PRECONCEPTION SCREENING GENE PANEL DG 2.17 (2198 genes)

Releasedate: 06-12-2019

Gene	*Median Coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
AAAS	109.1	100.0%	99.8%	Achalasia-addisonianism-alacrimia syndrome, 231550
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
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
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

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
ACACA	111.9	98.3%	97.6%	No OMIM disease ID
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
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
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
ACTL6B	144.9	100.0%	100.0%	Epileptic encephalopathy, early infantile, 76, 618468 Intellectual developmental disorder with severe speech and ambulation defects, 618470
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
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
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
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
ADGRG1	159.1	100.0%	100.0%	Polymicrogyria, bilateral perisylvian, 615752 Polymicrogyria, bilateral frontoparietal, 606854
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
ADK	101.3	99.9%	97.3%	Hypermethioninemia due to adenosine kinase deficiency, 614300
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
AFG3L2	100.8	95.7%	85.1%	Spastic ataxia 5, autosomal recessive, 614487 Spinocerebellar ataxia 28, 610246
AGA	144.3	100.0%	100.0%	Aspartylglucosaminuria, 208400
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
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
AHI1	125.5	99.9%	97.6%	Joubert syndrome 3, 608629
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
AIMP1	80.4	99.1%	91.4%	Leukodystrophy, hypomyelinating, 3, 260600
AIMP2	126.1	97.0%	89.6%	Leukodystrophy, hypomyelinating, 17, 618006
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
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
ALAD	101.8	99.4%	95.4%	Porphyria, acute hepatic, 612740
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
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
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
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
ANAPC1	74.5	59.5%	56.7%	Rothmund-Thomson syndrome, type 1, 618625
ANGPTL3	88.9	99.0%	95.0%	Hypobetalipoproteinemia, familial, 2, 605019

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
ANKS6	101.2	99.3%	96.5%	Nephronophthisis 16, 615382
ANO10	106.0	98.6%	96.5%	Spinocerebellar ataxia, autosomal recessive 10, 613728
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
ANTXR1	112.9	99.3%	97.3%	GAPO syndrome, 230740
ANTXR2	117.5	99.8%	97.7%	Hyaline fibromatosis syndrome, 228600
AP1S1	105.8	100.0%	99.9%	MEDNIK syndrome, 609313
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
APC2	142.2	100.0%	99.6%	?Sotos syndrome 3, 617169 Cortical dysplasia, complex, with other brain malformations 10, 618677
APOB	158.4	100.0%	99.8%	Hypobetalipoproteinemia, 615558 Hypercholesterolemia, familial, 2, 144010
APOC2	112.2	100.0%	100.0%	Hyperlipoproteinemia, type Ib, 207750
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
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
ARFGEF2	129.2	99.9%	98.9%	Periventricular heterotopia with microcephaly, 608097
ARG1	158.2	100.0%	100.0%	Argininemia, 207800

ARHGDIA	226.3	100.0%	99.9%	Nephrotic syndrome, type 8, 615244
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
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
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
ARMC4	107.7	94.4%	93.5%	Ciliary dyskinesia, primary, 23, 615451
ARMC9	129.0	100.0%	99.4%	Joubert syndrome 30, 617622
ARNT2	127.7	100.0%	100.0%	?Webb-Dattani syndrome, 615926
ARSA	154.9	100.0%	100.0%	Metachromatic leukodystrophy, 250100
ARSB	111.3	100.0%	99.4%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARV1	108.7	100.0%	99.7%	Epileptic encephalopathy, early infantile, 38, 617020
ASAH1	124.7	99.6%	96.8%	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASCC1	122.4	95.7%	92.1%	Barrett esophagus/esophageal adenocarcinoma, 614266 ?Spinal muscular atrophy with congenital bone fractures 2, 616867
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
ASS1	106.1	95.4%	88.7%	Citrullinemia, 215700
ATAD1	63.3	98.8%	91.0%	Hyperekplexia 4, 618011
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
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
ATOH7	210.8	99.8%	99.2%	Persistent hyperplastic primary vitreous, autosomal recessive, 221900
ATP13A2	149.6	100.0%	99.8%	Kufor-Rakeb syndrome, 606693 Spastic paraplegia 78, autosomal recessive, 617225
ATP2A1	157.9	100.0%	100.0%	Brody myopathy, 601003

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
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
ATP6V1E1	67.0	91.8%	86.3%	Cutis laxa, autosomal recessive, type IIC, 617402
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
AUH	135.7	100.0%	99.8%	3-methylglutaconic aciduria, type I, 250950
AURKC	72.7	99.5%	93.6%	Spermatogenic failure 5, 243060
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
BANF1	52.9	97.3%	86.5%	Nestor-Guillermo progeria syndrome, 614008
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
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
BCS1L	160.0	100.0%	100.0%	Leigh syndrome, 256000 GRACILE syndrome, 603358 Bjornstad syndrome, 262000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BFSP1	105.8	100.0%	99.3%	Cataract 33, multiple types, 611391
BFSP2	106.9	100.0%	99.2%	Cataract 12, multiple types, 611597
BHLHA9	41.5	97.8%	83.4%	Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432 ?Camptosynpolydactyly, complex, 607539
BIN1	125.8	100.0%	99.5%	Centronuclear myopathy 2, 255200
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
BMPER	132.4	100.0%	99.6%	Diaphanospondylodysostosis, 608022
BMPR1B	141.6	100.0%	100.0%	Brachydactyly, type A2, 112600 Brachydactyly, type A1, D, 616849 Acromesomelic dysplasia, Demirhan type, 609441
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
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
BRF1	120.2	99.9%	98.8%	Cerebellofaciodental syndrome, 616202
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
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
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
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
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
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
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
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
CACNA1B	147.4	99.8%	98.6%	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497
CACNA1D	135.3	98.0%	97.8%	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896
CACNA2D4	103.4	99.2%	97.4%	Retinal cone dystrophy 4, 610478
CAD	147.4	100.0%	99.6%	Epileptic encephalopathy, early infantile, 50, 616457
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
CAPN10	130.9	100.0%	99.9%	No OMIM disease ID
CAPN3	103.8	99.4%	97.3%	Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600 Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129
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
CARD9	152.2	100.0%	99.9%	Candidiasis, familial, 2, autosomal recessive, 212050
CARS2	138.8	100.0%	100.0%	Combined oxidative phosphorylation deficiency 27, 616672
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
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
CAVIN1	200.3	100.0%	100.0%	Lipodystrophy, congenital generalized, type 4, 613327
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
CCDC39	82.4	99.6%	96.0%	Ciliary dyskinesia, primary, 14, 613807
CCDC40	120.1	99.5%	98.5%	Ciliary dyskinesia, primary, 15, 613808
CCDC65	84.8	99.8%	97.8%	Ciliary dyskinesia, primary, 27, 615504
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
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
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
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
CD40	157.9	100.0%	100.0%	Immunodeficiency with hyper-IgM, type 3, 606843
CD59	153.6	94.1%	86.4%	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
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
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
CDC45	148.0	99.6%	98.4%	Meier-Gorlin syndrome 7, 617063
CDC6	142.2	100.0%	99.7%	?Meier-Gorlin syndrome 5, 613805
CDCA7	117.7	100.0%	99.7%	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910
CDH11	131.7	100.0%	100.0%	Elsahy-Waters syndrome, 211380
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
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
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
CEBPE	113.9	100.0%	100.0%	Specific granule deficiency, 245480
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
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
CEP89	130.0	98.2%	95.5%	No OMIM disease ID
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
CFAP53	135.5	99.2%	97.0%	Heterotaxy, visceral, 6, autosomal recessive, 614779
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
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
CFTR	112.9	99.4%	97.3%	Cystic fibrosis, 219700 Congenital bilateral absence of vas deferens, 277180 Sweat chloride elevation without CF, 0
CHAT	125.5	97.1%	89.5%	Myasthenic syndrome, congenital, 6, presynaptic, 254210
CHKB	126.8	100.0%	100.0%	Muscular dystrophy, congenital, megaconial type, 602541
CHMP1A	133.0	100.0%	99.8%	Pontocerebellar hypoplasia, type 8, 614961
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
CHRNB1	134.8	100.0%	99.8%	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314 Myasthenic syndrome, congenital, 2A, slow-channel, 616313
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
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
CIB2	218.3	100.0%	99.5%	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
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
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
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

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
CLIC5	103.2	100.0%	99.9%	?Deafness, autosomal recessive 103, 616042
CLIP1	122.0	99.9%	99.1%	No OMIM Disease ID
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
CLRN1	140.6	100.0%	99.5%	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902
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
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
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
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
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
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

COL4A3	93.8	99.6%	97.9%	Hematuria, benign familial, 141200 Alport syndrome 2, autosomal recessive, 203780 Alport syndrome 3, autosomal dominant, 104200
COL4A4	96.5	99.7%	98.0%	Alport syndrome 2, autosomal recessive, 203780 Hematuria, familial benign, 141200
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
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
COLEC10	119.9	100.0%	100.0%	3MC syndrome 3, 248340
COLEC11	197.8	100.0%	100.0%	3MC syndrome 2, 265050
COLQ	106.4	99.9%	98.2%	Myasthenic syndrome, congenital, 5, 603034
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
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
COX6B1	143.0	100.0%	100.0%	Mitochondrial complex IV deficiency, 220110
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
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
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
CREB3L1	145.4	100.0%	99.9%	Osteogenesis imperfecta, type XVI, 616229
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
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
CRYBB1	132.7	100.0%	99.9%	Cataract 17, multiple types, 611544
CRYBB3	149.6	100.0%	100.0%	Cataract 22, 609741
CSF1R	121.9	100.0%	99.4%	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
CSF2RB	136.0	99.9%	98.7%	Surfactant metabolism dysfunction, pulmonary, 5, 614370
CSPP1	117.4	100.0%	99.4%	Joubert syndrome 21, 615636
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
CTC1	113.5	100.0%	99.6%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTDP1	141.7	96.2%	88.2%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
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
CUBN	103.2	99.6%	98.0%	Megaloblastic anemia-1, Finnish type, 261100
CUL7	139.6	100.0%	100.0%	3-M syndrome 1, 273750
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
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
CYC1	167.9	99.7%	97.3%	Mitochondrial complex III deficiency, nuclear type 6, 615453
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
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
CYP2C8	91.2	99.1%	95.2%	No OMIM disease ID
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
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
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
DCPS	140.6	100.0%	99.9%	Al-Raqad syndrome, 616459
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
DDX59	143.3	100.0%	99.7%	Orofaciodigital syndrome V, 174300
DEAF1	124.1	100.0%	99.1%	Mental retardation, autosomal dominant 24, 615828 ?Dyskinesia, seizures, and intellectual developmental disorder, 617171
DENND5A	101.5	99.8%	98.9%	Epileptic encephalopathy, early infantile, 49, 617281
DES	138.0	100.0%	100.0%	Cardiomyopathy, dilated, 1I, 604765 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 Myopathy, myofibrillar, 1, 601419
DFNB59	116.0	100.0%	99.2%	Deafness, autosomal recessive 59, 610220
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
DIAPH1	104.4	100.0%	99.8%	Seizures, cortical blindness, microcephaly syndrome, 616632 Deafness, autosomal dominant 1, 124900
DIS3L2	151.0	100.0%	99.9%	Perlman syndrome, 267000
DLAT	104.9	100.0%	99.4%	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	117.2	100.0%	99.9%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLL3	122.2	97.9%	93.8%	Spondylocostal dysostosis 1, autosomal recessive, 277300
DLX5	159.7	100.0%	99.8%	?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600
DMGDH	135.4	100.0%	99.8%	Dimethylglycine dehydrogenase deficiency, 605850
DMP1	135.8	100.0%	99.9%	Hypophosphatemic rickets, AR, 241520
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
DNAH11	132.3	99.9%	98.9%	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH5	115.4	100.0%	99.3%	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
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
DNAJB2	129.2	100.0%	100.0%	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881
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
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
DNASE1L3	118.0	99.9%	99.7%	Systemic lupus erythematosus 16, 614420

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
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
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
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
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
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
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
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
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
DYM	102.4	97.4%	95.7%	Smith-McCort dysplasia, 607326 Dyggve-Melchior-Clausen disease, 223800
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
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
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
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
EDNRB	126.2	96.4%	91.5%	ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580
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
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
EGR2	140.2	100.0%	100.0%	Dejerine-Sottas disease, 145900 Hypomyelinating neuropathy, congenital, 1, 605253 Charcot-Marie-Tooth disease, type 1D, 607678
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
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
ELMOD3	144.7	100.0%	99.8%	?Deafness, autosomal recessive 88, 615429
ELOVL4	103.3	100.0%	99.6%	Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
ELP1	118.4	99.9%	98.3%	Dysautonomia, familial, 223900
ELP2	118.6	99.8%	97.6%	Mental retardation, autosomal recessive 58, 617270
EMC1	111.0	100.0%	99.2%	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
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
ENAM	144.6	100.0%	100.0%	Amelogenesis imperfecta, type IC, 204650 Amelogenesis imperfecta, type IB, 104500
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
EPB41	122.1	99.6%	97.5%	Elliptocytosis-1, 611804

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
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
ERBB3	120.0	100.0%	99.3%	?Lethal congenital contractural syndrome 2, 607598
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
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

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
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
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
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
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
EYS	130.0	99.8%	97.3%	Retinitis pigmentosa 25, 602772
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
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
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
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
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
FASTKD2	116.5	99.9%	98.0%	?Mitochondrial complex IV deficiency, 220110
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
FBP1	114.0	100.0%	99.2%	Fructose-1,6-bisphosphatase deficiency, 229700
FBXL4	165.9	100.0%	100.0%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FBXO31	123.6	100.0%	98.7%	?Mental retardation, autosomal recessive 45, 615979
FBXO7	157.7	100.0%	99.5%	Parkinson disease 15, autosomal recessive, 260300
FCGR3A	184.5	99.7%	98.1%	Immunodeficiency 20, 615707
FCN3	135.3	100.0%	100.0%	Immunodeficiency due to ficolin 3 deficiency, 613860

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
FGD4	105.3	99.8%	98.3%	Charcot-Marie-Tooth disease, type 4H, 609311
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
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
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
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
FLG	158.7	100.0%	99.9%	Ichthyosis vulgaris, 146700
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
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
FOLR1	115.7	100.0%	100.0%	Neurodegeneration due to cerebral folate transport deficiency, 613068
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
FOXI1	209.3	100.0%	100.0%	Enlarged vestibular aqueduct, 600791
FOXN1	149.7	100.0%	99.8%	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
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
FRRS1L	100.0	89.3%	81.8%	Epileptic encephalopathy, early infantile, 37, 616981
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
FTCD	129.8	99.1%	96.1%	Glutamate formiminotransferase deficiency, 229100
FTO	99.1	83.8%	83.7%	Growth retardation, developmental delay, facial dysmorphism, 612938
FUCA1	135.9	100.0%	100.0%	Fucosidosis, 230000
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
FYCO1	135.2	100.0%	100.0%	Cataract 18, autosomal recessive, 610019

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
GAA	180.3	100.0%	100.0%	Glycogen storage disease II, 232300
GAB1	154.4	100.0%	99.8%	?Deafness, autosomal recessive 26, 605428
GAD1	114.6	100.0%	99.8%	?Cerebral palsy, spastic quadriplegic, 1, 603513
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
GAS2L2	183.2	100.0%	99.8%	?Ciliary dyskinesia, primary, 41, 618449
GAS8	134.7	100.0%	99.7%	Ciliary dyskinesia, primary, 33, 616726
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
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
GDF1	62.1	99.4%	91.2%	Right atrial isomerism (Ivemark), 208530 Congenital heart defects, multiple types, 6, 613854
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
GEMIN4	159.5	100.0%	99.8%	Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities, 617913
GFER	103.0	100.0%	99.9%	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076
GFM1	104.4	100.0%	98.9%	Combined oxidative phosphorylation deficiency 1, 609060
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
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
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
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
GLIS2	148.5	100.0%	100.0%	Nephronophthisis 7, 611498
GLIS3	133.1	100.0%	99.6%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
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
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
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

GNAT2	106.3	100.0%	98.5%	Achromatopsia 4, 613856
GNB3	159.3	100.0%	100.0%	Night blindness, congenital stationary, type 1H, 617024
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
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
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
GPC6	132.5	100.0%	100.0%	Omodysplasia 1, 258315
GPD1	94.7	100.0%	99.6%	Hypertriglyceridemia, transient infantile, 614480
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
GPR179	163.2	100.0%	100.0%	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
GPR88	172.4	100.0%	100.0%	?Chorea, childhood-onset, with psychomotor retardation, 616939
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
GRHPR	106.8	85.1%	82.3%	Hyperoxaluria, primary, type II, 260000
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
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
GSS	98.9	100.0%	99.6%	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900
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
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
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
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
HAX1	146.3	100.0%	100.0%	Neutropenia, severe congenital 3, autosomal recessive, 610738
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
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
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
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
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
HNMT	132.7	100.0%	99.7%	Mental retardation, autosomal recessive 51, 616739
HOGA1	163.0	100.0%	99.5%	Hyperoxaluria, primary, type III, 613616
HOXA1	184.7	100.0%	100.0%	Athabaskan brainstem dysgenesis syndrome, 601536 Bosley-Salih-Alorainy syndrome, 601536
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
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
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

HSD11B2	183.2	95.8%	89.4%	Apparent mineralocorticoid excess, 218030
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
HSPA9	83.8	88.2%	84.2%	Even-plus syndrome, 616854 Anemia, sideroblastic, 4, 182170
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
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
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
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
IDH3B	136.5	95.8%	95.4%	Retinitis pigmentosa 46, 612572
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
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
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
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
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
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
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
IL17RA	167.0	100.0%	100.0%	Immunodeficiency 51, 613953
IL17RC	139.8	100.0%	100.0%	Candidiasis, familial, 9, 616445
IL1RN	145.3	100.0%	99.9%	Interleukin 1 receptor antagonist deficiency, 612852
IL21R	160.2	100.0%	100.0%	Immunodeficiency 56, 615207
IL2RA	106.8	100.0%	99.2%	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367
IL36RN	102.7	100.0%	99.9%	Psoriasis 14, pustular, 614204
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
IMPG2	127.1	99.5%	98.0%	Macular dystrophy, vitelliform, 5, 616152 Retinitis pigmentosa 56, 613581
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
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
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
IRAK4	100.7	99.7%	94.9%	IRAK4 deficiency, 607676 Invasive pneumococcal disease, recurrent isolated, 1, 610799
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
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
ITPA	142.5	100.0%	100.0%	Epileptic encephalopathy, early infantile, 35, 616647
IVD	106.7	100.0%	100.0%	Isovaleric acidemia, 243500
IYD	108.7	99.3%	94.8%	Thyroid dyshormonogenesis 4, 274800
JAGN1	129.4	100.0%	100.0%	Neutropenia, severe congenital, 6, autosomal recessive, 616022
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
JUP	137.0	100.0%	99.9%	Arrhythmogenic right ventricular dysplasia 12, 611528 Naxos disease, 601214
KALRN	130.7	99.9%	99.5%	No OMIM Disease ID
KANK2	180.1	100.0%	100.0%	Nephrotic syndrome, type 16, 617783 Palmoplantar keratoderma and woolly hair, 616099
KARS	109.9	100.0%	99.3%	Deafness, autosomal recessive 89, 613916 ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641
KATNB1	170.5	100.0%	100.0%	Lissencephaly 6, with microcephaly, 616212
KCNE1	398.8	100.0%	100.0%	Long QT syndrome 5, 613695 Jervell and Lange-Nielsen syndrome 2, 612347
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
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
KCNV2	154.7	100.0%	100.0%	Retinal cone dystrophy 3B, 610356
KCTD7	171.1	95.0%	95.0%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
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
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
KIAA1109	137.8	99.8%	98.9%	Alkuraya-Kucinskas syndrome, 617822
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
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
KIF7	120.4	99.3%	96.6%	?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131
KISS1R	180.8	100.0%	100.0%	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 ?Precocious puberty, central, 1, 176400
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
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
KLK4	176.5	100.0%	99.8%	Amelogenesis imperfecta, type IIA1, 204700
KLKB1	132.4	99.9%	99.2%	Fletcher factor (prekallikrein) deficiency, 612423
KMT2B	156.7	98.1%	94.7%	Dystonia 28, childhood-onset, 617284
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
KRT10	137.0	100.0%	99.0%	Epidermolytic hyperkeratosis, 113800 Ichthyosis with confetti, 609165 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602

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
KRT18	38.7	91.9%	74.0%	Cirrhosis, cryptogenic, 215600
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
KRT8	40.9	91.3%	73.9%	Cirrhosis, cryptogenic, 215600
KRT85	112.7	99.1%	96.1%	Ectodermal dysplasia 4, hair/nail type, 602032
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
L2HGDH	123.6	99.2%	97.2%	L-2-hydroxyglutaric aciduria, 236792
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
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
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
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
LDHA	57.3	97.7%	89.0%	Glycogen storage disease XI, 612933
LDLRAP1	162.0	100.0%	99.9%	Hypercholesterolemia, familial, 4, 603813
LEMD2	117.7	100.0%	99.5%	Cataract 46, juvenile-onset, 212500
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
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
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
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
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
LMNB2	157.9	99.6%	98.3%	?Epilepsy, progressive myoclonic, 9, 616540
LMOD3	134.2	100.0%	99.9%	Nemaline myopathy 10, 616165
LONP1	164.8	100.0%	100.0%	CODAS syndrome, 600373
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
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
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
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
LRRC6	138.2	99.5%	96.2%	Ciliary dyskinesia, primary, 19, 614935
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
LTC4S	96.2	94.6%	79.7%	No OMIM Disease ID
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
LZTFL1	116.5	99.9%	99.2%	Bardet-Biedl syndrome 17, 615994
MAB21L2	265.2	100.0%	100.0%	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877
MAD2L2	150.7	100.0%	99.9%	?Fanconi anemia, complementation group V, 617243
MAG	175.3	100.0%	99.9%	Spastic paraplegia 75, autosomal recessive, 616680
MAGI2	93.7	95.1%	92.1%	Nephrotic syndrome, type 15, 617609
MAK	134.1	99.2%	96.5%	Retinitis pigmentosa 62, 614181
MALT1	128.7	93.6%	89.3%	Immunodeficiency 12, 615468
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
MAP3K20	110.7	99.9%	98.8%	Split-foot malformation with mesoaxial polydactyly, 616890 Centronuclear myopathy 6 with fiber-type disproportion, 617760
MAPKBP1	144.1	100.0%	100.0%	Nephronophthisis 20, 617271
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
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
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
MBOAT7	121.9	100.0%	99.9%	Mental retardation, autosomal recessive 57, 617188
MC2R	160.2	100.0%	99.4%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
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
MCM4	141.3	100.0%	99.5%	Immunodeficiency 54, 609981
MCM5	131.9	100.0%	100.0%	?Meier-Gorlin syndrome 8, 617564

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
MECR	114.2	100.0%	99.7%	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
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
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
MEOX1	114.4	100.0%	98.8%	Klippel-Feil syndrome 2, 214300
MERTK	133.5	99.5%	99.0%	Retinitis pigmentosa 38, 613862
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
METTL23	117.0	100.0%	100.0%	Mental retardation, autosomal recessive 44, 615942
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
MICU1	103.4	98.7%	96.2%	Myopathy with extrapyramidal signs, 615673
MIPEP	100.6	99.8%	96.1%	Combined oxidative phosphorylation deficiency 31, 617228
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
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
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
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
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
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
MOGS	157.9	100.0%	100.0%	Congenital disorder of glycosylation, type IIb, 606056
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
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
MRAP	173.5	100.0%	100.0%	Glucocorticoid deficiency 2, 607398
MRE11	48.1	97.7%	83.5%	Ataxia-telangiectasia-like disorder 1, 604391
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
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
MSH6	171.5	100.0%	99.9%	Mismatch repair cancer syndrome, 276300 Colorectal cancer, hereditary nonpolyposis, type 5, 614350
MSMO1	47.0	96.2%	87.7%	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MSRB3	138.7	99.9%	99.3%	Deafness, autosomal recessive 74, 613718
MSTO1	107.9	99.5%	97.1%	Myopathy, mitochondrial, and ataxia, 617675

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
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
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
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
MVK	130.3	90.5%	90.4%	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377
MYBPC1	129.2	100.0%	99.4%	Arthrogryposis, distal, type 1B, 614335 Myopathy, congenital, with tremor, 618524 Lethal congenital contracture syndrome 4, 614915
MYD88	219.1	100.0%	99.9%	Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260 Macroglobulinemia, Waldenstrom, somatic, 153600
MYH2	113.6	100.0%	99.7%	Proximal myopathy and ophthalmoplegia, 605637
MYL3	106.8	100.0%	100.0%	Cardiomyopathy, hypertrophic, 8, 608751
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
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
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
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
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
NDE1	95.8	100.0%	99.8%	Lissencephaly 4 (with microcephaly), 614019 ?Microhydranencephaly, 605013
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
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
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
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
NEUROG3	205.9	100.0%	100.0%	Diarrhea 4, malabsorptive, congenital, 610370
NFASC	128.3	100.0%	99.8%	Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356
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
NHP2	135.0	100.0%	99.8%	Dyskeratosis congenita, autosomal recessive 2, 613987
NIN	130.6	99.9%	99.2%	?Seckel syndrome 7, 614851
NIPAL4	137.3	100.0%	99.7%	Ichthyosis, congenital, autosomal recessive 6, 612281
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
NLRP1	126.8	99.5%	97.9%	Palmoplantar carcinoma, multiple self-healing, 615225 Autoinflammation with arthritis and dyskeratosis, 617388
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
NOP10	124.6	100.0%	100.0%	Dyskeratosis congenita, autosomal recessive 1, 224230
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
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
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
NRXN1	147.9	97.6%	97.3%	Pitt-Hopkins-like syndrome 2, 614325
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
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
NTHL1	134.4	100.0%	100.0%	Familial adenomatous polyposis 3, 616415
NTRK1	144.9	100.0%	99.7%	Medullary thyroid carcinoma, familial, 155240 Insensitivity to pain, congenital, with anhidrosis, 256800
NUBPL	102.4	98.9%	94.2%	Mitochondrial complex I deficiency, nuclear type 21, 618242
NUP107	126.3	99.9%	98.0%	Galloway-Mowat syndrome 7, 618348 ?Ovarian dysgenesis 6, 618078 Nephrotic syndrome, type 11, 616730
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
NUP62	124.5	100.0%	100.0%	Striatonigral degeneration, infantile, 271930
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
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
OGDH	181.7	100.0%	99.6%	No OMIM disease ID
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
OPLAH	158.6	100.0%	100.0%	5-oxoprolinase deficiency, 260005
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
OSGEP	104.4	100.0%	97.8%	Galloway-Mowat syndrome 3, 617729
OSTM1	113.8	97.8%	92.1%	Osteopetrosis, autosomal recessive 5, 259720
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
OXCT1	123.6	99.7%	98.2%	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050
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
PAH	128.9	100.0%	100.0%	Phenylketonuria, 261600
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
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

PATL2	100.1	99.8%	96.3%	Oocyte maturation defect 4, 617743
PAX1	212.4	98.6%	94.1%	?Otofaciocervical syndrome 2, 615560
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
PAX7	147.9	100.0%	100.0%	Myopathy, congenital, progressive, with scoliosis, 618578 Rhabdomyosarcoma 2, alveolar, 268220
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
PCK1	128.7	100.0%	100.0%	?Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680
PCK2	177.6	100.0%	100.0%	No OMIM disease ID
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
PCYT1A	97.1	99.1%	95.7%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDE10A	107.4	81.5%	80.3%	Dyskinesia, limb and orofacial, infantile-onset, 616921 Striatal degeneration, autosomal dominant, 616922
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
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
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
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
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
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
PGM1	134.8	100.0%	99.8%	Congenital disorder of glycosylation, type It, 614921
PGM3	148.4	100.0%	99.9%	Immunodeficiency 23, 615816
PHGDH	116.2	100.0%	99.6%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
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
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
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
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
PIGT	169.4	98.1%	98.1%	?Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
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
PIK3R1	125.7	99.9%	98.8%	SHORT syndrome, 269880 Immunodeficiency 36, 616005 ?Agammaglobulinemia 7, autosomal recessive, 615214
PIK3R5	130.3	100.0%	100.0%	Ataxia-oculomotor apraxia 3, 615217
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
PKD1L1	113.1	100.0%	99.5%	Heterotaxy, visceral, 8, autosomal, 617205
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
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
PLCB1	134.5	100.0%	99.8%	Epileptic encephalopathy, early infantile, 12, 613722
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
PLD1	116.6	100.0%	99.6%	Cardiac valvular defect, developmental, 212093
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
PLK4	145.8	99.9%	98.1%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
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
PLPBP	97.2	99.9%	97.2%	Epilepsy, early-onset, vitamin B6-dependent, 617290
PMM2	130.3	100.0%	99.7%	Congenital disorder of glycosylation, type Ia, 212065
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

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
POGLUT1	100.2	100.0%	99.1%	?Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232 Dowling-Degos disease 4, 615696
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
POLH	121.6	100.0%	99.2%	Xeroderma pigmentosum, variant type, 278750
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
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
POU1F1	109.5	99.9%	98.2%	Pituitary hormone deficiency, combined, 1, 613038
PPA2	92.7	98.0%	90.2%	?Sudden cardiac failure, alcohol-induced, 617223 Sudden cardiac failure, infantile, 617222
PPIB	118.3	100.0%	100.0%	Osteogenesis imperfecta, type IX, 259440
PPP1R15B	130.2	100.0%	100.0%	Microcephaly, short stature, and impaired glucose metabolism 2, 616817
PPT1	140.2	90.3%	89.2%	Ceroid lipofuscinosis, neuronal, 1, 256730
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
PRDM5	136.5	99.9%	98.2%	Brittle cornea syndrome 2, 614170
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
PRKCD	177.4	100.0%	100.0%	Autoimmune lymphoproliferative syndrome, type III, 615559
PRKDC	98.5	99.4%	96.4%	Immunodeficiency 26, with or without neurologic abnormalities, 615966
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
PRMT7	128.8	100.0%	99.9%	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157

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
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
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
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
PSPH	122.1	100.0%	99.8%	Phosphoserine phosphatase deficiency, 614023
PTF1A	138.4	100.0%	99.7%	Pancreatic and cerebellar agenesis, 609069 Pancreatic agenesis 2, 615935
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
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
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
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
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
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
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
RAD50	100.6	96.4%	89.5%	Nijmegen breakage syndrome-like disorder, 613078
RAD51C	141.9	100.0%	99.7%	Fanconi anemia, complementation group O, 613390
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
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
RAX	156.7	100.0%	99.9%	Microphthalmia, isolated 3, 611038

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
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
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
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
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
RELB	120.3	99.7%	96.5%	?Immunodeficiency 53, 617585
RELN	131.8	100.0%	99.6%	Lissencephaly 2 (Norman-Roberts type), 257320
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
RETREG1	132.5	100.0%	98.6%	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
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

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
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
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
RLBP1	129.0	100.0%	99.9%	Fundus albipunctatus, 136880 Bothnia retinal dystrophy, 607475 Retinitis punctata albescens, 136880 Newfoundland rod-cone dystrophy, 607476
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
RNASET2	109.5	95.4%	89.8%	Leukoencephalopathy, cystic, without megalencephaly, 612951
RNF168	187.1	100.0%	99.4%	RIDDLE syndrome, 611943
RNF216	128.1	100.0%	98.3%	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840
ROBO3	114.9	99.7%	98.0%	Gaze palsy, familial horizontal, with progressive scoliosis, 1, 607313
ROGDI	141.6	100.0%	99.9%	Kohlschutter-Tonz syndrome, 226750
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
RORC	132.1	100.0%	100.0%	Immunodeficiency 42, 616622
RP1	112.2	91.5%	91.0%	Retinitis pigmentosa 1, 180100
RPE65	133.0	100.0%	99.8%	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794
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
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
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
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
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
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
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
SALL2	143.2	100.0%	100.0%	?Coloboma, ocular, autosomal recessive, 216820
SAMD9	161.7	100.0%	100.0%	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455
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
SASS6	73.5	99.9%	98.1%	?Microcephaly 14, primary, autosomal recessive, 616402
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
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
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
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
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
SECISBP2	109.6	99.5%	96.6%	Thyroid hormone metabolism, abnormal, 609698
SELENON	143.9	85.3%	84.0%	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310
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
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
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
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
SETX	153.0	100.0%	99.7%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 Amyotrophic lateral sclerosis 4, juvenile, 602433
SFRP4	136.0	100.0%	99.1%	Pyle disease, 265900
SFTPB	101.4	100.0%	99.7%	Surfactant metabolism dysfunction, pulmonary, 1, 265120
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
SGCG	117.7	100.0%	100.0%	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700
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
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
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
SIL1	138.5	99.4%	96.7%	Marinesco-Sjogren syndrome, 248800
SIX6	303.9	100.0%	100.0%	Optic disc anomalies with retinal and/or macular dystrophy, 212550
SKIV2L	150.1	100.0%	99.9%	Trichohepatoenteric syndrome 2, 614602
SLC10A2	124.2	100.0%	99.9%	Bile acid malabsorption, primary, 613291
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
SLC13A5	155.1	100.0%	99.9%	Epileptic encephalopathy, early infantile, 25, 615905
SLC17A5	136.7	98.7%	95.1%	Sialic acid storage disorder, infantile, 269920 Salla disease, 604369
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
SLC1A4	159.5	100.0%	99.8%	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC22A12	131.0	100.0%	99.9%	Hypouricemia, renal, 220150
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
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
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
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
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

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
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
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
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
SLC39A8	144.7	100.0%	99.8%	Congenital disorder of glycosylation, type IIn, 616721
SLC3A1	147.7	100.0%	99.7%	Cystinuria, 220100
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
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
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
SLC6A3	142.7	100.0%	99.9%	Parkinsonism-dystonia, infantile, 1, 613135
SLC6A5	137.1	100.0%	99.9%	Hyperekplexia 3, 614618
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
SLCO2A1	104.4	100.0%	98.7%	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
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
SMARCAL1	119.6	100.0%	99.8%	Schimke immunoosseous dysplasia, 242900
SMARCD2	99.5	87.5%	86.1%	Specific granule deficiency 2, 617475
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

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
SNAI2	106.3	100.0%	99.1%	Waardenburg syndrome, type 2D, 608890 Piebaldism, 172800
SNAP29	182.5	100.0%	100.0%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNIP1	140.1	100.0%	99.6%	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501
SNORD118	NC	NC	NC	Leukoencephalopathy, brain calcifications, and cysts, 614561
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
SOST	207.3	100.0%	99.5%	Sclerosteosis 1, 269500 Van Buchem disease, 239100 Craniodiaphyseal dysplasia, autosomal dominant, 122860
SOX18	59.1	97.4%	85.4%	Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940 Hypotrichosis-lymphedema-telangiectasia syndrome, 607823
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
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
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
SPINK5	128.1	100.0%	99.2%	Netherton syndrome, 256500
SPINT2	71.9	99.8%	93.0%	Diarrhea 3, secretory sodium, congenital, syndromic, 270420
SPR	159.7	100.0%	100.0%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRTN	164.4	100.0%	100.0%	Ruijs-Aalfs syndrome, 616200

SPTA1	107.6	99.9%	98.8%	Pyropoikilocytosis, 266140 Elliptocytosis-2, 130600 Spherocytosis, type 3, 270970
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
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
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
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
STAMBP	96.0	99.9%	97.6%	Microcephaly-capillary malformation syndrome, 614261
STAR	146.4	100.0%	100.0%	Lipoid adrenal hyperplasia, 201710
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
STAT5B	119.7	99.9%	98.8%	Leukemia, acute promyelocytic, somatic, 102578 Growth hormone insensitivity with immunodeficiency, 245590
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
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
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
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
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
SULT2B1	135.3	100.0%	100.0%	Ichthyosis, congenital, autosomal recessive 14, 617571
SUMF1	91.7	99.9%	97.6%	Multiple sulfatase deficiency, 272200
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
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
SYNE4	92.6	100.0%	99.2%	Deafness, autosomal recessive 76, 615540
SYNJ1	126.3	99.9%	98.5%	Epileptic encephalopathy, early infantile, 53, 617389 Parkinson disease 20, early-onset, 615530
SYT14	103.1	60.6%	58.1%	?Spinocerebellar ataxia, autosomal recessive 11, 614229
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
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
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
TAF6	138.7	100.0%	99.5%	Alazami-Yuan syndrome, 617126
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
TARS2	93.9	99.7%	97.1%	?Combined oxidative phosphorylation deficiency 21, 615918
TAT	119.7	100.0%	99.9%	Tyrosinemia, type II, 276600
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
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
TBX15	114.2	100.0%	100.0%	Cousin syndrome, 260660
TBX19	168.3	100.0%	100.0%	Adrenocorticotropic hormone deficiency, 201400
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
TCIRG1	149.6	99.6%	98.0%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	157.6	100.0%	100.0%	Transcobalamin II deficiency, 275350
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
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

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
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
TF	106.6	100.0%	99.8%	Atransferrinemia, 209300
TFAM	67.1	91.8%	70.6%	?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156
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
TGM1	153.3	100.0%	100.0%	Ichthyosis, congenital, autosomal recessive 1, 242300
TGM5	152.1	100.0%	99.9%	Peeling skin syndrome 2, 609796
TH	106.8	100.0%	99.2%	Segawa syndrome, recessive, 605407
THOC6	253.7	100.0%	100.0%	Beaulieu-Boycott-Innes syndrome, 613680
THRB	146.5	100.0%	99.3%	Thyroid hormone resistance, 188570 Thyroid hormone resistance, selective pituitary, 145650 Thyroid hormone resistance, autosomal recessive, 274300
TIMM50	133.8	100.0%	99.4%	3-methylglutaconic aciduria, type IX, 617698
TIMMDC1	167.0	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 31, 618251
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
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

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
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
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
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
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
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
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
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
TNNI3	112.2	99.6%	96.4%	Cardiomyopathy, hypertrophic, 7, 613690 ?Cardiomyopathy, dilated, 2A, 611880

				Cardiomyopathy, dilated, 1FF, 613286 Cardiomyopathy, familial restrictive, 1, 115210
TNNT1	113.8	100.0%	99.4%	Nemaline myopathy 5, Amish type, 605355
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
TOR1AIP1	142.0	99.2%	97.1%	?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072
TP53RK	93.0	99.8%	97.7%	Galloway-Mowat syndrome 4, 617730
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
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
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
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
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
TRH	130.3	99.9%	98.4%	No OMIM disease ID
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
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
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
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
TRPM1	134.7	100.0%	99.3%	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TRPM6	128.5	99.9%	99.0%	Hypomagnesemia 1, intestinal, 602014
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
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
TSPAN12	132.9	100.0%	99.8%	Exudative vitreoretinopathy 5, 613310
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
TTC19	84.9	98.8%	86.6%	Mitochondrial complex III deficiency, nuclear type 2, 615157
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
TTPA	109.2	99.3%	95.6%	Ataxia with isolated vitamin E deficiency, 277460
TUBA8	136.6	100.0%	99.5%	Cortical dysplasia, complex, with other brain malformations 8, 613180
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
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
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
UBA5	79.3	96.9%	83.9%	?Spinocerebellar ataxia, autosomal recessive 24, 617133 Epileptic encephalopathy, early infantile, 44, 617132
UBE2T	92.1	100.0%	99.4%	Fanconi anemia, complementation group T, 616435
UBE3B	119.7	100.0%	99.8%	Kaufman oculocerebrofacial syndrome, 244450
UBR1	118.2	99.9%	98.9%	Johanson-Blizzard syndrome, 243800
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

UGT1A1	192.9	100.0%	100.0%	Hyperbilirubinemia, familial transient neonatal, 237900 Crigler-Najjar syndrome, type I, 218800 Crigler-Najjar syndrome, type II, 606785
UMPS	156.7	100.0%	98.8%	Orotic aciduria, 258900
UNC13D	120.2	99.9%	99.2%	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
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
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
UVSSA	140.1	99.6%	99.5%	UV-sensitive syndrome 3, 614640
VAC14	107.4	100.0%	98.8%	Striatonigral degeneration, childhood-onset, 617054
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
VDR	116.5	99.0%	96.4%	Rickets, vitamin D-resistant, type IIA, 277440
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
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
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
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
VPS53	117.2	91.3%	89.9%	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	126.7	99.9%	98.3%	Pontocerebellar hypoplasia type 1A, 607596
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
WARS2	142.1	99.9%	99.1%	Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710
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
WDPCP	105.7	97.1%	93.6%	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
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
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
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

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
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
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
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
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
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
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
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
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
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
ZBTB16	161.1	100.0%	100.0%	Skeletal defects, genital hypoplasia, and mental retardation, 612447 Leukemia, acute promyelocytic, PL2F/RARA type, 0
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
ZFYVE26	110.8	99.9%	99.0%	Spastic paraplegia 15, autosomal recessive, 270700
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
ZNF335	147.5	100.0%	99.9%	Microcephaly 10, primary, autosomal recessive, 615095
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
ZNF469	180.5	100.0%	100.0%	Brittle cornea syndrome 1, 229200
ZNF513	153.8	100.0%	100.0%	?Retinitis pigmentosa 58, 613617
ZNHIT3	141.9	74.4%	74.4%	PEHO syndrome, 260565
ZP1	194.3	100.0%	100.0%	Oocyte maturation defect 1, 615774

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