PRECONCEPTION SCREENING GENE PANEL DG 2.18 (2337 genes)

Releasedate: 20-04-2020

Gene	Agilent V5 covered >10x	Agilent V5 covered > 20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype description and OMIM disease ID
AAAS	100%	99,90%	100%	100%	Achalasia-addisonianism-alacrimia syndrome, 231550
AARS	100%	99,90%	100%	100%	Epileptic encephalopathy, early infantile, 29, 616339 Charcot-Marie-Tooth disease, axonal, type 2N, 613287
AARS2	100%	99,40%	100%	100%	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889
AASS	100%	99,70%	100%	100%	Hyperlysinemia, 238700
ABAT	100%	99,40%	100%	100%	GABA-transaminase deficiency, 613163
ABCA1	99,90%	99,10%	100%	100%	Tangier disease, 205400 HDL deficiency, familial, 1, 604091
ABCA12	99,50%	98,70%	100%	100%	Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500 Ichthyosis, congenital, autosomal recessive 4A, 601277
ABCA3	99,90%	99,30%	100%	100%	Surfactant metabolism dysfunction, pulmonary, 3, 610921
ABCA4	99,90%	99,30%	100%	100%	Retinal dystrophy, early-onset severe, 248200 Stargardt disease 1, 248200 Fundus flavimaculatus, 248200 Cone-rod dystrophy 3, 604116 Retinitis pigmentosa 19, 601718
ABCB11	100%	99,70%	100%	100%	Cholestasis, progressive familial intrahepatic 2, 601847 Cholestasis, benign recurrent intrahepatic, 2, 605479
ABCB4	99,90%	99,60%	100%	100%	Gallbladder disease 1, 600803 Cholestasis, intrahepatic, of pregnancy, 3, 614972 Cholestasis, progressive familial intrahepatic 3, 602347
ABCC2	100%	99,90%	100%	100%	Dubin-Johnson syndrome, 237500
ABCC6	93,60%	92,40%	100%	100%	Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850 Arterial calcification, generalized, of infancy, 2, 614473
ABCC8	100%	99,80%	100%	100%	Diabetes mellitus, permanent neonatal, 606176 Diabetes mellitus, noninsulin-dependent, 125853 Diabetes mellitus, transient neonatal 2, 610374 Hyperinsulinemic hypoglycemia, familial, 1, 256450 Hypoglycemia of infancy, leucine-sensitive, 240800
ABCD3	99,80%	97,70%	100%	100%	?Bile acid synthesis defect, congenital, 5, 616278
ABCD4	99,90%	98,60%	100%	100%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	100%	100%	100%	100%	Sitosterolemia 2, 618666
ABCG8	99,10%	97,30%	100%	100%	Sitosterolemia 1, 210250
ABHD12	98,70%	92,30%	100%	99,30%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ABHD5	100%	100%	100%	100%	Chanarin-Dorfman syndrome, 275630

ACACA	98,40%	98,10%	100%	100%	No OMIM disease ID
ACAD8	100%	100%	100%	100%	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	100%	99,90%	100%	100%	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADM	99,80%	99,00%	100%	100%	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	99,90%	98,20%	100%	100%	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	100%	99,20%	100%	100%	2-methylbutyrylglycinuria, 610006
ACADVL	99,40%	97,30%	100%	100%	VLCAD deficiency, 201475
ACAN	96,50%	92,70%	98,90%	98,70%	Short stature and advanced bone age, with or without early-onset osteoarthritis and/or osteochondritis dissecans, 165800 Spondyloepimetaphyseal dysplasia, aggrecan type, 612813 ?Spondyloepiphyseal dysplasia, Kimberley type, 608361
ACAT1	99,90%	97,50%	100%	100%	Alpha-methylacetoacetic aciduria, 203750
ACD	100%	99,90%	100%	100%	?Dyskeratosis congenita, autosomal dominant 6, 616553 ?Dyskeratosis congenita, autosomal recessive 7, 616553
ACE	99,90%	98,40%	100%	100%	Renal tubular dysgenesis, 267430
ACER3	99,80%	98,60%	100%	100%	?Leukodystrophy, progressive, early childhood-onset, 617762
ACO2	96,30%	90,30%	100%	100%	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559
ACOX1	100%	99,90%	100%	100%	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACOX2	100%	99,20%	100%	100%	Bile acid synthesis defect, congenital, 6, 617308
ACP5	99,80%	98,30%	100%	100%	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACSF3	100%	99,90%	100%	100%	Combined malonic and methylmalonic aciduria, 614265
ACTA1	99,60%	92,30%	100%	100%	Myopathy, actin, congenital, with cores, 161800 Nemaline myopathy 3, autosomal dominant or recessive, 161800 Myopathy, congenital, with fiber-type disproportion 1, 255310 Myopathy, actin, congenital, with excess of thin myofilaments, 161800 ?Myopathy, scapulohumeroperoneal, 616852
ACTL6B	100%	99,80%	100%	100%	Epileptic encephalopathy, early infantile, 76, 618468 Intellectual developmental disorder with severe speech and ambulation defects, 618470
ACY1	100%	98,80%	100%	100%	Aminoacylase 1 deficiency, 609924
ADA	100%	99,70%	100%	100%	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADA2	100%	99,00%	100%	100%	?Sneddon syndrome, 182410 Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688
ADAM17	99,90%	99,00%	100%	100%	?Inflammatory skin and bowel disease, neonatal, 1, 614328
ADAM22	99,90%	99,50%	100%	100%	?Epileptic encephalopathy, early infantile, 61, 617933
ADAM9	99,80%	99,10%	100%	100%	Cone-rod dystrophy 9, 612775
ADAMTS10	99,90%	98,50%	100%	100%	Weill-Marchesani syndrome 1, recessive, 277600

ADAMTS13	97,10%	93,80%	99,90%	99,50%	Thrombotic thrombocytopenic purpura, hereditary, 274150
ADAMTS17	92,80%	89,00%	97,60%	95,80%	Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTS18	100%	99,70%	100%	100%	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
ADAMTS2	99,00%	96,60%	98,00%	97,80%	Ehlers-Danlos syndrome, dermatosparaxis type, 225410
ADAMTS3	100%	100%	100%	100%	Hennekam lymphangiectasia-lymphedema syndrome 3, 618154
ADAMTSL2	97,10%	93,30%	99,80%	99,40%	Geleophysic dysplasia 1, 231050
ADAMTSL4	100%	99,20%	100%	100%	Ectopia lentis et pupillae, 225200 Ectopia lentis, isolated, autosomal recessive, 225100
ADAR	100%	99,80%	100%	100%	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
ADAT3	100%	99,70%	100%	100%	Mental retardation, autosomal recessive 36, 615286
ADCY1	95,20%	93,80%	98,50%	97,90%	?Deafness, autosomal recessive 44, 610154
ADCY6	100%	100%	100%	100%	?Lethal congenital contracture syndrome 8, 616287
ADD3	99,90%	99,50%	100%	100%	Cerebral palsy, spastic quadriplegic, 3, 617008
ADGRG1	100%	100%	100%	100%	Polymicrogyria, bilateral perisylvian, 615752 Polymicrogyria, bilateral frontoparietal, 606854
ADGRG6	99,90%	99,00%	100%	100%	Lethal congenital contracture syndrome 9, 616503
ADGRV1	99,60%	98,60%	100%	100%	Usher syndrome, type 2C, 605472 ?Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472
ADK	99,50%	95,80%	100%	100%	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADPRHL2	100%	99,80%	100%	100%	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
ADSL	99,20%	98,70%	100%	100%	Adenylosuccinase deficiency, 103050
ADSSL1	90,20%	87,50%	100%	100%	Myopathy, distal, 5, 617030
AEBP1	100%	100%	100%	100%	Ehlers-Danlos syndrome, classic-like, 2, 618000
AFG3L2	95,00%	91,10%	100%	99,90%	Spastic ataxia 5, autosomal recessive, 614487 Spinocerebellar ataxia 28, 610246
AGA	100%	100%	100%	100%	Aspartylglucosaminuria, 208400
AGBL5	99,90%	99,30%	100%	100%	Retinitis pigmentosa 75, 617023
AGK	99,90%	97,60%	100%	100%	Sengers syndrome, 212350 Cataract 38, autosomal recessive, 614691
AGL	100%	99,40%	100%	100%	Glycogen storage disease IIIb, 232400 Glycogen storage disease IIIa, 232400
AGPAT2	99,60%	96,10%	100%	100%	Lipodystrophy, congenital generalized, type 1, 608594
AGPS	99,30%	95,40%	100%	99,90%	Rhizomelic chondrodysplasia punctata, type 3, 600121
AGRN	96,90%	92,60%	100%	99,90%	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120
AGT	100%	100%	100%	100%	Renal tubular dysgenesis, 267430

AGTPBP1	96,00%	94,10%	100%	100%	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276
AGTR1	92,00%	91,80%	100%	100%	Renal tubular dysgenesis, 267430
AGXT	100%	100%	100%	100%	Hyperoxaluria, primary, type 1, 259900
AHCY	100%	99,20%	100%	100%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHI1	99,70%	97,90%	100%	100%	Joubert syndrome 3, 608629
AHR	100%	99,60%	100%	100%	?Retinitis pigmentosa 85, 618345
AHSG	99,90%	99,50%	100%	100%	?Alopecia-mental retardation syndrome 1, 203650
AICDA	100%	100%	100%	100%	Immunodeficiency with hyper-IgM, type 2, 605258
AIMP1	99,20%	94,50%	100%	99,90%	Leukodystrophy, hypomyelinating, 3, 260600
AIMP2	88,90%	86,00%	100%	100%	Leukodystrophy, hypomyelinating, 17, 618006
AIPL1	100%	99,80%	100%	100%	Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393 Cone-rod dystrophy, 604393
AIRE	100%	99,80%	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	98,80%	94,50%	100%	100%	Reticular dysgenesis, 267500
AKR1C2	94,90%	89,20%	100%	100%	46XY sex reversal 8, 614279
AKR1D1	100%	99,40%	100%	100%	Bile acid synthesis defect, congenital, 2, 235555
ALAD	99,30%	94,10%	100%	100%	Porphyria, acute hepatic, 612740
ALB	100%	99,40%	100%	100%	Analbuminemia, 616000
ALDH18A1	100%	99,90%	100%	100%	Cutis laxa, autosomal recessive, type IIIA, 219150 Cutis laxa, autosomal dominant 3, 616603 Spastic paraplegia 9B, autosomal recessive, 616586 Spastic paraplegia 9A, autosomal dominant, 601162
ALDH1A3	97,20%	94,50%	100%	99,90%	Microphthalmia, isolated 8, 615113
ALDH3A2	95,30%	94,60%	100%	100%	Sjogren-Larsson syndrome, 270200
ALDH4A1	100%	99,40%	100%	100%	Hyperprolinemia, type II, 239510
ALDH5A1	91,00%	81,50%	100%	100%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	100%	99,90%	100%	100%	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	94,40%	88,80%	100%	100%	Epilepsy, pyridoxine-dependent, 266100
ALDOA	75,50%	74,00%	100%	100%	Glycogen storage disease XII, 611881
ALDOB	100%	99,10%	100%	100%	Fructose intolerance, hereditary, 229600
ALG1	53,00%	45,80%	100%	100%	Congenital disorder of glycosylation, type Ik, 608540
ALG11	96,80%	96,80%	96,80%	96,80%	Congenital disorder of glycosylation, type Ip, 613661
ALG12	100%	100%	100%	100%	Congenital disorder of glycosylation, type Ig, 607143
ALG14	100%	99,90%	100%	100%	?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227

ALG2	100%	100%	100%	100%	Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 ?Congenital disorder of glycosylation, type Ii, 607906
ALG3	100%	99,70%	100%	100%	Congenital disorder of glycosylation, type Id, 601110
ALG6	98,60%	94,80%	100%	100%	Congenital disorder of glycosylation, type Ic, 603147
ALG8	97,20%	95,60%	96,60%	96,60%	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	100%	99,70%	100%	100%	Gillessen-Kaesbach-Nishimura syndrome, 263210 Congenital disorder of glycosylation, type Il, 608776
ALKBH8	99,80%	98,90%	100%	100%	Intellectual developmental disorder, autosomal recessive 71, 618504
ALMS1	99,80%	99,50%	100%	100%	Alstrom syndrome, 203800
ALOX12B	100%	100%	100%	100%	Ichthyosis, congenital, autosomal recessive 2, 242100
ALOXE3	100%	99,50%	100%	100%	Ichthyosis, congenital, autosomal recessive 3, 606545
ALPK3	97,80%	94,60%	100%	100%	Cardiomyopathy, familial hypertrophic 27, 618052
ALPL	100%	100%	100%	100%	Hypophosphatasia, adult, 146300 Odontohypophosphatasia, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500
ALS2	100%	99,90%	100%	100%	Primary lateral sclerosis, juvenile, 606353 Amyotrophic lateral sclerosis 2, juvenile, 205100 Spastic paralysis, infantile onset ascending, 607225
ALX1	99,70%	97,10%	100%	100%	Frontonasal dysplasia 3, 613456
ALX3	77,90%	73,30%	100%	100%	Frontonasal dysplasia 1, 136760
ALX4	100%	99,30%	100%	100%	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597
AMACR	100%	100%	100%	100%	Bile acid synthesis defect, congenital, 4, 214950 Alpha-methylacyl-CoA racemase deficiency, 614307
AMBN	99,80%	98,50%	100%	100%	Amelogenesis imperfecta, type IF, 616270
AMN	89,70%	80,00%	100%	100%	Megaloblastic anemia-1, Norwegian type, 261100
AMPD1	99,90%	98,60%	100%	100%	Myopathy due to myoadenylate deaminase deficiency, 615511
AMPD2	99,80%	98,90%	100%	100%	?Spastic paraplegia 63, 615686 Pontocerebellar hypoplasia, type 9, 615809
AMT	100%	100%	100%	100%	Glycine encephalopathy, 605899
ANAPC1	59,40%	57,70%	100%	99,90%	Rothmund-Thomson syndrome, type 1, 618625
ANGPTL3	98,80%	95,40%	100%	100%	Hypobetalipoproteinemia, familial, 2, 605019
ANK1	100%	99,40%	100%	100%	Spherocytosis, type 1, 182900
ANK3	99,30%	99,00%	100%	100%	?Mental retardation, autosomal recessive, 37, 615493
ANKH	100%	100%	100%	100%	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000

ANKLE2	99,90%	98,60%	100%	99,80%	Microcephaly 16, primary, autosomal recessive, 616681
ANKS6	93,80%	89,50%	97,90%	95,80%	Nephronophthisis 16, 615382
ANO10	99,80%	97,90%	100%	100%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANO5	99,50%	97,30%	100%	100%	Miyoshi muscular dystrophy 3, 613319 Gnathodiaphyseal dysplasia, 166260 Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307
ANO6	99,90%	98,70%	100%	100%	Scott syndrome, 262890
ANTXR1	99,70%	97,90%	100%	100%	GAPO syndrome, 230740
ANTXR2	100%	98,20%	100%	100%	Hyaline fibromatosis syndrome, 228600
AP1S1	99,90%	99,50%	100%	100%	MEDNIK syndrome, 609313
AP3B1	99,20%	95,80%	100%	100%	Hermansky-Pudlak syndrome 2, 608233
AP3B2	99,40%	95,10%	100%	100%	Epileptic encephalopathy, early infantile, 48, 617276
AP3D1	99,80%	98,60%	100%	100%	?Hermansky-Pudlak syndrome 10, 617050
AP4B1	99,90%	98,70%	100%	100%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	99,80%	98,70%	100%	100%	Stuttering, familial persistent, 1, 184450 Spastic paraplegia 51, autosomal recessive, 613744
AP4M1	99,90%	98,90%	100%	100%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	78,90%	71,30%	87,90%	87,90%	Spastic paraplegia 52, autosomal recessive, 614067
AP5Z1	100%	99,80%	100%	100%	Spastic paraplegia 48, autosomal recessive, 613647
APC2	97,60%	92,70%	99,90%	99,10%	?Sotos syndrome 3, 617169 Cortical dysplasia, complex, with other brain malformations 10, 618677
APOB	99,80%	99,30%	100%	100%	Hypobetalipoproteinemia, 615558 Hypercholesterolemia, familial, 2, 144010
APOC2	100%	100%	100%	100%	Hyperlipoproteinemia, type Ib, 207750
APOE	98,90%	90,70%	100%	100%	Hyperlipoproteinemia, type III, 617347 Lipoprotein glomerulopathy, 611771 Sea-blue histiocyte disease, 269600 Alzheimer disease 2, 104310
APOPT1	81,90%	80,70%	93,50%	93,40%	Mitochondrial complex IV deficiency, 220110
APRT	100%	99,50%	100%	100%	Adenine phosphoribosyltransferase deficiency, 614723
APTX	94,90%	92,50%	100%	100%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
AQP2	100%	98,60%	100%	100%	Diabetes insipidus, nephrogenic, 125800
ARFGEF2	99,90%	99,10%	100%	100%	Periventricular heterotopia with microcephaly, 608097
ARG1	100%	100%	100%	100%	Argininemia, 207800
ARHGDIA	100%	100%	100%	100%	Nephrotic syndrome, type 8, 615244
ARHGEF18	95,40%	92,30%	100%	100%	Retinitis pigmentosa 78, 617433
ARHGEF2	100%	99,90%	100%	100%	?Neurodevelopmental disorder with midbrain and hindbrain malformations, 617523

ARL13B	100%	99,20%	100%	100%	Joubert syndrome 8, 612291
ARL2BP	95,90%	88,30%	100%	100%	Retinitis pigmentosa with or without situs inversus, 615434
ARL3	100%	98,40%	100%	100%	Joubert syndrome 35, 618161 Retinitis pigmentosa 83, 618173
ARL6	99,90%	98,60%	100%	100%	?Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151
ARL6IP1	99,40%	92,60%	100%	100%	?Spastic paraplegia 61, autosomal recessive, 615685
ARMC4	95,60%	93,50%	100%	100%	Ciliary dyskinesia, primary, 23, 615451
ARMC9	100%	99,80%	100%	100%	Joubert syndrome 30, 617622
ARNT2	100%	100%	100%	99,60%	?Webb-Dattani syndrome, 615926
ARPC1B	100%	100%	100%	100%	Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease, 617718
ARSA	100%	99,80%	100%	100%	Metachromatic leukodystrophy, 250100
ARSB	96,90%	88,30%	100%	100%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARSG	100%	99,50%	100%	100%	Usher syndrome, type IV, 618144
ARV1	100%	99,90%	100%	100%	Epileptic encephalopathy, early infantile, 38, 617020
ASAH1	99,70%	98,60%	100%	100%	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASCC1	97,70%	94,40%	91,10%	91,10%	Barrett esophagus/esophageal adenocarcinoma, 614266 Spinal muscular atrophy with congenital bone fractures 2, 616867
ASL	100%	99,60%	100%	100%	Argininosuccinic aciduria, 207900
ASNS	99,40%	95,20%	100%	100%	Asparagine synthetase deficiency, 615574
ASPA	99,90%	98,30%	100%	100%	Canavan disease, 271900
ASPH	99,90%	98,80%	100%	100%	Traboulsi syndrome, 601552
ASPM	99,70%	98,20%	100%	100%	Microcephaly 5, primary, autosomal recessive, 608716
ASS1	95,40%	87,90%	100%	100%	Citrullinemia, 215700
ATAD1	99,60%	95,10%	100%	100%	Hyperekplexia 4, 618011
ATAD3A	91,90%	83,20%	100%	100%	Harel-Yoon syndrome, 617183 ?Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810
ATCAY	100%	99,80%	100%	100%	Ataxia, cerebellar, Cayman type, 601238
ATF6	100%	99,90%	100%	100%	Achromatopsia 7, 616517
ATG5	99,40%	97,80%	100%	100%	?Spinocerebellar ataxia, autosomal recessive 25, 617584
ATIC	99,90%	99,30%	100%	100%	AICA-ribosiduria due to ATIC deficiency, 608688
ATM	99,80%	98,10%	100%	100%	Ataxia-telangiectasia, 208900 Lymphoma, mantle cell, somatic, 0 Lymphoma, B-cell non-Hodgkin, somatic, 0 T-cell prolymphocytic leukemia, somatic, 0
ATOH7	96,00%	91,20%	99,10%	94,40%	Persistent hyperplastic primary vitreous, autosomal recessive, 221900

ATP13A2	100%	99,50%	100%	100%	Kufor-Rakeb syndrome, 606693 Spastic paraplegia 78, autosomal recessive, 617225
ATP2A1	100%	100%	100%	100%	Brody myopathy, 601003
ATP5A1	95,20%	87,60%	100%	100%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4, 615228 ?Combined oxidative phosphorylation deficiency 22, 616045
ATP5D	96,20%	89,30%	100%	100%	Mitochondrial complex V (ATP synthase) deficiency, 618120
ATP5E	100%	100%	100%	100%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053
ATP6V0A2	100%	99,50%	100%	100%	Wrinkly skin syndrome, 278250 Cutis laxa, autosomal recessive, type IIA, 219200
ATP6V0A4	100%	99,90%	100%	100%	Renal tubular acidosis, distal, autosomal recessive, 602722
ATP6V1A	99,90%	98,70%	100%	100%	Epileptic encephalopathy, infantile or early childhood, 3, 618012 Cutis laxa, autosomal recessive, type IID, 617403
ATP6V1B1	100%	100%	100%	100%	Renal tubular acidosis with deafness, 267300
ATP6V1E1	93,10%	88,30%	100%	100%	Cutis laxa, autosomal recessive, type IIC, 617402
ATP7B	99,90%	99,20%	100%	100%	Wilson disease, 277900
ATP8A2	100%	99,70%	100%	100%	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268
ATP8B1	96,50%	94,00%	100%	100%	Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, progressive familial intrahepatic 1, 211600 Cholestasis, benign recurrent intrahepatic, 243300
ATPAF2	100%	100%	100%	100%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
ATR	99,90%	99,40%	100%	100%	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
AUH	100%	99,80%	100%	100%	3-methylglutaconic aciduria, type I, 250950
AURKC	100%	99,20%	100%	100%	Spermatogenic failure 5, 243060
B2M	100%	100%	100%	100%	Immunodeficiency 43, 241600 ?Amyloidosis, familial visceral, 105200
B3GALNT2	93,80%	89,40%	92,50%	92,50%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B3GALT6	75,70%	69,70%	89,80%	81,60%	Al-Gazali syndrome, 609465 Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B3GAT3	99,90%	98,20%	94,80%	94,80%	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B3GLCT	99,60%	96,30%	99,90%	99,20%	Peters-plus syndrome, 261540
B4GALNT1	99,30%	95,00%	100%	100%	Spastic paraplegia 26, autosomal recessive, 609195
B4GALT1	100%	99,80%	100%	100%	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	99,80%	97,40%	99,90%	98,60%	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	92,20%	92,00%	100%	100%	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120
B9D2	100%	100%	100%	100%	Joubert syndrome 34, 614175 ?Meckel syndrome 10, 614175
BAAT	99,80%	98,40%	100%	100%	Hypercholanemia, familial, 607748
BANF1	98,30%	86,60%	100%	100%	Nestor-Guillermo progeria syndrome, 614008
BBIP1	98,60%	92,40%	100%	100%	?Bardet-Biedl syndrome 18, 615995
BBS1	100%	100%	100%	100%	Bardet-Biedl syndrome 1, 209900
BBS10	100%	99,80%	100%	100%	Bardet-Biedl syndrome 10, 615987
BBS12	100%	100%	100%	100%	Bardet-Biedl syndrome 12, 615989
BBS2	100%	99,50%	100%	100%	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	99,90%	99,30%	100%	100%	Bardet-Biedl syndrome 4, 615982
BBS5	99,00%	93,90%	100%	100%	Bardet-Biedl syndrome 5, 615983
BBS7	98,70%	95,50%	100%	100%	Bardet-Biedl syndrome 7, 615984
BBS9	99,70%	97,60%	100%	100%	Bardet-Biedl syndrome 9, 615986
BCKDHA	99,90%	99,20%	100%	100%	Maple syrup urine disease, type Ia, 248600
BCKDHB	99,50%	94,40%	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%	Leigh syndrome, 256000 GRACILE syndrome, 603358 Bjornstad syndrome, 262000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BFSP1	99,00%	89,90%	100%	99,90%	Cataract 33, multiple types, 611391
BFSP2	99,80%	97,60%	100%	100%	Cataract 12, multiple types, 611597
BHLHA9	70,90%	50,40%	99,80%	97,30%	Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432 ?Camptosynpolydactyly, complex, 607539
BIN1	99,60%	95,70%	100%	100%	Centronuclear myopathy 2, 255200
BLM	99,80%	98,30%	100%	100%	Bloom syndrome, 210900
BLNK	97,10%	95,50%	100%	100%	?Agammaglobulinemia 4, 613502
BLOC1S3	98,50%	81,30%	100%	100%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	99,90%	97,10%	94,90%	94,90%	?Hermansky-pudlak syndrome 9, 614171
BLVRA	100%	99,40%	100%	100%	Hyperbiliverdinemia, 614156
BMP1	100%	100%	100%	100%	Osteogenesis imperfecta, type XIII, 614856
BMPER	100%	99,80%	100%	100%	Diaphanospondylodysostosis, 608022

BMPR1B	100%	99,90%	100%	100%	Brachydactyly, type A2, 112600 Brachydactyly, type A1, D, 616849 Acromesomelic dysplasia, Demirhan type, 609441
BOLA3	99,40%	90,20%	100%	100%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BPGM	100%	100%	100%	100%	Erythrocytosis, familial, 8, 222800
BRAT1	99,70%	98,20%	100%	100%	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498 Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056
BRCA1	99,40%	98,80%	100%	100%	Fanconi anemia, complementation group S, 617883
BRCA2	99,80%	98,50%	100%	100%	Wilms tumor, 194070 Fanconi anemia, complementation group D1, 605724
BRF1	99,90%	98,40%	100%	100%	Cerebellofaciodental syndrome, 616202
BRIP1	99,90%	99,00%	100%	100%	Fanconi anemia, complementation group J, 609054
BSCL2	100%	100%	100%	100%	Lipodystrophy, congenital generalized, type 2, 269700 Silver spastic paraplegia syndrome, 270685 Neuropathy, distal hereditary motor, type VA, 600794 Encephalopathy, progressive, with or without lipodystrophy, 615924
BSND	100%	100%	100%	100%	Bartter syndrome, type 4a, 602522 Sensorineural deafness with mild renal dysfunction, 602522
BTD	100%	99,90%	100%	100%	Biotinidase deficiency, 253260
BUB1B	99,60%	98,90%	100%	100%	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300
BVES	99,90%	98,80%	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 25, 616812
C12orf4	100%	99,30%	100%	100%	Mental retardation, autosomal recessive 66, 618221
C12orf57	100%	98,90%	100%	100%	Temtamy syndrome, 218340
C12orf65	99,80%	98,50%	100%	100%	Spastic paraplegia 55, autosomal recessive, 615035 Combined oxidative phosphorylation deficiency 7, 613559
C15orf41	100%	99,80%	96,30%	96,30%	Dyserythropoietic anemia, congenital, type Ib, 615631
C19orf12	100%	99,80%	100%	100%	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
C19orf70	100%	99,70%	100%	99,70%	Combined oxidative phosphorylation deficiency 37, 618329
C1QA	100%	100%	100%	100%	C1q deficiency, 613652
C1QB	100%	100%	100%	100%	C1q deficiency, 613652
C1QBP	86,90%	77,30%	100%	100%	Combined oxidative phosphorylation deficiency 33, 617713
C1QC	100%	99,20%	100%	100%	C1q deficiency, 613652
C1S	99,90%	99,00%	99,50%	97,70%	Ehlers-Danlos syndrome, periodontal type, 2, 617174 C1s deficiency, 613783
C2	100%	100%	100%	100%	C2 deficiency, 217000

C21orf2	100%	99,30%	100%	100%	Spondylometaphyseal dysplasia, axial, 602271 Retinal dystrophy with macular staphyloma, 617547
C21orf59	100%	99,70%	100%	100%	Ciliary dyskinesia, primary, 26, 615500
C2CD3	95,80%	95,60%	95,90%	95,90%	Orofaciodigital syndrome XIV, 615948
C2orf71	99,60%	98,50%	100%	100%	Retinitis pigmentosa 54, 613428
C3	99,90%	99,20%	100%	100%	C3 deficiency, 613779
C4A	98,40%	95,10%	99,60%	99,20%	C4a deficiency, 614380
C4B	99,20%	96,90%	99,90%	99,80%	C4B deficiency, 614379
C4orf26	100%	100%	100%	100%	Amelogenesis imperfecta, type IIA4, 614832
C5	99,90%	98,50%	100%	100%	C5 deficiency, 609536
C5orf42	99,70%	98,40%	100%	100%	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
C7orf43	100%	99,40%	100%	100%	?Microcephaly 25, primary, autosomal recessive, 618351
C8A	100%	99,60%	100%	100%	C8 deficiency, type I, 613790
C8B	100%	99,20%	100%	100%	C8 deficiency, type II, 613789
C8orf37	100%	99,40%	100%	100%	Retinitis pigmentosa 64, 614500 Bardet-Biedl syndrome 21, 617406 Cone-rod dystrophy 16, 614500
C9	99,90%	99,50%	100%	100%	C9 deficiency, 613825
CA12	100%	100%	100%	100%	Hyperchlorhidrosis, isolated, 143860
CA2	100%	100%	100%	100%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA5A	99,70%	97,10%	100%	100%	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CA8	99,60%	97,30%	100%	100%	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CABP2	75,90%	68,00%	100%	100%	Deafness, autosomal recessive 93, 614899
CABP4	100%	99,90%	100%	100%	Cone-rod synaptic disorder, congenital nonprogressive, 610427
CACNA1B	97,50%	95,70%	99,10%	97,70%	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497
CACNA1D	98,00%	97,90%	100%	100%	Sinoatrial node dysfunction and deafness, 614896 Primary aldosteronism, seizures, and neurologic abnormalities, 615474
CACNA2D2	94,00%	93,20%	99,20%	97,60%	Cerebellar atrophy with seizures and variable developmental delay, 618501
CACNA2D4	98,90%	97,70%	100%	100%	Retinal cone dystrophy 4, 610478
CAD	100%	99,20%	100%	100%	Epileptic encephalopathy, early infantile, 50, 616457
CAMK2A	100%	99,60%	100%	100%	?Mental retardation, autosomal recessive 63, 618095 Mental retardation, autosomal dominant 53, 617798
CANT1	100%	99,90%	100%	100%	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719
CAPN1	100%	100%	100%	100%	Spastic paraplegia 76, autosomal recessive, 616907
CAPN10	100%	99,60%	100%	100%	No OMIM disease ID

CAPN3	100%	99,30%	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600 Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129
CARD11	100%	99,90%	100%	100%	Immunodeficiency 11B with atopic dermatitis, 617638 B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11A, 615206
CARD9	99,90%	98,40%	100%	100%	Candidiasis, familial, 2, autosomal recessive, 212050
CARS2	100%	100%	100%	99,20%	Combined oxidative phosphorylation deficiency 27, 616672
CASP14	100%	100%	100%	100%	Ichthyosis, congenital, autosomal recessive 12, 617320
CASP8	95,60%	95,40%	95,60%	95,60%	?Autoimmune lymphoproliferative syndrome, type IIB, 607271 Hepatocellular carcinoma, somatic, 114550
CASQ2	100%	100%	100%	100%	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CASR	100%	99,90%	100%	100%	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 Hypocalciuric hypercalcemia, type I, 145980 Hypocalcemia, autosomal dominant, 601198 Hyperparathyroidism, neonatal, 239200
CAST	98,30%	95,40%	100%	100%	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295
CAT	100%	100%	100%	100%	Acatalasemia, 614097
CATSPER1	100%	100%	100%	100%	Spermatogenic failure 7, 612997
CAV1	100%	100%	100%	100%	Pulmonary hypertension, primary, 3, 615343 Lipodystrophy, familial partial, type 7, 606721 ?Lipodystrophy, congenital generalized, type 3, 612526
CAVIN1	100%	100%	100%	100%	Lipodystrophy, congenital generalized, type 4, 613327
CBS	99,80%	98,30%	100%	100%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CBX2	100%	99,80%	100%	100%	?46XY sex reversal 5, 613080
CC2D1A	100%	99,30%	100%	100%	Mental retardation, autosomal recessive 3, 608443
CC2D2A	99,70%	97,70%	98,20%	98,20%	Meckel syndrome 6, 612284 Joubert syndrome 9, 612285 COACH syndrome, 216360
CCBE1	99,80%	98,80%	100%	100%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CCDC103	100%	100%	100%	100%	Ciliary dyskinesia, primary, 17, 614679
CCDC114	100%	100%	100%	100%	Ciliary dyskinesia, primary, 20, 615067
CCDC115	95,30%	90,00%	100%	100%	Congenital disorder of glycosylation, type IIo, 616828
CCDC151	100%	99,70%	100%	100%	Ciliary dyskinesia, primary, 30, 616037
CCDC174	99,50%	97,10%	100%	100%	Hypotonia, infantile, with psychomotor retardation, 616816
CCDC39	99,50%	96,50%	100%	100%	Ciliary dyskinesia, primary, 14, 613807
CCDC40	99,10%	98,10%	100%	100%	Ciliary dyskinesia, primary, 15, 613808
CCDC47	99,40%	97,50%	100%	100%	Trichohepatoneurodevelopmental syndrome, 618268

CCDC65	99,60%	97,10%	100%	100%	Ciliary dyskinesia, primary, 27, 615504
CCDC8	100%	100%	100%	100%	3-M syndrome 3, 614205
CCDC88A	98,90%	95,40%	100%	100%	?PEHO syndrome-like, 617507
CCDC88C	100%	99,30%	100%	100%	?Spinocerebellar ataxia 40, 616053 Hydrocephalus, congenital, 1, 236600
CCNO	100%	99,20%	100%	100%	Ciliary dyskinesia, primary, 29, 615872
CCT5	100%	99,70%	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%	100%	100%	100%	?Immunodeficiency 25, 610163
CD27	99,90%	96,90%	100%	100%	Lymphoproliferative syndrome 2, 615122
CD2AP	99,90%	98,80%	100%	100%	Glomerulosclerosis, focal segmental, 3, 607832
CD320	100%	99,80%	100%	100%	Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646
CD3D	100%	100%	100%	100%	Immunodeficiency 19, 615617
CD3E	100%	99,50%	100%	100%	Immunodeficiency 18, SCID variant, 615615 Immunodeficiency 18, 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	92,20%	84,30%	99,20%	97,10%	Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300
CD59	95,10%	86,60%	79,50%	79,50%	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%	99,90%	100%	100%	Immunodeficiency, common variable, 6, 613496
CD8A	100%	99,80%	100%	100%	CD8 deficiency, familial, 608957
CDAN1	100%	99,60%	100%	100%	Dyserythropoietic anemia, congenital, type Ia, 224120
CDC14A	100%	98,90%	100%	100%	Deafness, autosomal recessive 32, with or without immotile sperm, 608653
CDC45	99,80%	98,50%	100%	100%	Meier-Gorlin syndrome 7, 617063
CDC6	100%	100%	100%	100%	?Meier-Gorlin syndrome 5, 613805
CDCA7	100%	99,60%	100%	100%	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910
CDH11	100%	100%	100%	100%	Elsahy-Waters syndrome, 211380
CDH23	100%	100%	100%	100%	Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1D, 601067
CDH3	100%	99,50%	100%	100%	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 Hypotrichosis, congenital, with juvenile macular dystrophy, 601553
CDHR1	99,20%	98,10%	100%	100%	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660

CDK10	100%	99,90%	100%	100%	Al Kaissi syndrome, 617694
CDK5	100%	100%	100%	100%	?Lissencephaly 7 with cerebellar hypoplasia, 616342
CDK5RAP2	99,80%	98,90%	100%	100%	Microcephaly 3, primary, autosomal recessive, 604804
CDK6	100%	99,60%	100%	100%	?Microcephaly 12, primary, autosomal recessive, 616080
CDSN	100%	100%	100%	100%	Hypotrichosis 2, 146520 Peeling skin syndrome 1, 270300
CDT1	99,70%	97,50%	100%	99,10%	Meier-Gorlin syndrome 4, 613804
CEACAM16	100%	99,50%	100%	100%	Deafness, autosomal recessive 113, 618410 Deafness, autosomal dominant 4B, 614614
CEBPE	100%	100%	100%	100%	Specific granule deficiency, 245480
CENPE	98,20%	92,20%	100%	100%	?Microcephaly 13, primary, autosomal recessive, 616051
CENPF	99,80%	98,50%	100%	100%	Stromme syndrome, 243605
CENPJ	100%	99,60%	100%	100%	Microcephaly 6, primary, autosomal recessive, 608393 ?Seckel syndrome 4, 613676
CEP104	100%	99,20%	100%	100%	Joubert syndrome 25, 616781
CEP120	100%	99,50%	100%	100%	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP135	99,10%	93,60%	100%	100%	Microcephaly 8, primary, autosomal recessive, 614673
CEP152	99,70%	98,20%	100%	100%	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823
CEP164	99,90%	98,30%	100%	100%	Nephronophthisis 15, 614845
CEP19	100%	100%	100%	100%	Morbid obesity and spermatogenic failure, 615703
CEP290	96,10%	90,00%	100%	100%	?Bardet-Biedl syndrome 14, 615991 Leber congenital amaurosis 10, 611755 Senior-Loken syndrome 6, 610189 Meckel syndrome 4, 611134 Joubert syndrome 5, 610188
CEP41	99,80%	97,40%	100%	100%	Joubert syndrome 15, 614464
CEP55	100%	99,80%	100%	100%	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500
CEP57	99,20%	93,00%	100%	100%	Mosaic variegated aneuploidy syndrome 2, 614114
CEP63	99,30%	96,50%	100%	100%	?Seckel syndrome 6, 614728
CEP78	99,70%	97,60%	100%	100%	Cone-rod dystrophy and hearing loss, 617236
CEP83	99,80%	97,40%	100%	100%	Nephronophthisis 18, 615862
CEP89	96,00%	94,50%	100%	100%	No OMIM disease ID
CERKL	99,50%	96,90%	100%	100%	Retinitis pigmentosa 26, 608380
CERS1	75,40%	63,70%	94,20%	86,40%	?Epilepsy, progressive myoclonic, 8, 616230

CERS3	99,90%	98,90%	100%	100%	Ichthyosis, congenital, autosomal recessive 9, 615023
CFAP53	99,60%	97,40%	100%	100%	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFD	89,30%	83,70%	100%	100%	Complement factor D deficiency, 613912
CFH	99,90%	99,00%	100%	99,90%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814
CFI	99,20%	96,80%	100%	100%	Complement factor I deficiency, 610984
CFL2	100%	99,60%	100%	100%	Nemaline myopathy 7, autosomal recessive, 610687
CFTR	99,60%	97,90%	100%	100%	Cystic fibrosis, 219700 Congenital bilateral absence of vas deferens, 277180 Sweat chloride elevation without CF, 0
CHAT	93,50%	85,70%	100%	100%	Myasthenic syndrome, congenital, 6, presynaptic, 254210
CHKB	100%	99,70%	100%	100%	Muscular dystrophy, congenital, megaconial type, 602541
CHMP1A	100%	99,80%	100%	100%	Pontocerebellar hypoplasia, type 8, 614961
CHP1	98,50%	89,10%	100%	100%	?Spastic ataxia 9, autosomal recessive, 618438
CHRM3	100%	100%	100%	100%	?Prune belly syndrome, 100100
CHRNA1	94,70%	94,00%	100%	100%	Myasthenic syndrome, congenital, 1A, slow-channel, 601462 Myasthenic syndrome, congenital, 1B, fast-channel, 608930 Multiple pterygium syndrome, lethal type, 253290
CHRNB1	100%	99,40%	100%	100%	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314 Myasthenic syndrome, congenital, 2A, slow-channel, 616313
CHRND	99,70%	97,90%	100%	100%	?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323 ?Myasthenic syndrome, congenital, 3A, slow-channel, 616321 Myasthenic syndrome, congenital, 3B, fast-channel, 616322 Multiple pterygium syndrome, lethal type, 253290
CHRNE	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%	Escobar syndrome, 265000 Multiple pterygium syndrome, lethal type, 253290
CHST11	100%	100%	100%	100%	?Osteochondrodysplasia, brachydactyly, and overlapping malformed digits, 618167
CHST14	99,90%	98,90%	100%	100%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	100%	99,40%	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	97,20%	95,70%	99,70%	98,00%	Temtamy preaxial brachydactyly syndrome, 605282
CHUK	100%	99,10%	100%	100%	Cocoon syndrome, 613630
CIB2	99,70%	97,00%	100%	99,90%	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869

CIDEC	100%	97,90%	100%	100%	?Lipodystrophy, familial partial, type 5, 615238
CIITA	100%	99,50%	100%	100%	Bare lymphocyte syndrome, type II, complementation group A, 209920
CISD2	83,40%	83,40%	100%	100%	Wolfram syndrome 2, 604928
CIT	100%	99,40%	100%	100%	Microcephaly 17, primary, autosomal recessive, 617090
CKAP2L	99,70%	98,60%	100%	100%	Filippi syndrome, 272440
CLCF1	100%	99,40%	100%	100%	Cold-induced sweating syndrome 2, 610313
CLCN1	100%	99,20%	100%	100%	Myotonia congenita, dominant, 160800 Myotonia congenita, recessive, 255700 Myotonia levior, recessive, 0
CLCN2	100%	99,50%	100%	100%	Leukoencephalopathy with ataxia, 615651 Hyperaldosteronism, familial, type II, 605635
CLCN7	99,70%	98,40%	100%	100%	Osteopetrosis, autosomal recessive 4, 611490 Osteopetrosis, autosomal dominant 2, 166600 Hypopigmentation, organomegaly, and delayed myelination and development, 618541
CLCNKB	99,10%	95,90%	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%	99,70%	100%	100%	Deafness, autosomal recessive 29, 614035
CLDN16	100%	100%	100%	100%	Hypomagnesemia 3, renal, 248250
CLDN19	98,50%	93,10%	100%	100%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLIC5	100%	99,90%	100%	100%	?Deafness, autosomal recessive 103, 616042
CLIP1	100%	99,00%	100%	100%	No OMIM disease ID
CLMP	100%	99,60%	100%	100%	Congenital short bowel syndrome, 615237
CLN3	92,50%	91,80%	92,50%	92,50%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	99,40%	95,50%	100%	100%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	99,90%	97,10%	100%	100%	Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300 Ceroid lipofuscinosis, neuronal, 6, 601780
CLN8	83,50%	83,50%	100%	100%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLP1	100%	100%	100%	100%	Pontocerebellar hypoplasia, type 10, 615803
CLPB	100%	99,90%	100%	100%	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CLPP	100%	99,10%	100%	100%	Perrault syndrome 3, 614129
CLRN1	100%	99,80%	100%	100%	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902
CNGA1	91,70%	86,30%	91,00%	91,00%	Retinitis pigmentosa 49, 613756
CNGA3	100%	99,70%	100%	100%	Achromatopsia 2, 216900

CNGB1	99,40%	97,50%	100%	100%	Retinitis pigmentosa 45, 613767
CNGB3	99,40%	95,90%	100%	100%	Macular degeneration, juvenile, 248200 Achromatopsia 3, 262300
CNNM2	100%	100%	100%	100%	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNNM4	99,80%	98,90%	99,70%	98,80%	Jalili syndrome, 217080
CNPY3	100%	99,30%	100%	100%	Epileptic encephalopathy, early infantile, 60, 617929
CNTN1	99,90%	98,90%	100%	100%	?Myopathy, congenital, Compton-North, 612540
CNTN2	92,70%	92,70%	100%	100%	?Epilepsy, myoclonic, familial adult, 5, 615400
CNTNAP1	100%	99,80%	100%	100%	Lethal congenital contracture syndrome 7, 616286 Hypomyelinating neuropathy, congenital, 3, 618186
CNTNAP2	100%	99,80%	100%	100%	Pitt-Hopkins like syndrome 1, 610042 Cortical dysplasia-focal epilepsy syndrome, 610042
COA5	99,10%	88,90%	85,20%	85,20%	?Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 3, 616500
COA6	99,90%	98,40%	100%	100%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 4, 616501
COA7	100%	100%	100%	100%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387
COASY	100%	100%	100%	100%	Pontocerebellar hypoplasia, type 12, 618266 Neurodegeneration with brain iron accumulation 6, 615643
COCH	100%	99,90%	100%	100%	?Deafness, autosomal recessive 110, 618094 Deafness, autosomal dominant 9, 601369
COG1	100%	100%	100%	100%	Congenital disorder of glycosylation, type IIg, 611209
COG2	99,90%	98,50%	100%	100%	?Congenital disorder of glycosylation, type IIq, 617395
COG4	100%	99,90%	100%	100%	Saul-Wilson syndrome, 618150 Congenital disorder of glycosylation, type IIj, 613489
COG5	99,70%	97,60%	100%	100%	Congenital disorder of glycosylation, type IIi, 613612
COG6	99,10%	93,90%	100%	100%	Shaheen syndrome, 615328 Congenital disorder of glycosylation, type III, 614576
COG7	100%	100%	100%	100%	Congenital disorder of glycosylation, type IIe, 608779
COG8	100%	99,60%	100%	100%	Congenital disorder of glycosylation, type IIh, 611182
COL11A1	99,20%	95,70%	100%	100%	Stickler syndrome, type II, 604841 Marshall syndrome, 154780 ?Deafness, autosomal dominant 37, 618533 Fibrochondrogenesis 1, 228520
COL11A2	100%	99,50%	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	100%	99,40%	100%	100%	Bethlem myopathy 2, 616471 ?Ullrich congenital muscular dystrophy 2, 616470
COL13A1	100%	99,80%	100%	100%	Myasthenic syndrome, congenital, 19, 616720
COL17A1	98,70%	96,80%	100%	100%	Epithelial recurrent erosion dystrophy, 122400 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa, junctional, localisata variant, 226650
COL18A1	98,10%	95,60%	100%	100%	Knobloch syndrome, type 1, 267750
COL1A2	99,40%	97,00%	100%	100%	Ehlers-Danlos syndrome, cardiac valvular type, 225320 Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type IV, 166220 Osteogenesis imperfecta, type III, 259420
COL25A1	99,50%	99,10%	99,90%	99,90%	Fibrosis of extraocular muscles, congenital, 5, 616219
COL27A1	99,90%	99,70%	100%	100%	Steel syndrome, 615155
COL3A1	99,60%	97,60%	100%	100%	Ehlers-Danlos syndrome, vascular type, 130050 Polymicrogyria with or without vascular-type EDS, 618343
COL4A3	98,70%	98,00%	100%	100%	Hematuria, benign familial, 141200 Alport syndrome 2, autosomal recessive, 203780 Alport syndrome 3, autosomal dominant, 104200
COL4A4	99,90%	98,20%	100%	100%	Alport syndrome 2, autosomal recessive, 203780 Hematuria, familial benign, 141200
COL6A1	100%	99,40%	100%	100%	Ullrich congenital muscular dystrophy 1, 254090 Bethlem myopathy 1, 158810
COL6A2	100%	99,80%	100%	100%	Bethlem myopathy 1, 158810 ?Myosclerosis, congenital, 255600 Ullrich congenital muscular dystrophy 1, 254090
COL6A3	100%	99,80%	100%	100%	Bethlem myopathy 1, 158810 Dystonia 27, 616411 Ullrich congenital muscular dystrophy 1, 254090
COL7A1	99,90%	99,10%	100%	100%	EBD inversa, 226600 Epidermolysis bullosa dystrophica, AR, 226600 Toenail dystrophy, isolated, 607523 EBD, Bart type, 132000 Transient bullous of the newborn, 131705 Epidermolysis bullosa dystrophica, AD, 131750 Epidermolysis bullosa pruriginosa, 604129 Epidermolysis bullosa, pretibial, 131850 EBD, localisata variant, 0

COL9A1	100%	99,20%	100%	100%	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	99,90%	99,00%	100%	100%	?Stickler syndrome, type V, 614284 Epiphyseal dysplasia, multiple, 2, 600204
COLEC10	100%	100%	100%	100%	3MC syndrome 3, 248340
COLEC11	100%	100%	100%	100%	3MC syndrome 2, 265050
COLGALT1	93,30%	89,00%	98,60%	97,00%	Brain small vessel disease 3, 618360
COLQ	100%	99,20%	100%	100%	Myasthenic syndrome, congenital, 5, 603034
COPB2	99,90%	99,30%	100%	100%	?Microcephaly 19, primary, autosomal recessive, 617800
COQ2	98,00%	95,30%	97,20%	97,20%	Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	90,90%	89,30%	100%	100%	Coenzyme Q10 deficiency, primary, 7, 616276
COQ6	99,90%	98,40%	100%	100%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	100%	99,80%	100%	100%	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ8A	100%	99,50%	100%	100%	Coenzyme Q10 deficiency, primary, 4, 612016
COQ8B	100%	99,30%	100%	100%	Nephrotic syndrome, type 9, 615573
COQ9	100%	97,90%	100%	100%	Coenzyme Q10 deficiency, primary, 5, 614654
CORO1A	100%	98,60%	100%	100%	Immunodeficiency 8, 615401
COX10	100%	100%	100%	100%	Mitochondrial complex IV deficiency, 220110 Leigh syndrome due to mitochondrial COX4 deficiency, 256000
COX14	100%	100%	100%	100%	?Mitochondrial complex IV deficiency, 220110
COX15	99,90%	98,80%	100%	100%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000
COX20	97,80%	88,30%	100%	100%	Mitochondrial complex IV deficiency, 220110
COX4I2	100%	100%	100%	100%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX6A1	100%	99,50%	100%	100%	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
COX6A2	99,20%	93,70%	100%	100%	Mitochondrial complex IV deficiency, 220110
COX6B1	100%	100%	100%	100%	Mitochondrial complex IV deficiency, 220110
COX8A	100%	100%	100%	100%	?Mitochondrial complex IV deficiency, 220110
CP	94,80%	88,90%	100%	100%	Hemosiderosis, systemic, due to aceruloplasminemia, 604290 Cerebellar ataxia, 604290
CPA6	99,60%	97,50%	100%	100%	Febrile seizures, familial, 11, 614418 Epilepsy, familial temporal lobe, 5, 614417
CPAMD8	95,80%	92,80%	99,90%	99,60%	Anterior segment dysgenesis 8, 617319
CPLX1	100%	100%	100%	100%	Epileptic encephalopathy, early infantile, 63, 617976
CPN1	99,90%	99,40%	100%	100%	Carboxypeptidase N deficiency, 212070
CPOX	99,90%	95,40%	100%	100%	Harderoporphyria, 121300 Coproporphyria, 121300

CPS1	100%	99,90%	100%	100%	Carbamoylphosphate synthetase I deficiency, 237300
CPT1A	100%	98,90%	100%	100%	CPT deficiency, hepatic, type IA, 255120
CPT2	98,20%	97,80%	100%	100%	CPT II deficiency, myopathic, stress-induced, 255110 CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836
CR2	100%	99,80%	100%	100%	Immunodeficiency, common variable, 7, 614699
CRADD	99,50%	96,30%	100%	100%	Mental retardation, autosomal recessive 34, with variant lissencephaly, 614499
CRAT	100%	99,80%	100%	100%	?Neurodegeneration with brain iron accumulation 8, 617917
CRB1	100%	99,90%	100%	100%	Pigmented paravenous chorioretinal atrophy, 172870 Retinitis pigmentosa-12, 600105 Leber congenital amaurosis 8, 613835
CRB2	98,50%	93,00%	100%	100%	Focal segmental glomerulosclerosis 9, 616220 Ventriculomegaly with cystic kidney disease, 219730
CRBN	88,20%	87,70%	97,00%	92,90%	Mental retardation, autosomal recessive 2, 607417
CREB3L1	100%	99,90%	100%	100%	Osteogenesis imperfecta, type XVI, 616229
CRIPT	98,10%	93,20%	100%	100%	Short stature with microcephaly and distinctive facies, 615789
CRLF1	91,00%	89,80%	97,90%	95,20%	Cold-induced sweating syndrome 1, 272430
CRTAP	99,80%	98,80%	100%	100%	Osteogenesis imperfecta, type VII, 610682
CRYAA	99,90%	97,50%	100%	100%	Cataract 9, multiple types, 604219
CRYAB	100%	99,20%	100%	100%	Myopathy, myofibrillar, 2, 608810 Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869
CRYBB1	100%	100%	100%	100%	Cataract 17, multiple types, 611544
CRYBB3	100%	100%	100%	100%	Cataract 22, 609741
CSF1R	99,90%	99,30%	100%	100%	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
CSF2RB	100%	99,00%	100%	100%	Surfactant metabolism dysfunction, pulmonary, 5, 614370
CSPP1	99,80%	98,70%	100%	100%	Joubert syndrome 21, 615636
CSTA	100%	99,80%	100%	100%	Peeling skin syndrome 4, 607936
CSTB	99,60%	89,80%	100%	100%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTC1	100%	99,60%	100%	100%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTDP1	88,40%	84,30%	100%	99,40%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTNNA2	100%	99,80%	100%	100%	Cortical dysplasia, complex, with other brain malformations 9, 618174
CTNS	100%	99,80%	100%	100%	Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, atypical nephropathic, 219800

CTPS1	100%	100%	100%	100%	Immunodeficiency 24, 615897
CTSA	100%	100%	100%	100%	Galactosialidosis, 256540
CTSC	100%	100%	100%	100%	Periodontitis 1, juvenile, 170650 Papillon-Lefevre syndrome, 245000 Haim-Munk syndrome, 245010
CTSD	98,40%	95,00%	100%	100%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	84,00%	79,30%	100%	99,90%	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362
CTSK	100%	99,90%	100%	100%	Pycnodysostosis, 265800
CTU2	99,70%	97,70%	100%	100%	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142
CUBN	99,70%	98,30%	100%	100%	Megaloblastic anemia-1, Finnish type, 261100
CUL7	100%	99,30%	100%	100%	3-M syndrome 1, 273750
CWC27	99,30%	96,50%	100%	100%	Retinitis pigmentosa with or without skeletal anomalies, 250410
CWF19L1	100%	99,80%	100%	100%	Spinocerebellar ataxia, autosomal recessive 17, 616127
CYB5A	100%	100%	100%	100%	Methemoglobinemia and ambiguous genitalia, 250790
CYB5R3	98,40%	98,00%	99,80%	98,90%	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYBA	95,00%	82,40%	100%	100%	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690
CYC1	97,50%	89,20%	99,90%	98,70%	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYP11A1	99,30%	96,10%	100%	100%	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	100%	100%	100%	100%	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 Aldosteronism, glucocorticoid-remediable, 103900
CYP11B2	100%	100%	100%	100%	Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 Aldosterone to renin ratio raised, 0
CYP17A1	100%	99,50%	100%	100%	17-alpha-hydroxylase/17,20-lyase deficiency, 202110 17,20-lyase deficiency, isolated, 202110
CYP19A1	98,80%	96,80%	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
CYP24A1	100%	99,90%	100%	100%	Hypercalcemia, infantile, 1, 143880
CYP26B1	100%	99,90%	100%	100%	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
CYP26C1	99,70%	97,10%	100%	99,80%	Focal facial dermal dysplasia 4, 614974
CYP27A1	98,90%	96,70%	100%	100%	Cerebrotendinous xanthomatosis, 213700
CYP27B1	99,90%	99,30%	100%	100%	Vitamin D-dependent rickets, type I, 264700
CYP2C8	99,90%	98,60%	100%	100%	No OMIM disease ID
CYP2R1	99,40%	95,60%	100%	100%	Rickets due to defect in vitamin D 25-hydroxylation, 600081

CYP2U1	94,80%	91,50%	100%	99,90%	Spastic paraplegia 56, autosomal recessive, 615030
CYP4F22	100%	99,40%	100%	100%	Ichthyosis, congenital, autosomal recessive 5, 604777
CYP4V2	99,90%	98,40%	100%	100%	Bietti crystalline corneoretinal dystrophy, 210370
CYP7B1	98,00%	92,80%	100%	100%	Spastic paraplegia 5A, autosomal recessive, 270800 Bile acid synthesis defect, congenital, 3, 613812
D2HGDH	99,20%	97,20%	100%	100%	D-2-hydroxyglutaric aciduria, 600721
DAG1	100%	100%	100%	100%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538
DARS	100%	99,30%	100%	100%	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2	100%	99,30%	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	99,80%	98,00%	100%	100%	Maple syrup urine disease, type II, 248600
DCAF17	98,90%	93,30%	100%	100%	Woodhouse-Sakati syndrome, 241080
DCC	100%	100%	100%	100%	Esophageal carcinoma, somatic, 133239 Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 Mirror movements 1 and/or agenesis of the corpus callosum, 157600 Colorectal cancer, somatic, 114500
DCDC2	100%	99,90%	100%	100%	Sclerosing cholangitis, neonatal, 617394 Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212
DCHS1	99,80%	99,10%	100%	100%	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390
DCLRE1C	100%	99,40%	100%	100%	Omenn syndrome, 603554 Severe combined immunodeficiency, Athabascan type, 602450
DCPS	100%	100%	100%	100%	Al-Raqad syndrome, 616459
DDB2	99,60%	97,50%	100%	100%	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DDC	99,70%	96,40%	100%	100%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	97,90%	95,80%	100%	100%	Spastic paraplegia 28, autosomal recessive, 609340
DDHD2	100%	99,60%	100%	100%	Spastic paraplegia 54, autosomal recessive, 615033
DDOST	100%	99,90%	100%	100%	?Congenital disorder of glycosylation, type Ir, 614507
DDR2	100%	99,90%	100%	100%	Warburg-Cinotti syndrome, 618175 Spondylometaepiphyseal dysplasia, short limb-hand type, 271665
DDRGK1	100%	99,90%	100%	100%	Spondyloepimetaphyseal dysplasia, Shohat type, 602557
DDX11	85,20%	80,70%	100%	100%	Warsaw breakage syndrome, 613398
DDX59	100%	100%	100%	100%	Orofaciodigital syndrome V, 174300
DEAF1	97,30%	88,80%	100%	98,70%	Mental retardation, autosomal dominant 24, 615828 ?Dyskinesia, seizures, and intellectual developmental disorder, 617171

DEGS1	100%	100%	100%	100%	Leukodystrophy, hypomyelinating, 18, 618404
DENND5A	100%	99,40%	100%	100%	Epileptic encephalopathy, early infantile, 49, 617281
DES	100%	99,70%	100%	100%	?Cardiomyopathy, dilated, 1I, 604765 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 Myopathy, myofibrillar, 1, 601419
DFNB59	100%	99,70%	100%	100%	Deafness, autosomal recessive 59, 610220
DGAT1	91,90%	87,60%	99,70%	98,60%	?Diarrhea 7, protein-losing enteropathy type, 615863
DGKE	99,80%	98,10%	100%	100%	Nephrotic syndrome, type 7, 615008
DGUOK	100%	99,40%	100%	100%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 Portal hypertension, noncirrhotic, 617068 Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHCR24	100%	100%	100%	100%	Desmosterolosis, 602398
DHCR7	100%	100%	100%	100%	Smith-Lemli-Opitz syndrome, 270400
DHDDS	99,00%	95,00%	95,20%	95,20%	Retinitis pigmentosa 59, 613861 ?Congenital disorder of glycosylation, type 1bb, 613861 Developmental delay and seizures with or without movement abnormalities, 617836
DHFR	92,10%	78,90%	100%	100%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHH	100%	100%	100%	100%	46XY sex reversal 7, 233420 46XY partial gonadal dysgenesis, with minifascicular neuropathy, 607080
DHODH	100%	100%	100%	100%	Miller syndrome, 263750
DHPS	100%	99,70%	93,30%	93,20%	Neurodevelopmental disorder with seizures and speech and walking impairment, 618480
DHTKD1	99,90%	98,90%	100%	100%	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DHX38	100%	99,30%	100%	100%	Retinitis pigmentosa 84, 618220
DIAPH1	99,80%	99,00%	99,50%	97,90%	Seizures, cortical blindness, microcephaly syndrome, 616632 Deafness, autosomal dominant 1, 124900
DIS3L2	100%	99,80%	100%	100%	Perlman syndrome, 267000
DLAT	100%	99,70%	100%	100%	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	100%	99,70%	100%	100%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLL3	92,10%	87,00%	100%	99,10%	Spondylocostal dysostosis 1, autosomal recessive, 277300
DLX5	100%	99,90%	100%	100%	?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600
DMGDH	100%	99,70%	100%	100%	Dimethylglycine dehydrogenase deficiency, 605850
DMP1	100%	99,90%	100%	100%	Hypophosphatemic rickets, AR, 241520
DMXL2	99,90%	99,10%	100%	100%	?Deafness, autosomal dominant 71, 617605 ?Polyendocrine-polyneuropathy syndrome, 616113 Epileptic encephalopathy, early infantile, 81, 618663
DNA2	99,80%	98,30%	100%	100%	?Seckel syndrome 8, 615807 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156

DNAAF1	100%	99,80%	100%	100%	Ciliary dyskinesia, primary, 13, 613193
DNAAF2	99,90%	98,90%	100%	100%	Ciliary dyskinesia, primary, 10, 612518
DNAAF3	99,50%	96,10%	100%	100%	Ciliary dyskinesia, primary, 2, 606763
DNAAF4	99,80%	97,00%	100%	100%	Ciliary dyskinesia, primary, 25, 615482
DNAAF5	84,60%	78,60%	99,10%	97,50%	Ciliary dyskinesia, primary, 18, 614874
DNAH1	100%	99,70%	100%	100%	?Ciliary dyskinesia, primary, 37, 617577 Spermatogenic failure 18, 617576
DNAH11	99,90%	99,00%	100%	100%	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH5	99,90%	99,30%	100%	100%	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAH9	99,50%	98,30%	100%	100%	Ciliary dyskinesia, primary, 40, 618300
DNAI1	100%	100%	100%	100%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	98,60%	96,20%	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	87,40%	87,40%	100%	100%	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC19	98,90%	96,20%	100%	100%	3-methylglutaconic aciduria, type V, 610198
DNAJC21	99,90%	99,00%	100%	100%	Bone marrow failure syndrome 3, 617052
DNAJC3	100%	99,70%	100%	100%	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192
DNAJC6	100%	99,40%	100%	100%	Parkinson disease 19b, early-onset, 615528 Parkinson disease 19a, juvenile-onset, 615528
DNAL1	99,00%	96,80%	100%	100%	Ciliary dyskinesia, primary, 16, 614017
DNASE1L3	100%	100%	100%	100%	Systemic lupus erythematosus 16, 614420
DNM1L	99,90%	98,50%	100%	100%	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 Optic atrophy 5, 610708
DNM2	98,10%	94,50%	100%	100%	Lethal congenital contracture syndrome 5, 615368 Charcot-Marie-Tooth disease, axonal type 2M, 606482 Centronuclear myopathy 1, 160150 Charcot-Marie-Tooth disease, dominant intermediate B, 606482
DNMBP	100%	99,70%	100%	100%	Cataract 48, 618415
DNMT3B	100%	100%	100%	100%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK2	100%	99,60%	100%	100%	Immunodeficiency 40, 616433
DOCK3	100%	99,00%	100%	100%	Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292
DOCK6	99,30%	98,90%	100%	100%	Adams-Oliver syndrome 2, 614219
DOCK7	99,80%	98,20%	100%	99,90%	Epileptic encephalopathy, early infantile, 23, 615859
DOCK8	100%	99,60%	100%	100%	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
DOK7	94,10%	91,30%	100%	100%	Myasthenic syndrome, congenital, 10, 254300 Fetal akinesia deformation sequence 3, 618389
DOLK	100%	100%	100%	100%	Congenital disorder of glycosylation, type Im, 610768

DONSON	91,70%	85,30%	100%	100%	Microcephaly-micromelia syndrome, 251230 Microcephaly, short stature, and limb abnormalities, 617604
DPAGT1	100%	100%	100%	100%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPH1	100%	99,90%	100%	100%	Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901
DPM1	98,20%	91,30%	99,70%	97,10%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	100%	98,70%	100%	100%	Congenital disorder of glycosylation, type Iu, 615042
DPM3	100%	100%	100%	100%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937
DPY19L2	74,50%	71,20%	100%	100%	Spermatogenic failure 9, 613958
DPYD	99,70%	97,70%	100%	100%	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270
DPYS	100%	99,90%	100%	100%	Dihydropyrimidinuria, 222748
DRAM2	100%	99,90%	100%	100%	Cone-rod dystrophy 21, 616502
DRC1	100%	99,50%	100%	100%	Ciliary dyskinesia, primary, 21, 615294
DSC2	99,80%	98,40%	100%	100%	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 Arrhythmogenic right ventricular dysplasia 11, 610476
DSC3	99,50%	96,80%	100%	100%	?Hypotrichosis and recurrent skin vesicles, 613102
DSE	99,00%	96,10%	100%	100%	Ehlers-Danlos syndrome, musculocontractural type 2, 615539
DSG4	100%	99,20%	100%	100%	Hypotrichosis 6, 607903
DSP	100%	99,60%	100%	100%	Keratosis palmoplantaris striata II, 612908 Skin fragility-woolly hair syndrome, 607655 Arrhythmogenic right ventricular dysplasia 8, 607450 Epidermolysis bullosa, lethal acantholytic, 609638 Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821
DST	99,90%	99,40%	100%	100%	?Neuropathy, hereditary sensory and autonomic, type VI, 614653 Epidermolysis bullosa simplex, autosomal recessive 2, 615425
DSTYK	99,90%	99,20%	100%	100%	Congenital anomalies of kidney and urinary tract 1, 610805 Spastic paraplegia 23, 270750
DTNBP1	99,80%	98,70%	100%	100%	Hermansky-Pudlak syndrome 7, 614076
DUOX2	96,70%	94,70%	100%	100%	Thyroid dyshormonogenesis 6, 607200
DUOXA2	100%	100%	100%	100%	Thyroid dyshormonogenesis 5, 274900
DYM	97,40%	96,50%	100%	100%	Smith-McCort dysplasia, 607326 Dyggve-Melchior-Clausen disease, 223800
DYNC1I2	84,40%	68,80%	100%	100%	Neurodevelopmental disorder with microcephaly and structural brain anomalies, 618492
DYNC2H1	98,80%	95,50%	100%	100%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYNC2LI1	99,70%	97,60%	100%	100%	Short-rib thoracic dysplasia 15 with polydactyly, 617088

DYSF	100%	99,90%	100%	100%	Miyoshi muscular dystrophy 1, 254130 Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601 Myopathy, distal, with anterior tibial onset, 606768
DZIP1L	99,90%	99,00%	100%	100%	Polycystic kidney disease 5, 617610
EARS2	99,80%	97,70%	100%	100%	Combined oxidative phosphorylation deficiency 12, 614924
ECEL1	95,40%	90,00%	100%	100%	Arthrogryposis, distal, type 5D, 615065
ECHS1	99,90%	99,00%	100%	100%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ECM1	100%	99,60%	100%	100%	Urbach-Wiethe disease, 247100
EDAR	100%	99,90%	100%	100%	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490 Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900
EDARADD	99,90%	98,80%	100%	100%	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941 Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940
EDC3	100%	99,90%	100%	100%	?Mental retardation, autosomal recessive 50, 616460
EDN1	100%	100%	100%	100%	Auriculocondylar syndrome 3, 615706 Question mark ears, isolated, 612798
EDN3	100%	99,90%	100%	100%	Waardenburg syndrome, type 4B, 613265 Central hypoventilation syndrome, congenital, 209880
EDNRB	98,00%	93,80%	100%	100%	Waardenburg syndrome, type 4A, 277580 ABCD syndrome, 600501
EFEMP2	100%	100%	100%	100%	Cutis laxa, autosomal recessive, type IB, 614437
EFL1	99,60%	98,50%	100%	100%	Shwachman-Diamond syndrome 2, 617941
EGF	99,90%	99,70%	100%	100%	Hypomagnesemia 4, renal, 611718
EGFR	100%	100%	100%	99,80%	?Inflammatory skin and bowel disease, neonatal, 2, 616069 Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980 Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980
EGR2	100%	100%	100%	100%	Dejerine-Sottas disease, 145900 Hypomyelinating neuropathy, congenital, 1, 605253 Charcot-Marie-Tooth disease, type 1D, 607678
EIF2AK3	97,20%	94,50%	100%	100%	Wolcott-Rallison syndrome, 226980
EIF2AK4	99,80%	98,60%	100%	100%	Pulmonary venoocclusive disease 2, 234810
EIF2B1	100%	100%	100%	100%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B2	99,90%	99,50%	100%	100%	Ovarioleukodystrophy, 603896 Leukoencephalopathy with vanishing white matter, 603896
EIF2B3	100%	100%	100%	100%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B4	100%	99,90%	100%	100%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B5	100%	99,00%	100%	100%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896

EIF3F	96,80%	84,10%	100%	100%	Mental retardation, autosomal recessive 67, 618295
EIF4A3	100%	99,50%	100%	100%	Robin sequence with cleft mandible and limb anomalies, 268305
ELAC2	100%	99,70%	100%	100%	Combined oxidative phosphorylation deficiency 17, 615440
ELMO2	99,90%	99,00%	100%	100%	Vascular malformation, primary intraosseous, 606893
ELMOD3	100%	99,80%	100%	100%	?Deafness, autosomal recessive 88, 615429
ELOVL4	100%	99,50%	100%	100%	Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
ELP1	99,80%	99,00%	100%	100%	Dysautonomia, familial, 223900
ELP2	99,90%	98,80%	100%	100%	Mental retardation, autosomal recessive 58, 617270
EMC1	100%	99,30%	100%	100%	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EMG1	100%	100%	100%	100%	Bowen-Conradi syndrome, 211180
EML1	99,70%	98,40%	100%	100%	Band heterotopia, 600348
EMP2	99,90%	96,70%	100%	100%	Nephrotic syndrome, type 10, 615861
ENAM	100%	100%	100%	100%	Amelogenesis imperfecta, type IC, 204650 Amelogenesis imperfecta, type IB, 104500
ENO3	100%	99,90%	100%	100%	?Glycogen storage disease XIII, 612932
ENPP1	96,40%	91,20%	98,70%	97,80%	Hypophosphatemic rickets, autosomal recessive, 2, 613312 Cole disease, 615522 Arterial calcification, generalized, of infancy, 1, 208000
ENTPD1	100%	100%	100%	100%	Spastic paraplegia 64, autosomal recessive, 615683
EOGT	79,40%	78,40%	91,90%	89,00%	Adams-Oliver syndrome 4, 615297
EPB41	99,40%	97,80%	100%	100%	Elliptocytosis-1, 611804
EPB42	100%	99,50%	100%	100%	Spherocytosis, type 5, 612690
EPCAM	98,60%	90,30%	99,80%	98,30%	Colorectal cancer, hereditary nonpolyposis, type 8, 613244 Diarrhea 5, with tufting enteropathy, congenital, 613217
EPG5	99,50%	98,50%	100%	100%	Vici syndrome, 242840
EPHX1	99,90%	98,80%	100%	100%	?Hypercholanemia, familial, 607748
EPM2A	94,20%	91,50%	100%	97,70%	Epilepsy, progressive myoclonic 2A (Lafora), 254780
EPO	99,90%	97,60%	100%	100%	Erythrocytosis, familial, 5, 617907 ?Diamond-Blackfan anemia-like, 617911
EPRS	100%	99,60%	100%	100%	Leukodystrophy, hypomyelinating, 15, 617951
EPS8	100%	99,10%	100%	100%	?Deafness, autosomal recessive 102, 615974
EPS8L2	96,10%	93,70%	100%	100%	Deafness autosomal recessive 106, 617637
ERAL1	100%	99,70%	100%	100%	Perrault syndrome 6, 617565
ERBB3	100%	99,80%	100%	100%	?Lethal congenital contractural syndrome 2, 607598
ERCC1	100%	99,30%	100%	100%	Cerebrooculofacioskeletal syndrome 4, 610758

ERCC2	100%	99,70%	100%	100%	Trichothiodystrophy 1, photosensitive, 601675 ?Cerebrooculofacioskeletal syndrome 2, 610756 Xeroderma pigmentosum, group D, 278730
ERCC3	100%	99,40%	100%	100%	Xeroderma pigmentosum, group B, 610651 Trichothiodystrophy 2, photosensitive, 616390
ERCC4	100%	99,90%	100%	100%	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 Fanconi anemia, complementation group Q, 615272 XFE progeroid syndrome, 610965 Xeroderma pigmentosum, group F, 278760
ERCC5	100%	99,60%	100%	100%	Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 Xeroderma pigmentosum, group G, 278780 Cerebrooculofacioskeletal syndrome 3, 616570
ERCC6	100%	100%	100%	100%	Cerebrooculofacioskeletal syndrome 1, 214150 Cockayne syndrome, type B, 133540 Premature ovarian failure 11, 616946 UV-sensitive syndrome 1, 600630 De Sanctis-Cacchione syndrome, 278800
ERCC6L2	100%	99,40%	100%	100%	Bone marrow failure syndrome 2, 615715
ERCC8	99,50%	95,80%	100%	100%	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621
ERGIC1	95,20%	94,60%	98,40%	98,40%	?Arthrogryposis multiplex congenita, neurogenic type, 208100
ERLIN1	100%	100%	100%	100%	Spastic paraplegia 62, 615681
ERLIN2	100%	99,90%	100%	100%	Spastic paraplegia 18, autosomal recessive, 611225
ESCO2	98,70%	95,20%	100%	100%	Roberts syndrome, 268300 SC phocomelia syndrome, 269000
ESPN	44,60%	35,80%	100%	99,80%	?Usher syndrome, type 1M, 618632 Deafness, autosomal recessive 36, 609006 Deafness, neurosensory, without vestibular involvement, autosomal dominant, 609006
ESR1	100%	99,80%	100%	100%	Estrogen resistance, 615363 Breast cancer, somatic, 114480
ESRP1	99,90%	98,90%	100%	100%	?Deafness, autosomal recessive 109, 618013
ESRRB	99,90%	98,00%	100%	100%	Deafness, autosomal recessive 35, 608565
ETFA	100%	100%	100%	100%	Glutaric acidemia IIA, 231680
ETFB	100%	99,80%	100%	100%	Glutaric acidemia IIB, 231680
ETFDH	100%	99,80%	100%	100%	Glutaric acidemia IIC, 231680
ETHE1	99,90%	97,40%	100%	100%	Ethylmalonic encephalopathy, 602473
EVC	93,90%	88,60%	96,90%	94,80%	Ellis-van Creveld syndrome, 225500 ?Weyers acrofacial dysostosis, 193530

EVC2	97,70%	96,10%	100%	100%	Weyers acrofacial dysostosis, 193530 Ellis-van Creveld syndrome, 225500
EXOC6B	99,10%	97,60%	100%	100%	Spondyloepimetaphyseal dysplasia with joint laxity, type 3, 618395
EXOSC2	100%	100%	100%	100%	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EXOSC3	99,50%	94,90%	100%	100%	Pontocerebellar hypoplasia, type 1B, 614678
EXOSC8	97,90%	91,20%	100%	100%	Pontocerebellar hypoplasia, type 1C, 616081
EXOSC9	99,70%	97,20%	100%	100%	Pontocerebellar hypoplasia, type 1D, 618065
EXPH5	100%	100%	100%	100%	Epidermolysis bullosa, nonspecific, autosomal recessive, 615028
EXT2	100%	99,30%	100%	100%	Exostoses, multiple, type 2, 133701 Seizures, scoliosis, and macrocephaly syndrome, 616682
EXTL3	100%	100%	100%	100%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
EYS	99,70%	98,20%	100%	100%	Retinitis pigmentosa 25, 602772
F10	99,80%	99,10%	100%	100%	Factor X deficiency, 227600
F11	100%	100%	100%	100%	Factor XI deficiency, autosomal dominant, 612416 Factor XI deficiency, autosomal recessive, 612416
F12	99,90%	98,80%	100%	100%	Factor XII deficiency, 234000 Angioedema, hereditary, type III, 610618
F13A1	100%	100%	100%	100%	Factor XIIIA deficiency, 613225
F13B	98,70%	93,50%	100%	100%	Factor XIIIB deficiency, 613235
F2	99,90%	97,90%	100%	100%	Hypoprothrombinemia, 613679 Dysprothrombinemia, 613679 Thrombophilia due to thrombin defect, 188050
F5	99,40%	98,50%	100%	100%	Factor V deficiency, 227400 Thrombophilia due to activated protein C resistance, 188055
F7	100%	100%	100%	100%	Factor VII deficiency, 227500
FA2H	92,00%	83,10%	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%	100%	100%	100%	Tyrosinemia, type I, 276700
FAM126A	100%	99,40%	100%	100%	Leukodystrophy, hypomyelinating, 5, 610532
FAM161A	100%	99,70%	100%	100%	Retinitis pigmentosa 28, 606068
FAM20A	99,60%	94,70%	100%	100%	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM20C	100%	100%	100%	99,80%	Raine syndrome, 259775
FAM46A	100%	99,70%	100%	100%	Osteogenesis imperfecta, type XVIII, 617952
FAN1	100%	99,80%	100%	100%	Interstitial nephritis, karyomegalic, 614817
FANCA	100%	99,40%	100%	100%	Fanconi anemia, complementation group A, 227650
FANCB	98,60%	94,10%	100%	100%	Fanconi anemia, complementation group B, 300514

FANCC	99,90%	99,30%	100%	100%	Fanconi anemia, complementation group C, 227645
FANCD2	99,50%	97,50%	98,80%	98,80%	Fanconi anemia, complementation group D2, 227646
FANCE	89,80%	85,10%	100%	99,90%	Fanconi anemia, complementation group E, 600901
FANCF	100%	100%	100%	100%	Fanconi anemia, complementation group F, 603467
FANCG	100%	99,90%	100%	100%	Fanconi anemia, complementation group G, 614082
FANCI	99,90%	99,20%	100%	100%	Fanconi anemia, complementation group I, 609053
FANCL	100%	98,60%	100%	100%	Fanconi anemia, complementation group L, 614083
FAR1	97,60%	92,80%	100%	100%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154
FARS2	100%	100%	100%	100%	Spastic paraplegia 77, autosomal recessive, 617046 Combined oxidative phosphorylation deficiency 14, 614946
FARSB	98,80%	94,60%	100%	100%	Rajab interstitial lung disease with brain calcifications, 613658
FASTKD2	99,80%	98,90%	100%	100%	?Mitochondrial complex IV deficiency, 220110
FAT4	100%	100%	100%	100%	Van Maldergem syndrome 2, 615546 Hennekam lymphangiectasia-lymphedema syndrome 2, 616006
FBLN5	91,80%	91,80%	91,80%	91,80%	Macular degeneration, age-related, 3, 608895 ?Cutis laxa, autosomal dominant 2, 614434 Neuropathy, hereditary, with or without age-related macular degeneration, 608895 Cutis laxa, autosomal recessive, type IA, 219100
FBP1	100%	99,50%	100%	100%	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	96,00%	93,10%	100%	99,90%	?Mental retardation, autosomal recessive 45, 615979
FBXO7	99,80%	97,90%	100%	100%	Parkinson disease 15, autosomal recessive, 260300
FCGR3A	99,00%	97,10%	100%	100%	Immunodeficiency 20, 615707
FCN3	100%	100%	100%	100%	Immunodeficiency due to ficolin 3 deficiency, 613860
FDFT1	97,70%	96,00%	100%	100%	Squalene synthase deficiency, 618156
FDX2	100%	100%	100%	100%	Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900
FDXR	100%	99,30%	100%	100%	Auditory neuropathy and optic atrophy, 617717
FECH	100%	100%	100%	100%	Protoporphyria, erythropoietic, 1, 177000
FERMT1	99,90%	97,90%	100%	100%	Kindler syndrome, 173650
FERMT3	100%	100%	100%	100%	Leukocyte adhesion deficiency, type III, 612840
FEZF1	100%	99,90%	100%	100%	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FGA	99,10%	97,20%	100%	100%	Dysfibrinogenemia, congenital, 616004 Amyloidosis, familial visceral, 105200 Hypodysfibrinogenemia, congenital, 616004 Afibrinogenemia, congenital, 202400

FGB	99,80%	99,10%	100%	100%	Afibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Hypofibrinogenemia, congenital, 202400
FGD4	99,90%	99,40%	100%	100%	Charcot-Marie-Tooth disease, type 4H, 609311
FGF20	97,50%	87,60%	100%	100%	?Renal hypodysplasia/aplasia 2, 615721
FGF23	99,60%	97,50%	100%	100%	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993 Hypophosphatemic rickets, autosomal dominant, 193100
FGF3	99,80%	95,10%	100%	100%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGG	99,70%	98,20%	100%	100%	Hypofibrinogenemia, congenital, 202400 Hypodysfibrinogenemia, 616004 Dysfibrinogenemia, congenital, 616004 Afibrinogenemia, congenital, 202400
FH	92,10%	88,30%	100%	100%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FIBP	100%	100%	100%	100%	Thauvin-Robinet-Faivre syndrome, 617107
FIG4	100%	99,80%	100%	100%	Yunis-Varon syndrome, 216340 ?Polymicrogyria, bilateral temporooccipital, 612691 Charcot-Marie-Tooth disease, type 4J, 611228 Amyotrophic lateral sclerosis 11, 612577
FITM2	100%	100%	100%	100%	Siddiqi syndrome, 618635
FKBP10	98,80%	97,20%	100%	100%	Bruck syndrome 1, 259450 Osteogenesis imperfecta, type XI, 610968
FKBP14	100%	99,90%	100%	100%	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557
FKRP	100%	100%	100%	99,90%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153
FKTN	99,70%	97,00%	100%	100%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800
FLAD1	100%	99,80%	100%	100%	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100
FLG	100%	99,90%	100%	100%	Ichthyosis vulgaris, 146700
FLNB	99,50%	98,80%	100%	100%	Larsen syndrome, 150250 Atelosteogenesis, type I, 108720 Boomerang dysplasia, 112310 Spondylocarpotarsal synostosis syndrome, 272460 Atelosteogenesis, type III, 108721

FLVCR1	100%	98,90%	100%	100%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FLVCR2	100%	100%	100%	100%	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790
FMN2	85,50%	82,50%	100%	100%	Mental retardation, autosomal recessive 47, 616193
FMO3	99,90%	99,70%	100%	100%	Trimethylaminuria, 602079
FOLR1	100%	100%	100%	100%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXE1	96,90%	78,50%	99,90%	99,10%	Bamforth-Lazarus syndrome, 241850
FOXE3	82,60%	72,00%	94,40%	87,80%	Cataract 34, multiple types, 612968 Anterior segment dysgenesis 2, multiple subtypes, 610256
FOXI1	100%	100%	100%	100%	Enlarged vestibular aqueduct, 600791
FOXN1	100%	99,60%	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%	99,90%	100%	100%	Mitochondrial complex I deficiency, nuclear type 19, 618241
FRAS1	100%	99,40%	100%	100%	Fraser syndrome 1, 219000
FREM1	99,90%	99,10%	100%	100%	Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485 Bifid nose with or without anorectal and renal anomalies, 608980
FREM2	100%	99,30%	100%	100%	Fraser syndrome 2, 617666 Cryptophthalmos, unilateral or bilateral, isolated, 123570
FRMD4A	90,70%	87,30%	96,60%	96,60%	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819
FRRS1L	79,70%	69,10%	99,20%	95,80%	Epileptic encephalopathy, early infantile, 37, 616981
FSHB	100%	100%	100%	100%	Hypogonadotropic hypogonadism 24 without anosmia, 229070
FSHR	99,50%	97,20%	100%	100%	Ovarian hyperstimulation syndrome, 608115 Ovarian dysgenesis 1, 233300 Ovarian response to FSH stimulation, 276400
FTCD	95,70%	91,00%	100%	100%	Glutamate formiminotransferase deficiency, 229100
FTO	83,80%	83,70%	94,20%	94,20%	Growth retardation, developmental delay, facial dysmorphism, 612938
FUCA1	100%	99,90%	100%	100%	Fucosidosis, 230000
FUK	97,70%	95,40%	100%	100%	Congenital disorder of glycosylation with defective fucosylation 2, 618324
FUT8	100%	99,20%	100%	100%	Congenital disorder of glycosylation with defective fucosylation 1, 618005
FXN	95,50%	80,10%	100%	100%	Friedreich ataxia with retained reflexes, 229300 Friedreich ataxia, 229300
FYCO1	100%	99,90%	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%	99,90%	100%	100%	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
GAA	100%	99,90%	100%	100%	Glycogen storage disease II, 232300

GAB1	100%	99,40%	100%	100%	?Deafness, autosomal recessive 26, 605428
GAD1	100%	99,90%	100%	100%	?Cerebral palsy, spastic quadriplegic, 1, 603513
GALC	99,80%	98,30%	100%	100%	Krabbe disease, 245200
GALE	100%	100%	100%	100%	Galactose epimerase deficiency, 230350
GALK1	100%	99,10%	100%	100%	Galactokinase deficiency with cataracts, 230200
GALNS	100%	99,80%	100%	100%	Mucopolysaccharidosis IVA, 253000
GALNT3	99,80%	99,00%	100%	100%	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GALT	100%	99,70%	100%	100%	Galactosemia, 230400
GAMT	93,10%	82,70%	100%	100%	Cerebral creatine deficiency syndrome 2, 612736
GAN	100%	99,60%	100%	100%	Giant axonal neuropathy-1, 256850
GAS2L2	100%	100%	100%	100%	?Ciliary dyskinesia, primary, 41, 618449
GAS8	99,90%	99,30%	100%	100%	Ciliary dyskinesia, primary, 33, 616726
GATM	100%	100%	100%	100%	Cerebral creatine deficiency syndrome 3, 612718
GBA	100%	100%	100%	100%	Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 Gaucher disease, type I, 230800 Gaucher disease, perinatal lethal, 608013 Gaucher disease, type II, 230900
GBA2	100%	99,70%	100%	100%	Spastic paraplegia 46, autosomal recessive, 614409
GBE1	100%	99,60%	100%	100%	Polyglucosan body disease, adult form, 263570 Glycogen storage disease IV, 232500
GCDH	100%	99,20%	100%	100%	Glutaricaciduria, type I, 231670
GCH1	99,90%	95,50%	100%	100%	Hyperphenylalaninemia, BH4-deficient, B, 233910 Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230
GCK	100%	100%	100%	100%	Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, permanent neonatal, 606176 MODY, type II, 125851 Hyperinsulinemic hypoglycemia, familial, 3, 602485
GCLC	99,80%	98,00%	100%	100%	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450
GCNT2	99,50%	99,50%	100%	100%	Adult i phenotype without cataract, 110800 Cataract 13 with adult i phenotype, 116700
GCSH	75,70%	68,90%	100%	100%	?Glycine encephalopathy, 605899
GDAP1	99,80%	99,30%	100%	100%	Charcot-Marie-Tooth disease, recessive intermediate, A, 608340 Charcot-Marie-Tooth disease, type 4A, 214400 Charcot-Marie-Tooth disease, axonal, type 2K, 607831 Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706
GDAP2	100%	99,20%	100%	100%	Spinocerebellar ataxia, autosomal recessive 27, 618369

GDF1	73,90%	54,00%	98,70%	92,00%	Right atrial isomerism (Ivemark), 208530 Congenital heart defects, multiple types, 6, 613854
GDF5	100%	100%	100%	100%	?Acromesomelic dysplasia, Hunter-Thompson type, 201250 Symphalangism, proximal, 1B, 615298 Brachydactyly, type A1, C, 615072 Chondrodysplasia, Grebe type, 200700 Brachydactyly, type A2, 112600 Du Pan syndrome, 228900 Brachydactyly, type C, 113100 Multiple synostoses syndrome 2, 610017
GEMIN4	100%	99,50%	100%	100%	Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities, 617913
GFER	99,60%	93,90%	100%	100%	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076
GFM1	99,90%	99,40%	100%	100%	Combined oxidative phosphorylation deficiency 1, 609060
GFM2	98,90%	95,20%	100%	100%	Combined oxidative phosphorylation deficiency 39, 618397
GFPT1	100%	99,40%	100%	100%	Myasthenia, congenital, 12, with tubular aggregates, 610542
GGCX	100%	99,90%	100%	100%	Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450 Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842
GGT1	19,90%	18,40%	100%	100%	?Glutathioninuria, 231950
GH1	100%	100%	100%	100%	Kowarski syndrome, 262650 Growth hormone deficiency, isolated, type IA, 262400 Growth hormone deficiency, isolated, type IB, 612781 Growth hormone deficiency, isolated, type II, 173100
GHR	99,60%	99,50%	99,80%	99,80%	Increased responsiveness to growth hormone, 604271 Laron dwarfism, 262500 Growth hormone insensitivity, partial, 604271
GHRHR	96,40%	96,10%	100%	100%	Growth hormone deficiency, isolated, type IV, 618157
GHSR	98,50%	95,80%	100%	100%	Growth hormone deficiency, isolated partial, 615925
GIF	100%	99,70%	100%	100%	Intrinsic factor deficiency, 261000
GINS1	99,30%	94,90%	100%	100%	Immunodeficiency 55, 617827
GIPC3	97,50%	94,20%	99,60%	98,10%	Deafness, autosomal recessive 15, 601869
GJA1	100%	100%	100%	100%	Erythrokeratodermia variabilis et progressiva 3, 617525 Craniometaphyseal dysplasia, autosomal recessive, 218400 Atrioventricular septal defect 3, 600309 Oculodentodigital dysplasia, 164200 Syndactyly, type III, 186100 Oculodentodigital dysplasia, autosomal recessive, 257850

					Hypoplastic left heart syndrome 1, 241550 Palmoplantar keratoderma with congenital alopecia, 104100
GJB2	100%	100%	100%	100%	Deafness, autosomal dominant 3A, 601544 Deafness, autosomal recessive 1A, 220290 Bart-Pumphrey syndrome, 149200 Vohwinkel syndrome, 124500 Keratoderma, palmoplantar, with deafness, 148350 Keratitis-ichthyosis-deafness syndrome, 148210 Hystrix-like ichthyosis with deafness, 602540
GJB6	100%	100%	100%	100%	Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290 Ectodermal dysplasia 2, Clouston type, 129500
GJC2	78,20%	58,70%	96,90%	91,40%	Spastic paraplegia 44, autosomal recessive, 613206 Lymphatic malformation 3, 613480 Leukodystrophy, hypomyelinating, 2, 608804
GLB1	99,90%	97,40%	100%	100%	GM1-gangliosidosis, type III, 230650 GM1-gangliosidosis, type I, 230500 Mucopolysaccharidosis type IVB (Morquio), 253010 GM1-gangliosidosis, type II, 230600
GLDC	89,90%	82,00%	100%	99,90%	Glycine encephalopathy, 605899
GLDN	94,60%	91,00%	100%	100%	Lethal congenital contracture syndrome 11, 617194
GLE1	100%	100%	100%	100%	Lethal congenital contracture syndrome 1, 253310 Congenital arthrogryposis with anterior horn cell disease, 611890
GLIS2	100%	99,80%	100%	100%	Nephronophthisis 7, 611498
GLIS3	100%	99,60%	100%	100%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GLRA1	100%	99,80%	100%	100%	Hyperekplexia 1, 149400
GLRB	99,20%	95,10%	100%	100%	Hyperekplexia 2, 614619
GLRX5	97,30%	89,10%	99,60%	95,40%	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GLS	96,30%	87,20%	100%	99,90%	?Infantile cataract, skin abnormalities, glutamate excess, and impaired intellectual development, 618339 Global developmental delay, progressive ataxia, and elevated glutamine, 618412 Epileptic encephalopathy, early infantile, 71, 618328
GLUL	99,90%	98,20%	100%	100%	Glutamine deficiency, congenital, 610015
GLYCTK	98,80%	97,30%	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 (congenital with brain and eye anomalies), type A, 14, 615350 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352
GNAT2	99,90%	99,00%	100%	100%	Achromatopsia 4, 613856
GNB3	100%	100%	100%	100%	Night blindness, congenital stationary, type 1H, 617024
GNB5	100%	98,80%	100%	100%	Intellectual developmental disorder with cardiac arrhythmia, 617173 Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182
GNE	100%	99,70%	100%	100%	Sialuria, 269921 Nonaka myopathy, 605820
GNMT	100%	100%	100%	100%	Glycine N-methyltransferase deficiency, 606664
GNPAT	99,70%	97,30%	100%	100%	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	100%	99,90%	100%	100%	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GNPTG	99,10%	94,30%	100%	99,90%	Mucolipidosis III gamma, 252605
GNRHR	100%	100%	100%	100%	Hypogonadotropic hypogonadism 7 without anosmia, 146110
GNS	98,40%	94,80%	100%	100%	Mucopolysaccharidosis type IIID, 252940
GORAB	100%	99,10%	100%	100%	Geroderma osteodysplasticum, 231070
GOSR2	95,90%	94,60%	100%	100%	Epilepsy, progressive myoclonic 6, 614018
GP1BA	98,60%	95,90%	100%	100%	Bernard-Soulier syndrome, type A1 (recessive), 231200 von Willebrand disease, platelet-type, 177820 Bernard-Soulier syndrome, type A2 (dominant), 153670
GP1BB	72,90%	59,60%	99,50%	95,00%	Giant platelet disorder, isolated, 231200 Bernard-Soulier syndrome, type B, 231200
GP6	100%	100%	94,90%	91,70%	Bleeding disorder, platelet-type, 11, 614201
GP9	96,50%	89,30%	100%	100%	Bernard-Soulier syndrome, type C, 231200
GPAA1	98,90%	95,90%	100%	100%	Glycosylphosphatidylinositol biosynthesis defect 15, 617810
GPC6	100%	100%	100%	100%	Omodysplasia 1, 258315
GPD1	100%	100%	100%	100%	Hypertriglyceridemia, transient infantile, 614480
GPHN	100%	99,50%	100%	100%	Molybdenum cofactor deficiency C, 615501
GPI	100%	100%	100%	100%	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470
GPIHBP1	100%	100%	100%	100%	Hyperlipoproteinemia, type 1D, 615947
GPNMB	100%	100%	100%	100%	Amyloidosis, primary localized cutaneous, 3, 617920
GPR179	100%	100%	100%	100%	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
GPR88	99,40%	95,10%	98,80%	94,90%	?Chorea, childhood-onset, with psychomotor retardation, 616939
GPSM2	99,90%	99,20%	100%	100%	Chudley-McCullough syndrome, 604213
GPT2	99,20%	93,60%	100%	99,80%	Mental retardation, autosomal recessive 49, 616281
GPX4	90,50%	85,80%	98,20%	94,90%	Spondylometaphyseal dysplasia, Sedaghatian type, 250220

GRAP	82,80%	78,30%	100%	100%	Deafness, autosomal recessive 114, 618456
GRHPR	84,20%	81,30%	100%	99,30%	Hyperoxaluria, primary, type II, 260000
GRID2	100%	99,80%	100%	100%	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRIK2	96,20%	95,40%	96,30%	96,30%	Mental retardation, autosomal recessive, 6, 611092
GRIN1	100%	100%	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%	99,70%	100%	100%	Fraser syndrome 3, 617667
GRK1	100%	100%	100%	100%	Oguchi disease-2, 613411
GRM1	100%	99,70%	100%	100%	Spinocerebellar ataxia 44, 617691 Spinocerebellar ataxia, autosomal recessive 13, 614831
GRM6	90,20%	80,60%	98,30%	96,30%	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270
GRN	100%	100%	100%	100%	Ceroid lipofuscinosis, neuronal, 11, 614706 Aphasia, primary progressive, 607485 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
GRXCR1	100%	99,80%	100%	100%	Deafness, autosomal recessive 25, 613285
GRXCR2	100%	100%	100%	100%	?Deafness, autosomal recessive 101, 615837
GSC	99,20%	92,40%	100%	100%	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471
GSS	100%	99,90%	100%	100%	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900
GSX2	100%	100%	100%	100%	Diencephalic-mesencephalic junction dysplasia syndrome 2, 618646
GTF2E2	100%	99,80%	100%	100%	Trichothiodystrophy 6, nonphotosensitive, 616943
GTF2H5	100%	99,60%	100%	100%	Trichothiodystrophy 3, photosensitive, 616395
GTPBP2	100%	99,30%	100%	99,90%	Jaberi-Elahi syndrome, 617988
GTPBP3	100%	99,80%	100%	100%	Combined oxidative phosphorylation deficiency 23, 616198
GUCY1A3	100%	99,80%	100%	100%	Moyamoya 6 with achalasia, 615750
GUCY2C	100%	99,60%	100%	100%	Diarrhea 6, 614616 Meconium ileus, 614665
GUCY2D	99,60%	96,20%	100%	100%	Cone-rod dystrophy 6, 601777 Leber congenital amaurosis 1, 204000 Night blindness, congenital stationary, type 1I, 618555 ?Choroidal dystrophy, central areolar 1, 215500
GUF1	99,70%	97,80%	100%	100%	?Epileptic encephalopathy, early infantile, 40, 617065
GUSB	92,90%	91,70%	100%	100%	Mucopolysaccharidosis VII, 253220
GYG1	99,90%	99,20%	100%	100%	?Glycogen storage disease XV, 613507 Polyglucosan body myopathy 2, 616199
GYS1	100%	98,60%	100%	100%	Glycogen storage disease 0, muscle, 611556

GYS2	99,80%	99,00%	100%	100%	Glycogen storage disease 0, liver, 240600
GZF1	100%	99,60%	100%	100%	Joint laxity, short stature, and myopia, 617662
H6PD	99,00%	99,00%	100%	100%	Cortisone reductase deficiency 1, 604931
HAAO	100%	99,80%	100%	100%	Vertebral, cardiac, renal, and limb defects syndrome 1, 617660
HACE1	100%	99,30%	100%	100%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADH	99,00%	97,50%	100%	100%	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HADHA	97,10%	91,30%	100%	100%	LCHAD deficiency, 609016 HELLP syndrome, maternal, of pregnancy, 609016 Fatty liver, acute, of pregnancy, 609016 Trifunctional protein deficiency, 609015
HADHB	98,80%	89,70%	100%	100%	Trifunctional protein deficiency, 609015
HAMP	100%	100%	100%	100%	Hemochromatosis, type 2B, 613313
HARS	100%	100%	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504
HARS2	100%	100%	100%	100%	?Perrault syndrome 2, 614926
HAVCR2	100%	100%	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%	Thalassemia, beta, 613985 Methmoglobinemia, beta type, 617971 Erythrocytosis 6, 617980 Heinz body anemia, 140700 Delta-beta thalassemia, 141749 Thalassemia-beta, dominant inclusion-body, 603902 Hereditary persistence of fetal hemoglobin, 141749 Sickle cell anemia, 603903
HELLS	97,80%	92,10%	100%	100%	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911
HEPACAM	86,00%	78,90%	100%	100%	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926
HEPHL1	100%	99,90%	100%	100%	?Abnormal hair, joint laxity, and developmental delay, 261990
HERC1	100%	100%	100%	100%	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
HERC2	79,90%	77,20%	100%	100%	Mental retardation, autosomal recessive 38, 615516
HES7	84,40%	53,90%	100%	100%	Spondylocostal dysostosis 4, autosomal recessive, 613686
HESX1	99,70%	97,30%	100%	100%	Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230 Growth hormone deficiency with pituitary anomalies, 182230

HEXA	93,80%	93,30%	100%	100%	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800
HEXB	99,60%	96,90%	100%	99,90%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HFE	100%	99,70%	100%	100%	Hemochromatosis, 235200
HFE2	100%	100%	100%	100%	Hemochromatosis, type 2A, 602390
HFM1	96,30%	89,60%	100%	100%	Premature ovarian failure 9, 615724
HGD	100%	100%	100%	100%	Alkaptonuria, 203500
HGF	100%	99,40%	100%	100%	Deafness, autosomal recessive 39, 608265
HGSNAT	86,40%	86,30%	91,20%	89,30%	Retinitis pigmentosa 73, 616544 Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930
HIBCH	98,20%	88,50%	100%	100%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HIKESHI	98,20%	90,40%	100%	100%	Leukodystrophy, hypomyelinating, 13, 616881
HINT1	98,30%	89,30%	100%	100%	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200
HK1	100%	100%	100%	100%	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285 Neurodevelopmental disorder with visual defects and brain anomalies, 618547 Retinitis pigmentosa 79, 617460
HLCS	100%	100%	100%	100%	Holocarboxylase synthetase deficiency, 253270
HMGCL	100%	99,80%	100%	100%	HMG-CoA lyase deficiency, 246450
HMGCS2	100%	99,60%	100%	100%	HMG-CoA synthase-2 deficiency, 605911
HMOX1	98,40%	89,90%	100%	100%	Heme oxygenase-1 deficiency, 614034
HMX1	62,40%	42,90%	99,70%	96,10%	Oculoauricular syndrome, 612109
HNMT	100%	99,80%	100%	100%	Mental retardation, autosomal recessive 51, 616739
HOGA1	100%	96,40%	100%	100%	Hyperoxaluria, primary, type III, 613616
HOXA1	100%	100%	100%	100%	Athabaskan brainstem dysgenesis syndrome, 601536 Bosley-Salih-Alorainy syndrome, 601536
HOXA2	100%	99,90%	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%	99,90%	100%	100%	Ectodermal dysplasia 9, hair/nail type, 614931
HPCA	100%	100%	100%	100%	Dystonia 2, torsion, autosomal recessive, 224500
HPD	100%	100%	100%	100%	Tyrosinemia, type III, 276710 Hawkinsinuria, 140350
HPGD	100%	98,90%	100%	100%	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	99,70%	97,50%	100%	100%	Hermansky-Pudlak syndrome 3, 614072
HPS4	100%	100%	100%	100%	Hermansky-Pudlak syndrome 4, 614073
HPS5	100%	99,70%	100%	100%	Hermansky-Pudlak syndrome 5, 614074
HPS6	97,10%	88,90%	100%	100%	Hermansky-Pudlak syndrome 6, 614075
HPSE2	100%	99,90%	100%	100%	Urofacial syndrome 1, 236730
HR	98,50%	95,60%	100%	100%	Hypotrichosis 4, 146550 Alopecia universalis, 203655 Atrichia with papular lesions, 209500
HSD11B2	86,00%	82,70%	99,90%	98,10%	Apparent mineralocorticoid excess, 218030
HSD17B3	100%	100%	100%	100%	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	96,00%	93,70%	96,60%	96,60%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B2	100%	99,70%	100%	100%	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810
HSD3B7	99,10%	95,50%	100%	100%	Bile acid synthesis defect, congenital, 1, 607765
HSPA9	88,50%	84,50%	100%	100%	Even-plus syndrome, 616854 Anemia, sideroblastic, 4, 182170
HSPD1	98,80%	93,70%	100%	100%	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233
HSPG2	99,20%	97,70%	100%	99,90%	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
HTRA1	83,90%	80,00%	96,50%	92,10%	Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779 CARASIL syndrome, 600142
HTRA2	100%	99,90%	100%	100%	3-methylglutaconic aciduria, type VIII, 617248
HYAL1	100%	100%	100%	100%	?Mucopolysaccharidosis type IX, 601492
HYDIN	99,90%	99,30%	100%	100%	Ciliary dyskinesia, primary, 5, 608647
HYLS1	100%	100%	100%	100%	Hydrolethalus syndrome, 236680
IARS	100%	99,60%	100%	100%	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093
IARS2	100%	99,90%	100%	100%	?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
IBA57	93,70%	90,10%	100%	100%	?Spastic paraplegia 74, autosomal recessive, 616451 Multiple mitochondrial dysfunctions syndrome 3, 615330
ICK	99,90%	98,70%	100%	100%	Endocrine-cerebroosteodysplasia, 612651
ICOS	99,90%	99,80%	100%	100%	Immunodeficiency, common variable, 1, 607594
IDH3B	95,40%	95,40%	100%	100%	Retinitis pigmentosa 46, 612572
IDUA	93,70%	86,80%	100%	100%	Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Is, 607016

IER3IP1	91,90%	82,60%	100%	100%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFNAR2	100%	99,70%	100%	100%	?Immunodeficiency 45, 616669
IFNGR1	100%	99,40%	100%	100%	Immunodeficiency 27A, mycobacteriosis, AR, 209950 Immunodeficiency 27B, mycobacteriosis, AD, 615978
IFNGR2	93,30%	93,20%	100%	99,80%	Immunodeficiency 28, mycobacteriosis, 614889
IFT122	100%	99,60%	100%	100%	Cranioectodermal dysplasia 1, 218330
IFT140	99,80%	98,80%	100%	100%	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	99,90%	99,10%	100%	100%	Retinitis pigmentosa 71, 616394 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 Short-rib thoracic dysplasia 18 with polydactyly, 617866 ?Retinitis pigmentosa 81, 617871
IFT52	100%	99,90%	100%	100%	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102
IFT57	99,90%	99,10%	100%	100%	?Orofaciodigital syndrome XVIII, 617927
IFT74	98,40%	93,90%	100%	100%	?Bardet-Biedl syndrome 20, 617119
IFT80	97,60%	88,20%	100%	100%	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IFT81	93,50%	90,10%	95,00%	94,90%	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IGF1	100%	99,90%	100%	100%	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IGF1R	100%	99,90%	100%	100%	Insulin-like growth factor I, resistance to, 270450
IGFALS	99,90%	99,60%	100%	100%	Acid-labile subunit, deficiency of, 615961
IGFBP7	92,70%	87,20%	100%	100%	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis, 614224
IGHM	100%	100%	100%	100%	Agammaglobulinemia 1, 601495
IGHMBP2	98,80%	95,10%	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	99,90%	96,90%	100%	100%	Agammaglobulinemia 2, 613500
IHH	100%	100%	100%	100%	Acrocapitofemoral dysplasia, 607778 Brachydactyly, type A1, 112500
IKBKB	99,70%	96,50%	97,20%	97,20%	Immunodeficiency 15A, 618204 Immunodeficiency 15B, 615592
IL10RA	100%	100%	100%	100%	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	99,80%	98,00%	100%	100%	Inflammatory bowel disease 25, early onset, autosomal recessive, 612567
IL11RA	100%	99,90%	100%	100%	Craniosynostosis and dental anomalies, 614188
IL12B	100%	99,30%	100%	100%	Immunodeficiency 29, mycobacteriosis, 614890
IL12RB1	98,90%	96,30%	94,10%	94,10%	Immunodeficiency 30, 614891

IL17RA	100%	99,40%	100%	100%	Immunodeficiency 51, 613953
IL17RC	100%	99,90%	100%	100%	Candidiasis, familial, 9, 616445
IL1RN	100%	100%	100%	100%	Interleukin 1 receptor antagonist deficiency, 612852
IL21R	100%	100%	100%	100%	Immunodeficiency 56, 615207
IL2RA	100%	99,70%	100%	100%	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367
IL2RB	100%	99,70%	100%	100%	Immunodeficiency 63 with lymphoproliferation and autoimmunity, 618495
IL36RN	100%	100%	100%	100%	Psoriasis 14, pustular, 614204
IL6ST	96,40%	90,30%	100%	100%	Hyper-IgE recurrent infection syndrome 4, autosomal recessive, 618523
IL7R	100%	99,80%	100%	100%	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
ILDR1	99,90%	98,50%	100%	100%	Deafness, autosomal recessive 42, 609646
IMPA1	97,00%	87,00%	100%	100%	Mental retardation, autosomal recessive 59, 617323
IMPAD1	100%	100%	100%	100%	Chondrodysplasia with joint dislocations, GPAPP type, 614078
IMPG2	99,80%	98,40%	100%	100%	Macular dystrophy, vitelliform, 5, 616152 Retinitis pigmentosa 56, 613581
INPP5E	97,10%	92,70%	100%	100%	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300
INPP5K	100%	100%	100%	100%	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404
INPPL1	98,40%	94,50%	99,90%	99,70%	Opsismodysplasia, 258480
INSR	97,80%	94,70%	99,90%	99,20%	Hyperinsulinemic hypoglycemia, familial, 5, 609968 Rabson-Mendenhall syndrome, 262190 Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Leprechaunism, 246200
MR AP	100%	100%	100%	100%	Glucocorticoid deficiency 2, 607398
MR E11	98,90%	93,30%	100%	100%	Ataxia-telangiectasia-like disorder 1, 604391
MR M2	100%	99,50%	99,00%	99,00%	?Mitochondrial DNA depletion syndrome 17, 618567
MR PL3	93,20%	87,20%	100%	100%	Combined oxidative phosphorylation deficiency 9, 614582
MR PL44	99,90%	98,70%	100%	100%	?Combined oxidative phosphorylation deficiency 16, 615395
MR PS14	100%	100%	100%	100%	?Combined oxidative phosphorylation deficiency 38, 618378
MR PS16	100%	99,60%	100%	100%	Combined oxidative phosphorylation deficiency 2, 610498
MR PS2	99,60%	96,90%	100%	100%	Combined oxidative phosphorylation deficiency 36, 617950

MR PS22	99,80%	99,10%	100%	100%	Combined oxidative phosphorylation deficiency 5, 611719 Ovarian dysgenesis 7, 618117
MR PS34	97,60%	92,00%	100%	100%	Combined oxidative phosphorylation deficiency 32, 617664
MR PS7	100%	100%	100%	100%	?Combined oxidative phosphorylation deficiency 34, 617872
INTS1	99,80%	98,50%	100%	100%	Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies, 618571
INTS8	99,90%	98,80%	100%	100%	?Neurodevelopmental disorder with cerebellar hypoplasia and spasticity, 618572
INTU	99,70%	98,10%	100%	100%	?Short-rib thoracic dysplasia 20 with polydactyly, 617925 ?Orofaciodigital syndrome XVII, 617926
INVS	100%	100%	100%	100%	Nephronophthisis 2, infantile, 602088
IQCB1	93,90%	85,00%	100%	100%	Senior-Loken syndrome 5, 609254
IRAK4	99,80%	97,70%	100%	100%	IRAK4 deficiency, 607676 Invasive pneumococcal disease, recurrent isolated, 1, 610799
IREB2	100%	99,80%	100%	100%	Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451
IRF7	100%	99,90%	100%	100%	?Immunodeficiency 39, 616345
IRF8	99,00%	95,70%	100%	100%	Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990 Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893
IRF9	100%	100%	100%	100%	Immunodeficiency 65, susceptibility to viral infections, 618648
IRX5	99,90%	98,20%	100%	99,80%	Hamamy syndrome, 611174
ISCA1	94,20%	85,90%	95,10%	95,10%	Multiple mitochondrial dysfunctions syndrome 5, 617613
ISCA2	100%	98,80%	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
ISPD	98,50%	94,80%	100%	99,40%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
ITCH	95,40%	95,20%	100%	99,00%	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITGA2B	99,70%	97,80%	100%	100%	Glanzmann thrombasthenia, 273800 Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Thrombocytopenia, neonatal alloimmune, BAK antigen related, 0
ITGA3	99,50%	97,40%	100%	100%	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748
ITGA6	99,90%	98,90%	100%	100%	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730
ITGA7	99,60%	98,00%	100%	100%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
ITGA8	99,90%	99,70%	100%	100%	Renal hypodysplasia/aplasia 1, 191830
ITGB2	100%	100%	100%	100%	Leukocyte adhesion deficiency, 116920
ITGB3	100%	99,40%	100%	100%	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Glanzmann thrombasthenia, 273800

					Purpura, posttransfusion, 0 Thrombocytopenia, neonatal alloimmune, 0
ITGB4	98,40%	96,20%	100%	100%	Epidermolysis bullosa of hands and feet, 131800 Epidermolysis bullosa, junctional, with pyloric atresia, 226730 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
ITGB6	97,20%	95,80%	100%	100%	Amelogenesis imperfecta, type IH, 616221
ITK	100%	98,90%	100%	100%	Lymphoproliferative syndrome 1, 613011
ITPA	100%	100%	100%	100%	Epileptic encephalopathy, early infantile, 35, 616647
ITPR1	100%	99,90%	100%	100%	Spinocerebellar ataxia 15, 606658 Gillespie syndrome, 206700 Spinocerebellar ataxia 29, congenital nonprogressive, 117360
IVD	100%	100%	100%	100%	Isovaleric acidemia, 243500
IYD	99,50%	95,70%	100%	100%	Thyroid dyshormonogenesis 4, 274800
JAGN1	100%	100%	100%	100%	Neutropenia, severe congenital, 6, autosomal recessive, 616022
JAK3	99,90%	98,70%	100%	100%	SCID, autosomal recessive, T-negative/B-positive type, 600802
JAM3	100%	99,90%	100%	100%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
JPH1	100%	99,90%	100%	100%	?Charcot-Marie-Tooth disease, axonal, autosomal dominant, type 2K, 607831
JUP	100%	99,50%	100%	100%	Arrhythmogenic right ventricular dysplasia 12, 611528 Naxos disease, 601214
KALRN	99,90%	99,60%	100%	100%	No OMIM disease ID
KANK2	100%	100%	100%	100%	Nephrotic syndrome, type 16, 617783 Palmoplantar keratoderma and woolly hair, 616099
KARS	100%	99,90%	100%	100%	Deafness, autosomal recessive 89, 613916 ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641
KATNB1	100%	99,90%	100%	100%	Lissencephaly 6, with microcephaly, 616212
KCNE1	100%	100%	100%	100%	Long QT syndrome 5, 613695 Jervell and Lange-Nielsen syndrome 2, 612347
KCNJ1	100%	100%	100%	100%	Bartter syndrome, type 2, 241200
KCNJ10	89,30%	89,00%	100%	100%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ11	100%	100%	100%	100%	Maturity-onset diabetes of the young, type 13, 616329 Diabetes, permanent neonatal, with or without neurologic features, 606176 Diabetes mellitus, transient neonatal, 3, 610582 Hyperinsulinemic hypoglycemia, familial, 2, 601820
KCNJ13	100%	100%	100%	100%	Leber congenital amaurosis 16, 614186 Snowflake vitreoretinal degeneration, 193230

KCNMA1	94,40%	93,60%	100%	100%	Liang-Wang syndrome, 618729 Cerebellar atrophy, developmental delay, and seizures, 617643 Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446
KCNQ1	95,50%	94,20%	100%	99,80%	Long QT syndrome 1, 192500 Jervell and Lange-Nielsen syndrome, 220400 Short QT syndrome 2, 609621 Atrial fibrillation, familial, 3, 607554
KCNV2	100%	99,90%	100%	100%	Retinal cone dystrophy 3B, 610356
KCTD7	95,00%	95,00%	100%	100%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM5B	99,50%	97,90%	97,70%	97,70%	Mental retardation, autosomal recessive 65, 618109
KERA	100%	100%	100%	100%	Cornea plana 2, autosomal recessive, 217300
KHDC3L	100%	99,80%	100%	100%	Hydatidiform mole, recurrent, 2, 614293
KIAA0556	100%	99,90%	100%	100%	Joubert syndrome 26, 616784
KIAA0586	97,30%	93,10%	95,80%	95,80%	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546
KIAA0753	100%	99,30%	100%	100%	?Orofaciodigital syndrome XV, 617127
KIAA1109	99,80%	99,20%	100%	100%	Alkuraya-Kucinskas syndrome, 617822
KIAA1549	97,90%	96,40%	98,80%	98,00%	Retinitis pigmentosa 86, 618613
KIF14	99,60%	97,70%	100%	100%	?Meckel syndrome 12, 616258 Microcephaly 20, primary, autosomal recessive, 617914
KIF1A	99,40%	97,10%	100%	100%	NESCAV syndrome, 614255 Spastic paraplegia 30, autosomal dominant, 610357 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal recessive, 610357
KIF1BP	96,10%	96,10%	96,10%	96,10%	Goldberg-Shprintzen megacolon syndrome, 609460
KIF1C	100%	100%	100%	100%	Spastic ataxia 2, autosomal recessive, 611302
KIF7	93,60%	90,60%	99,10%	97,80%	?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131
KISS1R	100%	99,50%	100%	100%	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 ?Precocious puberty, central, 1, 176400
KIZ	100%	99,20%	100%	100%	Retinitis pigmentosa 69, 615780
KL	98,20%	97,20%	98,50%	97,50%	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994
KLC2	99,20%	97,90%	100%	100%	Spastic paraplegia, optic atrophy, and neuropathy, 609541
KLHL3	100%	99,30%	100%	100%	Pseudohypoaldosteronism, type IID, 614495
KLHL40	100%	100%	100%	100%	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	100%	99,90%	100%	100%	Nemaline myopathy 9, 615731

KLHL7	99,90%	99,80%	100%	100%	Retinitis pigmentosa 42, 612943 PERCHING syndrome, 617055
KLK4	100%	100%	100%	100%	Amelogenesis imperfecta, type IIA1, 204700
KLKB1	100%	99,50%	100%	100%	Fletcher factor (prekallikrein) deficiency, 612423
KMT2B	95,80%	94,00%	98,70%	97,90%	Dystonia 28, childhood-onset, 617284
KNL1	99,20%	98,10%	98,90%	98,80%	Microcephaly 4, primary, autosomal recessive, 604321
KPTN	100%	100%	100%	100%	Mental retardation, autosomal recessive 41, 615637
KRT10	100%	99,30%	100%	100%	Epidermolytic hyperkeratosis, 113800 Ichthyosis with confetti, 609165 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602
KRT14	89,00%	81,90%	100%	100%	Naegeli-Franceschetti-Jadassohn syndrome, 161000 Dermatopathia pigmentosa reticularis, 125595 Epidermolysis bullosa simplex, Koebner type, 131900 Epidermolysis bullosa simplex, recessive 1, 601001 Epidermolysis bullosa simplex, Dowling-Meara type, 131760 Epidermolysis bullosa simplex, Weber-Cockayne type, 131800
KRT18	86,70%	70,90%	100%	100%	Cirrhosis, cryptogenic, 215600
KRT5	100%	100%	100%	100%	Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 Epidermolysis bullosa simplex-MCR, 609352 Epidermolysis bullosa simplex-MP, 131960 Dowling-Degos disease 1, 179850 Epidermolysis bullosa simplex, Koebner type, 131900 Epidermolysis bullosa simplex, Dowling-Meara type, 131760 Epidermolysis bullosa simplex, recessive 1, 601001
KRT8	90,60%	69,60%	100%	100%	Cirrhosis, cryptogenic, 215600
KRT85	99,00%	93,60%	100%	100%	Ectodermal dysplasia 4, hair/nail type, 602032
KY	100%	99,70%	100%	100%	Myopathy, myofibrillar, 7, 617114
KYNU	99,60%	97,10%	100%	100%	Vertebral, cardiac, renal, and limb defects syndrome 2, 617661 ?Hydroxykynureninuria, 236800
L2HGDH	99,00%	97,20%	100%	100%	L-2-hydroxyglutaric aciduria, 236792
LAMA1	100%	99,70%	100%	100%	Poretti-Boltshauser syndrome, 615960
LAMA2	100%	99,60%	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855
LAMA3	100%	99,70%	100%	100%	Epidermolysis bullosa, junctional, Herlitz type, 226700 Laryngoonychocutaneous syndrome, 245660 Epidermolysis bullosa, generalized atrophic benign, 226650
LAMB1	100%	99,90%	100%	100%	Lissencephaly 5, 615191

LAMB2	100%	99,70%	100%	100%	Pierson syndrome, 609049 Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199
LAMB3	100%	99,60%	100%	100%	Amelogenesis imperfecta, type IA, 104530 Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LAMC2	99,80%	98,00%	100%	100%	Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa, junctional, Herlitz type, 226700
LAMC3	98,60%	97,10%	100%	99,60%	Cortical malformations, occipital, 614115
LAMTOR2	100%	99,70%	100%	100%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LARGE1	100%	99,60%	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	88,50%	78,40%	100%	100%	Alazami syndrome, 615071
LARS	99,80%	98,40%	100%	100%	?Infantile liver failure syndrome 1, 615438
LARS2	100%	100%	100%	100%	Perrault syndrome 4, 615300 ?Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LAT	100%	99,20%	100%	100%	Immunodeficiency 52, 617514
LBR	99,40%	94,50%	100%	100%	Pelger-Huet anomaly, 169400 Greenberg skeletal dysplasia, 215140 ?Reynolds syndrome, 613471 Pelger-Huet anomaly with mild skeletal anomalies, 618019
LCA5	99,90%	99,20%	100%	100%	Leber congenital amaurosis 5, 604537
LCAT	99,00%	93,80%	100%	100%	Norum disease, 245900 Fish-eye disease, 136120
LCK	98,90%	96,60%	100%	100%	?Immunodeficiency 22, 615758
LCT	99,80%	98,50%	100%	100%	Lactase deficiency, congenital, 223000
LDHA	95,00%	91,70%	100%	100%	Glycogen storage disease XI, 612933
LDHD	100%	99,50%	100%	100%	D-lactic aciduria, 245450
LDLRAP1	98,80%	94,20%	100%	100%	Hypercholesterolemia, familial, 4, 603813
LEMD2	98,70%	92,00%	100%	100%	Cataract 46, juvenile-onset, 212500
LEP	99,90%	97,30%	100%	100%	Obesity, morbid, due to leptin deficiency, 614962
LEPR	94,30%	92,60%	94,60%	94,60%	Obesity, morbid, due to leptin receptor deficiency, 614963
LFNG	87,90%	86,40%	92,20%	87,70%	Spondylocostal dysostosis 3, autosomal recessive, 609813
LGI4	99,90%	97,90%	100%	100%	Arthrogryposis multiplex congenita, neurogenic, with myelin defect, 617468
LHB	90,40%	38,90%	100%	100%	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300
LHCGR	94,10%	92,30%	100%	100%	Leydig cell adenoma, somatic, with precocious puberty, 176410 Precocious puberty, male, 176410 Luteinizing hormone resistance, female, 238320

					Leydig cell hypoplasia with pseudohermaphroditism, 238320 Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320
LHFPL5	100%	100%	100%	100%	Deafness, autosomal recessive 67, 610265
LHX3	96,60%	96,50%	100%	100%	Pituitary hormone deficiency, combined, 3, 221750
LIAS	100%	99,10%	100%	100%	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIFR	99,70%	98,00%	100%	100%	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LIG4	100%	99,90%	100%	100%	LIG4 syndrome, 606593
LIM2	100%	100%	100%	100%	Cataract 19, multiple types, 615277
LIMS2	93,00%	92,70%	99,80%	98,90%	?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827
LINGO1	100%	100%	100%	100%	Mental retardation, autosomal recessive 64, 618103
LINS1	100%	99,10%	100%	100%	Mental retardation, autosomal recessive 27, 614340
LIPA	99,20%	95,20%	95,20%	95,20%	Wolman disease, 278000 Cholesteryl ester storage disease, 278000
LIPC	100%	99,40%	100%	100%	Hepatic lipase deficiency, 614025
LIPE	100%	99,00%	100%	100%	Lipodystrophy, familial partial, type 6, 615980
LIPH	100%	99,80%	100%	100%	Hypotrichosis 7, 604379 Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379
LIPN	100%	98,90%	100%	100%	Ichthyosis, congenital, autosomal recessive 8, 613943
LIPT1	100%	99,90%	100%	100%	Lipoyltransferase 1 deficiency, 616299
LIPT2	94,90%	75,20%	100%	100%	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668
LMAN1	99,80%	99,20%	100%	100%	Combined factor V and VIII deficiency, 227300
LMAN2L	100%	99,70%	100%	100%	?Mental retardation, autosomal recessive, 52, 616887
LMBR1	98,10%	96,20%	98,70%	98,70%	Triphalangeal thumb-polysyndactyly syndrome, 174500 Syndactyly, type IV, 186200 Triphalangeal thumb, type I, 174500 Acheiropody, 200500 Laurin-Sandrow syndrome, 135750 Hypoplastic or aplastic tibia with polydactyly, 188740 Polydactyly, preaxial type II, 174500
LMBRD1	98,50%	93,90%	100%	100%	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LMF1	100%	99,60%	100%	100%	Lipase deficiency, combined, 246650
LMNA	97,40%	91,90%	100%	100%	Muscular dystrophy, congenital, 613205 Lipodystrophy, familial partial, type 2, 151660 Charcot-Marie-Tooth disease, type 2B1, 605588 Cardiomyopathy, dilated, 1A, 115200 Heart-hand syndrome, Slovenian type, 610140 Hutchinson-Gilford progeria, 176670 Restrictive dermopathy, lethal, 275210

					Mandibuloacral dysplasia, 248370 Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 Malouf syndrome, 212112
LMNB2	97,50%	94,30%	98,20%	96,90%	?Epilepsy, progressive myoclonic, 9, 616540
LMOD3	100%	99,70%	100%	100%	Nemaline myopathy 10, 616165
LNPK	98,40%	92,80%	93,30%	93,30%	Neurodevelopmental disorder with epilepsy and hypoplasia of the corpus callosum, 618090
LONP1	100%	99,80%	100%	100%	CODAS syndrome, 600373
LOXHD1	100%	99,70%	100%	100%	Deafness, autosomal recessive 77, 613079
LPAR6	99,60%	97,80%	100%	100%	Hypotrichosis 8, 278150 Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150
LPIN1	99,60%	97,30%	100%	100%	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	100%	100%	100%	100%	Majeed syndrome, 609628
LPL	100%	100%	100%	100%	Lipoprotein lipase deficiency, 238600 Combined hyperlipidemia, familial, 144250
LRAT	100%	100%	100%	100%	Retinal dystrophy, early-onset severe, 613341 Leber congenital amaurosis 14, 613341 Retinitis pigmentosa, juvenile, 613341
LRBA	99,90%	99,60%	100%	100%	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRIG2	99,60%	98,80%	100%	100%	Urofacial syndrome 2, 615112
LRIT3	93,90%	91,90%	100%	100%	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRMDA	96,80%	95,60%	99,60%	99,60%	Albinism, oculocutaneous, type VII, 615179
LRP1	99,70%	98,90%	100%	100%	?Keratosis pilaris atrophicans, 604093
LRP2	100%	99,90%	100%	100%	Donnai-Barrow syndrome, 222448
LRP4	99,10%	98,80%	100%	100%	?Myasthenic syndrome, congenital, 17, 616304 Sclerosteosis 2, 614305 Cenani-Lenz syndactyly syndrome, 212780
LRP5	98,50%	98,10%	100%	99,70%	van Buchem disease, type 2, 607636 Exudative vitreoretinopathy 4, 601813 Hyperostosis, endosteal, 144750 Osteosclerosis, 144750 Polycystic liver disease 4 with or without kidney cysts, 617875 Osteoporosis-pseudoglioma syndrome, 259770 Osteopetrosis, autosomal dominant 1, 607634
LRPAP1	100%	100%	100%	100%	Myopia 23, autosomal recessive, 615431
LRPPRC	99,90%	99,10%	100%	100%	Leigh syndrome, French-Canadian type, 220111
LRRC56	100%	99,00%	100%	100%	Ciliary dyskinesia, primary, 39, 618254
LRRC6	99,20%	96,30%	100%	100%	Ciliary dyskinesia, primary, 19, 614935

LRSAM1	100%	99,90%	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2P, 614436
LRTOMT	99,30%	94,20%	93,30%	93,10%	Deafness, autosomal recessive 63, 611451
LSS	100%	99,90%	100%	100%	Alopecia-mental retardation syndrome 4, 618840 Cataract 44, 616509 Hypotrichosis 14, 618275
LTBP2	99,90%	99,00%	100%	100%	Glaucoma 3, primary congenital, D, 613086 ?Weill-Marchesani syndrome 3, recessive, 614819 Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750
LTBP3	99,60%	98,10%	100%	100%	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
LTBP4	99,90%	97,50%	100%	100%	Cutis laxa, autosomal recessive, type IC, 613177
LTC4S	74,20%	68,50%	100%	100%	No OMIM disease ID
LYRM4	68,50%	66,20%	66,30%	66,30%	?Combined oxidative phosphorylation deficiency 19, 615595
LYRM7	95,90%	86,20%	100%	100%	Mitochondrial complex III deficiency, nuclear type 8, 615838
LYST	99,60%	98,30%	100%	100%	Chediak-Higashi syndrome, 214500
LZTFL1	99,90%	99,20%	100%	100%	Bardet-Biedl syndrome 17, 615994
LZTR1	100%	99,90%	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%	99,90%	100%	100%	?Fanconi anemia, complementation group V, 617243
MAG	100%	100%	100%	100%	Spastic paraplegia 75, autosomal recessive, 616680
MAGI2	94,50%	92,40%	94,70%	93,30%	Nephrotic syndrome, type 15, 617609
MAK	98,70%	96,80%	100%	100%	Retinitis pigmentosa 62, 614181
MALT1	91,20%	89,40%	100%	100%	Immunodeficiency 12, 615468
MAN1B1	100%	99,70%	100%	99,90%	Mental retardation, autosomal recessive 15, 614202
MAN2B1	99,80%	97,90%	100%	100%	Mannosidosis, alpha-, types I and II, 248500
MANBA	99,80%	98,40%	100%	100%	Mannosidosis, beta, 248510
MAP3K20	100%	99,50%	100%	100%	Split-foot malformation with mesoaxial polydactyly, 616890 Centronuclear myopathy 6 with fiber-type disproportion, 617760
MAPKBP1	100%	100%	100%	100%	Nephronophthisis 20, 617271
MAPT	100%	99,50%	100%	100%	Pick disease, 172700 Dementia, frontotemporal, with or without parkinsonism, 600274 Supranuclear palsy, progressive, 601104 Supranuclear palsy, progressive atypical, 260540
MARS	99,70%	97,40%	100%	100%	Interstitial lung and liver disease, 615486 Charcot-Marie-Tooth disease, axonal, type 2U, 616280

MARS2	100%	100%	100%	100%	Spastic ataxia 3, autosomal recessive, 611390 ?Combined oxidative phosphorylation deficiency 25, 616430
MARVELD2	99,20%	96,10%	100%	100%	Deafness, autosomal recessive 49, 610153
MASP1	100%	99,90%	100%	100%	3MC syndrome 1, 257920
MAT1A	99,70%	97,70%	100%	100%	Methionine adenosyltransferase deficiency, autosomal recessive, 250850 Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850
MATN3	84,70%	84,60%	100%	100%	?Spondyloepimetaphyseal dysplasia, 608728 Epiphyseal dysplasia, multiple, 5, 607078
MBOAT7	100%	99,50%	100%	100%	Mental retardation, autosomal recessive 57, 617188
MBTPS1	99,60%	98,40%	100%	100%	?Spondyloepiphyseal dysplasia, Kondo-Fu type, 618392
MC2R	99,90%	98,30%	100%	100%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MCCC1	100%	99,80%	100%	100%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	99,90%	98,40%	100%	100%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	100%	100%	100%	100%	Methylmalonyl-CoA epimerase deficiency, 251120
MCFD2	99,50%	96,90%	100%	100%	Factor V and factor VIII, combined deficiency of, 613625
MCM3AP	99,90%	99,10%	100%	100%	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124
MCM4	100%	99,50%	100%	100%	Immunodeficiency 54, 609981
MCM5	100%	99,70%	100%	100%	?Meier-Gorlin syndrome 8, 617564
MCM9	99,90%	99,80%	100%	100%	Ovarian dysgenesis 4, 616185
MCOLN1	99,80%	98,40%	100%	100%	Mucolipidosis IV, 252650
MCPH1	100%	99,40%	100%	100%	Microcephaly 1, primary, autosomal recessive, 251200
MDH2	98,00%	97,90%	100%	100%	Epileptic encephalopathy, early infantile, 51, 617339
MECR	100%	98,90%	100%	100%	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MED17	96,30%	93,50%	100%	100%	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	100%	99,70%	100%	100%	Mental retardation, autosomal recessive 18, 614249
MED25	100%	99,80%	100%	100%	?Charcot-Marie-Tooth disease, type 2B2, 605589 Basel-Vanagait-Smirin-Yosef syndrome, 616449
MEFV	99,90%	98,60%	96,40%	96,40%	Familial Mediterranean fever, AR, 249100 Familial Mediterranean fever, AD, 134610
MEGF10	100%	100%	100%	100%	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399
MEGF8	99,90%	99,00%	100%	100%	Carpenter syndrome 2, 614976
MEOX1	100%	98,90%	100%	100%	Klippel-Feil syndrome 2, 214300
MERTK	99,50%	98,80%	99,10%	99,10%	Retinitis pigmentosa 38, 613862
MESP2	93,90%	86,90%	97,50%	97,50%	Spondylocostal dysostosis 2, autosomal recessive, 608681

MET	100%	99,50%	100%	100%	Hepatocellular carcinoma, childhood type, somatic, 114550 ?Deafness, autosomal recessive 97, 616705 Renal cell carcinoma, papillary, 1, familial and somatic, 605074
METTL23	100%	100%	100%	100%	Mental retardation, autosomal recessive 44, 615942
MFF	94,30%	89,90%	100%	100%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFN2	100%	99,90%	100%	100%	Hereditary motor and sensory neuropathy VIA, 601152 Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260
MFRP	100%	100%	100%	100%	Nanophthalmos 2, 609549 Microphthalmia, isolated 5, 611040
MFSD2A	99,70%	98,50%	100%	100%	Microcephaly 15, primary, autosomal recessive, 616486
MFSD8	100%	99,70%	100%	100%	Macular dystrophy with central cone involvement, 616170 Ceroid lipofuscinosis, neuronal, 7, 610951
MGAT2	100%	100%	100%	100%	Congenital disorder of glycosylation, type IIa, 212066
MGME1	100%	100%	100%	100%	Mitochondrial DNA depletion syndrome 11, 615084
MGP	98,70%	95,10%	100%	100%	Keutel syndrome, 245150
MICU1	98,90%	95,20%	100%	100%	Myopathy with extrapyramidal signs, 615673
MIPEP	99,20%	96,50%	100%	100%	Combined oxidative phosphorylation deficiency 31, 617228
MITF	100%	99,90%	100%	100%	COMMAD syndrome, 617306 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 Tietz albinism-deafness syndrome, 103500
MKKS	83,20%	83,20%	90,70%	90,70%	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKS1	99,80%	97,90%	100%	100%	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000
MLC1	100%	99,00%	100%	100%	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MLH1	100%	99,90%	100%	100%	Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320 Colorectal cancer, hereditary nonpolyposis, type 2, 609310
MLPH	100%	98,80%	100%	100%	Griscelli syndrome, type 3, 609227
MLYCD	96,00%	90,40%	100%	98,90%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	100%	100%	100%	100%	Methylmalonic aciduria, vitamin B12-responsive, 251100
MMAB	100%	99,60%	100%	100%	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110
MMACHC	100%	100%	100%	100%	Methylmalonic aciduria and homocystinuria, cblC type, 277400

MMADHC	94,40%	83,50%	89,70%	89,70%	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410
MME	99,80%	98,70%	98,00%	98,00%	Charcot-Marie-Tooth disease, axonal, type 2T, 617017 ?Spinocerebellar ataxia 43, 617018
MMP13	95,20%	92,20%	92,40%	92,40%	Metaphyseal anadysplasia 1, 602111 Metaphyseal dysplasia, Spahr type, 250400 Spondyloepimetaphyseal dysplasia, Missouri type, 602111
MMP14	100%	98,90%	100%	100%	?Winchester syndrome, 277950
MMP2	100%	100%	100%	100%	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP20	100%	100%	100%	100%	Amelogenesis imperfecta, type IIA2, 612529
MMP21	99,90%	98,80%	100%	100%	Heterotaxy, visceral, 7, autosomal, 616749
MMP9	99,10%	96,10%	100%	100%	Metaphyseal anadysplasia 2, 613073
MOCOS	99,80%	97,70%	100%	100%	Xanthinuria, type II, 603592
MOCS1	99,20%	95,40%	100%	100%	Molybdenum cofactor deficiency A, 252150
MOCS2	99,60%	99,50%	100%	100%	Molybdenum cofactor deficiency B, 252160
MOGS	100%	99,90%	100%	100%	Congenital disorder of glycosylation, type IIb, 606056
MPC1	100%	99,60%	100%	100%	Mitochondrial pyruvate carrier deficiency, 614741
MPDU1	100%	100%	100%	100%	Congenital disorder of glycosylation, type If, 609180
MPDZ	99,80%	98,80%	100%	100%	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219
MPI	100%	99,90%	100%	100%	Congenital disorder of glycosylation, type Ib, 602579
MPL	100%	99,50%	100%	100%	Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498 Myelofibrosis with myeloid metaplasia, somatic, 254450
MPLKIP	100%	99,40%	100%	100%	Trichothiodystrophy 4, nonphotosensitive, 234050
MPO	100%	99,90%	100%	100%	Myeloperoxidase deficiency, 254600
MPV17	100%	97,20%	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MPZ	100%	100%	100%	100%	Charcot-Marie-Tooth disease, type 2J, 607736 Charcot-Marie-Tooth disease, type 1B, 118200 Dejerine-Sottas disease, 145900 Hypomyelinating neuropathy, congenital, 2, 618184 Charcot-Marie-Tooth disease, dominant intermediate D, 607791 Roussy-Levy syndrome, 180800 Charcot-Marie-Tooth disease, type 2I, 607677
MPZL2	100%	99,90%	100%	100%	Deafness, autosomal recessive 111, 618145
MS4A1	99,80%	98,80%	100%	100%	Immunodeficiency, common variable, 5, 613495

MSH2	99,80%	97,70%	100%	100%	Muir-Torre syndrome, 158320 Mismatch repair cancer syndrome, 276300 Colorectal cancer, hereditary nonpolyposis, type 1, 120435
MSH3	100%	99,20%	100%	100%	Familial adenomatous polyposis 4, 617100 Endometrial carcinoma, somatic, 608089
MSH6	100%	99,80%	100%	100%	Mismatch repair cancer syndrome, 276300 Colorectal cancer, hereditary nonpolyposis, type 5, 614350
MSMO1	96,30%	88,90%	100%	100%	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MSRB3	100%	99,40%	100%	100%	Deafness, autosomal recessive 74, 613718
MSTO1	99,60%	96,70%	100%	100%	Myopathy, mitochondrial, and ataxia, 617675
MTFMT	100%	99,80%	100%	100%	Combined oxidative phosphorylation deficiency 15, 614947 Mitochondrial complex I deficiency, nuclear type 27, 618248
MTHFD1	100%	99,50%	100%	100%	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780
MTHFR	97,30%	96,00%	100%	100%	Homocystinuria due to MTHFR deficiency, 236250
MTHFS	75,00%	74,90%	100%	100%	Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367
MTMR 2	100%	99,00%	100%	100%	Charcot-Marie-Tooth disease, type 4B1, 601382
MTO1	91,30%	90,40%	91,60%	91,40%	Combined oxidative phosphorylation deficiency 10, 614702
MTPAP	99,50%	96,10%	100%	100%	?Spastic ataxia 4, autosomal recessive, 613672
MTR	100%	100%	100%	100%	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940
MTRR	100%	99,60%	100%	100%	Homocystinuria-megaloblastic anemia, cbl E type, 236270
MTTP	100%	99,60%	100%	100%	Abetalipoproteinemia, 200100
MUSK	100%	99,90%	100%	100%	Fetal akinesia deformation sequence 1, 208150 Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325
MUT	99,80%	98,30%	100%	100%	Methylmalonic aciduria, mut(0) type, 251000
MUTYH	100%	100%	100%	100%	Gastric cancer, somatic, 613659 Adenomas, multiple colorectal, 608456
MVK	90,90%	90,50%	90,50%	90,50%	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377
MYBPC1	99,90%	99,50%	100%	100%	Arthrogryposis, distal, type 1B, 614335 Myopathy, congenital, with tremor, 618524 Lethal congenital contracture syndrome 4, 614915
MYD88	100%	100%	100%	100%	Macroglobulinemia, Waldenstrom, somatic, 153600 Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260
MYF5	100%	100%	100%	100%	Ophthalmoplegia, external, with rib and vertebral anomalies, 618155
MYH2	99,90%	99,40%	100%	100%	Proximal myopathy and ophthalmoplegia, 605637

MYH3	99,90%	99,00%	100%	100%	Contractures, pterygia, and spondylocarpotarsal fusion syndrome 1B, 618469 Arthrogryposis, distal, type 2B3 (Sheldon-Hall), 618436 Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700 Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A, 178110
MYL1	100%	99,70%	100%	100%	Myopathy, congenital, with fast-twitch (type II) fiber atrophy, 618414
MYL3	100%	100%	100%	100%	Cardiomyopathy, hypertrophic, 8, 608751
MYLK	100%	99,90%	100%	100%	Aortic aneurysm, familial thoracic 7, 613780 Megacystis-microcolon-intestinal hypoperistalsis syndrome, 249210
MYMK	100%	100%	100%	100%	Carey-Fineman-Ziter syndrome, 254940
MYO15A	98,80%	97,00%	100%	99,90%	Deafness, autosomal recessive 3, 600316
MYO18B	100%	99,10%	100%	100%	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549
MYO1E	99,90%	99,50%	100%	100%	Glomerulosclerosis, focal segmental, 6, 614131
MYO3A	99,60%	96,60%	100%	100%	Deafness, autosomal recessive 30, 607101
MYO5A	99,80%	98,90%	100%	100%	Griscelli syndrome, type 1, 214450
MYO5B	99,10%	96,20%	100%	100%	Microvillus inclusion disease, 251850
MYO6	99,50%	96,60%	100%	100%	Deafness, autosomal recessive 37, 607821 Deafness, autosomal dominant 22, 606346 Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346
MYO7A	99,30%	97,40%	100%	100%	Deafness, autosomal recessive 2, 600060 Deafness, autosomal dominant 11, 601317 Usher syndrome, type 1B, 276900
MYO9A	99,90%	99,10%	100%	100%	Myasthenic syndrome, congenital, 24, presynaptic, 618198
MYPN	100%	99,70%	100%	100%	Cardiomyopathy, dilated, 1KK, 615248 Cardiomyopathy, familial restrictive, 4, 615248 Nemaline myopathy 11, autosomal recessive, 617336 Cardiomyopathy, hypertrophic, 22, 615248
MYSM1	100%	99,10%	100%	100%	Bone marrow failure syndrome 4, 618116
NADK2	99,90%	97,20%	99,00%	96,30%	2,4-dienoyl-CoA reductase deficiency, 616034
NAGA	100%	100%	100%	100%	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241
NAGLU	92,90%	89,90%	99,90%	99,20%	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491
NAGS	99,70%	95,00%	100%	100%	N-acetylglutamate synthase deficiency, 237310
NALCN	100%	99,50%	99,80%	99,80%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419 Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266
NANS	100%	99,90%	100%	100%	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442

NARS2	98,30%	97,40%	100%	100%	?Deafness, autosomal recessive 94, 618434 Combined oxidative phosphorylation deficiency 24, 616239
NAT8L	99,50%	94,20%	95,60%	90,80%	?N-acetylaspartate deficiency, 614063
NAXD	100%	100%	100%	100%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321
NAXE	100%	99,80%	100%	100%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NBAS	100%	99,60%	100%	100%	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NBEAL2	99,40%	99,30%	100%	100%	Gray platelet syndrome, 139090
NBN	99,90%	98,60%	100%	100%	Aplastic anemia, 609135 Nijmegen breakage syndrome, 251260 Leukemia, acute lymphoblastic, 613065
NCAPD2	100%	99,70%	100%	100%	?Microcephaly 21, primary, autosomal recessive, 617983
NCAPD3	99,90%	98,90%	100%	100%	Microcephaly 22, primary, autosomal recessive, 617984
NCAPG2	99,90%	99,20%	100%	100%	Khan-Khan-Katsanis syndrome, 618460
NCAPH	100%	100%	100%	100%	?Microcephaly 23, primary, autosomal recessive, 617985
NCF1	26,00%	25,80%	100%	99,80%	Chronic granulomatous disease due to deficiency of NCF-1, 233700
NCF2	99,90%	98,30%	100%	100%	Chronic granulomatous disease due to deficiency of NCF-2, 233710
NCF4	100%	100%	100%	100%	?Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III, 613960
NDE1	100%	100%	100%	100%	Lissencephaly 4 (with microcephaly), 614019 ?Microhydranencephaly, 605013
NDRG1	100%	100%	100%	100%	Charcot-Marie-Tooth disease, type 4D, 601455
NDST1	100%	100%	100%	100%	Mental retardation, autosomal recessive 46, 616116
NDUFA10	99,80%	98,60%	100%	100%	Mitochondrial complex I deficiency, nuclear type 22, 618243
NDUFA11	100%	100%	100%	99,80%	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFA12	100%	100%	100%	100%	?Mitochondrial complex I deficiency, nuclear type 23, 618244
NDUFA13	92,20%	89,20%	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	99,90%	96,50%	100%	100%	Mitochondrial complex I deficiency, nuclear type 26, 618247
NDUFAF1	100%	100%	100%	100%	Mitochondrial complex I deficiency, nuclear type 11, 618234
NDUFAF2	95,00%	83,40%	100%	99,90%	Mitochondrial complex I deficiency, nuclear type 10, 618233
NDUFAF3	100%	99,90%	100%	100%	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF4	99,80%	98,20%	100%	100%	Mitochondrial complex I deficiency, nuclear type 15, 618237
NDUFAF5	100%	99,50%	100%	100%	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFAF6	100%	96,80%	100%	100%	Mitochondrial complex I deficiency, nuclear type 17, 618239
NDUFB11	99,50%	96,50%	100%	99,50%	Linear skin defects with multiple congenital anomalies 3, 300952 ?Mitochondrial complex I deficiency, nuclear type 30, 301021

NDUFB3	95,80%	80,50%	100%	100%	Mitochondrial complex I deficiency, nuclear type 25, 618246
NDUFB8	100%	99,90%	100%	100%	Mitochondrial complex I deficiency, nuclear type 32, 618252
NDUFB9	98,40%	95,50%	100%	100%	?Mitochondrial complex I deficiency, nuclear type 24, 618245
NDUFS1	100%	99,50%	100%	100%	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	100%	100%	100%	100%	Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	90,70%	90,60%	91,90%	90,70%	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	100%	99,40%	100%	100%	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS6	100%	99,90%	100%	100%	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFS7	100%	99,20%	100%	100%	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFS8	100%	99,40%	100%	100%	Mitochondrial complex I deficiency, nuclear type 2, 618222
NDUFV1	98,00%	96,10%	100%	100%	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	86,90%	76,90%	100%	100%	Mitochondrial complex I deficiency, nuclear type 7, 618229
NEB	83,00%	82,60%	99,90%	99,90%	Nemaline myopathy 2, autosomal recessive, 256030
NECAP1	100%	100%	100%	100%	?Epileptic encephalopathy, early infantile, 21, 615833
NECTIN1	100%	99,90%	100%	100%	Orofacial cleft 7, 225060 Cleft lip/palate-ectodermal dysplasia syndrome, 225060
NECTIN4	100%	100%	100%	100%	Ectodermal dysplasia-syndactyly syndrome 1, 613573
NEK1	99,80%	98,00%	100%	100%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NEK2	99,70%	95,50%	96,10%	96,10%	?Retinitis pigmentosa 67, 615565
NEK8	100%	99,90%	100%	100%	?Nephronophthisis 9, 613824 Renal-hepatic-pancreatic dysplasia 2, 615415
NEK9	100%	99,60%	100%	100%	Lethal congenital contracture syndrome 10, 617022 Nevus comedonicus, somatic, 617025 ?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262
NEU1	99,70%	97,70%	100%	100%	Sialidosis, type II, 256550 Sialidosis, type I, 256550
NEUROG3	100%	100%	100%	100%	Diarrhea 4, malabsorptive, congenital, 610370
NFASC	100%	99,90%	100%	100%	Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356
NFU1	98,80%	90,80%	100%	100%	Multiple mitochondrial dysfunctions syndrome 1, 605711
NGF	100%	100%	100%	100%	Neuropathy, hereditary sensory and autonomic, type V, 608654
NGLY1	100%	99,80%	100%	100%	Congenital disorder of deglycosylation, 615273
NHEJ1	100%	96,20%	100%	100%	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
NHLRC1	100%	98,70%	100%	100%	Epilepsy, progressive myoclonic 2B (Lafora), 254780
NHLRC2	99,60%	98,20%	100%	100%	FINCA syndrome, 618278
NHP2	100%	100%	100%	100%	Dyskeratosis congenita, autosomal recessive 2, 613987
NIN	100%	99,50%	100%	100%	?Seckel syndrome 7, 614851

NIPAL4	100%	99,10%	100%	100%	Ichthyosis, congenital, autosomal recessive 6, 612281
NKX2-6	100%	99,50%	100%	100%	Persistent truncus arteriosus, 217095 Conotruncal heart malformations, 217095
NKX3-2	99,80%	97,00%	100%	100%	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330
NKX6-2	89,00%	81,80%	100%	100%	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560
NLRP1	99,60%	98,00%	100%	100%	Palmoplantar carcinoma, multiple self-healing, 615225 Autoinflammation with arthritis and dyskeratosis, 617388 ?Respiratory papillomatosis, juvenile recurrent, congenital, 618803
NLRP7	100%	99,60%	100%	100%	Hydatidiform mole, recurrent, 1, 231090
NME8	99,20%	95,30%	100%	100%	Ciliary dyskinesia, primary, 6, 610852
NMNAT1	100%	99,20%	98,30%	95,60%	Leber congenital amaurosis 9, 608553
NNT	100%	99,40%	100%	100%	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736
NOP10	100%	99,80%	100%	100%	Dyskeratosis congenita, autosomal recessive 1, 224230
NPC1	99,60%	98,70%	100%	100%	Niemann-Pick disease, type D, 257220 Niemann-Pick disease, type C1, 257220
NPC2	100%	99,60%	100%	100%	Niemann-pick disease, type C2, 607625
NPHP1	100%	99,00%	100%	100%	Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900 Joubert syndrome 4, 609583
NPHP3	99,70%	98,40%	100%	100%	Meckel syndrome 7, 267010 Renal-hepatic-pancreatic dysplasia 1, 208540 Nephronophthisis 3, 604387
NPHP4	100%	99,80%	100%	100%	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
NPHS1	99,80%	99,10%	100%	100%	Nephrotic syndrome, type 1, 256300
NPHS2	100%	99,50%	100%	100%	Nephrotic syndrome, type 2, 600995
NPPA	100%	100%	100%	100%	Atrial standstill 2, 615745 Atrial fibrillation, familial, 6, 612201
NPR2	100%	99,60%	100%	100%	Short stature with nonspecific skeletal abnormalities, 616255 Epiphyseal chondrodysplasia, Miura type, 615923 Acromesomelic dysplasia, Maroteaux type, 602875
NR0B2	100%	99,30%	100%	100%	Obesity, mild, early-onset, 601665
NR1H4	99,80%	98,50%	100%	100%	Cholestasis, progressive familial intrahepatic, 5, 617049
NR2E3	100%	99,60%	100%	100%	Enhanced S-cone syndrome, 268100 Retinitis pigmentosa 37, 611131
NRXN1	97,40%	96,90%	100%	99,80%	Pitt-Hopkins-like syndrome 2, 614325
NSMCE2	99,70%	98,20%	100%	100%	Seckel syndrome 10, 617253
NSMCE3	100%	100%	100%	100%	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241

NSUN2	96,00%	95,10%	100%	100%	Mental retardation, autosomal recessive 5, 611091
NT5C2	98,00%	96,50%	100%	100%	Spastic paraplegia 45, autosomal recessive, 613162
NT5C3A	97,90%	88,30%	100%	100%	Anemia, hemolytic, due to UMPH1 deficiency, 266120
NT5E	100%	99,90%	100%	100%	Calcification of joints and arteries, 211800
NTHL1	100%	99,80%	100%	100%	Familial adenomatous polyposis 3, 616415
NTRK1	99,80%	98,20%	100%	100%	Insensitivity to pain, congenital, with anhidrosis, 256800
NUBPL	99,70%	98,40%	100%	100%	Mitochondrial complex I deficiency, nuclear type 21, 618242
NUP107	99,80%	98,50%	100%	100%	Galloway-Mowat syndrome 7, 618348 ?Ovarian dysgenesis 6, 618078 Nephrotic syndrome, type 11, 616730
NUP133	99,70%	98,30%	100%	100%	Nephrotic syndrome, type 18, 618177 ?Galloway-Mowat syndrome 8, 618349
NUP160	100%	99,90%	100%	100%	?Nephrotic syndrome, type 19, 618178
NUP205	99,90%	99,40%	100%	100%	?Nephrotic syndrome, type 13, 616893
NUP214	100%	99,70%	100%	100%	Leukemia, acute myeloid, somatic, 601626 Leukemia, T-cell acute lymphoblastic, somatic, 613065
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	100%	100%	100%	100%	Fetal akinesia deformation sequence 4, 618393
NUP93	98,00%	94,20%	95,50%	95,50%	Nephrotic syndrome, type 12, 616892
NUS1	60,00%	44,50%	100%	100%	Mental retardation, autosomal dominant 55, with seizures, 617831 ?Congenital disorder of glycosylation, type 1aa, 617082
NXN	100%	100%	99,90%	99,50%	Robinow syndrome, autosomal recessive 2, 618529
OAT	85,20%	76,30%	100%	100%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OBSL1	100%	99,30%	100%	100%	3-M syndrome 2, 612921
OCA2	99,90%	98,70%	100%	100%	Albinism, oculocutaneous, type II, 203200 Albinism, brown oculocutaneous, 203200
OCLN	100%	100%	100%	100%	Pseudo-TORCH syndrome 1, 251290
OGDH	100%	99,90%	100%	100%	No OMIM disease ID
OPA1	99,70%	97,60%	100%	100%	Behr syndrome, 210000 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896
OPA3	100%	99,00%	100%	100%	Optic atrophy 3 with cataract, 165300 3-methylglutaconic aciduria, type III, 258501
OPLAH	100%	99,80%	100%	100%	5-oxoprolinase deficiency, 260005

ORAI1	95,80%	92,80%	97,20%	92,40%	Myopathy, tubular aggregate, 2, 615883 Immunodeficiency 9, 612782
ORC1	100%	99,40%	100%	100%	Meier-Gorlin syndrome 1, 224690
ORC4	98,70%	93,60%	100%	100%	Meier-Gorlin syndrome 2, 613800
ORC6	100%	99,90%	100%	100%	Meier-Gorlin syndrome 3, 613803
OSGEP	100%	99,40%	100%	100%	Galloway-Mowat syndrome 3, 617729
OSTM1	98,60%	94,00%	100%	100%	Osteopetrosis, autosomal recessive 5, 259720
OTOA	99,40%	97,60%	100%	99,90%	Deafness, autosomal recessive 22, 607039
OTOF	100%	99,90%	100%	100%	Auditory neuropathy, autosomal recessive, 1, 601071 Deafness, autosomal recessive 9, 601071
OTOG	99,40%	98,60%	100%	99,90%	Deafness, autosomal recessive 18B, 614945
OTOGL	99,50%	97,40%	100%	100%	Deafness, autosomal recessive 84B, 614944
OTUD6B	99,90%	98,80%	100%	100%	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452
OTULIN	92,60%	86,50%	99,20%	95,00%	Autoinflammation, panniculitis, and dermatosis syndrome, 617099
OXCT1	99,80%	98,10%	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	99,80%	98,00%	100%	100%	Myopia, high, with cataract and vitreoretinal degeneration, 614292
P4HTM	99,00%	97,40%	100%	99,40%	Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493
PAH	100%	100%	100%	100%	Phenylketonuria, 261600
PAM16	65,30%	65,20%	82,90%	82,90%	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320
PANK2	100%	99,30%	100%	100%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PAPSS2	100%	99,50%	100%	100%	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847
PARK7	100%	100%	100%	100%	Parkinson disease 7, autosomal recessive early-onset, 606324
PARN	100%	99,90%	100%	100%	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371 Dyskeratosis congenita, autosomal recessive 6, 616353
PARS2	100%	100%	100%	100%	Epileptic encephalopathy, early infantile, 75, 618437
PATL2	100%	99,00%	100%	100%	Oocyte maturation defect 4, 617743
PAX1	92,40%	87,90%	100%	99,60%	Otofaciocervical syndrome 2, 615560
PAX3	100%	99,90%	100%	100%	Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220 Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820
PAX7	100%	100%	100%	100%	Myopathy, congenital, progressive, with scoliosis, 618578 Rhabdomyosarcoma 2, alveolar, 268220

PC	99,80%	97,30%	100%	100%	Pyruvate carboxylase deficiency, 266150
PCBD1	100%	99,60%	100%	99,70%	Hyperphenylalaninemia, BH4-deficient, D, 264070
PCCA	99,50%	96,70%	100%	100%	Propionicacidemia, 606054
PCCB	97,90%	96,00%	98,70%	96,20%	Propionicacidemia, 606054
PCDH12	100%	100%	100%	100%	Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280
PCDH15	98,60%	97,50%	100%	100%	Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1F, 602083 Deafness, autosomal recessive 23, 609533
PCK1	100%	100%	100%	100%	?Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680
PCK2	100%	100%	100%	100%	No OMIM disease ID
PCLO	99,70%	98,70%	100%	100%	?Pontocerebellar hypoplasia, type 3, 608027
PCNA	100%	98,40%	100%	100%	?Ataxia-telangiectasia-like disorder 2, 615919
PCNT	99,60%	97,10%	100%	100%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PCSK1	100%	99,50%	100%	100%	Obesity with impaired prohormone processing, 600955
PCYT1A	98,90%	95,50%	100%	100%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDE10A	81,20%	80,50%	100%	100%	Dyskinesia, limb and orofacial, infantile-onset, 616921 Striatal degeneration, autosomal dominant, 616922
PDE6A	100%	99,60%	100%	100%	Retinitis pigmentosa 43, 613810
PDE6B	100%	99,90%	100%	100%	Night blindness, congenital stationary, autosomal dominant 2, 163500 Retinitis pigmentosa-40, 613801
PDE6C	99,90%	97,80%	100%	100%	Cone dystrophy 4, 613093
PDE6D	100%	100%	100%	100%	?Joubert syndrome 22, 615665
PDE6G	100%	99,70%	100%	100%	Retinitis pigmentosa 57, 613582
PDE6H	100%	97,90%	100%	100%	Retinal cone dystrophy 3, 610024 Achromatopsia 6, 610024
PDHB	99,10%	97,50%	100%	100%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	99,90%	99,40%	100%	100%	Lacticacidemia due to PDX1 deficiency, 245349
PDP1	100%	100%	100%	100%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	94,70%	87,60%	97,30%	96,60%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	99,80%	97,10%	100%	100%	Coenzyme Q10 deficiency, primary, 3, 614652
PDX1	93,00%	82,40%	100%	100%	Pancreatic agenesis 1, 260370 MODY, type IV, 606392
PDXK	79,30%	76,60%	99,40%	96,70%	Neuropathy, hereditary motor and sensory, type VIC, with optic atrophy, 618511
PDZD7	98,80%	96,30%	100%	100%	Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472 Deafness, autosomal recessive 57, 618003
PEPD	100%	98,80%	100%	100%	Prolidase deficiency, 170100
PET100	100%	99,60%	100%	100%	Mitochondrial complex IV deficiency, 220110

PEX1	99,90%	99,40%	100%	100%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	96,80%	89,70%	100%	99,90%	Peroxisome biogenesis disorder 6B, 614871 Peroxisome biogenesis disorder 6A (Zellweger), 614870
PEX11B	100%	99,60%	100%	100%	?Peroxisome biogenesis disorder 14B, 614920
PEX12	100%	100%	100%	100%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	100%	100%	100%	100%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	96,70%	90,80%	100%	100%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	97,90%	94,20%	100%	100%	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	99,90%	98,50%	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%	100%	100%	100%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	100%	99,30%	100%	100%	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370
PEX5	99,90%	99,00%	100%	100%	Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716 Peroxisome biogenesis disorder 2A (Zellweger), 214110
PEX6	94,50%	86,70%	100%	100%	Peroxisome biogenesis disorder 4B, 614863 Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862
PEX7	87,80%	80,70%	91,30%	91,30%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PFKM	100%	99,50%	100%	100%	Glycogen storage disease VII, 232800
PGAM2	100%	100%	100%	100%	Glycogen storage disease X, 261670
PGAP1	99,00%	94,40%	100%	100%	Mental retardation, autosomal recessive 42, 615802
PGAP2	100%	99,90%	100%	100%	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGAP3	63,50%	59,60%	100%	100%	Hyperphosphatasia with mental retardation syndrome 4, 615716
PGM1	100%	100%	100%	100%	Congenital disorder of glycosylation, type It, 614921
PGM3	100%	99,80%	100%	100%	Immunodeficiency 23, 615816
PHGDH	99,90%	98,80%	100%	100%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHKB	99,90%	99,20%	100%	100%	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750

PHKG2	100%	99,90%	100%	100%	Glycogen storage disease IXc, 613027 Cirrhosis due to liver phosphorylase kinase deficiency, 0
PHOX2A	91,60%	72,70%	100%	99,80%	Fibrosis of extraocular muscles, congenital, 2, 602078
PHYH	100%	99,60%	100%	100%	Refsum disease, 266500
PI4KA	92,60%	88,80%	99,90%	99,90%	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531
TAC3	100%	99,60%	100%	100%	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACO1	98,40%	93,00%	100%	100%	Mitochondrial complex IV deficiency, 220110
TACR3	100%	100%	100%	100%	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
TACSTD2	99,00%	96,40%	100%	100%	Corneal dystrophy, gelatinous drop-like, 204870
TAF13	100%	100%	100%	100%	Mental retardation, autosomal recessive 60, 617432
TAF2	99,90%	98,60%	100%	100%	Mental retardation, autosomal recessive 40, 615599
TAF6	99,80%	98,90%	100%	100%	Alazami-Yuan syndrome, 617126
TALDO1	100%	97,90%	100%	100%	Transaldolase deficiency, 606003
TANGO2	100%	99,30%	100%	100%	metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TAP1	100%	99,20%	100%	100%	Bare lymphocyte syndrome, type I, 604571
TAP2	99,90%	99,30%	100%	100%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	96,50%	95,50%	96,60%	96,60%	Bare lymphocyte syndrome, type I, 604571
TAPT1	91,70%	86,90%	98,50%	94,80%	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897
TARS2	100%	99,30%	100%	100%	?Combined oxidative phosphorylation deficiency 21, 615918
TAT	100%	100%	100%	100%	Tyrosinemia, type II, 276600
TBC1D20	94,20%	94,20%	100%	99,90%	Warburg micro syndrome 4, 615663
TBC1D23	99,70%	97,20%	100%	100%	Pontocerebellar hypoplasia, type 11, 617695
PIBF1	99,50%	96,20%	100%	100%	Joubert syndrome 33, 617767
PIEZO1	99,90%	98,80%	100%	100%	Lymphatic malformation 6, 616843 Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380
PIEZO2	100%	99,50%	100%	100%	Arthrogryposis, distal, with impaired proprioception and touch, 617146 Arthrogryposis, distal, type 5, 108145 ?Marden-Walker syndrome, 248700 Arthrogryposis, distal, type 3, 114300
PIGB	99,90%	97,80%	100%	100%	Epileptic encephalopathy, early infantile, 80, 618580
PIGC	99,20%	90,90%	100%	100%	Glycosylphosphatidylinositol biosynthesis defect 16, 617816
PIGG	100%	99,70%	100%	100%	Mental retardation, autosomal recessive 53, 616917
PIGH	82,10%	68,10%	75,20%	74,40%	Glycosylphosphatidylinositol biosynthesis defect 17, 618010
PIGL	100%	100%	100%	100%	CHIME syndrome, 280000
PIGM	100%	100%	100%	100%	Glycosylphosphatidylinositol deficiency, 610293

PIGN	93,80%	91,50%	98,80%	98,80%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	100%	99,90%	100%	100%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGP	95,80%	87,30%	100%	100%	Epileptic encephalopathy, early infantile, 55, 617599
PIGQ	92,80%	90,80%	100%	100%	Epileptic encephalopathy, early infantile, 77, 618548
PIGS	100%	100%	100%	100%	Glycosylphosphatidylinositol biosynthesis defect 18, 618143
PIGT	98,10%	98,10%	100%	100%	?Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGU	100%	99,10%	100%	100%	Glycosylphosphatidylinositol biosynthesis defect 21, 618590
PIGV	100%	100%	100%	100%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIGW	100%	99,80%	100%	100%	Glycosylphosphatidylinositol biosynthesis defect 11, 616025
PIGY	100%	99,90%	100%	100%	Hyperphosphatasia with mental retardation syndrome 6, 616809
PIK3C2A	99,20%	96,90%	100%	100%	Oculoskeletodental syndrome, 618440
PIK3R1	99,80%	99,00%	100%	100%	SHORT syndrome, 269880 Immunodeficiency 36, 616005 ?Agammaglobulinemia 7, autosomal recessive, 615214
PIK3R5	100%	99,90%	100%	100%	Ataxia-oculomotor apraxia 3, 615217
PINK1	90,70%	86,90%	99,90%	99,40%	Parkinson disease 6, early onset, 605909
PIP5K1C	98,00%	95,80%	99,90%	99,80%	Lethal congenital contractural syndrome 3, 611369
PKD1L1	100%	99,80%	100%	100%	Heterotaxy, visceral, 8, autosomal, 617205
PKHD1	100%	99,60%	100%	100%	Polycystic kidney disease 4, with or without hepatic disease, 263200
PKLR	100%	99,20%	100%	100%	Pyruvate kinase deficiency, 266200 Adenosine triphosphate, elevated, of erythrocytes, 102900
PKP1	100%	99,10%	100%	100%	Ectodermal dysplasia/skin fragility syndrome, 604536
PLA2G6	99,90%	98,30%	100%	100%	Infantile neuroaxonal dystrophy 1, 256600 Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217
PLA2G7	99,90%	99,00%	100%	100%	Platelet-activating factor acetylhydrolase deficiency, 614278
PLAA	100%	99,20%	100%	100%	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527
PLCB1	100%	99,80%	100%	100%	Epileptic encephalopathy, early infantile, 12, 613722
PLCD1	99,90%	97,80%	100%	100%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCE1	99,90%	99,30%	100%	100%	Nephrotic syndrome, type 3, 610725
PLD1	100%	99,60%	100%	100%	Cardiac valvular defect, developmental, 212093
PLEC	100%	99,80%	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723 Epidermolysis bullosa simplex with pyloric atresia, 612138 Epidermolysis bullosa simplex with muscular dystrophy, 226670 ?Epidermolysis bullosa simplex with nail dystrophy, 616487 Epidermolysis bullosa simplex, Ogna type, 131950

PLEKHG2	100%	99,30%	100%	100%	Leukodystrophy and acquired microcephaly with or without dystonia, 616763
PLEKHG5	98,90%	94,60%	100%	99,90%	Spinal muscular atrophy, distal, autosomal recessive, 4, 611067 Charcot-Marie-Tooth disease, recessive intermediate C, 615376
PLEKHM1	100%	99,80%	100%	100%	Osteopetrosis, autosomal dominant 3, 618107 ?Osteopetrosis, autosomal recessive 6, 611497
PLG	87,80%	87,50%	100%	100%	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PLK4	99,90%	98,20%	100%	100%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLOD1	100%	98,40%	100%	100%	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD2	99,30%	97,30%	100%	100%	Bruck syndrome 2, 609220
PLOD3	99,80%	98,00%	100%	100%	Lysyl hydroxylase 3 deficiency, 612394
PLPBP	98,20%	90,10%	100%	100%	Epilepsy, early-onset, vitamin B6-dependent, 617290
PLVAP	100%	100%	100%	100%	Diarrhea 10, protein-losing enteropathy type, 618183
PMM2	100%	100%	100%	100%	Congenital disorder of glycosylation, type Ia, 212065
PMPCA	97,70%	94,20%	100%	100%	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMPCB	100%	99,70%	100%	100%	Multiple mitochondrial dysfunctions syndrome 6, 617954
PMS2	84,30%	82,80%	100%	100%	Mismatch repair cancer syndrome, 276300 Colorectal cancer, hereditary nonpolyposis, type 4, 614337
PNKP	100%	100%	100%	100%	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNLIP	100%	99,80%	100%	100%	?Pancreatic lipase deficiency, 614338
PNP	99,80%	98,90%	100%	100%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA1	100%	100%	100%	100%	Ichthyosis, congenital, autosomal recessive 10, 615024
PNPLA2	99,70%	96,10%	100%	100%	Neutral lipid storage disease with myopathy, 610717
PNPLA6	100%	99,70%	100%	100%	Spastic paraplegia 39, autosomal recessive, 612020 Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 ?Laurence-Moon syndrome, 245800
PNPLA8	100%	99,80%	100%	100%	?Mitochondrial myopathy with lactic acidosis, 251950
PNPO	99,90%	97,70%	100%	100%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
PNPT1	97,70%	89,70%	100%	100%	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	99,80%	98,80%	100%	100%	Cone-rod dystrophy 20, 615973
POGLUT1	99,40%	94,60%	100%	100%	?Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232 Dowling-Degos disease 4, 615696

POLE	100%	99,80%	100%	100%	FILS syndrome, 615139 IMAGE-I syndrome, 618336
POLG	100%	99,30%	100%	100%	Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
POLH	100%	99,60%	100%	100%	Xeroderma pigmentosum, variant type, 278750
POLR1C	99,30%	95,50%	90,70%	90,70%	Treacher Collins syndrome 3, 248390 Leukodystrophy, hypomyelinating, 11, 616494
POLR1D	91,60%	91,60%	100%	100%	Treacher Collins syndrome 2, 613717
POLR3A	100%	99,70%	100%	100%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 Wiedemann-Rautenstrauch syndrome, 264090
POLR3B	99,90%	98,60%	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%	99,90%	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 (congenital with brain and eye anomalies, type A, 8, 614830 Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135
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	100%	99,10%	100%	100%	Proteasome-associated autoinflammatory syndrome 2, 618048 Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952
POMT1	99,30%	97,50%	100%	100%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155
POMT2	99,40%	96,40%	100%	100%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156
POP1	100%	99,70%	100%	100%	Anauxetic dysplasia 2, 617396
POR	99,80%	98,60%	100%	100%	Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571 Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750
POU1F1	100%	99,20%	100%	100%	Pituitary hormone deficiency, combined, 1, 613038

PPA2	98,70%	94,00%	100%	100%	?Sudden cardiac failure, alcohol-induced, 617223 Sudden cardiac failure, infantile, 617222
PPCS	99,80%	99,50%	100%	100%	Cardiomyopathy, dilated, 2C, 618189
PPIB	100%	100%	100%	100%	Osteogenesis imperfecta, type IX, 259440
PPIP5K2	98,90%	95,20%	100%	100%	Deafness, autosomal recessive 100, 618422
PPM1K	100%	100%	100%	100%	?Maple syrup urine disease, mild variant, 615135
PPP1R15B	100%	99,60%	100%	100%	Microcephaly, short stature, and impaired glucose metabolism 2, 616817
PPP2R3C	99,60%	94,90%	100%	100%	Spermatogenic failure 36, 618420 Gonadal dysgenesis, dysmorphic facies, retinal dystrophy, and myopathy, 618419
PPT1	90,30%	90,30%	82,50%	82,50%	Ceroid lipofuscinosis, neuronal, 1, 256730
PRCD	100%	100%	100%	100%	Retinitis pigmentosa 36, 610599
PRDM12	90,80%	88,00%	93,40%	91,70%	Neuropathy, hereditary sensory and autonomic, type VIII, 616488
PRDM5	99,90%	99,20%	100%	100%	Brittle cornea syndrome 2, 614170
PRDM8	92,90%	88,60%	100%	99,80%	?Epilepsy, progressive myoclonic, 10, 616640
PRDX1	100%	100%	100%	100%	Methylmalonic aciduria and homocystinuria, cblC type, digenic, 277400
PREPL	99,80%	98,20%	100%	100%	Myasthenic syndrome, congenital, 22, 616224
PRF1	91,20%	90,80%	100%	100%	Aplastic anemia, 609135 Lymphoma, non-Hodgkin, 605027 Hemophagocytic lymphohistiocytosis, familial, 2, 603553
PRG4	87,40%	80,90%	100%	100%	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250
PRICKLE1	100%	100%	100%	100%	Epilepsy, progressive myoclonic 1B, 612437
PRKCD	100%	100%	100%	100%	Autoimmune lymphoproliferative syndrome, type III, 615559
PRKDC	99,70%	98,00%	100%	100%	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PRKN	79,80%	78,80%	89,80%	89,80%	Parkinson disease, juvenile, type 2, 600116 Ovarian cancer, somatic, 167000 Adenocarcinoma of lung, somatic, 211980
PRKRA	100%	99,40%	100%	100%	Dystonia 16, 612067
PRMT7	100%	99,90%	100%	100%	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157
PROC	100%	100%	100%	100%	Thrombophilia due to protein C deficiency, autosomal dominant, 176860 Thrombophilia due to protein C deficiency, autosomal recessive, 612304
PRODH	85,00%	80,60%	100%	100%	Hyperprolinemia, type I, 239500
PROM1	97,20%	96,10%	100%	100%	Retinitis pigmentosa 41, 612095 Stargardt disease 4, 603786 Cone-rod dystrophy 12, 612657 Macular dystrophy, retinal, 2, 608051
PROP1	92,60%	82,60%	100%	100%	Pituitary hormone deficiency, combined, 2, 262600

PROS1	98,80%	96,10%	100%	100%	Thrombophilia due to protein S deficiency, autosomal recessive, 614514 Thrombophilia due to protein S deficiency, autosomal dominant, 612336
PRSS12	100%	99,90%	100%	100%	Mental retardation, autosomal recessive 1, 249500
PRSS56	99,90%	96,40%	100%	100%	Microphthalmia, isolated 6, 613517
PRUNE1	100%	99,90%	100%	100%	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481
PRX	100%	100%	100%	100%	Charcot-Marie-Tooth disease, type 4F, 614895 Dejerine-Sottas disease, 145900
PSAP	100%	100%	100%	100%	Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Combined SAP deficiency, 611721 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PSAT1	95,30%	81,60%	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	99,90%	98,50%	100%	100%	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040
PSMB9	99,90%	97,70%	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	95,80%	85,60%	98,60%	93,30%	Pancreatic and cerebellar agenesis, 609069 Pancreatic agenesis 2, 615935
PTH1R	100%	98,70%	100%	100%	Metaphyseal chondrodysplasia, Murk Jansen type, 156400 Failure of tooth eruption, primary, 125350 Eiken syndrome, 600002 Chondrodysplasia, Blomstrand type, 215045
PTPN14	99,70%	97,40%	100%	100%	Choanal atresia and lymphedema, 613611
PTPRC	99,00%	95,10%	100%	100%	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971
PTPRO	99,90%	99,40%	100%	100%	Nephrotic syndrome, type 6, 614196
PTPRQ	94,60%	92,50%	92,80%	92,70%	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	99,90%	99,10%	100%	100%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUS1	100%	99,50%	99,60%	97,20%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PUS3	100%	100%	100%	100%	Mental retardation, autosomal recessive 55, 617051
PUS7	100%	99,80%	100%	100%	Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342
PXDN	100%	99,60%	100%	100%	Anterior segment dysgenesis 7, with sclerocornea, 269400
PYCR1	99,90%	97,70%	100%	100%	Cutis laxa, autosomal recessive, type IIIB, 614438 Cutis laxa, autosomal recessive, type IIB, 612940
PYCR2	100%	99,10%	100%	100%	Leukodystrophy, hypomyelinating, 10, 616420

PYGL	100%	100%	100%	100%	Glycogen storage disease VI, 232700
PYGM	100%	99,90%	100%	100%	McArdle disease, 232600
PYROXD1	95,20%	83,90%	100%	100%	Myopathy, myofibrillar, 8, 617258
QARS	100%	100%	100%	100%	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	100%	99,70%	100%	100%	Hyperphenylalaninemia, BH4-deficient, C, 261630
RAB18	99,50%	97,40%	100%	100%	Warburg micro syndrome 3, 614222
RAB23	100%	99,50%	100%	100%	Carpenter syndrome, 201000
RAB27A	100%	100%	100%	100%	Griscelli syndrome, type 2, 607624
RAB28	99,70%	96,00%	100%	100%	Cone-rod dystrophy 18, 615374
RAB33B	100%	100%	100%	100%	Smith-McCort dysplasia 2, 615222
RAB3GAP1	99,40%	98,90%	99,40%	99,40%	Warburg micro syndrome 1, 600118
RAB3GAP2	99,50%	97,00%	100%	100%	Warburg micro syndrome 2, 614225 Martsolf syndrome, 212720
RAD50	97,50%	91,60%	100%	100%	Nijmegen breakage syndrome-like disorder, 613078
RAD51C	100%	99,80%	100%	100%	Fanconi anemia, complementation group O, 613390
RAG1	100%	100%	100%	100%	Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457 Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889 Combined cellular and humoral immune defects with granulomas, 233650
RAG2	100%	100%	100%	100%	Severe combined immunodeficiency, B cell-negative, 601457 Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554
RAPSN	100%	99,70%	100%	100%	Fetal akinesia deformation sequence 2, 618388 Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326
RARB	100%	100%	100%	100%	Microphthalmia, syndromic 12, 615524
RARS	94,20%	91,60%	94,40%	94,30%	Leukodystrophy, hypomyelinating, 9, 616140
RARS2	100%	99,80%	100%	100%	Pontocerebellar hypoplasia, type 6, 611523
RASGRP1	100%	99,60%	100%	100%	Immunodeficiency 64, 618534
RASGRP2	99,70%	97,30%	100%	100%	?Bleeding disorder, platelet-type, 18, 615888
RAX	96,00%	87,00%	100%	98,40%	Microphthalmia, isolated 3, 611038
RBBP8	100%	99,70%	100%	100%	Jawad syndrome, 251255 Seckel syndrome 2, 606744 Pancreatic carcinoma, somatic, 0
RBCK1	99,90%	98,20%	100%	100%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RBM28	100%	100%	100%	100%	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RBM8A	99,80%	97,90%	100%	100%	Thrombocytopenia-absent radius syndrome, 274000

RBP3	100%	100%	100%	100%	?Retinitis pigmentosa 66, 615233
RBP4	99,90%	97,70%	100%	100%	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RCBTB1	99,90%	99,50%	100%	100%	Retinal dystrophy with or without extraocular anomalies, 617175
RD3	100%	100%	100%	100%	Leber congenital amaurosis 12, 610612
RDH11	100%	99,00%	100%	100%	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108
RDH12	100%	98,60%	100%	100%	Leber congenital amaurosis 13, 612712
RDH5	100%	99,90%	100%	100%	Fundus albipunctatus, 136880
RDX	89,10%	71,50%	100%	100%	Deafness, autosomal recessive 24, 611022
RECQL4	99,80%	98,10%	100%	99,90%	RAPADILINO syndrome, 266280 Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2,, 268400
REEP2	99,90%	98,60%	100%	100%	?Spastic paraplegia 72, autosomal dominant, 615625 ?Spastic paraplegia 72, autosomal recessive, 615625
REEP6	100%	100%	100%	100%	Retinitis pigmentosa 77, 617304
RELB	98,80%	88,70%	100%	100%	?Immunodeficiency 53, 617585
RELN	100%	99,80%	100%	100%	Lissencephaly 2 (Norman-Roberts type), 257320
REN	100%	100%	100%	100%	Renal tubular dysgenesis, 267430 Hyperuricemic nephropathy, familial juvenile 2, 613092
REPS1	99,60%	97,50%	100%	100%	?Neurodegeneration with brain iron accumulation 7, 617916
RETREG1	98,80%	95,10%	100%	100%	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
RFC1	99,90%	98,90%	100%	100%	Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome, 614575
RFT1	99,80%	99,60%	100%	100%	Congenital disorder of glycosylation, type In, 612015
RFWD3	100%	99,80%	100%	100%	?Fanconi anemia, complementation group W, 617784
RFX5	99,70%	98,10%	100%	100%	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFX6	100%	99,60%	100%	100%	Mitchell-Riley syndrome, 615710
RFXANK	100%	99,50%	100%	100%	MHC class II deficiency, complementation group B, 209920
RFXAP	99,30%	97,00%	100%	99,90%	Bare lymphocyte syndrome, type II, complementation group D, 209920
RGR	100%	99,10%	100%	100%	Retinitis pigmentosa 44, 613769
RHO	100%	100%	100%	100%	Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis punctata albescens, 136880 Retinitis pigmentosa 4, autosomal dominant or recessive, 613731
RIN2	100%	100%	100%	100%	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075
RINT1	99,90%	98,60%	100%	100%	Infantile liver failure syndrome 3, 618641

RMRP	NC	NC	NC	NC	Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RIPK1	100%	99,00%	100%	100%	Immunodeficiency 57 with autoinflammation, 618108 Autoinflammation with episodic fever and lymphadenopathy, 618852
RIPK4	100%	99,90%	100%	100%	Popliteal pterygium syndrome, Bartsocas-Papas type, 263650 CHAND syndrome, 214350
RIPOR2	100%	99,80%	100%	100%	?Deafness, autosomal recessive 104, 616515
RIPPLY2	100%	97,90%	100%	100%	?Spondylocostal dysostosis 6, 616566
RLBP1	100%	99,90%	100%	100%	Fundus albipunctatus, 136880 Retinitis punctata albescens, 136880 Bothnia retinal dystrophy, 607475 Newfoundland rod-cone dystrophy, 607476
RMND1	100%	98,60%	100%	100%	Combined oxidative phosphorylation deficiency 11, 614922
RNASEH1	98,50%	95,30%	100%	100%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479
RNASEH2A	100%	100%	100%	100%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	96,00%	92,50%	100%	99,80%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	100%	99,50%	100%	100%	Aicardi-Goutieres syndrome 3, 610329
RNASET2	97,40%	93,10%	100%	100%	Leukoencephalopathy, cystic, without megalencephaly, 612951
RNF168	100%	99,80%	100%	100%	RIDDLE syndrome, 611943
RNF216	99,80%	98,70%	100%	100%	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840
RNPC3	91,50%	70,70%	100%	100%	?Growth hormone deficiency, isolated, type V, 618160
ROBO3	98,90%	96,10%	100%	100%	Gaze palsy, familial horizontal, with progressive scoliosis, 1, 607313
ROGDI	98,40%	95,20%	99,90%	99,10%	Kohlschutter-Tonz syndrome, 226750
ROR1	97,00%	96,80%	99,90%	99,30%	?Deafness, autosomal recessive 108, 617654
ROR2	100%	99,90%	97,00%	97,00%	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RORC	100%	100%	100%	100%	Immunodeficiency 42, 616622
RP1	91,50%	90,60%	100%	100%	Retinitis pigmentosa 1, 180100
RPE65	99,80%	97,80%	100%	100%	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 87 with choroidal involvement, 618697 Retinitis pigmentosa 20, 613794
RPGRIP1	100%	99,90%	100%	100%	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826
RPGRIP1L	96,70%	95,70%	100%	99,50%	COACH syndrome, 216360 Meckel syndrome 5, 611561 Joubert syndrome 7, 611560
RPIA	98,60%	94,90%	100%	100%	Ribose 5-phosphate isomerase deficiency, 608611

RRM2B	100%	99,70%	100%	100%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075
RSPH1	100%	100%	100%	100%	Ciliary dyskinesia, primary, 24, 615481
RSPH3	99,90%	98,80%	100%	100%	Ciliary dyskinesia, primary, 32, 616481
RSPH4A	98,10%	95,60%	100%	100%	Ciliary dyskinesia, primary, 11, 612649
RSPH9	99,90%	97,90%	100%	100%	Ciliary dyskinesia, primary, 12, 612650
RSPO1	100%	99,90%	100%	100%	Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644 Palmoplantar hyperkeratosis and true hermaphroditism, 610644
RSPO2	97,10%	90,70%	100%	100%	Tetraamelia syndrome 2, 618021 ?Humerofemoral hypoplasia with radiotibial ray deficiency, 618022
RSPO4	100%	100%	100%	100%	Anonychia congenita, 206800
RSPRY1	100%	100%	100%	100%	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723
RSRC1	99,80%	96,80%	100%	100%	Intellectual developmental disorder, autosomal recessive 70, 618402
RTEL1	99,50%	96,80%	100%	100%	Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 Dyskeratosis congenita, autosomal dominant 4, 615190
RTN4IP1	99,90%	98,70%	100%	100%	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
RTTN	98,90%	98,00%	100%	100%	Microcephaly, short stature, and polymicrogyria with seizures, 614833
RUBCN	99,40%	97,50%	100%	100%	?Spinocerebellar ataxia, autosomal recessive 15, 615705
RUSC2	100%	100%	100%	100%	Mental retardation, autosomal recessive 61, 617773
RYR1	96,90%	93,90%	99,40%	99,00%	Central core disease, 117000 King-Denborough syndrome, 145600 Minicore myopathy with external ophthalmoplegia, 255320 Neuromuscular disease, congenital, with uniform type 1 fiber, 117000
S1PR2	99,40%	96,90%	100%	100%	Deafness, autosomal recessive 68, 610419
SACS	100%	100%	100%	100%	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAG	100%	100%	100%	100%	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758
SALL2	100%	100%	100%	100%	?Coloboma, ocular, autosomal recessive, 216820
SAMD9	100%	99,80%	100%	100%	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455
SAMHD1	100%	99,60%	100%	100%	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SAR1B	97,00%	89,70%	100%	100%	Chylomicron retention disease, 246700
SARS	100%	99,30%	100%	100%	?Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709
SARS2	95,80%	94,60%	100%	100%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845

SASH1	99,90%	98,70%	100%	100%	?Cancer, alopecia, pigment dyscrasia, onychodystrophy, and keratoderma, 618373 Dyschromatosis universalis hereditaria 1, 127500
SASS6	99,90%	98,50%	100%	100%	?Microcephaly 14, primary, autosomal recessive, 616402
SBDS	100%	100%	100%	100%	Shwachman-Diamond syndrome, 260400
SBF1	99,00%	97,70%	100%	100%	Charcot-Marie-Tooth disease, type 4B3, 615284
SBF2	99,90%	99,40%	100%	100%	Charcot-Marie-Tooth disease, type 4B2, 604563
SC5D	100%	99,50%	100%	100%	Lathosterolosis, 607330
SCAPER	99,70%	98,20%	100%	100%	Intellectual developmental disorder and retinitis pigmentosa, 618195
SCARB2	100%	99,80%	100%	100%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCARF2	95,40%	86,30%	99,80%	99,20%	Van den Ende-Gupta syndrome, 600920
SCN1B	98,00%	96,40%	99,80%	99,30%	Epileptic encephalopathy, early infantile, 52, 617350 Atrial fibrillation, familial, 13, 615377 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233 Brugada syndrome 5, 612838
SCN4A	100%	99,60%	100%	100%	Paramyotonia congenita, 168300 Hyperkalemic periodic paralysis, type 2, 170500 Myotonia congenita, atypical, acetazolamide-responsive, 608390 Myasthenic syndrome, congenital, 16, 614198 Hypokalemic periodic paralysis, type 2, 613345
SCN9A	99,30%	97,80%	100%	100%	Small fiber neuropathy, 133020 HSAN2D, autosomal recessive, 243000 Paroxysmal extreme pain disorder, 167400 Epilepsy, generalized, with febrile seizures plus, type 7, 613863 Insensitivity to pain, congenital, 243000 Erythermalgia, primary, 133020 Febrile seizures, familial, 3B, 613863
SCNN1A	99,70%	98,20%	100%	100%	Pseudohypoaldosteronism, type I, 264350 ?Liddle syndrome 3, 618126 Bronchiectasis with or without elevated sweat chloride 2, 613021
SCNN1B	100%	99,70%	100%	100%	Pseudohypoaldosteronism, type I, 264350 Liddle syndrome 1, 177200 Bronchiectasis with or without elevated sweat chloride 1, 211400
SCNN1G	99,80%	98,30%	100%	100%	Liddle syndrome 2, 618114 Pseudohypoaldosteronism, type I, 264350 Bronchiectasis with or without elevated sweat chloride 3, 613071
SCO1	97,10%	93,80%	100%	100%	Mitochondrial complex IV deficiency, 220110

SCO2	100%	100%	100%	100%	Myopia 6, 608908 Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377
SCP2	100%	99,20%	100%	100%	?Leukoencephalopathy with dystonia and motor neuropathy, 613724
SCYL1	100%	100%	100%	100%	Spinocerebellar ataxia, autosomal recessive 21, 616719
SDCCAG8	100%	99,90%	100%	100%	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SDHA	85,80%	80,40%	100%	100%	Leigh syndrome, 256000 Paragangliomas 5, 614165 Cardiomyopathy, dilated, 1GG, 613642 Mitochondrial respiratory chain complex II deficiency, 252011
SDHAF1	99,90%	93,20%	100%	100%	Mitochondrial complex II deficiency, 252011
SDHD	54,00%	51,60%	80,10%	80,10%	Paragangliomas 1, with or without deafness, 168000 Mitochondrial complex II deficiency, 252011 Paraganglioma and gastric stromal sarcoma, 606864 Pheochromocytoma, 171300
SDR9C7	100%	100%	100%	100%	Ichthyosis, congenital, autosomal recessive 13, 617574
SEC23A	99,70%	98,20%	100%	100%	Craniolenticulosutural dysplasia, 607812
SEC23B	99,90%	99,30%	100%	100%	?Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SEC24D	100%	99,70%	100%	100%	Cole-Carpenter syndrome 2, 616294
SEC31A	99,30%	97,10%	100%	100%	?Neurodevelopmental disorder with spastic quadriplegia, optic atrophy, seizures, and structural brain anomalies, 618651
SECISBP2	99,80%	97,40%	100%	100%	Thyroid hormone metabolism, abnormal, 609698
SELENON	84,50%	84,00%	87,70%	85,10%	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310
SEMA4A	100%	99,80%	100%	100%	Cone-rod dystrophy 10, 610283 Retinitis pigmentosa 35, 610282
SEPSECS	100%	100%	100%	100%	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	99,90%	99,50%	100%	100%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPINA1	100%	100%	100%	100%	Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490 Emphysema-cirrhosis, due to AAT deficiency, 613490 Emphysema due to AAT deficiency, 613490
SERPINA6	100%	100%	100%	100%	Corticosteroid-binding globulin deficiency, 611489
SERPINB6	95,90%	95,90%	100%	100%	?Deafness, autosomal recessive 91, 613453
SERPINB7	100%	99,90%	100%	100%	Palmoplantar keratoderma, Nagashima type, 615598
SERPINB8	95,00%	95,00%	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%	100%	100%	100%	Osteogenesis imperfecta, type VI, 613982
SERPINF2	100%	99,80%	100%	100%	Alpha-2-plasmin inhibitor deficiency, 262850
SERPING1	99,70%	97,50%	100%	100%	Angioedema, hereditary, types I and II, 106100 Complement component 4, partial deficiency of, 120790
SERPINH1	100%	98,30%	100%	100%	Osteogenesis imperfecta, type X, 613848
SETX	100%	99,80%	100%	100%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 Amyotrophic lateral sclerosis 4, juvenile, 602433
SFRP4	100%	99,80%	100%	100%	Pyle disease, 265900
SFTPB	99,50%	96,80%	100%	100%	Surfactant metabolism dysfunction, pulmonary, 1, 265120
SFXN4	99,90%	98,90%	100%	100%	Combined oxidative phosphorylation deficiency 18, 615578
SGCA	100%	99,90%	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099
SGCB	97,70%	96,50%	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286
SGCD	100%	98,90%	100%	100%	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287
SGCG	100%	99,20%	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700
SGO1	99,90%	98,90%	100%	100%	Chronic atrial and intestinal dysrhythmia, 616201
SGPL1	100%	100%	100%	100%	Nephrotic syndrome, type 14, 617575
SGSH	94,40%	94,10%	100%	100%	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SH3PXD2B	100%	100%	100%	100%	Frank-ter Haar syndrome, 249420
SH3TC2	100%	99,70%	100%	100%	Charcot-Marie-Tooth disease, type 4C, 601596 Mononeuropathy of the median nerve, mild, 613353
SI	99,20%	96,10%	100%	100%	Sucrase-isomaltase deficiency, congenital, 222900
SIGMAR1	100%	100%	100%	100%	?Amyotrophic lateral sclerosis 16, juvenile, 614373 ?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726
SIK3	99,80%	98,70%	99,30%	98,10%	?Spondyloepimetaphyseal dysplasia, Krakow type, 618162
SIL1	99,20%	96,70%	100%	100%	Marinesco-Sjogren syndrome, 248800
SIX6	100%	100%	100%	100%	Optic disc anomalies with retinal and/or macular dystrophy, 212550
SKIV2L	100%	99,80%	100%	100%	Trichohepatoenteric syndrome 2, 614602
SLC10A2	100%	100%	100%	100%	Bile acid malabsorption, primary, 613291
SLC10A7	99,70%	98,00%	100%	100%	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363
SLC11A2	100%	99,90%	100%	100%	Anemia, hypochromic microcytic, with iron overload 1, 206100
SLC12A1	100%	99,90%	100%	100%	Bartter syndrome, type 1, 601678
SLC12A3	100%	99,90%	100%	100%	Gitelman syndrome, 263800
SLC12A5	83,90%	83,80%	97,40%	97,40%	Epileptic encephalopathy, early infantile, 34, 616645
SLC12A6	100%	100%	100%	100%	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC13A3	99,40%	97,50%	100%	100%	Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate, 618384
SLC13A5	100%	99,90%	100%	100%	Epileptic encephalopathy, early infantile, 25, 615905

SLC16A1	100%	99,30%	100%	100%	Monocarboxylate transporter 1 deficiency, 616095 Hyperinsulinemic hypoglycemia, familial, 7, 610021 Erythrocyte lactate transporter defect, 245340
SLC17A5	99,60%	97,00%	100%	100%	Sialic acid storage disorder, infantile, 269920 Salla disease, 604369
SLC18A2	100%	99,70%	100%	100%	?Parkinsonism-dystonia, infantile, 2, 618049
SLC18A3	100%	100%	100%	100%	Myasthenic syndrome, congenital, 21, presynaptic, 617239
SLC19A2	100%	99,70%	100%	100%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	100%	99,80%	98,70%	98,70%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A1	99,90%	99,60%	100%	100%	Dicarboxylic aminoaciduria, 222730
SLC1A4	99,00%	95,80%	100%	100%	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC22A12	100%	99,80%	100%	100%	Hypouricemia, renal, 220150
SLC22A5	100%	100%	100%	100%	Carnitine deficiency, systemic primary, 212140
SLC24A1	100%	99,90%	100%	100%	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
SLC24A4	100%	99,80%	100%	100%	Amelogenesis imperfecta, type IIA5, 615887
SLC24A5	99,90%	99,10%	100%	100%	Albinism, oculocutaneous, type VI, 113750
SLC25A1	95,80%	88,60%	99,50%	97,80%	Myasthenic syndrome, congenital, 23, presynaptic, 618197 Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A12	99,90%	99,50%	100%	100%	Epileptic encephalopathy, early infantile, 39, 612949
SLC25A13	100%	99,70%	100%	100%	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A15	99,80%	98,10%	100%	100%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A19	100%	98,50%	100%	100%	Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710 Microcephaly, Amish type, 607196
SLC25A20	100%	100%	100%	100%	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A22	98,60%	95,80%	100%	100%	Epileptic encephalopathy, early infantile, 3, 609304
SLC25A26	100%	99,50%	100%	100%	Combined oxidative phosphorylation deficiency 28, 616794
SLC25A3	99,80%	98,00%	100%	100%	Mitochondrial phosphate carrier deficiency, 610773
SLC25A38	99,70%	97,10%	100%	100%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC25A4	100%	100%	100%	100%	Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283
SLC25A42	96,50%	93,20%	100%	100%	metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416
SLC25A46	99,70%	97,30%	100%	100%	Neuropathy, hereditary motor and sensory, type VIB, 616505
SLC26A1	100%	99,60%	100%	100%	?Nephrolithiasis, calcium oxalate, 167030

SLC26A2	100%	100%	100%	100%	De la Chapelle dysplasia, 256050 Atelosteogenesis, type II, 256050 Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 Diastrophic dysplasia, 222600 Achondrogenesis Ib, 600972 Epiphyseal dysplasia, multiple, 4, 226900
SLC26A3	100%	99,50%	100%	100%	Diarrhea 1, secretory chloride, congenital, 214700
SLC26A4	100%	99,70%	100%	100%	Pendred syndrome, 274600 Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791
SLC26A5	99,10%	96,80%	100%	100%	?Deafness, autosomal recessive 61, 613865
SLC27A4	100%	99,80%	100%	100%	Ichthyosis prematurity syndrome, 608649
SLC29A3	100%	99,60%	100%	100%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC2A1	92,80%	92,80%	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	97,70%	97,70%	100%	100%	Arterial tortuosity syndrome, 208050
SLC2A2	100%	100%	100%	100%	Fanconi-Bickel syndrome, 227810
SLC2A9	99,80%	96,10%	100%	100%	Hypouricemia, renal, 2, 612076
SLC30A10	100%	100%	100%	100%	Hypermanganesemia with dystonia 1, 613280
SLC30A9	98,80%	94,20%	100%	100%	?Birk-Landau-Perez syndrome, 617595
SLC33A1	99,90%	98,90%	100%	100%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC34A1	99,90%	99,10%	100%	100%	Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286 Hypercalcemia, infantile, 2, 616963 ?Fanconi renotubular syndrome 2, 613388
SLC34A2	100%	100%	100%	100%	Pulmonary alveolar microlithiasis, 265100
SLC34A3	100%	99,40%	100%	100%	Hypophosphatemic rickets with hypercalciuria, 241530
SLC35A1	100%	99,70%	100%	100%	Congenital disorder of glycosylation, type IIf, 603585
SLC35A3	80,70%	78,60%	81,10%	81,00%	?Arthrogryposis, mental retardation, and seizures, 615553
SLC35C1	99,90%	98,70%	100%	100%	Congenital disorder of glycosylation, type IIc, 266265
SLC35D1	100%	97,70%	100%	100%	Schneckenbecken dysplasia, 269250
SLC37A4	100%	99,20%	100%	100%	Glycogen storage disease Ic, 232240 Glycogen storage disease Ib, 232220
SLC38A8	99,90%	97,30%	100%	100%	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218
SLC39A13	99,80%	98,20%	100%	100%	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350
SLC39A14	100%	99,40%	93,50%	93,50%	?Hyperostosis cranalis interna, 144755 Hypermanganesemia with dystonia 2, 617013

SLC39A4	99,50%	95,50%	100%	100%	Acrodermatitis enteropathica, 201100
SLC39A8	100%	99,70%	100%	100%	Congenital disorder of glycosylation, type IIn, 616721
SLC3A1	100%	99,80%	96,60%	96,60%	Cystinuria, 220100
SLC45A1	100%	99,60%	100%	100%	Intellectual developmental disorder with neuropsychiatric features, 617532
SLC45A2	100%	99,90%	100%	100%	Albinism, oculocutaneous, type IV, 606574
SLC46A1	99,90%	98,50%	100%	100%	Folate malabsorption, hereditary, 229050
SLC4A1	100%	99,80%	96,10%	96,10%	Spherocytosis, type 4, 612653 Cryohydrocytosis, 185020 Ovalocytosis, SA type, 166900 Renal tubular acidosis, distal, AD, 179800 Renal tubular acidosis, distal, AR, 611590
SLC4A11	100%	99,90%	100%	100%	Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy, autosomal recessive, 217700 Corneal endothelial dystrophy and perceptive deafness, 217400
SLC4A4	99,80%	99,20%	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%	Brown-Vialetto-Van Laere syndrome 1, 211530 ?Fazio-Londe disease, 211500
SLC5A1	100%	100%	100%	100%	Glucose/galactose malabsorption, 606824
SLC5A2	100%	100%	100%	100%	Renal glucosuria, 233100
SLC5A5	100%	99,80%	100%	100%	Thyroid dyshormonogenesis 1, 274400
SLC5A7	100%	99,90%	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 HEARTnup disorder, 234500 Hyperglycinuria, 138500
SLC6A3	100%	100%	100%	100%	Parkinsonism-dystonia, infantile, 1, 613135
SLC6A5	100%	100%	100%	100%	Hyperekplexia 3, 614618
SLC6A9	100%	100%	100%	100%	Glycine encephalopathy with normal serum glycine, 617301
SLC7A14	100%	100%	100%	100%	Retinitis pigmentosa 68, 615725
SLC7A7	100%	99,90%	100%	100%	Lysinuric protein intolerance, 222700
SLC7A9	100%	99,90%	100%	100%	Cystinuria, 220100
SLC9A1	100%	100%	100%	100%	?Lichtenstein-Knorr syndrome, 616291
SLC9A3	100%	99,70%	100%	99,90%	Diarrhea 8, secretory sodium, congenital, 616868
SLCO2A1	100%	99,40%	100%	100%	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
SLITRK6	100%	100%	100%	100%	Deafness and myopia, 221200

SLURP1	100%	99,30%	100%	100%	Meleda disease, 248300
SLX4	100%	99,80%	100%	100%	Fanconi anemia, complementation group P, 613951
SMARCAL1	100%	99,90%	100%	100%	Schimke immunoosseous dysplasia, 242900
SMARCD2	87,00%	85,90%	99,60%	97,00%	Specific granule deficiency 2, 617475
SMG9	100%	100%	100%	100%	Heart and brain malformation syndrome, 616920
SMN1	99,50%	94,70%	94,60%	94,60%	Spinal muscular atrophy-2, 253550 Spinal muscular atrophy-3, 253400 Spinal muscular atrophy-1, 253300 Spinal muscular atrophy-4, 271150
SMOC1	99,90%	98,40%	100%	100%	Microphthalmia with limb anomalies, 206920
SMOC2	77,00%	76,70%	100%	100%	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SMPD1	100%	100%	100%	100%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SMPD4	99,40%	94,20%	100%	100%	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622
SNAI2	100%	99,10%	100%	100%	Waardenburg syndrome, type 2D, 608890 Piebaldism, 172800
SNAP29	100%	100%	100%	100%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNIP1	98,90%	97,10%	100%	100%	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501
SNORD118	NC	NC	NC	NC	Leukoencephalopathy, brain calcifications, and cysts, 614561
SNX10	96,20%	95,70%	100%	99,60%	Osteopetrosis, autosomal recessive 8, 615085
SNX14	99,60%	95,90%	100%	100%	Spinocerebellar ataxia, autosomal recessive 20, 616354
SOBP	97,50%	92,90%	97,00%	95,30%	Mental retardation, anterior maxillary protrusion, and strabismus, 613671
SOD1	100%	99,90%	100%	100%	Amyotrophic lateral sclerosis 1, 105400 Spastic tetraplegia and axial hypotonia, progressive, 618598
SOST	100%	99,50%	100%	100%	Sclerosteosis 1, 269500 Van Buchem disease, 239100 Craniodiaphyseal dysplasia, autosomal dominant, 122860
SOX18	70,70%	55,20%	96,10%	92,60%	Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940 Hypotrichosis-lymphedema-telangiectasia syndrome, 607823
SP110	100%	100%	100%	100%	Hepatic venoocclusive disease with immunodeficiency, 235550
SP7	100%	99,80%	100%	100%	Osteogenesis imperfecta, type XII, 613849
SPAG1	99,30%	95,80%	99,90%	98,60%	Ciliary dyskinesia, primary, 28, 615505
SPARC	100%	100%	100%	100%	Osteogenesis imperfecta, type XVII, 616507
SPART	99,70%	96,80%	100%	100%	Troyer syndrome, 275900
SPATA5	100%	99,70%	100%	100%	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPATA7	99,80%	98,20%	100%	100%	Retinitis pigmentosa, juvenile, autosomal recessive, 604232 Leber congenital amaurosis 3, 604232

SPEG	96,40%	89,50%	99,70%	99,70%	Centronuclear myopathy 5, 615959
SPG11	100%	99,30%	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360 Amyotrophic lateral sclerosis 5, juvenile, 602099
SPG21	99,40%	96,80%	100%	100%	Mast syndrome, 248900
SPG7	94,90%	92,60%	100%	100%	Spastic paraplegia 7, autosomal recessive, 607259
SPINK5	99,90%	99,50%	100%	100%	Netherton syndrome, 256500
SPINT2	98,50%	83,80%	100%	100%	Diarrhea 3, secretory sodium, congenital, syndromic, 270420
SPNS2	92,10%	89,30%	97,60%	95,70%	?Deafness, autosomal recessive 115, 618457
SPR	99,80%	96,30%	100%	100%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRTN	100%	100%	100%	100%	Ruijs-Aalfs syndrome, 616200
SPTA1	99,90%	99,20%	100%	100%	Pyropoikilocytosis, 266140 Elliptocytosis-2, 130600 Spherocytosis, type 3, 270970
SPTB	100%	100%	100%	100%	Elliptocytosis-3, 617948 Spherocytosis, type 2, 616649 Anemia, neonatal hemolytic, fatal or near-fatal, 617948
SPTBN2	100%	99,30%	99,90%	99,90%	Spinocerebellar ataxia, autosomal recessive 14, 615386 Spinocerebellar ataxia 5, 600224
SPTBN4	97,30%	91,00%	100%	100%	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519
SQSTM1	98,80%	95,50%	100%	100%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 Myopathy, distal, with rimmed vacuoles, 617158 Paget disease of bone 3, 167250
SRD5A2	99,90%	99,00%	100%	100%	Pseudovaginal perineoscrotal hypospadias, 264600
SRD5A3	99,90%	99,10%	100%	100%	Kahrizi syndrome, 612713 Congenital disorder of glycosylation, type Iq, 612379
ST14	99,90%	98,60%	100%	100%	Ichthyosis, congenital, autosomal recessive 11, 602400
ST3GAL3	100%	99,80%	100%	100%	Mental retardation, autosomal recessive 12, 611090 ?Epileptic encephalopathy, early infantile, 15, 615006
ST3GAL5	85,00%	84,20%	98,70%	98,40%	Salt and pepper developmental regression syndrome, 609056
STAC3	100%	100%	100%	100%	Myopathy, congenital, Baily-Bloch, 255995
STAMBP	100%	99,40%	100%	100%	Microcephaly-capillary malformation syndrome, 614261
STAR	100%	100%	100%	100%	Lipoid adrenal hyperplasia, 201710
STAT1	99,90%	98,80%	100%	100%	Immunodeficiency 31C, autosomal dominant, 614162 Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796
STAT2	100%	99,90%	100%	100%	Immunodeficiency 44, 616636

STAT5B	100%	98,50%	100%	100%	Leukemia, acute promyelocytic, somatic, 102578 Growth hormone insensitivity with immunodeficiency, 245590
STIL	100%	99,80%	100%	100%	Microcephaly 7, primary, autosomal recessive, 612703
STIM1	99,80%	98,00%	100%	100%	Myopathy, tubular aggregate, 1, 160565 Immunodeficiency 10, 612783 Stormorken syndrome, 185070
STK4	100%	99,80%	100%	100%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STN1	100%	100%	100%	100%	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341
STRA6	100%	99,80%	100%	100%	Microphthalmia, syndromic 9, 601186 Microphthalmia, isolated, with coloboma 8, 601186
STRADA	100%	98,90%	100%	100%	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087
STRC	99,90%	98,00%	100%	100%	Deafness, autosomal recessive 16, 603720
STT3A	100%	100%	100%	100%	?Congenital disorder of glycosylation, type Iw, 615596
STT3B	100%	99,60%	100%	100%	?Congenital disorder of glycosylation, type Ix, 615597
STUB1	100%	98,70%	100%	100%	Spinocerebellar ataxia, autosomal recessive 16, 615768 ?Spinocerebellar ataxia 48, 618093
STX11	100%	100%	100%	100%	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STXBP2	82,10%	79,70%	99,30%	97,10%	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
SUCLA2	94,30%	86,60%	100%	100%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	99,90%	99,80%	100%	100%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUFU	100%	100%	100%	100%	Basal cell nevus syndrome, 109400 Medulloblastoma, desmoplastic, 155255 Joubert syndrome 32, 617757
SULT2B1	100%	100%	100%	100%	Ichthyosis, congenital, autosomal recessive 14, 617571
SUMF1	97,50%	90,80%	100%	100%	Multiple sulfatase deficiency, 272200
SUOX	100%	100%	100%	100%	Sulfite oxidase deficiency, 272300
SURF1	89,40%	88,20%	100%	100%	Leigh syndrome, due to COX IV deficiency, 256000 Charcot-Marie-Tooth disease, type 4K, 616684
SVBP	100%	100%	100%	100%	Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569
SYNE1	98,30%	98,00%	98,80%	98,80%	Spinocerebellar ataxia, autosomal recessive 8, 610743 Arthrogryposis multiplex congenita, myogenic type, 618484 Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998
SYNE4	99,70%	97,00%	100%	100%	Deafness, autosomal recessive 76, 615540
SYNJ1	99,90%	99,40%	100%	100%	Epileptic encephalopathy, early infantile, 53, 617389 Parkinson disease 20, early-onset, 615530
SYT14	61,90%	58,90%	95,90%	95,90%	?Spinocerebellar ataxia, autosomal recessive 11, 614229

SZT2	99,60%	99,50%	100%	99,90%	Epileptic encephalopathy, early infantile, 18, 615476
T	99,40%	96,90%	100%	100%	Sacral agenesis with vertebral anomalies, 615709
TBC1D24	100%	100%	100%	100%	Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp, 608105 DOORS syndrome, 220500 Deafness, autosomal dominant 65, 616044 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021 Deafness , autosomal recessive 86, 614617
TBC1D7	100%	99,30%	100%	100%	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000
TBCD	96,20%	94,40%	100%	100%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TBCE	99,80%	97,30%	100%	100%	Kenny-Caffey syndrome, type 1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
TBCK	99,10%	96,80%	100%	100%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900
TBX15	100%	99,90%	100%	100%	Cousin syndrome, 260660
TBX19	100%	100%	100%	100%	Adrenocorticotropic hormone deficiency, 201400
TBX6	99,50%	95,40%	100%	100%	Spondylocostal dysostosis 5, 122600
TBXAS1	100%	100%	100%	100%	Ghosal hematodiaphyseal syndrome, 231095
TCAP	100%	100%	100%	100%	Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954
TCIRG1	97,60%	90,10%	100%	100%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	100%	100%	100%	100%	Transcobalamin II deficiency, 275350
TCTEX1D2	100%	100%	100%	100%	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405
TCTN1	96,70%	93,00%	94,70%	94,70%	Joubert syndrome 13, 614173
TCTN2	100%	99,50%	100%	100%	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	100%	100%	100%	100%	Orofaciodigital syndrome IV, 258860 Joubert syndrome 18, 614815
TDP1	99,90%	99,50%	100%	100%	?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250
TDP2	100%	99,40%	100%	100%	Spinocerebellar ataxia, autosomal recessive 23, 616949
TDRD7	99,90%	99,10%	100%	100%	Cataract 36, 613887
TECPR2	100%	100%	100%	100%	Spastic paraplegia 49, autosomal recessive, 615031
TECR	100%	98,90%	100%	100%	Mental retardation, autosomal recessive 14, 614020
TECRL	96,30%	89,30%	100%	100%	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021
TECTA	100%	99,90%	100%	100%	Deafness, autosomal dominant 8/12, 601543 Deafness, autosomal recessive 21, 603629
TELO2	99,70%	96,20%	100%	100%	You-Hoover-Fong syndrome, 616954

TENM3	100%	99,70%	100%	100%	Microphthalmia, syndromic 15, 615145 ?Microphthalmia, isolated, with coloboma 9, 615145
TF	100%	100%	100%	100%	Atransferrinemia, 209300
TFAM	97,50%	83,50%	100%	100%	?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156
TFG	96,90%	96,30%	100%	100%	?Spastic paraplegia 57, autosomal recessive, 615658 Hereditary motor and sensory neuropathy, Okinawa type, 604484
TFR2	99,10%	97,80%	100%	100%	Hemochromatosis, type 3, 604250
TFRC	100%	99,80%	100%	100%	Immunodeficiency 46, 616740
TG	100%	99,40%	100%	100%	Thyroid dyshormonogenesis 3, 274700
TGDS	99,40%	96,80%	100%	100%	Catel-Manzke syndrome, 616145
TGFB1	100%	99,90%	100%	100%	Camurati-Engelmann disease, 131300 Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213
TGM1	100%	99,90%	100%	100%	Ichthyosis, congenital, autosomal recessive 1, 242300
TGM5	100%	99,70%	100%	100%	Peeling skin syndrome 2, 609796
TH	99,30%	96,10%	100%	100%	Segawa syndrome, recessive, 605407
THOC6	100%	100%	100%	100%	Beaulieu-Boycott-Innes syndrome, 613680
THRB	100%	99,70%	100%	100%	Thyroid hormone resistance, 188570 Thyroid hormone resistance, selective pituitary, 145650 Thyroid hormone resistance, autosomal recessive, 274300
TIMM50	98,30%	94,40%	100%	100%	3-methylglutaconic aciduria, type IX, 617698
TIMMDC1	100%	100%	100%	100%	Mitochondrial complex I deficiency, nuclear type 31, 618251
TJP2	94,00%	93,60%	100%	100%	Hypercholanemia, familial, 607748 Cholestasis, progressive familial intrahepatic 4, 615878
TK2	99,20%	96,30%	100%	100%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560 ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069
TKT	98,70%	97,80%	98,70%	98,70%	Short stature, developmental delay, and congenital heart defects, 617044
TLE6	100%	98,80%	100%	100%	Preimplantation embryonic lethality, 616814
TMC1	99,70%	97,10%	100%	100%	Deafness, autosomal recessive 7, 600974 Deafness, autosomal dominant 36, 606705
TMC6	100%	99,30%	100%	100%	Epidermodysplasia verruciformis, 226400
TMC8	100%	98,70%	100%	100%	Epidermodysplasia verruciformis 2, 618231
TMCO1	88,00%	87,40%	88,00%	88,00%	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	96,30%	84,40%	100%	100%	Optic atrophy 7, 612989
TMEM126B	99,80%	97,40%	100%	100%	Mitochondrial complex I deficiency, nuclear type 29, 618250

TMEM132E	96,90%	93,50%	100%	100%	?Deafness, autosomal recessive 99, 618481
TMEM138	100%	99,10%	100%	100%	Joubert syndrome 16, 614465
TMEM165	100%	100%	100%	100%	Congenital disorder of glycosylation, type IIk, 614727
TMEM199	100%	99,90%	100%	100%	Congenital disorder of glycosylation, type IIp, 616829
TMEM216	99,90%	98,10%	100%	100%	Meckel syndrome 2, 603194 Joubert syndrome 2, 608091
TMEM231	100%	99,60%	100%	100%	Meckel syndrome 11, 615397 Joubert syndrome 20, 614970
TMEM237	100%	99,90%	100%	100%	Joubert syndrome 14, 614424
TMEM260	97,50%	93,40%	100%	100%	Structural heart defects and renal anomalies syndrome, 617478
TMEM38B	100%	99,90%	100%	100%	Osteogenesis imperfecta, type XIV, 615066
TMEM5	99,50%	96,80%	100%	99,90%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
TMEM67	99,50%	95,00%	100%	99,90%	Meckel syndrome 3, 607361 ?RHYNS syndrome, 602152 Nephronophthisis 11, 613550 COACH syndrome, 216360 Joubert syndrome 6, 610688
TMEM70	98,00%	93,90%	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	99,20%	95,10%	100%	100%	Deafness, autosomal recessive 6, 600971
TMPRSS15	98,50%	95,20%	100%	100%	Enterokinase deficiency, 226200
TMPRSS3	100%	99,90%	100%	100%	Deafness, autosomal recessive 8/10, 601072
TMPRSS6	99,90%	99,10%	100%	100%	Iron-refractory iron deficiency anemia, 206200
TMTC3	99,60%	96,50%	100%	100%	Lissencephaly 8, 617255
TNFRSF11A	94,60%	93,30%	99,20%	98,00%	Osteolysis, familial expansile, 174810 Osteopetrosis, autosomal recessive 7, 612301
TNFRSF11B	100%	100%	100%	100%	Paget disease of bone 5, juvenile-onset, 239000
TNFRSF13B	100%	100%	100%	100%	Immunoglobulin A deficiency 2, 609529 Immunodeficiency, common variable, 2, 240500
TNFRSF13C	80,10%	75,40%	100%	99,90%	Immunodeficiency, common variable, 4, 613494
TNFRSF4	99,40%	95,40%	100%	100%	?Immunodeficiency 16, 615593
TNFSF11	100%	99,90%	100%	100%	Osteopetrosis, autosomal recessive 2, 259710
TNIK	100%	99,30%	100%	100%	Mental retardation, autosomal recessive 54, 617028
TNNI3	99,70%	95,20%	100%	100%	Cardiomyopathy, hypertrophic, 7, 613690 ?Cardiomyopathy, dilated, 2A, 611880 Cardiomyopathy, dilated, 1FF, 613286 Cardiomyopathy, familial restrictive, 1, 115210

TNNT1	99,90%	97,60%	100%	100%	Nemaline myopathy 5, Amish type, 605355
TNXB	99,60%	95,10%	100%	100%	Ehlers-Danlos syndrome, classic-like, 1, 606408 Vesicoureteral reflux 8, 615963
TOE1	100%	100%	100%	100%	Pontocerebellar hypoplasia, type 7, 614969
TONSL	99,80%	97,80%	100%	100%	Spondyloepimetaphyseal dysplasia, sponastrime type, 271510
TOP3A	100%	98,70%	100%	100%	Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097 ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098
TOR1AIP1	99,90%	98,00%	100%	100%	?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072
TP53RK	92,50%	79,60%	100%	100%	Galloway-Mowat syndrome 4, 617730
TPI1	99,80%	97,50%	100%	100%	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512
TPK1	99,80%	99,00%	100%	100%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TPM3	89,20%	87,20%	100%	100%	CAP myopathy 1, 609284 Nemaline myopathy 1, autosomal dominant or recessive, 609284 Myopathy, congenital, with fiber-type disproportion, 255310
TPO	99,90%	98,20%	100%	100%	Thyroid dyshormonogenesis 2A, 274500
TPP1	100%	100%	100%	100%	Spinocerebellar ataxia, autosomal recessive 7, 609270 Ceroid lipofuscinosis, neuronal, 2, 204500
TPRKB	81,10%	75,90%	81,90%	81,90%	Galloway-Mowat syndrome 5, 617731
TPRN	87,90%	79,30%	94,40%	89,80%	Deafness, autosomal recessive 79, 613307
TRAC	100%	100%	100%	100%	Immunodeficiency 7, TCR-alpha/beta deficient, 615387
TRAF3IP1	99,60%	97,60%	100%	100%	Senior-Loken syndrome 9, 616629
TRAIP	100%	100%	100%	100%	Seckel syndrome 9, 616777
TRAK1	100%	99,60%	100%	100%	Epileptic encephalopathy, early infantile, 68, 618201
TRAPPC11	100%	99,20%	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356
TRAPPC12	100%	99,60%	100%	100%	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669
TRAPPC2L	100%	100%	100%	100%	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331
TRAPPC6B	99,90%	98,00%	100%	100%	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862
TRAPPC9	100%	99,60%	100%	100%	Mental retardation, autosomal recessive 13, 613192
TRDN	96,20%	86,60%	100%	100%	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441
TREM2	100%	99,80%	100%	100%	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193
TREX1	100%	100%	100%	100%	Vasculopathy, retinal, with cerebral leukodystrophy, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
TRH	99,60%	96,50%	100%	100%	No OMIM disease ID
TRIM2	93,90%	93,30%	93,90%	93,90%	Charcot-Marie-Tooth disease, type 2R, 615490
TRIM32	100%	100%	100%	100%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110

TRIM36	100%	99,20%	100%	100%	?Anencephaly, 206500
TRIM37	98,60%	98,10%	98,70%	98,70%	Mulibrey nanism, 253250
TRIOBP	97,80%	96,10%	99,90%	99,60%	Deafness, autosomal recessive 28, 609823
TRIP11	98,40%	94,00%	100%	100%	Osteochondrodysplasia, 184260 Achondrogenesis, type IA, 200600
TRIP13	100%	100%	100%	100%	Mosaic variegated aneuploidy syndrome 3, 617598
TRIP4	100%	99,10%	100%	100%	Spinal muscular atrophy with congenital bone fractures 1, 616866 ?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066
TRIT1	100%	100%	100%	100%	Combined oxidative phosphorylation deficiency 35, 617873
TRMT1	99,40%	96,20%	100%	100%	Mental retardation, autosomal recessive 68, 618302
TRMT10A	100%	99,70%	100%	100%	Microcephaly, short stature, and impaired glucose metabolism 1, 616033
TRMT10C	100%	100%	100%	99,90%	Combined oxidative phosphorylation deficiency 30, 616974
TRMT5	100%	99,30%	100%	100%	Combined oxidative phosphorylation deficiency 26, 616539
TRMU	100%	100%	100%	99,90%	Liver failure, transient infantile, 613070
TRNT1	99,50%	96,50%	100%	100%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 Retinitis pigmentosa and erythrocytic microcytosis, 616959
TRPM1	100%	99,80%	100%	100%	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TRPM6	99,90%	99,50%	100%	100%	Hypomagnesemia 1, intestinal, 602014
TSEN15	99,80%	97,50%	100%	100%	Pontocerebellar hypoplasia, type 2F, 617026
TSEN2	100%	99,60%	100%	100%	Pontocerebellar hypoplasia type 2B, 612389
TSEN34	90,80%	86,40%	100%	100%	?Pontocerebellar hypoplasia type 2C, 612390
TSEN54	96,30%	94,30%	99,90%	98,90%	Pontocerebellar hypoplasia type 4, 225753 Pontocerebellar hypoplasia type 2A, 277470 ?Pontocerebellar hypoplasia type 5, 610204
TSFM	100%	99,50%	94,90%	94,90%	Combined oxidative phosphorylation deficiency 3, 610505
TSHB	100%	100%	100%	100%	Hypothyroidism, congenital, nongoitrous 4, 275100
TSHR	99,90%	99,30%	100%	100%	Hyperthyroidism, nonautoimmune, 609152 Hypothyroidism, congenital, nongoitrous, 1, 275200 Hyperthyroidism, familial gestational, 603373 Thyroid adenoma, hyperfunctioning, somatic, 0 Thyroid carcinoma with thyrotoxicosis, 0
TSPAN12	100%	99,80%	100%	100%	Exudative vitreoretinopathy 5, 613310
TSPEAR	100%	99,20%	97,90%	97,90%	Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180 ?Deafness, autosomal recessive 98, 614861
TSPYL1	100%	100%	100%	100%	Sudden infant death with dysgenesis of the testes syndrome, 608800
TTC19	81,50%	73,80%	100%	99,20%	Mitochondrial complex III deficiency, nuclear type 2, 615157

TTC21B	99,90%	99,30%	100%	100%	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
TTC25	100%	100%	100%	100%	Ciliary dyskinesia, primary, 35, 617092
TTC37	100%	99,30%	100%	100%	Trichohepatoenteric syndrome 1, 222470
TTC7A	99,30%	95,40%	100%	100%	Gastrointestinal defects and immunodeficiency syndrome, 243150
TTC8	99,60%	98,10%	100%	100%	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
TTI2	100%	100%	100%	100%	Mental retardation, autosomal recessive 39, 615541
TTLL5	100%	99,70%	100%	100%	Cone-rod dystrophy 19, 615860
TTPA	94,70%	87,10%	100%	100%	Ataxia with isolated vitamin E deficiency, 277460
TUB	99,40%	97,10%	100%	100%	?Retinal dystrophy and obesity, 616188
TUBA8	99,90%	99,50%	100%	100%	Cortical dysplasia, complex, with other brain malformations 8, 613180
TUBGCP4	99,20%	96,40%	100%	100%	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TUBGCP6	100%	99,30%	100%	100%	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
TUFM	100%	99,00%	100%	100%	Combined oxidative phosphorylation deficiency 4, 610678
TULP1	100%	99,50%	100%	100%	Retinitis pigmentosa 14, 600132 Leber congenital amaurosis 15, 613843
TUSC3	100%	99,50%	100%	100%	Mental retardation, autosomal recessive 7, 611093
TWIST2	100%	100%	100%	100%	Barber-Say syndrome, 209885 Ablepharon-macrostomia syndrome, 200110 Focal facial dermal dysplasia 3, Setleis type, 227260
TWNK	100%	100%	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%	100%	100%	100%	?Combined oxidative phosphorylation deficiency 29, 616811
TXNL4A	100%	99,40%	100%	100%	Burn-McKeown syndrome, 608572
TYK2	99,90%	99,00%	100%	100%	Immunodeficiency 35, 611521
TYMP	100%	97,00%	100%	100%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYR	100%	100%	100%	100%	Waardenburg syndrome/albinism, digenic, 103470 Albinism, oculocutaneous, type IB, 606952 Albinism, oculocutaneous, type IA, 203100
TYROBP	100%	100%	100%	100%	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770
TYRP1	100%	99,80%	100%	100%	Albinism, oculocutaneous, type III, 203290
UBA5	97,80%	86,80%	100%	100%	?Spinocerebellar ataxia, autosomal recessive 24, 617133 Epileptic encephalopathy, early infantile, 44, 617132
UBE2T	100%	99,90%	100%	100%	Fanconi anemia, complementation group T, 616435
UBE3B	100%	99,90%	100%	100%	Kaufman oculocerebrofacial syndrome, 244450

UBR1	99,90%	99,10%	98,00%	98,00%	Johanson-Blizzard syndrome, 243800
UCHL1	99,80%	92,50%	100%	100%	Spastic paraplegia 79, autosomal recessive, 615491
UFC1	100%	100%	100%	100%	Neurodevelopmental disorder with spasticity and poor growth, 618076
UFM1	74,00%	69,40%	100%	100%	Leukodystrophy, hypomyelinating, 14, 617899
UGT1A1	100%	100%	100%	100%	Hyperbilirubinemia, familial transient neonatal, 237900 Crigler-Najjar syndrome, type I, 218800 Crigler-Najjar syndrome, type II, 606785
UMPS	100%	99,40%	97,00%	97,00%	Orotic aciduria, 258900
UNC13D	99,70%	98,10%	100%	100%	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC80	100%	99,50%	100%	100%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801
UNG	100%	98,80%	99,90%	99,30%	Immunodeficiency with hyper IgM, type 5, 608106
UPB1	100%	100%	100%	100%	Beta-ureidopropionase deficiency, 613161
UQCC2	100%	99,70%	100%	100%	Mitochondrial complex III deficiency, nuclear type 7, 615824
UQCC3	100%	98,70%	100%	100%	?Mitochondrial complex III deficiency, nuclear type 9, 616111
UQCRB	99,40%	95,10%	100%	100%	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC2	99,90%	99,30%	100%	100%	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRQ	100%	100%	100%	100%	Mitochondrial complex III deficiency, nuclear type 4, 615159
UROC1	100%	100%	100%	100%	?Urocanase deficiency, 276880
UROD	98,90%	96,10%	100%	100%	Porphyria, hepatoerythropoietic, 176100 Porphyria cutanea tarda, 176100
UROS	100%	99,90%	100%	100%	Porphyria, congenital erythropoietic, 263700
USB1	100%	99,40%	100%	100%	Poikiloderma with neutropenia, 604173
USH1C	100%	99,80%	100%	100%	Deafness, autosomal recessive 18A, 602092 Usher syndrome, type 1C, 276904
USH1G	99,60%	97,90%	100%	100%	Usher syndrome, type 1G, 606943
USH2A	100%	99,80%	100%	100%	Retinitis pigmentosa 39, 613809 Usher syndrome, type 2A, 276901
USP18	95,90%	95,90%	100%	100%	Pseudo-TORCH syndrome 2, 617397
USP45	99,60%	98,10%	100%	100%	?Leber congenital amaurosis 19, 618513
UVSSA	99,30%	98,80%	99,40%	99,30%	UV-sensitive syndrome 3, 614640
VAC14	99,90%	98,50%	100%	100%	Striatonigral degeneration, childhood-onset, 617054
VAMP1	100%	100%	100%	100%	Spastic ataxia 1, autosomal dominant, 108600 Myasthenic syndrome, congenital, 25, 618323
VARS	100%	99,90%	100%	100%	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802
VARS2	100%	99,40%	100%	100%	Combined oxidative phosphorylation deficiency 20, 615917
VAX1	97,50%	91,50%	95,70%	91,70%	?Microphthalmia, syndromic 11, 614402
VDR	99,90%	99,00%	100%	100%	Rickets, vitamin D-resistant, type IIA, 277440

VHL	96,30%	91,40%	100%	100%	Pheochromocytoma, 171300 Erythrocytosis, familial, 2, 263400 von Hippel-Lindau syndrome, 193300 Renal cell carcinoma, somatic, 144700 Hemangioblastoma, cerebellar, somatic, 0
VIPAS39	100%	100%	100%	100%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VKORC1	100%	100%	93,00%	93,00%	Warfarin resistance, 122700 Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473
VLDLR	100%	99,80%	100%	100%	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VPS11	94,90%	93,60%	100%	100%	Leukodystrophy, hypomyelinating, 12, 616683
VPS13A	99,40%	95,60%	100%	100%	Choreoacanthocytosis, 200150
VPS13B	99,50%	98,20%	99,50%	99,40%	Cohen syndrome, 216550
VPS13C	99,40%	96,90%	100%	100%	Parkinson disease 23, autosomal recessive, early onset, 616840
VPS13D	100%	99,70%	100%	100%	Spinocerebellar ataxia, autosomal recessive 4, 607317
VPS33A	97,30%	95,70%	95,80%	95,80%	Mucopolysaccharidosis-plus syndrome, 617303
VPS33B	100%	100%	100%	100%	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
VPS37A	91,30%	78,20%	100%	100%	Spastic paraplegia 53, autosomal recessive, 614898
VPS45	99,20%	95,60%	95,10%	95,10%	Neutropenia, severe congenital, 5, autosomal recessive, 615285
VPS51	95,00%	83,20%	100%	100%	Pontocerebellar hypoplasia, type 13, 618606
VPS53	91,50%	90,70%	100%	99,30%	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	99,70%	98,50%	100%	100%	Pontocerebellar hypoplasia type 1A, 607596
VSX2	100%	99,30%	100%	100%	Microphthalmia with coloboma 3, 610092 Microphthalmia, isolated 2, 610093
VWA3B	100%	99,70%	100%	100%	?Spinocerebellar ataxia, autosomal recessive 22, 616948
VWF	99,80%	98,60%	100%	100%	von Willebrand disease, type 1, 193400 von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 von Willibrand disease, type 3, 277480
WARS2	100%	99,40%	100%	100%	Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710
WASHC4	99,10%	95,50%	100%	100%	?Mental retardation, autosomal recessive 43, 615817
WASHC5	100%	99,80%	100%	100%	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, autosomal dominant, 603563
WBP2	100%	99,70%	100%	100%	Deafness, autosomal recessive 107, 617639
WDPCP	98,20%	94,40%	98,10%	98,10%	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR19	100%	99,40%	100%	100%	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307

					?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WDR34	100%	99,60%	100%	100%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
WDR35	99,80%	98,90%	100%	100%	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 Cranioectodermal dysplasia 2, 613610
WDR4	100%	100%	100%	100%	Microcephaly, growth deficiency, seizures, and brain malformations, 618346 Galloway-Mowat syndrome 6, 618347
WDR45B	98,00%	89,20%	100%	100%	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977
WDR60	99,50%	97,00%	100%	100%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WDR62	100%	99,50%	100%	100%	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
WDR72	96,80%	96,40%	96,90%	96,90%	Amelogenesis imperfecta, type IIA3, 613211
WDR73	100%	100%	100%	100%	Galloway-Mowat syndrome 1, 251300
WDR81	100%	100%	100%	100%	Hydrocephalus, congenital, 3, with brain anomalies, 617967 Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185
WEE2	100%	99,60%	100%	100%	Oocyte maturation defect 5, 617996
WFS1	100%	99,90%	100%	100%	?Cataract 41, 116400 Deafness, autosomal dominant 6/14/38, 600965 Wolfram-like syndrome, autosomal dominant, 614296 Wolfram syndrome 1, 222300
WHRN	99,80%	98,10%	100%	100%	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
WIPF1	100%	99,90%	100%	100%	?Wiskott-Aldrich syndrome 2, 614493
WIPI2	100%	99,30%	100%	100%	?Intellectual developmental disorder with short stature and variable skeletal anomalies, 618453
WISP3	100%	100%	100%	100%	Spondyloepiphyseal dysplasia tarda with progressive arthropathy, 208230 Arthropathy, progressive pseudorheumatoid, of childhood, 208230
WNK1	99,90%	99,60%	100%	100%	Pseudohypoaldosteronism, type IIC, 614492 Neuropathy, hereditary sensory and autonomic, type II, 201300
WNT1	99,30%	95,30%	100%	100%	Osteogenesis imperfecta, type XV, 615220
WNT10A	100%	99,40%	100%	100%	Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400 Odontoonychodermal dysplasia, 257980
WNT10B	100%	99,40%	100%	100%	Split-hand/foot malformation 6, 225300 Tooth agenesis, selective, 8, 617073
WNT3	100%	99,60%	100%	100%	?Tetra-amelia syndrome 1, 273395
WNT4	99,10%	94,80%	98,90%	96,20%	?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%	100%	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	99,90%	98,80%	100%	100%	Werner syndrome, 277700
WWOX	100%	100%	100%	100%	Esophageal squamous cell carcinoma, somatic, 133239 Spinocerebellar ataxia, autosomal recessive 12, 614322 Epileptic encephalopathy, early infantile, 28, 616211
XDH	100%	99,90%	100%	100%	Xanthinuria, type I, 278300
XPA	99,60%	95,60%	100%	100%	Xeroderma pigmentosum, group A, 278700
XPC	100%	100%	100%	100%	Xeroderma pigmentosum, group C, 278720
XPNPEP3	100%	100%	100%	100%	Nephronophthisis-like nephropathy 1, 613159
XRCC1	100%	98,80%	100%	100%	?Spinocerebellar ataxia, autosomal recessive 26, 617633
XRCC2	99,80%	97,40%	100%	100%	?Fanconi anemia, complementation group U, 617247
XRCC4	99,90%	99,30%	100%	100%	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	97,40%	89,60%	98,10%	94,80%	Desbuquois dysplasia 2, 615777
XYLT2	100%	98,30%	96,70%	96,70%	Spondyloocular syndrome, 605822
YARS2	100%	99,80%	100%	100%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
YME1L1	99,00%	95,20%	100%	100%	?Optic atrophy 11, 617302
YY1AP1	99,30%	98,20%	100%	100%	Grange syndrome, 602531
ZAP70	100%	99,30%	100%	100%	Immunodeficiency 48, 269840 Autoimmune disease, multisystem, infantile-onset, 2, 617006
ZBTB11	99,90%	99,60%	100%	100%	Intellectual developmental disorder, autosomal recessive 69, 618383
ZBTB16	100%	99,90%	100%	100%	Skeletal defects, genital hypoplasia, and mental retardation, 612447 Leukemia, acute promyelocytic, PL2F/RARA type, 0
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	99,90%	98,90%	100%	100%	Mental retardation, autosomal recessive 56, 617125
ZFYVE26	100%	99,10%	100%	100%	Spastic paraplegia 15, autosomal recessive, 270700
ZMPSTE24	100%	99,90%	100%	100%	Restrictive dermopathy, lethal, 275210 Mandibuloacral dysplasia with type B lipodystrophy, 608612
ZMYND10	100%	100%	100%	100%	Ciliary dyskinesia, primary, 22, 615444
ZNF142	100%	99,90%	100%	100%	Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425
ZNF335	100%	99,90%	100%	100%	Microcephaly 10, primary, autosomal recessive, 615095
ZNF341	97,20%	95,00%	100%	100%	Hyper-IgE recurrent infection syndrome 3, autosomal recessive, 618282
ZNF408	100%	100%	100%	100%	?Exudative vitreoretinopathy 6, 616468 Retinitis pigmentosa 72, 616469

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	74,40%	74,40%	74,60%	74,40%	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.
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-DNA coding genes.
non-DNA coding genes are covered, but as coverage statistics are based on DNA coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : April 20th , 2020.
This list is accurate for panel version DG 2.18
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors