124.3	96.2%	95.5%	Osteopetrosis, autosomal recessive 8, 615085
SNX14	79.9	99.7%	93.7%	Spinocerebellar ataxia, autosomal recessive 20, 616354
SOBP	199.7	98.9%	98.8%	Mental retardation, anterior maxillary protrusion, and strabismus, 613671
SOD1	129.8	100.0%	99.7%	Amyotrophic lateral sclerosis 1, 105400 Spastic tetraplegia and axial hypotonia, progressive, 618598
SOHLH1	111.4	100.0%	98.6%	Ovarian dysgenesis 5, 617690 Spermatogenic failure 32, 618115
SON	134.3	99.2%	95.4%	ZTTK syndrome, 617140
SOS1	100.6	99.7%	96.7%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOS2	98.0	99.7%	97.7%	Noonan syndrome 9, 616559
SOST	207.3	100.0%	99.5%	Sclerosteosis 1, 269500 Van Buchem disease, 239100 Craniodiaphyseal dysplasia, autosomal dominant, 122860
SOX10	101.7	100.0%	99.8%	Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 PCWH syndrome, 609136 Waardenburg syndrome, type 4C, 613266
SOX11	225.3	100.0%	100.0%	Coffin-Siris syndrome 9, 615866
SOX17	147.6	100.0%	100.0%	Vesicoureteral reflux 3, 613674
SOX18	59.1	97.4%	85.4%	Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940 Hypotrichosis-lymphedema-telangiectasia syndrome, 607823
SOX2	261.8	100.0%	100.0%	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SOX3	87.5	99.0%	95.2%	Panhypopituitarism, X-linked, 312000 Mental retardation, X-linked, with isolated growth hormone deficiency, 300123
SOX4	122.0	100.0%	99.9%	Coffin-Siris syndrome 10, 618506
SOX5	92.0	99.8%	96.6%	Lamb-Shaffer syndrome, 616803
SOX9	181.5	100.0%	100.0%	Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290 Campomelic dysplasia with autosomal sex reversal, 114290
SP110	112.3	100.0%	99.9%	Hepatic venoocclusive disease with immunodeficiency, 235550
SP7	164.6	100.0%	99.5%	Osteogenesis imperfecta, type XII, 613849
SPAG1	102.2	98.7%	93.2%	Ciliary dyskinesia, primary, 28, 615505
SPARC	144.6	100.0%	100.0%	Osteogenesis imperfecta, type XVII, 616507
SPART	132.9	100.0%	98.4%	Troyer syndrome, 275900
SPAST	94.9	99.7%	96.9%	Spastic paraplegia 4, autosomal dominant, 182601

SPATA16	135.6	100.0%	99.2%	?Spermatogenic failure 6, 102530
SPATA5	142.5	100.0%	99.8%	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPATA7	120.5	99.7%	97.1%	Retinitis pigmentosa, juvenile, autosomal recessive, 604232 Leber congenital amaurosis 3, 604232
SPECC1L	133.2	100.0%	99.7%	Hypertelorism, Teebi type, 145420 ?Facial clefting, oblique, 1, 600251 Opitz GBBB syndrome, type II, 145410
SPEG	145.0	99.5%	98.1%	Centronuclear myopathy 5, 615959
SPG11	118.8	99.9%	98.5%	Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360 Amyotrophic lateral sclerosis 5, juvenile, 602099
SPG21	120.3	99.6%	97.2%	Mast syndrome, 248900
SPG7	123.7	99.8%	97.8%	Spastic paraplegia 7, autosomal recessive, 607259
SPINK1	86.2	100.0%	100.0%	Pancreatitis, hereditary, 167800 Tropical calcific pancreatitis, 608189
SPINK2	109.2	99.3%	99.2%	?Spermatogenic failure 29, 618091
SPINK5	128.1	100.0%	99.2%	Netherton syndrome, 256500
SPINT2	71.9	99.8%	93.0%	Diarrhea 3, secretory sodium, congenital, syndromic, 270420
SPNS2	157.7	97.5%	94.0%	?Deafness, autosomal recessive 115, 618457
SPR	159.7	100.0%	100.0%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRED1	143.0	99.8%	98.1%	Legius syndrome, 611431
SPRTN	164.4	100.0%	100.0%	Ruijs-Aalfs syndrome, 616200
SPRY4	187.2	100.0%	100.0%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
SPTA1	107.6	99.9%	98.8%	Pyropoikilocytosis, 266140 Elliptocytosis-2, 130600 Spherocytosis, type 3, 270970
SPTAN1	118.5	99.1%	98.6%	Epileptic encephalopathy, early infantile, 5, 613477
SPTB	155.6	100.0%	100.0%	Elliptocytosis-3, 617948 Spherocytosis, type 2, 616649 Anemia, neonatal hemolytic, fatal or near-fatal, 617948
SPTBN2	141.6	100.0%	99.9%	Spinocerebellar ataxia, autosomal recessive 14, 615386 Spinocerebellar ataxia 5, 600224
SPTBN4	117.1	99.9%	99.0%	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519
SPTLC1	108.0	98.4%	91.8%	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	146.3	100.0%	100.0%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SQSTM1	129.7	100.0%	99.6%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145

				Myopathy, distal, with rimmed vacuoles, 617158 Paget disease of bone 3, 167250
SRC	135.9	100.0%	99.8%	Colon cancer, advanced, somatic, 114500 ?Thrombocytopenia 6, 616937
SRCAP	166.7	100.0%	99.8%	Floating-Harbor syndrome, 136140
SRD5A2	92.3	100.0%	97.6%	Pseudovaginal perineoscrotal hypospadias, 264600
SRD5A3	149.2	99.9%	98.5%	Kahrizi syndrome, 612713 Congenital disorder of glycosylation, type Iq, 612379
SRP72	69.4	95.7%	84.1%	Bone marrow failure syndrome 1, 614675
SRPX2	64.2	99.6%	94.4%	?Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643
SRY	31.7	50.0%	50.0%	46XY sex reversal 1, 400044
SSR4	118.5	100.0%	100.0%	Congenital disorder of glycosylation, type Iy, 300934
SSTR5	203.5	100.0%	100.0%	Somatostatin analog, resistance to, 0
SSX1	89.4	82.0%	79.6%	?Sarcoma, synovial, 300813
SSX2	60.7	62.3%	57.7%	?Sarcoma, synovial, 300813
ST14	170.8	100.0%	100.0%	Ichthyosis, congenital, autosomal recessive 11, 602400
ST3GAL3	143.4	100.0%	99.8%	Mental retardation, autosomal recessive 12, 611090 ?Epileptic encephalopathy, early infantile, 15, 615006
ST3GAL5	104.4	89.3%	85.5%	Salt and pepper developmental regression syndrome, 609056
STAC3	122.8	100.0%	100.0%	Myopathy, congenital, Baily-Bloch, 255995
STAG1	108.3	99.8%	96.9%	Mental retardation, autosomal dominant 47, 617635
STAG2	72.0	96.8%	86.9%	Mullegama-Klein-Martinez syndrome, 301022
STAG3	105.4	93.5%	92.8%	Premature ovarian failure 8, 615723
STAMBP	96.0	99.9%	97.6%	Microcephaly-capillary malformation syndrome, 614261
STAR	146.4	100.0%	100.0%	Lipoid adrenal hyperplasia, 201710
STARD7	105.4	100.0%	98.9%	Epilepsy, familial adult myoclonic, 2, 607876
STAT1	116.6	99.2%	97.2%	Immunodeficiency 31C, autosomal dominant, 614162 Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796
STAT2	117.1	100.0%	99.9%	Immunodeficiency 44, 616636
STAT3	106.9	100.0%	99.4%	Hyper-IgE recurrent infection syndrome, 147060 Autoimmune disease, multisystem, infantile-onset, 1, 615952
STAT5B	119.7	99.9%	98.8%	Leukemia, acute promyelocytic, somatic, 102578 Growth hormone insensitivity with immunodeficiency, 245590
STEAP3	185.9	100.0%	99.6%	?Anemia, hypochromic microcytic, with iron overload 2, 615234
STIL	153.2	100.0%	99.7%	Microcephaly 7, primary, autosomal recessive, 612703

STIM1	129.2	99.8%	97.1%	Myopathy, tubular aggregate, 1, 160565 Immunodeficiency 10, 612783 Stormorken syndrome, 185070
STK11	142.7	100.0%	100.0%	Testicular tumor, somatic, 273300 Peutz-Jeghers syndrome, 175200 Pancreatic cancer, somatic, 260350 Melanoma, malignant, somatic, 0
STK4	124.5	100.0%	99.7%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STN1	82.8	100.0%	99.6%	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341
STOX1	149.6	89.7%	89.6%	Preeclampsia/eclampsia 4, 609404
STRA6	125.5	100.0%	99.9%	Microphthalmia, syndromic 9, 601186 Microphthalmia, isolated, with coloboma 8, 601186
STRADA	112.5	100.0%	99.5%	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087
STRC	109.0	100.0%	99.1%	Deafness, autosomal recessive 16, 603720
STS	81.1	99.6%	96.3%	Ichthyosis, X-linked, 308100
STT3A	125.4	100.0%	99.9%	?Congenital disorder of glycosylation, type Iw, 615596
STT3B	127.0	100.0%	99.8%	?Congenital disorder of glycosylation, type Ix, 615597
STUB1	193.3	100.0%	99.8%	Spinocerebellar ataxia, autosomal recessive 16, 615768 ?Spinocerebellar ataxia 48, 618093
STX11	330.7	100.0%	100.0%	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STX16	116.6	99.9%	99.0%	Pseudohypoparathyroidism, type IB, 603233
STX1B	173.7	100.0%	100.0%	Generalized epilepsy with febrile seizures plus, type 9, 616172
STXBP1	108.2	96.8%	96.5%	Epileptic encephalopathy, early infantile, 4, 612164
STXBP2	110.2	84.1%	80.8%	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
SUCLA2	57.8	91.5%	82.6%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	104.2	100.0%	99.7%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUFU	141.7	100.0%	100.0%	Basal cell nevus syndrome, 109400 Medulloblastoma, desmoplastic, 155255 Joubert syndrome 32, 617757
SUGCT	126.9	99.0%	94.8%	Glutaric aciduria III, 231690
SULT2B1	135.3	100.0%	100.0%	Ichthyosis, congenital, autosomal recessive 14, 617571
SUMF1	91.7	99.9%	97.6%	Multiple sulfatase deficiency, 272200
SUMO1	18.5	61.9%	40.9%	?Orofacial cleft 10, 613705
SUN5	103.5	100.0%	99.9%	Spermatogenic failure 16, 617187
SUOX	180.8	100.0%	100.0%	Sulfite oxidase deficiency, 272300

SURF1	89.9	93.5%	89.1%	Leigh syndrome, due to COX IV deficiency, 256000 Charcot-Marie-Tooth disease, type 4K, 616684
SVBP	114.3	100.0%	100.0%	Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569
SYCE1	124.9	99.9%	98.2%	?Premature ovarian failure 12, 616947 ?Spermatogenic failure 15, 616950
SYCP3	82.9	99.8%	98.0%	Spermatogenic failure 4, 270960 Pregnancy loss, recurrent, 4, 270960
SYN1	73.1	93.6%	84.0%	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNE1	123.5	98.3%	97.8%	Spinocerebellar ataxia, autosomal recessive 8, 610743 Arthrogryposis multiplex congenita, myogenic type, 618484 Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998
SYNE2	112.1	99.6%	97.9%	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999
SYNE4	92.6	100.0%	99.2%	Deafness, autosomal recessive 76, 615540
SYNGAP1	152.9	98.5%	98.0%	Mental retardation, autosomal dominant 5, 612621
SYNJ1	126.3	99.9%	98.5%	Epileptic encephalopathy, early infantile, 53, 617389 Parkinson disease 20, early-onset, 615530
SYP	87.1	99.9%	98.6%	Mental retardation, X-linked 96, 300802
SYT1	153.0	100.0%	99.1%	Baker-Gordon syndrome, 618218
SYT14	103.1	60.6%	58.1%	?Spinocerebellar ataxia, autosomal recessive 11, 614229
SYT2	100.4	99.9%	98.6%	Myasthenic syndrome, congenital, 7, presynaptic, 616040
SZT2	146.3	99.6%	99.5%	Epileptic encephalopathy, early infantile, 18, 615476
T	158.5	99.6%	97.2%	Sacral agenesis with vertebral anomalies, 615709
TAB2	171.5	100.0%	99.7%	Congenital heart defects, nonsyndromic, 2, 614980
TAC3	63.9	99.9%	95.4%	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACO1	103.0	99.9%	95.8%	Mitochondrial complex IV deficiency, 220110
TACR3	153.6	100.0%	99.6%	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
TACSTD2	314.2	100.0%	100.0%	Corneal dystrophy, gelatinous drop-like, 204870
TAF1	89.0	99.4%	96.2%	Dystonia-Parkinsonism, X-linked, 314250 Mental retardation, X-linked, syndromic 33, 300966
TAF13	96.3	100.0%	100.0%	Mental retardation, autosomal recessive 60, 617432
TAF2	110.2	99.8%	98.1%	Mental retardation, autosomal recessive 40, 615599
TAF4B	127.3	99.2%	94.8%	?Spermatogenic failure 13, 615841
TAF6	138.7	100.0%	99.5%	Alazami-Yuan syndrome, 617126
TAL1	67.5	96.5%	88.3%	Leukemia, T-cell acute lymphocytic, somatic, 613065
TAL2	114.5	100.0%	100.0%	Leukemia, T-cell acute lymphocytic, somatic, 613065
TALDO1	158.9	100.0%	99.8%	Transaldolase deficiency, 606003

TANGO2	139.6	100.0%	100.0%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TAP1	133.9	99.9%	97.7%	Bare lymphocyte syndrome, type I, 604571
TAP2	101.4	99.6%	98.7%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	130.6	96.6%	96.6%	Bare lymphocyte syndrome, type I, 604571
TAPT1	87.2	99.3%	93.9%	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897
TARDBP	129.8	100.0%	100.0%	Frontotemporal lobar degeneration, TARDBP-related, 612069 Amyotrophic lateral sclerosis 10, with or without FTD, 612069
TARS	109.6	99.8%	97.8%	Trichothiodystrophy 7, nonphotosensitive, 618546
TARS2	93.9	99.7%	97.1%	?Combined oxidative phosphorylation deficiency 21, 615918
TAT	119.7	100.0%	99.9%	Tyrosinemia, type II, 276600
TAZ	125.3	99.3%	96.2%	Barth syndrome, 302060
TBC1D20	121.0	97.4%	94.5%	Warburg micro syndrome 4, 615663
TBC1D23	89.8	98.8%	94.7%	Pontocerebellar hypoplasia, type 11, 617695
TBC1D24	199.9	100.0%	100.0%	Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp, 608105 DOORS syndrome, 220500 Deafness, autosomal dominant 65, 616044 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021 Deafness , autosomal recessive 86, 614617
TBC1D7	99.0	99.9%	99.0%	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000
TBC1D8B	90.8	98.2%	92.5%	Nephrotic syndrome, type 20, 301028
TBCD	145.8	98.8%	95.5%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TBCE	117.0	99.3%	95.6%	Kenny-Caffey syndrome, type 1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
TBCK	97.5	99.2%	94.9%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900
TBK1	98.9	99.8%	97.4%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439
TBL1X	117.5	95.9%	90.1%	Hypothyroidism, congenital, nongoitrous, 8, 301033
TBL1XR1	61.2	93.8%	78.4%	Pierpont syndrome, 602342 Mental retardation, autosomal dominant 41, 616944
TBL1Y	39.4	49.3%	45.7%	?Deafness, Y-linked 2, 400047
TBP	103.4	100.0%	99.8%	Spinocerebellar ataxia 17, 607136
TBR1	184.5	100.0%	100.0%	Intellectual developmental disorder with autism and speech delay, 606053
TBX1	114.2	93.7%	88.3%	Velocardiofacial syndrome, 192430 DiGeorge syndrome, 188400

				Tetralogy of Fallot, 187500 Conotruncal anomaly face syndrome, 217095
TBX15	114.2	100.0%	100.0%	Cousin syndrome, 260660
TBX18	111.5	99.9%	98.4%	Congenital anomalies of kidney and urinary tract 2, 143400
TBX19	168.3	100.0%	100.0%	Adrenocorticotropic hormone deficiency, 201400
TBX2	184.1	100.0%	100.0%	Vertebral anomalies and variable endocrine and T-cell dysfunction, 618223
TBX20	112.3	100.0%	100.0%	Atrial septal defect 4, 611363
TBX21	122.6	100.0%	99.7%	Asthma and nasal polyps, 208550
TBX22	104.3	99.0%	94.5%	?Abruzzo-Erickson syndrome, 302905 Cleft palate with ankyloglossia, 303400
TBX3	109.8	99.9%	98.7%	Ulnar-mammary syndrome, 181450
TBX4	187.7	99.8%	98.2%	Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension, 147891
TBX5	145.6	100.0%	100.0%	Holt-Oram syndrome, 142900
TBX6	135.0	99.8%	97.5%	Spondylocostal dysostosis 5, 122600
TBXAS1	135.5	100.0%	100.0%	Ghosal hematodiaphyseal syndrome, 231095
TCAP	113.4	100.0%	100.0%	Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954
TCF12	137.8	100.0%	99.9%	Craniosynostosis 3, 615314
TCF20	134.1	100.0%	100.0%	Developmental delay with variable intellectual impairment and behavioral abnormalities, 618430
TCF3	100.2	99.4%	96.9%	Agammaglobulinemia 8, autosomal dominant, 616941
TCF4	111.6	100.0%	99.8%	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954
TCHH	183.8	100.0%	100.0%	?Uncombable hair syndrome 3, 617252
TCIRG1	149.6	99.6%	98.0%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	157.6	100.0%	100.0%	Transcobalamin II deficiency, 275350
TCOF1	119.1	99.9%	99.3%	Treacher Collins syndrome 1, 154500
TCTEX1D2	126.1	99.9%	99.0%	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405
TCTN1	96.0	95.7%	92.6%	Joubert syndrome 13, 614173
TCTN2	127.0	100.0%	99.0%	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	121.0	100.0%	100.0%	Orofaciodigital syndrome IV, 258860 Joubert syndrome 18, 614815
TDGF1	127.1	99.8%	95.5%	Forebrain defects, 0
TDP1	105.7	100.0%	99.5%	?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250
TDP2	175.8	100.0%	99.9%	Spinocerebellar ataxia, autosomal recessive 23, 616949
TDRD7	139.3	99.9%	99.0%	Cataract 36, 613887
TDRD9	114.8	100.0%	98.9%	?Spermatogenic failure 30, 618110

TEAD1	136.1	100.0%	99.7%	Sveinsson chorioretinal atrophy, 108985
TECPR2	147.6	100.0%	100.0%	Spastic paraplegia 49, autosomal recessive, 615031
TECR	139.6	100.0%	99.9%	Mental retardation, autosomal recessive 14, 614020
TECRL	71.0	96.0%	87.5%	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021
TECTA	176.7	100.0%	99.9%	Deafness, autosomal dominant 8/12, 601543 Deafness, autosomal recessive 21, 603629
TEK	151.2	100.0%	99.8%	Glaucoma 3, primary congenital, E, 617272 Venous malformations, multiple cutaneous and mucosal, 600195
TELO2	136.2	99.9%	98.7%	You-Hoover-Fong syndrome, 616954
TENM3	155.3	99.8%	99.4%	Microphthalmia, syndromic 15, 615145 ?Microphthalmia, isolated, with coloboma 9, 615145
TENM4	128.6	100.0%	99.8%	Essential tremor, hereditary, 5, 616736
TERC	NC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550
TET2	173.2	100.0%	100.0%	Myelodysplastic syndrome, somatic, 614286
TEX11	71.3	93.1%	83.7%	Spermatogenic failure, X-linked, 2, 309120
TEX14	98.5	100.0%	98.4%	?Spermatogenic failure 23, 617707
TEX15	114.2	100.0%	99.7%	Spermatogenic failure 25, 617960
TF	106.6	100.0%	99.8%	Atransferrinemia, 209300
TFAM	67.1	91.8%	70.6%	?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156
TFAP2A	124.1	100.0%	99.3%	Branchiooculofacial syndrome, 113620
TFAP2B	186.8	99.7%	97.6%	Char syndrome, 169100 Patent ductus arteriosus 2, 617035
TFE3	95.0	99.8%	97.1%	Renal cell carcinoma, papillary, 1, 300854
TFG	112.1	97.1%	95.6%	?Spastic paraplegia 57, autosomal recessive, 615658 Hereditary motor and sensory neuropathy, Okinawa type, 604484
TFR2	138.0	99.8%	98.7%	Hemochromatosis, type 3, 604250
TFRC	133.1	99.9%	99.0%	Immunodeficiency 46, 616740
TG	123.6	100.0%	99.2%	Thyroid dyshormonogenesis 3, 274700
TGDS	84.3	99.3%	95.0%	Catel-Manzke syndrome, 616145
TGFB1	114.1	100.0%	99.5%	Camurati-Engelmann disease, 131300 Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213
TGFB2	179.0	100.0%	99.8%	Loeys-Dietz syndrome 4, 614816
TGFB3	149.0	100.0%	100.0%	Loeys-Dietz syndrome 5, 615582 Arrhythmogenic right ventricular dysplasia 1, 107970
TGFBI	118.3	99.9%	98.8%	Corneal dystrophy, lattice type IIIA, 608471 Corneal dystrophy, Groenouw type I, 121900 Corneal dystrophy, lattice type I, 122200

				Corneal dystrophy, Reis-Bucklers type, 608470 Corneal dystrophy, Thiel-Behnke type, 602082 Corneal dystrophy, epithelial basement membrane, 121820 Corneal dystrophy, Avellino type, 607541
TGFBR1	156.6	97.3%	94.3%	Loeys-Dietz syndrome 1, 609192
TGFBR2	169.1	100.0%	100.0%	Esophageal cancer, somatic, 133239 Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Loeys-Dietz syndrome 2, 610168
TGIF1	150.3	100.0%	100.0%	Holoprosencephaly 4, 142946
TGM1	153.3	100.0%	100.0%	Ichthyosis, congenital, autosomal recessive 1, 242300
TGM3	144.3	99.9%	98.5%	?Uncombable hair syndrome 2, 617251
TGM5	152.1	100.0%	99.9%	Peeling skin syndrome 2, 609796
TGM6	140.6	99.9%	98.8%	Spinocerebellar ataxia 35, 613908
TH	106.8	100.0%	99.2%	Segawa syndrome, recessive, 605407
THAP1	144.9	100.0%	100.0%	Dystonia 6, torsion, 602629
THBD	208.2	100.0%	100.0%	Thrombophilia due to thrombomodulin defect, 614486
THOC2	80.5	98.6%	92.0%	Mental retardation, X-linked 12/35, 300957
THOC6	253.7	100.0%	100.0%	Beaulieu-Boycott-Innes syndrome, 613680
THPO	102.6	100.0%	99.9%	Thrombocythemia 1, 187950
THRA	185.3	100.0%	99.9%	Hypothyroidism, congenital, nongoitrous, 6, 614450
THRB	146.5	100.0%	99.3%	Thyroid hormone resistance, 188570 Thyroid hormone resistance, selective pituitary, 145650 Thyroid hormone resistance, autosomal recessive, 274300
TIA1	128.5	99.9%	96.8%	Welander distal myopathy, 604454
TIMM50	133.8	100.0%	99.4%	3-methylglutaconic aciduria, type IX, 617698
TIMM8A	50.3	95.4%	80.0%	Mohr-Tranebjaerg syndrome, 304700
TIMMDC1	167.0	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 31, 618251
TIMP3	148.2	100.0%	100.0%	Sorsby fundus dystrophy, 136900
TINF2	190.9	100.0%	100.0%	Revesz syndrome, 268130 Dyskeratosis congenita, autosomal dominant 3, 613990
TJP2	114.9	94.0%	93.6%	Hypercholanemia, familial, 607748 Cholestasis, progressive familial intrahepatic 4, 615878
TK2	111.5	100.0%	99.8%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560 ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069
TKT	124.8	98.7%	98.3%	Short stature, developmental delay, and congenital heart defects, 617044
TLE6	128.3	100.0%	99.3%	Preimplantation embryonic lethality, 616814
TLK2	89.8	98.5%	92.8%	Mental retardation, autosomal dominant 57, 618050

TLL1	128.5	100.0%	99.9%	Atrial septal defect 6, 613087
TMC1	111.5	99.9%	97.4%	Deafness, autosomal recessive 7, 600974 Deafness, autosomal dominant 36, 606705
TMC6	102.1	100.0%	99.8%	Epidermodysplasia verruciformis, 226400
TMC8	148.5	100.0%	99.9%	Epidermodysplasia verruciformis 2, 618231
TMCO1	82.6	87.9%	87.3%	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980
TMEM106B	115.3	100.0%	98.8%	Leukodystrophy, hypomyelinating, 16, 617964
TMEM107	161.8	100.0%	100.0%	Orofaciodigital syndrome XVI, 617563 Meckel syndrome 13, 617562 ?Joubert syndrome 29, 617562
TMEM126A	100.7	95.6%	79.5%	Optic atrophy 7, 612989
TMEM126B	87.8	99.6%	96.7%	Mitochondrial complex I deficiency, nuclear type 29, 618250
TMEM132E	139.1	99.9%	98.0%	?Deafness, autosomal recessive 99, 618481
TMEM138	87.8	100.0%	99.0%	Joubert syndrome 16, 614465
TMEM165	159.2	99.9%	99.7%	Congenital disorder of glycosylation, type IIk, 614727
TMEM173	104.9	99.6%	94.7%	STING-associated vasculopathy, infantile-onset, 615934
TMEM199	127.8	100.0%	99.9%	Congenital disorder of glycosylation, type IIp, 616829
TMEM216	92.0	99.9%	96.9%	Meckel syndrome 2, 603194 Joubert syndrome 2, 608091
TMEM231	112.1	100.0%	99.7%	Meckel syndrome 11, 615397 Joubert syndrome 20, 614970
TMEM237	114.5	99.9%	98.8%	Joubert syndrome 14, 614424
TMEM240	184.6	100.0%	100.0%	Spinocerebellar ataxia 21, 607454
TMEM260	117.1	99.9%	97.8%	Structural heart defects and renal anomalies syndrome, 617478
TMEM38B	110.2	100.0%	99.1%	Osteogenesis imperfecta, type XIV, 615066
TMEM43	138.5	99.8%	98.3%	Emery-Dreifuss muscular dystrophy 7, AD, 614302 Arrhythmogenic right ventricular dysplasia 5, 604400
TMEM5	167.7	99.8%	96.8%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
TMEM67	80.6	99.3%	93.5%	Meckel syndrome 3, 607361 ?RHYNS syndrome, 602152 Nephronophthisis 11, 613550 COACH syndrome, 216360 Joubert syndrome 6, 610688
TMEM70	117.3	99.9%	98.5%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMEM94	180.9	100.0%	100.0%	Intellectual developmental disorder with cardiac defects and dysmorphic facies, 618316
TMEM98	131.6	99.1%	96.8%	Nanophthalmos 4, 615972
TMIE	112.7	100.0%	100.0%	Deafness, autosomal recessive 6, 600971

TMPRSS15	104.8	98.4%	94.2%	Enterokinase deficiency, 226200
TMPRSS3	103.4	100.0%	99.6%	Deafness, autosomal recessive 8/10, 601072
TMPRSS6	117.1	100.0%	99.8%	Iron-refractory iron deficiency anemia, 206200
TMTC3	90.1	99.9%	97.3%	Lissencephaly 8, 617255
TNC	154.7	100.0%	99.9%	Deafness, autosomal dominant 56, 615629
TNFAIP3	158.7	100.0%	100.0%	Autoinflammatory syndrome, familial, Behcet-like, 616744
TNFRSF10B	118.0	100.0%	99.9%	Squamous cell carcinoma, head and neck, 275355
TNFRSF11A	139.5	96.4%	95.6%	Osteolysis, familial expansile, 174810 Osteopetrosis, autosomal recessive 7, 612301
TNFRSF11B	175.0	100.0%	100.0%	Paget disease of bone 5, juvenile-onset, 239000
TNFRSF13B	109.3	100.0%	99.8%	Immunoglobulin A deficiency 2, 609529 Immunodeficiency, common variable, 2, 240500
TNFRSF13C	115.9	96.9%	86.5%	Immunodeficiency, common variable, 4, 613494
TNFRSF1A	121.5	92.8%	91.4%	Periodic fever, familial, 142680
TNFRSF4	89.9	99.9%	98.2%	?Immunodeficiency 16, 615593
TNFSF11	133.0	100.0%	100.0%	Osteopetrosis, autosomal recessive 2, 259710
TNIK	107.7	99.9%	98.9%	Mental retardation, autosomal recessive 54, 617028
TNNC1	160.8	100.0%	100.0%	Cardiomyopathy, hypertrophic, 13, 613243 Cardiomyopathy, dilated, 1Z, 611879
TNNI2	169.0	100.0%	100.0%	Arthrogryposis, distal, type 2B1, 601680
TNNI3	112.2	99.6%	96.4%	Cardiomyopathy, hypertrophic, 7, 613690 ?Cardiomyopathy, dilated, 2A, 611880 Cardiomyopathy, dilated, 1FF, 613286 Cardiomyopathy, familial restrictive, 1, 115210
TNNI3K	103.8	100.0%	99.6%	Cardiac conduction disease with or without dilated cardiomyopathy, 616117
TNNT1	113.8	100.0%	99.4%	Nemaline myopathy 5, Amish type, 605355
TNNT2	114.9	100.0%	100.0%	Cardiomyopathy, familial restrictive, 3, 612422 Cardiomyopathy, hypertrophic, 2, 115195 Left ventricular noncompaction 6, 601494 Cardiomyopathy, dilated, 1D, 601494
TNNT3	160.3	100.0%	99.8%	Arthrogryposis, distal, type 2B2, 618435
TNPO3	116.9	100.0%	100.0%	Muscular dystrophy, limb-girdle, autosomal dominant 2, 608423
TNRC6A	131.5	100.0%	99.7%	?Epilepsy, familial adult myoclonic, 6, 618074
TNXB	119.2	99.8%	97.6%	Ehlers-Danlos syndrome, classic-like, 1, 606408 Vesicoureteral reflux 8, 615963
TOE1	153.2	100.0%	100.0%	Pontocerebellar hypoplasia, type 7, 614969
TONSL	142.9	100.0%	99.7%	Spondyloepimetaphyseal dysplasia, sponastrime type, 271510

TOP1	95.9	99.7%	97.4%	DNA topoisomerase I, camptothecin-resistant, 0
TOP2A	117.1	100.0%	99.2%	DNA topoisomerase II, resistance to inhibition of, by amsacrine, 0
TOP3A	128.8	99.7%	97.7%	Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097 ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098
TOPORS	181.9	100.0%	100.0%	Retinitis pigmentosa 31, 609923
TOR1A	147.8	100.0%	100.0%	Dystonia-1, torsion, 128100
TOR1AIP1	142.0	99.2%	97.1%	?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072
TP53	96.8	99.9%	99.0%	Breast cancer, somatic, 114480 Li-Fraumeni syndrome, 151623 Pancreatic cancer, somatic, 260350 Bone marrow failure syndrome 5, 618165 Nasopharyngeal carcinoma, somatic, 607107 Hepatocellular carcinoma, somatic, 114550
TP53RK	93.0	99.8%	97.7%	Galloway-Mowat syndrome 4, 617730
TP63	169.9	100.0%	100.0%	Limb-mammary syndrome, 603543 Orofacial cleft 8, 618149 Split-hand/foot malformation 4, 605289 Hay-Wells syndrome, 106260 Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 Rapp-Hodgkin syndrome, 129400 ADULT syndrome, 103285
TPI1	120.2	99.8%	97.4%	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512
TPK1	96.3	99.7%	97.1%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TPM1	120.1	100.0%	99.8%	Left ventricular noncompaction 9, 611878 Cardiomyopathy, hypertrophic, 3, 115196 Cardiomyopathy, dilated, 1Y, 611878
TPM2	114.5	100.0%	99.8%	Nemaline myopathy 4, autosomal dominant, 609285 Arthrogryposis, distal, type 2B4, 108120 Arthrogryposis, distal, type 1A, 108120 CAP myopathy 2, 609285
TPM3	76.2	89.4%	88.3%	CAP myopathy 1, 609284 Nemaline myopathy 1, autosomal dominant or recessive, 609284 Myopathy, congenital, with fiber-type disproportion, 255310
TPO	146.9	100.0%	99.9%	Thyroid dyshormonogenesis 2A, 274500
TPP1	130.2	100.0%	100.0%	Spinocerebellar ataxia, autosomal recessive 7, 609270 Ceroid lipofuscinosis, neuronal, 2, 204500
TPRKB	57.9	81.5%	74.8%	Galloway-Mowat syndrome 5, 617731
TPRN	113.1	92.8%	88.3%	Deafness, autosomal recessive 79, 613307

TRAC	132.2	100.0%	100.0%	Immunodeficiency 7, TCR-alpha/beta deficient, 615387
TRAF3IP1	87.5	99.1%	96.7%	Senior-Loken syndrome 9, 616629
TRAF3IP2	118.0	100.0%	98.4%	?Candidiasis, familial, 8, 615527
TRAF7	178.6	99.9%	98.7%	Cardiac, facial, and digital anomalies with developmental delay, 618164
TRAIP	128.8	100.0%	100.0%	Seckel syndrome 9, 616777
TRAK1	159.8	100.0%	99.7%	Epileptic encephalopathy, early infantile, 68, 618201
TRAPPC11	124.3	99.9%	99.0%	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356
TRAPPC12	178.4	100.0%	100.0%	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669
TRAPPC2	59.0	84.8%	64.8%	Spondyloepiphyseal dysplasia tarda, 313400
TRAPPC2L	211.9	100.0%	100.0%	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331
TRAPPC6B	72.6	100.0%	97.2%	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862
TRAPPC9	133.0	100.0%	99.8%	Mental retardation, autosomal recessive 13, 613192
TRDN	78.9	95.2%	83.7%	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441
TREH	153.4	98.4%	93.9%	Trehalase deficiency, 612119
TREM2	135.8	100.0%	100.0%	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193
TREX1	261.9	100.0%	100.0%	Vasculopathy, retinal, with cerebral leukodystrophy, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
TRHR	179.7	99.9%	99.1%	Hypothyroidism, congenital, nongoitrous, 7, 618573
TRIM2	144.0	93.9%	93.4%	Charcot-Marie-Tooth disease, type 2R, 615490
TRIM32	132.8	100.0%	100.0%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRIM36	136.1	100.0%	99.1%	?Anencephaly, 206500
TRIM37	112.8	98.5%	97.4%	Mulibrey nanism, 253250
TRIM44	124.5	100.0%	100.0%	?Aniridia 3, 617142
TRIO	129.9	99.3%	97.5%	Mental retardation, autosomal dominant 44, 617061
TRIOBP	173.9	99.6%	98.1%	Deafness, autosomal recessive 28, 609823
TRIP11	87.5	97.2%	91.6%	Osteochondrodysplasia, 184260 Achondrogenesis, type IA, 200600
TRIP12	132.7	100.0%	99.3%	Mental retardation, autosomal dominant 49, 617752
TRIP13	131.4	100.0%	100.0%	Mosaic variegated aneuploidy syndrome 3, 617598
TRIP4	105.3	100.0%	98.9%	Spinal muscular atrophy with congenital bone fractures 1, 616866 ?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066
TRIT1	107.1	100.0%	99.9%	Combined oxidative phosphorylation deficiency 35, 617873
TRMT1	130.7	99.9%	98.4%	Mental retardation, autosomal recessive 68, 618302
TRMT10A	116.1	100.0%	99.9%	Microcephaly, short stature, and impaired glucose metabolism 1, 616033
TRMT10C	138.5	100.0%	100.0%	Combined oxidative phosphorylation deficiency 30, 616974

TRMT5	181.7	99.9%	98.2%	Combined oxidative phosphorylation deficiency 26, 616539
TRMU	106.5	100.0%	99.5%	Liver failure, transient infantile, 613070
TRNT1	100.7	99.2%	95.3%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 Retinitis pigmentosa and erythrocytic microcytosis, 616959
TRPA1	82.2	94.5%	87.3%	?Episodic pain syndrome, familial, 1, 615040
TRPC3	156.9	99.9%	98.4%	?Spinocerebellar ataxia 41, 616410
TRPC6	94.2	98.0%	95.8%	Glomerulosclerosis, focal segmental, 2, 603965
TRPM1	134.7	100.0%	99.3%	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TRPM4	152.9	100.0%	100.0%	Erythrokeratodermia veriabilis et progressiva 6, 618531 Progressive familial heart block, type IB, 604559
TRPM6	128.5	99.9%	99.0%	Hypomagnesemia 1, intestinal, 602014
TRPS1	160.6	100.0%	99.9%	Trichorhinophalangeal syndrome, type I, 190350 Trichorhinophalangeal syndrome, type III, 190351
TRPV3	131.6	99.8%	98.6%	?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400 Olmsted syndrome, 614594
TRPV4	150.4	100.0%	100.0%	Spinal muscular atrophy, distal, congenital nonprogressive, 600175 Spondylometaphyseal dysplasia, Kozlowski type, 184252 Parastremmatic dwarfism, 168400 SED, Maroteaux type, 184095 Scapuloperoneal spinal muscular atrophy, 181405 Metatropic dysplasia, 156530 Digital arthropathy-brachydactyly, familial, 606835 Hereditary motor and sensory neuropathy, type IIc, 606071 Brachyolmia type 3, 113500 ?Avascular necrosis of femoral head, primary, 2, 617383
TRPV6	156.9	100.0%	100.0%	Hyperparathyroidism, transient neonatal, 618188
TRRAP	145.6	99.7%	99.2%	Developmental delay with or without dysmorphic facies and autism, 618454
TSC1	117.4	99.6%	98.4%	Tuberous sclerosis-1, 191100 Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690
TSC2	155.5	100.0%	100.0%	Tuberous sclerosis-2, 613254 ?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690
TSEN15	93.1	99.9%	96.9%	Pontocerebellar hypoplasia, type 2F, 617026
TSEN2	100.3	100.0%	99.1%	Pontocerebellar hypoplasia type 2B, 612389
TSEN34	87.9	99.3%	95.0%	?Pontocerebellar hypoplasia type 2C, 612390

TSEN54	129.0	99.7%	97.9%	Pontocerebellar hypoplasia type 4, 225753 Pontocerebellar hypoplasia type 2A, 277470 ?Pontocerebellar hypoplasia type 5, 610204
TSFM	123.3	100.0%	99.6%	Combined oxidative phosphorylation deficiency 3, 610505
TSGA10	108.8	100.0%	99.1%	?Spermatogenic failure 26, 617961
TSHB	222.2	100.0%	100.0%	Hypothyroidism, congenital, nongoitrous 4, 275100
TSHR	158.0	100.0%	99.0%	Hyperthyroidism, nonautoimmune, 609152 Hypothyroidism, congenital, nongoitrous, 1, 275200 Hyperthyroidism, familial gestational, 603373 Thyroid adenoma, hyperfunctioning, somatic, 0 Thyroid carcinoma with thyrotoxicosis, 0
TSHZ1	160.9	98.8%	98.7%	Aural atresia, congenital, 607842
TSPAN12	132.9	100.0%	99.8%	Exudative vitreoretinopathy 5, 613310
TSPAN7	111.9	100.0%	99.6%	Mental retardation, X-linked 58, 300210
TSPEAR	151.2	100.0%	99.9%	Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180 ?Deafness, autosomal recessive 98, 614861
TSPYL1	159.7	100.0%	100.0%	Sudden infant death with dysgenesis of the testes syndrome, 608800
TSR2	81.3	100.0%	99.4%	?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946
TTBK2	110.8	99.9%	98.1%	Spinocerebellar ataxia 11, 604432
TTC19	84.9	98.8%	86.6%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTC21A	127.5	100.0%	99.9%	Spermatogenic failure 37, 618429
TTC21B	115.1	99.9%	98.8%	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
TTC25	100.1	100.0%	99.7%	Ciliary dyskinesia, primary, 35, 617092
TTC37	131.5	99.9%	98.9%	Trichohepatoenteric syndrome 1, 222470
TTC7A	123.2	99.9%	98.9%	Gastrointestinal defects and immunodeficiency syndrome, 243150
TTC8	116.8	99.7%	97.8%	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
TTI2	100.1	100.0%	99.9%	Mental retardation, autosomal recessive 39, 615541
TTLL5	138.8	99.9%	98.9%	Cone-rod dystrophy 19, 615860
TTN	165.0	98.6%	98.1%	Cardiomyopathy, dilated, 1G, 604145 Tibial muscular dystrophy, tardive, 600334 Salih myopathy, 611705 Cardiomyopathy, familial hypertrophic, 9, 613765 Myopathy, myofibrillar, 9, with early respiratory failure, 603689 Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807
TTPA	109.2	99.3%	95.6%	Ataxia with isolated vitamin E deficiency, 277460

TTR	125.9	94.6%	94.6%	Amyloidosis, hereditary, transthyretin-related, 105210 Carpal tunnel syndrome, familial, 115430
TUB	112.3	100.0%	99.4%	?Retinal dystrophy and obesity, 616188
TUBA1A	82.5	99.9%	97.8%	Lissencephaly 3, 611603
TUBA3D	109.2	100.0%	97.4%	Keratoconus 9, 617928
TUBA4A	173.6	100.0%	100.0%	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia, 616208
TUBA8	136.6	100.0%	99.5%	Cortical dysplasia, complex, with other brain malformations 8, 613180
TUBB	123.3	98.0%	94.4%	Symmetric circumferential skin creases, congenital, 1, 156610 Cortical dysplasia, complex, with other brain malformations 6, 615771
TUBB1	164.6	100.0%	100.0%	Macrothrombocytopenia, autosomal dominant, TUBB1-related, 613112
TUBB2A	83.6	99.8%	98.0%	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB2B	88.3	100.0%	99.9%	Cortical dysplasia, complex, with other brain malformations 7, 610031
TUBB3	135.6	99.9%	99.1%	Fibrosis of extraocular muscles, congenital, 3A, 600638 Cortical dysplasia, complex, with other brain malformations 1, 614039
TUBB4A	114.3	97.8%	95.9%	Leukodystrophy, hypomyelinating, 6, 612438 Dystonia 4, torsion, autosomal dominant, 128101
TUBB4B	96.6	100.0%	100.0%	Leber congenital amaurosis with early-onset deafness, 617879
TUBB6	97.5	92.7%	90.7%	?Facial palsy, congenitla, with ptosis and velopharyngeal dysfunction, 617732
TUBB8	23.3	89.2%	58.1%	Oocyte maturation defect 2, 616780
TUBG1	162.6	100.0%	100.0%	Cortical dysplasia, complex, with other brain malformations 4, 615412
TUBGCP4	108.4	97.8%	95.1%	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TUBGCP6	169.9	100.0%	99.7%	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
TUFM	141.1	100.0%	99.6%	Combined oxidative phosphorylation deficiency 4, 610678
TULP1	128.8	100.0%	99.7%	Retinitis pigmentosa 14, 600132 Leber congenital amaurosis 15, 613843
TUSC3	155.7	99.9%	99.5%	Mental retardation, autosomal recessive 7, 611093
TWIST1	185.6	100.0%	100.0%	Robinow-Sorauf syndrome, 180750 Craniosynostosis 1, 123100 Sweeney-Cox syndrome, 617746 Saethre-Chotzen syndrome with or without eyelid anomalies, 101400
TWIST2	159.1	100.0%	100.0%	Barber-Say syndrome, 209885 Ablepharon-macrostomia syndrome, 200110 Focal facial dermal dysplasia 3, Setleis type, 227260
TWNK	170.3	100.0%	100.0%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 Perrault syndrome 5, 616138
TXN2	65.7	100.0%	99.8%	?Combined oxidative phosphorylation deficiency 29, 616811

TXNL4A	138.1	100.0%	98.5%	Burn-McKeown syndrome, 608572
TXNRD2	121.5	96.7%	95.6%	?Glucocorticoid deficiency 5, 617825
TYK2	142.1	100.0%	99.7%	Immunodeficiency 35, 611521
TYMP	138.6	100.0%	100.0%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYR	153.5	100.0%	100.0%	Waardenburg syndrome/albinism, digenic, 103470 Albinism, oculocutaneous, type IB, 606952 Albinism, oculocutaneous, type IA, 203100
TYROBP	94.9	100.0%	100.0%	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770
TYRP1	155.1	100.0%	99.9%	Albinism, oculocutaneous, type III, 203290
UBA1	139.8	99.7%	98.4%	Spinal muscular atrophy, X-linked 2, infantile, 301830
UBA5	79.3	96.9%	83.9%	?Spinocerebellar ataxia, autosomal recessive 24, 617133 Epileptic encephalopathy, early infantile, 44, 617132
UBAP1	133.7	99.3%	96.3%	Spastic paraplegia 80, autosomal dominant, 618418
UBE2A	122.1	99.6%	97.4%	Mental retardation, X-linked syndromic, Nascimento-type, 300860
UBE2T	92.1	100.0%	99.4%	Fanconi anemia, complementation group T, 616435
UBE3A	80.9	98.8%	92.5%	Angelman syndrome, 105830
UBE3B	119.7	100.0%	99.8%	Kaufman oculocerebrofacial syndrome, 244450
UBIAD1	202.0	99.9%	98.2%	Corneal dystrophy, Schnyder type, 121800
UBQLN2	132.2	100.0%	99.8%	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857
UBR1	118.2	99.9%	98.9%	Johanson-Blizzard syndrome, 243800
UBTF	127.3	100.0%	99.8%	Neurodegeneration, childhood-onset, with brain atrophy, 617672
UCHL1	104.7	99.7%	95.9%	Spastic paraplegia 79, autosomal recessive, 615491
UFC1	125.9	100.0%	100.0%	Neurodevelopmental disorder with spasticity and poor growth, 618076
UFM1	109.2	72.0%	69.8%	Leukodystrophy, hypomyelinating, 14, 617899
UFSP2	126.6	100.0%	99.5%	?Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974 ?Hip dysplasia, Beukes type, 142669
UGT1A1	192.9	100.0%	100.0%	Hyperbilirubinemia, familial transient neonatal, 237900 Crigler-Najjar syndrome, type I, 218800 Crigler-Najjar syndrome, type II, 606785
UMOD	123.1	97.7%	95.8%	Glomerulocystic kidney disease with hyperuricemia and isosthenuria, 609886 Medullary cystic kidney disease 2, 603860 Hyperuricemic nephropathy, familial juvenile 1, 162000
UMPS	156.7	100.0%	98.8%	Orotic aciduria, 258900
UNC119	126.4	100.0%	99.9%	?Immunodeficiency 13, 615518 ?Cone-rod dystrophy, 0
UNC13D	120.2	99.9%	99.2%	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC45B	121.5	100.0%	99.3%	?Cataract 43, 616279

UNC80	114.4	100.0%	99.6%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801
UNG	127.2	98.9%	95.1%	Immunodeficiency with hyper IgM, type 5, 608106
UPB1	150.9	100.0%	100.0%	Beta-ureidopropionase deficiency, 613161
UPF3B	60.7	94.1%	84.6%	Mental retardation, X-linked, syndromic 14, 300676
UQCC2	146.3	99.8%	98.8%	Mitochondrial complex III deficiency, nuclear type 7, 615824
UQCC3	140.4	100.0%	100.0%	?Mitochondrial complex III deficiency, nuclear type 9, 616111
UQCRB	106.1	98.8%	94.8%	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC2	105.8	99.9%	98.3%	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRQ	172.4	100.0%	100.0%	Mitochondrial complex III deficiency, nuclear type 4, 615159
UROC1	143.4	100.0%	99.9%	?Urocanase deficiency, 276880
UROD	139.7	99.6%	96.7%	Porphyria, hepatoerythropoietic, 176100 Porphyria cutanea tarda, 176100
UROS	104.6	100.0%	99.9%	Porphyria, congenital erythropoietic, 263700
USB1	122.0	99.8%	98.2%	Poikiloderma with neutropenia, 604173
USH1C	99.1	100.0%	99.3%	Deafness, autosomal recessive 18A, 602092 Usher syndrome, type 1C, 276904
USH1G	221.0	99.9%	99.3%	Usher syndrome, type 1G, 606943
USH2A	130.8	100.0%	99.8%	Retinitis pigmentosa 39, 613809 Usher syndrome, type 2A, 276901
USP18	151.7	95.9%	95.9%	Pseudo-TORCH syndrome 2, 617397
USP27X	167.4	100.0%	100.0%	Mental retardation, X-linked 105, 300984
USP45	93.8	99.7%	98.1%	?Leber congenital amaurosis 19, 618513
USP8	65.8	98.4%	89.5%	Pituitary adenoma 4, ACTH-secreting, somatic, 219090
USP9X	92.5	97.9%	91.6%	Mental retardation, X-linked 99, 300919 Mental retardation, X-linked 99, syndromic, female-restricted, 300968
USP9Y	28.1	47.8%	41.3%	Spermatogenic failure, Y-linked, 2, 415000
UVSSA	140.1	99.6%	99.5%	UV-sensitive syndrome 3, 614640
VAC14	107.4	100.0%	98.8%	Striatonigral degeneration, childhood-onset, 617054
VAMP1	142.3	100.0%	100.0%	Spastic ataxia 1, autosomal dominant, 108600 Myasthenic syndrome, congenital, 25, 618323
VANGL1	157.4	100.0%	100.0%	Caudal regression syndrome, 600145
VANGL2	171.4	100.0%	99.6%	Neural tube defects, 182940
VAPB	94.9	100.0%	99.4%	Spinal muscular atrophy, late-onset, Finkel type, 182980 Amyotrophic lateral sclerosis 8, 608627
VARS	142.4	100.0%	99.9%	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802
VARS2	130.9	100.0%	99.8%	Combined oxidative phosphorylation deficiency 20, 615917
VAX1	108.2	100.0%	99.4%	?Microphthalmia, syndromic 11, 614402

VCAN	155.3	100.0%	100.0%	Wagner syndrome 1, 143200
VCL	105.4	100.0%	99.2%	Cardiomyopathy, dilated, 1W, 611407 Cardiomyopathy, hypertrophic, 15, 613255
VCP	103.9	100.0%	99.4%	Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320 Charcot-Marie-Tooth disease, type 2Y, 616687 Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954
VDR	116.5	99.0%	96.4%	Rickets, vitamin D-resistant, type IIA, 277440
VEGFC	166.9	100.0%	100.0%	Lymphatic malformation 4, 615907
VHL	182.8	100.0%	99.8%	Pheochromocytoma, 171300 Erythrocytosis, familial, 2, 263400 von Hippel-Lindau syndrome, 193300 Renal cell carcinoma, somatic, 144700 Hemangioblastoma, cerebellar, somatic, 0
VIM	138.3	99.7%	98.0%	Cataract 30, pulverulent, 116300
VIPAS39	114.4	100.0%	99.9%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VKORC1	161.2	100.0%	100.0%	Warfarin resistance, 122700 Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473
VLDLR	145.5	100.0%	100.0%	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VMA21	97.5	99.4%	95.0%	Myopathy, X-linked, with excessive autophagy, 310440
VPS11	125.6	95.5%	93.8%	Leukodystrophy, hypomyelinating, 12, 616683
VPS13A	74.8	99.2%	94.2%	Choreoacanthocytosis, 200150
VPS13B	135.9	99.4%	97.8%	Cohen syndrome, 216550
VPS13C	106.4	99.4%	95.8%	Parkinson disease 23, autosomal recessive, early onset, 616840
VPS13D	141.7	100.0%	99.8%	Spinocerebellar ataxia, autosomal recessive 4, 607317
VPS33A	106.3	96.6%	94.7%	Mucopolysaccharidosis-plus syndrome, 617303
VPS33B	111.7	100.0%	100.0%	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
VPS37A	61.7	89.1%	74.8%	Spastic paraplegia 53, autosomal recessive, 614898
VPS45	127.2	97.3%	94.1%	Neutropenia, severe congenital, 5, autosomal recessive, 615285
VPS51	109.0	99.2%	95.7%	Pontocerebellar hypoplasia, type 13, 618606
VPS53	117.2	91.3%	89.9%	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	126.7	99.9%	98.3%	Pontocerebellar hypoplasia type 1A, 607596
VSX1	73.0	99.0%	92.6%	Keratoconus 1, 148300 ?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195
VSX2	134.2	100.0%	100.0%	Microphthalmia with coloboma 3, 610092 Microphthalmia, isolated 2, 610093
VWA3B	130.0	100.0%	99.3%	?Spinocerebellar ataxia, autosomal recessive 22, 616948

VWF	105.1	100.0%	99.3%	von Willebrand disease, type 1, 193400 von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 von Willibrand disease, type 3, 277480
WAC	142.4	100.0%	99.2%	Desanto-Shinawi syndrome, 616708
WARS	101.4	99.8%	97.9%	Neuronopathy, distal hereditary motor, type IX, 617721
WARS2	142.1	99.9%	99.1%	Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710
WAS	75.4	95.3%	84.4%	Thrombocytopenia, X-linked, intermittent, 313900 Thrombocytopenia, X-linked, 313900 Wiskott-Aldrich syndrome, 301000 Neutropenia, severe congenital, X-linked, 300299
WASHC4	104.0	98.9%	94.9%	?Mental retardation, autosomal recessive 43, 615817
WASHC5	133.1	100.0%	99.5%	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, autosomal dominant, 603563
WBP2	101.8	100.0%	99.5%	Deafness, autosomal recessive 107, 617639
WDFY3	125.5	100.0%	99.3%	?Microcephaly 18, primary, autosomal dominant, 617520
WDPCP	105.7	97.1%	93.6%	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR11	116.0	97.3%	96.1%	Hypogonadotropic hypogonadism 14 with or without anosmia, 614858
WDR19	125.3	100.0%	99.4%	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WDR26	97.7	99.9%	98.4%	Skraban-Deardorff syndrome, 617616
WDR34	129.6	100.0%	100.0%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
WDR35	137.8	99.5%	98.3%	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 Cranioectodermal dysplasia 2, 613610
WDR36	125.5	99.8%	97.6%	Glaucoma 1, open angle, G, 609887
WDR37	152.1	100.0%	98.9%	Neurooculocardiogenitourinary syndrome, 618652
WDR4	152.7	100.0%	100.0%	Microcephaly, growth deficiency, seizures, and brain malformations, 618346 Galloway-Mowat syndrome 6, 618347
WDR45	74.7	97.1%	90.6%	Neurodegeneration with brain iron accumulation 5, 300894
WDR45B	75.6	97.4%	90.3%	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977
WDR60	111.7	99.8%	97.8%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WDR62	166.4	100.0%	100.0%	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
WDR66	122.3	100.0%	99.9%	Spermatogenic failure 33, 618152
WDR72	123.2	96.9%	96.1%	Amelogenesis imperfecta, type IIA3, 613211

WDR73	164.4	100.0%	100.0%	Galloway-Mowat syndrome 1, 251300
WDR81	205.5	100.0%	100.0%	Hydrocephalus, congenital, 3, with brain anomalies, 617967 Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185
WEE2	101.7	100.0%	99.2%	Oocyte maturation defect 5, 617996
WFS1	210.0	100.0%	99.8%	?Cataract 41, 116400 Deafness, autosomal dominant 6/14/38, 600965 Wolfram-like syndrome, autosomal dominant, 614296 Wolfram syndrome 1, 222300
WHRN	145.9	100.0%	99.3%	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
WIPF1	95.7	100.0%	99.3%	?Wiskott-Aldrich syndrome 2, 614493
WIPI2	123.1	99.8%	97.7%	?Intellectual developmental disorder with short stature and variable skeletal anomalies, 618453
WISP3	118.0	100.0%	100.0%	Spondyloepiphyseal dysplasia tarda with progressive arthropathy, 208230 Arthropathy, progressive pseudorheumatoid, of childhood, 208230
WNK1	138.8	100.0%	99.6%	Pseudohypoaldosteronism, type IIC, 614492 Neuropathy, hereditary sensory and autonomic, type II, 201300
WNK4	157.3	100.0%	99.6%	Pseudohypoaldosteronism, type IIB, 614491
WNT1	289.4	100.0%	100.0%	Osteogenesis imperfecta, type XV, 615220
WNT10A	159.7	100.0%	100.0%	Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400 Odontoonychodermal dysplasia, 257980
WNT10B	176.5	100.0%	100.0%	Split-hand/foot malformation 6, 225300 Tooth agenesis, selective, 8, 617073
WNT2B	139.0	100.0%	98.0%	Diarrhea 9, 618168
WNT3	186.5	100.0%	99.9%	?Tetra-amelia syndrome 1, 273395
WNT4	254.1	99.9%	99.1%	?SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330
WNT5A	174.9	100.0%	100.0%	Robinow syndrome, autosomal dominant 1, 180700
WNT7A	218.7	100.0%	100.0%	Fuhrmann syndrome, 228930 Ulna and fibula, absence of, with severe limb deficiency, 276820
WRAP53	178.7	100.0%	100.0%	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	120.3	100.0%	98.7%	Werner syndrome, 277700
WT1	96.5	100.0%	99.6%	Mesothelioma, somatic, 156240 Wilms tumor, type 1, 194070 Frasier syndrome, 136680 Denys-Drash syndrome, 194080 Meacham syndrome, 608978 Nephrotic syndrome, type 4, 256370

WWOX	122.0	100.0%	100.0%	Esophageal squamous cell carcinoma, somatic, 133239 Spinocerebellar ataxia, autosomal recessive 12, 614322 Epileptic encephalopathy, early infantile, 28, 616211
XDH	98.4	100.0%	99.8%	Xanthinuria, type I, 278300
XIAP	94.7	93.1%	88.3%	Lymphoproliferative syndrome, X-linked, 2, 300635
XIST	NC	NC	NC	X-inactivation, familial skewed, 300087
XK	88.8	99.9%	99.5%	McLeod syndrome with or without chronic granulomatous disease, 300842
XPA	73.5	99.9%	97.6%	Xeroderma pigmentosum, group A, 278700
XPC	151.8	100.0%	99.9%	Xeroderma pigmentosum, group C, 278720
XPNPEP3	103.7	100.0%	99.9%	Nephronophthisis-like nephropathy 1, 613159
XPR1	126.1	100.0%	99.9%	Basal ganglia calcification, idiopathic, 6, 616413
XRCC1	122.7	100.0%	99.6%	?Spinocerebellar ataxia, autosomal recessive 26, 617633
XRCC2	169.7	99.8%	95.1%	?Fanconi anemia, complementation group U, 617247
XRCC4	139.7	100.0%	99.2%	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	138.1	100.0%	99.4%	Desbuquois dysplasia 2, 615777
XYLT2	161.8	99.9%	98.7%	Spondyloocular syndrome, 605822
YAP1	97.9	98.5%	94.0%	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433
YARS	110.2	100.0%	99.4%	Charcot-Marie-Tooth disease, dominant intermediate C, 608323
YARS2	188.2	100.0%	99.4%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
YME1L1	102.3	98.1%	92.4%	?Optic atrophy 11, 617302
YWHAG	181.4	100.0%	100.0%	Epileptic encephalopathy, early infantile, 56, 617665
YY1	146.4	100.0%	99.1%	Gabriele-de Vries syndrome, 617557
YY1AP1	152.8	98.4%	97.0%	Grange syndrome, 602531
ZAP70	206.3	100.0%	99.9%	Immunodeficiency 48, 269840 Autoimmune disease, multisystem, infantile-onset, 2, 617006
ZBTB11	165.5	100.0%	99.6%	Intellectual developmental disorder, autosomal recessive 69, 618383
ZBTB16	161.1	100.0%	100.0%	Skeletal defects, genital hypoplasia, and mental retardation, 612447 Leukemia, acute promyelocytic, PL2F/RARA type, 0
ZBTB18	185.2	99.9%	99.4%	Mental retardation, autosomal dominant 22, 612337
ZBTB20	199.5	100.0%	100.0%	Primrose syndrome, 259050
ZBTB24	160.7	100.0%	100.0%	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069
ZBTB42	152.6	100.0%	100.0%	?Lethal congenital contracture syndrome 6, 616248
ZC3H14	153.9	99.9%	98.4%	Mental retardation, autosomal recessive 56, 617125
ZC4H2	74.3	99.6%	95.6%	Wieacker-Wolff syndrome, 314580
ZDHHC9	50.3	98.4%	89.1%	Mental retardation, X-linked syndromic, Raymond type, 300799
ZEB1	155.3	100.0%	99.8%	Corneal dystrophy, posterior polymorphous, 3, 609141 Corneal dystrophy, Fuchs endothelial, 6, 613270

ZEB2	145.0	99.7%	98.6%	Mowat-Wilson syndrome, 235730
ZFHX2	144.4	100.0%	99.9%	?Marsili syndrome, 147430
ZFHX3	130.6	100.0%	99.9%	Prostate cancer, somatic, 176807
ZFP57	113.4	100.0%	99.3%	Diabetes mellitus, transient neonatal, 1, 601410
ZFPM2	160.7	100.0%	99.9%	46XY sex reversal 9, 616067 Diaphragmatic hernia 3, 610187 Tetralogy of Fallot, 187500
ZFYVE26	110.8	99.9%	99.0%	Spastic paraplegia 15, autosomal recessive, 270700
ZFYVE27	117.1	100.0%	100.0%	Spastic paraplegia 33, autosomal dominant, 610244
ZIC1	319.7	100.0%	100.0%	Craniosynostosis 6, 616602
ZIC2	190.6	98.4%	96.3%	Holoprosencephaly 5, 609637
ZIC3	155.4	100.0%	99.8%	Congenital heart defects, nonsyndromic, 1, X-linked, 306955 Heterotaxy, visceral, 1, X-linked, 306955 VACTERL association, X-linked, 314390
ZMPSTE24	126.2	100.0%	99.7%	Restrictive dermopathy, lethal, 275210 Mandibuloacral dysplasia with type B lipodystrophy, 608612
ZMYND10	132.9	100.0%	100.0%	Ciliary dyskinesia, primary, 22, 615444
ZMYND11	119.7	100.0%	99.6%	Mental retardation, autosomal dominant 30, 616083
ZMYND15	164.7	100.0%	99.9%	?Spermatogenic failure 14, 615842
ZNF141	121.7	100.0%	100.0%	?Polydactyly, postaxial, type A6, 615226
ZNF142	145.6	100.0%	99.9%	Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425
ZNF148	157.2	100.0%	99.9%	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260
ZNF335	147.5	100.0%	99.9%	Microcephaly 10, primary, autosomal recessive, 615095
ZNF341	129.0	99.1%	97.4%	Hyper-IgE recurrent infection syndrome 3, autosomal recessive, 618282
ZNF408	162.5	100.0%	100.0%	?Exudative vitreoretinopathy 6, 616468 Retinitis pigmentosa 72, 616469
ZNF423	215.2	100.0%	100.0%	Nephronophthisis 14, 614844 Joubert syndrome 19, 614844
ZNF462	164.9	100.0%	99.8%	Weiss-Kruszka syndrome, 618619
ZNF469	180.5	100.0%	100.0%	Brittle cornea syndrome 1, 229200
ZNF513	153.8	100.0%	100.0%	?Retinitis pigmentosa 58, 613617
ZNF644	153.0	100.0%	99.9%	Myopia 21, autosomal dominant, 614167
ZNF687	187.1	100.0%	100.0%	Paget disease of bone 6, 616833
ZNF711	110.7	99.7%	97.4%	Mental retardation, X-linked 97, 300803
ZNF750	197.7	100.0%	99.9%	Seborrhea-like dermatitis with psoriasiform elements, 610227
ZNHIT3	141.9	74.4%	74.4%	PEHO syndrome, 260565
ZP1	194.3	100.0%	100.0%	Oocyte maturation defect 1, 615774

ZP2	119.5	99.8%	99.0%	Oocyte maturation defect 6, 618353
ZP3	157.9	100.0%	100.0%	Oocyte maturation defect 3, 617712
ZSWIM6	127.2	97.5%	95.6%	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 11th , 2019.
This list is accurate for panel version DG 2.17
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors