PRE CONCEPTION GENE PANEL DG 3.2.0 (2363 genes)

Releasedate: 16-09-2021

Gene	Agilent V5 covered >10x	Agilent V5 covered >20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype Description and OMIM disease ID
AAAS	100	99,4	100	100	Achalasia-addisonianism-alacrimia syndrome, 231550
AARS1	100	99,7	100	99,9	Developmental and epileptic encephalopathy 29, 616339 Charcot-Marie-Tooth disease, axonal, type 2N, 613287
AARS2	100	99,4	100	100	Leukoencephalopathy, progressive, with ovarian failure, 615889 Combined oxidative phosphorylation deficiency 8, 614096
AASS	99,9	99,8	100	99,9	Hyperlysinemia, 238700
ABAT	99,9	97,8	100	100	GABA-transaminase deficiency, 613163
ABCA1	99,8	97,8	100	100	Tangier disease, 205400 HDL deficiency, familial, 1, 604091
ABCA3	99,8	99	100	100	Surfactant metabolism dysfunction, pulmonary, 3, 610921
ABCA4	99,9	99	96,5	96,5	Retinal dystrophy, early-onset severe, 248200 Retinitis pigmentosa 19, 601718 Cone-rod dystrophy 3, 604116 Fundus flavimaculatus, 248200 Stargardt disease 1, 248200
ABCB11	100	99,4	100	100	Cholestasis, benign recurrent intrahepatic, 2, 605479 Cholestasis, progressive familial intrahepatic 2, 601847
ABCB4	99,9	99,3	100	100	Gallbladder disease 1, 600803 Cholestasis, intrahepatic, of pregnancy, 3, 614972 Cholestasis, progressive familial intrahepatic 3, 602347
ABCC2	100	99,9	100	100	Dubin-Johnson syndrome, 237500
ABCC6	93,6	92,5	100	100	Pseudoxanthoma elasticum, 264800 Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, forme fruste, 177850
ABCC8	100	99,5	100	100	Diabetes mellitus, permanent neonatal 3, with or without neurologic features, 618857 Diabetes mellitus, transient neonatal 2, 610374 Diabetes mellitus, noninsulin-dependent, 125853 Hypoglycemia of infancy, leucine-sensitive, 240800 Hyperinsulinemic hypoglycemia, familial, 1, 256450
ABCD3	99,4	98	100	100	?Bile acid synthesis defect, congenital, 5, 616278
ABCD4	99,8	97,7	100	100	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	99,9	99,9	100	100	Sitosterolemia 2, 618666

ABCG8	99	97,1	100	100	Sitosterolemia 1, 210250
ABHD12	91,7	86	100	99,8	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ABHD5	100	100	100	100	Chanarin-Dorfman syndrome, 275630
ACACA	98,4	97,9	100	100	No OMIM disease ID
ACAD8	100	100	100	100	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	100	99,8	100	100	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADM	99,8	97,9	100	100	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	100	99,4	100	100	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	99,8	97,9	100	100	2-methylbutyrylglycinuria, 610006
ACADVL	99,7	96,6	100	100	VLCAD deficiency, 201475
ACAN	96,9	92,5	98,9	98,7	?Spondyloepiphyseal dysplasia, Kimberley type, 608361 Short stature and advanced bone age, with or without early-onset osteoarthritis and/or osteochondritis dissecans, 165800 Spondyloepimetaphyseal dysplasia, aggrecan type, 612813
ACAT1	99,6	97,9	100	99,7	Alpha-methylacetoacetic aciduria, 203750
ACD	100	99,9	100	100	?Dyskeratosis congenita, autosomal recessive 7, 616553 ?Dyskeratosis congenita, autosomal dominant 6, 616553
ACE	99,9	98,4	100	99,9	Renal tubular dysgenesis, 267430
ACER3	99,5	99	100	100	?Leukodystrophy, progressive, early childhood-onset, 617762
ACO2	94,1	86,3	100	100	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559
ACOX1	100	99,3	100	100	Mitchell syndrome, 618960 Peroxisomal acyl-CoA oxidase deficiency, 264470
ACOX2	100	99	100	100	Bile acid synthesis defect, congenital, 6, 617308
ACP5	99,9	98,9	100	100	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACSF3	100	99,5	100	100	Combined malonic and methylmalonic aciduria, 614265
ACTA1	98,2	89,5	100	100	?Myopathy, scapulohumeroperoneal, 616852 Nemaline myopathy 3, autosomal dominant or recessive, 161800 Myopathy, actin, congenital, with excess of thin myofilaments, 161800 Myopathy, actin, congenital, with cores, 161800 Myopathy, congenital, with fiber-type disproportion 1, 255310
ACTL6B	100	100	100	100	Developmental and epileptic encephalopathy 76, 618468 Intellectual developmental disorder with severe speech and ambulation defects, 618470

ACY1	100	99,7	100	100	Aminoacylase 1 deficiency, 609924
ADA	99,7	96,1	100	100	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADA2	99,9	97,3	100	100	Sneddon syndrome, 182410 Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688
ADAM17	99,6	98	100	99,9	?Inflammatory skin and bowel disease, neonatal, 1, 614328
ADAM22	99,8	99,6	100	100	Developmental and epileptic encephalopathy 61, 617933
ADAM9	99,7	98	100	100	Cone-rod dystrophy 9, 612775
ADAMTS10	100	99,9	100	100	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS13	97	94,3	99,8	98,9	Thrombotic thrombocytopenic purpura, hereditary, 274150
ADAMTS17	92,9	88,8	97,9	95,9	Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTS18	100	99,6	100	100	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
ADAMTS2	99,8	97,5	98	97,7	Ehlers-Danlos syndrome, dermatosparaxis type, 225410
ADAMTS3	100	100	100	100	Hennekam lymphangiectasia-lymphedema syndrome 3, 618154
ADAMTSL2	98	94,7	99,9	99,8	Geleophysic dysplasia 1, 231050
ADAMTSL4	99,8	98,4	100	100	Ectopia lentis et pupillae, 225200 Ectopia lentis, isolated, autosomal recessive, 225100
ADAR	100	99,4	100	100	Dyschromatosis symmetrica hereditaria, 127400 Aicardi-Goutieres syndrome 6, 615010
ADARB1	97,2	95,3	95,1	95,1	Neurodevelopmental disorder with hypotonia, microcephaly, and seizures, 618862
ADAT3	100	100	100	100	Neurodevelopmental disorder with brain abnormalities, poor growth, and dysmorphic facies, 615286
ADCY1	95,7	94,3	98,4	97,7	?Deafness, autosomal recessive 44, 610154
ADCY6	100	100	100	100	Lethal congenital contracture syndrome 8, 616287
ADD3	99,8	98,8	100	100	Cerebral palsy, spastic quadriplegic, 3, 617008
ADGRG1	100	100	100	100	Polymicrogyria, bilateral frontoparietal, 606854 Polymicrogyria, bilateral perisylvian, 615752
ADGRG6	99,7	98,7	100	100	Lethal congenital contracture syndrome 9, 616503
ADGRV1	99,5	98,4	100	100	Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472 ?Febrile seizures, familial, 4, 604352
ADK	83,3	79,7	84,5	84,5	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADPRS	100	99,9	100	100	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170

ADSL	99,2	98,6	100	100	Adenylosuccinase deficiency, 103050
ADSS1	90,8	87	100	100	Myopathy, distal, 5, 617030
AEBP1	100	100	100	100	Ehlers-Danlos syndrome, classic-like, 2, 618000
AFG3L2	94,6	86,3	100	100	Spastic ataxia 5, autosomal recessive, 614487 Optic atrophy 12, 618977 Spinocerebellar ataxia 28, 610246
AGA	100	99,9	100	100	Aspartylglucosaminuria, 208400
AGBL5	99,6	98,4	100	100	Retinitis pigmentosa 75, 617023
AGK	90,4	87,9	91,2	91,1	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AGL	99,8	99,5	100	100	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGPAT2	99	94	100	100	Lipodystrophy, congenital generalized, type 1, 608594
AGPS	98,8	95,2	100	99,4	Rhizomelic chondrodysplasia punctata, type 3, 600121
AGRN	97,6	92,6	100	99,9	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120
AGT	100	99,9	100	100	Renal tubular dysgenesis, 267430
AGTPBP1	96,3	94,2	100	100	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276
AGTR1	91,9	91,8	100	100	Renal tubular dysgenesis, 267430
AGXT	100	100	100	100	Hyperoxaluria, primary, type 1, 259900
AHCY	99,9	98,8	100	100	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHI1	99,4	97,4	100	100	Joubert syndrome 3, 608629
AHR	99,2	98,6	100	100	?Retinitis pigmentosa 85, 618345
AHSG	100	99,8	100	100	?Alopecia-mental retardation syndrome 1, 203650
AICDA	100	99,9	100	100	Immunodeficiency with hyper-IgM, type 2, 605258
AIMP1	99,2	92,5	100	99,9	Leukodystrophy, hypomyelinating, 3, 260600
AIMP2	89,4	86	100	99,9	Leukodystrophy, hypomyelinating, 17, 618006
AIPL1	100	99,2	100	100	Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393 Cone-rod dystrophy, 604393
AIRE	100	99,9	100	100	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AK1	100	100	100	100	Hemolytic anemia due to adenylate kinase deficiency, 612631

AK2	98,7	95,2	100	99,7	Reticular dysgenesis, 267500
AKR1D1	99,8	98,6	100	100	Bile acid synthesis defect, congenital, 2, 235555
ALAD	97,8	92,5	100	100	Porphyria, acute hepatic, 612740
ALB	99,8	99,4	100	100	Analbuminemia, 616000
ALDH18A1	100	99,9	100	100	Spastic paraplegia 9A, autosomal dominant, 601162 Cutis laxa, autosomal recessive, type IIIA, 219150 Spastic paraplegia 9B, autosomal recessive, 616586 Cutis laxa, autosomal dominant 3, 616603
ALDH1A3	97,6	94,6	100	100	Microphthalmia, isolated 8, 615113
ALDH3A2	88,8	88,4	93,2	93,2	Sjogren-Larsson syndrome, 270200
ALDH4A1	100	99,7	100	100	Hyperprolinemia, type II, 239510
ALDH5A1	92,4	83,5	100	100	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	100	99,6	100	100	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	91,1	84,5	100	100	Epilepsy, pyridoxine-dependent, 266100
ALDOA	98,9	96,3	100	100	Glycogen storage disease XII, 611881
ALDOB	98,8	95,7	100	100	Fructose intolerance, hereditary, 229600
ALG1	53,6	46,9	100	100	Congenital disorder of glycosylation, type Ik, 608540
ALG11	96,8	96,8	96,8	96,8	Congenital disorder of glycosylation, type Ip, 613661
ALG12	100	99,9	100	100	Congenital disorder of glycosylation, type Ig, 607143
ALG14	100	99,9	100	100	Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies, 619031 Myopathy, epilepsy, and progressive cerebral atrophy, 619036 ?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227
ALG2	100	100	100	100	?Congenital disorder of glycosylation, type Ii, 607906 Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228
ALG3	100	99,5	100	100	Congenital disorder of glycosylation, type Id, 601110
ALG6	98,2	93,7	100	99,9	Congenital disorder of glycosylation, type Ic, 603147
ALG8	96,6	95,9	96,6	96,6	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	99,9	99,3	100	100	Gillessen-Kaesbach-Nishimura syndrome, 263210 Congenital disorder of glycosylation, type Il, 608776
ALKBH8	99,7	98,6	100	100	Intellectual developmental disorder, autosomal recessive 71, 618504
ALMS1	99,7	99,5	100	100	Alstrom syndrome, 203800

ALOX12B	100	99,7	100	100	Ichthyosis, congenital, autosomal recessive 2, 242100
ALOXE3	99,9	99	100	100	Ichthyosis, congenital, autosomal recessive 3, 606545
ALPK3	98,1	95,1	100	100	Cardiomyopathy, familial hypertrophic 27, 618052
ALPL	100	99,4	100	100	Odontohypophosphatasia, 146300 Hypophosphatasia, infantile, 241500 Hypophosphatasia, childhood, 241510 Hypophosphatasia, adult, 146300
ALS2	99,9	99,8	100	100	Primary lateral sclerosis, juvenile, 606353 Spastic paralysis, infantile onset ascending, 607225 Amyotrophic lateral sclerosis 2, juvenile, 205100
ALX1	99,6	95,2	100	100	Frontonasal dysplasia 3, 613456
ALX3	80,2	72,8	100	100	Frontonasal dysplasia 1, 136760
ALX4	100	99,9	100	100	Parietal foramina 2, 609597 Frontonasal dysplasia 2, 613451
AMACR	100	100	100	100	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
AMBN	99,6	97,7	100	99,9	Amelogenesis imperfecta, type IF, 616270
AMN	92,5	82,9	100	100	Imerslund-Grasbeck syndrome 2, 618882
AMPD1	100	99,5	100	100	Myopathy due to myoadenylate deaminase deficiency, 615511
AMPD2	99,8	99	100	100	?Spastic paraplegia 63, 615686 Pontocerebellar hypoplasia, type 9, 615809
AMT	100	100	100	100	Glycine encephalopathy, 605899
ANAPC1	58,8	56,8	100	99,9	Rothmund-Thomson syndrome, type 1, 618625
ANGPTL3	98,6	92	100	100	Hypobetalipoproteinemia, familial, 2, 605019
ANK1	99,9	98,7	100	100	Spherocytosis, type 1, 182900
ANK3	99,3	99	100	100	Mental retardation, autosomal recessive, 37, 615493
ANKH	100	99,9	100	100	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000
ANKLE2	100	99,3	100	99,7	Microcephaly 16, primary, autosomal recessive, 616681
ANKS6	94,2	89,7	97	95	Nephronophthisis 16, 615382
ANO10	99,2	96,6	100	100	Spinocerebellar ataxia, autosomal recessive 10, 613728

ANO5	99,2	96,9	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307 Miyoshi muscular dystrophy 3, 613319 Gnathodiaphyseal dysplasia, 166260
ANO6	99,2	96,8	100	100	Scott syndrome, 262890
ANTXR1	99,9	99,1	100	100	GAPO syndrome, 230740
ANTXR2	99,5	97,2	100	100	Hyaline fibromatosis syndrome, 228600
AP1B1	100	99,4	100	100	Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150
AP1S1	99,9	99,4	100	100	MEDNIK syndrome, 609313
AP3B1	99,2	96,4	100	99,9	Hermansky-Pudlak syndrome 2, 608233
AP3B2	93,3	89,8	99,9	99	Developmental and epileptic encephalopathy 48, 617276
AP3D1	99,6	98,4	100	100	?Hermansky-Pudlak syndrome 10, 617050
AP4B1	99,8	98,5	100	100	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	99,5	98,6	100	99,9	Stuttering, familial persistent, 1, 184450 Spastic paraplegia 51, autosomal recessive, 613744
AP4M1	99,8	97,8	100	100	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	78,9	71	87,9	87,9	Spastic paraplegia 52, autosomal recessive, 614067
AP5Z1	100	99,9	100	100	Spastic paraplegia 48, autosomal recessive, 613647
APC2	98,3	94,8	99,7	98,5	Cortical dysplasia, complex, with other brain malformations 10, 618677 ?Sotos syndrome 3, 617169
APOC2	100	100	100	100	Hyperlipoproteinemia, type Ib, 207750
APOE	100	98,8	100	100	Alzheimer disease 2, 104310 Sea-blue histiocyte disease, 269600 Lipoprotein glomerulopathy, 611771 Hyperlipoproteinemia, type III, 617347
APRT	100	100	100	100	Adenine phosphoribosyltransferase deficiency, 614723
APTX	94,1	90,6	100	100	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
AQP2	100	99,2	100	100	Diabetes insipidus, nephrogenic, 2, 125800
ARFGEF2	99,7	98,7	100	100	Periventricular heterotopia with microcephaly, 608097
ARG1	92,9	92,9	92,9	92,7	Argininemia, 207800
ARHGDIA	100	100	100	100	Nephrotic syndrome, type 8, 615244
ARHGEF18	98,8	93,9	100	100	Retinitis pigmentosa 78, 617433

ARHGEF2	93	92,6	100	100	?Neurodevelopmental disorder with midbrain and hindbrain malformations, 617523
ARL13B	100	99,3	100	100	Joubert syndrome 8, 612291
ARL2BP	94,9	87,3	100	100	Retinitis pigmentosa with or without situs inversus, 615434
ARL3	99,7	96,5	100	100	Retinitis pigmentosa 83, 618173 Joubert syndrome 35, 618161
ARL6	99,1	98,4	100	100	Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151
ARL6IP1	98,5	81,5	100	100	?Spastic paraplegia 61, autosomal recessive, 615685
ARMC9	99,9	99,4	100	100	Joubert syndrome 30, 617622
ARNT2	100	100	100	99,5	?Webb-Dattani syndrome, 615926
ARPC1B	100	100	100	100	Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia, 617718
ARSA	100	99,8	100	100	Metachromatic leukodystrophy, 250100
ARSB	98,8	91	100	100	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARSG	99,9	98	100	100	Usher syndrome, type IV, 618144
ARV1	99,9	98,8	100	99,9	Developmental and epileptic encephalopathy 38, 617020
ASAH1	99,1	97,3	100	100	Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 Farber lipogranulomatosis, 228000
ASCC1	92,5	89,4	87,1	87	Spinal muscular atrophy with congenital bone fractures 2, 616867 Barrett esophagus/esophageal adenocarcinoma, 614266
ASL	100	99,7	100	100	Argininosuccinic aciduria, 207900
ASNS	98,1	91,2	100	100	Asparagine synthetase deficiency, 615574
ASPA	99,9	99,1	100	100	Canavan disease, 271900
ASPH	99,6	98,6	100	99,9	Traboulsi syndrome, 601552
ASPM	99,4	97,9	100	99,9	Microcephaly 5, primary, autosomal recessive, 608716
ASS1	93,2	83,2	100	100	Citrullinemia, 215700
ATAD1	99,1	91,6	100	100	Hyperekplexia 4, 618011
ATAD3A	91,4	86,7	100	100	Harel-Yoon syndrome, 617183 Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810
ATCAY	100	100	100	100	Ataxia, cerebellar, Cayman type, 601238
ATF6	99,9	99,5	100	100	Achromatopsia 7, 616517
ATG5	98,4	96,4	100	100	?Spinocerebellar ataxia, autosomal recessive 25, 617584

ATIC	99,8	99,1	100	100	AICA-ribosiduria due to ATIC deficiency, 608688
ATM	99,4	97,1	100	100	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic, T-cell prolymphocytic leukemia, somatic, Lymphoma, mantle cell, somatic,
ATOH7	97,6	92,6	99,2	94,1	Persistent hyperplastic primary vitreous, autosomal recessive, 221900
ATP13A2	100	99,6	100	100	Spastic paraplegia 78, autosomal recessive, 617225 Kufor-Rakeb syndrome, 606693
ATP2A1	100	100	100	100	Brody myopathy, 601003
ATP5F1A	92,2	83	100	100	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4, 615228 ?Combined oxidative phosphorylation deficiency 22, 616045
ATP5F1D	97,4	91,8	100	100	Mitochondrial complex V (ATP synthase) deficiency, 618120
ATP5F1E	100	100	100	100	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053
ATP6V0A2	99,9	98,7	100	100	Wrinkly skin syndrome, 278250 Cutis laxa, autosomal recessive, type IIA, 219200
ATP6V0A4	100	99,3	100	100	Distal renal tubular acidosis 3, with or without sensorineural hearing loss, 602722
ATP6V1A	99,8	98,4	100	100	Cutis laxa, autosomal recessive, type IID, 617403 Developmental and epileptic encephalopathy 93, 618012
ATP6V1B1	100	100	100	100	Distal renal tubular acidosis 2 with progressive sensorineural hearing loss, 267300
ATP6V1E1	92,5	86,1	100	100	Cutis laxa, autosomal recessive, type IIC, 617402
ATP7B	99,9	99,2	100	100	Wilson disease, 277900
ATP8A2	100	99,6	100	100	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268
ATP8B1	96,6	93,5	100	100	Cholestasis, progressive familial intrahepatic 1, 211600 Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, benign recurrent intrahepatic, 243300
ATPAF2	100	99,9	100	100	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
ATR	99,7	99	100	100	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
AUH	99,7	99,4	100	99,9	3-methylglutaconic aciduria, type I, 250950
AURKC	99,9	97,2	100	100	Spermatogenic failure 5, 243060
B2M	100	100	100	100	?Amyloidosis, familial visceral, 105200 Immunodeficiency 43, 241600
B3GALNT2	94,3	89,8	92,5	92,5	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181

B3GALT6	77	73	91,7	81	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640 Al-Gazali syndrome, 609465
B3GAT3	99,4	96,6	95,4	94,8	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B3GLCT	99,7	98,2	100	99,4	Peters-plus syndrome, 261540
B4GALNT1	98,3	93,5	100	100	Spastic paraplegia 26, autosomal recessive, 609195
B4GALT1	100	99,3	100	100	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	99,7	96,8	100	99,4	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
B4GAT1	100	100	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
B9D1	85,2	85,2	95,8	94	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120
B9D2	100	100	100	100	?Meckel syndrome 10, 614175 Joubert syndrome 34, 614175
BAAT	99,5	97,5	100	100	Hypercholanemia, familial, 607748 Bile acid conjugation defect 1, 619232
BANF1	95,3	78,1	100	100	Nestor-Guillermo progeria syndrome, 614008
BBIP1	95,7	87,4	100	100	?Bardet-Biedl syndrome 18, 615995
BBS1	100	100	100	100	Bardet-Biedl syndrome 1, 209900
BBS10	100	99,9	100	100	Bardet-Biedl syndrome 10, 615987
BBS12	100	100	100	100	Bardet-Biedl syndrome 12, 615989
BBS2	99,4	98	100	100	Retinitis pigmentosa 74, 616562 Bardet-Biedl syndrome 2, 615981
BBS4	99,9	98,9	100	99,9	Bardet-Biedl syndrome 4, 615982
BBS5	98,4	94,7	100	100	Bardet-Biedl syndrome 5, 615983
BBS7	99	96,5	100	99,9	Bardet-Biedl syndrome 7, 615984
BBS9	92	89	95,8	95,8	Bardet-Biedl syndrome 9, 615986
BCKDHA	99,8	97,9	100	100	Maple syrup urine disease, type Ia, 248600
BCKDHB	99,8	95,4	100	100	Maple syrup urine disease, type Ib, 248600
BCKDK	100	100	100	100	Branched-chain ketoacid dehydrogenase kinase deficiency, 614923
BCL10	100	100	100	100	?Immunodeficiency 37, 616098 Lymphoma, MALT, somatic, 137245

BCS1L	100	100	100	100	GRACILE syndrome, 603358 Mitochondrial complex III deficiency, nuclear type 1, 124000 Bjornstad syndrome, 262000
BFSP1	99,8	94	100	100	Cataract 33, multiple types, 611391
BFSP2	99,7	97,2	100	100	Cataract 12, multiple types, 611597
BHLHA9	72,5	53,5	99,6	96,9	?Camptosynpolydactyly, complex, 607539 Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432
BIN1	99,7	96	100	100	Centronuclear myopathy 2, 255200
BLM	99,3	97,7	100	100	Bloom syndrome, 210900
BLNK	96,9	92,6	100	100	?Agammaglobulinemia 4, 613502
BLOC1S3	99,9	90,3	100	100	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	99,3	97,6	100	100	?Hermansky-Pudlak syndrome 9, 614171
BLVRA	99,8	97,8	100	100	Hyperbiliverdinemia, 614156
BMP1	100	100	100	100	Osteogenesis imperfecta, type XIII, 614856
BMPER	100	99,6	100	100	Diaphanospondylodysostosis, 608022
BMPR1B	99,9	99,9	100	100	Acromesomelic dysplasia, Demirhan type, 609441 Brachydactyly, type A2, 112600 Brachydactyly, type A1, D, 616849
BOLA3	99	86,7	100	100	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BPGM	100	100	100	100	Erythrocytosis, familial, 8, 222800
IMPAD1	100	99,9	100	100	Chondrodysplasia with joint dislocations, GPAPP type, 614078
BRAT1	99,9	98,9	100	100	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056 Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BRCA1	99,4	98,4	100	100	Fanconi anemia, complementation group S, 617883
BRCA2	99,1	98,2	100	100	Fanconi anemia, complementation group D1, 605724 Wilms tumor, 194070
BRF1	99,8	98,4	100	100	Cerebellofaciodental syndrome, 616202
BRIP1	99,4	98,5	100	100	Fanconi anemia, complementation group J, 609054
BSCL2	100	99,9	100	100	Lipodystrophy, congenital generalized, type 2, 269700 Neuropathy, distal hereditary motor, type VC, 619112 Silver spastic paraplegia syndrome, 270685 Encephalopathy, progressive, with or without lipodystrophy, 615924

BSND	100	99,9	100	100	Sensorineural deafness with mild renal dysfunction, 602522 Bartter syndrome, type 4a, 602522
BTD	83	82,9	83,1	83,1	Biotinidase deficiency, 253260
BUB1B	99,3	98,3	100	100	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300
BVES	99,4	98,6	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 25, 616812
C12orf4	99,6	99,5	100	100	Mental retardation, autosomal recessive 66, 618221
C12orf57	100	98,6	100	100	Temtamy syndrome, 218340
C19orf12	100	99,8	100	100	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
C1QA	100	100	100	100	C1q deficiency, 613652
C1QB	100	100	100	100	C1q deficiency, 613652
C1QBP	84,3	70,6	100	100	Combined oxidative phosphorylation deficiency 33, 617713
C1QC	100	99,7	100	100	C1q deficiency, 613652
C1S	99,9	98,8	99,7	97,7	C1s deficiency, 613783 Ehlers-Danlos syndrome, periodontal type, 2, 617174
C2CD3	95,8	95,4	95,9	95,9	Orofaciodigital syndrome XIV, 615948
C3	99,9	98,5	100	100	C3 deficiency, 613779
C4A	98,3	94,9	99,5	99,1	C4a deficiency, 614380
C4B	98,8	95,6	100	99,9	C4B deficiency, 614379
C5	99,6	98,2	100	99,8	C5 deficiency, 609536
C8A	100	99,4	100	100	C8 deficiency, type I, 613790
C8B	99,9	98,6	100	100	C8 deficiency, type II, 613789
C9	99,7	99,2	100	100	C9 deficiency, 613825
CA12	100	99,9	100	100	Hyperchlorhidrosis, isolated, 143860
CA2	100	100	100	100	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA5A	87,6	85,6	87,7	87,7	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CA8	99,4	96,6	100	100	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CABP2	75,4	64,9	100	100	Deafness, autosomal recessive 93, 614899
CABP4	100	99,6	100	100	Cone-rod synaptic disorder, congenital nonprogressive, 610427
CACNA1B	98	96,1	99,3	98,2	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497

CACNA1D	97,9	97,7	100	100	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896
CACNA2D2	93,8	93,1	99,6	98,4	Cerebellar atrophy with seizures and variable developmental delay, 618501
CAD	99,8	98,5	100	100	Developmental and epileptic encephalopathy 50, 616457
CAMK2A	99,8	98,4	99,9	99,6	Mental retardation, autosomal dominant 53, 617798 ?Mental retardation, autosomal recessive 63, 618095
CANT1	100	100	100	100	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719
CAPN1	100	100	100	100	Spastic paraplegia 76, autosomal recessive, 616907
CAPN10	100	99,4	100	100	No OMIM disease ID
CAPN3	97,7	96,3	97,9	97,9	Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600 Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129
CARD11	100	99,9	100	100	B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11B with atopic dermatitis, 617638 Immunodeficiency 11A, 615206
CARD9	100	99	100	100	Candidiasis, familial, 2, autosomal recessive, 212050
CARS2	100	100	100	99,5	Combined oxidative phosphorylation deficiency 27, 616672
CASP14	100	99,9	100	100	Ichthyosis, congenital, autosomal recessive 12, 617320
CASP8	95,6	95,4	95,6	95,6	Hepatocellular carcinoma, somatic, 114550 ?Autoimmune lymphoproliferative syndrome, type IIB, 607271
CASQ2	100	99,8	100	100	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CASR	100	99,5	100	100	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 Hyperparathyroidism, neonatal, 239200 Hypocalcemia, autosomal dominant, 601198 Hypocalciuric hypercalcemia, type I, 145980
CAST	99	95,4	100	100	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295
CAT	100	100	100	100	Acatalasemia, 614097
CATSPER1	100	99,8	100	100	Spermatogenic failure 7, 612997
CAV1	100	100	100	100	?Lipodystrophy, congenital generalized, type 3, 612526 Pulmonary hypertension, primary, 3, 615343 Lipodystrophy, familial partial, type 7, 606721
CAVIN1	100	100	100	100	Lipodystrophy, congenital generalized, type 4, 613327
CBLIF	100	99,6	100	100	Intrinsic factor deficiency, 261000

CBS	99,9	98,5	100	100	Thrombosis, hyperhomocysteinemic, 236200 Homocystinuria, B6-responsive and nonresponsive types, 236200
CBX2	100	100	100	100	?46XY sex reversal 5, 613080
CC2D1A	100	99,6	100	100	Mental retardation, autosomal recessive 3, 608443
CC2D2A	98,3	96,6	97,1	97	COACH syndrome 2, 619111 Meckel syndrome 6, 612284 Joubert syndrome 9, 612285
CCBE1	99,9	98,8	100	100	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CCDC103	100	100	100	100	Ciliary dyskinesia, primary, 17, 614679
CCDC115	95,8	90,2	100	100	Congenital disorder of glycosylation, type IIo, 616828
CCDC174	99,5	96,7	100	100	Hypotonia, infantile, with psychomotor retardation, 616816
CCDC39	99	96,2	100	99,9	Ciliary dyskinesia, primary, 14, 613807
CCDC40	99,1	98,2	100	100	Ciliary dyskinesia, primary, 15, 613808
CCDC47	99,9	97,4	100	100	Trichohepatoneurodevelopmental syndrome, 618268
CCDC65	97	92,5	100	100	Ciliary dyskinesia, primary, 27, 615504
CCDC8	100	100	100	100	3-M syndrome 3, 614205
CCDC88A	95,9	91,8	97,5	97,3	?PEHO syndrome-like, 617507
CCDC88C	99,9	99,3	100	100	?Spinocerebellar ataxia 40, 616053 Hydrocephalus, congenital, 1, 236600
CCN6	84,6	84,6	84,9	84,6	Progressive pseudorheumatoid dysplasia, 208230
CCNO	100	99	100	100	Ciliary dyskinesia, primary, 29, 615872
CCT5	99,9	99,3	100	100	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CD151	100	100	100	100	Nephropathy with pretibial epidermolysis bullosa and deafness, 609057
CD19	100	100	100	100	Immunodeficiency, common variable, 3, 613493
CD247	100	99,4	100	100	?Immunodeficiency 25, 610163
CD27	99,9	98,3	100	100	Lymphoproliferative syndrome 2, 615122
CD2AP	99,6	98,8	100	99,9	Glomerulosclerosis, focal segmental, 3, 607832
CD320	100	100	100	99,9	Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646
CD3D	100	99,9	100	100	Immunodeficiency 19, 615617
CD3E	100	98,8	100	100	Immunodeficiency 18, 615615 Immunodeficiency 18, SCID variant, 615615

CD3G	100	100	100	100	Immunodeficiency 17, CD3 gamma deficient, 615607
CD40	100	100	100	100	Immunodeficiency with hyper-IgM, type 3, 606843
CD55	91,5	82,6	95	92,6	Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300
CD59	75,5	67	64,5	64,5	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD79A	100	100	100	100	Agammaglobulinemia 3, 613501
CD79B	100	100	100	100	Agammaglobulinemia 6, 612692
CD81	100	99,6	100	99,8	Immunodeficiency, common variable, 6, 613496
CD8A	100	100	100	100	CD8 deficiency, familial, 608957
CDAN1	100	99,9	100	100	Dyserythropoietic anemia, congenital, type Ia, 224120
CDC14A	99,9	99,2	100	100	Deafness, autosomal recessive 32, with or without immotile sperm, 608653
CDC45	99,8	98,5	100	100	Meier-Gorlin syndrome 7, 617063
CDC6	100	99,9	100	100	?Meier-Gorlin syndrome 5, 613805
CDCA7	100	99,6	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910
CDH11	100	100	100	100	Elsahy-Waters syndrome, 211380
CDH23	100	100	100	100	Usher syndrome, type 1D, 601067 Usher syndrome, type 1D/F digenic, 601067 Deafness, autosomal recessive 12, 601386
CDH3	100	99,1	100	100	Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280
CDHR1	99,5	98,1	100	100	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660
C15orf41	85,9	85,7	100	100	Dyserythropoietic anemia, congenital, type Ib, 615631
CDK10	100	99,5	100	100	Al Kaissi syndrome, 617694
CDK5	100	99,5	100	100	?Lissencephaly 7 with cerebellar hypoplasia, 616342
CDK5RAP2	99,6	98,5	100	100	Microcephaly 3, primary, autosomal recessive, 604804
CDK6	99,9	99,1	100	100	?Microcephaly 12, primary, autosomal recessive, 616080
CDSN	100	99,8	100	100	Hypotrichosis 2, 146520 Peeling skin syndrome 1, 270300
CDT1	99,7	97,8	99,9	98	Meier-Gorlin syndrome 4, 613804
CEACAM16	100	99,2	100	100	Deafness, autosomal dominant 4B, 614614 Deafness, autosomal recessive 113, 618410

CEBPE	100	100	100	100	Specific granule deficiency, 245480
CENPE	96,7	90	100	99,7	?Microcephaly 13, primary, autosomal recessive, 616051
CENPF	99,4	96,9	100	100	Stromme syndrome, 243605
CENPJ	99,8	98,7	100	100	Microcephaly 6, primary, autosomal recessive, 608393 ?Seckel syndrome 4, 613676
CEP104	99,9	98	100	100	Joubert syndrome 25, 616781
CEP120	99,9	99,6	100	100	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300 Joubert syndrome 31, 617761
CEP135	98,3	90,1	100	99,9	Microcephaly 8, primary, autosomal recessive, 614673
CEP152	99,5	98	100	100	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823
CEP164	99,8	98,2	100	100	Nephronophthisis 15, 614845
CEP19	100	100	100	100	Morbid obesity and spermatogenic failure, 615703
CEP290	96,2	90,8	100	99,9	Leber congenital amaurosis 10, 611755 Joubert syndrome 5, 610188 Senior-Loken syndrome 6, 610189 ?Bardet-Biedl syndrome 14, 615991 Meckel syndrome 4, 611134
CEP41	98,8	93,4	100	100	Joubert syndrome 15, 614464
CEP55	100	99,8	100	100	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500
CEP57	97,6	89,3	100	100	Mosaic variegated aneuploidy syndrome 2, 614114
CEP63	98,6	94,9	100	100	?Seckel syndrome 6, 614728
CEP78	98,8	95,6	100	99,9	Cone-rod dystrophy and hearing loss, 617236
CEP83	99	96,6	100	99,9	Nephronophthisis 18, 615862
CERKL	99,3	96,5	100	100	Retinitis pigmentosa 26, 608380
CERS1	79,1	65,2	93,2	86,5	?Epilepsy, progressive myoclonic, 8, 616230
CERS3	99,7	98	100	100	Ichthyosis, congenital, autosomal recessive 9, 615023
CFAP298	99,6	96,7	100	100	Ciliary dyskinesia, primary, 26, 615500
CFAP410	100	99,6	100	100	Retinal dystrophy with macular staphyloma, 617547 Spondylometaphyseal dysplasia, axial, 602271
C8orf37	99,7	99,6	100	100	Retinitis pigmentosa 64, 614500 Cone-rod dystrophy 16, 614500 Bardet-Biedl syndrome 21, 617406

CFAP53	99,3	96,6	100	100	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFD	90,9	84,6	100	100	Complement factor D deficiency, 613912
CFH	99,8	98,5	100	99,9	Basal laminar drusen, 126700 Complement factor H deficiency, 609814
CFI	99,3	96	100	99,9	Complement factor I deficiency, 610984
CFL2	99,5	99	100	99,9	Nemaline myopathy 7, autosomal recessive, 610687
CFTR	99,5	97,9	100	100	Cystic fibrosis, 219700 Congenital bilateral absence of vas deferens, 277180 Sweat chloride elevation without CF,
CHAT	93,1	85,1	100	99,9	Myasthenic syndrome, congenital, 6, presynaptic, 254210
CHKB	100	99,6	100	100	Muscular dystrophy, congenital, megaconial type, 602541
CHMP1A	100	99,6	100	100	Pontocerebellar hypoplasia, type 8, 614961
CHP1	97,1	85,7	100	100	?Spastic ataxia 9, autosomal recessive, 618438
CHRM3	100	100	100	100	Prune belly syndrome, 100100
CHRNA1	100	99,6	100	100	Myasthenic syndrome, congenital, 1B, fast-channel, 608930 Myasthenic syndrome, congenital, 1A, slow-channel, 601462 Multiple pterygium syndrome, lethal type, 253290
CHRNB1	100	99,5	100	100	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314 Myasthenic syndrome, congenital, 2A, slow-channel, 616313
CHRND	99,4	97,4	100	100	?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323 Multiple pterygium syndrome, lethal type, 253290 Myasthenic syndrome, congenital, 3B, fast-channel, 616322 ?Myasthenic syndrome, congenital, 3A, slow-channel, 616321
CHRNE	100	100	100	100	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	100	100	100	100	Multiple pterygium syndrome, lethal type, 253290 Escobar syndrome, 265000
CHST11	100	100	100	100	?Osteochondrodysplasia, brachydactyly, and overlapping malformed digits, 618167
CHST14	99,9	98,8	100	100	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	100	99,9	100	100	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHST6	100	100	100	100	Macular corneal dystrophy, 217800
CHST8	100	100	100	100	?Peeling skin syndrome 3, 616265

CHSY1	97,8	96,3	99,3	96,9	Temtamy preaxial brachydactyly syndrome, 605282
CHUK	99,7	98,1	100	100	?Popliteal pterygium syndrome, Bartsocas-Papas type 2, 619339 Cocoon syndrome, 613630
CIB2	99,3	96,2	100	99,9	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CIDEC	99,9	98,3	100	100	?Lipodystrophy, familial partial, type 5, 615238
CIITA	100	99,4	100	100	Bare lymphocyte syndrome, type II, complementation group A, 209920
CILK1	99,6	98	100	99,8	Endocrine-cerebroosteodysplasia, 612651
CISD2	83,4	83,4	100	100	Wolfram syndrome 2, 604928
CIT	99,8	98,2	100	100	Microcephaly 17, primary, autosomal recessive, 617090
CKAP2L	99,5	98,3	100	100	Filippi syndrome, 272440
CLCF1	100	99,5	100	100	Cold-induced sweating syndrome 2, 610313
CLCN1	99,9	98,8	100	100	Myotonia congenita, recessive, 255700 Myotonia congenita, dominant, 160800 Myotonia levior, recessive,
CLCN2	100	99,3	100	100	Leukoencephalopathy with ataxia, 615651 Hyperaldosteronism, familial, type II, 605635
CLCN7	99,4	97,8	100	100	Hypopigmentation, organomegaly, and delayed myelination and development, 618541 Osteopetrosis, autosomal recessive 4, 611490 Osteopetrosis, autosomal dominant 2, 166600
CLCNKB	98,7	95,3	100	100	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090
CLDN1	100	100	100	100	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626
CLDN10	100	100	100	100	HELIX syndrome, 617671
CLDN14	100	99,5	100	100	Deafness, autosomal recessive 29, 614035
CLDN16	100	100	100	100	Hypomagnesemia 3, renal, 248250
CLDN19	98,3	92,9	100	100	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLIC5	89,5	87,3	100	100	?Deafness, autosomal recessive 103, 616042
CLIP1	99,8	98,7	100	100	No OMIM disease ID
CLMP	100	99,5	100	100	Congenital short bowel syndrome, 615237
CLN3	92,5	92,4	92,5	92,5	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	69	66,3	71,8	71,6	Ceroid lipofuscinosis, neuronal, 5, 256731

CLN6	99,9	98,9	100	100	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, 4A (Kufs type), autosomal recessive, 204300
CLN8	83,5	83,5	100	100	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 Ceroid lipofuscinosis, neuronal, 8, 600143
CLP1	100	100	100	100	Pontocerebellar hypoplasia, type 10, 615803
CLPB	94,9	94	100	100	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CLPP	100	99,5	100	100	Perrault syndrome 3, 614129
CLRN1	100	99,7	100	100	Usher syndrome, type 3A, 276902 Retinitis pigmentosa 61, 614180
CNGA1	91	85,6	91	90,9	Retinitis pigmentosa 49, 613756
CNGA3	100	99,8	100	100	Achromatopsia 2, 216900
CNGB1	99	97,1	100	100	Retinitis pigmentosa 45, 613767
CNGB3	97,8	90,6	100	100	Achromatopsia 3, 262300
CNNM2	100	99,9	100	100	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNNM4	99,9	99	99,7	99	Jalili syndrome, 217080
CNPY3	100	99,7	100	100	Developmental and epileptic encephalopathy 60, 617929
CNTN1	99,7	98,6	100	100	?Myopathy, congenital, Compton-North, 612540
CNTN2	92,7	92,7	100	100	?Epilepsy, myoclonic, familial adult, 5, 615400
CNTNAP1	100	99,8	100	100	Lethal congenital contracture syndrome 7, 616286 Hypomyelinating neuropathy, congenital, 3, 618186
CNTNAP2	100	99,5	100	100	Pitt-Hopkins like syndrome 1, 610042 Cortical dysplasia-focal epilepsy syndrome, 610042
COA5	94,4	83	85,2	85,2	?Mitochondrial complex IV, deficiency, nuclear type 9, 616500
COA6	99,6	96,3	100	100	Mitochondrial complex IV deficiency, nuclear type 13, 616501
COA7	100	100	100	100	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387
COA8	81,9	80,8	93,7	93,4	Mitochondrial complex IV deficiency, nuclear type 17, 619061
COASY	100	100	100	100	Pontocerebellar hypoplasia, type 12, 618266 Neurodegeneration with brain iron accumulation 6, 615643
COCH	95	93,5	100	99,9	Deafness, autosomal dominant 9, 601369 ?Deafness, autosomal recessive 110, 618094
COG1	100	99,9	100	100	Congenital disorder of glycosylation, type IIg, 611209
COG2	99,8	98,9	100	100	?Congenital disorder of glycosylation, type IIq, 617395

COG4	100	100	100	100	Congenital disorder of glycosylation, type IIj, 613489 Saul-Wilson syndrome, 618150
COG5	99,1	96,8	100	100	Congenital disorder of glycosylation, type IIi, 613612
COG6	98,5	93,1	100	100	Shaheen syndrome, 615328 Congenital disorder of glycosylation, type IIl, 614576
COG7	100	99,4	100	100	Congenital disorder of glycosylation, type IIe, 608779
COG8	98,6	95,3	100	100	Congenital disorder of glycosylation, type IIh, 611182
COL11A1	96	92,7	100	99,9	Fibrochondrogenesis 1, 228520 Stickler syndrome, type II, 604841 Marshall syndrome, 154780 Deafness, autosomal dominant 37, 618533
COL11A2	100	99,6	100	100	Deafness, autosomal dominant 13, 601868 Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150 Fibrochondrogenesis 2, 614524 Deafness, autosomal recessive 53, 609706 Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840
COL12A1	99,9	99,3	100	100	Bethlem myopathy 2, 616471 ?Ullrich congenital muscular dystrophy 2, 616470
COL13A1	93,9	93,5	100	100	Myasthenic syndrome, congenital, 19, 616720
COL17A1	99	96,3	100	100	Epithelial recurrent erosion dystrophy, 122400 Epidermolysis bullosa, junctional, localisata variant, 226650 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
COL18A1	98,2	95,7	100	100	Knobloch syndrome, type 1, 267750 Glaucoma, primary closed-angle, 618880
COL1A2	98,7	95,7	100	100	Osteogenesis imperfecta, type III, 259420 Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821 Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2, 619120 Ehlers-Danlos syndrome, cardiac valvular type, 225320 Osteogenesis imperfecta, type IV, 166220 Osteogenesis imperfecta, type II, 166210
COL25A1	95,6	95,3	99,9	99,9	Fibrosis of extraocular muscles, congenital, 5, 616219
COL27A1	99,8	99,4	100	100	Steel syndrome, 615155
COL3A1	99,6	96,2	100	100	Ehlers-Danlos syndrome, vascular type, 130050 Polymicrogyria with or without vascular-type EDS, 618343

COL4A3	98,9	97,4	100	100	Hematuria, benign familial, 141200 Alport syndrome 3, autosomal dominant, 104200 Alport syndrome 2, autosomal recessive, 203780
COL4A4	99,6	97,4	100	100	Hematuria, familial benign, 141200 Alport syndrome 2, autosomal recessive, 203780
COL6A1	100	99,7	100	100	Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090
COL6A2	100	99,8	100	100	Bethlem myopathy 1, 158810 ?Myosclerosis, congenital, 255600 Ullrich congenital muscular dystrophy 1, 254090
COL6A3	100	99,7	100	100	Ullrich congenital muscular dystrophy 1, 254090 Dystonia 27, 616411 Bethlem myopathy 1, 158810
COL7A1	99,6	98,5	100	100	Epidermolysis bullosa, pretibial, 131850 Transient bullous of the newborn, 131705 EBD, Bart type, 132000 Epidermolysis bullosa dystrophica, AD, 131750 Epidermolysis bullosa pruriginosa, 604129 EBD inversa, 226600 Epidermolysis bullosa dystrophica, AR, 226600 Toenail dystrophy, isolated, 607523 EBD, localisata variant,
COL9A1	99,9	98,6	100	100	Stickler syndrome, type IV, 614134 ?Epiphyseal dysplasia, multiple, 6, 614135
COL9A2	99,9	98,9	100	100	Epiphyseal dysplasia, multiple, 2, 600204 ?Stickler syndrome, type V, 614284
COLEC10	100	99,9	100	100	3MC syndrome 3, 248340
COLEC11	100	100	100	100	3MC syndrome 2, 265050
COLGALT1	94,3	90,3	99,1	97,9	Brain small vessel disease 3, 618360
COLQ	99,8	97,1	100	100	Myasthenic syndrome, congenital, 5, 603034
COPB2	99,6	98,2	100	100	?Microcephaly 19, primary, autosomal recessive, 617800
COQ2	97,6	96,7	97,2	97,2	Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	91	89,7	100	100	Coenzyme Q10 deficiency, primary, 7, 616276
COQ6	99,9	98,5	100	100	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	100	99,6	100	100	?Coenzyme Q10 deficiency, primary, 8, 616733

COQ8A	100	99,6	100	100	Coenzyme Q10 deficiency, primary, 4, 612016
COQ8B	100	99,2	100	100	Nephrotic syndrome, type 9, 615573
COQ9	100	98,7	100	100	Coenzyme Q10 deficiency, primary, 5, 614654
CORO1A	99,9	98,9	100	99,9	Immunodeficiency 8, 615401
COX10	100	99,9	100	100	Mitochondrial complex IV deficiency, nuclear type 3, 619046
COX14	100	100	100	100	?Mitochondrial complex IV deficiency, nuclear type 10, 619053
COX15	99,9	97,8	100	100	Mitochondrial complex IV deficiency, nuclear type 6, 615119
COX20	95,7	82,4	100	99,9	Mitochondrial complex IV deficiency, nuclear type 11, 619054
COX4I2	100	99,9	100	100	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX6A1	100	99,9	100	100	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
COX6A2	99,8	94	100	100	Mitochondrial complex IV deficiency, nuclear type 18, 619062
COX6B1	100	100	100	100	Mitochondrial complex IV deficiency, nuclear type 7, 619051
COX8A	100	100	100	100	?Mitochondrial complex IV deficiency, nuclear type 15, 619059
CP	92,6	85,2	100	99,9	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290
CPA6	99,2	96,1	100	100	Febrile seizures, familial, 11, 614418 Epilepsy, familial temporal lobe, 5, 614417
CPAMD8	95,9	92,6	99,9	99,6	Anterior segment dysgenesis 8, 617319
CPLANE1	99,4	98,2	100	100	Orofaciodigital syndrome VI, 277170 Joubert syndrome 17, 614615
CPLX1	100	100	100	100	Developmental and epileptic encephalopathy 63, 617976
CPN1	99,9	98,7	100	100	Carboxypeptidase N deficiency, 212070
CPOX	99,8	97,2	100	100	Coproporphyria, 121300 Harderoporphyria, 618892
CPS1	100	100	100	100	Carbamoylphosphate synthetase I deficiency, 237300
CPT1A	99,8	97,6	100	100	CPT deficiency, hepatic, type IA, 255120
CPT2	98,2	97,4	100	100	CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 CPT II deficiency, myopathic, stress-induced, 255110
CR2	100	99,9	100	100	Immunodeficiency, common variable, 7, 614699
CRADD	99,9	97,5	100	100	Mental retardation, autosomal recessive 34, with variant lissencephaly, 614499

CRAT	100	99,9	100	100	?Neurodegeneration with brain iron accumulation 8, 617917
CRB1	100	99,9	100	100	Leber congenital amaurosis 8, 613835 Retinitis pigmentosa-12, 600105 Pigmented paravenous chorioretinal atrophy, 172870
CRB2	98,9	94,2	100	100	Focal segmental glomerulosclerosis 9, 616220 Ventriculomegaly with cystic kidney disease, 219730
CRBN	87,9	87,8	96,3	91,8	Mental retardation, autosomal recessive 2, 607417
CREB3L1	100	99,9	100	100	Osteogenesis imperfecta, type XVI, 616229
CRIPT	98,9	93,5	100	100	Short stature with microcephaly and distinctive facies, 615789
CRLF1	91,1	90,3	97,9	95,7	Cold-induced sweating syndrome 1, 272430
CRPPA	98,4	94,7	100	99,8	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
CRTAP	100	99,4	100	100	Osteogenesis imperfecta, type VII, 610682
CRYAA	99,9	98	100	100	Cataract 9, multiple types, 604219
CRYAB	100	98,2	100	100	Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869 Myopathy, myofibrillar, 2, 608810 Cataract 16, multiple types, 613763 Cardiomyopathy, dilated, 1II, 615184
CRYBB1	100	99,9	100	100	Cataract 17, multiple types, 611544
CRYBB3	100	99,9	100	100	Cataract 22, 609741
CSF1R	100	99,6	100	100	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
CSF2RB	99,9	99	100	100	Surfactant metabolism dysfunction, pulmonary, 5, 614370
CSPP1	99,7	98,1	100	100	Joubert syndrome 21, 615636
CSTA	99,7	99,3	100	100	Peeling skin syndrome 4, 607936
CSTB	99,6	90,5	100	100	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTC1	100	99,1	100	100	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTDP1	88,7	85	100	99,8	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTNNA2	99,9	99,7	100	100	Cortical dysplasia, complex, with other brain malformations 9, 618174
CTNS	100	99,3	100	100	Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, atypical nephropathic, 219800

CTPS1	93	93	93	93	Immunodeficiency 24, 615897
CTSA	100	99,6	100	100	Galactosialidosis, 256540
CTSC	100	100	100	100	Periodontitis 1, juvenile, 170650 Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000
CTSD	98,4	95	100	100	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	83,9	78,9	100	100	Ceroid lipofuscinosis, neuronal, 13 (Kufs type), autosomal dominant, 615362
CTSK	100	99,2	100	100	Pycnodysostosis, 265800
CTU2	100	98,7	100	100	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142
CUBN	99,2	97,1	100	100	Imerslund-Grasbeck syndrome 1, 261100
CUL7	100	99,1	100	100	3-M syndrome 1, 273750
CWC27	99,5	95,9	100	100	Retinitis pigmentosa with or without skeletal anomalies, 250410
CWF19L1	100	99,6	100	100	Spinocerebellar ataxia, autosomal recessive 17, 616127
CYB5A	100	100	100	100	Methemoglobinemia and ambiguous genitalia, 250790
CYB5R3	99,1	98,1	99,6	98,5	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYBA	96	82,5	100	100	Chronic granulomatous disease 4, autosomal recessive, 233690
CYC1	98,3	89,5	100	99,4	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYP11A1	99,2	94,5	100	100	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	100	99,9	100	100	Aldosteronism, glucocorticoid-remediable, 103900 Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010
CYP11B2	100	99,9	100	100	Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 Aldosterone to renin ratio raised,
CYP17A1	99,9	98,5	100	100	17,20-lyase deficiency, isolated, 202110 17-alpha-hydroxylase/17,20-lyase deficiency, 202110
CYP19A1	98,3	95,7	100	100	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300
CYP1B1	100	100	100	100	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 Anterior segment dysgenesis 6, multiple subtypes, 617315
CYP21A2	97,4	91,1	100	100	Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910 Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910
CYP24A1	100	100	100	100	Hypercalcemia, infantile, 1, 143880

CYP26B1	100	100	100	100	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
CYP26C1	99,8	98,5	100	99,9	Focal facial dermal dysplasia 4, 614974
CYP27A1	99,7	98,1	100	100	Cerebrotendinous xanthomatosis, 213700
CYP27B1	100	99,8	100	100	Vitamin D-dependent rickets, type I, 264700
CYP2C8	99,9	97,5	100	100	No OMIM disease ID
CYP2R1	99,5	96	100	100	Rickets due to defect in vitamin D 25-hydroxylation deficiency, 600081
CYP2U1	95,3	92	100	99,9	Spastic paraplegia 56, autosomal recessive, 615030
CYP4F22	100	98,8	100	100	Ichthyosis, congenital, autosomal recessive 5, 604777
CYP4V2	99,6	97	100	100	Bietti crystalline corneoretinal dystrophy, 210370
CYP7B1	98,1	92,7	100	100	Spastic paraplegia 5A, autosomal recessive, 270800 Bile acid synthesis defect, congenital, 3, 613812
D2HGDH	99,7	98,2	100	100	D-2-hydroxyglutaric aciduria, 600721
DAG1	100	99,9	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818
DARS1	99,5	99,3	100	99,9	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2	94,8	93,8	100	100	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBH	100	100	100	100	Orthostatic hypotension 1, due to DBH deficiency, 223360
DBT	99,1	96,1	100	100	Maple syrup urine disease, type II, 248600
DCAF17	98,5	93,4	100	100	Woodhouse-Sakati syndrome, 241080
DCC	100	100	100	100	Mirror movements 1 and/or agenesis of the corpus callosum, 157600 Esophageal carcinoma, somatic, 133239 Colorectal cancer, somatic, 114500 Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542
DCDC2	100	99,9	100	100	Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212 Sclerosing cholangitis, neonatal, 617394
DCHS1	99,9	99,4	100	100	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390
DCLRE1C	99,8	98,2	100	99,9	Severe combined immunodeficiency, Athabascan type, 602450 Omenn syndrome, 603554
DCPS	91,3	91,2	100	100	Al-Raqad syndrome, 616459
DDB2	99,7	97,7	100	100	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740

DDC	99,2	95	100	100	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	98,5	96,5	100	100	Spastic paraplegia 28, autosomal recessive, 609340
DDHD2	99,7	99,5	100	100	Spastic paraplegia 54, autosomal recessive, 615033
DDOST	100	99,8	100	100	?Congenital disorder of glycosylation, type Ir, 614507
DDR2	100	99,6	100	100	Warburg-Cinotti syndrome, 618175 Spondylometaepiphyseal dysplasia, short limb-hand type, 271665
DDRGK1	100	100	100	100	Spondyloepimetaphyseal dysplasia, Shohat type, 602557
DDX11	84,9	80	100	100	Warsaw breakage syndrome, 613398
DDX59	100	99,8	100	100	Orofaciodigital syndrome V, 174300
DEAF1	99	94,5	99,9	98,2	Vulto-van Silfout-de Vries syndrome, 615828 Neurodevelopmental disorder with hypotonia, impaired expressive language, and with or without seizures, 617171
DEGS1	100	100	100	100	Leukodystrophy, hypomyelinating, 18, 618404
DENND5A	99,8	98,7	100	100	Developmental and epileptic encephalopathy 49, 617281
DES	100	99,6	100	100	Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419
DGAT1	91,8	87,6	99,9	98,9	?Diarrhea 7, protein-losing enteropathy type, 615863
DGKE	99,7	98,5	100	100	Nephrotic syndrome, type 7, 615008
DGUOK	99,9	98,8	100	100	Portal hypertension, noncirrhotic, 1, 617068 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHCR24	97,7	97,7	97,7	97,7	Desmosterolosis, 602398
DHCR7	100	100	100	100	Smith-Lemli-Opitz syndrome, 270400
DHDDS	99,4	95,6	95,2	95,2	Developmental delay and seizures with or without movement abnormalities, 617836 ?Congenital disorder of glycosylation, type 1bb, 613861 Retinitis pigmentosa 59, 613861
DHFR	88,9	76,3	100	100	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHH	100	100	100	100	46XY gonadal dysgenesis with minifascicular neuropathy, 607080 46XY sex reversal 7, 233420
DHODH	100	99,9	100	100	Miller syndrome, 263750
DHPS	100	99,8	93,3	93,2	Neurodevelopmental disorder with seizures and speech and walking impairment, 618480

DHTKD1	99,8	98,8	100	100	?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025 Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750
DHX38	100	99,1	100	100	Retinitis pigmentosa 84, 618220
DIAPH1	99,8	98,4	99,9	99	Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632
DIS3L2	100	100	100	100	Perlman syndrome, 267000
DLAT	99,8	99,3	100	99,9	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	99,9	99,7	100	99,9	Dihydrolipoamide dehydrogenase deficiency, 246900
DLL3	93	87,8	100	99,5	Spondylocostal dysostosis 1, autosomal recessive, 277300
DLX5	99,9	98,2	100	100	Split-hand/foot malformation 1, 183600 ?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600
DMGDH	99,9	99,7	100	100	Dimethylglycine dehydrogenase deficiency, 605850
DMP1	99,9	99,9	100	100	Hypophosphatemic rickets, AR, 241520
DMXL2	99,7	98,9	100	99,9	Developmental and epileptic encephalopathy 81, 618663 ?Deafness, autosomal dominant 71, 617605 ?Polyendocrine-polyneuropathy syndrome, 616113
DNA2	99,6	96,9	100	100	?Seckel syndrome 8, 615807 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156
DNAAF1	100	99,4	100	100	Ciliary dyskinesia, primary, 13, 613193
LRRC6	99,3	96,9	100	100	Ciliary dyskinesia, primary, 19, 614935
DNAAF2	99,7	98,4	100	100	Ciliary dyskinesia, primary, 10, 612518
DNAAF3	99,5	96,3	100	100	Ciliary dyskinesia, primary, 2, 606763
DNAAF4	99,4	94,7	100	99,7	Ciliary dyskinesia, primary, 25, 615482
DNAAF5	85,6	78,8	98,8	96,9	Ciliary dyskinesia, primary, 18, 614874
DNAH1	99,9	99,6	100	100	Spermatogenic failure 18, 617576 ?Ciliary dyskinesia, primary, 37, 617577
DNAH11	99,8	98,8	100	100	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH5	99,9	98,9	100	100	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAH9	99,5	97,8	100	100	Ciliary dyskinesia, primary, 40, 618300
DNAI1	100	99,9	100	100	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	98,2	95,8	100	100	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJB2	100	100	100	100	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881

DNAJC12	87,4	87,3	100	100	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC19	99,3	92,2	100	100	3-methylglutaconic aciduria, type V, 610198
DNAJC21	99,5	97,4	100	100	Bone marrow failure syndrome 3, 617052
DNAJC3	99,7	99,7	100	99,9	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192
DNAJC6	99,8	98,7	100	100	Parkinson disease 19a, juvenile-onset, 615528 Parkinson disease 19b, early-onset, 615528
DNAL1	99,6	98,3	100	99,2	Ciliary dyskinesia, primary, 16, 614017
DNASE1L3	100	100	100	100	Systemic lupus erythematosus 16, 614420
DNM1L	99,6	98,3	100	100	Optic atrophy 5, 610708 Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388
DNM2	98,6	93,9	100	100	Centronuclear myopathy 1, 160150 Charcot-Marie-Tooth disease, axonal type 2M, 606482 Charcot-Marie-Tooth disease, dominant intermediate B, 606482 Lethal congenital contracture syndrome 5, 615368
DNMBP	99,9	98,9	100	100	Cataract 48, 618415
DNMT3B	100	99,9	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 Facioscapulohumeral muscular dystrophy 4, digenic, 619478
DOCK2	99,8	98,8	100	100	Immunodeficiency 40, 616433
DOCK3	99,9	98,9	100	100	Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292
DOCK6	99,4	98,7	100	100	Adams-Oliver syndrome 2, 614219
DOCK7	99,6	98,3	100	99,9	Developmental and epileptic encephalopathy 23, 615859
DOCK8	100	99	100	100	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
DOK7	94,9	92	100	100	Fetal akinesia deformation sequence 3, 618389 Myasthenic syndrome, congenital, 10, 254300
DOLK	100	100	100	100	Congenital disorder of glycosylation, type Im, 610768
DONSON	93,8	85,8	100	100	Microcephaly, short stature, and limb abnormalities, 617604 Microcephaly-micromelia syndrome, 251230
DPAGT1	100	99,8	100	100	Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750 Congenital disorder of glycosylation, type Ij, 608093
DPH1	100	99,9	100	100	Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901
DPM1	97,4	90,9	98,6	94,6	Congenital disorder of glycosylation, type Ie, 608799
DPM2	100	97,7	100	100	Congenital disorder of glycosylation, type Iu, 615042

DPM3	100	100	100	100	?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937
DPY19L2	73,7	69,3	100	99,9	Spermatogenic failure 9, 613958
DPYD	99,5	96,5	100	100	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270
DPYS	100	100	100	100	Dihydropyrimidinuria, 222748
DRAM2	100	99,9	100	100	Cone-rod dystrophy 21, 616502
DRC1	99,9	98,3	100	100	Ciliary dyskinesia, primary, 21, 615294
DSC2	99,4	97,4	100	99,9	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 Arrhythmogenic right ventricular dysplasia 11, 610476
DSC3	98,9	96,7	100	100	Hypotrichosis and recurrent skin vesicles, 613102
DSE	98,4	95,4	100	100	Ehlers-Danlos syndrome, musculocontractural type 2, 615539
DSG4	99,8	99	100	100	Hypotrichosis 6, 607903
DSP	99,9	99,4	100	100	Arrhythmogenic right ventricular dysplasia 8, 607450 Skin fragility-woolly hair syndrome, 607655 Epidermolysis bullosa, lethal acantholytic, 609638 Keratosis palmoplantaris striata II, 612908 Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821 Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676
DST	95,3	94,5	95,6	95,6	Epidermolysis bullosa simplex, autosomal recessive 2, 615425 ?Neuropathy, hereditary sensory and autonomic, type VI, 614653
DSTYK	99,9	98,8	100	100	Congenital anomalies of kidney and urinary tract 1, 610805 Spastic paraplegia 23, 270750
DTNBP1	99,7	98	100	99,9	Hermansky-Pudlak syndrome 7, 614076
DUOX2	97,2	94,8	100	100	Thyroid dyshormonogenesis 6, 607200
DUOXA2	100	100	100	100	Thyroid dyshormonogenesis 5, 274900
DYM	97	95,6	100	100	Smith-McCort dysplasia, 607326 Dyggve-Melchior-Clausen disease, 223800
DYNC1I2	84	66	100	100	Neurodevelopmental disorder with microcephaly and structural brain anomalies, 618492
DYNC2H1	98,6	95,2	100	99,8	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
WDR60	99,3	95,8	100	100	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WDR34	100	99,8	100	100	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633

DYNC2LI1	99,6	98,4	100	100	Short-rib thoracic dysplasia 15 with polydactyly, 617088
TCTEX1D2	100	99,8	100	100	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405
DYSF	100	99,8	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601 Miyoshi muscular dystrophy 1, 254130 Myopathy, distal, with anterior tibial onset, 606768
DZIP1L	99,8	98	100	100	Polycystic kidney disease 5, 617610
EARS2	99,8	98	100	100	Combined oxidative phosphorylation deficiency 12, 614924
ECEL1	95,9	91,8	100	100	Arthrogryposis, distal, type 5D, 615065
ECHS1	100	99,4	100	100	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ECM1	100	99,6	100	100	Urbach-Wiethe disease, 247100
EDAR	100	99,7	100	100	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490 Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900
EDARADD	99,7	98,9	100	100	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941 Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940
EDC3	100	99,5	100	100	?Mental retardation, autosomal recessive 50, 616460
EDN1	100	99,4	100	100	Question mark ears, isolated, 612798 Auriculocondylar syndrome 3, 615706
EDN3	98,8	98,8	100	100	Waardenburg syndrome, type 4B, 613265
EDNRB	96,3	92,5	100	100	ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580
EFEMP2	100	100	100	100	Cutis laxa, autosomal recessive, type IB, 614437
EFL1	99,3	97,7	100	100	Shwachman-Diamond syndrome 2, 617941
EGF	99,9	99,8	100	100	?Hypomagnesemia 4, renal, 611718
EGFR	100	100	100	100	?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	100	100	100	100	Dejerine-Sottas disease, 145900 Charcot-Marie-Tooth disease, type 1D, 607678 Hypomyelinating neuropathy, congenital, 1, 605253
EIF2AK3	98,2	95,5	100	99,8	Wolcott-Rallison syndrome, 226980
EIF2AK4	99,6	97,8	100	100	Pulmonary venoocclusive disease 2, 234810
EIF2B1	100	99,8	100	100	Leukoencephalopathy with vanishing white matter, 603896

EIF2B2	100	98,1	100	100	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B3	100	100	100	100	Leukoencephalopathy with vanishing white matter, 603896
EIF2B4	100	99,5	100	100	Ovarioleukodystrophy, 603896 Leukoencephalopathy with vanishing white matter, 603896
EIF2B5	99,8	98,5	100	100	Ovarioleukodystrophy, 603896 Leukoencephalopathy with vanishing white matter, 603896
EIF3F	97,1	82,5	100	100	Mental retardation, autosomal recessive 67, 618295
EIF4A3	100	99,2	100	100	Robin sequence with cleft mandible and limb anomalies, 268305
ELAC2	100	99,2	100	100	Combined oxidative phosphorylation deficiency 17, 615440
ELMO2	100	99	100	100	Vascular malformation, primary intraosseous, 606893
ELMOD3	100	100	100	100	?Deafness, autosomal recessive 88, 615429 ?Deafness, autosomal dominant 81, 619500
ELOVL4	99,7	98,9	100	99,9	Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
ELP1	99,8	98,9	100	100	Dysautonomia, familial, 223900
ELP2	99,8	98,3	100	99,9	Mental retardation, autosomal recessive 58, 617270
EMC1	99,9	98	100	100	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EMG1	100	100	100	100	Bowen-Conradi syndrome, 211180
EML1	99,6	98,1	100	100	Band heterotopia, 600348
EMP2	98,9	93	100	100	Nephrotic syndrome, type 10, 615861
ENAM	100	100	100	100	Amelogenesis imperfecta, type IC, 204650 Amelogenesis imperfecta, type IB, 104500
ENO3	100	100	100	100	?Glycogen storage disease XIII, 612932
ENPP1	96,5	90,6	98,8	97,8	Hypophosphatemic rickets, autosomal recessive, 2, 613312 Arterial calcification, generalized, of infancy, 1, 208000 Cole disease, 615522
ENTPD1	100	99,8	100	100	Spastic paraplegia 64, autosomal recessive, 615683
EOGT	79,3	77,8	91,8	88,3	Adams-Oliver syndrome 4, 615297
EPB41	85,4	83,8	100	100	Elliptocytosis-1, 611804
EPB42	99,9	98,5	100	100	Spherocytosis, type 5, 612690

EPCAM	97,5	89,6	100	100	Colorectal cancer, hereditary nonpolyposis, type 8, 613244 Diarrhea 5, with tufting enteropathy, congenital, 613217
EPG5	99,2	97,8	100	100	Vici syndrome, 242840
EPHX1	99,8	97,8	100	100	No OMIM disease ID
EPM2A	93,9	91,2	99,6	96,3	Epilepsy, progressive myoclonic 2A (Lafora), 254780
EPO	99,9	97,8	100	100	Erythrocytosis, familial, 5, 617907 ?Diamond-Blackfan anemia-like, 617911
EPRS1	99,8	99,4	100	100	Leukodystrophy, hypomyelinating, 15, 617951
EPS8	96,9	96	100	100	?Deafness, autosomal recessive 102, 615974
EPS8L2	84,7	82,3	88	88	Deafness autosomal recessive 106, 617637
ERAL1	100	99,6	100	100	Perrault syndrome 6, 617565
ERBB3	100	99,3	100	100	?Lethal congenital contractural syndrome 2, 607598 Visceral neuropathy, familial, 1, autosomal recessive, 243180
ERCC1	100	96,4	100	100	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	100	99,4	100	100	Xeroderma pigmentosum, group D, 278730 Trichothiodystrophy 1, photosensitive, 601675 ?Cerebrooculofacioskeletal syndrome 2, 610756
ERCC3	96,8	95,6	100	100	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651
ERCC4	100	99,9	100	100	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 XFE progeroid syndrome, 610965 Xeroderma pigmentosum, group F, 278760 Fanconi anemia, complementation group Q, 615272
ERCC5	99,9	99	100	100	Xeroderma pigmentosum, group G, 278780 Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	100	100	100	100	UV-sensitive syndrome 1, 600630 Cerebrooculofacioskeletal syndrome 1, 214150 Cockayne syndrome, type B, 133540 De Sanctis-Cacchione syndrome, 278800 Premature ovarian failure 11, 616946
ERCC6L2	99,6	98,6	100	100	Bone marrow failure syndrome 2, 615715
ERCC8	99	94,8	100	100	UV-sensitive syndrome 2, 614621 Cockayne syndrome, type A, 216400
ERGIC1	95,3	94,6	98,4	98,4	?Arthrogryposis multiplex congenita 2, neurogenic type, 208100

ERLIN1	100	100	100	100	Spastic paraplegia 62, 615681
ERLIN2	100	99,1	100	99,9	Spastic paraplegia 18, autosomal recessive, 611225
ESCO2	98,5	94,6	100	99,7	Juberg-Hayward syndrome, 216100 Roberts-SC phocomelia syndrome, 268300
ESPN	48,5	38,4	100	99,9	Deafness, neurosensory, without vestibular involvement, autosomal dominant, 609006 Deafness, autosomal recessive 36, 609006 ?Usher syndrome, type 1M, 618632
ESR1	100	99,8	100	100	Breast cancer, somatic, 114480 Estrogen resistance, 615363
ESRP1	99,9	98,4	100	100	?Deafness, autosomal recessive 109, 618013
ESRRB	96,2	93	100	100	Deafness, autosomal recessive 35, 608565
ETFA	99,8	99,6	100	99,9	Glutaric acidemia IIA, 231680
ETFB	100	99,9	100	100	Glutaric acidemia IIB, 231680
ETFDH	99,8	99,4	100	100	Glutaric acidemia IIC, 231680
ETHE1	99,3	93,3	100	100	Ethylmalonic encephalopathy, 602473
EVC	94,2	91,4	97,5	95,1	Ellis-van Creveld syndrome, 225500 ?Weyers acrofacial dysostosis, 193530
EVC2	98	96,2	100	100	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530
EXOC6B	98,1	97,1	99,9	99,5	Spondyloepimetaphyseal dysplasia with joint laxity, type 3, 618395
EXOSC2	100	99,9	100	100	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EXOSC3	98,1	90,5	100	100	Pontocerebellar hypoplasia, type 1B, 614678
EXOSC8	98,7	90	100	100	Pontocerebellar hypoplasia, type 1C, 616081
EXOSC9	99,3	94,7	100	99,9	Pontocerebellar hypoplasia, type 1D, 618065
EXPH5	100	99,9	100	100	Epidermolysis bullosa, nonspecific, autosomal recessive, 615028
EXT2	99,9	99	100	100	Seizures, scoliosis, and macrocephaly syndrome, 616682 Exostoses, multiple, type 2, 133701
EXTL3	100	100	100	100	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
EYS	99,5	97,5	100	100	Retinitis pigmentosa 25, 602772
F10	99,8	98,4	100	100	Factor X deficiency, 227600
F11	100	100	100	100	Factor XI deficiency, autosomal dominant, 612416 Factor XI deficiency, autosomal recessive, 612416

F12	100	98,6	100	100	Angioedema, hereditary, 3, 610618 Factor XII deficiency, 234000
F13A1	100	100	100	100	Factor XIIIA deficiency, 613225
F13B	98,3	92,8	100	99,9	Factor XIIIB deficiency, 613235
F2	99,9	97,8	100	100	Hypoprothrombinemia, 613679 Dysprothrombinemia, 613679 Thrombophilia due to thrombin defect, 188050
F5	99,9	98,4	100	100	Thrombophilia due to activated protein C resistance, 188055 Factor V deficiency, 227400
F7	100	100	100	100	Factor VII deficiency, 227500
FA2H	92,4	82,6	100	100	Spastic paraplegia 35, autosomal recessive, 612319
FADD	100	100	100	100	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations, 613759
FAH	100	99,5	100	99,9	Tyrosinemia, type I, 276700
FAM126A	99,5	99,4	100	100	Leukodystrophy, hypomyelinating, 5, 610532
FAM161A	99,8	99,5	100	100	Retinitis pigmentosa 28, 606068
FAM20A	99,6	94,4	100	100	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM20C	100	100	100	99,7	Raine syndrome, 259775
FAN1	100	99,8	100	100	Interstitial nephritis, karyomegalic, 614817
FANCA	99,9	98,7	100	100	Fanconi anemia, complementation group A, 227650
FANCB	98	91,7	100	99,6	Fanconi anemia, complementation group B, 300514
FANCC	96,9	95,7	97,3	97,3	Fanconi anemia, complementation group C, 227645
FANCD2	98,7	95,9	98,8	98,8	Fanconi anemia, complementation group D2, 227646
FANCE	90,7	85,5	100	100	Fanconi anemia, complementation group E, 600901
FANCF	100	100	100	100	Fanconi anemia, complementation group F, 603467
FANCG	100	99,9	100	100	Fanconi anemia, complementation group G, 614082
FANCI	99,8	98,6	100	100	Fanconi anemia, complementation group I, 609053
FANCL	99,4	97,6	100	100	Fanconi anemia, complementation group L, 614083
FAR1	97,4	94	100	100	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154 Cataracts, spastic paraparesis, and speech delay, 619338
FARS2	100	100	100	100	Combined oxidative phosphorylation deficiency 14, 614946 Spastic paraplegia 77, autosomal recessive, 617046

FARSB	98	92,9	100	100	Rajab interstitial lung disease with brain calcifications 1, 613658
FASTKD2	99,6	98,6	100	100	Combined oxidative phosphorylation deficiency 44, 618855
FAT4	100	100	100	100	Van Maldergem syndrome 2, 615546 Hennekam lymphangiectasia-lymphedema syndrome 2, 616006
FBLN5	91,8	91,7	91,8	91,8	Cutis laxa, autosomal recessive, type IA, 219100 Macular degeneration, age-related, 3, 608895 Neuropathy, hereditary, with or without age-related macular degeneration, 608895 ?Cutis laxa, autosomal dominant 2, 614434
FBP1	93,6	91,3	93,7	93,7	Fructose-1,6-bisphosphatase deficiency, 229700
FBXL3	100	100	100	100	Intellectual developmental disorder with short stature, facial anomalies, and speech defects, 606220
FBXL4	100	100	100	100	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FBXO31	97	94,2	100	100	?Mental retardation, autosomal recessive 45, 615979
FBXO7	99,8	98,3	100	100	Parkinson disease 15, autosomal recessive, 260300
FCGR3A	98,6	96,8	100	99,9	Immunodeficiency 20, 615707
FCN3	100	99,3	100	100	Immunodeficiency due to ficolin 3 deficiency, 613860
FCSK	98	96,1	100	100	Congenital disorder of glycosylation with defective fucosylation 2, 618324
FDFT1	98,5	96,7	100	100	Squalene synthase deficiency, 618156
FDX2	100	100	100	100	Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900
FDXR	100	98,6	100	100	Auditory neuropathy and optic atrophy, 617717
FECH	99,9	99,8	100	100	Protoporphyria, erythropoietic, 1, 177000
FERMT1	99,1	95,8	100	99,9	Kindler syndrome, 173650
FERMT3	100	100	100	100	Leukocyte adhesion deficiency, type III, 612840
FEZF1	100	100	100	100	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FGA	99,1	96,8	100	100	Hypodysfibrinogenemia, congenital, 616004 Dysfibrinogenemia, congenital, 616004 Amyloidosis, familial visceral, 105200 Afibrinogenemia, congenital, 202400
FGB	99,8	98,8	100	100	Hypofibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Afibrinogenemia, congenital, 202400
FGD4	99,8	98,8	100	100	Charcot-Marie-Tooth disease, type 4H, 609311
FGF20	95,8	88,4	100	100	?Renal hypodysplasia/aplasia 2, 615721

FGF23	99,4	96,7	100	100	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993 Hypophosphatemic rickets, autosomal dominant, 193100
FGF3	100	98,7	100	100	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGG	99,4	97,5	100	100	Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, 616004 Hypofibrinogenemia, congenital, 202400 Afibrinogenemia, congenital, 202400
FH	93,2	87,2	100	100	Leiomyomatosis and renal cell cancer, 150800 Fumarase deficiency, 606812
FIBP	100	99,8	100	100	Thauvin-Robinet-Faivre syndrome, 617107
FIG4	99,8	99,7	100	100	Yunis-Varon syndrome, 216340 ?Polymicrogyria, bilateral temporooccipital, 612691 Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228
FITM2	100	100	100	100	Siddiqi syndrome, 618635
FKBP10	98,9	97,3	100	100	Osteogenesis imperfecta, type XI, 610968 Bruck syndrome 1, 259450
FKBP14	99,8	98,7	100	100	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557
FKRP	100	100	100	100	Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153
FKTN	99,8	95,2	100	100	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Cardiomyopathy, dilated, 1X, 611615
FLAD1	100	99,7	100	100	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100
FLG	99,9	99,9	100	100	Ichthyosis vulgaris, 146700
FLNB	99,4	98,7	100	100	Larsen syndrome, 150250 Atelosteogenesis, type I, 108720 Atelosteogenesis, type III, 108721 Spondylocarpotarsal synostosis syndrome, 272460 Boomerang dysplasia, 112310
FLVCR1	99,7	98,3	100	99,9	Ataxia, posterior column, with retinitis pigmentosa, 609033
FLVCR2	100	100	100	100	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790
FMN2	85,5	82,8	100	100	Mental retardation, autosomal recessive 47, 616193

FMO3	99,9	99,7	100	100	Trimethylaminuria, 602079
FOLR1	100	99,9	100	100	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXE1	97,9	82,2	100	98,9	Bamforth-Lazarus syndrome, 241850
FOXE3	87,6	79	95,8	89,2	Anterior segment dysgenesis 2, multiple subtypes, 610256 Cataract 34, multiple types, 612968
FOXI1	100	100	100	100	Enlarged vestibular aqueduct, 600791
FOXN1	100	99,1	100	100	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806 T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXRED1	100	99,6	100	100	Mitochondrial complex I deficiency, nuclear type 19, 618241
FRAS1	100	99,2	100	100	Fraser syndrome 1, 219000
FREM1	99,8	98,4	100	100	Manitoba oculotrichoanal syndrome, 248450 Bifid nose with or without anorectal and renal anomalies, 608980 Trigonocephaly 2, 614485
FREM2	99,8	98,7	100	100	Fraser syndrome 2, 617666 Cryptophthalmos, unilateral or bilateral, isolated, 123570
FRMD4A	91,3	89,4	96,6	96,6	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819
FRRS1L	82,5	73,7	98,8	93,9	Developmental and epileptic encephalopathy 37, 616981
FSHB	100	100	100	100	Hypogonadotropic hypogonadism 24 without anosmia, 229070
FSHR	99,2	97	100	100	Ovarian response to FSH stimulation, 276400 Ovarian hyperstimulation syndrome, 608115 Ovarian dysgenesis 1, 233300
FTCD	97,7	93,2	100	100	Glutamate formiminotransferase deficiency, 229100
FTO	83,8	83,7	94,2	94,2	Growth retardation, developmental delay, facial dysmorphism, 612938
FUCA1	100	100	100	100	Fucosidosis, 230000
FUT8	99,8	98,9	100	100	Congenital disorder of glycosylation with defective fucosylation 1, 618005
FXN	98,3	84,7	100	100	Friedreich ataxia with retained reflexes, 229300 Friedreich ataxia, 229300
FYCO1	100	100	100	100	Cataract 18, autosomal recessive, 610019
FZD6	100	100	100	100	Nail disorder, nonsyndromic congenital, 1, 161050
G6PC	100	100	100	100	Glycogen storage disease Ia, 232200
G6PC3	100	99,9	100	100	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
GAA	100	99,9	100	100	Glycogen storage disease II, 232300

GAB1	99,9	98,9	100	100	?Deafness, autosomal recessive 26, 605428
GAD1	100	99,3	100	100	Developmental and epileptic encephalopathy 89, 619124
GALC	99,7	97,6	100	100	Krabbe disease, 245200
GALE	100	100	100	100	Galactose epimerase deficiency, 230350
GALK1	100	99,2	100	100	Galactokinase deficiency with cataracts, 230200
GALNS	100	99,3	100	100	Mucopolysaccharidosis IVA, 253000
GALNT2	99,8	97,1	100	100	Congenital disorder of glycosylation, type IIt, 618885
GALNT3	99,8	98,7	100	100	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GALT	100	99,6	100	100	Galactosemia, 230400
GAMT	95	82,7	100	100	Cerebral creatine deficiency syndrome 2, 612736
GAN	99,9	98,8	100	100	Giant axonal neuropathy-1, 256850
GAS2L2	100	99,9	100	100	?Ciliary dyskinesia, primary, 41, 618449
GAS8	99,9	99,6	100	100	Ciliary dyskinesia, primary, 33, 616726
GATM	100	100	100	100	Cerebral creatine deficiency syndrome 3, 612718 Fanconi renotubular syndrome 1, 134600
GBA	100	100	100	100	Gaucher disease, type II, 230900 Gaucher disease, type IIIC, 231005 Gaucher disease, type III, 231000 Gaucher disease, type I, 230800 Gaucher disease, perinatal lethal, 608013
GBA2	100	99,5	100	100	Spastic paraplegia 46, autosomal recessive, 614409
GBE1	99,9	99,7	100	100	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GCDH	100	99,2	100	100	Glutaricaciduria, type I, 231670
GCH1	99,9	97,3	100	100	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GCK	95,4	95,4	95,2	92,6	MODY, type II, 125851 Diabetes mellitus, permanent neonatal 1, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485 Diabetes mellitus, noninsulin-dependent, late onset, 125853
GCLC	99,4	97,1	100	99,9	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450
GCNT2	99,5	99,5	100	100	Adult i phenotype without cataract, 110800 Cataract 13 with adult i phenotype, 116700

GCSH	75,7	64,4	100	100	?Glycine encephalopathy, 605899
GDAP1	99,7	98,4	100	100	Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706 Charcot-Marie-Tooth disease, recessive intermediate, A, 608340 Charcot-Marie-Tooth disease, axonal, type 2K, 607831 Charcot-Marie-Tooth disease, type 4A, 214400
GDAP2	99,7	99	100	100	Spinocerebellar ataxia, autosomal recessive 27, 618369
GDF1	80,8	59	98,5	92	Congenital heart defects, multiple types, 6, 613854 Right atrial isomerism (Ivemark), 208530
GDF5	100	100	100	100	Du Pan syndrome, 228900 Multiple synostoses syndrome 2, 610017 Symphalangism, proximal, 1B, 615298 ?Acromesomelic dysplasia, Hunter-Thompson type, 201250 Brachydactyly, type A2, 112600 Brachydactyly, type C, 113100 Chondrodysplasia, Grebe type, 200700 Brachydactyly, type A1, C, 615072
GEMIN4	99,9	99,2	100	100	Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities, 617913
GFER	99,8	97,6	100	100	Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076
GFM1	99,7	98,7	100	100	Combined oxidative phosphorylation deficiency 1, 609060
GFM2	98,1	93,7	100	100	Combined oxidative phosphorylation deficiency 39, 618397
GFPT1	99,9	99,4	100	100	Myasthenia, congenital, 12, with tubular aggregates, 610542
GGCX	100	99,6	100	100	Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450 Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842
GGT1	19,7	18,2	100	100	?Glutathioninuria, 231950
GH1	100	100	100	100	Kowarski syndrome, 262650 Growth hormone deficiency, isolated, type II, 173100 Growth hormone deficiency, isolated, type IB, 612781 Growth hormone deficiency, isolated, type IA, 262400
GHR	99,5	99,5	99,5	99,5	Laron dwarfism, 262500 Increased responsiveness to growth hormone, 604271 Growth hormone insensitivity, partial, 604271
GHRHR	96,5	96,4	100	99,9	Growth hormone deficiency, isolated, type IV, 618157
GHSR	98,7	95,6	100	100	Growth hormone deficiency, isolated partial, 615925
GINS1	98,4	93,4	100	100	Immunodeficiency 55, 617827

GIPC3	24,9	23,2	100	99,6	Deafness, autosomal recessive 15, 601869
GJA1	100	100	100	100	Erythrokeratodermia variabilis et progressiva 3, 617525 Craniometaphyseal dysplasia, autosomal recessive, 218400 Oculodentodigital dysplasia, 164200 Hypoplastic left heart syndrome 1, 241550 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100 Oculodentodigital dysplasia, autosomal recessive, 257850 Atrioventricular septal defect 3, 600309
GJB2	100	100	100	100	Keratoderma, palmoplantar, with deafness, 148350 Deafness, autosomal recessive 1A, 220290 Deafness, autosomal dominant 3A, 601544 Hystrix-like ichthyosis with deafness, 602540 Bart-Pumphrey syndrome, 149200 Keratitis-ichthyosis-deafness syndrome, 148210 Vohwinkel syndrome, 124500
GJB6	100	100	100	100	Ectodermal dysplasia 2, Clouston type, 129500 Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290
GJC2	82,3	64,5	97,8	93,2	Lymphatic malformation 3, 613480 Spastic paraplegia 44, autosomal recessive, 613206 Leukodystrophy, hypomyelinating, 2, 608804
GLB1	99,2	92,8	100	100	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010 GM1-gangliosidosis, type II, 230600
GLDC	88,9	77,8	100	99,9	Glycine encephalopathy, 605899
GLDN	95,8	91,5	100	100	Lethal congenital contracture syndrome 11, 617194
GLE1	100	99,9	100	100	Lethal congenital contracture syndrome 1, 253310 Congenital arthrogryposis with anterior horn cell disease, 611890
GLIS2	100	99,9	100	100	Nephronophthisis 7, 611498
GLIS3	98,5	97,4	100	100	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GLRA1	100	99,8	100	100	Hyperekplexia 1, 149400
GLRB	99,1	95,3	100	100	Hyperekplexia 2, 614619

GLRX5	97,2	89,6	99,3	95,2	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GLS	96,9	88,5	100	99,9	Global developmental delay, progressive ataxia, and elevated glutamine, 618412 ?Infantile cataract, skin abnormalities, glutamate excess, and impaired intellectual development, 618339 Developmental and epileptic encephalopathy 71, 618328
GLUL	73	69	100	100	Glutamine deficiency, congenital, 610015
GLYCTK	98,7	97,3	100	100	D-glyceric aciduria, 220120
GM2A	100	100	100	100	GM2-gangliosidosis, AB variant, 272750
GMPPA	100	100	100	100	Alacrima, achalasia, and mental retardation syndrome, 615510
GMPPB	100	100	100	100	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350
GNAT2	99,9	97,9	100	100	Achromatopsia 4, 613856
GNB3	100	100	100	100	Night blindness, congenital stationary, type 1H, 617024
GNB5	99,9	96,5	100	100	Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182 Intellectual developmental disorder with cardiac arrhythmia, 617173
GNE	100	99,5	100	100	Sialuria, 269921 Nonaka myopathy, 605820
GNMT	100	100	100	100	Glycine N-methyltransferase deficiency, 606664
GNPAT	99,5	95,6	100	100	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	99,9	99,7	100	100	Mucolipidosis III alpha/beta, 252600 Mucolipidosis II alpha/beta, 252500
GNPTG	99,8	96,6	100	100	Mucolipidosis III gamma, 252605
GNRHR	100	100	100	100	Hypogonadotropic hypogonadism 7 without anosmia, 146110
GNS	99,2	94,6	100	100	Mucopolysaccharidosis type IIID, 252940
GORAB	99,7	97,2	100	100	Geroderma osteodysplasticum, 231070
GOSR2	96	95,1	100	100	Epilepsy, progressive myoclonic 6, 614018
GP1BA	97,6	94,3	100	100	Bernard-Soulier syndrome, type A1 (recessive), 231200 Bernard-Soulier syndrome, type A2 (dominant), 153670 von Willebrand disease, platelet-type, 177820
GP1BB	77,8	66,9	100	99,5	Giant platelet disorder, isolated, 231200 Bernard-Soulier syndrome, type B, 231200
GP6	100	99,9	97,7	93,5	Bleeding disorder, platelet-type, 11, 614201

GP9	98,1	91,8	100	100	Bernard-Soulier syndrome, type C, 231200
GPAA1	98,6	95,5	100	100	Glycosylphosphatidylinositol biosynthesis defect 15, 617810
GPC6	100	99,9	100	100	Omodysplasia 1, 258315
GPD1	100	99,9	100	100	Hypertriglyceridemia, transient infantile, 614480
GPHN	99,9	99,1	100	100	Molybdenum cofactor deficiency C, 615501
GPI	100	99,3	100	100	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470
GPIHBP1	100	99,9	100	100	Hyperlipoproteinemia, type 1D, 615947
GPNMB	95,5	95,5	95,5	95,5	Amyloidosis, primary localized cutaneous, 3, 617920
GPR179	100	100	100	100	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
GPR88	99,8	97,1	98,7	95,2	?Chorea, childhood-onset, with psychomotor retardation, 616939
GPSM2	99,9	99,3	100	100	Chudley-McCullough syndrome, 604213
GPT2	99,4	95,3	100	100	Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281
GPX4	91,3	88,4	98,8	96,1	Spondylometaphyseal dysplasia, Sedaghatian type, 250220
GRAP	81,6	77,3	100	100	Deafness, autosomal recessive 114, 618456
GRHPR	83,3	79,2	100	99,3	Hyperoxaluria, primary, type II, 260000
GRID2	100	99,8	100	100	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRIK2	96,1	95,3	96,3	96,3	Mental retardation, autosomal recessive, 6, 611092
GRIN1	100	99,9	100	100	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	100	99,3	100	100	Fraser syndrome 3, 617667
GRK1	100	100	100	100	Oguchi disease-2, 613411
GRM1	100	99,5	100	100	Spinocerebellar ataxia, autosomal recessive 13, 614831 Spinocerebellar ataxia 44, 617691
GRM6	93,1	83,6	98,4	96,3	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270
GRN	100	100	100	100	Aphasia, primary progressive, 607485 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706
GRXCR1	99,8	99,5	100	100	Deafness, autosomal recessive 25, 613285
GRXCR2	100	100	100	100	?Deafness, autosomal recessive 101, 615837

GSC	98,9	93	100	100	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471
GSS	96,5	96,3	100	100	Hemolytic anemia due to glutathione synthetase deficiency, 231900 Glutathione synthetase deficiency, 266130
GSX2	100	100	100	100	Diencephalic-mesencephalic junction dysplasia syndrome 2, 618646
GTF2E2	99,6	99,5	100	99,9	Trichothiodystrophy 6, nonphotosensitive, 616943
GTF2H5	72,2	71,7	72,5	72,5	Trichothiodystrophy 3, photosensitive, 616395
GTPBP2	99,8	98,5	100	100	Jaberi-Elahi syndrome, 617988
GTPBP3	100	99,9	100	100	Combined oxidative phosphorylation deficiency 23, 616198
GUCY1A1	99,9	99,8	100	100	Moyamoya 6 with achalasia, 615750
GUCY2C	99,9	99,4	100	100	Diarrhea 6, 614616 Meconium ileus, 614665
GUCY2D	100	98,7	100	100	Cone-rod dystrophy 6, 601777 ?Choroidal dystrophy, central areolar 1, 215500 Leber congenital amaurosis 1, 204000 Night blindness, congenital stationary, type 1I, 618555
GUF1	99,3	97,9	100	100	?Developmental and epileptic encephalopathy 40, 617065
GUSB	92,5	90,1	100	100	Mucopolysaccharidosis VII, 253220
GYG1	99,6	97,4	100	100	?Glycogen storage disease XV, 613507 Polyglucosan body myopathy 2, 616199
GYS1	100	98	100	100	Glycogen storage disease 0, muscle, 611556
GYS2	99,9	99,4	100	100	Glycogen storage disease 0, liver, 240600
GZF1	100	99,7	100	100	Joint laxity, short stature, and myopia, 617662
H6PD	99	99	100	100	Cortisone reductase deficiency 1, 604931
HAAO	100	100	100	100	Vertebral, cardiac, renal, and limb defects syndrome 1, 617660
HACE1	99,7	99,3	100	99,9	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADH	99,2	97,7	100	100	Hyperinsulinemic hypoglycemia, familial, 4, 609975 3-hydroxyacyl-CoA dehydrogenase deficiency, 231530
HADHA	95,5	88,3	100	100	HELLP syndrome, maternal, of pregnancy, 609016 Mitochondrial trifunctional protein deficiency, 609015 LCHAD deficiency, 609016 Fatty liver, acute, of pregnancy, 609016
HADHB	97,7	87	100	99,9	Trifunctional protein deficiency, 609015
HAMP	100	100	100	100	Hemochromatosis, type 2B, 613313

HARS1	100	100	100	100	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504
HARS2	100	99,5	100	99,8	Perrault syndrome 2, 614926
HAVCR2	100	99,8	100	100	T-cell lymphoma, subcutaneous panniculitis-like, 618398
HAX1	100	100	100	100	Neutropenia, severe congenital 3, autosomal recessive, 610738
HBB	100	100	100	100	Methemoglobinemia, beta type, 617971 Thalassemia-beta, dominant inclusion-body, 603902 Sickle cell anemia, 603903 Thalassemia, beta, 613985 Delta-beta thalassemia, 141749 Hereditary persistence of fetal hemoglobin, 141749 Heinz body anemia, 140700 Erythrocytosis 6, 617980
HELLS	98,2	91,9	100	99,9	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911
HEPACAM	86,8	78,5	100	100	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926
HEPHL1	100	99,5	100	100	?Abnormal hair, joint laxity, and developmental delay, 261990
HERC1	100	99,9	100	100	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
HERC2	79,7	76,7	100	100	Mental retardation, autosomal recessive 38, 615516
HES7	75,6	44,1	100	100	Spondylocostal dysostosis 4, autosomal recessive, 613686
HESX1	99,3	97,3	100	100	Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230 Growth hormone deficiency with pituitary anomalies, 182230
HEXA	93,8	93,1	100	100	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800
HEXB	99,4	96,6	100	100	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HFM1	95,4	89,8	100	99,9	Premature ovarian failure 9, 615724
HGD	100	99,7	100	100	Alkaptonuria, 203500
HGF	99,7	99,6	100	100	Deafness, autosomal recessive 39, 608265
HGSNAT	86,4	86,2	91,3	89,1	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544
HIBCH	98,2	84,5	100	100	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HIKESHI	98,9	91,5	100	100	Leukodystrophy, hypomyelinating, 13, 616881

HINT1	95,2	82,5	100	100	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200
HJV	100	100	100	100	Hemochromatosis, type 2A, 602390
HK1	100	99,9	100	100	Retinitis pigmentosa 79, 617460 Neuropathy, hereditary motor and sensory, Russe type, 605285 Neurodevelopmental disorder with visual defects and brain anomalies, 618547 Hemolytic anemia due to hexokinase deficiency, 235700
HLCS	100	100	100	100	Holocarboxylase synthetase deficiency, 253270
HMGCL	100	99,4	100	100	HMG-CoA lyase deficiency, 246450
HMGCS2	100	99,7	100	100	HMG-CoA synthase-2 deficiency, 605911
HMOX1	97,7	90,1	100	100	Heme oxygenase-1 deficiency, 614034
HMX1	64	43,1	99,8	96,8	Oculoauricular syndrome, 612109
HNMT	99,9	99,5	100	100	Mental retardation, autosomal recessive 51, 616739
HOGA1	99,5	95,5	100	100	Hyperoxaluria, primary, type III, 613616
HOXA1	100	100	100	100	Bosley-Salih-Alorainy syndrome, 601536 Athabaskan brainstem dysgenesis syndrome, 601536
HOXA2	99,9	99,3	100	100	Microtia with or without hearing impairment (AD), 612290 ?Microtia, hearing impairment, and cleft palate (AR), 612290
HOXB1	100	100	100	100	Facial paresis, hereditary congenital, 3, 614744
HOXC13	100	100	100	100	Ectodermal dysplasia 9, hair/nail type, 614931
HPCA	100	100	100	100	Dystonia 2, torsion, autosomal recessive, 224500
HPD	100	99,8	100	100	Hawkinsinuria, 140350 Tyrosinemia, type III, 276710
HPDL	100	100	100	100	Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026 Spastic paraplegia 83, autosomal recessive, 619027
HPGD	99,5	99,3	100	99,7	?Digital clubbing, isolated congenital, 119900 Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100 Cranioosteoarthropathy, 259100
HPS1	100	100	100	100	Hermansky-Pudlak syndrome 1, 203300
HPS3	99,8	97,1	100	100	Hermansky-Pudlak syndrome 3, 614072
HPS4	100	100	100	100	Hermansky-Pudlak syndrome 4, 614073
HPS5	99,9	99,3	100	100	Hermansky-Pudlak syndrome 5, 614074
HPS6	97,7	88,7	100	100	Hermansky-Pudlak syndrome 6, 614075

HPSE2	100	99,5	100	100	Urofacial syndrome 1, 236730
HR	98,9	96,2	100	100	Atrichia with papular lesions, 209500 Alopecia universalis, 203655
HSD11B2	87,6	83,8	99,9	97,6	Apparent mineralocorticoid excess, 218030
HSD17B3	97,8	97,8	100	100	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	95,3	92,8	96,6	96,6	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B2	100	99,7	100	100	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810
HSD3B7	98,9	95	100	100	Bile acid synthesis defect, congenital, 1, 607765
HSPA9	87,1	82,8	100	100	Even-plus syndrome, 616854 Anemia, sideroblastic, 4, 182170
HSPD1	96,7	90	100	100	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233
HSPG2	99,2	97,5	100	99,8	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
HTRA1	77,9	73,2	87,5	83,2	CARASIL syndrome, 600142 Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779
HTRA2	100	99,6	100	100	3-methylglutaconic aciduria, type VIII, 617248
HYAL1	100	100	100	100	?Mucopolysaccharidosis type IX, 601492
HYDIN	99,8	98,7	100	100	Ciliary dyskinesia, primary, 5, 608647
HYLS1	100	100	100	100	Hydrolethalus syndrome, 236680
IARS1	99,9	99,4	100	100	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093
IARS2	99,9	99,8	100	100	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
IBA57	95,4	91,7	100	100	Multiple mitochondrial dysfunctions syndrome 3, 615330 ?Spastic paraplegia 74, autosomal recessive, 616451
ICOS	99,9	99,8	100	99,9	Immunodeficiency, common variable, 1, 607594
IDH3B	95,4	95,4	100	100	Retinitis pigmentosa 46, 612572
IDUA	94,6	87,4	100	100	Mucopolysaccharidosis Is, 607016 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Ih, 607014
IER3IP1	92	80,2	100	100	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFNAR2	99,8	98,8	100	100	?Immunodeficiency 45, 616669

IFNGR1	98	97,3	100	100	Immunodeficiency 27A, mycobacteriosis, AR, 209950 Immunodeficiency 27B, mycobacteriosis, AD, 615978
IFNGR2	93,7	93,2	100	99,5	Immunodeficiency 28, mycobacteriosis, 614889
IFT122	99,9	99,2	100	100	Cranioectodermal dysplasia 1, 218330
IFT140	99,9	99,2	100	100	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 Retinitis pigmentosa 80, 617781
IFT172	99,6	98,6	100	100	Retinitis pigmentosa 71, 616394 Bardet-Biedl syndrome 20, 619471 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	100	100	100	100	Bardet-Biedl syndrome 19, 615996
IFT43	100	100	100	100	?Cranioectodermal dysplasia 3, 614099 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866
IFT52	100	99,9	100	99,9	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102
IFT57	99,9	99	100	99,7	?Orofaciodigital syndrome XVIII, 617927
IFT74	98,6	96,2	100	99,9	?Bardet-Biedl syndrome 22, 617119
IFT80	97,2	85,7	100	99,9	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IFT81	92,9	89,6	94,9	94,6	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IGF1	99,8	99,8	100	100	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IGF1R	100	99,7	100	100	Insulin-like growth factor I, resistance to, 270450
IGFALS	100	100	100	100	Acid-labile subunit, deficiency of, 615961
IGFBP7	93,6	88,5	100	100	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis, 614224
IGHM	100	100	100	100	Agammaglobulinemia 1, 601495
IGHMBP2	99,3	96,9	100	100	Neuronopathy, distal hereditary motor, type VI, 604320 Charcot-Marie-Tooth disease, axonal, type 2S, 616155
IGKC	100	100	100	100	Kappa light chain deficiency, 614102
IGLL1	100	99,7	100	100	Agammaglobulinemia 2, 613500
IHH	100	100	100	100	Acrocapitofemoral dysplasia, 607778 Brachydactyly, type A1, 112500
IKBKB	99	96,1	100	100	Immunodeficiency 15B, 615592 Immunodeficiency 15A, 618204
IL10RA	100	99,9	100	100	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	99,9	97,8	100	100	Inflammatory bowel disease 25, early onset, autosomal recessive, 612567

IL11RA	100	99,6	100	100	Craniosynostosis and dental anomalies, 614188
IL12B	100	99,1	100	100	Immunodeficiency 29, mycobacteriosis, 614890
IL12RB1	98,7	96,1	94,1	94,1	Immunodeficiency 30, 614891
IL17RA	100	99,8	100	100	Immunodeficiency 51, 613953
IL17RC	100	99,9	100	100	Candidiasis, familial, 9, 616445
IL1RN	100	99,9	100	100	Interleukin 1 receptor antagonist deficiency, 612852
IL21R	100	100	100	100	Immunodeficiency 56, 615207
IL2RA	100	99,1	100	100	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367
IL2RB	100	99,8	100	100	Immunodeficiency 63 with lymphoproliferation and autoimmunity, 618495
IL36RN	100	99,9	100	100	Psoriasis 14, pustular, 614204
IL6ST	94,9	89,4	100	100	Hyper-IgE recurrent infection syndrome 4, autosomal recessive, 618523
IL7R	99,9	99,3	100	100	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
ILDR1	99,1	97,1	100	100	Deafness, autosomal recessive 42, 609646
IMPA1	96,1	86,4	100	99,8	Mental retardation, autosomal recessive 59, 617323
IMPG2	99,4	97,9	100	100	Retinitis pigmentosa 56, 613581 Macular dystrophy, vitelliform, 5, 616152
INPP5E	96,9	93,2	100	100	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INPP5K	100	99,7	100	100	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404
INPPL1	98,6	94,4	100	99,9	Opsismodysplasia, 258480
INSR	97,3	93	100	99,6	Rabson-Mendenhall syndrome, 262190 Leprechaunism, 246200 Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968
INTS1	99,8	98,6	100	100	Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies, 618571
INTS8	99,6	98,8	100	99,9	?Neurodevelopmental disorder with cerebellar hypoplasia and spasticity, 618572
INTU	99,9	98,6	100	100	?Orofaciodigital syndrome XVII, 617926 ?Short-rib thoracic dysplasia 20 with polydactyly, 617925
INVS	100	99,8	100	100	Nephronophthisis 2, infantile, 602088
IQCB1	92,8	82,8	100	100	Senior-Loken syndrome 5, 609254
IQSEC1	88,6	86,1	97,6	94,8	Intellectual developmental disorder with short stature and behavioral abnormalities, 618687

IRAK4	99,5	95,7	100	99,8	Immunodeficiency 67, 607676
IREB2	99,9	99,8	100	100	Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451
IRF7	100	99,8	100	100	?Immunodeficiency 39, 616345
IRF8	98,7	96	100	100	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893 Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990
IRF9	100	99,9	100	100	Immunodeficiency 65, susceptibility to viral infections, 618648
IRX5	100	98,8	100	99,9	Hamamy syndrome, 611174
ISCA1	89,5	76,2	95,1	95,1	Multiple mitochondrial dysfunctions syndrome 5, 617613
ISCA2	99,8	96,5	100	100	Multiple mitochondrial dysfunctions syndrome 4, 616370
ISCU	100	100	100	100	Myopathy with lactic acidosis, hereditary, 255125
ISG15	100	100	100	100	Immunodeficiency 38, 616126
ITCH	91,5	90,8	95,3	93,1	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITGA2B	99,7	97,2	100	100	Glanzmann thrombasthenia 1, 273800 Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Thrombocytopenia, neonatal alloimmune, BAK antigen related,
ITGA3	99,6	97,9	100	100	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748
ITGA6	99,8	98	100	100	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730
ITGA7	99,7	97,9	100	100	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
ITGA8	99,9	99,3	100	100	Renal hypodysplasia/aplasia 1, 191830
ITGB2	97,2	97,2	97,2	97,2	Leukocyte adhesion deficiency, 116920
ITGB3	100	99	100	100	Bleeding disorder, platelet-type, 24, autosomal dominant, 619271 Glanzmann thrombasthenia 2, 619267 Thrombocytopenia, neonatal alloimmune, Purpura, posttransfusion,
ITGB4	98,7	96,4	100	100	Epidermolysis bullosa of hands and feet, 131800 Epidermolysis bullosa, junctional, with pyloric atresia, 226730 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
ITGB6	97,4	95,7	100	100	Amelogenesis imperfecta, type IH, 616221
ITK	99,8	98,6	100	100	Lymphoproliferative syndrome 1, 613011
ITPA	100	100	100	100	Developmental and epileptic encephalopathy 35, 616647

ITPR1	100	99,5	100	100	Gillespie syndrome, 206700 Spinocerebellar ataxia 29, congenital nonprogressive, 117360 Spinocerebellar ataxia 15, 606658
IVD	100	99,9	100	100	Isovaleric acidemia, 243500
IYD	99,4	94,5	100	100	Thyroid dyshormonogenesis 4, 274800
JAGN1	100	100	100	99,2	Neutropenia, severe congenital, 6, autosomal recessive, 616022
JAK3	99,5	97,6	100	100	SCID, autosomal recessive, T-negative/B-positive type, 600802
JAM2	99,9	99,7	92,3	92,3	Basal ganglia calcification, idiopathic, 8, autosomal recessive, 618824
JAM3	100	100	100	100	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
JPH1	100	99,8	100	100	?Charcot-Marie-Tooth disease, axonal, autosomal dominant, type 2K, 607831
JUP	100	99,8	100	100	Naxos disease, 601214 ?Arrhythmogenic right ventricular dysplasia 12, 611528
KALRN	99,9	99,4	100	100	No OMIM disease ID
KANK2	100	100	100	100	Nephrotic syndrome, type 16, 617783 Palmoplantar keratoderma and woolly hair, 616099
KARS1	99,9	98,9	100	100	Deafness, autosomal recessive 89, 613916 Leukoencephalopathy, progressive, infantile-onset, with or without deafness, 619147 ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness, congenital, and adult-onset progressive leukoencephalopathy, 619196
KATNB1	100	100	100	100	Lissencephaly 6, with microcephaly, 616212
KIAA0556	100	99,6	100	100	Joubert syndrome 26, 616784
KCNE1	100	100	100	100	Jervell and Lange-Nielsen syndrome 2, 612347 Long QT syndrome 5, 613695
KCNJ1	100	100	100	100	Bartter syndrome, type 2, 241200
KCNJ10	89,2	88,5	100	100	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ11	100	100	100	100	Diabetes, permanent neonatal 2, with or without neurologic features, 618856 Maturity-onset diabetes of the young, type 13, 616329 Diabetes mellitus, transient neonatal 3, 610582 Hyperinsulinemic hypoglycemia, familial, 2, 601820
KCNJ13	100	100	100	100	Snowflake vitreoretinal degeneration, 193230 Leber congenital amaurosis 16, 614186

KCNMA1	94	93	100	100	Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446 Cerebellar atrophy, developmental delay, and seizures, 617643 Liang-Wang syndrome, 618729
KCNQ1	93,5	90,6	99,9	99,4	Short QT syndrome 2, 609621 Atrial fibrillation, familial, 3, 607554 Long QT syndrome 1, 192500 Jervell and Lange-Nielsen syndrome, 220400
KCNV2	100	99,7	100	100	Retinal cone dystrophy 3B, 610356
KCTD7	95	95	100	100	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM5B	93,4	90,7	94,7	93,3	Mental retardation, autosomal recessive 65, 618109
KERA	100	100	100	100	Cornea plana 2, autosomal recessive, 217300
KHDC3L	100	99,8	100	100	Hydatidiform mole, recurrent, 2, 614293
KIAA0586	97,1	92	95,8	95,7	Short-rib thoracic dysplasia 14 with polydactyly, 616546 Joubert syndrome 23, 616490
KIAA0753	99,9	98,9	100	100	?Orofaciodigital syndrome XV, 617127 ?Joubert syndrome 38, 619476 Short-rib thoracic dysplasia 21 without polydactyly, 619479
KIAA1109	99,8	99	100	100	Alkuraya-Kucinskas syndrome, 617822
KIAA1549	97,8	96,3	99	98,4	Retinitis pigmentosa 86, 618613
KIF14	99,2	96,8	100	99,9	Microcephaly 20, primary, autosomal recessive, 617914 ?Meckel syndrome 12, 616258
KIF1A	97,4	95,3	98	98	NESCAV syndrome, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal dominant, 610357 Spastic paraplegia 30, autosomal recessive, 610357
KIF1C	100	99,7	100	100	Spastic ataxia 2, autosomal recessive, 611302
KIF7	93,6	91,9	99,7	98,6	Joubert syndrome 12, 200990 Acrocallosal syndrome, 200990 ?Hydrolethalus syndrome 2, 614120 ?Al-Gazali-Bakalinova syndrome, 607131
KIFBP	96,1	96	96,1	96,1	Goldberg-Shprintzen megacolon syndrome, 609460
KISS1R	100	99,6	100	100	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 ?Precocious puberty, central, 1, 176400
KIZ	99,8	98,4	100	100	Retinitis pigmentosa 69, 615780

KL	98,5	97,5	98,7	97,9	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994
KLC2	99,2	98	100	100	Spastic paraplegia, optic atrophy, and neuropathy, 609541
KLHL3	100	98,9	100	100	Pseudohypoaldosteronism, type IID, 614495
KLHL40	100	100	100	100	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	99,9	99,4	100	100	Nemaline myopathy 9, 615731
KLHL7	99,8	99,7	100	100	Retinitis pigmentosa 42, 612943 PERCHING syndrome, 617055
KLK4	100	100	100	100	Amelogenesis imperfecta, type IIA1, 204700
KLKB1	99,8	99,3	100	99,9	Fletcher factor (prekallikrein) deficiency, 612423
KMT2B	96,2	94	98,5	97,8	Dystonia 28, childhood-onset, 617284
KNL1	99,1	97,3	98,9	98,7	Microcephaly 4, primary, autosomal recessive, 604321
KPTN	100	100	100	100	Mental retardation, autosomal recessive 41, 615637
KRT10	99,9	99,1	100	100	Epidermolytic hyperkeratosis, 113800 Ichthyosis with confetti, 609165 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602
KRT14	89,6	81,6	100	100	Epidermolysis bullosa simplex, recessive 1, 601001 Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 Dermatopathia pigmentosa reticularis, 125595 Epidermolysis bullosa simplex, Koebner type, 131900 Naegeli-Franceschetti-Jadassohn syndrome, 161000 Epidermolysis bullosa simplex, Dowling-Meara type, 131760
KRT18	83,2	66,1	100	100	Cirrhosis, cryptogenic, 215600
KRT5	100	99,7	100	100	Dowling-Degos disease 1, 179850 Epidermolysis bullosa simplex-MP, 131960 Epidermolysis bullosa simplex, Koebner type, 131900 Epidermolysis bullosa simplex-MCR, 609352 Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 Epidermolysis bullosa simplex, recessive 1, 601001 Epidermolysis bullosa simplex, Dowling-Meara type, 131760
KRT8	90,3	69,5	100	100	Cirrhosis, cryptogenic, 215600
KRT85	99,1	94,5	100	100	Ectodermal dysplasia 4, hair/nail type, 602032
KY	100	99,6	100	100	Myopathy, myofibrillar, 7, 617114
KYNU	99,5	96,7	100	100	?Hydroxykynureninuria, 236800 Vertebral, cardiac, renal, and limb defects syndrome 2, 617661

L2HGDH	98,9	96,4	100	100	L-2-hydroxyglutaric aciduria, 236792
LAMA1	99,9	99,3	100	100	Poretti-Boltshauser syndrome, 615960
LAMA2	99,9	99,1	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855
LAMA3	99,9	99,3	100	100	Laryngoonychocutaneous syndrome, 245660 Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, generalized atrophic benign, 226650
LAMB1	100	99,6	100	100	Lissencephaly 5, 615191
LAMB2	99,9	99,3	100	100	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 Pierson syndrome, 609049
LAMB3	99,9	98,8	100	100	Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa, junctional, Herlitz type, 226700 Amelogenesis imperfecta, type IA, 104530
LAMC2	99,4	96,8	100	100	Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa, junctional, Herlitz type, 226700
LAMC3	98,9	97,5	100	99,8	Cortical malformations, occipital, 614115
LAMTOR2	100	100	100	100	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LARGE1	100	99,7	100	100	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	86,6	75,4	100	99,9	Alazami syndrome, 615071
LARS1	99,4	97,2	100	99,9	?Infantile liver failure syndrome 1, 615438
LARS2	100	100	100	100	Perrault syndrome 4, 615300 Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LAT	100	99,4	100	100	Immunodeficiency 52, 617514
LBR	97,9	91	100	100	Pelger-Huet anomaly, 169400 Pelger-Huet anomaly with mild skeletal anomalies, 618019 ?Reynolds syndrome, 613471 Greenberg skeletal dysplasia, 215140
LCA5	99,6	97,9	100	100	Leber congenital amaurosis 5, 604537
LCAT	98,8	93,3	100	100	Fish-eye disease, 136120 Norum disease, 245900
LCK	98,2	96,1	100	100	?Immunodeficiency 22, 615758
LCT	99,6	97,4	100	100	Lactase deficiency, congenital, 223000
LDHA	94,4	89,3	100	100	Glycogen storage disease XI, 612933

LDHD	100	99,6	100	100	D-lactic aciduria with susceptibility to gout, 245450
LDLRAP1	98,9	94	100	100	Hypercholesterolemia, familial, 4, 603813
LEMD2	99,9	96,1	100	100	Marbach-Rustad progeroid syndrome, 619322 Cataract 46, juvenile-onset, 212500
LEP	100	99,6	100	100	Obesity, morbid, due to leptin deficiency, 614962
LEPR	94,1	92,3	94,6	94,5	Obesity, morbid, due to leptin receptor deficiency, 614963
LFNG	88,6	86,5	92	87,3	Spondylocostal dysostosis 3, autosomal recessive, 609813
LGI4	99,7	97,9	100	100	Arthrogryposis multiplex congenita 1, neurogenic, with myelin defect, 617468
LHB	91,7	42,8	100	100	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300
LHCGR	96,6	92,4	100	100	Leydig cell adenoma, somatic, with precocious puberty, 176410 Leydig cell hypoplasia with pseudohermaphroditism, 238320 Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320 Luteinizing hormone resistance, female, 238320 Precocious puberty, male, 176410
LHFPL5	100	100	100	100	Deafness, autosomal recessive 67, 610265
LHX3	96,6	96,2	100	100	Pituitary hormone deficiency, combined, 3, 221750
LIAS	99,8	98,9	100	100	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIFR	99,3	97,8	100	99,9	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LIG4	99,8	99,3	100	100	LIG4 syndrome, 606593
LIM2	100	99,8	100	100	Cataract 19, multiple types, 615277
LIMS2	94,1	92,7	100	99,7	?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827
LINGO1	100	100	100	100	Mental retardation, autosomal recessive 64, 618103
LINS1	99,8	98,8	100	100	Mental retardation, autosomal recessive 27, 614340
LIPA	96,9	94,6	95,2	95,2	Wolman disease, 278000 Cholesteryl ester storage disease, 278000
LIPC	100	99,4	100	100	Hepatic lipase deficiency, 614025
LIPE	100	99,2	100	100	Lipodystrophy, familial partial, type 6, 615980
LIPH	100	99	100	100	Hypotrichosis 7, 604379 Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379
LIPN	99,6	99,4	100	100	Ichthyosis, congenital, autosomal recessive 8, 613943
LIPT1	99,7	99,5	100	100	Lipoyltransferase 1 deficiency, 616299

LIPT2	98,4	82,4	100	100	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668
LMAN1	99,8	98,5	100	100	Combined factor V and VIII deficiency, 227300
LMAN2L	100	99,5	100	100	?Mental retardation, autosomal recessive, 52, 616887
LMBR1	98,1	94,7	98,7	98,5	Triphalangeal thumb, type I, 174500 Syndactyly, type IV, 186200 Laurin-Sandrow syndrome, 135750 Hypoplastic or aplastic tibia with polydactyly, 188740 Polydactyly, preaxial type II, 174500 Acheiropody, 200500 Triphalangeal thumb-polysyndactyly syndrome, 174500
LMBRD1	94,1	89,1	96,1	95,7	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LMF1	100	99,7	100	100	Lipase deficiency, combined, 246650
LMNA	96,1	90,6	100	100	Mandibuloacral dysplasia, 248370 Heart-hand syndrome, Slovenian type, 610140 Cardiomyopathy, dilated, 1A, 115200 Restrictive dermopathy, lethal, 275210 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 Charcot-Marie-Tooth disease, type 2B1, 605588 Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 Hutchinson-Gilford progeria, 176670 Lipodystrophy, familial partial, type 2, 151660 Muscular dystrophy, congenital, 613205 Malouf syndrome, 212112
LMNB2	98,3	95,5	97,9	96,7	Microcephaly 27, primary, autosomal dominant, 619180 ?Epilepsy, progressive myoclonic, 9, 616540
LMOD3	99,8	99	100	100	Nemaline myopathy 10, 616165
LNPK	97,2	92,1	93,3	93,2	Neurodevelopmental disorder with epilepsy and hypoplasia of the corpus callosum, 618090
LONP1	100	99,9	100	100	CODAS syndrome, 600373
LOXHD1	99,9	98,7	100	100	Deafness, autosomal recessive 77, 613079
LPAR6	99,3	96,8	100	100	Hypotrichosis 8, 278150 Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150
LPIN1	99,4	97,2	100	100	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	99,9	99,7	100	100	Majeed syndrome, 609628
LPL	100	100	100	100	Lipoprotein lipase deficiency, 238600 Combined hyperlipidemia, familial, 144250

LRAT	100	100	100	100	Leber congenital amaurosis 14, 613341 Retinal dystrophy, early-onset severe, 613341 Retinitis pigmentosa, juvenile, 613341
LRBA	99,9	99,7	100	100	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRIG2	99,8	99,2	100	100	Urofacial syndrome 2, 615112
LRIT3	94,1	92,2	100	100	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRMDA	97,4	95,5	99,6	99,6	Albinism, oculocutaneous, type VII, 615179
LRP1	99,8	99,1	100	100	?Keratosis pilaris atrophicans, 604093
LRP2	100	99,8	100	100	Donnai-Barrow syndrome, 222448
LRP4	99,1	98,4	100	100	?Myasthenic syndrome, congenital, 17, 616304 Sclerosteosis 2, 614305 Cenani-Lenz syndactyly syndrome, 212780
LRP5	99,2	98,2	99,8	99,2	Osteopetrosis, autosomal dominant 1, 607634 Hyperostosis, endosteal, 144750 Osteosclerosis, 144750 Polycystic liver disease 4 with or without kidney cysts, 617875 Osteoporosis-pseudoglioma syndrome, 259770 Exudative vitreoretinopathy 4, 601813 van Buchem disease, type 2, 607636
LRPAP1	100	99,9	100	100	Myopia 23, autosomal recessive, 615431
LRPPRC	99,7	99,3	100	99,9	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111
LRRC56	99,8	99	100	100	Ciliary dyskinesia, primary, 39, 618254
LRSAM1	100	100	100	100	Charcot-Marie-Tooth disease, axonal, type 2P, 614436
LRTOMT	100	99,2	100	100	Deafness, autosomal recessive 63, 611451
LSS	100	99,4	100	100	Hypotrichosis 14, 618275 Cataract 44, 616509 Alopecia-mental retardation syndrome 4, 618840
LTBP2	99,8	98,9	100	100	Glaucoma 3, primary congenital, D, 613086 Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750 ?Weill-Marchesani syndrome 3, recessive, 614819
LTBP3	99,8	98,6	100	99,9	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
LTBP4	99,9	98,1	100	100	Cutis laxa, autosomal recessive, type IC, 613177
LTC4S	76,4	69	100	100	No OMIM disease ID

LYRM4	66,7	65,6	66,3	66,3	?Combined oxidative phosphorylation deficiency 19, 615595
LYRM7	95,6	86,5	100	99,2	Mitochondrial complex III deficiency, nuclear type 8, 615838
LYST	99,4	97,8	100	100	Chediak-Higashi syndrome, 214500
LZTFL1	99,7	99,4	100	99,9	Bardet-Biedl syndrome 17, 615994
LZTR1	100	99,9	100	100	Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
MAB21L1	100	100	100	100	Cerebellar, ocular, craniofacial, and genital syndrome, 618479
MAB21L2	100	100	100	100	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877
MAD2L2	100	99,9	100	100	?Fanconi anemia, complementation group V, 617243
MADD	100	99,2	100	100	Neurodevelopmental disorder with dysmorphic facies, impaired speech and hypotonia, 619005 DEEAH syndrome, 619004
MAG	100	100	100	100	Spastic paraplegia 75, autosomal recessive, 616680
MAGI2	94,2	91	94,7	93,3	Nephrotic syndrome, type 15, 617609
MAK	99,7	97,9	100	100	Retinitis pigmentosa 62, 614181
MALT1	90,9	87,7	100	99,9	Immunodeficiency 12, 615468
MAN1B1	100	99,7	100	100	Rafiq syndrome, 614202
MAN2B1	99,6	97,4	100	100	Mannosidosis, alpha-, types I and II, 248500
MANBA	87,1	84,9	100	99,9	Mannosidosis, beta, 248510
MAP3K20	99,9	99,2	100	99,9	Centronuclear myopathy 6 with fiber-type disproportion, 617760 Split-foot malformation with mesoaxial polydactyly, 616890
MAPKBP1	100	100	100	100	Nephronophthisis 20, 617271
MAPT	99,9	98,9	100	100	Supranuclear palsy, progressive, 601104 Supranuclear palsy, progressive atypical, 260540 Dementia, frontotemporal, with or without parkinsonism, 600274 Pick disease, 172700
MARS1	99	96,1	100	100	Interstitial lung and liver disease, 615486 Charcot-Marie-Tooth disease, axonal, type 2U, 616280
MARS2	100	100	100	100	?Combined oxidative phosphorylation deficiency 25, 616430 Spastic ataxia 3, autosomal recessive, 611390
MARVELD2	98,5	94,9	100	100	Deafness, autosomal recessive 49, 610153
MASP1	100	99,6	100	100	3MC syndrome 1, 257920

MAT1A	99,9	98,5	100	100	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MATN3	84,8	84,1	100	100	Spondyloepimetaphyseal dysplasia, Borochowitz-Cormier-Daire type, 608728 Epiphyseal dysplasia, multiple, 5, 607078
MBOAT7	100	99,3	100	100	Mental retardation, autosomal recessive 57, 617188
MBTPS1	99,4	97,3	100	100	?Spondyloepiphyseal dysplasia, Kondo-Fu type, 618392
MC2R	99,7	97,4	100	100	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MCCC1	99,9	98,7	100	100	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	99,9	99,1	100	100	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	100	100	100	100	Methylmalonyl-CoA epimerase deficiency, 251120
MCFD2	99,4	94,9	100	100	Factor V and factor VIII, combined deficiency of, 613625
MCM3AP	99,9	99,1	100	100	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124
MCM4	95,3	95	95,5	95,5	Immunodeficiency 54, 609981
MCM5	100	99,7	100	100	?Meier-Gorlin syndrome 8, 617564
MCM9	99,9	99	100	100	Ovarian dysgenesis 4, 616185
MCOLN1	99,8	98,8	100	100	Mucolipidosis IV, 252650
MCPH1	99,8	98,6	100	100	Microcephaly 1, primary, autosomal recessive, 251200
MDH2	98	98	100	100	Developmental and epileptic encephalopathy 51, 617339
MECR	100	98,7	100	100	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MED17	95,8	92,4	100	100	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	99,9	99	100	100	Mental retardation, autosomal recessive 18, 614249
MED25	100	99,9	100	99,9	Basel-Vanagait-Smirin-Yosef syndrome, 616449
MEFV	99,6	97,6	96,4	96,4	Neutrophilic dermatosis, acute febrile, 608068 Familial Mediterranean fever, AR, 249100 Familial Mediterranean fever, AD, 134610
MEGF10	100	99,9	100	100	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399
MEGF8	100	99,2	100	100	Carpenter syndrome 2, 614976
MEOX1	100	98	100	100	Klippel-Feil syndrome 2, 214300
MERTK	99,4	98,6	99,1	99,1	Retinitis pigmentosa 38, 613862

MESD	100	98,8	100	100	Osteogenesis imperfecta, type XX, 618644
MESP2	95,7	90,6	97,5	97,5	Spondylocostal dysostosis 2, autosomal recessive, 608681
MET	100	99,4	100	100	Renal cell carcinoma, papillary, 1, familial and somatic, 605074 Hepatocellular carcinoma, childhood type, somatic, 114550 ?Deafness, autosomal recessive 97, 616705
METTL23	100	100	100	100	Mental retardation, autosomal recessive 44, 615942
MFF	93,9	89,4	100	100	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFN2	100	99,8	100	100	Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260 Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 Hereditary motor and sensory neuropathy VIA, 601152
MFRP	100	100	100	100	Microphthalmia, isolated 5, 611040 Nanophthalmos 2, 609549
MFSD2A	99,5	97,3	100	100	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities, 616486
MFSD8	99,6	99,4	100	100	Macular dystrophy with central cone involvement, 616170 Ceroid lipofuscinosis, neuronal, 7, 610951
MGAT2	100	99,9	100	100	Congenital disorder of glycosylation, type IIa, 212066
MGME1	100	99,9	100	100	Mitochondrial DNA depletion syndrome 11, 615084
MGP	98,7	93,6	100	100	Keutel syndrome, 245150
MICOS13	100	98,9	100	99,9	Combined oxidative phosphorylation deficiency 37, 618329
MICU1	97,3	92,2	100	100	Myopathy with extrapyramidal signs, 615673
MIPEP	99,5	97,1	100	100	Combined oxidative phosphorylation deficiency 31, 617228
MITF	100	99,9	100	100	Waardenburg syndrome, type 2A, 193510 Tietz albinism-deafness syndrome, 103500 Waardenburg syndrome/ocular albinism, digenic, 103470 COMMAD syndrome, 617306
MKKS	100	100	100	100	McKusick-Kaufman syndrome, 236700 Bardet-Biedl syndrome 6, 605231
MKS1	99,4	96,3	100	100	Bardet-Biedl syndrome 13, 615990 Meckel syndrome 1, 249000 Joubert syndrome 28, 617121
MLC1	100	98,8	100	100	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MLH1	100	99,9	100	100	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Muir-Torre syndrome, 158320 Mismatch repair cancer syndrome 1, 276300

MLPH	100	98,5	100	100	Griscelli syndrome, type 3, 609227
MLYCD	96,8	92,5	100	99,4	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	100	100	100	100	Methylmalonic aciduria, vitamin B12-responsive, cblA type, 251100
MMAB	100	99,9	100	100	Methylmalonic aciduria, vitamin B12-responsive, cblB type, 251110
MMACHC	100	100	100	100	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	91,6	81,3	89,7	89,7	Methylmalonic aciduria, cblD type, variant 2, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Homocystinuria, cblD type, variant 1, 277410
MME	99,7	98,6	98	97,9	?Spinocerebellar ataxia 43, 617018 Charcot-Marie-Tooth disease, axonal, type 2T, 617017
MMP13	93,6	92,2	92,4	92,3	?Spondyloepimetaphyseal dysplasia, Missouri type, 602111 Metaphyseal anadysplasia 1, 602111 Metaphyseal dysplasia, Spahr type, 250400
MMP14	100	99,4	100	100	?Winchester syndrome, 277950
MMP2	100	100	100	100	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP20	99,9	99,3	100	100	Amelogenesis imperfecta, type IIA2, 612529
MMP21	99,8	99,2	100	100	Heterotaxy, visceral, 7, autosomal, 616749
MMP9	99,6	96,8	100	100	Metaphyseal anadysplasia 2, 613073
MMUT	99,7	98,2	100	100	Methylmalonic aciduria, mut(0) type, 251000
MOCOS	99,9	97,8	100	100	Xanthinuria, type II, 603592
MOCS1	98,9	95,5	100	100	Molybdenum cofactor deficiency A, 252150
MOCS2	99,4	99,4	100	100	Molybdenum cofactor deficiency B, 252160
MOGS	100	99,9	100	100	Congenital disorder of glycosylation, type IIb, 606056
MPC1	100	99,6	100	100	Mitochondrial pyruvate carrier deficiency, 614741
MPDU1	100	99,2	100	100	Congenital disorder of glycosylation, type If, 609180
MPDZ	99,8	98,5	100	100	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219
MPI	100	99,5	100	100	Congenital disorder of glycosylation, type Ib, 602579
MPL	100	99,8	100	100	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498
MPLKIP	100	99,4	100	100	Trichothiodystrophy 4, nonphotosensitive, 234050

MPO	99,9	98,7	100	100	Myeloperoxidase deficiency, 254600
MPV17	100	98,7	100	100	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MPZ	85,6	81,9	100	100	Charcot-Marie-Tooth disease, type 2I, 607677 Dejerine-Sottas disease, 145900 Charcot-Marie-Tooth disease, type 1B, 118200 Roussy-Levy syndrome, 180800 Charcot-Marie-Tooth disease, dominant intermediate D, 607791 Hypomyelinating neuropathy, congenital, 2, 618184 Charcot-Marie-Tooth disease, type 2J, 607736
MPZL2	100	99,9	100	100	Deafness, autosomal recessive 111, 618145
MRAP	100	100	100	100	Glucocorticoid deficiency 2, 607398
MRE11	98,2	88,6	100	100	Ataxia-telangiectasia-like disorder 1, 604391
MRM2	100	98,9	98,9	98,9	?Mitochondrial DNA depletion syndrome 17, 618567
MRPL3	91,7	82,1	100	100	Combined oxidative phosphorylation deficiency 9, 614582
MRPL44	99,5	97,4	100	100	?Combined oxidative phosphorylation deficiency 16, 615395
MRPS14	100	100	100	100	?Combined oxidative phosphorylation deficiency 38, 618378
MRPS16	100	98,8	100	100	Combined oxidative phosphorylation deficiency 2, 610498
MRPS2	99,6	97	100	100	Combined oxidative phosphorylation deficiency 36, 617950
MRPS22	99,7	98,3	100	100	Ovarian dysgenesis 7, 618117 Combined oxidative phosphorylation deficiency 5, 611719
MRPS34	98,6	93,3	100	100	Combined oxidative phosphorylation deficiency 32, 617664
MRPS7	100	100	100	100	?Combined oxidative phosphorylation deficiency 34, 617872
MS4A1	99,5	96,5	100	99,9	?Immunodeficiency, common variable, 5, 613495
MSH2	98,5	94,5	100	100	Muir-Torre syndrome, 158320 Colorectal cancer, hereditary nonpolyposis, type 1, 120435 Mismatch repair cancer syndrome 2, 619096
MSH3	97,8	97,1	100	100	Familial adenomatous polyposis 4, 617100 Endometrial carcinoma, somatic, 608089
MSH6	100	99,3	100	100	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 Mismatch repair cancer syndrome 3, 619097
MSMO1	93,1	86,8	100	100	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MSRB3	99,8	99,8	100	100	Deafness, autosomal recessive 74, 613718

MSTO1	99	96,3	100	100	Myopathy, mitochondrial, and ataxia, 617675
MTFMT	99,9	99,5	100	100	Combined oxidative phosphorylation deficiency 15, 614947 Mitochondrial complex I deficiency, nuclear type 27, 618248
MTHFD1	99,9	98,4	100	100	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780
MTHFR	97,3	95,9	100	100	Homocystinuria due to MTHFR deficiency, 236250
MTHFS	75	75	100	100	Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367
MTMR2	99,5	98,4	100	100	Charcot-Marie-Tooth disease, type 4B1, 601382
MTO1	90,9	88,8	92,8	91,4	Combined oxidative phosphorylation deficiency 10, 614702
MTPAP	99,1	94,1	100	100	?Spastic ataxia 4, autosomal recessive, 613672
MTR	100	99,9	100	100	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940
C12orf65	99	94,5	100	100	Spastic paraplegia 55, autosomal recessive, 615035 Combined oxidative phosphorylation deficiency 7, 613559
MTRR	99,8	98,4	100	100	Homocystinuria-megaloblastic anemia, cbl E type, 236270
MTTP	99,9	99,2	100	100	Abetalipoproteinemia, 200100
MUSK	100	99,9	100	100	Fetal akinesia deformation sequence 1, 208150 Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325
MUTYH	100	100	100	100	Adenomas, multiple colorectal, 608456 Gastric cancer, somatic, 613659
MVK	91,4	90,5	90,5	90,5	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377
MYBPC1	99,8	99,1	100	99,9	Myopathy, congenital, with tremor, 618524 Lethal congenital contracture syndrome 4, 614915 Arthrogryposis, distal, type 1B, 614335
MYD88	100	99,5	100	100	Macroglobulinemia, Waldenstrom, somatic, 153600 Immunodeficiency 68, 612260
MYF5	100	100	100	100	Ophthalmoplegia, external, with rib and vertebral anomalies, 618155
MYH2	99,9	99,1	100	100	Proximal myopathy and ophthalmoplegia, 605637
MYH3	99,9	98,4	100	100	Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A, 178110 Contractures, pterygia, and spondylocarpotarsal fusion syndrome 1B, 618469 Arthrogryposis, distal, type 2B3 (Sheldon-Hall), 618436 Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700
MYL1	99,8	99,1	100	100	Myopathy, congenital, with fast-twitch (type II) fiber atrophy, 618414

MYL3	100	100	100	100	Cardiomyopathy, hypertrophic, 8, 608751
MYLK	100	99,6	100	100	Megacystis-microcolon-intestinal hypoperistalsis syndrome 1, 249210 Aortic aneurysm, familial thoracic 7, 613780
MYMK	100	100	100	100	Carey-Fineman-Ziter syndrome, 254940
MYO15A	99,1	97,6	100	99,8	Deafness, autosomal recessive 3, 600316
MYO18B	100	99,3	100	100	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549
MYO1E	99,9	98,6	100	100	Glomerulosclerosis, focal segmental, 6, 614131
MYO3A	99,1	95,4	100	99,9	Deafness, autosomal recessive 30, 607101
MYO5A	99,6	98,3	100	100	Griscelli syndrome, type 1, 214450
MYO5B	98,5	94,8	100	100	Diarrhea 2, with microvillus atrophy, 251850
MYO6	99,1	96,3	100	100	Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346 Deafness, autosomal dominant 22, 606346 Deafness, autosomal recessive 37, 607821
MYO7A	99,7	98,3	100	100	Deafness, autosomal recessive 2, 600060 Usher syndrome, type 1B, 276900 Deafness, autosomal dominant 11, 601317
MYO9A	99,8	98,9	100	100	Myasthenic syndrome, congenital, 24, presynaptic, 618198
MYORG	100	100	100	100	Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317
MYPN	100	99,5	100	100	Cardiomyopathy, hypertrophic, 22, 615248 Cardiomyopathy, familial restrictive, 4, 615248 Cardiomyopathy, dilated, 1KK, 615248 Nemaline myopathy 11, autosomal recessive, 617336
MYSM1	96,1	95,4	96,4	96,3	Bone marrow failure syndrome 4, 618116
NADK2	99,7	99,3	99,5	96,8	2,4-dienoyl-CoA reductase deficiency, 616034
NADSYN1	100	100	100	100	Vertebral, cardiac, renal, and limb defects syndrome 3, 618845
NAGA	100	100	100	100	Schindler disease, type I, 609241 Kanzaki disease, 609242 Schindler disease, type III, 609241
NAGLU	93,8	91,7	99,9	98,7	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NAGS	99,9	97,9	100	100	N-acetylglutamate synthase deficiency, 237310
NALCN	99,7	98,9	99,8	99,7	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419

NANS	100	99,9	100	100	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NARS2	97,9	97,1	100	99,9	Combined oxidative phosphorylation deficiency 24, 616239 ?Deafness, autosomal recessive 94, 618434
NAT8L	100	98	92,9	87,1	?N-acetylaspartate deficiency, 614063
NAXD	100	99,9	100	100	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321
NAXE	100	98,6	100	100	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NBAS	99,9	99,3	100	100	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 Infantile liver failure syndrome 2, 616483
NBEAL2	99,5	99,3	100	100	Gray platelet syndrome, 139090
NBN	99,2	97,8	100	99,9	Leukemia, acute lymphoblastic, 613065 Aplastic anemia, 609135 Nijmegen breakage syndrome, 251260
NCAPD2	99,9	99	100	100	?Microcephaly 21, primary, autosomal recessive, 617983
NCAPD3	99,7	98	100	100	Microcephaly 22, primary, autosomal recessive, 617984
NCAPG2	99,8	99	100	100	Khan-Khan-Katsanis syndrome, 618460
NCAPH	100	100	100	100	?Microcephaly 23, primary, autosomal recessive, 617985
NCF1	26	25,7	100	99,9	Chronic granulomatous disease 1, autosomal recessive, 233700
NCF2	99,8	97	100	100	Chronic granulomatous disease 2, autosomal recessive, 233710
NCF4	100	100	100	100	Chronic granulomatous disease 3, autosomal recessive, 613960
NDE1	100	99,4	100	100	Lissencephaly 4 (with microcephaly), 614019 ?Microhydranencephaly, 605013
NDRG1	100	99,9	100	100	Charcot-Marie-Tooth disease, type 4D, 601455
NDST1	100	100	100	100	Mental retardation, autosomal recessive 46, 616116
NDUFA10	99,9	98,6	100	100	Mitochondrial complex I deficiency, nuclear type 22, 618243
NDUFA11	100	99,8	100	99,9	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFA12	99,6	99,6	100	100	Mitochondrial complex I deficiency, nuclear type 23, 618244
NDUFA13	92,2	90	100	100	?Mitochondrial complex I deficiency, nuclear type 28, 618249
NDUFA2	100	100	100	100	Mitochondrial complex I deficiency, nuclear type 13, 618235
NDUFA6	100	100	100	100	Mitochondrial complex I deficiency, nuclear type 33, 618253
NDUFA9	99,3	95,2	100	100	Mitochondrial complex I deficiency, nuclear type 26, 618247
NDUFAF1	100	100	100	100	Mitochondrial complex I deficiency, nuclear type 11, 618234

NDUFAF2	91	77,5	100	99,6	Mitochondrial complex I deficiency, nuclear type 10, 618233
NDUFAF3	100	99,9	100	100	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF4	99,6	96,9	100	99,8	Mitochondrial complex I deficiency, nuclear type 15, 618237
NDUFAF5	99,7	99,1	100	100	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFAF6	99,3	96,9	100	99,9	Mitochondrial complex I deficiency, nuclear type 17, 618239 Fanconi renotubular syndrome 5, 618913
NDUFB11	99,1	94,8	99,9	99,1	Linear skin defects with multiple congenital anomalies 3, 300952 ?Mitochondrial complex I deficiency, nuclear type 30, 301021
NDUFB3	88,6	71	100	100	Mitochondrial complex I deficiency, nuclear type 25, 618246
NDUFB8	100	99,5	100	100	Mitochondrial complex I deficiency, nuclear type 32, 618252
NDUFB9	96,1	91,5	98,7	98,7	?Mitochondrial complex I deficiency, nuclear type 24, 618245
NDUFS1	99,9	99,1	100	99,9	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	100	100	100	100	Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	90,7	90,6	92,8	90,7	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	99,7	99,7	100	100	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS6	100	99,8	100	100	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFS7	100	99,7	100	100	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFS8	100	99,1	100	100	Mitochondrial complex I deficiency, nuclear type 2, 618222
NDUFV1	99	97	100	100	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	85,8	78,7	100	100	Mitochondrial complex I deficiency, nuclear type 7, 618229
NEB	82,9	82,5	99,9	99,8	Nemaline myopathy 2, autosomal recessive, 256030 Arthrogryposis multiplex congenita 6, 619334
NECAP1	100	100	100	100	Developmental and epileptic encephalopathy 21, 615833
NECTIN1	100	99,7	100	100	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060
NECTIN4	100	99,9	100	100	Ectodermal dysplasia-syndactyly syndrome 1, 613573
NEK1	99,5	98,2	100	99,9	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NEK2	99,3	93,4	96,1	96,1	?Retinitis pigmentosa 67, 615565
NEK8	100	99,8	100	100	Renal-hepatic-pancreatic dysplasia 2, 615415 ?Nephronophthisis 9, 613824

NEK9	99,9	99	100	100	?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262 Nevus comedonicus, somatic, 617025 Lethal congenital contracture syndrome 10, 617022
NEPRO	99,8	99,5	100	100	Anauxetic dysplasia 3, 618853
NEU1	99,3	96,1	100	100	Sialidosis, type II, 256550 Sialidosis, type I, 256550
NEUROG3	100	100	100	100	Diarrhea 4, malabsorptive, congenital, 610370
NFASC	100	99,5	100	100	Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356
NFU1	98,7	87,7	100	100	Multiple mitochondrial dysfunctions syndrome 1, 605711
NGF	100	100	100	100	Neuropathy, hereditary sensory and autonomic, type V, 608654
NGLY1	99,8	99,7	100	100	Congenital disorder of deglycosylation, 615273
NHEJ1	99,8	97,2	100	100	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
NHLRC1	100	99,8	100	100	Epilepsy, progressive myoclonic 2B (Lafora), 254780
NHLRC2	99,7	97,8	100	99,9	FINCA syndrome, 618278
NHP2	100	100	100	100	Dyskeratosis congenita, autosomal recessive 2, 613987
NIN	99,9	99,5	99,1	99,1	?Seckel syndrome 7, 614851
NIPAL4	100	98,9	100	100	Ichthyosis, congenital, autosomal recessive 6, 612281
NKX2-6	100	100	100	100	Persistent truncus arteriosus, 217095 Conotruncal heart malformations, 217095
NKX3-2	100	99,3	100	100	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330
NKX6-2	88,2	81,9	100	100	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560
NLRP1	99,3	97,2	100	100	?Respiratory papillomatosis, juvenile recurrent, congenital, 618803 Autoinflammation with arthritis and dyskeratosis, 617388 Palmoplantar carcinoma, multiple self-healing, 615225
NLRP7	99,9	99	100	100	Hydatidiform mole, recurrent, 1, 231090
NME8	98,9	94,2	100	100	Ciliary dyskinesia, primary, 6, 610852
NMNAT1	100	99,2	99,4	96,7	Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis, 619260 Leber congenital amaurosis 9, 608553
NNT	96,4	96	96,4	96,4	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736
NOP10	100	99,2	100	100	Dyskeratosis congenita, autosomal recessive 1, 224230

NPC1	99,9	99	100	100	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220
NPC2	100	99,2	100	100	Niemann-pick disease, type C2, 607625
NPHP1	99,8	99,1	100	100	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NPHP3	99,6	98,5	100	99,9	Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540 Meckel syndrome 7, 267010
NPHP4	100	99,8	100	100	Senior-Loken syndrome 4, 606996 Nephronophthisis 4, 606966
NPHS1	99,7	99	100	100	Nephrotic syndrome, type 1, 256300
NPHS2	100	99,6	100	99,9	Nephrotic syndrome, type 2, 600995
NPPA	100	100	100	100	Atrial standstill 2, 615745 Atrial fibrillation, familial, 6, 612201
NPR2	100	99,2	100	100	Acromesomelic dysplasia, Maroteaux type, 602875 Epiphyseal chondrodysplasia, Miura type, 615923 Short stature with nonspecific skeletal abnormalities, 616255
NR0B2	100	99,6	100	100	Obesity, mild, early-onset, 601665
NR1H4	99,6	98,6	100	100	Cholestasis, progressive familial intrahepatic, 5, 617049
NR2E3	100	99,6	100	100	Retinitis pigmentosa 37, 611131 Enhanced S-cone syndrome, 268100
NRROS	100	100	100	100	Seizures, early-onset, with neurodegeneration and brain calcification, 618875
NRXN1	97,5	96,9	99,9	99,7	Pitt-Hopkins-like syndrome 2, 614325
NSMCE2	99,5	98,7	100	100	Seckel syndrome 10, 617253
NSMCE3	100	100	100	100	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241
NSUN2	95,5	92,9	100	100	Mental retardation, autosomal recessive 5, 611091
NT5C2	97,7	94,6	100	100	Spastic paraplegia 45, autosomal recessive, 613162
NT5C3A	94,6	82,2	100	100	Anemia, hemolytic, due to UMPH1 deficiency, 266120
NT5E	100	100	100	100	Calcification of joints and arteries, 211800
NTHL1	100	99,9	100	100	Familial adenomatous polyposis 3, 616415
NTNG2	99,1	97,3	100	99,6	Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718
NTRK1	99,9	98,5	100	100	Insensitivity to pain, congenital, with anhidrosis, 256800

NUBPL	99,5	96,9	100	100	Mitochondrial complex I deficiency, nuclear type 21, 618242
NUP107	99,7	98,4	100	99,9	?Ovarian dysgenesis 6, 618078 Galloway-Mowat syndrome 7, 618348 Nephrotic syndrome, type 11, 616730
NUP133	99,4	97,3	100	100	?Galloway-Mowat syndrome 8, 618349 Nephrotic syndrome, type 18, 618177
NUP160	100	99,8	100	100	?Nephrotic syndrome, type 19, 618178
NUP188	99,9	99,1	100	100	Sandestig-Stefanova syndrome, 618804
NUP205	99,9	99,3	100	99,9	?Nephrotic syndrome, type 13, 616893
NUP214	99,8	99	100	100	Leukemia, T-cell acute lymphoblastic, somatic, 613065 Leukemia, acute myeloid, somatic, 601626
NUP37	100	100	100	100	?Microcephaly 24, primary, autosomal recessive, 618179
NUP62	100	100	100	100	Striatonigral degeneration, infantile, 271930
NUP85	100	100	100	100	Nephrotic syndrome, type 17, 618176
NUP88	99,8	99,8	100	100	Fetal akinesia deformation sequence 4, 618393
NUP93	96,7	92,7	95,5	95,5	Nephrotic syndrome, type 12, 616892
NUS1	56,5	42	100	99,9	Mental retardation, autosomal dominant 55, with seizures, 617831 ?Congenital disorder of glycosylation, type 1aa, 617082
NXN	100	100	100	99,7	Robinow syndrome, autosomal recessive 2, 618529
OAT	82	73	100	100	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OBSL1	100	99,8	100	100	3-M syndrome 2, 612921
OCA2	99,9	98,3	100	100	Albinism, brown oculocutaneous, 203200 Albinism, oculocutaneous, type II, 203200
OCLN	100	99,9	100	100	Pseudo-TORCH syndrome 1, 251290
CCDC114	100	99,8	100	100	Ciliary dyskinesia, primary, 20, 615067
ARMC4	92,4	89,9	96,3	96,2	Ciliary dyskinesia, primary, 23, 615451
CCDC151	100	99,7	100	100	Ciliary dyskinesia, primary, 30, 616037
TTC25	100	99,7	100	100	Ciliary dyskinesia, primary, 35, 617092
ODAPH	100	100	100	100	Amelogenesis imperfecta, type IIA4, 614832
OGDH	100	99,8	100	100	No OMIM disease ID
OPA1	99,5	96,7	100	99,9	Optic atrophy plus syndrome, 125250 Optic atrophy 1, 165500

					Behr syndrome, 210000 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896
OPA3	100	99,5	100	100	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPLAH	100	99,8	100	100	5-oxoprolinase deficiency, 260005
ORAI1	99,3	97,1	99,4	96,7	Immunodeficiency 9, 612782 Myopathy, tubular aggregate, 2, 615883
ORC1	99,9	97,9	100	100	Meier-Gorlin syndrome 1, 224690
ORC4	96,8	90,6	100	100	Meier-Gorlin syndrome 2, 613800
ORC6	100	99,8	100	100	Meier-Gorlin syndrome 3, 613803
OSGEP	99,8	95,7	100	100	Galloway-Mowat syndrome 3, 617729
OSTM1	98,7	92,9	100	100	Osteopetrosis, autosomal recessive 5, 259720
OTOA	99,7	98,2	100	100	Deafness, autosomal recessive 22, 607039
OTOF	100	99,8	100	100	Auditory neuropathy, autosomal recessive, 1, 601071 Deafness, autosomal recessive 9, 601071
OTOG	99,3	98,5	100	100	Deafness, autosomal recessive 18B, 614945
OTOGL	99,3	97,1	100	99,9	Deafness, autosomal recessive 84B, 614944
OTUD6B	99,7	98,6	100	99,8	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452
OTULIN	92,9	87	98,8	94,1	Autoinflammation, panniculitis, and dermatosis syndrome, 617099
OXCT1	99,4	97,6	100	100	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050
P2RY12	100	100	100	100	Bleeding disorder, platelet-type, 8, 609821
P3H1	100	100	100	100	Osteogenesis imperfecta, type VIII, 610915
P3H2	99,9	98	100	100	Myopia, high, with cataract and vitreoretinal degeneration, 614292
P4HTM	99,3	97,6	100	99,6	Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493
PAH	100	100	100	100	Phenylketonuria, 261600
PAM16	65,3	65,2	82,9	82,9	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320
PANK2	100	99,7	100	100	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PAPSS2	99,8	98	100	100	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847
PARK7	100	99,8	100	100	Parkinson disease 7, autosomal recessive early-onset, 606324

PARN	81,1	80,4	88,3	87,6	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
PARS2	100	100	100	100	Developmental and epileptic encephalopathy 75, 618437
PATL2	99,9	95	100	100	Oocyte maturation defect 4, 617743
PAX1	92,6	87,5	100	99,7	Otofaciocervical syndrome 2, 615560
PAX3	100	99,8	100	100	Craniofacial-deafness-hand syndrome, 122880 Waardenburg syndrome, type 3, 148820 Waardenburg syndrome, type 1, 193500 Rhabdomyosarcoma 2, alveolar, 268220
PAX7	100	100	100	100	Rhabdomyosarcoma 2, alveolar, 268220 Myopathy, congenital, progressive, with scoliosis, 618578
PC	99,7	98	100	100	Pyruvate carboxylase deficiency, 266150
PCARE	99,6	98,1	100	100	Retinitis pigmentosa 54, 613428
PCBD1	100	99,8	100	100	Hyperphenylalaninemia, BH4-deficient, D, 264070
PCCA	98,9	93,4	100	100	Propionicacidemia, 606054
PCCB	96,7	95,4	99	96,2	Propionicacidemia, 606054
PCDH12	100	100	100	100	Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280
PCDH15	97,9	96,8	100	100	Usher syndrome, type 1D/F digenic, 601067 Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1F, 602083
PCK1	100	100	100	100	?Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680
PCK2	100	100	100	100	No OMIM disease ID
PCLO	99,4	98,3	100	100	?Pontocerebellar hypoplasia, type 3, 608027
PCNA	99,8	98,3	100	100	?Ataxia-telangiectasia-like disorder 2, 615919
PCNT	99,3	96,5	100	100	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PCSK1	99,9	99,4	100	100	Obesity with impaired prohormone processing, 600955
PCYT1A	99,2	95,7	100	100	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDE10A	65,7	64,3	86,5	83,2	Striatal degeneration, autosomal dominant, 616922 Dyskinesia, limb and orofacial, infantile-onset, 616921
PDE6A	100	99,5	100	100	Retinitis pigmentosa 43, 613810
PDE6B	100	99,8	100	100	Retinitis pigmentosa-40, 613801 Night blindness, congenital stationary, autosomal dominant 2, 163500

PDE6C	99,7	97	100	100	Cone dystrophy 4, 613093
PDE6D	100	99,9	100	100	Joubert syndrome 22, 615665
PDE6G	100	100	100	100	Retinitis pigmentosa 57, 613582
PDE6H	99,8	86,1	100	100	Retinal cone dystrophy 3, 610024 Achromatopsia 6, 610024
PDHB	99,2	96,8	100	100	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	99,8	99,6	100	100	Lacticacidemia due to PDX1 deficiency, 245349
PDP1	100	100	100	100	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	95,2	87,8	97,4	97,4	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	98,4	94,3	100	100	Coenzyme Q10 deficiency, primary, 3, 614652
PDX1	94,8	82,2	100	99,9	Pancreatic agenesis 1, 260370 MODY, type IV, 606392
PDXK	79,4	77,1	99,9	99,1	Neuropathy, hereditary motor and sensory, type VIC, with optic atrophy, 618511
PDZD7	96	91,5	100	99,7	Deafness, autosomal recessive 57, 618003 Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472
PEPD	100	99,4	100	100	Prolidase deficiency, 170100
PET100	100	99,2	100	100	Mitochondrial complex IV deficiency, nuclear type 12, 619055
PEX1	99,8	99,4	100	100	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	98,8	90,6	100	100	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX11B	100	98,3	100	100	Peroxisome biogenesis disorder 14B, 614920
PEX12	100	100	100	100	Peroxisome biogenesis disorder 3B, 266510 Peroxisome biogenesis disorder 3A (Zellweger), 614859
PEX13	100	100	100	100	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	95,8	89,4	100	100	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	97,1	93,9	100	100	Peroxisome biogenesis disorder 8B, 614877 Peroxisome biogenesis disorder 8A (Zellweger), 614876
PEX19	99	94,4	100	100	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	100	100	100	100	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867

PEX26	100	99,8	100	100	Peroxisome biogenesis disorder 7B, 614873 Peroxisome biogenesis disorder 7A (Zellweger), 614872
PEX3	99,4	99,2	100	100	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370
PEX5	99,9	98,8	100	100	Peroxisome biogenesis disorder 2B, 202370 Peroxisome biogenesis disorder 2A (Zellweger), 214110 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	96,4	88	100	100	Peroxisome biogenesis disorder 4B, 614863 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Heimler syndrome 2, 616617
PEX7	88	81	91,3	91,2	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PFKM	100	99,7	100	100	Glycogen storage disease VII, 232800
PGAM2	100	100	100	100	Glycogen storage disease X, 261670
PGAP1	98,7	94,6	100	99,8	Neurodevelopmental disorder with dysmorphic features, spasticity, and brain abnormalities, 615802
PGAP2	100	99,9	100	100	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGAP3	62,6	58,1	100	100	Hyperphosphatasia with mental retardation syndrome 4, 615716
PGM1	94,2	94,1	94,2	94,2	Congenital disorder of glycosylation, type It, 614921
PGM3	99,9	99,7	91,7	91,7	Immunodeficiency 23, 615816
PHGDH	99,9	98,2	100	100	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHKB	99,7	99,1	100	100	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750
PHKG2	100	99,9	100	100	Glycogen storage disease IXc, 613027
PHOX2A	92,8	74,4	100	100	Fibrosis of extraocular muscles, congenital, 2, 602078
PHYH	100	98,9	100	100	Refsum disease, 266500
PI4KA	92,6	88,7	100	99,9	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531
PIBF1	99,1	95	100	99,9	Joubert syndrome 33, 617767
PIEZO1	99,9	98,9	100	100	Lymphatic malformation 6, 616843 Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380
PIEZO2	99,8	99,2	100	100	Arthrogryposis, distal, type 5, 108145 Arthrogryposis, distal, with impaired proprioception and touch, 617146 Arthrogryposis, distal, type 3, 114300 ?Marden-Walker syndrome, 248700

PIGB	99,5	97,3	100	100	Developmental and epileptic encephalopathy 80, 618580
PIGC	96	86,2	100	100	Glycosylphosphatidylinositol biosynthesis defect 16, 617816
PIGG	100	99,6	100	100	Mental retardation, autosomal recessive 53, 616917
PIGH	81,9	64,4	75,9	74,4	Glycosylphosphatidylinositol biosynthesis defect 17, 618010
PIGK	98,8	94,2	100	100	Neurodevelopmental disorder with hypotonia and cerebellar atrophy, with or without seizures, 618879
PIGL	100	99,6	100	100	CHIME syndrome, 280000
PIGM	100	100	100	100	Glycosylphosphatidylinositol deficiency, 610293
PIGN	93,1	89,6	98,8	98,6	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	100	99,8	100	100	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGP	95,6	85,5	100	99,9	Developmental and epileptic encephalopathy 55, 617599
PIGQ	93,4	91,6	100	100	Developmental and epileptic encephalopathy 77, 618548
PIGS	100	99,6	100	100	Developmental and epileptic encephalopathy 95, 618143
PIGT	98,1	98	100	100	?Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGU	100	99,5	100	98,9	Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis, 618590
PIGV	100	100	100	100	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIGW	100	99,7	100	100	Glycosylphosphatidylinositol biosynthesis defect 11, 616025
PIGY	100	100	100	100	Hyperphosphatasia with mental retardation syndrome 6, 616809
PIK3C2A	99	95,9	100	100	Oculoskeletodental syndrome, 618440
PIK3R1	99,7	98,4	100	100	Immunodeficiency 36, 616005 ?Agammaglobulinemia 7, autosomal recessive, 615214 SHORT syndrome, 269880
PIK3R5	100	99,9	100	100	Ataxia-oculomotor apraxia 3, 615217
PINK1	91	85,8	100	99,5	Parkinson disease 6, early onset, 605909
PIP5K1C	99,2	96,7	99,9	99,2	Lethal congenital contractural syndrome 3, 611369
PJVK	100	99,7	100	99,9	Deafness, autosomal recessive 59, 610220
PKD1L1	100	99,3	100	100	Heterotaxy, visceral, 8, autosomal, 617205
PKHD1	100	99,6	100	100	Polycystic kidney disease 4, with or without hepatic disease, 263200
PKLR	99,9	98	100	100	Adenosine triphosphate, elevated, of erythrocytes, 102900 Pyruvate kinase deficiency, 266200

PKP1	99,9	98,6	100	100	Ectodermal dysplasia/skin fragility syndrome, 604536
PLA2G6	92,1	90,7	92,3	92,3	Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217 Infantile neuroaxonal dystrophy 1, 256600
PLA2G7	99,8	99,3	100	100	Platelet-activating factor acetylhydrolase deficiency, 614278
PLAA	99,6	98,4	100	100	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527
PLCB1	99,9	99,4	100	100	Developmental and epileptic encephalopathy 12, 613722
PLCB4	99,8	98,7	100	100	Auriculocondylar syndrome 2, 614669
PLCD1	99,9	97,3	100	100	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCE1	99,8	98,9	100	100	Nephrotic syndrome, type 3, 610725
PLD1	99,8	98,7	100	100	Cardiac valvular defect, developmental, 212093
PLEC	100	99,9	100	100	?Epidermolysis bullosa simplex with nail dystrophy, 616487 Epidermolysis bullosa simplex, Ogna type, 131950 Epidermolysis bullosa simplex with pyloric atresia, 612138 Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723 Epidermolysis bullosa simplex with muscular dystrophy, 226670
PLEKHG2	99,8	98,1	100	100	Leukodystrophy and acquired microcephaly with or without dystonia, 616763
PLEKHG5	96,1	93,4	96,3	96,2	Spinal muscular atrophy, distal, autosomal recessive, 4, 611067 Charcot-Marie-Tooth disease, recessive intermediate C, 615376
PLEKHM1	100	99,9	100	100	?Osteopetrosis, autosomal recessive 6, 611497 Osteopetrosis, autosomal dominant 3, 618107
PLG	87,8	87,6	100	100	Dysplasminogenemia, 217090 Angioedema, hereditary, 4, 619360 Plasminogen deficiency, type I, 217090
PLK4	99,4	98,4	100	100	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLOD1	100	98,2	100	100	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD2	99,2	98,1	100	100	Bruck syndrome 2, 609220
PLOD3	100	98,7	100	100	Lysyl hydroxylase 3 deficiency, 612394
PLPBP	95,1	88,9	100	99,9	Epilepsy, early-onset, vitamin B6-dependent, 617290
PLVAP	100	100	100	100	Diarrhea 10, protein-losing enteropathy type, 618183
PMM2	99,8	99,8	100	100	Congenital disorder of glycosylation, type Ia, 212065
PMPCA	97,6	93,5	100	100	Spinocerebellar ataxia, autosomal recessive 2, 213200

PMPCB	99,9	99,2	100	100	Multiple mitochondrial dysfunctions syndrome 6, 617954
PMS2	83,9	81,6	100	100	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 Mismatch repair cancer syndrome 4, 619101
PNKP	100	100	100	100	?Charcot-Marie-Tooth disease, type 2B2, 605589 Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNLIP	99,9	99,6	100	100	?Pancreatic lipase deficiency, 614338
PNP	99,8	98,7	100	100	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA1	100	100	100	100	Ichthyosis, congenital, autosomal recessive 10, 615024
PNPLA2	99,8	96,1	100	100	Neutral lipid storage disease with myopathy, 610717
PNPLA6	100	99,8	100	100	Spastic paraplegia 39, autosomal recessive, 612020 Oliver-McFarlane syndrome, 275400 ?Laurence-Moon syndrome, 245800 Boucher-Neuhauser syndrome, 215470
PNPLA8	99,7	99,5	100	99,9	?Mitochondrial myopathy with lactic acidosis, 251950
PNPO	99,9	97,1	100	100	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
PNPT1	96,9	86,1	100	99,9	Deafness, autosomal recessive 70, 614934 Combined oxidative phosphorylation deficiency 13, 614932
POC1A	100	100	100	100	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POC1B	99,5	97,3	100	100	Cone-rod dystrophy 20, 615973
POGLUT1	99,8	95,9	100	100	Dowling-Degos disease 4, 615696 ?Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232
POLE	100	99,5	100	100	FILS syndrome, 615139 IMAGE-I syndrome, 618336
POLG	99,9	98,8	100	100	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
POLH	100	99,1	100	100	Xeroderma pigmentosum, variant type, 278750
POLR1C	89,6	84,8	82,8	82,8	Leukodystrophy, hypomyelinating, 11, 616494 Treacher Collins syndrome 3, 248390
POLR1D	91,6	91,6	100	99,8	Treacher Collins syndrome 2, 613717

POLR3A	99,9	99	100	100	Wiedemann-Rautenstrauch syndrome, 264090 Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	99,7	97,6	100	100	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMC	100	100	100	100	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734
POMGNT1	100	99,8	100	100	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	100	100	100	100	Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830
POMK	100	100	100	100	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249
POMP	99,7	98,8	100	100	Proteasome-associated autoinflammatory syndrome 2, 618048 Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952
POMT1	99,5	97,3	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155
POMT2	99,8	97,3	100	100	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150
POP1	100	99,3	100	100	Anauxetic dysplasia 2, 617396
POR	99,5	98	100	100	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571
POU1F1	99,9	98,2	100	100	Pituitary hormone deficiency, combined, 1, 613038
PPA2	97,3	88,6	100	100	?Sudden cardiac failure, alcohol-induced, 617223 Sudden cardiac failure, infantile, 617222
PPCS	100	99,1	100	100	Cardiomyopathy, dilated, 2C, 618189
PPIB	100	99,9	100	100	Osteogenesis imperfecta, type IX, 259440
PPIL1	100	100	100	100	Pontocerebellar hypoplasia, type 14, 619301
PPIP5K2	98,2	94,8	100	99,9	Deafness, autosomal recessive 100, 618422
PPM1K	100	100	100	100	?Maple syrup urine disease, mild variant, 615135
PPP1R15B	100	99,6	100	100	Microcephaly, short stature, and impaired glucose metabolism 2, 616817
PPP1R21	99,3	95,5	100	100	Neurodevelopmental disorder with hypotonia, facial dysmorphism, and brain abnormalities, 619383

PPP2R3C	98,3	89,4	100	99,8	Gonadal dysgenesis, dysmorphic facies, retinal dystrophy, and myopathy, 618419 Spermatogenic failure 36, 618420
PPT1	90,3	89,9	82,5	82,5	Ceroid lipofuscinosis, neuronal, 1, 256730
PRCD	100	100	100	100	Retinitis pigmentosa 36, 610599
PRDM12	91,7	89,6	92,8	91	Neuropathy, hereditary sensory and autonomic, type VIII, 616488
PRDM5	99,8	98,4	100	100	Brittle cornea syndrome 2, 614170
PRDM8	93,5	89,2	100	100	?Epilepsy, progressive myoclonic, 10, 616640
PRDX1	100	99,9	100	100	Methylmalonic aciduria and homocystinuria, cblC type, digenic, 277400
PREPL	98,9	96,6	100	100	Myasthenic syndrome, congenital, 22, 616224
PRF1	91,2	90,1	100	100	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Aplastic anemia, 609135 Lymphoma, non-Hodgkin, 605027
PRG4	92,7	83,2	100	100	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250
PRICKLE1	100	99,9	100	100	Epilepsy, progressive myoclonic 1B, 612437
PRKCD	100	99,9	100	100	Autoimmune lymphoproliferative syndrome, type III, 615559
PRKDC	99,2	96,9	100	100	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PRKN	66,9	65,8	75,4	75,3	Adenocarcinoma of lung, somatic, 211980 Parkinson disease, juvenile, type 2, 600116 Ovarian cancer, somatic, 167000
PRKRA	99,8	99,5	100	100	Dystonia 16, 612067
PRMT7	100	99,9	100	100	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157
PROC	100	100	100	100	Thrombophilia due to protein C deficiency, autosomal recessive, 612304 Thrombophilia due to protein C deficiency, autosomal dominant, 176860
PRODH	84	80,2	100	100	Hyperprolinemia, type I, 239500
PROM1	97,2	96,5	100	99,9	Macular dystrophy, retinal, 2, 608051 Retinitis pigmentosa 41, 612095 Stargardt disease 4, 603786 Cone-rod dystrophy 12, 612657
PROP1	91	80,2	100	100	Pituitary hormone deficiency, combined, 2, 262600
PROS1	96,4	89,4	98,4	98,4	Thrombophilia due to protein S deficiency, autosomal dominant, 612336 Thrombophilia due to protein S deficiency, autosomal recessive, 614514
PRSS12	100	99,9	100	100	Mental retardation, autosomal recessive 1, 249500
PRSS56	100	98	100	100	Microphthalmia, isolated 6, 613517

PRUNE1	93,6	93,1	93,6	93,6	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481
PRX	96,1	95,8	97,5	96,4	Charcot-Marie-Tooth disease, type 4F, 614895 Dejerine-Sottas disease, 145900
PSAP	100	99,6	100	100	Combined SAP deficiency, 611721 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900 Gaucher disease, atypical, 610539
PSAT1	92	75,1	100	100	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992
PSMB4	100	100	100	100	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591
PSMB8	99,8	97,5	100	100	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040
PSMB9	99,5	95,4	100	100	?Proteasome-associated autoinflammatory syndrome 3, digenic, 617591
PSMC3IP	100	100	100	100	Ovarian dysgenesis 3, 614324
PSPH	100	100	100	100	Phosphoserine phosphatase deficiency, 614023
PTF1A	98,8	91,1	98,7	92,9	Pancreatic and cerebellar agenesis, 609069 Pancreatic agenesis 2, 615935
PTH1R	99,6	95,9	100	100	Metaphyseal chondrodysplasia, Murk Jansen type, 156400 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Chondrodysplasia, Blomstrand type, 215045
PTPN14	99,4	96,9	100	100	Choanal atresia and lymphedema, 613611
PTPN23	100	100	100	100	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity, 618890
PTPRC	98,8	93,9	100	99,9	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971
PTPRO	99,7	98,9	100	100	Nephrotic syndrome, type 6, 614196
PTPRQ	94,4	92,5	92,8	92,2	Deafness, autosomal dominant 73, 617663 Deafness, autosomal recessive 84A, 613391
PTRH2	100	100	100	100	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PTS	99,5	99	100	99,9	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUS1	99,9	98	99,9	98,2	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PUS3	100	100	100	100	Neurodevelopmental disorder with microcephaly and gray sclerae, 617051
PUS7	99,8	99,7	100	100	Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342
PXDN	99,9	99,6	100	100	Anterior segment dysgenesis 7, with sclerocornea, 269400

PYCR1	100	98,2	100	100	Cutis laxa, autosomal recessive, type IIIB, 614438 Cutis laxa, autosomal recessive, type IIB, 612940
PYCR2	100	99,3	100	100	Leukodystrophy, hypomyelinating, 10, 616420
PYGL	100	100	100	100	Glycogen storage disease VI, 232700
PYGM	100	100	100	100	McArdle disease, 232600
PYROXD1	92,1	78,7	100	100	Myopathy, myofibrillar, 8, 617258
QARS1	100	100	100	100	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	100	98,9	100	100	Hyperphenylalaninemia, BH4-deficient, C, 261630
RAB18	98,9	94,5	99,9	99,8	Warburg micro syndrome 3, 614222
RAB23	99,7	99,7	100	100	Carpenter syndrome, 201000
RAB27A	99,5	99,5	100	99,9	Griscelli syndrome, type 2, 607624
RAB28	99,2	97,2	100	99,9	Cone-rod dystrophy 18, 615374
RAB33B	85	85	100	100	Smith-McCort dysplasia 2, 615222
RAB3GAP1	99,2	98,7	99,4	99,3	Martsolf syndrome 2, 619420 Warburg micro syndrome 1, 600118
RAB3GAP2	99,1	96,3	100	99,9	Martsolf syndrome 1, 212720 Warburg micro syndrome 2, 614225
RAD50	96,6	89,7	100	100	Nijmegen breakage syndrome-like disorder, 613078
RAD51C	99,8	99,4	100	100	Fanconi anemia, complementation group O, 613390
RAG1	100	100	100	100	Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457 Combined cellular and humoral immune defects with granulomas, 233650 Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889
RAG2	100	100	100	100	Severe combined immunodeficiency, B cell-negative, 601457 Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554
RALGAPA1	73,6	61,2	100	99,9	Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation, 618797
RAPSN	100	99,6	100	100	Fetal akinesia deformation sequence 2, 618388 Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326
RARB	100	99,8	100	100	Microphthalmia, syndromic 12, 615524
RARS1	94,1	91,8	94,4	94,2	Leukodystrophy, hypomyelinating, 9, 616140

RARS2	99,7	98,6	100	100	Pontocerebellar hypoplasia, type 6, 611523
RASGRP1	100	99,6	100	100	Immunodeficiency 64, 618534
RASGRP2	100	98,3	100	100	?Bleeding disorder, platelet-type, 18, 615888
RAX	98,1	89,3	99,9	97,7	Microphthalmia, isolated 3, 611038
RBBP8	99,7	99,4	100	99,9	Seckel syndrome 2, 606744 Jawad syndrome, 251255 Pancreatic carcinoma, somatic,
RBCK1	99,9	98,3	100	100	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RBM28	100	100	100	100	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RBM8A	99,6	95,3	100	100	Thrombocytopenia-absent radius syndrome, 274000
RBP3	100	100	100	100	?Retinitis pigmentosa 66, 615233
RBP4	99,6	96,3	100	100	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RCBTB1	99,7	98,6	100	100	Retinal dystrophy with or without extraocular anomalies, 617175
RD3	100	100	100	100	Leber congenital amaurosis 12, 610612
RDH11	99,6	96,9	100	100	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108
RDH12	99,3	95,4	100	100	Leber congenital amaurosis 13, 612712
RDH5	100	100	100	100	Fundus albipunctatus, 136880
RDX	87,2	69,3	100	99,9	Deafness, autosomal recessive 24, 611022
RECQL4	99,9	98,6	100	100	Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2, 268400 RAPADILINO syndrome, 266280
REEP2	99,8	98	100	100	?Spastic paraplegia 72, autosomal dominant, 615625 ?Spastic paraplegia 72, autosomal recessive, 615625
REEP6	100	99,9	97,9	90,8	Retinitis pigmentosa 77, 617304
RELB	99	91,5	100	100	?Immunodeficiency 53, 617585
RELN	100	99,6	100	100	Lissencephaly 2 (Norman-Roberts type), 257320
REN	100	100	100	100	Renal tubular dysgenesis, 267430 Tubulointerstitial kidney disease, autosomal dominant, 4, 613092
REPS1	98,5	96,1	100	99,9	?Neurodegeneration with brain iron accumulation 7, 617916
RETREG1	99,1	96,1	100	100	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
RFC1	99,5	97,4	100	99,9	Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome, 614575

RFT1	99,7	98,4	100	100	Congenital disorder of glycosylation, type In, 612015
RFWD3	100	99,4	100	100	?Fanconi anemia, complementation group W, 617784
RFX5	99,7	98,1	100	100	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFX6	99,9	99,5	100	100	Mitchell-Riley syndrome, 615710
RFXANK	100	99,7	100	100	MHC class II deficiency, complementation group B, 209920
RFXAP	99,9	98,6	100	100	Bare lymphocyte syndrome, type II, complementation group D, 209920
RGR	99	97,6	99	99	Retinitis pigmentosa 44, 613769
RHO	100	100	100	100	Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 Retinitis punctata albescens, 136880
RIMS2	96,6	94,5	97,8	97,7	Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970
RIN2	100	99,7	100	100	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075
RINT1	99,6	97,6	100	99,9	Infantile liver failure syndrome 3, 618641
RIPK1	99,8	98,5	100	100	Immunodeficiency 57 with autoinflammation, 618108 Autoinflammation with episodic fever and lymphadenopathy, 618852
RIPK4	100	99,9	100	100	CHAND syndrome, 214350 Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650
RIPOR2	99,9	99,3	100	100	?Deafness, autosomal recessive 104, 616515
RIPPLY2	99	94,7	100	99,9	?Spondylocostal dysostosis 6, 616566
RLBP1	100	99,8	100	100	Bothnia retinal dystrophy, 607475 Newfoundland rod-cone dystrophy, 607476 Retinitis punctata albescens, 136880 Fundus albipunctatus, 136880
RMND1	99,7	97,2	100	99,9	Combined oxidative phosphorylation deficiency 11, 614922
RMRP	NC	NC	NC	NC	Anauxetic dysplasia 1, 607095 Metaphyseal dysplasia without hypotrichosis, 250460 Cartilage-hair hypoplasia, 250250
RNASEH1	98,7	95,8	100	100	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479
RNASEH2A	100	99,7	100	100	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	81	78,2	91	90,9	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	100	100	100	100	Aicardi-Goutieres syndrome 3, 610329
RNASET2	95,7	91	100	100	Leukoencephalopathy, cystic, without megalencephaly, 612951

RNF168	99,9	99,6	100	100	RIDDLE syndrome, 611943
RNF216	99,6	98,2	100	100	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840
RNPC3	94	75,1	100	100	?Growth hormone deficiency, isolated, type V, 618160
ROBO3	99	95,9	100	100	Gaze palsy, familial horizontal, with progressive scoliosis, 1, 607313
ROGDI	98,6	95,2	99,9	98,1	Kohlschutter-Tonz syndrome, 226750
ROR1	97,2	96,8	100	99,4	?Deafness, autosomal recessive 108, 617654
ROR2	100	99,4	97	97	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RORC	100	100	100	100	Immunodeficiency 42, 616622
RP1	91,2	90,7	100	100	Retinitis pigmentosa 1, 180100
RPE65	99,9	98,7	100	100	Retinitis pigmentosa 20, 613794 Retinitis pigmentosa 87 with choroidal involvement, 618697 Leber congenital amaurosis 2, 204100
RPGRIP1	100	99,7	100	100	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826
RPGRIP1L	96,5	95,3	100	99,4	Joubert syndrome 7, 611560 Meckel syndrome 5, 611561 ?COACH syndrome 3, 619113
RPIA	99,1	96,1	100	100	Ribose 5-phosphate isomerase deficiency, 608611
RRM2B	100	99,8	100	99,9	Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077
RSPH1	99,9	99,9	100	100	Ciliary dyskinesia, primary, 24, 615481
RSPH3	99,6	98,4	100	99,9	Ciliary dyskinesia, primary, 32, 616481
RSPH4A	98,2	95,4	100	100	Ciliary dyskinesia, primary, 11, 612649
RSPH9	99,7	96,3	100	100	Ciliary dyskinesia, primary, 12, 612650
RSPO1	100	99,9	100	100	Palmoplantar hyperkeratosis and true hermaphroditism, 610644 Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644
RSPO2	94,8	88,3	100	100	?Humerofemoral hypoplasia with radiotibial ray deficiency, 618022 Tetraamelia syndrome 2, 618021
RSPO4	100	99,2	100	100	Anonychia congenita, 206800
RSPRY1	99,9	99,9	100	100	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723
RSRC1	99	94,6	100	100	Intellectual developmental disorder, autosomal recessive 70, 618402

RTEL1	99,7	97,2	100	100	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190
RTN4IP1	99,6	97,3	100	100	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
RTTN	98,6	97,6	100	100	Microcephaly, short stature, and polymicrogyria with seizures, 614833
RUBCN	99,7	97,9	100	100	Spinocerebellar ataxia, autosomal recessive 15, 615705
RUSC2	100	100	100	100	Mental retardation, autosomal recessive 61, 617773
RXYLT1	99,2	95,9	100	99,9	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
RYR1	97,1	94	99,4	99	Neuromuscular disease, congenital, with uniform type 1 fiber, 117000 Central core disease, 117000 King-Denborough syndrome, 145600 Minicore myopathy with external ophthalmoplegia, 255320
S1PR2	99	96,4	100	100	Deafness, autosomal recessive 68, 610419
SACS	99,9	99,9	100	100	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAG	100	100	100	100	Retinitis pigmentosa 47, 613758 Oguchi disease-1, 258100
SALL2	100	100	100	100	?Coloboma, ocular, autosomal recessive, 216820
SAMD9	99,9	99,8	100	100	Tumoral calcinosis, familial, normophosphatemic, 610455 Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041 MIRAGE syndrome, 617053
SAMHD1	98,5	97,9	100	100	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SAR1B	94,8	88,6	100	100	Chylomicron retention disease, 246700
SARS1	100	99,3	100	100	?Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709
SARS2	95,7	94,5	100	100	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SASH1	99,8	98,3	100	100	Dyschromatosis universalis hereditaria 1, 127500 ?Cancer, alopecia, pigment dyscrasia, onychodystrophy, and keratoderma, 618373
SASS6	99,1	98,1	100	99,7	?Microcephaly 14, primary, autosomal recessive, 616402
SBDS	100	99,9	100	100	Shwachman-Diamond syndrome, 260400
SBF1	99,1	98	100	100	Charcot-Marie-Tooth disease, type 4B3, 615284
SBF2	99,7	98,6	100	100	Charcot-Marie-Tooth disease, type 4B2, 604563
SC5D	99,9	99,1	100	100	Lathosterolosis, 607330
SCAPER	99,5	97,1	100	99,9	Intellectual developmental disorder and retinitis pigmentosa, 618195

SCARB2	99,9	99,4	100	100	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCARF2	97,4	88,9	99,8	99,2	Van den Ende-Gupta syndrome, 600920
SCN1B	98,2	96,3	99,7	98,9	Generalized epilepsy with febrile seizures plus, type 1, 604233 Developmental and epileptic encephalopathy 52, 617350 Cardiac conduction defect, nonspecific, 612838 Atrial fibrillation, familial, 13, 615377 Brugada syndrome 5, 612838
SCN4A	99,9	99,4	100	100	Paramyotonia congenita, 168300 Hypokalemic periodic paralysis, type 2, 613345 Myotonia congenita, atypical, acetazolamide-responsive, 608390 Myasthenic syndrome, congenital, 16, 614198 Hyperkalemic periodic paralysis, type 2, 170500
SCN9A	99,1	97	100	100	Erythermalgia, primary, 133020 Insensitivity to pain, congenital, 243000 Small fiber neuropathy, 133020 Paroxysmal extreme pain disorder, 167400 Neuropathy, hereditary sensory and autonomic, type IID, 243000
SCNN1A	99,7	97,5	100	100	Pseudohypoaldosteronism, type I, 264350 ?Liddle syndrome 3, 618126 Bronchiectasis with or without elevated sweat chloride 2, 613021
SCNN1B	100	99,8	100	100	Bronchiectasis with or without elevated sweat chloride 1, 211400 Pseudohypoaldosteronism, type I, 264350 Liddle syndrome 1, 177200
SCNN1G	99,8	98,4	100	100	Bronchiectasis with or without elevated sweat chloride 3, 613071 Pseudohypoaldosteronism, type I, 264350 Liddle syndrome 2, 618114
SCO1	97,6	94,4	100	100	Mitochondrial complex IV deficiency, nuclear type 4, 619048
SCO2	100	100	100	100	Myopia 6, 608908 Mitochondrial complex IV deficiency, nuclear type 2, 604377
SCP2	99,9	97,9	100	100	?Leukoencephalopathy with dystonia and motor neuropathy, 613724
SCYL1	100	99,9	100	100	Spinocerebellar ataxia, autosomal recessive 21, 616719
SDCCAG8	99,8	99,8	100	100	Senior-Loken syndrome 7, 613615 Bardet-Biedl syndrome 16, 615993
SDHA	84,5	77,9	100	100	Cardiomyopathy, dilated, 1GG, 613642 Mitochondrial complex II deficiency, nuclear type 1, 252011

					Neurodegeneration with ataxia and late-onset optic atrophy, 619259 Paragangliomas 5, 614165
SDHAF1	100	98,4	100	100	Mitochondrial complex II deficiency, nuclear type 2, 619166
SDHB	100	100	100	100	Paragangliomas 4, 115310 Mitochondrial complex II deficiency, nuclear type 4, 619224 Gastrointestinal stromal tumor, 606764 Pheochromocytoma, 171300 Paraganglioma and gastric stromal sarcoma, 606864
SDHD	53,8	49	80,1	80,1	Paragangliomas 1, with or without deafness, 168000 Paraganglioma and gastric stromal sarcoma, 606864 Mitochondrial complex II deficiency, nuclear type 3, 619167 Pheochromocytoma, 171300
SDR9C7	100	100	100	100	Ichthyosis, congenital, autosomal recessive 13, 617574
SEC23A	99,7	97	100	100	Craniolenticulosutural dysplasia, 607812
SEC23B	99,9	99,1	100	100	?Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SEC24D	99,9	99,3	100	100	Cole-Carpenter syndrome 2, 616294
SEC31A	99	96,2	100	100	?Neurodevelopmental disorder with spastic quadriplegia, optic atrophy, seizures, and structural brain anomalies, 618651
SECISBP2	99,5	95,9	100	100	Thyroid hormone metabolism, abnormal, 609698
SELENON	84,3	84	87,8	85,1	Myopathy, congenital, with fiber-type disproportion, 255310 Muscular dystrophy, rigid spine, 1, 602771
SEMA4A	100	99,4	100	100	Retinitis pigmentosa 35, 610282 Cone-rod dystrophy 10, 610283
SEPSECS	99,9	99,6	100	100	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	99,6	99,5	100	99,9	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPINA1	100	100	100	100	Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490 Emphysema due to AAT deficiency, 613490 Emphysema-cirrhosis, due to AAT deficiency, 613490
SERPINA6	100	100	100	100	Corticosteroid-binding globulin deficiency, 611489
SERPINB6	93,4	93,4	100	100	?Deafness, autosomal recessive 91, 613453
SERPINB7	100	99,8	100	99,6	Palmoplantar keratoderma, Nagashima type, 615598
SERPINB8	95	95	100	100	Peeling skin syndrome 5, 617115
SERPINC1	100	100	100	100	Thrombophilia due to antithrombin III deficiency, 613118

SERPINE1	100	100	100	100	Plasminogen activator inhibitor-1 deficiency, 613329
SERPINF1	100	99,9	100	100	Osteogenesis imperfecta, type VI, 613982
SERPINF2	100	99,9	100	100	Alpha-2-plasmin inhibitor deficiency, 262850
SERPING1	99,6	96,4	100	100	Angioedema, hereditary, 1 and 2, 106100 Complement component 4, partial deficiency of, 120790
SERPINH1	99,8	98	100	100	Osteogenesis imperfecta, type X, 613848
SETX	99,8	99,6	100	100	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 Amyotrophic lateral sclerosis 4, juvenile, 602433
SFRP4	99,8	99,1	100	100	Pyle disease, 265900
SFTPB	100	99,2	100	100	Surfactant metabolism dysfunction, pulmonary, 1, 265120
SFXN4	99,6	97,4	100	100	Combined oxidative phosphorylation deficiency 18, 615578
SGCA	100	99,6	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099
SGCB	97,8	96,5	100	99,9	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286
SGCD	99,6	96,5	100	100	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287
SGCG	100	99,4	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700
SGO1	99,5	99	100	100	Chronic atrial and intestinal dysrhythmia, 616201
SGPL1	100	100	100	100	Nephrotic syndrome, type 14, 617575
SGSH	94,8	94,1	100	100	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SH3PXD2B	100	99,9	100	100	Frank-ter Haar syndrome, 249420
SH3TC2	100	99,4	100	100	Charcot-Marie-Tooth disease, type 4C, 601596 Mononeuropathy of the median nerve, mild, 613353
SI	99	95,9	100	99,9	Sucrase-isomaltase deficiency, congenital, 222900
SIGMAR1	100	100	100	100	?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726 ?Amyotrophic lateral sclerosis 16, juvenile, 614373
SIK3	99,7	98	99,3	98	?Spondyloepimetaphyseal dysplasia, Krakow type, 618162
SIL1	98,7	96	100	100	Marinesco-Sjogren syndrome, 248800
SIX6	100	100	100	100	Optic disc anomalies with retinal and/or macular dystrophy, 212550
SKIV2L	100	99,5	100	100	Trichohepatoenteric syndrome 2, 614602
SLC10A2	100	100	100	100	?Bile acid malabsorption, primary, 1, 613291
SLC10A7	99,5	98,1	100	100	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363

SLC11A2	98,1	97,4	100	100	Anemia, hypochromic microcytic, with iron overload 1, 206100
SLC12A1	96,2	96	96,2	96,2	Bartter syndrome, type 1, 601678
SLC12A2	94,4	92,4	100	100	Kilquist syndrome, 619080 Delpire-McNeill syndrome, 619083 Deafness, autosomal dominant 78, 619081
SLC12A3	100	100	100	100	Gitelman syndrome, 263800
SLC12A5	83,9	83,8	97,4	97,4	Developmental and epileptic encephalopathy 34, 616645
SLC12A6	100	100	100	100	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC13A3	99,8	97,9	100	100	Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate, 618384
SLC13A5	100	100	100	100	Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905
SLC16A1	100	98,6	100	100	Hyperinsulinemic hypoglycemia, familial, 7, 610021 Erythrocyte lactate transporter defect, 245340 Monocarboxylate transporter 1 deficiency, 616095
SLC17A5	99,6	96,2	100	100	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC18A2	99,9	99,6	100	100	?Parkinsonism-dystonia, infantile, 2, 618049
SLC18A3	100	100	100	100	Myasthenic syndrome, congenital, 21, presynaptic, 617239
SLC19A2	100	98,5	100	100	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	97,8	97	98,7	98,7	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A1	100	99,5	100	100	Dicarboxylic aminoaciduria, 222730
SLC1A4	99,6	97	100	100	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC22A12	100	99,8	100	100	Hypouricemia, renal, 220150
SLC22A5	100	99,6	100	100	Carnitine deficiency, systemic primary, 212140
SLC24A1	100	99,9	100	100	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
SLC24A4	100	99,5	100	100	Amelogenesis imperfecta, type IIA5, 615887
SLC24A5	99,6	98,5	100	100	Albinism, oculocutaneous, type VI, 113750
SLC25A1	96,9	89,8	99,7	98,2	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182 Myasthenic syndrome, congenital, 23, presynaptic, 618197
SLC25A12	100	99,2	100	100	Developmental and epileptic encephalopathy 39, 612949
SLC25A13	100	99,4	100	100	Citrullinemia, type II, neonatal-onset, 605814 Citrullinemia, adult-onset type II, 603471
SLC25A15	99,3	96,6	100	100	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970

SLC25A19	99,9	98	100	100	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A20	100	98,9	100	100	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A22	99,2	96,5	100	100	Developmental and epileptic encephalopathy 3, 609304
SLC25A26	99,8	98,1	100	100	Combined oxidative phosphorylation deficiency 28, 616794
SLC25A3	99,7	96,9	100	100	Mitochondrial phosphate carrier deficiency, 610773
SLC25A38	97,4	93,3	100	100	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC25A4	100	99,8	100	100	Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283 Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184
SLC25A42	97,1	94,3	100	100	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416
SLC25A46	99,7	98,6	100	100	Neuropathy, hereditary motor and sensory, type VIB, 616505 Pontocerebellar hypoplasia, type 1E, 619303
SLC26A1	100	99,7	100	100	?Nephrolithiasis, calcium oxalate, 167030
SLC26A2	100	100	100	100	Epiphyseal dysplasia, multiple, 4, 226900 De la Chapelle dysplasia, 256050 Diastrophic dysplasia, 222600 Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 Achondrogenesis Ib, 600972 Atelosteogenesis, type II, 256050
SLC26A3	100	99,5	100	100	Diarrhea 1, secretory chloride, congenital, 214700
SLC26A4	99,9	99,7	100	100	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791 Pendred syndrome, 274600
SLC26A5	98,7	95,9	100	100	?Deafness, autosomal recessive 61, 613865
SLC27A4	100	99,9	100	100	Ichthyosis prematurity syndrome, 608649
SLC29A3	100	99,5	100	100	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC2A1	92,8	92,7	100	100	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	97,7	97,7	100	100	Arterial tortuosity syndrome, 208050
SLC2A2	100	99,8	100	100	Fanconi-Bickel syndrome, 227810
SLC2A9	99,3	95	100	100	Hypouricemia, renal, 2, 612076

SLC30A10	100	100	100	100	Hypermanganesemia with dystonia 1, 613280
SLC30A9	98,7	94,3	100	99,9	?Birk-Landau-Perez syndrome, 617595
SLC33A1	99,8	98,5	100	99,8	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC34A1	100	99,7	100	100	?Fanconi renotubular syndrome 2, 613388 Hypercalcemia, infantile, 2, 616963 Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286
SLC34A2	100	99,6	100	100	Pulmonary alveolar microlithiasis, 265100
SLC34A3	100	99,3	100	100	Hypophosphatemic rickets with hypercalciuria, 241530
SLC35A1	99,7	99,3	100	100	Congenital disorder of glycosylation, type IIf, 603585
SLC35A3	80,4	78,8	81	80,9	?Arthrogryposis, mental retardation, and seizures, 615553
SLC35C1	100	99,4	100	100	Congenital disorder of glycosylation, type IIc, 266265
SLC35D1	99,6	97,6	100	99,2	Schneckenbecken dysplasia, 269250
SLC37A4	99,8	97,6	100	100	Glycogen storage disease Ib, 232220 Congenital disorder of glycosylation, type IIw, 619525 Glycogen storage disease Ic, 232240
SLC38A8	99,9	97,9	100	100	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218
SLC39A13	99,9	97,9	100	100	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350
SLC39A14	100	99	93,5	93,5	?Hyperostosis cranalis interna, 144755 Hypermanganesemia with dystonia 2, 617013
SLC39A4	99,3	96,3	100	100	Acrodermatitis enteropathica, 201100
SLC39A8	100	99,7	100	100	Congenital disorder of glycosylation, type IIn, 616721
SLC3A1	100	99,7	96,6	96,6	Cystinuria, 220100
SLC44A1	98,2	98,1	100	99,9	Neurodegeneration, childhood-onset, with ataxia, tremor, optic atrophy, and cognitive decline, 618868
SLC45A1	100	100	100	100	Intellectual developmental disorder with neuropsychiatric features, 617532
SLC45A2	100	99,9	100	100	Albinism, oculocutaneous, type IV, 606574
SLC46A1	100	98,5	100	100	Folate malabsorption, hereditary, 229050
SLC4A1	100	99,9	96,1	96,1	Distal renal tubular acidosis 1, 179800 Spherocytosis, type 4, 612653 Distal renal tubular acidosis 4 with hemolytic anemia, 611590 Cryohydrocytosis, 185020 Ovalocytosis, SA type, 166900

SLC4A11	100	99,9	100	100	Corneal endothelial dystrophy, autosomal recessive, 217700 Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy and perceptive deafness, 217400
SLC4A4	99,9	99,4	100	100	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC52A2	100	100	100	100	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	100	100	100	100	?Fazio-Londe disease, 211500 Brown-Vialetto-Van Laere syndrome 1, 211530
SLC5A1	100	99,8	100	100	Glucose/galactose malabsorption, 606824
SLC5A2	100	100	100	100	Renal glucosuria, 233100
SLC5A5	100	99,9	100	100	Thyroid dyshormonogenesis 1, 274400
SLC5A7	100	100	100	100	Neuronopathy, distal hereditary motor, type VIIA, 158580 Myasthenic syndrome, congenital, 20, presynaptic, 617143
SLC6A17	100	100	100	100	Mental retardation, autosomal recessive 48, 616269
SLC6A19	100	100	100	100	Iminoglycinuria, digenic, 242600 Hartnup disorder, 234500 Hyperglycinuria, 138500
SLC6A3	100	99,9	100	100	Parkinsonism-dystonia, infantile, 1, 613135
SLC6A5	100	99,9	100	100	Hyperekplexia 3, 614618
SLC6A9	100	99,6	100	100	Glycine encephalopathy with normal serum glycine, 617301
SLC7A14	100	100	100	100	Retinitis pigmentosa 68, 615725
SLC7A7	100	99,9	100	100	Lysinuric protein intolerance, 222700
SLC7A9	100	99,4	100	100	Cystinuria, 220100
SLC9A1	100	100	100	100	Lichtenstein-Knorr syndrome, 616291
SLC9A3	90,5	86	96	93,6	Diarrhea 8, secretory sodium, congenital, 616868
SLCO2A1	99,9	98	100	100	Hypertrophic osteoarthropathy, primary, autosomal dominant, 167100 Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
SLITRK6	100	99,9	100	100	Deafness and myopia, 221200
SLURP1	100	99,4	100	100	Meleda disease, 248300
SLX4	100	99,9	100	100	Fanconi anemia, complementation group P, 613951
SMARCAL1	100	99,8	100	100	Schimke immunoosseous dysplasia, 242900
SMARCD2	87	85,8	99,9	98,6	Specific granule deficiency 2, 617475
SMG9	100	100	100	100	Heart and brain malformation syndrome, 616920

SMN1	99,7	96,1	94,6	94,6	Spinal muscular atrophy-2, 253550 Spinal muscular atrophy-4, 271150 Spinal muscular atrophy-3, 253400 Spinal muscular atrophy-1, 253300
SMO	98,9	94,7	100	100	Pallister-Hall-like syndrome, 241800 Basal cell carcinoma, somatic, 605462 Curry-Jones syndrome, somatic mosaic, 601707
SMOC1	99,8	98,2	100	100	Microphthalmia with limb anomalies, 206920
SMOC2	76,7	74,9	100	100	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SMPD1	100	99,9	100	100	Niemann-Pick disease, type B, 607616 Niemann-Pick disease, type A, 257200
SMPD4	99,6	95	100	100	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622
SNAI2	99,9	98	100	100	Waardenburg syndrome, type 2D, 608890 Piebaldism, 172800
SNAP29	100	100	100	100	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNIP1	99,2	97,3	100	100	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501
SNORD118	NC	NC	NC	NC	Leukoencephalopathy, brain calcifications, and cysts, 614561
SNX10	96,2	95,9	99,9	99,3	Osteopetrosis, autosomal recessive 8, 615085
SNX14	98,9	93,6	100	100	Spinocerebellar ataxia, autosomal recessive 20, 616354
SOBP	98,5	95,9	97,4	95,5	Mental retardation, anterior maxillary protrusion, and strabismus, 613671
SOD1	100	100	100	100	Spastic tetraplegia and axial hypotonia, progressive, 618598 Amyotrophic lateral sclerosis 1, 105400
SORD	90,6	89,4	98,4	95,1	Sorbitol dehydrogenase deficiency with peripheral neuropathy, 618912
SOST	100	99,6	100	100	Sclerosteosis 1, 269500 Craniodiaphyseal dysplasia, autosomal dominant, 122860
SOX18	75,2	55,5	95,7	91,7	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823 Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940
SP110	100	100	100	100	Hepatic venoocclusive disease with immunodeficiency, 235550
SP7	99,9	99,2	100	100	Osteogenesis imperfecta, type XII, 613849
SPAG1	98,7	93,9	99,6	97,9	Ciliary dyskinesia, primary, 28, 615505
SPARC	100	100	100	100	Osteogenesis imperfecta, type XVII, 616507
SPART	99,7	96,4	100	100	Troyer syndrome, 275900
SPATA5	99,8	99,5	100	100	Epilepsy, hearing loss, and mental retardation syndrome, 616577

SPATA7	99,6	98,2	100	100	Retinitis pigmentosa, juvenile, autosomal recessive, 604232 Leber congenital amaurosis 3, 604232
SPEG	97,2	91,1	99,7	99,7	Centronuclear myopathy 5, 615959
SPG11	99,8	99	100	100	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360
SPG21	98,9	94,7	100	100	Mast syndrome, 248900
SPG7	90,4	86,7	100	100	Spastic paraplegia 7, autosomal recessive, 607259
SPINK5	99,8	99,6	100	99,9	Netherton syndrome, 256500
SPINT2	97,5	78,8	100	100	Diarrhea 3, secretory sodium, congenital, syndromic, 270420
SPNS2	92,3	89,6	96,7	95	?Deafness, autosomal recessive 115, 618457
SPR	100	99,4	100	100	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRTN	100	100	100	100	Ruijs-Aalfs syndrome, 616200
SPTA1	99,9	98,8	100	100	Spherocytosis, type 3, 270970 Elliptocytosis-2, 130600 Pyropoikilocytosis, 266140
SPTB	100	99,9	100	100	Anemia, neonatal hemolytic, fatal or near-fatal, 617948 Elliptocytosis-3, 617948 Spherocytosis, type 2, 616649
SPTBN2	100	99,4	100	99,9	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia, autosomal recessive 14, 615386
SPTBN4	98,1	92,1	100	100	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519
SQSTM1	99,8	97,8	100	100	Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 Myopathy, distal, with rimmed vacuoles, 617158 Paget disease of bone 3, 167250
SRD5A2	100	98,8	100	100	Pseudovaginal perineoscrotal hypospadias, 264600
SRD5A3	100	99,1	100	100	Kahrizi syndrome, 612713 Congenital disorder of glycosylation, type Iq, 612379
ST14	99,9	98,9	100	100	Ichthyosis, congenital, autosomal recessive 11, 602400
ST3GAL3	68,8	68,2	95,3	95,2	Developmental and epileptic encephalopathy 15, 615006 Intellectual developmental disorder, autosomal recessive 12, 611090
ST3GAL5	85,9	84	98,7	98,6	Salt and pepper developmental regression syndrome, 609056
STAC3	100	100	100	100	Myopathy, congenital, Baily-Bloch, 255995

STAMBP	99,4	96,4	100	100	Microcephaly-capillary malformation syndrome, 614261
STAR	100	99,9	100	100	Lipoid adrenal hyperplasia, 201710
STAT1	93,1	90,2	95,7	95	Immunodeficiency 31C, chronic mucocutaneous candidiasis, autosomal dominant, 614162 Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796
STAT2	100	99,4	100	100	Pseudo-TORCH syndrome 3, 618886 Immunodeficiency 44, 616636
STAT5B	99,9	98,1	100	100	Growth hormone insensitivity with immune dysregulation 1, autosomal recessive, 245590 Growth hormone insensitivity with immune dysregulation 2, autosomal dominant, 618985 Leukemia, acute promyelocytic, somatic, 102578
STIL	99,9	99,7	100	100	Microcephaly 7, primary, autosomal recessive, 612703
STIM1	99,9	97,5	100	100	Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070 Immunodeficiency 10, 612783
STK4	99,9	99,7	100	100	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STN1	99,9	99,8	100	100	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341
STRA6	100	99,9	100	100	Microphthalmia, syndromic 9, 601186 Microphthalmia, isolated, with coloboma 8, 601186
STRADA	100	99	100	100	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087
STRC	99,9	98,3	100	100	Deafness, autosomal recessive 16, 603720
STT3A	100	100	100	100	Congenital disorder of glycosylation, type Iw, 615596
STT3B	99,7	99,4	100	100	?Congenital disorder of glycosylation, type Ix, 615597
STUB1	100	98,2	100	100	Spinocerebellar ataxia 48, 618093 Spinocerebellar ataxia, autosomal recessive 16, 615768
STX11	100	100	100	100	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STXBP2	82,4	79,9	99,7	98	Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease, 613101
SUCLA2	88,8	79,4	99,9	99,8	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	100	99,7	100	99,8	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUFU	100	100	100	100	Joubert syndrome 32, 617757 Medulloblastoma, desmoplastic, 155255 Basal cell nevus syndrome, 109400
SULT2B1	100	100	100	100	Ichthyosis, congenital, autosomal recessive 14, 617571

SUMF1	98,3	92,5	100	100	Multiple sulfatase deficiency, 272200
SUOX	100	100	100	100	Sulfite oxidase deficiency, 272300
SURF1	89,5	88,1	100	100	Charcot-Marie-Tooth disease, type 4K, 616684 Mitochondrial complex IV deficiency, nuclear type 1, 220110
SVBP	100	100	100	100	Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569
SYNE1	98,1	97,5	98,8	98,8	Arthrogryposis multiplex congenita 3, myogenic type, 618484 Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 Spinocerebellar ataxia, autosomal recessive 8, 610743
SYNE4	99,9	97,5	100	100	Deafness, autosomal recessive 76, 615540
SYNJ1	99,7	98,1	100	100	Parkinson disease 20, early-onset, 615530 Developmental and epileptic encephalopathy 53, 617389
SYT14	61	60,4	100	100	?Spinocerebellar ataxia, autosomal recessive 11, 614229
SZT2	99,6	99,3	100	99,9	Developmental and epileptic encephalopathy 18, 615476
TAC3	99,9	93,6	100	100	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACO1	98,9	93,7	100	100	Mitochondrial complex IV deficiency, nuclear type 8, 619052
TACR3	100	100	100	100	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
TACSTD2	99,1	96,6	100	100	Corneal dystrophy, gelatinous drop-like, 204870
TAF13	99,6	99,1	100	99,9	Mental retardation, autosomal recessive 60, 617432
TAF1C	100	100	100	100	No OMIM disease ID
TAF2	99,5	98,6	100	99,9	Mental retardation, autosomal recessive 40, 615599
TAF6	99,6	98,1	100	100	Alazami-Yuan syndrome, 617126
TALDO1	100	98	100	100	Transaldolase deficiency, 606003
TANGO2	100	99,3	100	100	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TAP1	100	97,6	100	100	Bare lymphocyte syndrome, type I, 604571
TAP2	99,9	98,6	100	100	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	96,5	95,1	96,6	96,6	Bare lymphocyte syndrome, type I, 604571
TAPT1	93	87,1	98,5	94,1	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897
TARS2	99,9	98,8	100	100	?Combined oxidative phosphorylation deficiency 21, 615918
TAT	100	100	100	100	Tyrosinemia, type II, 276600
TBC1D20	94,3	93,9	100	99,7	Warburg micro syndrome 4, 615663

TBC1D23	98,7	94,5	100	99,7	Pontocerebellar hypoplasia, type 11, 617695
TBC1D24	100	100	100	100	Deafness, autosomal recessive 86, 614617 Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp, 608105 Myoclonic epilepsy, infantile, familial, 605021 Deafness, autosomal dominant 65, 616044 Developmental and epileptic encephalopathy 16, 615338 DOORS syndrome, 220500
TBC1D7	99,7	99,3	100	100	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000
TBCD	95,5	93,3	100	100	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TBCE	99,7	96,6	100	100	Kenny-Caffey syndrome, type 1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
TBCK	99,4	95,8	100	99,9	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900
TBX15	100	99,7	100	100	Cousin syndrome, 260660
TBX19	100	100	100	100	Adrenocorticotropic hormone deficiency, 201400
TBX6	99,2	94,8	100	100	Spondylocostal dysostosis 5, 122600
TBXAS1	100	100	100	100	Ghosal hematodiaphyseal syndrome, 231095
TBXT	99,3	94,9	100	100	Sacral agenesis with vertebral anomalies, 615709
TCAP	100	100	100	100	Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954
TCIRG1	98,5	93,4	100	100	Osteopetrosis, autosomal recessive 1, 259700
TCN2	100	100	100	100	Transcobalamin II deficiency, 275350
TCTN1	96,8	92,8	94,7	94,7	Joubert syndrome 13, 614173
TCTN2	99,9	99,1	100	100	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	100	100	100	100	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TDP1	99,9	99,4	100	100	?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250
TDP2	99,6	99,5	100	99,9	Spinocerebellar ataxia, autosomal recessive 23, 616949
TDRD7	99,9	99,3	100	100	Cataract 36, 613887
TECPR2	100	100	100	100	Spastic paraplegia 49, autosomal recessive, 615031
TECR	100	98,5	100	100	Mental retardation, autosomal recessive 14, 614020

TECRL	97,5	91,7	100	99,4	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021
TECTA	100	99,8	100	100	Deafness, autosomal dominant 8/12, 601543 Deafness, autosomal recessive 21, 603629
TELO2	99,9	98	100	100	You-Hoover-Fong syndrome, 616954
TENM3	100	99,7	100	100	Microphthalmia, syndromic 15, 615145 ?Microphthalmia, isolated, with coloboma 9, 615145
TENT5A	100	99,5	100	100	Osteogenesis imperfecta, type XVIII, 617952
TF	100	99,9	100	100	Atransferrinemia, 209300
TFAM	98	78,5	100	100	?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156
TFG	97	96	100	99,9	?Spastic paraplegia 57, autosomal recessive, 615658 Hereditary motor and sensory neuropathy, Okinawa type, 604484
TFR2	99,3	96,9	100	100	Hemochromatosis, type 3, 604250
TFRC	99,9	99,6	100	100	Immunodeficiency 46, 616740
TG	99,9	98,5	100	100	Thyroid dyshormonogenesis 3, 274700
TGDS	99,4	95,9	100	99,9	Catel-Manzke syndrome, 616145
TGFB1	100	98,8	100	100	Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213 Camurati-Engelmann disease, 131300
TGM1	100	99,5	100	100	Ichthyosis, congenital, autosomal recessive 1, 242300
TGM5	100	99,4	100	100	Peeling skin syndrome 2, 609796
TH	99,8	98	100	100	Segawa syndrome, recessive, 605407
THOC6	100	100	100	100	Beaulieu-Boycott-Innes syndrome, 613680
THRB	100	99,6	100	100	Thyroid hormone resistance, autosomal recessive, 274300 Thyroid hormone resistance, 188570 Thyroid hormone resistance, selective pituitary, 145650
TIMM50	98,4	95	100	100	3-methylglutaconic aciduria, type IX, 617698
TIMMDC1	99,9	99,8	100	100	Mitochondrial complex I deficiency, nuclear type 31, 618251
TJP2	92,8	92,3	98,8	98,8	Hypercholanemia, familial 1, 607748 Cholestasis, progressive familial intrahepatic 4, 615878
TK2	99	96	100	100	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560 ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069
TKT	98,6	96,8	98,7	98,7	Short stature, developmental delay, and congenital heart defects, 617044
TLE6	99,9	97,8	100	100	Preimplantation embryonic lethality, 616814

TMC1	99,8	96,6	100	99,9	Deafness, autosomal dominant 36, 606705 Deafness, autosomal recessive 7, 600974
TMC6	100	99,6	100	100	Epidermodysplasia verruciformis, 226400
TMC8	99,9	98,9	100	100	Epidermodysplasia verruciformis 2, 618231
TMCO1	87,8	87	88	87,9	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980
TMEM107	100	100	100	100	Orofaciodigital syndrome XVI, 617563 Meckel syndrome 13, 617562 ?Joubert syndrome 29, 617562
TMEM126A	95,4	80	100	100	Optic atrophy 7, 612989
TMEM126B	99,6	97,2	100	100	Mitochondrial complex I deficiency, nuclear type 29, 618250
TMEM132E	97,5	94,8	100	100	Deafness, autosomal recessive 99, 618481
TMEM138	99,8	93,1	100	100	Joubert syndrome 16, 614465
TMEM165	100	100	100	100	Congenital disorder of glycosylation, type IIk, 614727
TMEM199	100	100	100	100	Congenital disorder of glycosylation, type IIp, 616829
TMEM216	98,5	92,8	100	100	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	100	99,3	100	100	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	99,8	99,3	100	100	Joubert syndrome 14, 614424
TMEM260	98,7	95,4	100	100	Structural heart defects and renal anomalies syndrome, 617478
TMEM38B	99,8	99,8	100	100	Osteogenesis imperfecta, type XIV, 615066
TMEM67	98,6	93,5	100	99,6	Nephronophthisis 11, 613550 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 ?RHYNS syndrome, 602152 COACH syndrome 1, 216360
TMEM70	98,4	94,6	100	100	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMEM94	100	100	100	100	Intellectual developmental disorder with cardiac defects and dysmorphic facies, 618316
TMIE	99,9	97,5	100	100	Deafness, autosomal recessive 6, 600971
TMPRSS15	98,2	95,7	100	99,9	Enterokinase deficiency, 226200
TMPRSS3	100	99,3	100	100	Deafness, autosomal recessive 8/10, 601072
TMPRSS6	100	99,3	100	100	Iron-refractory iron deficiency anemia, 206200

TMTC3	98,7	95,8	100	100	Lissencephaly 8, 617255
TNFRSF11A	94,9	93,8	99,1	97,7	Osteopetrosis, autosomal recessive 7, 612301 Osteolysis, familial expansile, 174810
TNFRSF11B	100	100	100	100	Paget disease of bone 5, juvenile-onset, 239000
TNFRSF13B	100	99,9	100	100	Immunodeficiency, common variable, 2, 240500 Immunoglobulin A deficiency 2, 609529
TNFRSF13C	85	75,6	100	100	Immunodeficiency, common variable, 4, 613494
TNFRSF4	97,7	89	100	100	?Immunodeficiency 16, 615593
TNFSF11	100	100	100	100	Osteopetrosis, autosomal recessive 2, 259710
TNIK	99,8	98,5	100	100	Mental retardation, autosomal recessive 54, 617028
TNNI3	99,6	95,5	100	100	?Cardiomyopathy, dilated, 2A, 611880 Cardiomyopathy, hypertrophic, 7, 613690 Cardiomyopathy, familial restrictive, 1, 115210 Cardiomyopathy, dilated, 1FF, 613286
TNNT1	99,6	97,1	100	100	Nemaline myopathy 5, Amish type, 605355
TNXB	98,7	93,9	100	100	Ehlers-Danlos syndrome, classic-like, 1, 606408 Vesicoureteral reflux 8, 615963
TOE1	100	100	100	100	Pontocerebellar hypoplasia, type 7, 614969
TONSL	99,9	98,4	100	100	Spondyloepimetaphyseal dysplasia, sponastrime type, 271510
TOP3A	99,6	96,5	100	100	?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098 Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097
TOR1AIP1	99,2	96,1	100	100	?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072
TP53RK	95,5	84,9	100	100	Galloway-Mowat syndrome 4, 617730
TPI1	99,8	98	100	100	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512
TPK1	99,5	97,2	100	100	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TPM3	87,7	84,3	100	100	CAP myopathy 1, 609284 Myopathy, congenital, with fiber-type disproportion, 255310 Nemaline myopathy 1, autosomal dominant or recessive, 609284
TPO	100	99,2	100	100	Thyroid dyshormonogenesis 2A, 274500
TPP1	100	100	100	100	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TPRKB	80,2	75,2	81,9	81,7	Galloway-Mowat syndrome 5, 617731
TPRN	89,7	83,4	94,5	88,3	Deafness, autosomal recessive 79, 613307

TRAC	100	100	100	100	Immunodeficiency 7, TCR-alpha/beta deficient, 615387
TRAF3IP1	98,7	95,4	100	100	Senior-Loken syndrome 9, 616629
TRAIP	100	100	100	99,9	Seckel syndrome 9, 616777
TRAK1	93,3	93,1	100	99,9	Developmental and epileptic encephalopathy 68, 618201
TRAPPC11	99,7	98,7	100	99,9	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356
TRAPPC12	100	99,9	100	100	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669
MAP11	100	98,8	100	100	?Microcephaly 25, primary, autosomal recessive, 618351
TRAPPC2L	100	100	100	100	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331
TRAPPC6B	99,2	96,4	100	100	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862
TRAPPC9	100	99,7	100	100	Mental retardation, autosomal recessive 13, 613192
TRDN	97,7	89,1	100	99,5	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441
TREM2	100	99,3	100	100	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193
TREX1	100	100	100	100	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
TRH	99,7	97,5	100	100	No OMIM disease ID
TRIM2	93,8	93,5	93,9	93,9	Charcot-Marie-Tooth disease, type 2R, 615490
TRIM32	100	99,9	100	100	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRIM36	99,8	98,7	100	100	?Anencephaly 1, 206500
TRIM37	98,3	97,1	98,7	98,6	Mulibrey nanism, 253250
TRIOBP	98,5	96,6	99,9	99,2	Deafness, autosomal recessive 28, 609823
TRIP11	97,2	92,6	100	99,9	Odontochondrodysplasia 1, 184260 Achondrogenesis, type IA, 200600
TRIP13	100	99,9	100	100	Oocyte maturation defect 9, 619011 Mosaic variegated aneuploidy syndrome 3, 617598
TRIP4	99,8	99	100	100	?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066 Spinal muscular atrophy with congenital bone fractures 1, 616866
TRIT1	100	100	100	100	Combined oxidative phosphorylation deficiency 35, 617873
TRMT1	99,5	96,2	100	100	Mental retardation, autosomal recessive 68, 618302
TRMT10A	99,7	99,5	100	100	Microcephaly, short stature, and impaired glucose metabolism 1, 616033

TRMT10C	100	99,9	100	99,9	Combined oxidative phosphorylation deficiency 30, 616974
TRMT5	99,8	99,1	100	100	Combined oxidative phosphorylation deficiency 26, 616539
TRMU	99,9	99,6	100	99,9	Liver failure, transient infantile, 613070
TRNT1	99,7	97,4	100	99,9	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 Retinitis pigmentosa and erythrocytic microcytosis, 616959
TRPM1	100	99,4	100	100	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TRPM6	99,9	99,1	100	100	Hypomagnesemia 1, intestinal, 602014
TSEN15	78,9	77	100	100	Pontocerebellar hypoplasia, type 2F, 617026
TSEN2	99,9	99,2	100	100	Pontocerebellar hypoplasia type 2B, 612389
TSEN34	92,1	85,6	100	100	?Pontocerebellar hypoplasia type 2C, 612390
TSEN54	96,7	94,8	99,9	99,2	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753 ?Pontocerebellar hypoplasia type 5, 610204
TSFM	100	99,3	94,9	94,9	Combined oxidative phosphorylation deficiency 3, 610505
TSHB	100	100	100	100	Hypothyroidism, congenital, nongoitrous 4, 275100
TSHR	95,9	95,1	100	100	Hyperthyroidism, familial gestational, 603373 Hyperthyroidism, nonautoimmune, 609152 Hypothyroidism, congenital, nongoitrous, 1, 275200 Thyroid adenoma, hyperfunctioning, somatic, Thyroid carcinoma with thyrotoxicosis,
TSPAN12	100	99,9	100	100	Exudative vitreoretinopathy 5, 613310
TSPEAR	100	99,7	100	100	?Deafness, autosomal recessive 98, 614861 Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180
TSPYL1	100	100	100	100	Sudden infant death with dysgenesis of the testes syndrome, 608800
TTC19	83,8	74,1	100	99,8	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTC21B	99,7	99,1	100	99,9	Short-rib thoracic dysplasia 4 with or without polydactyly, 613819 Nephronophthisis 12, 613820
TTC37	99,7	98,8	100	100	Trichohepatoenteric syndrome 1, 222470
TTC7A	99,6	97,1	100	100	Gastrointestinal defects and immunodeficiency syndrome, 243150
TTC8	99,5	98	100	100	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
TTI2	100	99,9	100	100	Mental retardation, autosomal recessive 39, 615541
TTLL5	99,9	98,1	100	100	Cone-rod dystrophy 19, 615860

TTPA	96,2	89,6	100	100	Ataxia with isolated vitamin E deficiency, 277460
TUB	99,8	97,1	100	100	?Retinal dystrophy and obesity, 616188
TUBA8	99,9	99,2	100	100	No OMIM disease ID
TUBGCP2	99,1	95,5	97	97	Pachygyria, microcephaly, developmental delay, and dysmorphic facies, with or without seizures, 618737
TUBGCP4	98,9	94,7	100	99,8	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TUBGCP6	100	99,4	100	100	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
TUFM	99,9	97,6	100	100	Combined oxidative phosphorylation deficiency 4, 610678
TULP1	99,8	98,2	100	100	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132
TUSC3	100	99,7	100	100	Mental retardation, autosomal recessive 7, 611093
TWIST2	100	99,9	100	100	Ablepharon-macrostomia syndrome, 200110 Barber-Say syndrome, 209885 Focal facial dermal dysplasia 3, Setleis type, 227260
TWNK	100	99,9	100	100	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 Perrault syndrome 5, 616138
TXN2	100	98,5	100	100	?Combined oxidative phosphorylation deficiency 29, 616811
TXNL4A	99,3	99,1	100	100	Burn-McKeown syndrome, 608572
TYK2	100	99,3	100	100	Immunodeficiency 35, 611521
TYMP	100	99,4	100	100	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYR	100	100	100	100	Albinism, oculocutaneous, type IB, 606952 Waardenburg syndrome/albinism, digenic, 103470 Albinism, oculocutaneous, type IA, 203100
TYROBP	100	100	100	100	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770
TYRP1	100	99,9	100	100	Albinism, oculocutaneous, type III, 203290
UBA5	97,4	86,6	100	100	?Spinocerebellar ataxia, autosomal recessive 24, 617133 Developmental and epileptic encephalopathy 44, 617132
UBE2T	99,9	99,3	100	100	Fanconi anemia, complementation group T, 616435
UBE3B	100	99,7	100	100	Kaufman oculocerebrofacial syndrome, 244450
UBR1	99,6	99,1	98	97,9	Johanson-Blizzard syndrome, 243800
UCHL1	99,3	90,5	100	100	Spastic paraplegia 79, autosomal recessive, 615491
UFC1	100	100	100	100	Neurodevelopmental disorder with spasticity and poor growth, 618076

UFM1	72,4	69,1	100	100	Leukodystrophy, hypomyelinating, 14, 617899
UGDH	99,7	99,1	100	100	Developmental and epileptic encephalopathy 84, 618792
UGT1A1	100	100	100	100	Crigler-Najjar syndrome, type I, 218800 Hyperbilirubinemia, familial transient neonatal, 237900 Crigler-Najjar syndrome, type II, 606785
UMPS	99,9	98,7	97	97	Orotic aciduria, 258900
UNC13D	99,3	97,4	100	100	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC80	97,9	97,1	100	100	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801
UNG	99,9	97,9	100	100	Immunodeficiency with hyper IgM, type 5, 608106
UPB1	100	100	100	100	Beta-ureidopropionase deficiency, 613161
UQCC2	99,9	98,5	100	100	Mitochondrial complex III deficiency, nuclear type 7, 615824
UQCC3	100	97,5	100	100	?Mitochondrial complex III deficiency, nuclear type 9, 616111
UQCRB	97,7	92,1	100	100	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC2	100	98,8	100	100	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRFS1	94,1	88,8	100	100	Mitochondrial complex III deficiency, nuclear type 10, 618775
UQCRQ	100	100	100	100	Mitochondrial complex III deficiency, nuclear type 4, 615159
UROC1	100	99,9	100	100	?Urocanase deficiency, 276880
UROD	98,5	95,5	100	100	Porphyria, hepatoerythropoietic, 176100 Porphyria cutanea tarda, 176100
UROS	100	99,9	100	100	Porphyria, congenital erythropoietic, 263700
USB1	100	98,8	100	100	Poikiloderma with neutropenia, 604173
USH1C	99,9	99,2	100	100	Usher syndrome, type 1C, 276904 Deafness, autosomal recessive 18A, 602092
USH1G	99,7	96,6	100	100	Usher syndrome, type 1G, 606943
USH2A	100	99,7	99,5	99,5	Usher syndrome, type 2A, 276901 Retinitis pigmentosa 39, 613809
USP18	95,9	95,9	100	100	Pseudo-TORCH syndrome 2, 617397
USP45	99,4	97,6	100	99,9	?Leber congenital amaurosis 19, 618513
UVSSA	100	100	100	100	UV-sensitive syndrome 3, 614640
VAC14	99,8	98,5	100	100	Striatonigral degeneration, childhood-onset, 617054

VAMP1	100	99,8	100	100	Myasthenic syndrome, congenital, 25, 618323 Spastic ataxia 1, autosomal dominant, 108600
VARS1	100	99,7	100	100	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802
VARS2	100	99	100	100	Combined oxidative phosphorylation deficiency 20, 615917
VAX1	98,9	93,9	96	92,3	?Microphthalmia, syndromic 11, 614402
VDR	96,7	94,4	99,5	97,7	Rickets, vitamin D-resistant, type IIA, 277440
VHL	95,5	90,6	100	100	Erythrocytosis, familial, 2, 263400 von Hippel-Lindau syndrome, 193300 Renal cell carcinoma, somatic, 144700 Pheochromocytoma, 171300 Hemangioblastoma, cerebellar, somatic,
VIPAS39	100	100	100	100	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VKORC1	100	99,8	93	93	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473 Warfarin resistance, 122700
VLDLR	100	100	100	100	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VPS11	94,6	92,2	100	100	Leukodystrophy, hypomyelinating, 12, 616683
VPS13A	98,4	94,6	100	99,9	Choreoacanthocytosis, 200150
VPS13B	99,4	97,8	99,4	99,3	Cohen syndrome, 216550
VPS13C	99	95,8	100	99,9	Parkinson disease 23, autosomal recessive, early onset, 616840
VPS13D	100	99,4	100	100	Spinocerebellar ataxia, autosomal recessive 4, 607317
VPS33A	91,9	89,9	89,9	89,9	Mucopolysaccharidosis-plus syndrome, 617303
VPS33B	100	99,9	100	100	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
VPS37A	91,3	76	100	100	Spastic paraplegia 53, autosomal recessive, 614898
VPS45	97,8	95,1	95,3	95,3	Neutropenia, severe congenital, 5, autosomal recessive, 615285
VPS51	96,5	84,1	100	100	Pontocerebellar hypoplasia, type 13, 618606
VPS53	91,1	89,9	100	99,4	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	99,4	97,8	100	100	Pontocerebellar hypoplasia type 1A, 607596
VSX2	100	99,2	100	100	Microphthalmia, isolated 2, 610093 Microphthalmia with coloboma 3, 610092
VWA1	85,4	77,7	99,6	96,1	Neuropathy, hereditary motor, with myopathic features, 619216
VWA3B	99,9	98,9	100	100	?Spinocerebellar ataxia, autosomal recessive 22, 616948

VWF	99,9	99	100	100	von Willebrand disease, type 1, 193400 von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 von Willebrand disease, type 3, 277480
WARS2	100	99,8	100	99,8	Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710
WASHC4	98,9	96	100	99,8	?Mental retardation, autosomal recessive 43, 615817
WASHC5	99,8	99,7	100	100	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, autosomal dominant, 603563
WBP2	100	99,2	100	100	Deafness, autosomal recessive 107, 617639
WDPCP	98	94,1	98,1	98	?Bardet-Biedl syndrome 15, 615992 Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR19	99,8	98,6	100	99,9	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 ?Cranioectodermal dysplasia 4, 614378
WDR35	99,6	98,4	100	100	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 Cranioectodermal dysplasia 2, 613610
WDR4	100	100	100	100	Galloway-Mowat syndrome 6, 618347 Microcephaly, growth deficiency, seizures, and brain malformations, 618346
WDR45B	94,8	80,3	100	100	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977
WDR62	100	99,9	100	100	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
WDR72	96,5	95,6	96,9	96,9	Amelogenesis imperfecta, type IIA3, 613211
WDR73	100	100	100	100	Galloway-Mowat syndrome 1, 251300
WDR81	100	100	100	100	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185 Hydrocephalus, congenital, 3, with brain anomalies, 617967
WEE2	99,9	99,6	100	100	Oocyte maturation defect 5, 617996
WFS1	100	99,8	100	100	Deafness, autosomal dominant 6/14/38, 600965 ?Cataract 41, 116400 Wolfram-like syndrome, autosomal dominant, 614296 Wolfram syndrome 1, 222300
WHRN	99,6	97,6	100	100	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
WIPF1	99,9	98,5	100	100	Wiskott-Aldrich syndrome 2, 614493
WIPI2	99,7	98,1	100	100	?Intellectual developmental disorder with short stature and variable skeletal anomalies, 618453

WNK1	99,8	99,3	100	100	Neuropathy, hereditary sensory and autonomic, type II, 201300 Pseudohypoaldosteronism, type IIC, 614492
WNT1	99,9	96,6	100	100	Osteogenesis imperfecta, type XV, 615220
WNT10A	100	98,9	100	100	Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400 Odontoonychodermal dysplasia, 257980
WNT10B	100	99,6	100	100	Tooth agenesis, selective, 8, 617073 Split-hand/foot malformation 6, 225300
WNT3	100	99,9	100	100	?Tetra-amelia syndrome 1, 273395
WNT4	97,8	93,6	99,3	96,5	?SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330
WNT7A	100	100	100	100	Fuhrmann syndrome, 228930 Ulna and fibula, absence of, with severe limb deficiency, 276820
WRAP53	100	100	100	99,9	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	99,3	98,2	100	99,9	Werner syndrome, 277700
WWOX	100	99,9	100	100	Esophageal squamous cell carcinoma, somatic, 133239 Developmental and epileptic encephalopathy 28, 616211 Spinocerebellar ataxia, autosomal recessive 12, 614322
XDH	100	99,8	100	100	Xanthinuria, type I, 278300
XPA	99,2	97,3	100	100	Xeroderma pigmentosum, group A, 278700
XPC	100	99,9	100	100	Xeroderma pigmentosum, group C, 278720
XPNPEP3	100	100	100	100	Nephronophthisis-like nephropathy 1, 613159
XRCC1	99,6	97,2	100	100	?Spinocerebellar ataxia, autosomal recessive 26, 617633
XRCC2	99,6	95,7	100	100	Spermatogenic failure, 619145 ?Premature ovarian failure 17, 619146 ?Fanconi anemia, complementation group U, 617247
XRCC4	99,7	98,4	100	100	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	97,8	91,1	97,7	94,1	Desbuquois dysplasia 2, 615777
XYLT2	99,9	97,1	96,7	96,7	Spondyloocular syndrome, 605822
YARS2	99,9	99,4	100	100	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
YIF1B	99,9	99,2	90,1	90,1	Kaya-Barakat-Masson syndrome, 619125
YME1L1	98,9	93,7	100	100	?Optic atrophy 11, 617302
YY1AP1	98,5	97	100	100	Grange syndrome, 602531

ZAP70	100	99,7	100	100	Immunodeficiency 48, 269840 Autoimmune disease, multisystem, infantile-onset, 2, 617006
ZBTB11	99,9	99,3	100	100	Intellectual developmental disorder, autosomal recessive 69, 618383
ZBTB16	100	100	100	100	Skeletal defects, genital hypoplasia, and mental retardation, 612447 Leukemia, acute promyelocytic, PL2F/RARA type,
ZBTB24	100	100	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069
ZBTB42	100	100	100	100	?Lethal congenital contracture syndrome 6, 616248
ZC3H14	99,7	98,2	100	99,9	Mental retardation, autosomal recessive 56, 617125
ZFYVE26	99,7	97,8	100	100	Spastic paraplegia 15, autosomal recessive, 270700
ZMPSTE24	99,6	99,4	100	99,9	Mandibuloacral dysplasia with type B lipodystrophy, 608612 Restrictive dermopathy, lethal, 275210
ZMYND10	100	100	100	100	Ciliary dyskinesia, primary, 22, 615444
ZNF142	100	99,7	100	100	Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425
ZNF335	100	99,7	100	100	Microcephaly 10, primary, autosomal recessive, 615095
ZNF341	97,3	95,9	100	100	Hyper-IgE recurrent infection syndrome 3, autosomal recessive, 618282
ZNF408	100	100	100	100	Retinitis pigmentosa 72, 616469 ?Exudative vitreoretinopathy 6, 616468
ZNF423	100	100	100	100	Nephronophthisis 14, 614844 Joubert syndrome 19, 614844
ZNF469	100	100	100	100	Brittle cornea syndrome 1, 229200
ZNF513	100	100	100	100	?Retinitis pigmentosa 58, 613617
ZNHIT3	74,4	74,4	75,7	74,4	PEHO syndrome, 260565
ZP1	100	100	100	100	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.
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.
Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.
TWIST is the chemistry used for (in-house) TURBO/RAPID WES analysis.
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 coverage denoting NC are non-protein coding genes.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : September 16th , 2021.

This list is accurate for panel version DG 3.2.0

Ad 1. “No OMIM Disease ID” signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} s ignify risk factors