MENDELIOME GENE PANEL DG 2.18 (3839 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
A4GALT	100%	100%	100%	100%	NOR polyagglutination syndrome, 111400
AAAS	100%	99,90%	100%	100%	Achalasia-addisonianism-alacrimia syndrome, 231550
AAGAB	100%	99,20%	100%	100%	Keratoderma, palmoplantar, punctate type IA, 148600
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
ABCA5	98,40%	92,30%	100%	100%	?Hypertrichosis, congenital generalized, with gingival hyperplasia, 135400
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
ABCB6	100%	99,80%	100%	100%	Dyschromatosis universalis hereditaria 3, 615402 Microphthalmia, isolated, with coloboma 7, 614497 Pseudohyperkalemia, familial, 2, due to red cell leak, 609153
ABCB7	99,50%	98,20%	99,80%	99,30%	Anemia, sideroblastic, with ataxia, 301310
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
ABCC9	100%	99,90%	100%	100%	Hypertrichotic osteochondrodysplasia, 239850 ?Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569
ABCD1	75,80%	71,60%	100%	100%	Adrenomyeloneuropathy, adult, 300100 Adrenoleukodystrophy, 300100
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
ABL1	100%	100%	100%	100%	Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 608232 Congenital heart defects and skeletal malformations syndrome, 617602
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
ACP4	97,20%	88,80%	100%	100%	Amelogenesis imperfecta, type IJ, 617297
ACP5	99,80%	98,30%	100%	100%	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACSF3	100%	99,90%	100%	100%	Combined malonic and methylmalonic aciduria, 614265

ACSL4	98,70%	94,60%	100%	100%	Mental retardation, X-linked 63, 300387
ACSL6	99,30%	97,80%	100%	100%	Myelodysplastic syndrome, 0 Myelogenous leukemia, acute, 0
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
ACTA2	100%	99,00%	100%	100%	Aortic aneurysm, familial thoracic 6, 611788 Multisystemic smooth muscle dysfunction syndrome, 613834 Moyamoya disease 5, 614042
ACTB	99,70%	96,10%	100%	100%	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACTC1	100%	99,70%	100%	100%	Left ventricular noncompaction 4, 613424 Atrial septal defect 5, 612794 Cardiomyopathy, dilated, 1R, 613424 Cardiomyopathy, hypertrophic, 11, 612098
ACTG1	100%	100%	100%	100%	Baraitser-Winter syndrome 2, 614583 Deafness, autosomal dominant 20/26, 604717
ACTG2	99,90%	98,20%	100%	100%	Visceral myopathy, 155310
ACTL6B	100%	99,80%	100%	100%	Epileptic encephalopathy, early infantile, 76, 618468 Intellectual developmental disorder with severe speech and ambulation defects, 618470
ACTN1	100%	100%	100%	100%	Bleeding disorder, platelet-type, 15, 615193
ACTN2	100%	100%	100%	100%	Myopathy, distal, 6, adult onset, 618655 Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158 Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158 Myopathy, congenital with structured cores and Z-line abnormalities, 618654
ACTN4	100%	99,30%	100%	100%	Glomerulosclerosis, focal segmental, 1, 603278
ACVR1	100%	100%	100%	100%	Fibrodysplasia ossificans progressiva, 135100
ACVR1B	99,70%	97,40%	100%	100%	Pancreatic cancer, somatic, 0
ACVR2B	98,30%	95,00%	100%	100%	Heterotaxy, visceral, 4, autosomal, 613751
ACVRL1	100%	98,90%	100%	100%	Telangiectasia, hereditary hemorrhagic, type 2, 600376
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
ADAM10	94,80%	93,90%	100%	100%	Reticulate acropigmentation of Kitamura, 615537

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
ADCY5	95,10%	91,20%	99,20%	98,00%	Dyskinesia, familial, with facial myokymia, 606703
ADCY6	100%	100%	100%	100%	?Lethal congenital contracture syndrome 8, 616287
ADD3	99,90%	99,50%	100%	100%	Cerebral palsy, spastic quadriplegic, 3, 617008
ADGRE2	96,80%	96,10%	99,10%	98,70%	Vibratory urticaria, 125630
ADGRG1	100%	100%	100%	100%	Polymicrogyria, bilateral perisylvian, 615752 Polymicrogyria, bilateral frontoparietal, 606854
ADGRG2	98,30%	92,70%	100%	100%	Congenital bilateral absence of vas deferens, X-linked, 300985
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
ADIPOQ	100%	100%	100%	100%	Adiponectin deficiency, 612556
ADK	99,50%	95,80%	100%	100%	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADNP	100%	100%	100%	100%	Helsmoortel-van der Aa syndrome, 615873
ADPRHL2	100%	99,80%	100%	100%	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
ADRB2	100%	99,70%	100%	100%	Beta-2-adrenoreceptor agonist, reduced response to, 0
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
AFF2	99,90%	99,40%	100%	99,80%	Mental retardation, X-linked, FRAXE type, 309548
AFF4	99,90%	98,90%	100%	100%	CHOPS syndrome, 616368

AFG3L2	95,00%	91,10%	100%	99,90%	Spastic ataxia 5, autosomal recessive, 614487 Spinocerebellar ataxia 28, 610246
AFP	96,90%	89,80%	100%	100%	Alpha-fetoprotein deficiency, 615969
AGA	100%	100%	100%	100%	Aspartylglucosaminuria, 208400
AGBL1	98,50%	98,50%	100%	100%	Corneal dystrophy, Fuchs endothelial, 8, 615523
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
AHDC1	100%	99,30%	100%	100%	Xia-Gibbs syndrome, 615829
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
AIFM1	99,90%	98,80%	100%	100%	Cowchock syndrome, 310490 Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 Combined oxidative phosphorylation deficiency 6, 300816 Deafness, X-linked 5, 300614
AIMP1	99,20%	94,50%	100%	99,90%	Leukodystrophy, hypomyelinating, 3, 260600
AIMP2	88,90%	86,00%	100%	100%	Leukodystrophy, hypomyelinating, 17, 618006
AIP	100%	99,00%	100%	100%	Pituitary adenoma 1, multiple types, 102200 Pituitary adenoma predisposition, 102200
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
AK7	99,70%	98,10%	100%	100%	?Spermatogenic failure 27, 617965
AKAP9	98,80%	95,50%	100%	100%	?Long QT syndrome 11, 611820
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
AKT1	100%	99,50%	100%	100%	Breast cancer, somatic, 114480 Cowden syndrome 6, 615109 Proteus syndrome, somatic, 176920 Ovarian cancer, somatic, 167000 Colorectal cancer, somatic, 114500
AKT2	100%	99,50%	100%	100%	Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900 Diabetes mellitus, type II, 125853
AKT3	98,70%	94,50%	100%	100%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937
AL032819.3	1,80%	0,00%	100%	100%	Diarrhea 11, malabsorptive, congenital, 618662
ALAD	99,30%	94,10%	100%	100%	Porphyria, acute hepatic, 612740
ALAS2	98,90%	94,90%	100%	100%	Protoporphyria, erythropoietic, X-linked, 300752 Anemia, sideroblastic, 1, 300751
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
ALDH2	100%	100%	100%	100%	Alcohol sensitivity, acute, 610251
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
ALG13	98,40%	92,60%	100%	99,60%	Epileptic encephalopathy, early infantile, 36, 300884 ?Congenital disorder of glycosylation, type Is, 300884
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
AMELX	99,90%	96,80%	100%	100%	Amelogenesis imperfecta, type 1E, 301200
AMER1	99,90%	98,50%	100%	100%	Osteopathia striata with cranial sclerosis, 300373
AMH	96,40%	83,80%	100%	99,80%	Persistent Mullerian duct syndrome, type I, 261550
AMHR2	100%	99,50%	100%	100%	Persistent Mullerian duct syndrome, type II, 261550
AMMECR1	100%	99,10%	100%	100%	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
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

AMTN	99,60%	98,60%	100%	100%	?Amelogenesis imperfecta, type IIIB, 617607
ANAPC1	59,40%	57,70%	100%	99,90%	Rothmund-Thomson syndrome, type 1, 618625
ANG	100%	100%	100%	100%	Amyotrophic lateral sclerosis 9, 611895
ANGPTL3	98,80%	95,40%	100%	100%	Hypobetalipoproteinemia, familial, 2, 605019
ANGPTL4	100%	99,20%	100%	100%	Plasma triglyceride level QTL, low, 615881
ANK1	100%	99,40%	100%	100%	Spherocytosis, type 1, 182900
ANK2	100%	100%	100%	100%	Cardiac arrhythmia, ankyrin-B-related, 600919 Long QT syndrome 4, 600919
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
ANKRD11	97,50%	94,80%	100%	100%	KBG syndrome, 148050
ANKRD26	95,00%	89,30%	97,20%	97,20%	Thrombocytopenia 2, 188000
ANKS6	93,80%	89,50%	97,90%	95,80%	Nephronophthisis 16, 615382
ANLN	98,70%	97,50%	100%	100%	Focal segmental glomerulosclerosis 8, 616032
ANO10	99,80%	97,90%	100%	100%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANO3	99,50%	97,80%	100%	100%	Dystonia 24, 615034
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
ANOS1	89,80%	88,90%	99,90%	99,40%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
ANTXR1	99,70%	97,90%	100%	100%	GAPO syndrome, 230740
ANTXR2	100%	98,20%	100%	100%	Hyaline fibromatosis syndrome, 228600
ANXA11	100%	98,50%	100%	100%	Amytrophic lateral sclerosis 23, 617839
AP1S1	99,90%	99,50%	100%	100%	MEDNIK syndrome, 609313
AP1S2	76,40%	67,90%	100%	100%	Mental retardation, X-linked syndromic 5, 304340
AP2M1	100%	100%	100%	100%	Intellectual developmental disorder 60 with seizures, 618587
AP2S1	90,40%	90,30%	100%	100%	Hypocalciuric hypercalcemia, type III, 600740
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
APC	100%	99,70%	100%	100%	Desmoid disease, hereditary, 135290 Adenomatous polyposis coli, 175100 Gardner syndrome, 175100 Hepatoblastoma, somatic, 114550 Colorectal cancer, somatic, 114500 Brain tumor-polyposis syndrome 2, 175100 Gastric cancer, somatic, 613659 Adenoma, periampullary, somatic, 0
APC2	97,60%	92,70%	99,90%	99,10%	?Sotos syndrome 3, 617169 Cortical dysplasia, complex, with other brain malformations 10, 618677
APCDD1	100%	99,80%	100%	100%	Hypotrichosis 1, 605389
APOA1	100%	100%	100%	100%	Hypoalphalipoproteinemia, primary, 2, with or without corneal clouding, 618463 Amyloidosis, 3 or more types, 105200 ApoA-I and apoC-III deficiency, combined, 618463
APOA2	84,20%	81,50%	100%	100%	Apolipoprotein A-II deficiency, 0
APOA5	100%	99,90%	100%	100%	Hyperchylomicronemia, late-onset, 144650
APOB	99,80%	99,30%	100%	100%	Hypobetalipoproteinemia, 615558 Hypercholesterolemia, familial, 2, 144010
APOC2	100%	100%	100%	100%	Hyperlipoproteinemia, type Ib, 207750
APOC3	100%	100%	100%	100%	Apolipoprotein C-III deficiency, 614028
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
APP	100%	99,90%	100%	100%	Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants, 605714 Alzheimer disease 1, familial, 104300
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
AQP5	100%	97,00%	100%	100%	Palmoplantar keratoderma, Bothnian type, 600231
AR	97,60%	93,20%	100%	99,20%	Hypospadias 1, X-linked, 300633 Androgen insensitivity, 300068 Androgen insensitivity, partial, with or without breast cancer, 312300 Spinal and bulbar muscular atrophy of Kennedy, 313200
ARCN1	97,00%	96,60%	96,90%	96,60%	Short stature, rhizomelic, with microcephaly, micrognathia, and developmental delay, 617164

ARF1	100%	100%	100%	100%	Periventricular nodular heterotopia 8, 618185
ARFGEF2	99,90%	99,10%	100%	100%	Periventricular heterotopia with microcephaly, 608097
ARG1	100%	100%	100%	100%	Argininemia, 207800
ARHGAP26	100%	99,90%	100%	100%	Leukemia, juvenile myelomonocytic, somatic, 607785
ARHGAP31	99,90%	98,80%	100%	100%	Adams-Oliver syndrome 1, 100300
ARHGDIA	100%	100%	100%	100%	Nephrotic syndrome, type 8, 615244
ARHGEF1	99,90%	98,40%	100%	100%	?Immunodeficiency 62, 618459
ARHGEF10	99,80%	98,00%	100%	100%	?Slowed nerve conduction velocity, AD, 608236
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
ARHGEF9	76,50%	74,10%	97,20%	97,10%	Epileptic encephalopathy, early infantile, 8, 300607
ARID1A	98,10%	96,40%	100%	100%	Coffin-Siris syndrome 2, 614607
ARID1B	99,50%	98,60%	99,90%	99,20%	Coffin-Siris syndrome 1, 135900
ARID2	99,80%	98,40%	100%	100%	Coffin-Siris syndrome 6, 617808
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
ARMC2	100%	99,20%	100%	100%	Spermatogenic failure 38, 618433
ARMC4	95,60%	93,50%	100%	100%	Ciliary dyskinesia, primary, 23, 615451
ARMC5	100%	99,40%	100%	100%	ACTH-independent macronodular adrenal hyperplasia 2, 615954
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
ARR3	100%	99,80%	100%	100%	Myopia 26, X-linked, female-limited, 301010
ARSA	100%	99,80%	100%	100%	Metachromatic leukodystrophy, 250100
ARSB	96,90%	88,30%	100%	100%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARSE	99,00%	93,00%	100%	99,90%	Chondrodysplasia punctata, X-linked recessive, 302950
ARSG	100%	99,50%	100%	100%	Usher syndrome, type IV, 618144
ARV1	100%	99,90%	100%	100%	Epileptic encephalopathy, early infantile, 38, 617020
ARX	81,00%	64,00%	91,50%	85,70%	Proud syndrome, 300004 Partington syndrome, 309510 Lissencephaly, X-linked 2, 300215 Epileptic encephalopathy, early infantile, 1, 308350

					Mental retardation, X-linked 29 and others, 300419 Hydranencephaly with abnormal genitalia, 300215
ASAH1	99,70%	98,60%	100%	100%	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASB10	99,40%	95,70%	100%	100%	Glaucoma 1, open angle, F, 603383
ASCC1	97,70%	94,40%	91,10%	91,10%	Barrett esophagus/esophageal adenocarcinoma, 614266 Spinal muscular atrophy with congenital bone fractures 2, 616867
ASCL1	100%	97,60%	100%	100%	Haddad syndrome, 209880 Central hypoventilation syndrome, congenital, 209880
ASH1L	98,70%	98,60%	98,70%	98,70%	Mental retardation, autosomal dominant 52, 617796
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
ASPSCR1	99,70%	97,80%	100%	100%	Alveolar soft-part sarcoma, 606243
ASS1	95,40%	87,90%	100%	100%	Citrullinemia, 215700
ASXL1	100%	99,50%	99,90%	99,90%	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ASXL2	99,70%	98,90%	100%	100%	Shashi-Pena syndrome, 617190
ASXL3	99,90%	99,70%	100%	100%	Bainbridge-Ropers syndrome, 615485
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
ATL1	100%	99,70%	100%	100%	Spastic paraplegia 3A, autosomal dominant, 182600 Neuropathy, hereditary sensory, type ID, 613708
ATL3	99,80%	98,30%	100%	100%	Neuropathy, hereditary sensory, type IF, 615632
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
ATN1	99,90%	98,20%	100%	100%	Dentatorubral-pallidoluysian atrophy, 125370 Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494

ATOH7	96,00%	91,20%	99,10%	94,40%	Persistent hyperplastic primary vitreous, autosomal recessive, 221900
ATP11C	98,70%	93,80%	100%	99,60%	?Hemolytic anemia, congenital, X-linked, 301015
ATP13A2	100%	99,50%	100%	100%	Kufor-Rakeb syndrome, 606693 Spastic paraplegia 78, autosomal recessive, 617225
ATP1A1	100%	100%	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2DD, 618036 Hypomagnesemia, seizures, and mental retardation 2, 618314
ATP1A2	100%	100%	100%	100%	Migraine, familial hemiplegic, 2, 602481 Migraine, familial basilar, 602481 Alternating hemiplegia of childhood 1, 104290
ATP1A3	100%	99,90%	100%	100%	CAPOS syndrome, 601338 Alternating hemiplegia of childhood 2, 614820 Dystonia-12, 128235
ATP2A1	100%	100%	100%	100%	Brody myopathy, 601003
ATP2A2	100%	100%	100%	100%	Acrokeratosis verruciformis, 101900 Darier disease, 124200
ATP2B3	99,50%	97,50%	100%	100%	?Spinocerebellar ataxia, X-linked 1, 302500
ATP2C1	100%	99,60%	100%	100%	Hailey-Hailey disease, 169600
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
ATP6AP1	98,20%	92,10%	100%	100%	Immunodeficiency 47, 300972
ATP6AP2	94,10%	76,60%	100%	100%	Congenital disorder of glycosylation, type IIr, 301045 Mental retardation, X-linked, syndromic, Hedera type, 300423 ?Parkinsonism with spasticity, X-linked, 300911
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
ATP6V1B2	100%	99,30%	100%	100%	Zimmermann-Laband syndrome 2, 616455 Deafness, congenital, with onychodystrophy, autosomal dominant, 124480
ATP6V1E1	93,10%	88,30%	100%	100%	Cutis laxa, autosomal recessive, type IIC, 617402
ATP7A	99,70%	97,50%	100%	100%	Occipital horn syndrome, 304150 Menkes disease, 309400 Spinal muscular atrophy, distal, X-linked 3, 300489
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
ATRX	99,40%	96,30%	100%	100%	Alpha-thalassemia/mental retardation syndrome, 301040 Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Mental retardation-hypotonic facies syndrome, X-linked, 309580
ATXN1	100%	99,70%	100%	100%	Spinocerebellar ataxia 1, 164400
ATXN10	99,90%	99,20%	100%	100%	Spinocerebellar ataxia 10, 603516
ATXN2	92,70%	85,70%	99,10%	96,80%	Spinocerebellar ataxia 2, 183090
ATXN3	94,50%	89,80%	95,80%	95,80%	Machado-Joseph disease, 109150
ATXN7	99,80%	97,60%	98,60%	97,40%	Spinocerebellar ataxia 7, 164500
ATXN8OS	NC	NC	NC	NC	Spinocerebellar ataxia 8, 608768
AUH	100%	99,80%	100%	100%	3-methylglutaconic aciduria, type I, 250950
AURKC	100%	99,20%	100%	100%	Spermatogenic failure 5, 243060
AUTS2	98,20%	95,80%	100%	100%	Mental retardation, autosomal dominant 26, 615834
AVIL	100%	99,90%	100%	100%	Nephrotic syndrome, type 21, 618594
AVP	84,90%	64,30%	100%	99,90%	Diabetes insipidus, neurohypophyseal, 125700
AVPR2	100%	99,40%	100%	100%	Nephrogenic syndrome of inappropriate antidiuresis, 300539 Diabetes insipidus, nephrogenic, 304800
AXIN1	100%	99,60%	100%	100%	Hepatocellular carcinoma, somatic, 114550 ?Caudal duplication anomaly, 607864
AXIN2	100%	99,90%	100%	99,90%	Colorectal cancer, somatic, 114500 Oligodontia-colorectal cancer syndrome, 608615
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
BACH2	100%	100%	100%	100%	Immunodeficiency 60, 618394
BAG3	100%	100%	100%	100%	Cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954
BANF1	98,30%	86,60%	100%	100%	Nestor-Guillermo progeria syndrome, 614008
BAP1	84,40%	83,00%	100%	100%	Tumor predisposition syndrome, 614327
BAX	98,00%	95,40%	100%	100%	T-cell acute lymphoblastic leukemia, somatic, 613065 Colorectal cancer, somatic, 114500
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
BCAP31	92,60%	83,20%	100%	99,90%	Deafness, dystonia, and cerebral hypomyelination, 300475
BCHE	100%	99,90%	100%	100%	Butyrylcholinesterase deficiency, 617936
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
BCL11A	99,80%	98,10%	100%	100%	Dias-Logan syndrome, 617101
BCL11B	99,10%	95,60%	98,80%	97,30%	Immunodeficiency 49, 617237 Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092
BCL2	100%	100%	100%	100%	Leukemia/lymphoma, B-cell, 2, 0
BCL7A	100%	100%	100%	100%	B-cell non-Hodgkin lymphoma, high-grade, 0

BCO1	100%	100%	100%	100%	?Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300
BCOR	99,60%	97,40%	100%	99,90%	Microphthalmia, syndromic 2, 300166
BCORL1	99,60%	97,90%	100%	100%	Shukla-Vernon syndrome, 301029
BCS1L	100%	100%	100%	100%	Leigh syndrome, 256000 GRACILE syndrome, 603358 Bjornstad syndrome, 262000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BDP1	98,80%	95,30%	100%	100%	?Deafness, autosomal recessive 112, 618257
BEAN1	98,70%	96,40%	92,20%	92,20%	Spinocerebellar ataxia 31, 117210
BEST1	99,40%	96,40%	99,90%	99,40%	Retinitis pigmentosa-50, 613194 Bestrophinopathy, autosomal recessive, 611809 Retinitis pigmentosa, concentric, 613194 Vitreoretinochoroidopathy, 193220 Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma, 193220 Macular dystrophy, vitelliform, 2, 153700
BFSP1	99,00%	89,90%	100%	99,90%	Cataract 33, multiple types, 611391
BFSP2	99,80%	97,60%	100%	100%	Cataract 12, multiple types, 611597
BGN	100%	100%	100%	100%	Meester-Loeys syndrome, 300989 Spondyloepimetaphyseal dysplasia, X-linked, 300106
BHLHA9	70,90%	50,40%	99,80%	97,30%	Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432 ?Camptosynpolydactyly, complex, 607539
BICD2	100%	99,70%	100%	100%	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291 Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290
BIN1	99,60%	95,70%	100%	100%	Centronuclear myopathy 2, 255200
BLK	100%	100%	100%	100%	Maturity-onset diabetes of the young, type 11, 613375
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
BMP15	100%	99,30%	100%	100%	Ovarian dysgenesis 2, 300510 Premature ovarian failure 4, 300510
BMP2	100%	100%	100%	100%	Brachydactyly, type A2, 112600 Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 617877
BMP4	100%	100%	100%	100%	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625
BMPER	100%	99,80%	100%	100%	Diaphanospondylodysostosis, 608022

BMPR1A	99,80%	96,60%	100%	100%	Polyposis syndrome, hereditary mixed, 2, 610069 Polyposis, juvenile intestinal, 174900 Juvenile polyposis syndrome, infantile form, 174900
BMPR1B	100%	99,90%	100%	100%	Brachydactyly, type A2, 112600 Brachydactyly, type A1, D, 616849 Acromesomelic dysplasia, Demirhan type, 609441
BMPR2	99,90%	99,90%	99,90%	99,90%	Pulmonary venoocclusive disease 1, 265450 Pulmonary hypertension, familial primary, 1, with or without HHT, 178600 Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600
BMS1	66,70%	66,40%	100%	100%	?Aplasia cutis congenita, nonsyndromic, 107600
BNC2	99,10%	99,10%	100%	100%	Lower urinary tract obstruction, congenital, 618612
BOLA3	99,40%	90,20%	100%	100%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BPGM	100%	100%	100%	100%	Erythrocytosis, familial, 8, 222800
BPTF	96,10%	94,20%	99,60%	98,60%	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, 617755
BRAF	95,60%	85,10%	100%	100%	Noonan syndrome 7, 613706 Cardiofaciocutaneous syndrome, 115150 Adenocarcinoma of lung, somatic, 211980 LEOPARD syndrome 3, 613707 Nonsmall cell lung cancer, somatic, 0 Melanoma, malignant, somatic, 0 Colorectal cancer, somatic, 0
BRAT1	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
BRDT	95,90%	91,60%	100%	100%	?Spermatogenic failure 21, 617644
BRF1	99,90%	98,40%	100%	100%	Cerebellofaciodental syndrome, 616202
BRIP1	99,90%	99,00%	100%	100%	Fanconi anemia, complementation group J, 609054
BRPF1	100%	100%	100%	100%	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333
BRWD3	99,30%	97,20%	100%	100%	Mental retardation, X-linked 93, 300659
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

BTK	100%	99,90%	100%	99,90%	Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200 Agammaglobulinemia, X-linked 1, 300755
BUB1	99,80%	98,80%	100%	100%	Colorectal cancer with chromosomal instability, somatic, 114500
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
C11orf70	99,30%	95,90%	100%	100%	Ciliary dyskinesia, primary, 38, 618063
C11orf80	98,20%	93,70%	92,50%	92,50%	Hydatidiform mole, recurrent, 4, 618432
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
C1GALT1C1	100%	99,50%	100%	100%	Tn polyagglutination syndrome, somatic, 300622
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
C1QTNF5	90,90%	78,50%	100%	100%	Retinal degeneration, late-onset, autosomal dominant, 605670
C1R	100%	100%	99,00%	96,90%	Ehlers-Danlos syndrome, periodontal type, 1, 130080
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
C6	100%	99,70%	100%	100%	C6 deficiency, 612446 Combined C6/C7 deficiency, 0
C7	100%	98,90%	100%	100%	C7 deficiency, 610102
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
C9orf72	99,90%	98,30%	100%	100%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1, 105550
CA12	100%	100%	100%	100%	Hyperchlorhidrosis, isolated, 143860
CA2	100%	100%	100%	100%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA4	100%	100%	100%	100%	Retinitis pigmentosa 17, 600852
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
CACNA1A	95,10%	91,90%	100%	100%	Spinocerebellar ataxia 6, 183086 Epileptic encephalopathy, early infantile, 42, 617106 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Episodic ataxia, type 2, 108500 Migraine, familial hemiplegic, 1, 141500
CACNA1B	97,50%	95,70%	99,10%	97,70%	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497
CACNA1C	99,90%	99,20%	100%	100%	Timothy syndrome, 601005 Long QT syndrome 8, 618447 Brugada syndrome 3, 611875
CACNA1D	98,00%	97,90%	100%	100%	Sinoatrial node dysfunction and deafness, 614896 Primary aldosteronism, seizures, and neurologic abnormalities, 615474
CACNA1E	100%	99,90%	100%	100%	Epileptic encephalopathy, early infantile, 69, 618285
CACNA1F	99,70%	97,50%	100%	100%	Cone-rod dystrophy, X-linked, 3, 300476 Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071 Aland Island eye disease, 300600
CACNA1G	100%	99,60%	100%	100%	Spinocerebellar ataxia 42, 616795 Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087
CACNA1H	98,70%	96,40%	100%	99,90%	Hyperaldosteronism, familial, type IV, 617027

CACNA1S	100%	99,90%	100%	100%	Hypokalemic periodic paralysis, type 1, 170400
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
CACNB2	100%	100%	100%	100%	Brugada syndrome 4, 611876
CACNB4	95,50%	94,30%	100%	100%	Episodic ataxia, type 5, 613855
CACNG2	100%	100%	100%	100%	?Mental retardation, autosomal dominant 10, 614256
CAD	100%	99,20%	100%	100%	Epileptic encephalopathy, early infantile, 50, 616457
CALM1	100%	99,40%	100%	100%	Long QT syndrome 14, 616247 Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916
CALM2	67,80%	65,10%	72,00%	72,00%	Long QT syndrome 15, 616249
CALR	94,80%	89,10%	100%	100%	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
CAMK2A	100%	99,60%	100%	100%	?Mental retardation, autosomal recessive 63, 618095 Mental retardation, autosomal dominant 53, 617798
CAMK2B	100%	99,80%	100%	100%	Mental retardation, autosomal dominant 54, 617799
CAMK2G	99,90%	98,10%	100%	100%	Mental retardation, autosomal dominant 59, 618522
CAMTA1	100%	99,50%	100%	100%	Cerebellar ataxia, nonprogressive, with mental retardation, 614756
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
CAPN3	100%	99,30%	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600 Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129
CAPN5	100%	100%	100%	100%	Vitreoretinopathy, neovascular inflammatory, 193235
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
CARD14	100%	99,10%	100%	100%	Pityriasis rubra pilaris, 173200 Psoriasis 2, 602723
CARD9	99,90%	98,40%	100%	100%	Candidiasis, familial, 2, autosomal recessive, 212050
CARMIL2	96,30%	94,50%	99,70%	98,20%	Immunodeficiency 58, 618131
CARS2	100%	100%	100%	99,20%	Combined oxidative phosphorylation deficiency 27, 616672
CASK	99,60%	96,80%	100%	100%	Mental retardation, with or without nystagmus, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 FG syndrome 4, 300422
CASP10	99,50%	97,30%	100%	100%	Lymphoma, non-Hodgkin, somatic, 605027 Gastric cancer, somatic, 613659 Autoimmune lymphoproliferative syndrome, type II, 603909
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
CASQ1	100%	99,50%	100%	100%	Myopathy, vacuolar, with CASQ1 aggregates, 616231
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
CAV3	100%	100%	100%	100%	Creatine phosphokinase, elevated serum, 123320 Long QT syndrome 9, 611818 Myopathy, distal, Tateyama type, 614321 Rippling muscle disease 2, 606072 Cardiomyopathy, familial hypertrophic, 192600
CAVIN1	100%	100%	100%	100%	Lipodystrophy, congenital generalized, type 4, 613327
CBL	97,30%	97,10%	100%	100%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
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
CCDC22	99,60%	96,50%	100%	100%	Ritscher-Schinzel syndrome 2, 300963
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
CCDC50	100%	99,70%	100%	100%	?Deafness, autosomal dominant 44, 607453
CCDC65	99,60%	97,10%	100%	100%	Ciliary dyskinesia, primary, 27, 615504
CCDC78	100%	100%	100%	100%	?Centronuclear myopathy 4, 614807
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
CCM2	98,70%	97,80%	100%	100%	Cerebral cavernous malformations-2, 603284
CCND2	100%	100%	100%	100%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938
CCNK	92,60%	89,80%	100%	99,80%	?Intellectual developmental disorder with hypertelorism and distinctive facies, 618147
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
CD164	99,10%	94,80%	100%	100%	?Deafness, autosomal dominant 66, 616969
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
CD36	99,70%	98,70%	100%	100%	Platelet glycoprotein IV deficiency, 608404
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
CD4	100%	99,90%	100%	100%	OKT4 epitope deficiency, 613949
CD40	100%	100%	100%	100%	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	97,30%	88,10%	100%	100%	Immunodeficiency, X-linked, with hyper-IgM, 308230
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
CD70	99,80%	97,70%	100%	100%	Lymphoproliferative syndrome 3, 618261
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
CD96	99,90%	99,70%	100%	100%	C syndrome, 211750
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
CDC42	97,80%	90,70%	100%	100%	Takenouchi-Kosaki syndrome, 616737
CDC45	99,80%	98,50%	100%	100%	Meier-Gorlin syndrome 7, 617063
CDC6	100%	100%	100%	100%	?Meier-Gorlin syndrome 5, 613805
CDC73	100%	99,40%	100%	100%	Parathyroid adenoma with cystic changes, 145001 Hyperparathyroidism-jaw tumor syndrome, 145001 Parathyroid carcinoma, 608266 Hyperparathyroidism, familial primary, 145000
CDCA7	100%	99,60%	100%	100%	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910
CDH1	99,20%	99,10%	96,10%	96,00%	Endometrial carcinoma, somatic, 608089 Blepharocheilodontic syndrome 1, 119580 Gastric cancer, hereditary diffuse, with or without cleft lip and/or palate, 137215 Ovarian cancer, somatic, 167000
CDH11	100%	100%	100%	100%	Elsahy-Waters syndrome, 211380
CDH15	99,90%	98,70%	100%	100%	Mental retardation, autosomal dominant 3, 612580
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
CDK13	98,00%	92,70%	100%	100%	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360
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
CDKL5	95,00%	93,50%	95,60%	95,00%	Epileptic encephalopathy, early infantile, 2, 300672
CDKN1B	100%	99,80%	100%	100%	Multiple endocrine neoplasia, type IV, 610755
CDKN1C	86,30%	74,80%	99,20%	96,90%	IMAGE syndrome, 614732 Beckwith-Wiedemann syndrome, 130650
CDKN2A	92,30%	92,10%	100%	100%	No OMIM disease ID
CDON	100%	99,60%	100%	100%	Holoprosencephaly 11, 614226
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

CEBPA	98,60%	83,90%	99,30%	94,70%	Leukemia, acute myeloid, somatic, 601626 ?Leukemia, acute myeloid, 601626
CEBPE	100%	100%	100%	100%	Specific granule deficiency, 245480
CEL	89,70%	88,00%	100%	99,80%	Maturity-onset diabetes of the young, type VIII, 609812
CELA2A	98,30%	95,50%	100%	100%	Abdominal obesity-metabolic syndrome 4, 618620
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
CEP250	100%	99,20%	100%	100%	Cone-rod dystrophy and hearing loss 2, 618358
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
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
CES1	99,80%	99,30%	99,90%	99,90%	Drug metabolism, altered, CES1-related, 618057
CETP	100%	99,90%	100%	100%	Hyperalphalipoproteinemia, 143470
CFAP43	99,90%	98,90%	100%	100%	Hydrocephalus, normal pressure, 1, 236690 Spermatogenic failure 19, 617592

CFAP44	99,80%	98,90%	100%	100%	?Spermatogenic failure 20, 617593
CFAP53	99,60%	97,40%	100%	100%	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFAP69	98,70%	93,50%	100%	100%	Spermatogenic failure 24, 617959
CFB	100%	100%	100%	100%	?Complement factor B deficiency, 615561
CFC1	84,20%	74,10%	100%	100%	Heterotaxy, visceral, 2, autosomal, 605376
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
CFHR5	99,60%	98,40%	100%	100%	Nephropathy due to CFHR5 deficiency, 614809
CFI	99,20%	96,80%	100%	100%	Complement factor I deficiency, 610984
CFL2	100%	99,60%	100%	100%	Nemaline myopathy 7, autosomal recessive, 610687
CFP	100%	99,00%	100%	100%	Properdin deficiency, X-linked, 312060
CFTR	99,60%	97,90%	100%	100%	Cystic fibrosis, 219700 Congenital bilateral absence of vas deferens, 277180 Sweat chloride elevation without CF, 0
CHAMP1	100%	100%	100%	100%	Mental retardation, autosomal dominant 40, 616579
CHAT	93,50%	85,70%	100%	100%	Myasthenic syndrome, congenital, 6, presynaptic, 254210
CHCHD10	59,10%	43,90%	100%	100%	Spinal muscular atrophy, Jokela type, 615048 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 ?Myopathy, isolated mitochondrial, autosomal dominant, 616209
CHCHD2	98,40%	83,80%	100%	100%	Parkinson disease 22, autosomal dominant, 616710
CHD1	99,30%	94,90%	100%	100%	Pilarowski-Bjornsson syndrome, 617682
CHD2	99,40%	99,20%	100%	100%	Epileptic encephalopathy, childhood-onset, 615369
CHD3	94,80%	92,60%	99,80%	99,50%	Snijders Blok-Campeau syndrome, 618205
CHD4	100%	99,90%	100%	100%	Sifrim-Hitz-Weiss syndrome, 617159
CHD7	100%	99,50%	100%	100%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
CHEK2	85,00%	81,50%	100%	100%	Li-Fraumeni syndrome, 609265 Osteosarcoma, somatic, 259500
CHKB	100%	99,70%	100%	100%	Muscular dystrophy, congenital, megaconial type, 602541
CHM	98,50%	94,50%	98,80%	97,40%	Choroideremia, 303100
CHMP1A	100%	99,80%	100%	100%	Pontocerebellar hypoplasia, type 8, 614961
CHMP2B	99,70%	96,70%	100%	100%	Amyotrophic lateral sclerosis 17, 614696 Dementia, familial, nonspecific, 600795
CHMP4B	100%	99,30%	100%	100%	Cataract 31, multiple types, 605387
CHN1	99,60%	98,70%	100%	100%	Duane retraction syndrome 2, 604356
CHP1	98,50%	89,10%	100%	100%	?Spastic ataxia 9, autosomal recessive, 618438

CHRDL1	100%	99,80%	100%	100%	Megalocornea 1, X-linked, 309300
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
CHRNA2	100%	100%	100%	100%	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA4	98,30%	96,20%	100%	100%	Epilepsy, nocturnal frontal lobe, 1, 600513
CHRNB1	100%	99,40%	100%	100%	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314 Myasthenic syndrome, congenital, 2A, slow-channel, 616313
CHRNB2	99,30%	96,00%	100%	100%	Epilepsy, nocturnal frontal lobe, 3, 605375
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
CIB1	97,30%	93,60%	100%	100%	Epidermodysplasia verruciformis 3, 618267
CIB2	99,70%	97,00%	100%	99,90%	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CIC	63,30%	63,30%	100%	99,90%	Mental retardation, autosomal dominant 45, 617600
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
CITED2	99,20%	99,00%	100%	100%	Atrial septal defect 8, 614433 Ventricular septal defect 2, 614431
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
CLCN4	99,90%	98,90%	100%	100%	Raynaud-Claes syndrome, 300114
CLCN5	99,90%	98,30%	100%	100%	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990 Dent disease, 300009 Hypophosphatemic rickets, 300554 Nephrolithiasis, type I, 310468
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
CLCNKA	99,80%	97,80%	100%	100%	Bartter syndrome, type 4b, digenic, 613090
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
CLEC7A	100%	100%	100%	100%	Candidiasis, familial, 4, autosomal recessive, 613108
CLIC2	99,90%	96,50%	100%	100%	?Mental retardation, X-linked, syndromic 32, 300886
CLIC5	100%	99,90%	100%	100%	?Deafness, autosomal recessive 103, 616042
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
CLPX	99,90%	99,40%	100%	100%	?Protoporphyria, erythropoietic, 2, 618015
CLRN1	100%	99,80%	100%	100%	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902

CLTC	100%	99,90%	100%	100%	Mental retardation, autosomal dominant 56, 617854
CNBP	100%	100%	100%	100%	Myotonic dystrophy 2, 602668
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
CNKSR2	98,60%	93,60%	100%	100%	Mental retardation, X-linked, syndromic, Houge type, 301008
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
CNOT1	100%	99,90%	100%	100%	Holoprosencephaly 12, with or without pancreatic agenesis, 618500
CNOT2	99,90%	99,50%	100%	100%	Intellectual developmental disorder with nasal speech, dysmorphic facies, and variable skeletal anomalies, 618608
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

COL10A1	100%	98,40%	100%	100%	Metaphyseal chondrodysplasia, Schmid type, 156500
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
COL1A1	99,90%	98,60%	100%	100%	Osteogenesis imperfecta, type I, 166200 Osteogenesis imperfecta, type IV, 166220 Osteogenesis imperfecta, type II, 166210 Caffey disease, 114000 Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060 Osteogenesis imperfecta, type III, 259420
COL1A2	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
COL2A1	100%	99,70%	100%	100%	Achondrogenesis, type II or hypochondrogenesis, 200610 Spondyloperipheral dysplasia, 271700 Kniest dysplasia, 156550 Stickler syndrome, type I, 108300 Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 Osteoarthritis with mild chondrodysplasia, 604864 Platyspondylic skeletal dysplasia, Torrance type, 151210 Avascular necrosis of the femoral head, 608805

					SED congenita, 183900 Legg-Calve-Perthes disease, 150600 SMED Strudwick type, 184250 Czech dysplasia, 609162 Stickler sydrome, type I, nonsyndromic ocular, 609508 Spondyloepiphyseal dysplasia, Stanescu type, 616583 Vitreoretinopathy with phalangeal epiphyseal dysplasia, 0
COL3A1	99,60%	97,60%	100%	100%	Ehlers-Danlos syndrome, vascular type, 130050 Polymicrogyria with or without vascular-type EDS, 618343
COL4A1	98,70%	97,40%	100%	100%	?Retinal arteries, tortuosity of, 180000 Brain small vessel disease with or without ocular anomalies, 175780 Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564
COL4A2	100%	99,60%	100%	100%	Brain small vessel disease 2, 614483
COL4A3	98,70%	98,00%	100%	100%	Hematuria, benign familial, 141200 Alport syndrome 2, autosomal recessive, 203780 Alport syndrome 3, autosomal dominant, 104200
COL4A3BP	99,60%	96,50%	100%	100%	Mental retardation, autosomal dominant 34, 616351
COL4A4	99,90%	98,20%	100%	100%	Alport syndrome 2, autosomal recessive, 203780 Hematuria, familial benign, 141200
COL4A5	97,80%	89,00%	100%	99,80%	Alport syndrome 1, X-linked, 301050
COL4A6	97,50%	93,30%	100%	99,90%	?Deafness, X-linked 6, 300914
COL5A1	98,80%	98,00%	100%	99,90%	Ehlers-Danlos syndrome, classic type, 1, 130000
COL5A2	100%	99,50%	100%	100%	Ehlers-Danlos syndrome, classic type, 2, 130010
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
COL8A2	99,90%	97,90%	100%	100%	Corneal dystrophy, posterior polymorphous 2, 609140 Corneal dystrophy, Fuchs endothelial, 1, 136800
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
COL9A3	98,70%	95,50%	99,70%	98,60%	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969
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
COMP	93,40%	92,30%	100%	100%	Epiphyseal dysplasia, multiple, 1, 132400 Pseudoachondroplasia, 177170
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
CORIN	100%	99,90%	100%	100%	Preeclampsia/eclampsia 5, 614595
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
COX7B	77,40%	48,80%	100%	100%	Linear skin defects with multiple congenital anomalies 2, 300887

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
CPT1C	100%	99,90%	100%	100%	?Spastic paraplegia 73, autosomal dominant, 616282
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
CREB1	99,70%	96,70%	100%	100%	Histiocytoma, angiomatoid fibrous, somatic, 612160
CREB3L1	100%	99,90%	100%	100%	Osteogenesis imperfecta, type XVI, 616229
CREBBP	99,70%	98,50%	100%	100%	Rubinstein-Taybi syndrome 1, 180849 Menke-Hennekam syndrome 1, 618332
CRELD1	99,90%	95,00%	100%	100%	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217
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
CRTC1	99,80%	99,70%	100%	100%	Mucoepidermoid salivary gland carcinoma, 0
CRX	100%	100%	100%	100%	Cone-rod retinal dystrophy-2, 120970 Leber congenital amaurosis 7, 613829
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
CRYBA1	100%	99,40%	100%	100%	Cataract 10, multiple types, 600881
CRYBA2	100%	100%	100%	100%	?Cataract 42, 115900
CRYBA4	100%	100%	100%	100%	Cataract 23, 610425
CRYBB1	100%	100%	100%	100%	Cataract 17, multiple types, 611544
CRYBB2	100%	100%	100%	100%	Cataract 3, multiple types, 601547
CRYBB3	100%	100%	100%	100%	Cataract 22, 609741
CRYGB	100%	99,60%	100%	100%	Cataract 39, multiple types, autosomal dominant, 615188
CRYGC	99,80%	96,90%	100%	100%	Cataract 2, multiple types, 604307
CRYGD	100%	98,90%	100%	100%	Cataract 4, multiple types, 115700
CRYGS	94,10%	86,60%	100%	100%	Cataract 20, multiple types, 116100
CRYM	100%	99,60%	100%	100%	Deafness, autosomal dominant 40, 616357
CSF1R	99,90%	99,30%	100%	100%	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
CSF2RA	89,90%	87,50%	95,60%	92,10%	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF2RB	100%	99,00%	100%	100%	Surfactant metabolism dysfunction, pulmonary, 5, 614370
CSF3R	99,60%	98,20%	100%	100%	Neutropenia, severe congenital, 7, autosomal recessive, 617014
CSNK1D	97,70%	95,10%	100%	100%	Advanced sleep-phase syndrome, familial, 2, 615224
CSNK2A1	95,90%	92,20%	92,30%	92,30%	Okur-Chung neurodevelopmental syndrome, 617062
CSPP1	99,80%	98,70%	100%	100%	Joubert syndrome 21, 615636
CSRP3	100%	99,00%	100%	100%	Cardiomyopathy, hypertrophic, 12, 612124 ?Cardiomyopathy, dilated, 1M, 607482
CST3	93,40%	66,00%	100%	100%	Cerebral amyloid angiopathy, 105150
CST6	98,20%	92,50%	100%	100%	?Ectodermal dysplasia 15, hypohidrotic/hair type, 618535
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
CTBP1	93,20%	86,90%	99,50%	98,60%	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915
CTC1	100%	99,60%	100%	100%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTCF	100%	99,30%	100%	100%	Mental retardation, autosomal dominant 21, 615502
CTDP1	88,40%	84,30%	100%	99,40%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTH	100%	100%	100%	100%	Cystathioninuria, 219500 Homocysteine, total plasma, elevated, 0
CTHRC1	93,80%	87,50%	100%	100%	Barrett esophagus/esophageal adenocarcinoma, 614266
CTLA4	100%	100%	100%	100%	Autoimmune lymphoproliferative syndrome, type V, 616100

CTNNA1	99,30%	98,10%	100%	100%	Macular dystrophy, patterned, 2, 608970
CTNNA2	100%	99,80%	100%	100%	Cortical dysplasia, complex, with other brain malformations 9, 618174
CTNNA3	100%	99,80%	100%	100%	Arrhythmogenic right ventricular dysplasia, familial, 13, 615616
CTNNB1	100%	100%	100%	100%	Ovarian cancer, somatic, 167000 Colorectal cancer, somatic, 114500 Medulloblastoma, somatic, 155255 Hepatocellular carcinoma, somatic, 114550 Pilomatricoma, somatic, 132600 Neurodevelopmental disorder with spastic diplegia and visual defects, 615075 Exudative vitreoretinopathy 7, 617572
CTNND1	100%	100%	100%	100%	Blepharocheilodontic syndrome 2, 617681
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
CUL3	99,90%	98,80%	100%	100%	Pseudohypoaldosteronism, type IIE, 614496
CUL4B	98,00%	90,90%	99,90%	99,20%	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
CUL7	100%	99,30%	100%	100%	3-M syndrome 1, 273750
CUX1	96,40%	94,80%	99,30%	98,40%	Global developmental delay with or without impaired intellectual development, 618330
CUX2	99,90%	99,10%	100%	100%	Epileptic encephalopathy, early infantile, 67, 618141
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
CXCR4	100%	100%	100%	100%	WHIM syndrome, 193670 Myelokathexis, isolated, 0
CXorf56	99,80%	96,70%	100%	100%	?Mental retardation, X-linked 107, 301013
CYB561	92,80%	92,60%	100%	99,90%	Orthostatic hypotension 2, 618182
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
CYBB	99,90%	99,30%	100%	100%	Immunodeficiency 34, mycobacteriosis, X-linked, 300645 Chronic granulomatous disease, X-linked, 306400
CYC1	97,50%	89,20%	99,90%	98,70%	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYCS	99,10%	94,90%	100%	100%	Thrombocytopenia 4, 612004
CYFIP2	100%	99,40%	100%	100%	Epileptic encephalopathy, early infantile, 65, 618008
CYLD	99,80%	98,00%	100%	100%	Cylindromatosis, familial, 132700 Brooke-Spiegler syndrome, 605041 Trichoepithelioma, multiple familial, 1, 601606
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
CYP21A2	97,80%	88,40%	100%	100%	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910
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
CYP2A6	100%	99,90%	100%	100%	Coumarin resistance, 122700
CYP2B6	99,90%	98,20%	100%	100%	Efavirenz, poor metabolism of, 614546
CYP2C19	100%	98,90%	100%	100%	Clopidogrel, impaired responsiveness to, 609535 Mephenytoin poor metabolizer, 609535 Proguanil poor metabolizer, 609535 Omeprazole poor metabolizer, 609535
CYP2C9	99,90%	98,40%	100%	100%	Warfarin sensitivity, 122700 Tolbutamide poor metabolizer, 0

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
DAB1	100%	100%	100%	100%	Spinocerebellar ataxia 37, 615945
DACT1	93,60%	91,10%	100%	99,90%	?Townes-Brocks syndrome 2, 617466
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
DCAF8	100%	99,90%	100%	100%	?Giant axonal neuropathy 2, autosomal dominant, 610100
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
DCN	95,70%	95,60%	95,70%	95,70%	Corneal dystrophy, congenital stromal, 610048
DCPS	100%	100%	100%	100%	Al-Raqad syndrome, 616459
DCTN1	100%	98,80%	100%	100%	Perry syndrome, 168605 Neuronopathy, distal hereditary motor, type VIIB, 607641
DCX	100%	99,90%	100%	100%	Subcortical laminal heterotopia, X-linked, 300067 Lissencephaly, X-linked, 300067
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
DDX3X	86,80%	84,90%	100%	100%	Mental retardation, X-linked 102, 300958
DDX58	99,90%	99,00%	100%	100%	Singleton-Merten syndrome 2, 616298
DDX59	100%	100%	100%	100%	Orofaciodigital syndrome V, 174300
DDX6	97,70%	88,70%	100%	100%	Intellectual developmental disorder with impaired language and dysmorphic facies, 618653
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
DEPDC5	100%	99,80%	100%	100%	Epilepsy, familial focal, with variable foci 1, 604364
DES	100%	99,70%	100%	100%	?Cardiomyopathy, dilated, 1I, 604765 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 Myopathy, myofibrillar, 1, 601419
DFNA5	100%	99,20%	100%	100%	Deafness, autosomal dominant 5, 600994
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

DHX30	100%	99,90%	100%	100%	Neurodevelopmental disorder with severe motor impairment and absent language, 617804
DHX38	100%	99,30%	100%	100%	Retinitis pigmentosa 84, 618220
DIABLO	100%	99,80%	100%	100%	Deafness, autosomal dominant 64, 614152
DIAPH1	99,80%	99,00%	99,50%	97,90%	Seizures, cortical blindness, microcephaly syndrome, 616632 Deafness, autosomal dominant 1, 124900
DIAPH2	95,90%	87,60%	99,90%	99,00%	?Premature ovarian failure 2A, 300511
DIAPH3	99,60%	97,00%	100%	100%	Auditory neuropathy, autosomal dominant, 1, 609129
DICER1	99,80%	99,00%	100%	100%	GLOW syndrome, somatic mosaic, 618272 Rhabdomyosarcoma, embryonal, 2, 180295 Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800 Pleuropulmonary blastoma, 601200
DIP2B	100%	99,30%	100%	100%	Mental retardation, FRA12A type, 136630
DIS3L2	100%	99,80%	100%	100%	Perlman syndrome, 267000
DKC1	99,80%	98,70%	100%	99,70%	Dyskeratosis congenita, X-linked, 305000
DLAT	100%	99,70%	100%	100%	Pyruvate dehydrogenase E2 deficiency, 245348
DLC1	100%	99,90%	100%	100%	Colorectal cancer, somatic, 114500
DLD	100%	99,70%	100%	100%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLG3	99,10%	93,20%	100%	100%	Mental retardation, X-linked 90, 300850
DLL3	92,10%	87,00%	100%	99,10%	Spondylocostal dysostosis 1, autosomal recessive, 277300
DLL4	100%	99,20%	100%	100%	Adams-Oliver syndrome 6, 616589
DLST	96,70%	90,30%	100%	100%	Paragangliomas 7, 618475
DLX3	99,90%	98,40%	100%	100%	Trichodontoosseous syndrome, 190320 Amelogenesis imperfecta, type IV, 104510
DLX4	100%	100%	100%	100%	?Orofacial cleft 15, 616788
DLX5	100%	99,90%	100%	100%	?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600
DMD	99,60%	98,60%	100%	100%	Cardiomyopathy, dilated, 3B, 302045 Becker muscular dystrophy, 300376 Duchenne muscular dystrophy, 310200
DMGDH	100%	99,70%	100%	100%	Dimethylglycine dehydrogenase deficiency, 605850
DMP1	100%	99,90%	100%	100%	Hypophosphatemic rickets, AR, 241520
DMPK	99,80%	98,40%	100%	100%	Myotonic dystrophy 1, 160900
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
DNAH17	100%	99,60%	100%	99,90%	Spermatogenic failure 39, 618643
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
DNAJB11	100%	99,50%	100%	100%	Polycystic kidney disease 6 with or without polycystic liver disease, 618061
DNAJB13	100%	100%	100%	100%	Ciliary dyskinesia, primary, 34, 617091
DNAJB2	100%	100%	100%	100%	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881
DNAJB6	96,50%	88,50%	100%	100%	Muscular dystrophy, limb-girdle, autosomal dominant 1, 603511
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
DNAJC5	100%	100%	100%	100%	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350
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
DNAL4	100%	98,90%	100%	100%	?Mirror movements 3, 616059
DNASE1L3	100%	100%	100%	100%	Systemic lupus erythematosus 16, 614420
DNM1	92,60%	89,10%	97,40%	97,40%	Epileptic encephalopathy, early infantile, 31, 616346
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
DNMT1	99,20%	99,00%	99,70%	99,20%	Neuropathy, hereditary sensory, type IE, 614116 Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121

DNMT3A	99,80%	98,60%	100%	100%	Heyn-Sproul-Jackson syndrome, 618724 Acute myeloid leukemia, somatic, 601626 Tatton-Brown-Rahman syndrome, 615879
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
DPF2	99,90%	98,40%	100%	100%	Coffin-Siris syndrome 7, 618027
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
DPP6	99,70%	97,80%	99,40%	97,60%	Mental retardation, autosomal dominant 33, 616311
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
DRD4	93,80%	81,80%	100%	99,70%	Autonomic nervous system dysfunction, 0
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
DSG1	99,30%	97,50%	100%	100%	Keratosis palmoplantaris striata I, AD, 148700 Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508

DSG2	100%	99,60%	100%	100%	Arrhythmogenic right ventricular dysplasia 10, 610193 Cardiomyopathy, dilated, 1BB, 612877
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
DSPP	96,80%	86,10%	100%	100%	Dentin dysplasia, type II, 125420 Deafness, autosomal dominant 39, with dentinogenesis, 605594 Dentinogenesis imperfecta, Shields type II, 125490 Dentinogenesis imperfecta, Shields type III, 125500
DST	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
DTNA	100%	99,90%	100%	100%	Left ventricular noncompaction 1, with or without congenital heart defects, 604169
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
DUSP6	100%	100%	100%	100%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
DVL1	97,20%	95,00%	100%	100%	Robinow syndrome, autosomal dominant 2, 616331
DVL3	100%	100%	100%	100%	Robinow syndrome, autosomal dominant 3, 616894
DYM	97,40%	96,50%	100%	100%	Smith-McCort dysplasia, 607326 Dyggve-Melchior-Clausen disease, 223800
DYNC1H1	99,90%	99,40%	100%	100%	Mental retardation, autosomal dominant 13, 614563 Charcot-Marie-Tooth disease, axonal, type 20, 614228 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600
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
DYRK1A	100%	100%	100%	100%	Mental retardation, autosomal dominant 7, 614104
DYRK1B	98,40%	94,00%	100%	100%	Abdominal obesity-metabolic syndrome 3, 615812
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
EBF3	100%	100%	100%	100%	Hypotonia, ataxia, and delayed development syndrome, 617330
EBP	99,70%	95,80%	100%	100%	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
ECE1	98,30%	97,90%	100%	100%	?Hirschsprung disease, cardiac defects, and autonomic dysfunction, 613870
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
EDA	98,10%	91,60%	100%	99,90%	Tooth agenesis, selective, X-linked 1, 313500 Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100
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
EDNRA	100%	100%	100%	100%	Mandibulofacial dysostosis with alopecia, 616367
EDNRB	98,00%	93,80%	100%	100%	Waardenburg syndrome, type 4A, 277580 ABCD syndrome, 600501
EED	99,10%	95,90%	100%	100%	Cohen-Gibson syndrome, 617561
EEF1A2	100%	100%	99,90%	99,10%	Epileptic encephalopathy, early infantile, 33, 616409 Mental retardation, autosomal dominant 38, 616393
EEF2	100%	99,90%	100%	100%	?Spinocerebellar ataxia 26, 609306
EFEMP1	100%	99,90%	100%	100%	Doyne honeycomb degeneration of retina, 126600
EFEMP2	100%	100%	100%	100%	Cutis laxa, autosomal recessive, type IB, 614437
EFL1	99,60%	98,50%	100%	100%	Shwachman-Diamond syndrome 2, 617941
EFNB1	100%	100%	100%	100%	Craniofrontonasal dysplasia, 304110
EFTUD2	100%	99,80%	100%	100%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
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
EGLN1	89,30%	82,20%	100%	100%	Erythrocytosis, familial, 3, 609820

EGR2	100%	100%	100%	100%	Dejerine-Sottas disease, 145900 Hypomyelinating neuropathy, congenital, 1, 605253 Charcot-Marie-Tooth disease, type 1D, 607678
EHHADH	100%	100%	100%	100%	?Fanconi renotubular syndrome 3, 615605
EHMT1	94,50%	93,70%	99,60%	99,50%	Kleefstra syndrome 1, 610253
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
EIF2S3	95,40%	89,10%	100%	100%	MEHMO syndrome, 300148
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
ELANE	99,70%	97,40%	100%	100%	Neutropenia, severe congenital 1, autosomal dominant, 202700 Neutropenia, cyclic, 162800
ELMO2	99,90%	99,00%	100%	100%	Vascular malformation, primary intraosseous, 606893
ELMOD3	100%	99,80%	100%	100%	?Deafness, autosomal recessive 88, 615429
ELN	99,80%	97,80%	100%	100%	Cutis laxa, autosomal dominant, 123700 Supravalvar aortic stenosis, 185500
ELOVL1	99,80%	97,60%	100%	100%	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527
ELOVL4	100%	99,50%	100%	100%	Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
ELOVL5	100%	99,80%	100%	100%	Spinocerebellar ataxia 38, 615957
ELP1	99,80%	99,00%	100%	100%	Dysautonomia, familial, 223900
ELP2	99,90%	98,80%	100%	100%	Mental retardation, autosomal recessive 58, 617270
ELP4	72,80%	70,20%	87,10%	87,10%	?Aniridia 2, 617141
EMC1	100%	99,30%	100%	100%	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EMD	99,90%	98,40%	100%	99,10%	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
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
EMX2	100%	100%	100%	100%	Schizencephaly, 269160
ENAM	100%	100%	100%	100%	Amelogenesis imperfecta, type IC, 204650 Amelogenesis imperfecta, type IB, 104500
ENG	99,60%	96,00%	100%	100%	Telangiectasia, hereditary hemorrhagic, type 1, 187300
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
EP300	99,80%	99,00%	100%	100%	Rubinstein-Taybi syndrome 2, 613684 Menke-Hennekam syndrome 2, 618333 Colorectal cancer, somatic, 114500
EPAS1	99,70%	98,10%	100%	100%	Erythrocytosis, familial, 4, 611783
EPB41	99,40%	97,80%	100%	100%	Elliptocytosis-1, 611804
EPB41L1	99,60%	95,80%	100%	100%	?Mental retardation, autosomal dominant 11, 614257
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
EPHA2	100%	99,50%	100%	100%	Cataract 6, multiple types, 116600
EPHB2	98,10%	98,10%	99,80%	98,80%	?Bleeding disorder, platelet-type, 22, 618462
EPHB4	100%	99,70%	100%	100%	Capillary malformation-arteriovenous malformation 2, 618196 Lymphatic malformation 7, 617300
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
ERBB2	98,40%	97,10%	100%	100%	Glioblastoma, somatic, 137800 Adenocarcinoma of lung, somatic, 211980 Gastric cancer, somatic, 613659 Ovarian cancer, somatic, 0

ERBB3	100%	99,80%	100%	100%	?Lethal congenital contractural syndrome 2, 607598
ERBB4	100%	99,50%	100%	100%	Amyotrophic lateral sclerosis 19, 615515
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
ERF	99,90%	98,50%	100%	100%	Craniosynostosis 4, 600775 Chitayat syndrome, 617180
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
ERMARD	99,90%	99,00%	100%	100%	?Periventricular nodular heterotopia 6, 615544
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
ESR2	100%	99,70%	100%	100%	?Ovarian dysgenesis 8, 618187
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
ETV6	100%	99,90%	100%	100%	Leukemia, acute myeloid, somatic, 601626 Thrombocytopenia 5, 616216
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
EWSR1	92,30%	85,30%	100%	100%	Neuroepithelioma, 612219 Ewing sarcoma, 612219
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
EXT1	99,90%	98,40%	100%	100%	Exostoses, multiple, type 1, 133700 Chondrosarcoma, 215300
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
EYA1	99,90%	99,70%	100%	100%	?Otofaciocervical syndrome, 166780 Anterior segment anomalies with or without cataract, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650 Branchiootic syndrome 1, 602588
EYA4	100%	99,70%	100%	100%	Deafness, autosomal dominant 10, 601316 ?Cardiomyopathy, dilated, 1J, 605362
EYS	99,70%	98,20%	100%	100%	Retinitis pigmentosa 25, 602772
EZH2	100%	99,50%	100%	100%	Weaver syndrome, 277590
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
F8	99,50%	98,30%	100%	99,90%	Hemophilia A, 306700
F9	99,60%	97,60%	99,90%	98,80%	Thrombophilia, X-linked, due to factor IX defect, 300807 Hemophilia B, 306900
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
FAM111A	99,90%	99,30%	100%	100%	Gracile bone dysplasia, 602361 Kenny-Caffey syndrome, type 2, 127000
FAM111B	100%	99,90%	100%	100%	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704
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
FAM58A	83,10%	78,50%	98,90%	94,70%	STAR syndrome, 300707
FAM83H	99,00%	95,00%	100%	100%	Amelogenesis imperfecta, type IIIA, 130900
FAM92A	90,60%	86,00%	100%	100%	?Polydactyly, postaxial, type A9, 618219
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
FANCM	99,60%	97,30%	100%	100%	Spermatogenic failure 28, 618086 ?Premature ovarian failure 15, 618096

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
FAS	100%	99,60%	100%	100%	Autoimmune lymphoproliferative syndrome, type IA, 601859 Squamous cell carcinoma, burn scar-related, somatic, 0
FASLG	100%	99,60%	100%	100%	Autoimmune lymphoproliferative syndrome, type IB, 601859
FASTKD2	99,80%	98,90%	100%	100%	?Mitochondrial complex IV deficiency, 220110
FAT2	100%	99,80%	100%	100%	Spinocerebellar ataxia 45, 617769
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
FBN1	100%	99,90%	100%	100%	Marfan lipodystrophy syndrome, 616914 Marfan syndrome, 154700 MASS syndrome, 604308 Ectopia lentis, familial, 129600 Acromicric dysplasia, 102370 Weill-Marchesani syndrome 2, dominant, 608328 Geleophysic dysplasia 2, 614185 Stiff skin syndrome, 184900
FBN2	100%	99,90%	100%	100%	Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118
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
FBXO11	96,90%	92,70%	100%	100%	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities, 618089
FBXO31	96,00%	93,10%	100%	99,90%	?Mental retardation, autosomal recessive 45, 615979
FBXO38	99,90%	99,30%	100%	100%	Neuronopathy, distal hereditary motor, type IID, 615575
FBXO7	99,80%	97,90%	100%	100%	Parkinson disease 15, autosomal recessive, 260300
FCGR3A	99,00%	97,10%	100%	100%	Immunodeficiency 20, 615707
FCGR3B	99,40%	98,20%	98,10%	98,00%	Neutropenia, alloimmune neonatal, 0
FCN3	100%	100%	100%	100%	Immunodeficiency due to ficolin 3 deficiency, 613860
FDFT1	97,70%	96,00%	100%	100%	Squalene synthase deficiency, 618156
FDPS	99,10%	93,50%	100%	100%	Porokeratosis 9, multiple types, 616631
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
FGD1	97,30%	92,80%	100%	100%	Mental retardation, X-linked syndromic 16, 305400 Aarskog-Scott syndrome, 305400
FGD4	99,90%	99,40%	100%	100%	Charcot-Marie-Tooth disease, type 4H, 609311
FGF10	100%	99,80%	100%	100%	Aplasia of lacrimal and salivary glands, 180920 LADD syndrome, 149730
FGF12	99,90%	98,10%	100%	100%	Epileptic encephalopathy, early infantile, 47, 617166
FGF14	100%	100%	100%	100%	Spinocerebellar ataxia 27, 609307
FGF16	100%	99,80%	100%	100%	Metacarpal 4-5 fusion, 309630
FGF17	100%	100%	100%	100%	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
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
FGF5	100%	100%	100%	100%	Trichomegaly, 190330
FGF8	98,20%	88,90%	100%	99,60%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGF9	100%	100%	100%	100%	Multiple synostoses syndrome 3, 612961
FGFR1	100%	99,90%	100%	100%	Pfeiffer syndrome, 101600 Jackson-Weiss syndrome, 123150 Trigonocephaly 1, 190440 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 HEARTsfield syndrome, 615465 Osteoglophonic dysplasia, 166250 Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001
FGFR2	97,70%	97,10%	100%	100%	Apert syndrome, 101200 Jackson-Weiss syndrome, 123150 Saethre-Chotzen syndrome, 101400

					Gastric cancer, somatic, 613659 Scaphocephaly, maxillary retrusion, and mental retardation, 609579 Bent bone dysplasia syndrome, 614592 LADD syndrome, 149730 Craniofacial-skeletal-dermatologic dysplasia, 101600 Pfeiffer syndrome, 101600 Crouzon syndrome, 123500 Beare-Stevenson cutis gyrata syndrome, 123790 Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Craniosynostosis, nonspecific, 0 Scaphocephaly and Axenfeld-Rieger anomaly, 0
FGFR3	99,80%	97,70%	100%	99,80%	Muenke syndrome, 602849 Nevus, epidermal, somatic, 162900 Thanatophoric dysplasia, type II, 187601 Bladder cancer, somatic, 109800 CATSHL syndrome, 610474 Crouzon syndrome with acanthosis nigricans, 612247 Hypochondroplasia, 146000 LADD syndrome, 149730 Achondroplasia, 100800 Thanatophoric dysplasia, type I, 187600 Colorectal cancer, somatic, 114500 Spermatocytic seminoma, somatic, 273300 Cervical cancer, somatic, 603956 SADDAN, 616482
FGG	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
FHL1	99,70%	95,80%	100%	100%	Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717 Scapuloperoneal myopathy, X-linked dominant, 300695 Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718 ?Uruguay faciocardiomusculoskeletal syndrome, 300280 Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 Myopathy, X-linked, with postural muscle atrophy, 300696
FIBP	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
FIGLA	99,70%	96,00%	100%	100%	Premature ovarian failure 6, 612310
FMR 1	96,10%	92,10%	100%	100%	Premature ovarian failure 1, 311360 Fragile X tremor/ataxia syndrome, 300623 Fragile X syndrome, 300624
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
FLCN	100%	100%	100%	100%	Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700 Birt-Hogg-Dube syndrome, 135150 Colorectal cancer, somatic, 114500
FLG	100%	99,90%	100%	100%	Ichthyosis vulgaris, 146700
FLG2	100%	100%	99,90%	99,90%	Peeling skin syndrome 6, 618084
FLI1	99,50%	98,20%	100%	100%	Bleeding disorder, platelet-type, 21, 617443
FLNA	100%	99,90%	100%	100%	Otopalatodigital syndrome, type I, 311300 Congenital short bowel syndrome, 300048 Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Cardiac valvular dysplasia, X-linked, 314400 ?FG syndrome 2, 300321 Heterotopia, periventricular, 1, 300049 Terminal osseous dysplasia, 300244 Frontometaphyseal dysplasia 1, 305620

FLNB	99,50%	98,80%	100%	100%	Larsen syndrome, 150250 Atelosteogenesis, type I, 108720 Boomerang dysplasia, 112310 Spondylocarpotarsal synostosis syndrome, 272460 Atelosteogenesis, type III, 108721
FLNC	100%	99,60%	100%	100%	Myopathy, myofibrillar, 5, 609524 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Cardiomyopathy, familial hypertrophic, 26, 0
FLRT3	100%	100%	100%	100%	Hypogonadotropic hypogonadism 21 with anosmia, 615271
FLT3	99,90%	98,90%	100%	100%	Leukemia, acute myeloid, somatic, 601626 Leukemia, acute lymphoblastic, somatic, 613065 Leukemia, acute myeloid, reduced survival in, somatic, 601626
FLT4	99,20%	98,30%	100%	100%	Congenital heart defects, multiple types, 7, 618780 Hemangioma, capillary infantile, somatic, 602089 Lymphatic malformation 1, 153100
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
FN1	100%	99,30%	100%	100%	Glomerulopathy with fibronectin deposits 2, 601894 Spondylometaphyseal dysplasia, corner fracture type, 184255
FOLR1	100%	100%	100%	100%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXC1	98,00%	89,60%	99,90%	98,50%	Axenfeld-Rieger syndrome, type 3, 602482 Anterior segment dysgenesis 3, multiple subtypes, 601631
FOXC2	100%	96,70%	100%	99,80%	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
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
FOXF1	99,90%	98,80%	100%	100%	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380
FOXG1	88,60%	82,10%	99,20%	96,40%	Rett syndrome, congenital variant, 613454
FOXI1	100%	100%	100%	100%	Enlarged vestibular aqueduct, 600791
FOXL2	99,70%	95,50%	99,80%	98,00%	Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 Premature ovarian failure 3, 608996 Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100
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

FOXO1	99,50%	93,90%	99,20%	96,90%	Rhabdomyosarcoma, alveolar, 268220
FOXP1	100%	99,80%	100%	100%	Mental retardation with language impairment and with or without autistic features, 613670
FOXP2	99,50%	99,20%	100%	100%	Speech-language disorder-1, 602081
FOXP3	99,20%	95,50%	100%	100%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790
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
FRMD7	99,90%	99,10%	100%	99,60%	Nystagmus 1, congenital, X-linked, 310700 Nystagmus, infantile periodic alternating, X-linked, 310700
FRMPD4	99,80%	98,50%	100%	100%	Mental retardation, X-linked 104, 300983
FRRS1L	79,70%	69,10%	99,20%	95,80%	Epileptic encephalopathy, early infantile, 37, 616981
FSCN2	100%	100%	100%	100%	Retinitis pigmentosa 30, 607921
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
FSIP2	99,60%	98,20%	100%	100%	Spermatogenic failure 34, 618153
FTCD	95,70%	91,00%	100%	100%	Glutamate formiminotransferase deficiency, 229100
FTH1	94,00%	76,60%	100%	100%	?Hemochromatosis, type 5, 615517
FTL	98,50%	89,40%	100%	100%	Hyperferritinemia-cataract syndrome, 600886 Neurodegeneration with brain iron accumulation 3, 606159 L-ferritin deficiency, dominant and recessive, 615604
FTO	83,80%	83,70%	94,20%	94,20%	Growth retardation, developmental delay, facial dysmorphism, 612938
FTSJ1	98,00%	93,80%	100%	100%	Mental retardation, X-linked 9/44, 309549
FUCA1	100%	99,90%	100%	100%	Fucosidosis, 230000
FUK	97,70%	95,40%	100%	100%	Congenital disorder of glycosylation with defective fucosylation 2, 618324
FUS	99,20%	96,40%	100%	100%	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 Essential tremor, hereditary, 4, 614782
FUT6	100%	100%	100%	100%	Fucosyltransferase 6 deficiency, 613852
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

FXYD2	100%	100%	100%	100%	Hypomagnesemia 2, renal, 154020
FYB1	99,40%	97,00%	100%	100%	Thrombocytopenia 3, 273900
FYCO1	100%	99,90%	100%	100%	Cataract 18, autosomal recessive, 610019
FZD2	99,90%	98,20%	100%	100%	Omodysplasia 2, 164745
FZD4	100%	100%	100%	100%	Exudative vitreoretinopathy 1, 133780 Retinopathy of prematurity, 133780
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
G6PD	99,30%	98,10%	100%	99,30%	Hemolytic anemia, G6PD deficient (favism), 300908
GAA	100%	99,90%	100%	100%	Glycogen storage disease II, 232300
GAB1	100%	99,40%	100%	100%	?Deafness, autosomal recessive 26, 605428
GABBR2	96,20%	92,00%	99,10%	98,40%	Neurodevelopmental disorder with poor language and loss of hand skills, 617903 Epileptic encephalopathy, early infantile, 59, 617904
GABRA1	100%	100%	100%	100%	Epileptic encephalopathy, early infantile, 19, 615744
GABRA2	99,70%	98,60%	100%	100%	Epileptic encephalopathy, early infantile, 78, 618557
GABRA5	100%	99,90%	100%	100%	Epileptic encephalopathy, early infantile, 79, 618559
GABRB1	100%	100%	100%	100%	Epileptic encephalopathy, early infantile, 45, 617153
GABRB2	100%	99,90%	100%	100%	Epileptic encephalopathy, infantile or early childhood, 2, 617829
GABRB3	99,60%	98,20%	100%	100%	Epileptic encephalopathy, early infantile, 43, 617113
GABRG2	90,80%	90,20%	93,00%	93,00%	Epileptic encephalopathy, early infantile, 74, 618396 Febrile seizures, familial, 8, 607681 Epilepsy, generalized, with febrile seizures plus, type 3, 607681
GAD1	100%	99,90%	100%	100%	?Cerebral palsy, spastic quadriplegic, 1, 603513
GAL	100%	99,80%	100%	100%	?Epilepsy, familial temporal lobe, 8, 616461
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
GANAB	99,90%	99,00%	100%	100%	Polycystic kidney disease 3, 600666
GARS	99,90%	99,10%	100%	100%	Charcot-Marie-Tooth disease, type 2D, 601472 Neuronopathy, distal hereditary motor, type VA, 600794

GAS2L2	100%	100%	100%	100%	?Ciliary dyskinesia, primary, 41, 618449
GAS8	99,90%	99,30%	100%	100%	Ciliary dyskinesia, primary, 33, 616726
GATA1	99,80%	98,40%	100%	100%	Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835 Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367 Thrombocytopenia with beta-thalassemia, X-linked, 314050
GATA2	100%	98,30%	100%	100%	Emberger syndrome, 614038 Immunodeficiency 21, 614172
GATA3	100%	100%	100%	100%	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GATA4	84,10%	74,50%	100%	99,90%	?Testicular anomalies with or without congenital heart disease, 615542 Tetralogy of Fallot, 187500 Atrioventricular septal defect 4, 614430 Atrial septal defect 2, 607941 Ventricular septal defect 1, 614429
GATA5	99,70%	93,70%	100%	100%	Congenital heart defects, multiple types, 5, 617912
GATA6	89,80%	83,00%	99,60%	98,00%	Pancreatic agenesis and congenital heart defects, 600001 Atrial septal defect 9, 614475 Atrioventricular septal defect 5, 614474 Persistent truncus arteriosus, 217095 Tetralogy of Fallot, 187500
GATAD1	99,90%	97,90%	100%	99,10%	?Cardiomyopathy, dilated, 2B, 614672
GATAD2B	100%	100%	100%	100%	Mental retardation, autosomal dominant 18, 615074
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
GCM2	100%	100%	100%	100%	Hyperparathyroidism 4, 617343 Hypoparathyroidism, familial isolated, 146200
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
GDF2	100%	100%	100%	100%	Telangiectasia, hereditary hemorrhagic, type 5, 615506
GDF3	100%	100%	100%	100%	Microphthalmia, isolated 7, 613704 Microphthalmia with coloboma 6, 613703 Klippel-Feil syndrome 3, autosomal dominant, 613702
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
GDF6	100%	99,90%	100%	99,40%	Leber congenital amaurosis 17, 615360 Klippel-Feil syndrome 1, autosomal dominant, 118100 Multiple synostoses syndrome 4, 617898 Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094
GDF9	100%	100%	100%	100%	?Premature ovarian failure 14, 618014
GDI1	99,80%	98,70%	100%	100%	Mental retardation, X-linked 41, 300849
GDNF	100%	100%	100%	100%	Central hypoventilation syndrome, 209880
GEMIN4	100%	99,50%	100%	100%	Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities, 617913
GFAP	91,80%	89,70%	100%	100%	Alexander disease, 203450
GFER	99,60%	93,90%	100%	100%	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076

GFI1	100%	99,20%	100%	100%	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 Neutropenia, severe congenital 2, autosomal dominant, 613107
GFI1B	98,50%	96,70%	100%	100%	Bleeding disorder, platelet-type, 17, 187900
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
GJA3	100%	99,70%	100%	100%	Cataract 14, multiple types, 601885
GJA5	100%	100%	100%	100%	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic (GJA5/SCN5A), 108770
GJA8	100%	100%	100%	100%	Cataract 1, multiple types, 116200
GJB1	100%	100%	100%	100%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
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
GJB3	100%	100%	100%	100%	Deafness, autosomal dominant 2B, 612644 Deafness, digenic, GJB2/GJB3, 220290 Erythrokeratodermia variabilis et progressiva 1, 133200 Deafness, autosomal recessive, 0 Deafness, autosomal dominant, with peripheral neuropathy, 0
GJB4	100%	100%	100%	100%	Erythrokeratodermia variabilis et progressiva 2, 617524
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
GK	88,90%	70,40%	100%	99,90%	Glycerol kinase deficiency, 307030
GLA	99,80%	96,60%	100%	100%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
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
GLI1	100%	99,80%	100%	100%	Polydactyly, postaxial, type A8, 618123 Polydactyly, preaxial I, 174400
GLI2	99,10%	97,40%	100%	99,80%	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829
GLI3	100%	99,50%	100%	100%	Polydactyly, postaxial, types A1 and B, 174200 Greig cephalopolysyndactyly syndrome, 175700 Polydactyly, preaxial, type IV, 174700 Pallister-Hall syndrome, 146510
GLIS2	100%	99,80%	100%	100%	Nephronophthisis 7, 611498
GLIS3	100%	99,60%	100%	100%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GLMN	99,30%	94,70%	100%	100%	Glomuvenous malformations, 138000

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
GLUD1	94,20%	82,90%	100%	100%	Hyperinsulinism-hyperammonemia syndrome, 606762
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
GMNN	99,80%	97,40%	100%	100%	Meier-Gorlin syndrome 6, 616835
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
GNA11	99,90%	95,00%	100%	100%	Hypocalciuric hypercalcemia, type II, 145981 Hypocalcemia, autosomal dominant 2, 615361
GNAI2	100%	100%	100%	100%	Ventricular tachycardia, idiopathic, 192605 Pituitary adenoma, ACTH-secreting, somatic, 0
GNAI3	99,30%	95,20%	100%	100%	Auriculocondylar syndrome 1, 602483
GNAL	96,80%	93,40%	100%	100%	Dystonia 25, 615073
GNAO1	93,80%	93,80%	100%	100%	Epileptic encephalopathy, early infantile, 17, 615473 Neurodevelopmental disorder with involuntary movements, 617493
GNAQ	84,30%	74,90%	100%	100%	Sturge-Weber syndrome, somatic, mosaic, 185300 Capillary malformations, congenital, 1, somatic, mosaic, 163000
GNAS	100%	99,90%	100%	99,90%	ACTH-independent macronodular adrenal hyperplasia, 219080 Pseudohypoparathyroidism Ic, 612462 Pseudohypoparathyroidism Ib, 603233 Pseudopseudohypoparathyroidism, 612463 McCune-Albright syndrome, somatic, mosaic, 174800 Osseous heteroplasia, progressive, 166350 Pituitary adenoma 3, multiple types, somatic, 617686 Pseudohypoparathyroidism Ia, 103580
GNAS-AS1	NC	NC	NC	NC	Pseudohypoparathyroidism, type IB, 603233
GNAT1	100%	100%	100%	100%	Night blindness, congenital stationary, type 1G, 616389 Night blindness, congenital stationary, autosomal dominant 3, 610444
GNAT2	99,90%	99,00%	100%	100%	Achromatopsia 4, 613856

GNB1	100%	100%	100%	100%	Mental retardation, autosomal dominant 42, 616973 Leukemia, acute lymphoblastic, somatic, 613065
GNB3	100%	100%	100%	100%	Night blindness, congenital stationary, type 1H, 617024
GNB4	100%	100%	100%	100%	Charcot-Marie-Tooth disease, dominant intermediate F, 615185
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
GNRH1	100%	93,70%	100%	100%	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841
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
GOT1	100%	99,30%	100%	100%	Aspartate aminotransferase, serum level of, QTL1, 614419
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
GPC3	99,10%	94,70%	100%	100%	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPC4	100%	99,60%	100%	100%	Keipert syndrome, 301026
GPC6	100%	100%	100%	100%	Omodysplasia 1, 258315
GPD1	100%	100%	100%	100%	Hypertriglyceridemia, transient infantile, 614480
GPD1L	100%	99,80%	100%	100%	Brugada syndrome 2, 611777
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

GPR101	100%	100%	100%	100%	Pituitary adenoma 2, GH-secreting, 300943
GPR143	85,80%	76,40%	99,80%	97,90%	Ocular albinism, type I, Nettleship-Falls type, 300500 Nystagmus 6, congenital, X-linked, 300814
GPR179	100%	100%	100%	100%	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
GPR68	99,50%	96,70%	100%	100%	Amelogenesis imperfecta, hypomaturation type, IIA6, 617217
GPR88	99,40%	95,10%	98,80%	94,90%	?Chorea, childhood-onset, with psychomotor retardation, 616939
GPRASP2	100%	99,70%	100%	100%	?Deafness, X-linked 7, 301018
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
GREB1L	100%	99,90%	100%	100%	Renal hypodysplasia/aplasia 3, 617805
GREM2	100%	100%	100%	100%	Tooth agenesis, selective, 9, 617275
GRHL2	100%	100%	100%	100%	Deafness, autosomal dominant 28, 608641 Corneal dystrophy, posterior polymorphous, 4, 618031 Ectodermal dysplasia/short stature syndrome, 616029
GRHL3	100%	100%	100%	100%	Van der Woude syndrome 2, 606713
GRHPR	84,20%	81,30%	100%	99,30%	Hyperoxaluria, primary, type II, 260000
GRIA3	99,70%	96,10%	100%	99,60%	Mental retardation, X-linked 94, 300699
GRIA4	99,80%	99,00%	100%	100%	Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864
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
GRIN2A	100%	100%	100%	100%	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570
GRIN2B	99,80%	99,20%	100%	100%	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation, autosomal dominant 6, 613970
GRIN2D	79,80%	65,40%	93,90%	88,70%	Epileptic encephalopathy, early infantile, 46, 617162
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
GSN	95,80%	93,50%	99,90%	99,30%	Amyloidosis, Finnish type, 105120
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
GUCA1A	100%	100%	100%	100%	Cone-rod dystrophy 14, 602093 Cone dystrophy-3, 602093
GUCA1B	100%	100%	100%	100%	Retinitis pigmentosa 48, 613827
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
GULOP	NC	NC	NC	NC	Scurvy, 0
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
H19	NC	NC	NC	NC	Wilms tumor 2, 194071 Silver-Russell syndrome, 180860 Beckwith-Wiedemann syndrome, 130650
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
HBA1	100%	99,80%	100%	100%	Hemoglobin H disease, nondeletional, 613978 Thalassemias, alpha-, 604131 Erythrocytosis, 7, 617981 Methemoglobinemia, alpha type, 617973 Heinz body anemias, alpha-, 140700
HBA2	98,90%	93,10%	100%	100%	Thalassemia, alpha-, 604131 Erythrocytosis 7, 617981 Heinz body anemia, 140700 Hemoglobin H disease, deletional and nondeletional, 613978
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
HBD	100%	100%	100%	100%	Thalassemia, delta-, 0 Thalassemia due to Hb Lepore, 0
HBG1	97,70%	94,90%	98,40%	97,00%	Fetal hemoglobin quantitative trait locus 1, 141749
HBG2	100%	100%	87,70%	87,70%	Fetal hemoglobin quantitative trait locus 1, 141749 Cyanosis, transient neonatal, 613977
HCCS	99,80%	97,60%	100%	100%	Linear skin defects with multiple congenital anomalies 1, 309801
HCFC1	98,30%	93,60%	100%	100%	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541

HCN1	100%	99,70%	100%	100%	Generalized epilepsy with febrile seizures plus, type 10, 618482 Epileptic encephalopathy, early infantile, 24, 615871
HCN4	100%	99,30%	100%	99,90%	Brugada syndrome 8, 613123 Sick sinus syndrome 2, 163800
HCRT	89,80%	81,10%	100%	99,90%	?Narcolepsy 1, 161400
HDAC6	99,50%	97,40%	100%	100%	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863
HDAC8	100%	99,80%	100%	100%	Cornelia de Lange syndrome 5, 300882
HECW2	100%	99,10%	100%	100%	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268
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
HIST1H1E	100%	100%	100%	100%	Rahman syndrome, 617537
HIVEP2	100%	100%	100%	100%	Mental retardation, autosomal dominant 43, 616977
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
HMBS	99,90%	99,40%	100%	100%	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000
HMGB3	78,60%	67,20%	100%	100%	?Microphthalmia, syndromic 13, 300915
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
HNF1A	100%	99,80%	100%	100%	MODY, type III, 600496 Hepatic adenoma, somatic, 142330 Renal cell carcinoma, 144700 Diabetes mellitus, insulin-dependent, 20, 612520
HNF1B	99,30%	96,10%	100%	100%	Renal cysts and diabetes syndrome, 137920 Diabetes mellitus, noninsulin-dependent, 125853
HNF4A	99,90%	99,00%	100%	100%	MODY, type I, 125850 Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026
HNMT	100%	99,80%	100%	100%	Mental retardation, autosomal recessive 51, 616739
HNRNPA1	98,80%	90,00%	100%	100%	Amyotrophic lateral sclerosis 20, 615426 ?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3, 615424
HNRNPA2B1	99,90%	99,40%	100%	100%	?Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2, 615422
HNRNPDL	97,30%	88,40%	100%	100%	Muscular dystrophy, limb-girdle, autosomal dominant 3, 609115
HNRNPH2	100%	100%	100%	100%	Mental retardation, X-linked, syndromic, Bain type, 300986
HNRNPK	91,50%	82,80%	100%	100%	Au-Kline syndrome, 616580
HNRNPU	99,90%	98,90%	100%	100%	Epileptic encephalopathy, early infantile, 54, 617391
HOGA1	100%	96,40%	100%	100%	Hyperoxaluria, primary, type III, 613616
HOMER2	99,50%	99,40%	100%	100%	?Deafness, autosomal dominant 68, 616707
HOXA1	100%	100%	100%	100%	Athabaskan brainstem dysgenesis syndrome, 601536 Bosley-Salih-Alorainy syndrome, 601536
HOXA11	97,10%	87,50%	100%	100%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
HOXA13	77,70%	69,00%	89,70%	79,70%	?Guttmacher syndrome, 176305 Hand-foot-uterus syndrome, 140000
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

HOXD10	100%	100%	100%	100%	Charcot-Marie-Tooth disease, foot deformity of, 192950 Vertical talus, congenital, 192950
HOXD13	99,90%	98,60%	100%	100%	Brachydactyly, type D, 113200 Brachydactyly, type E, 113300 ?Brachydactyly-syndactyly syndrome, 610713 Syndactyly, type V, 186300 Synpolydactyly 1, 186000
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
HPRT1	99,30%	91,80%	100%	99,30%	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322
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
HRAS	100%	100%	100%	100%	Nevus sebaceous or woolly hair nevus, somatic, 162900 Congenital myopathy with excess of muscle spindles, 218040 Bladder cancer, somatic, 109800 Thyroid carcinoma, follicular, somatic, 188470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Spitz nevus or nevus spilus, somatic, 137550 Costello syndrome, 218040
HRG	95,00%	94,20%	100%	100%	Thrombophilia due to HRG deficiency, 613116
HS6ST2	97,60%	96,60%	100%	100%	?Paganini-Miozzo syndrome, 301025
HSD11B1	100%	99,60%	100%	100%	Cortisone reductase deficiency 2, 614662
HSD11B2	86,00%	82,70%	99,90%	98,10%	Apparent mineralocorticoid excess, 218030
HSD17B10	100%	99,10%	100%	100%	HSD10 mitochondrial disease, 300438
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
HSF4	99,60%	97,20%	100%	100%	Cataract 5, multiple types, 116800
HSPA9	88,50%	84,50%	100%	100%	Even-plus syndrome, 616854 Anemia, sideroblastic, 4, 182170
HSPB1	98,80%	91,60%	100%	100%	Neuronopathy, distal hereditary motor, type IIB, 608634 Charcot-Marie-Tooth disease, axonal, type 2F, 606595
HSPB3	100%	100%	100%	100%	?Neuronopathy, distal hereditary motor, type IIC, 613376
HSPB8	100%	100%	100%	100%	Neuronopathy, distal hereditary motor, type IIA, 158590 Charcot-Marie-Tooth disease, axonal, type 2L, 608673
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
HTR1A	100%	100%	100%	100%	Periodic fever, menstrual cycle dependent, 614674
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
HTT	97,80%	96,60%	100%	99,90%	Huntington disease, 143100 Lopes-Maciel-Rodan syndrome, 617435
HUWE1	99,20%	95,80%	100%	100%	Mental retardation, X-linked syndromic, Turner type, 309590
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
HYOU1	100%	99,50%	100%	100%	?Immunodeficiency 59 and hypoglycemia, 233600
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
IDH2	99,70%	97,40%	100%	99,80%	D-2-hydroxyglutaric aciduria 2, 613657
IDH3B	95,40%	95,40%	100%	100%	Retinitis pigmentosa 46, 612572

IDS	99,90%	98,00%	100%	100%	Mucopolysaccharidosis II, 309900
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
IFIH1	99,70%	98,40%	100%	100%	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFITM5	99,30%	95,60%	100%	100%	Osteogenesis imperfecta, type V, 610967
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
IGBP1	99,50%	96,20%	100%	100%	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472
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
IGF2	100%	100%	100%	100%	?Growth restriction, severe, with distinctive facies, 616489
IGF2R	98,90%	97,30%	99,90%	99,70%	Hepatocellular carcinoma, somatic, 114550
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
IGHG2	68,80%	49,60%	100%	100%	IgG2 deficiency, selective, 0
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
IGSF1	99,50%	96,30%	100%	100%	Hypothyroidism, central, and testicular enlargement, 300888
IGSF3	95,40%	94,00%	100%	100%	?Lacrimal duct defect, 149700
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
IKBKG	84,10%	77,20%	100%	100%	Immunodeficiency 33, 300636 Incontinentia pigmenti, 308300 Immunodeficiency, isolated, 300584 Ectodermal dysplasia and immunodeficiency 1, 300291 Invasive pneumococcal disease, recurrent isolated, 2, 300640 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301
IKZF1	100%	100%	100%	100%	Immunodeficiency, common variable, 13, 616873
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
IL17F	99,90%	97,20%	100%	100%	?Candidiasis, familial, 6, autosomal dominant, 613956
IL17RA	100%	99,40%	100%	100%	Immunodeficiency 51, 613953
IL17RC	100%	99,90%	100%	100%	Candidiasis, familial, 9, 616445
IL17RD	99,90%	99,10%	100%	100%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
IL1RAPL1	99,80%	98,60%	100%	100%	Mental retardation, X-linked 21/34, 300143
IL1RN	100%	100%	100%	100%	Interleukin 1 receptor antagonist deficiency, 612852
IL21	99,40%	95,70%	100%	100%	?Immunodeficiency, common variable, 11, 615767
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
IL2RG	99,80%	97,10%	100%	100%	Severe combined immunodeficiency, X-linked, 300400 Combined immunodeficiency, X-linked, moderate, 312863
IL31RA	99,90%	99,90%	100%	100%	?Amyloidosis, primary localized cutaneous, 2, 613955
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
IMPDH1	87,90%	80,20%	100%	100%	Leber congenital amaurosis 11, 613837 Retinitis pigmentosa 10, 180105
IMPG1	99,70%	98,50%	100%	100%	Macular dystrophy, vitelliform, 4, 616151
IMPG2	99,80%	98,40%	100%	100%	Macular dystrophy, vitelliform, 5, 616152 Retinitis pigmentosa 56, 613581
INF2	86,70%	83,80%	100%	100%	Glomerulosclerosis, focal segmental, 5, 613237 Charcot-Marie-Tooth disease, dominant intermediate E, 614455
ING1	100%	99,90%	100%	100%	Squamous cell carcinoma, head and neck, somatic, 275355
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
INS	99,90%	97,90%	100%	100%	Maturity-onset diabetes of the young, type 10, 613370 Hyperproinsulinemia, 616214 Diabetes mellitus, insulin-dependent, 2, 125852 Diabetes mellitus, permanent neonatal, 606176
INSL3	80,60%	78,30%	80,70%	80,70%	Cryptorchidism, 219050
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
MRAP	100%	100%	100%	100%	Glucocorticoid deficiency 2, 607398
MRAS	100%	99,60%	100%	100%	Noonan syndrome 11, 618499
MRE11	98,90%	93,30%	100%	100%	Ataxia-telangiectasia-like disorder 1, 604391
MRM2	100%	99,50%	99,00%	99,00%	?Mitochondrial DNA depletion syndrome 17, 618567
MRPL3	93,20%	87,20%	100%	100%	Combined oxidative phosphorylation deficiency 9, 614582
MRPL44	99,90%	98,70%	100%	100%	?Combined oxidative phosphorylation deficiency 16, 615395
MRPS14	100%	100%	100%	100%	?Combined oxidative phosphorylation deficiency 38, 618378

MRPS16	100%	99,60%	100%	100%	Combined oxidative phosphorylation deficiency 2, 610498
MRPS2	99,60%	96,90%	100%	100%	Combined oxidative phosphorylation deficiency 36, 617950
MRPS22	99,80%	99,10%	100%	100%	Combined oxidative phosphorylation deficiency 5, 611719 Ovarian dysgenesis 7, 618117
MRPS34	97,60%	92,00%	100%	100%	Combined oxidative phosphorylation deficiency 32, 617664
MRPS7	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
IQCE	99,90%	98,80%	100%	100%	?Polydactyly, postaxial, type A7, 617642
IQSEC2	96,80%	88,60%	99,40%	98,40%	Mental retardation, X-linked 1/78, 309530
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
IRF1	100%	100%	100%	100%	Gastric cancer, somatic, 613659 Nonsmall cell lung cancer, somatic, 211980 Myelogenous leukemia, acute, 0 Myelodysplastic syndrome, preleukemic, 0
IRF2BP2	93,90%	77,70%	100%	99,90%	?Immunodeficiency, common variable, 14, 617765
IRF2BPL	99,50%	95,00%	99,90%	99,20%	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088
IRF6	99,60%	95,90%	100%	100%	Popliteal pterygium syndrome 1, 119500 van der Woude syndrome, 119300
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
IRS4	100%	100%	100%	100%	Hypothyroidism, congenital, nongoitrous, 9, 301035
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
ITM2B	100%	99,80%	100%	100%	Dementia, familial British, 176500 ?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079 Dementia, familial Danish, 117300
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
ITPR2	99,90%	99,30%	100%	100%	?Anhidrosis, isolated, with normal sweat glands, 106190
IVD	100%	100%	100%	100%	Isovaleric acidemia, 243500
IYD	99,50%	95,70%	100%	100%	Thyroid dyshormonogenesis 4, 274800
JAG1	97,70%	96,80%	100%	100%	Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500 ?Deafness, congenital heart defects, and posterior embryotoxon, 617992
JAGN1	100%	100%	100%	100%	Neutropenia, severe congenital, 6, autosomal recessive, 616022
JAK2	98,10%	95,80%	100%	100%	Myelofibrosis, somatic, 254450 Thrombocythemia 3, 614521 Polycythemia vera, somatic, 263300

					Leukemia, acute myeloid, somatic, 601626 Erythrocytosis, somatic, 133100
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
JPH2	95,50%	80,30%	100%	100%	Cardiomyopathy, hypertrophic, 17, 613873
JPH3	100%	99,80%	100%	100%	Huntington disease-like 2, 606438
JUP	100%	99,50%	100%	100%	Arrhythmogenic right ventricular dysplasia 12, 611528 Naxos disease, 601214
KANK1	100%	100%	100%	100%	Cerebral palsy, spastic quadriplegic, 2, 612900
KANK2	100%	100%	100%	100%	Nephrotic syndrome, type 16, 617783 Palmoplantar keratoderma and woolly hair, 616099
KANSL1	99,90%	99,20%	100%	100%	Koolen-De Vries syndrome, 610443
KARS	100%	99,90%	100%	100%	Deafness, autosomal recessive 89, 613916 ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641
KAT6A	100%	99,80%	100%	100%	Arboleda-Tham syndrome, 616268
KAT6B	99,90%	99,00%	100%	100%	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KATNB1	100%	99,90%	100%	100%	Lissencephaly 6, with microcephaly, 616212
KBTBD13	99,80%	95,80%	100%	100%	Nemaline myopathy 6, autosomal dominant, 609273
KCNA1	100%	99,90%	100%	100%	Episodic ataxia/myokymia syndrome, 160120
KCNA2	100%	99,60%	100%	100%	Epileptic encephalopathy, early infantile, 32, 616366
KCNA4	100%	100%	100%	100%	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284
KCNA5	100%	98,50%	100%	100%	Atrial fibrillation, familial, 7, 612240
KCNB1	100%	99,60%	100%	100%	Epileptic encephalopathy, early infantile, 26, 616056
KCNC1	100%	100%	100%	100%	Epilepsy, progressive myoclonic 7, 616187
KCNC3	81,10%	69,40%	94,70%	89,00%	Spinocerebellar ataxia 13, 605259
KCND3	100%	99,40%	100%	100%	Brugada syndrome 9, 616399 Spinocerebellar ataxia 19, 607346
KCNE1	100%	100%	100%	100%	Long QT syndrome 5, 613695 Jervell and Lange-Nielsen syndrome 2, 612347
KCNE2	100%	97,20%	100%	100%	Atrial fibrillation, familial, 4, 611493 Long QT syndrome 6, 613693
KCNE3	100%	100%	100%	100%	?Brugada syndrome 6, 613119
KCNH1	98,70%	98,70%	98,70%	98,70%	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500

KCNH2	95,80%	91,90%	100%	100%	Long QT syndrome 2, 613688 Short QT syndrome 1, 609620
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
KCNJ2	100%	100%	100%	100%	Short QT syndrome 3, 609622 Atrial fibrillation, familial, 9, 613980 Andersen syndrome, 170390
KCNJ5	100%	100%	100%	100%	Long QT syndrome 13, 613485 Hyperaldosteronism, familial, type III, 613677
KCNJ6	100%	100%	100%	100%	Keppen-Lubinsky syndrome, 614098
KCNK3	97,50%	95,00%	100%	100%	Pulmonary hypertension, primary, 4, 615344
KCNK4	99,10%	97,40%	100%	100%	Facial dysmorphism, hypertrichosis, epilepsy, intellectual/developmental delay, and gingival overgrowth syndrome, 618381
KCNK9	100%	100%	100%	100%	Birk-Barel mental retardation dysmorphism syndrome, 612292
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
KCNN3	100%	99,70%	100%	100%	Zimmermann-Laband syndrome 3, 618658
KCNN4	100%	99,40%	100%	100%	Dehydrated hereditary stomatocytosis 2, 616689
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
KCNQ1OT1	NC	NC	NC	NC	Beckwith-Wiedemann syndrome, 130650
KCNQ2	91,30%	89,80%	100%	100%	Epileptic encephalopathy, early infantile, 7, 613720 Seizures, benign neonatal, 1, 121200 Myokymia, 121200
KCNQ3	100%	99,40%	99,80%	99,10%	Seizures, benign neonatal, 2, 121201
KCNQ4	97,00%	95,70%	96,40%	93,90%	Deafness, autosomal dominant 2A, 600101
KCNQ5	97,80%	95,50%	100%	100%	Mental retardation, autosomal dominant 46, 617601

KCNT1	96,00%	95,20%	98,60%	97,30%	Epilepsy, nocturnal frontal lobe, 5, 615005 Epileptic encephalopathy, early infantile, 14, 614959
KCNT2	99,40%	97,10%	100%	100%	?Epileptic encephalopathy, early infantile, 57, 617771
KCNV2	100%	99,90%	100%	100%	Retinal cone dystrophy 3B, 610356
KCTD1	100%	99,70%	100%	100%	Scalp-ear-nipple syndrome, 181270
KCTD17	100%	99,00%	100%	100%	Dystonia 26, myoclonic, 616398
KCTD7	95,00%	95,00%	100%	100%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDF1	100%	99,80%	100%	100%	?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/nail type, 617337
KDM1A	98,20%	95,20%	100%	100%	Cleft palate, psychomotor retardation, and distinctive facial features, 616728
KDM5B	99,50%	97,90%	97,70%	97,70%	Mental retardation, autosomal recessive 65, 618109
KDM5C	99,80%	97,90%	100%	100%	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
KDM6A	96,10%	88,70%	100%	99,90%	Kabuki syndrome 2, 300867
KDM6B	98,80%	97,90%	100%	100%	Neurodevelopmental disorder with coarse facies and mild distal skeletal abnormalities, 618505
KDR	100%	99,80%	100%	100%	Hemangioma, capillary infantile, somatic, 602089
KDSR	100%	99,50%	100%	100%	Erythrokeratodermia variabilis et progressiva 4, 617526
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
KIAA0825	99,40%	97,70%	100%	100%	Polydactyly, postaxial, type A10, 618498
KIAA1109	99,80%	99,20%	100%	100%	Alkuraya-Kucinskas syndrome, 617822
KIAA1161	100%	100%	100%	100%	Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317
KIAA1549	97,90%	96,40%	98,80%	98,00%	Retinitis pigmentosa 86, 618613
KIDINS220	100%	100%	100%	100%	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296
KIF11	97,60%	94,80%	100%	100%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
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
KIF1B	100%	99,60%	100%	100%	Pheochromocytoma, 171300 ?Charcot-Marie-Tooth disease, type 2A1, 118210
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

KIF21A	99,90%	99,30%	100%	100%	Fibrosis of extraocular muscles, congenital, 1, 135700 Fibrosis of extraocular muscles, congenital, 3B, 135700
KIF22	100%	100%	100%	100%	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546
KIF2A	99,60%	95,60%	100%	100%	Cortical dysplasia, complex, with other brain malformations 3, 615411
KIF4A	99,40%	95,70%	100%	100%	?Mental retardation, X-linked 100, 300923
KIF5A	100%	99,90%	100%	100%	Myoclonus, intractable, neonatal, 617235 Spastic paraplegia 10, autosomal dominant, 604187
KIF5C	99,90%	98,80%	99,80%	99,80%	Cortical dysplasia, complex, with other brain malformations 2, 615282
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
KISS1	100%	98,30%	100%	100%	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842
KISS1R	100%	99,50%	100%	100%	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 ?Precocious puberty, central, 1, 176400
KIT	100%	99,60%	100%	100%	Gastrointestinal stromal tumor, familial, 606764 Mastocytosis, cutaneous, 154800 Germ cell tumors, somatic, 273300 Leukemia, acute myeloid, somatic, 601626 Mastocytosis, systemic, somatic, 154800 Piebaldism, 172800
KITLG	100%	98,50%	100%	100%	Hyperpigmentation with or without hypopigmentation, 145250 Deafness, autosomal dominant 69, unilateral or asymmetric, 616697
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
KLF1	100%	97,80%	100%	100%	Blood group--Lutheran inhibitor, 111150 Dyserythropoietic anemia, congenital, type IV, 613673
KLF11	100%	99,40%	100%	100%	Maturity-onset diabetes of the young, type VII, 610508
KLF6	100%	100%	100%	100%	Gastric cancer, somatic, 613659 Prostate cancer, somatic, 176807
KLHL10	100%	100%	100%	100%	Spermatogenic failure 11, 615081
KLHL15	100%	99,70%	100%	100%	Mental retardation, X-linked 103, 300982
KLHL24	100%	100%	100%	100%	Epidermolysis bullosa simplex, generalized, with scarring and hair loss, 617294
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
KLLN	100%	100%	100%	100%	Cowden syndrome 4, 615107
KMT2A	100%	99,90%	99,90%	99,40%	Wiedemann-Steiner syndrome, 605130
KMT2B	95,80%	94,00%	98,70%	97,90%	Dystonia 28, childhood-onset, 617284
KMT2C	92,20%	91,00%	100%	100%	Kleefstra syndrome 2, 617768
KMT2D	100%	99,40%	100%	100%	Kabuki syndrome 1, 147920
KMT2E	99,80%	98,50%	100%	100%	O'Donnell-Luria-Rodan syndrome, 618512
KMT5B	99,90%	99,10%	100%	100%	Mental retardation, autosomal dominant 51, 617788
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
KRAS	99,50%	96,90%	100%	100%	Oculoectodermal syndrome, somatic, 600268 Leukemia, acute myeloid, somatic, 601626 Breast cancer, somatic, 114480 RAS-associated autoimmune leukoproliferative disorder, 614470 Cardiofaciocutaneous syndrome 2, 615278 Arteriovenous malformation of the brain, somatic, 108010 Bladder cancer, somatic, 109800 Pancreatic carcinoma, somatic, 260350 Lung cancer, somatic, 211980 Gastric cancer, somatic, 137215 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Noonan syndrome 3, 609942
KREMEN1	97,70%	94,40%	99,50%	97,90%	Ectodermal dysplasia 13, hair/tooth type, 617392
KRIT1	100%	99,30%	100%	100%	Cavernous malformations of CNS and retina, 116860 Cerebral cavernous malformations-1, 116860 Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations, 116860
KRT1	98,70%	95,60%	100%	100%	Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602 Keratosis palmoplantaris striata III, 607654 Palmoplantar keratoderma, epidermolytic, 144200 Palmoplantar keratoderma, nonepidermolytic, 600962 Ichthyosis histrix, Curth-Macklin type, 146590 Epidermolytic hyperkeratosis, 113800

KRT10	100%	99,30%	100%	100%	Epidermolytic hyperkeratosis, 113800 Ichthyosis with confetti, 609165 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602
KRT12	99,70%	97,80%	100%	100%	Meesmann corneal dystrophy 1, 122100
KRT13	100%	100%	100%	100%	White sponge nevus 2, 615785
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
KRT16	74,20%	56,50%	100%	100%	Palmoplantar keratoderma, nonepidermolytic, focal, 613000 Pachyonychia congenita 1, 167200
KRT17	39,80%	22,80%	100%	100%	Pachyonychia congenita 2, 167210 Steatocystoma multiplex, 184500
KRT18	86,70%	70,90%	100%	100%	Cirrhosis, cryptogenic, 215600
KRT2	100%	99,80%	100%	100%	Ichthyosis bullosa of Siemens, 146800
KRT25	100%	100%	100%	100%	Woolly hair, autosomal recessive 3, 616760
KRT3	100%	100%	100%	100%	Meesmann corneal dystrophy 2, 618767
KRT4	100%	99,70%	100%	100%	White sponge nevus 1, 193900
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
KRT6A	92,30%	87,70%	100%	100%	Pachyonychia congenita 3, 615726
KRT6B	93,60%	88,60%	100%	100%	Pachyonychia congenita 4, 615728
KRT6C	88,30%	81,30%	99,90%	99,80%	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735
KRT71	100%	100%	100%	100%	?Hypotrichosis 13, 615896
KRT74	100%	100%	100%	100%	?Ectodermal dysplasia 7, hair/nail type, 614929 Woolly hair, autosomal dominant, 194300 ?Hypotrichosis 3, 613981
KRT8	90,60%	69,60%	100%	100%	Cirrhosis, cryptogenic, 215600
KRT81	99,20%	94,20%	100%	100%	Monilethrix, 158000
KRT83	96,60%	84,40%	100%	100%	Erythrokeratodermia variabilis et progressiva 5, 617756 Monilethrix, 158000

KRT85	99,00%	93,60%	100%	100%	Ectodermal dysplasia 4, hair/nail type, 602032
KRT86	99,70%	96,30%	100%	100%	Monilethrix, 158000
KRT9	99,20%	95,00%	100%	100%	Palmoplantar keratoderma, epidermolytic, 144200
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
L1CAM	99,90%	99,10%	100%	100%	MASA syndrome, 303350 Hydrocephalus with Hirschsprung disease, 307000 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 Corpus callosum, partial agenesis of, 304100 CRASH syndrome, 303350 Hydrocephalus due to aqueductal stenosis, 307000
L2HGDH	99,00%	97,20%	100%	100%	L-2-hydroxyglutaric aciduria, 236792
LAGE3	95,90%	85,10%	100%	100%	Galloway-Mowat syndrome 2, X-linked, 301006
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
LAMA4	100%	99,90%	100%	100%	Cardiomyopathy, dilated, 1JJ, 615235
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
LAMP2	99,20%	95,60%	100%	100%	Danon disease, 300257
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

LAS1L	99,70%	97,30%	100%	100%	Wilson-Turner syndrome, 309585
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
LDB3	95,40%	94,70%	100%	100%	Cardiomyopathy, hypertrophic, 24, 601493 Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 Myopathy, myofibrillar, 4, 609452 Left ventricular noncompaction 3, 601493
LDHA	95,00%	91,70%	100%	100%	Glycogen storage disease XI, 612933
LDHD	100%	99,50%	100%	100%	D-lactic aciduria, 245450
LDLR	100%	99,30%	100%	100%	Hypercholesterolemia, familial, 1, 143890 LDL cholesterol level QTL2, 143890
LDLRAP1	98,80%	94,20%	100%	100%	Hypercholesterolemia, familial, 4, 603813
LEF1	100%	100%	100%	100%	Sebaceous tumors, somatic, 0
LEMD2	98,70%	92,00%	100%	100%	Cataract 46, juvenile-onset, 212500
LEMD3	99,90%	98,70%	100%	100%	Osteopoikilosis with or without melorheostosis, 166700 Buschke-Ollendorff syndrome, 166700
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
LGI1	98,50%	97,50%	100%	100%	Epilepsy, familial temporal lobe, 1, 600512
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
LHX4	100%	100%	100%	100%	Pituitary hormone deficiency, combined, 4, 262700

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
LITAF	98,20%	92,70%	100%	100%	Charcot-Marie-Tooth disease, type 1C, 601098
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
LMNB1	99,90%	98,90%	100%	100%	Leukodystrophy, adult-onset, autosomal dominant, 169500
LMNB2	97,50%	94,30%	98,20%	96,90%	?Epilepsy, progressive myoclonic, 9, 616540
LMOD3	100%	99,70%	100%	100%	Nemaline myopathy 10, 616165
LMX1B	99,60%	96,30%	100%	100%	Nail-patella syndrome, 161200
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
LOR	99,00%	80,80%	100%	100%	Vohwinkel syndrome with ichthyosis, 604117
LOX	100%	99,60%	100%	100%	Aortic aneurysm, familial thoracic 10, 617168
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
LPP	100%	100%	100%	100%	Leukemia, acute myeloid, 601626 Lipoma, 0
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
LRP12	100%	99,80%	100%	100%	Oculopharyngodistal myopathy 1, 164310
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
LRP6	100%	99,90%	100%	100%	Tooth agenesis, selective, 7, 616724
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
LRRC8A	100%	99,80%	100%	100%	?Agammaglobulinemia 5, 613506
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
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
LYZ	100%	100%	100%	100%	Amyloidosis, renal, 105200
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
LZTS1	100%	99,80%	100%	100%	Esophageal squamous cell carcinoma, somatic, 133239
MAB21L1	100%	100%	100%	100%	Cerebellar, ocular, craniofacial, and genital syndrome, 618479
MAB21L2	100%	100%	100%	100%	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877
MACF1	100%	99,60%	100%	100%	Lissencephaly 9 with complex brainstem malformation, 618325
MAD1L1	100%	97,60%	100%	100%	Prostate cancer, somatic, 176807 Lymphoma, somatic, 0
MAD2L2	100%	99,90%	100%	100%	?Fanconi anemia, complementation group V, 617243
MAF	83,50%	78,00%	88,60%	82,20%	Ayme-Gripp syndrome, 601088 Cataract 21, multiple types, 610202
MAFA	93,60%	71,50%	99,40%	95,80%	Insulinomatosis and diabetes mellitus, 147630

MAFB	100%	99,40%	100%	100%	Duane retraction syndrome 3, 617041 Multicentric carpotarsal osteolysis syndrome, 166300
MAG	100%	100%	100%	100%	Spastic paraplegia 75, autosomal recessive, 616680
MAGED2	99,80%	97,70%	100%	99,90%	Bartter syndrome, type 5, antenatal, transient, 300971
MAGEL2	93,00%	87,20%	100%	100%	Schaaf-Yang syndrome, 615547
MAGI2	94,50%	92,40%	94,70%	93,30%	Nephrotic syndrome, type 15, 617609
MAGT1	98,50%	96,50%	98,70%	98,70%	Congenital disorder of glycosylation, type Icc, 301031 Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853
MAK	98,70%	96,80%	100%	100%	Retinitis pigmentosa 62, 614181
MALT1	91,20%	89,40%	100%	100%	Immunodeficiency 12, 615468
MAML2	100%	99,60%	100%	100%	Mucoepidermoid salivary gland carcinoma, 0
MAMLD1	99,80%	98,20%	100%	100%	Hypospadias 2, X-linked, 300758
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
MAOA	100%	99,70%	99,80%	98,50%	Brunner syndrome, 300615
MAP2K1	99,80%	97,10%	100%	100%	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	98,50%	95,10%	100%	100%	Cardiofaciocutaneous syndrome 4, 615280
MAP3K1	96,10%	91,60%	99,70%	98,30%	46XY sex reversal 6, 613762
MAP3K20	100%	99,50%	100%	100%	Split-foot malformation with mesoaxial polydactyly, 616890 Centronuclear myopathy 6 with fiber-type disproportion, 617760
MAP3K7	100%	99,60%	100%	100%	Cardiospondylocarpofacial syndrome, 157800 Frontometaphyseal dysplasia 2, 617137
MAP3K8	100%	99,90%	100%	100%	Lung cancer, somatic, 211980
MAPK8IP3	100%	99,60%	100%	100%	Neurodevelopmental disorder with or without variable brain abnormalities, 618443
MAPKAPK3	100%	99,60%	100%	100%	?Macular dystrophy, patterned, 3, 617111
MAPKBP1	100%	100%	100%	100%	Nephronophthisis 20, 617271
MAPRE2	100%	99,30%	100%	100%	Symmetric circumferential skin creases, congenital, 2, 616734
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
MARCH6	99,90%	98,40%	100%	100%	Epilepsy, familial adult myoclonic, 3, 613608
MARK3	99,90%	98,80%	100%	100%	?Visual impairment and progressive phthisis bulbi, 618283
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
MASP2	100%	99,60%	100%	100%	MASP2 deficiency, 613791
MAST1	100%	99,50%	100%	100%	Mega-corpus-callosum syndrome with cerebellar hypoplasia and cortical malformations, 618273
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
MATR3	97,00%	93,40%	100%	100%	Amyotrophic lateral sclerosis 21, 606070
MBD5	99,90%	99,90%	100%	100%	Mental retardation, autosomal dominant 1, 156200
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
MBTPS2	100%	99,00%	100%	100%	Osteogenesis imperfecta, type XIX, 301014 ?Olmsted syndrome, X-linked, 300918 IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800
MC2R	99,90%	98,30%	100%	100%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MC4R	100%	100%	100%	100%	Obesity (BMIQ20), 618406
MCC	100%	99,70%	100%	100%	Colorectal cancer, somatic, 114500
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
MCM2	100%	100%	100%	100%	?Deafness, autosomal dominant 70, 616968
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
MCM6	100%	100%	100%	100%	Lactase persistence/nonpersistence, 223100
MCM8	100%	99,60%	94,40%	94,40%	?Premature ovarian failure 10, 612885
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
MECOM	100%	99,90%	100%	100%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738

MECP2	100%	98,70%	100%	99,90%	Mental retardation, X-linked syndromic, Lubs type, 300260 Encephalopathy, neonatal severe, 300673 Mental retardation, X-linked, syndromic 13, 300055 Rett syndrome, atypical, 312750 Rett syndrome, 312750 Rett syndrome, preserved speech variant, 312750
MECR	100%	98,90%	100%	100%	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MED12	99,80%	96,70%	100%	100%	Ohdo syndrome, X-linked, 300895 Lujan-Fryns syndrome, 309520 Opitz-Kaveggia syndrome, 305450
MED13L	100%	99,80%	100%	100%	Transposition of the great arteries, dextro-looped 1, 608808 Mental retardation and distinctive facial features with or without cardiac defects, 616789
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
MEF2C	99,90%	96,00%	100%	100%	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
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
MEI1	100%	99,30%	100%	100%	Hydatidiform mole, recurrent, 3, 618431
MEIOB	100%	98,60%	100%	100%	?Spermatogenic failure 22, 617706
MEIS2	100%	100%	100%	100%	Cleft palate, cardiac defects, and mental retardation, 600987
MEN1	99,90%	99,10%	100%	100%	Multiple endocrine neoplasia 1, 131100 Angiofibroma, somatic, 0 Adrenal adenoma, somatic, 0 Parathyroid adenoma, somatic, 0 Lipoma, somatic, 0 Carcinoid tumor of lung, 0
MEOX1	100%	98,90%	100%	100%	Klippel-Feil syndrome 2, 214300
MERTK	99,50%	98,80%	99,10%	99,10%	Retinitis pigmentosa 38, 613862
MESD	100%	99,90%	100%	100%	Osteogenesis imperfecta, type XX, 618644
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

METTL13	99,90%	99,30%	100%	100%	No OMIM disease ID
METTL23	100%	100%	100%	100%	Mental retardation, autosomal recessive 44, 615942
MFAP5	99,90%	97,60%	100%	100%	Aortic aneurysm, familial thoracic 9, 616166
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
MIB1	100%	99,90%	100%	100%	Left ventricular noncompaction 7, 615092
MICU1	98,90%	95,20%	100%	100%	Myopathy with extrapyramidal signs, 615673
MID1	99,80%	98,70%	100%	100%	Opitz GBBB syndrome, type I, 300000
MID2	99,80%	98,70%	100%	100%	?Mental retardation, X-linked 101, 300928
MIP	100%	98,90%	100%	100%	Cataract 15, multiple types, 615274
MIPEP	99,20%	96,50%	100%	100%	Combined oxidative phosphorylation deficiency 31, 617228
MIR140	NC	NC	NC	NC	Spondyloepiphyseal dysplasia, Nishimura type, 618618
MIR17HG	NC	NC	NC	NC	Feingold syndrome 2, 614326
MIR184	NC	NC	NC	NC	EDICT syndrome, 614303
MIR204	NC	NC	NC	NC	?Retinal dystrophy and iris coloboma with or without cataract, 616722
MIR96	NC	NC	NC	NC	Deafness, autosomal dominant 50, 613074
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
MKL1	98,50%	97,20%	100%	100%	?Immunodeficiency 66, 618847
MKRN3	100%	100%	100%	100%	Precocious puberty, central, 2, 615346
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
MLH3	100%	100%	100%	100%	Colorectal cancer, somatic, 114500 Colorectal cancer, hereditary nonpolyposis, type 7, 614385
MLLT10	96,80%	95,50%	97,10%	97,10%	Leukemia, acute myeloid, 601626
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
MMP1	100%	98,90%	100%	100%	COPD, rate of decline of lung function in, 606963
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
MMP19	100%	99,50%	100%	100%	Cavitary optic disc anomalies, 611543
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
MN1	100%	99,30%	100%	100%	Meningioma, 607174 CEBALID syndrome, 618774
MNX1	68,20%	58,30%	87,40%	79,20%	Currarino syndrome, 176450
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
MOG	100%	99,50%	100%	100%	?Narcolepsy 7, 614250
MOGS	100%	99,90%	100%	100%	Congenital disorder of glycosylation, type IIb, 606056
MORC2	100%	99,80%	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688

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
MPIG6B	100%	100%	100%	100%	?Thrombocytopenia, anemia, and myelofibrosis, 617441
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
MSH5	100%	100%	100%	100%	?Premature ovarian failure 13, 617442
MSH6	100%	99,80%	100%	100%	Mismatch repair cancer syndrome, 276300 Colorectal cancer, hereditary nonpolyposis, type 5, 614350
MSL3	97,00%	88,30%	100%	100%	Basilicata-Akhtar syndrome, 301032
MSMO1	96,30%	88,90%	100%	100%	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MSN	99,00%	95,70%	100%	100%	Immunodeficiency 50, 300988
MSR1	99,90%	99,50%	100%	100%	Barrett esophagus/esophageal adenocarcinoma, 614266
MSRB3	100%	99,40%	100%	100%	Deafness, autosomal recessive 74, 613718
MSTN	100%	100%	100%	100%	Muscle hypertrophy, 614160
MSTO1	99,60%	96,70%	100%	100%	Myopathy, mitochondrial, and ataxia, 617675

MSX1	96,90%	89,30%	100%	100%	Orofacial cleft 5, 608874 Ectodermal dysplasia 3, Witkop type, 189500 Tooth agenesis, selective, 1, with or without orofacial cleft, 106600
MSX2	100%	99,40%	100%	100%	Parietal foramina 1, 168500 Craniosynostosis 2, 604757 Parietal foramina with cleidocranial dysplasia, 168550
MTAP	99,10%	93,50%	100%	100%	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250
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
MTM1	99,00%	93,30%	100%	100%	Myotubular myopathy, X-linked, 310400
MTO1	91,30%	90,40%	91,60%	91,40%	Combined oxidative phosphorylation deficiency 10, 614702
MTOR	100%	99,50%	100%	100%	Smith-Kingsmore syndrome, 616638 Focal cortical dysplasia, type II, somatic, 607341
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
MUC1	92,40%	83,60%	100%	99,90%	Medullary cystic kidney disease 1, 174000
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
MVD	99,90%	98,30%	100%	100%	Porokeratosis 7, multiple types, 614714
MVK	90,90%	90,50%	90,50%	90,50%	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377
MXI1	98,60%	95,30%	97,70%	94,10%	Prostate cancer, somatic, 176807 Neurofibrosarcoma, somatic, 0
MYBPC1	99,90%	99,50%	100%	100%	Arthrogryposis, distal, type 1B, 614335 Myopathy, congenital, with tremor, 618524 Lethal congenital contracture syndrome 4, 614915

MYBPC3	99,90%	97,60%	100%	100%	Cardiomyopathy, hypertrophic, 4, 115197 Cardiomyopathy, dilated, 1MM, 615396 Left ventricular noncompaction 10, 615396
MYC	65,90%	64,40%	100%	100%	Burkitt lymphoma, somatic, 113970
MYCN	100%	99,90%	99,30%	96,70%	Feingold syndrome 1, 164280
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
MYH11	100%	100%	100%	100%	Aortic aneurysm, familial thoracic 4, 132900
MYH14	98,40%	94,00%	100%	100%	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369 Deafness, autosomal dominant 4A, 600652
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
MYH6	99,40%	97,10%	100%	100%	Atrial septal defect 3, 614089 Cardiomyopathy, hypertrophic, 14, 613251 Cardiomyopathy, dilated, 1EE, 613252
MYH7	99,60%	97,30%	100%	100%	Myopathy, myosin storage, autosomal recessive, 255160 Left ventricular noncompaction 5, 613426 Laing distal myopathy, 160500 Myopathy, myosin storage, autosomal dominant, 608358 Cardiomyopathy, dilated, 1S, 613426 Scapuloperoneal syndrome, myopathic type, 181430 Cardiomyopathy, hypertrophic, 1, 192600
MYH8	100%	99,60%	100%	100%	Carney complex variant, 608837 Trismus-pseudocamptodactyly syndrome, 158300
MYH9	100%	99,30%	100%	100%	Deafness, autosomal dominant 17, 603622 Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100
MYL1	100%	99,70%	100%	100%	Myopathy, congenital, with fast-twitch (type II) fiber atrophy, 618414
MYL2	100%	99,80%	100%	100%	Cardiomyopathy, hypertrophic, 10, 608758
MYL3	100%	100%	100%	100%	Cardiomyopathy, hypertrophic, 8, 608751
MYL4	100%	100%	100%	100%	?Atrial fibrillation, familial, 18, 617280
MYLK	100%	99,90%	100%	100%	Aortic aneurysm, familial thoracic 7, 613780 Megacystis-microcolon-intestinal hypoperistalsis syndrome, 249210
MYLK2	100%	100%	100%	100%	Cardiomyopathy, hypertrophic, 1, digenic, 192600

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
MYOC	100%	98,60%	100%	100%	Glaucoma 1A, primary open angle, 137750
MYOT	100%	99,60%	100%	100%	Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
MYOZ2	100%	100%	100%	100%	Cardiomyopathy, hypertrophic, 16, 613838
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
MYRF	99,30%	98,50%	100%	100%	Cardiac-urogenital syndrome, 618280 Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113
MYSM1	100%	99,10%	100%	100%	Bone marrow failure syndrome 4, 618116
MYT1L	99,80%	99,00%	100%	100%	Mental retardation, autosomal dominant 39, 616521
NAA10	99,70%	98,50%	99,90%	99,90%	Ogden syndrome, 300855 ?Microphthalmia, syndromic 1, 309800
NAA15	95,80%	91,00%	96,80%	96,70%	Mental retardation, autosomal dominant 50, 617787
NACC1	100%	99,80%	100%	100%	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393
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
NANOS1	99,60%	95,10%	95,20%	88,80%	Spermatogenic failure 12, 615413
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
NCSTN	100%	99,80%	100%	100%	Acne inversa, familial, 1, 142690
NDE1	100%	100%	100%	100%	Lissencephaly 4 (with microcephaly), 614019 ?Microhydranencephaly, 605013
NDN	98,70%	89,10%	100%	100%	Prader-Willi syndrome, 176270
NDP	100%	99,70%	100%	100%	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600
NDRG1	100%	100%	100%	100%	Charcot-Marie-Tooth disease, type 4D, 601455
NDST1	100%	100%	100%	100%	Mental retardation, autosomal recessive 46, 616116
NDUFA1	99,90%	99,30%	100%	100%	Mitochondrial complex I deficiency, nuclear type 12, 301020
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
NEDD4L	72,00%	71,50%	100%	100%	Periventricular nodular heterotopia 7, 617201
NEFH	93,40%	84,50%	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2CC, 616924
NEFL	99,90%	98,20%	100%	100%	Charcot-Marie-Tooth disease, type 1F, 607734 Charcot-Marie-Tooth disease, dominant intermediate G, 617882 Charcot-Marie-Tooth disease, type 2E, 607684
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
NEUROD1	100%	99,10%	100%	100%	Maturity-onset diabetes of the young 6, 606394
NEUROD2	100%	99,90%	100%	100%	Epileptic encephalopathy, early infantile, 72, 618374
NEUROG3	100%	100%	100%	100%	Diarrhea 4, malabsorptive, congenital, 610370
NEXMIF	100%	99,50%	100%	100%	Mental retardation, X-linked 98, 300912
NEXN	92,00%	77,50%	100%	99,90%	Cardiomyopathy, hypertrophic, 20, 613876 Cardiomyopathy, dilated, 1CC, 613122
NF1	92,60%	90,20%	100%	100%	Watson syndrome, 193520 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321 Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210
NF2	100%	99,90%	100%	100%	Meningioma, NF2-related, somatic, 607174 Schwannomatosis, somatic, 162091 Neurofibromatosis, type 2, 101000
NFASC	100%	99,90%	100%	100%	Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356
NFE2L2	100%	99,90%	100%	100%	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744
NFIA	100%	99,60%	100%	100%	Brain malformations with or without urinary tract defects, 613735
NFIB	97,40%	96,50%	100%	100%	Macrocephaly, acquired, with impaired intellectual development, 618286
NFIX	100%	99,50%	99,60%	98,70%	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753
NFKB1	100%	99,40%	100%	100%	Immunodeficiency, common variable, 12, 616576
NFKB2	98,80%	95,60%	100%	100%	Immunodeficiency, common variable, 10, 615577
NFKBIA	95,20%	88,00%	100%	100%	Ectodermal dysplasia and immunodeficiency 2, 612132
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
NHS	95,40%	93,90%	100%	99,80%	Nance-Horan syndrome, 302350 Cataract 40, X-linked, 302200

NIN	100%	99,50%	100%	100%	?Seckel syndrome 7, 614851
NIPA1	100%	100%	99,80%	98,50%	Spastic paraplegia 6, autosomal dominant, 600363
NIPAL4	100%	99,10%	100%	100%	Ichthyosis, congenital, autosomal recessive 6, 612281
NIPBL	98,90%	97,00%	100%	100%	Cornelia de Lange syndrome 1, 122470
NKX2-1	98,60%	85,60%	100%	100%	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978
NKX2-5	100%	99,70%	100%	100%	Ventricular septal defect 3, 614432 Hypoplastic left heart syndrome 2, 614435 Conotruncal heart malformations, variable, 217095 Tetralogy of Fallot, 187500 Hypothyroidism, congenital nongoitrous, 5, 225250 Atrial septal defect 7, with or without AV conduction defects, 108900
NKX2-6	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
NLGN4X	99,90%	98,90%	100%	99,90%	Mental retardation, X-linked, 300495
NLRC4	100%	100%	100%	100%	Autoinflammation with infantile enterocolitis, 616050 ?Familial cold autoinflammatory syndrome 4, 616115
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
NLRP12	100%	99,90%	100%	100%	Familial cold autoinflammatory syndrome 2, 611762
NLRP3	100%	99,90%	100%	100%	Familial cold inflammatory syndrome 1, 120100 Muckle-Wells syndrome, 191900 CINCA syndrome, 607115 Deafness, autosomal dominant 34, with or without inflammation, 617772 Keratoendothelitis fugax hereditaria, 148200
NLRP7	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
NOBOX	99,90%	98,40%	100%	99,80%	Premature ovarian failure 5, 611548
NOD2	100%	99,90%	100%	100%	Blau syndrome, 186580
NODAL	100%	100%	100%	100%	Heterotaxy, visceral, 5, 270100
NOG	100%	100%	100%	100%	Tarsal-carpal coalition syndrome, 186570 Symphalangism, proximal, 1A, 185800 Stapes ankylosis with broad thumbs and toes, 184460

					Multiple synostoses syndrome 1, 186500 Brachydactyly, type B2, 611377
NOL3	93,70%	84,20%	100%	100%	?Myoclonus, familial, 1, 614937
NONO	100%	98,40%	100%	100%	Mental retardation, X-linked, syndromic 34, 300967
NOP10	100%	99,80%	100%	100%	Dyskeratosis congenita, autosomal recessive 1, 224230
NOP56	99,80%	98,60%	100%	100%	Spinocerebellar ataxia 36, 614153
NOTCH1	99,20%	97,20%	100%	100%	Aortic valve disease 1, 109730 Adams-Oliver syndrome 5, 616028
NOTCH2	100%	99,50%	100%	100%	Hajdu-Cheney syndrome, 102500 Alagille syndrome 2, 610205
NOTCH3	94,00%	90,20%	99,90%	99,40%	?Myofibromatosis, infantile 2, 615293 Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310 Lateral meningocele syndrome, 130720
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
NPM1	98,20%	85,30%	100%	100%	Leukemia, acute myeloid, somatic, 601626
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
NPRL2	100%	100%	100%	100%	Epilepsy, familial focal, with variable foci 2, 617116
NPRL3	100%	99,60%	100%	100%	Epilepsy, familial focal, with variable foci 3, 617118
NR0B1	100%	99,50%	100%	100%	Adrenal hypoplasia, congenital, 300200 46XY sex reversal 2, dosage-sensitive, 300018
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
NR2F1	100%	100%	99,10%	95,10%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NR2F2	100%	98,50%	100%	100%	Congenital heart defects, multiple types, 4, 615779
NR3C1	100%	99,90%	100%	100%	Glucocorticoid resistance, 615962
NR3C2	100%	99,70%	100%	100%	Pseudohypoaldosteronism type I, autosomal dominant, 177735 Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115
NR4A3	99,90%	98,00%	100%	100%	Chondrosarcoma, extraskeletal myxoid, 612237
NR5A1	100%	100%	100%	100%	Adrenocortical insufficiency, 612964 46, XX sex reversal 4, 617480 Premature ovarian failure 7, 612964 Spermatogenic failure 8, 613957 46XY sex reversal 3, 612965
NRAS	100%	100%	100%	100%	Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Colorectal cancer, somatic, 114500 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Thyroid carcinoma, follicular, somatic, 188470 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224
NRIP1	100%	100%	100%	100%	?Congenital anomalies of kidney and urinary tract 3, 618270
NRL	99,50%	94,80%	100%	100%	Retinitis pigmentosa 27, 613750 Retinal degeneration, autosomal recessive, clumped pigment type, 0
NRXN1	97,40%	96,90%	100%	99,80%	Pitt-Hopkins-like syndrome 2, 614325
NSD1	100%	99,90%	100%	100%	Sotos syndrome 1, 117550
NSDHL	100%	98,70%	100%	100%	CHILD syndrome, 308050 CK syndrome, 300831
NSMCE2	99,70%	98,20%	100%	100%	Seckel syndrome 10, 617253
NSMCE3	100%	100%	100%	100%	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241
NSMF	96,10%	95,60%	100%	100%	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
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
NTF4	99,60%	93,00%	100%	100%	Glaucoma 1, open angle, 1O, 613100
NTHL1	100%	99,80%	100%	100%	Familial adenomatous polyposis 3, 616415
NTN1	100%	99,70%	100%	100%	Mirror movements 4, 618264

NTRK1	99,80%	98,20%	100%	100%	Insensitivity to pain, congenital, with anhidrosis, 256800
NTRK2	100%	99,90%	100%	100%	Obesity, hyperphagia, and developmental delay, 613886 Epileptic encephalopathy, early infantile, 58, 617830
NUBPL	99,70%	98,40%	100%	100%	Mitochondrial complex I deficiency, nuclear type 21, 618242
NUMA1	100%	99,80%	100%	100%	Leukemia, acute promyelocytic, somatic, 612376
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
NUP155	99,20%	97,40%	100%	100%	?Atrial fibrillation 15, 615770
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
NUTM2B-AS1	NC	NC	NC	NC	?Oculopharyngeal myopathy with leukoencephalopathy 1, 618637
NXN	100%	100%	99,90%	99,50%	Robinow syndrome, autosomal recessive 2, 618529
NYX	96,30%	94,10%	99,70%	98,80%	Night blindness, congenital stationary (complete), 1A, X-linked, 310500
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
OCRL	99,90%	98,60%	100%	99,90%	Lowe syndrome, 309000 Dent disease 2, 300555
OFD1	88,00%	73,70%	100%	99,90%	Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Simpson-Golabi-Behmel syndrome, type 2, 300209
OGG1	100%	99,80%	100%	100%	Renal cell carcinoma, clear cell, somatic, 144700

OGT	99,90%	99,00%	100%	100%	Mental retardation, X-linked 106, 300997
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
OPCML	99,60%	99,60%	100%	100%	Ovarian cancer, somatic, 167000
OPHN1	99,50%	97,60%	99,90%	98,80%	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
OPLAH	100%	99,80%	100%	100%	5-oxoprolinase deficiency, 260005
OPN1LW	67,20%	60,60%	98,30%	98,10%	Blue cone monochromacy, 303700 Colorblindness, protan, 303900
OPN1MW	66,30%	58,50%	98,90%	97,50%	Colorblindness, deutan, 303800 Blue cone monochromacy, 303700
OPN1SW	100%	100%	100%	100%	Colorblindness, tritan, 190900
OPTN	100%	99,90%	100%	100%	Glaucoma 1, open angle, E, 137760 Amyotrophic lateral sclerosis 12, 613435
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
OSBPL2	100%	100%	100%	100%	Deafness, autosomal dominant 67, 616340
OSGEP	100%	99,40%	100%	100%	Galloway-Mowat syndrome 3, 617729
OSMR	100%	99,70%	100%	100%	Amyloidosis, primary localized cutaneous, 1, 105250
OSTM1	98,60%	94,00%	100%	100%	Osteopetrosis, autosomal recessive 5, 259720
OTC	100%	100%	100%	100%	Ornithine transcarbamylase deficiency, 311250
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
OTX2	100%	99,70%	100%	100%	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125

OVOL2	95,70%	89,50%	100%	100%	Corneal dystrophy, posterior polymorphous, 1, 122000
OXCT1	99,80%	98,10%	100%	100%	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050
P2RX2	100%	100%	100%	100%	Deafness, autosomal dominant 41, 608224
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
P4HA2	100%	99,20%	100%	100%	Myopia 25, autosomal dominant, 617238
P4HB	94,60%	94,00%	100%	100%	Cole-Carpenter syndrome 1, 112240
P4HTM	99,00%	97,40%	100%	99,40%	Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493
PABPN1	66,30%	56,90%	100%	99,10%	Oculopharyngeal muscular dystrophy, 164300
PACS1	98,80%	96,90%	100%	100%	Schuurs-Hoeijmakers syndrome, 615009
PACS2	99,30%	96,20%	100%	99,80%	Epileptic encephalopathy, early infantile, 66, 618067
PADI3	100%	100%	100%	100%	Uncombable hair syndrome, 191480
PADI6	100%	99,60%	100%	100%	Preimplantation embryonic lethality 2, 617234
PAFAH1B1	94,10%	87,10%	100%	100%	Subcortical laminar heterotopia, 607432 Lissencephaly 1, 607432
PAH	100%	100%	100%	100%	Phenylketonuria, 261600
PAK1	100%	99,60%	100%	100%	Intellectual developmental disorder with macrocephaly, seizures, and speech delay, 618158
PAK3	99,30%	95,90%	100%	99,80%	Mental retardation, X-linked 30/47, 300558
PALB2	100%	100%	100%	100%	Fanconi anemia, complementation group N, 610832
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
PANX1	100%	100%	100%	100%	Oocyte maturation defect 7, 618550
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
PAX2	100%	99,90%	100%	100%	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330
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
PAX4	100%	99,80%	100%	100%	Maturity-onset diabetes of the young, type IX, 612225 Diabetes mellitus, type 2, 125853
PAX6	100%	100%	100%	100%	Optic nerve hypoplasia, 165550 ?Coloboma, ocular, 120200 Foveal hypoplasia 1, 136520 Aniridia, 106210 Keratitis, 148190 ?Coloboma of optic nerve, 120430 ?Morning glory disc anomaly, 120430 Cataract with late-onset corneal dystrophy, 106210 Anterior segment dysgenesis 5, multiple subtypes, 604229
PAX7	100%	100%	100%	100%	Myopathy, congenital, progressive, with scoliosis, 618578 Rhabdomyosarcoma 2, alveolar, 268220
PAX8	100%	99,80%	100%	100%	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700
PAX9	99,70%	99,60%	100%	100%	Tooth agenesis, selective, 3, 604625
PBX1	100%	99,40%	100%	100%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
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
PCDH19	100%	98,90%	100%	100%	Epileptic encephalopathy, early infantile, 9, 300088
PCGF2	100%	99,50%	100%	100%	Turnpenny-Fry syndrome, 618371
PCK1	100%	100%	100%	100%	?Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680
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
PCSK9	95,00%	91,90%	100%	100%	Hypercholesterolemia, familial, 3, 603776
PCYT1A	98,90%	95,50%	100%	100%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDCD10	99,90%	98,90%	100%	100%	Cerebral cavernous malformations 3, 603285

PDE10A	81,20%	80,50%	100%	100%	Dyskinesia, limb and orofacial, infantile-onset, 616921 Striatal degeneration, autosomal dominant, 616922
PDE11A	99,90%	99,70%	100%	100%	Pigmented nodular adrenocortical disease, primary, 2, 610475
PDE1C	100%	99,60%	100%	100%	?Deafness, autosomal dominant 74, 618140
PDE3A	99,90%	99,40%	100%	100%	Hypertension and brachydactyly syndrome, 112410
PDE4D	95,70%	93,50%	100%	99,80%	Acrodysostosis 2, with or without hormone resistance, 614613
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
PDE8B	99,90%	99,70%	100%	100%	Striatal degeneration, autosomal dominant, 609161 Pigmented nodular adrenocortical disease, primary, 3, 614190
PDGFB	100%	99,30%	100%	100%	Dermatofibrosarcoma protuberans, 607907 Basal ganglia calcification, idiopathic, 5, 615483 Meningioma, SIS-related, 607174
PDGFRA	100%	100%	100%	100%	Gastrointestinal stromal tumor/GIST-plus syndrome, somatic or familial, 175510 Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685
PDGFRB	99,20%	97,50%	100%	100%	Basal ganglia calcification, idiopathic, 4, 615007 Kosaki overgrowth syndrome, 616592 Myofibromatosis, infantile, 1, 228550 Premature aging syndrome, Penttinen type, 601812
PDGFRL	100%	100%	100%	100%	Hepatocellular cancer, somatic, 114550 Colorectal cancer, somatic, 114500
PDHA1	99,40%	97,10%	100%	100%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
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
PDK3	99,50%	97,20%	100%	100%	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905
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
PDYN	100%	100%	100%	100%	Spinocerebellar ataxia 23, 610245

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
PER2	100%	99,90%	100%	100%	?Advanced sleep phase syndrome, familial, 1, 604348
PER3	99,90%	98,80%	100%	100%	?Advanced sleep phase syndrome, familial, 3, 616882
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
PFN1	100%	100%	100%	100%	Amyotrophic lateral sclerosis 18, 614808
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
PGK1	92,80%	79,30%	100%	100%	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	100%	100%	100%	100%	Congenital disorder of glycosylation, type It, 614921
PGM3	100%	99,80%	100%	100%	Immunodeficiency 23, 615816
PHACTR1	100%	99,70%	100%	100%	Epileptic encephalopathy, early infantile, 70, 618298
PHC1	99,90%	99,20%	100%	100%	?Microcephaly 11, primary, autosomal recessive, 615414
PHEX	100%	99,60%	99,90%	99,20%	Hypophosphatemic rickets, X-linked dominant, 307800
PHF6	97,80%	88,30%	99,90%	98,90%	Borjeson-Forssman-Lehmann syndrome, 301900
PHF8	99,70%	96,80%	100%	100%	Mental retardation syndrome, X-linked, Siderius type, 300263
PHGDH	99,90%	98,80%	100%	100%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHIP	98,60%	96,10%	100%	99,70%	Chung-Jansen syndrome, 617991
PHKA1	99,20%	95,30%	100%	99,90%	Muscle glycogenosis, 300559
PHKA2	100%	99,70%	100%	99,60%	Glycogen storage disease, type IXa2, 306000 Glycogen storage disease, type IXa1, 306000
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
PHOX2B	100%	99,70%	99,50%	97,80%	Neuroblastoma with Hirschsprung disease, 613013 Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, 209880
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
TAB2	100%	99,70%	100%	100%	Congenital heart defects, nonsyndromic, 2, 614980
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
TAF1	99,80%	97,70%	100%	100%	Dystonia-Parkinsonism, X-linked, 314250 Mental retardation, X-linked, syndromic 33, 300966
TAF13	100%	100%	100%	100%	Mental retardation, autosomal recessive 60, 617432
TAF2	99,90%	98,60%	100%	100%	Mental retardation, autosomal recessive 40, 615599
TAF4B	97,40%	93,10%	100%	100%	?Spermatogenic failure 13, 615841
TAF6	99,80%	98,90%	100%	100%	Alazami-Yuan syndrome, 617126
TAL1	87,10%	73,10%	100%	100%	Leukemia, T-cell acute lymphocytic, somatic, 613065
TAL2	100%	100%	100%	100%	Leukemia, T-cell acute lymphocytic, somatic, 613065

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
TARDBP	100%	100%	100%	100%	Frontotemporal lobar degeneration, TARDBP-related, 612069 Amyotrophic lateral sclerosis 10, with or without FTD, 612069
TARS	99,90%	98,10%	100%	100%	Trichothiodystrophy 7, nonphotosensitive, 618546
TARS2	100%	99,30%	100%	100%	?Combined oxidative phosphorylation deficiency 21, 615918
TAT	100%	100%	100%	100%	Tyrosinemia, type II, 276600
TAZ	99,20%	96,50%	100%	100%	Barth syndrome, 302060
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
PICALM	99,50%	96,20%	100%	100%	Leukemia, acute myeloid, somatic, 601626
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
PIGA	93,80%	86,70%	100%	100%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
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
PIH1D3	99,10%	92,50%	100%	100%	Ciliary dyskinesia, primary, 36, X-linked, 300991
PIK3C2A	99,20%	96,90%	100%	100%	Oculoskeletodental syndrome, 618440
PIK3CA	100%	99,80%	100%	100%	Ovarian cancer, somatic, 167000 Colorectal cancer, somatic, 114500 CLAPO syndrome, somatic, 613089 Cowden syndrome 5, 615108 Hepatocellular carcinoma, somatic, 114550 Breast cancer, somatic, 114480 Macrodactyly, somatic, 155500 Keratosis, seborrheic, somatic, 182000 Gastric cancer, somatic, 613659 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Nevus, epidermal, somatic, 162900 CLOVE syndrome, somatic, 612918 Nonsmall cell lung cancer, somatic, 211980
PIK3CD	98,80%	96,90%	100%	100%	Immunodeficiency 14, 615513
PIK3R1	99,80%	99,00%	100%	100%	SHORT syndrome, 269880 Immunodeficiency 36, 616005 ?Agammaglobulinemia 7, autosomal recessive, 615214
PIK3R2	90,70%	89,60%	99,30%	96,10%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PIK3R5	100%	99,90%	100%	100%	Ataxia-oculomotor apraxia 3, 615217
PIKFYVE	99,90%	99,40%	100%	100%	Corneal fleck dystrophy, 121850
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
PITPNM3	99,50%	98,70%	100%	100%	Cone-rod dystrophy 5, 600977
PITX1	96,70%	92,00%	100%	100%	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800
PITX2	99,90%	97,70%	100%	100%	Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550 Anterior segment dysgenesis 4, 137600
PITX3	100%	98,40%	100%	100%	Anterior segment dysgenesis 1, multiple subtypes, 107250 Cataract 11, syndromic, autosomal recessive, 610623 Cataract 11, multiple types, 610623

PKD1	39,20%	30,00%	99,20%	98,90%	Polycystic kidney disease 1, 173900
PKD1L1	100%	99,80%	100%	100%	Heterotaxy, visceral, 8, autosomal, 617205
PKD2	95,50%	91,10%	99,30%	97,70%	Polycystic kidney disease 2, 613095
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
PKP2	95,40%	88,60%	95,00%	95,00%	Arrhythmogenic right ventricular dysplasia 9, 609040
PLA2G4A	99,90%	99,40%	100%	100%	Gastrointestinal ulceration, recurrent, with dysfunctional platelets, 618372
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
PLAG1	100%	100%	100%	100%	Adenomas, salivary gland pleomorphic, somatic, 181030
PLAU	100%	99,80%	100%	100%	Quebec platelet disorder, 601709
PLCB1	100%	99,80%	100%	100%	Epileptic encephalopathy, early infantile, 12, 613722
PLCB4	99,90%	98,80%	100%	100%	Auriculocondylar syndrome 2, 614669
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
PLCG2	100%	99,80%	100%	100%	Familial cold autoinflammatory syndrome 3, 614468 Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878
PLCZ1	99,90%	96,90%	100%	100%	?Spermatogenic failure 17, 617214
PLD1	100%	99,60%	100%	100%	Cardiac valvular defect, developmental, 212093
PLD3	99,90%	99,10%	100%	100%	?Spinocerebellar ataxia 46, 617770
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

PLIN1	99,60%	94,90%	100%	99,50%	Lipodystrophy, familial partial, type 4, 613877
PLK4	99,90%	98,20%	100%	100%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLN	100%	100%	100%	100%	Cardiomyopathy, hypertrophic, 18, 613874 Cardiomyopathy, dilated, 1P, 609909
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
PLP1	100%	99,20%	100%	100%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PLPBP	98,20%	90,10%	100%	100%	Epilepsy, early-onset, vitamin B6-dependent, 617290
PLPP6	99,20%	93,70%	100%	100%	No OMIM disease ID
PLS3	97,70%	96,10%	97,20%	97,20%	Bone mineral density QTL18, osteoporosis, 300910
PLVAP	100%	100%	100%	100%	Diarrhea 10, protein-losing enteropathy type, 618183
PMFBP1	99,90%	99,30%	100%	100%	Spermatogenic failure 31, 618112
PML	100%	99,80%	100%	100%	Leukemia, acute promyelocytic, PML/RARA type, 0
PMM2	100%	100%	100%	100%	Congenital disorder of glycosylation, type Ia, 212065
PMP2	100%	100%	100%	100%	Charcot-Marie-Tooth disease, demyelinating, type 1G, 618279
PMP22	99,20%	95,20%	95,40%	88,50%	Dejerine-Sottas disease, 145900 ?Neuropathy, inflammatory demyelinating, 139393 Charcot-Marie-Tooth disease, type 1E, 118300 Roussy-Levy syndrome, 180800 Neuropathy, recurrent, with pressure palsies, 162500 Charcot-Marie-Tooth disease, type 1A, 118220
PMPCA	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
PMVK	100%	100%	100%	100%	Porokeratosis 1, multiple types, 175800
PNKD	100%	99,90%	100%	100%	Paroxysmal nonkinesigenic dyskinesia 1, 118800
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
POF1B	95,60%	86,90%	100%	99,80%	?Premature ovarian failure 2B, 300604
POFUT1	100%	99,00%	100%	100%	Dowling-Degos disease 2, 615327
POGLUT1	99,40%	94,60%	100%	100%	?Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232 Dowling-Degos disease 4, 615696
POGZ	99,40%	99,00%	100%	100%	White-Sutton syndrome, 616364
POLA1	99,30%	95,40%	100%	99,90%	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 Van Esch-O'Driscoll syndrome, 301030
POLD1	98,50%	95,20%	100%	100%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381
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
POLG2	99,60%	98,00%	100%	99,90%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131 Mitochondrial DNA depletion syndrome 16 (hepatic type), 618528
POLH	100%	99,60%	100%	100%	Xeroderma pigmentosum, variant type, 278750
POLR1A	100%	99,40%	100%	100%	Acrofacial dysostosis, Cincinnati type, 616462
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
POLR2A	100%	100%	100%	100%	Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603
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
PORCN	100%	99,10%	100%	100%	Focal dermal hypoplasia, 305600
POU1F1	100%	99,20%	100%	100%	Pituitary hormone deficiency, combined, 1, 613038
POU3F3	73,20%	59,60%	94,90%	83,80%	Snijders Blok-Fisher syndrome, 618604
POU3F4	100%	100%	100%	100%	Deafness, X-linked 2, 304400
POU4F3	100%	100%	100%	100%	Deafness, autosomal dominant 15, 602459
PPA2	98,70%	94,00%	100%	100%	?Sudden cardiac failure, alcohol-induced, 617223 Sudden cardiac failure, infantile, 617222
PPARG	100%	100%	100%	100%	Carotid intimal medial thickness 1, 609338 Insulin resistance, severe, digenic, 604367 Obesity, severe, 601665 Lipodystrophy, familial partial, type 3, 604367
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
PPM1D	100%	99,90%	100%	100%	Breast cancer, somatic, 114480 Jansen de Vries syndrome, 617450
PPM1K	100%	100%	100%	100%	?Maple syrup urine disease, mild variant, 615135
PPOX	99,70%	96,80%	100%	100%	Porphyria variegata, 176200
PPP1CB	99,90%	99,30%	100%	100%	Noonan syndrome-like disorder with loose anagen hair 2, 617506

PPP1R15B	100%	99,60%	100%	100%	Microcephaly, short stature, and impaired glucose metabolism 2, 616817
PPP1R3A	99,70%	99,20%	100%	100%	Insulin resistance, severe, digenic, 125853
PPP2CA	100%	100%	100%	100%	Neurodevelopmental disorder and language delay with or without structural brain abnormalities, 618354
PPP2R1A	91,60%	91,50%	93,60%	93,60%	Mental retardation, autosomal dominant 36, 616362
PPP2R1B	100%	99,90%	100%	100%	Lung cancer, somatic, 211980
PPP2R2B	100%	99,40%	100%	100%	Spinocerebellar ataxia 12, 604326
PPP2R3C	99,60%	94,90%	100%	100%	Spermatogenic failure 36, 618420 Gonadal dysgenesis, dysmorphic facies, retinal dystrophy, and myopathy, 618419
PPP2R5D	100%	100%	100%	100%	Mental retardation, autosomal dominant 35, 616355
PPP3CA	99,80%	98,40%	100%	100%	Epileptic encephalopathy, infantile or early childhood, 1, 617711 Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265
PPT1	90,30%	90,30%	82,50%	82,50%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	100%	100%	100%	100%	Renpenning syndrome, 309500
PRCC	99,90%	98,50%	100%	100%	Renal cell carcinoma, papillary, 605074
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
PRDM16	99,80%	99,10%	100%	100%	Left ventricular noncompaction 8, 615373 Cardiomyopathy, dilated, 1LL, 615373
PRDM5	99,90%	99,20%	100%	100%	Brittle cornea syndrome 2, 614170
PRDM6	95,80%	87,80%	100%	100%	Patent ductus arteriosus 3, 617039
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
PRIMPOL	97,50%	94,60%	100%	100%	Myopia 22, autosomal dominant, 615420
PRKACA	80,10%	79,30%	100%	100%	Cushing syndrome, ACTH-independent adrenal, somatic, 615830
PRKACG	100%	99,90%	100%	100%	?Bleeding disorder, platelet-type, 19, 616176
PRKAG2	99,10%	97,50%	100%	100%	Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200 Cardiomyopathy, hypertrophic 6, 600858
PRKAR1A	99,30%	93,50%	100%	100%	Myxoma, intracardiac, 255960 Carney complex, type 1, 160980 Pigmented nodular adrenocortical disease, primary, 1, 610489

					Acrodysostosis 1, with or without hormone resistance, 101800 Adrenocortical tumor, somatic, 0
PRKCA	100%	100%	100%	100%	Pituitary tumor, invasive, 0
PRKCD	100%	100%	100%	100%	Autoimmune lymphoproliferative syndrome, type III, 615559
PRKCG	99,90%	98,40%	100%	100%	Spinocerebellar ataxia 14, 605361
PRKCSH	99,80%	95,40%	100%	100%	Polycystic liver disease 1, 174050
PRKD1	99,60%	98,70%	100%	100%	Congenital heart defects and ectodermal dysplasia, 617364
PRKDC	99,70%	98,00%	100%	100%	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PRKG1	99,80%	98,30%	100%	100%	Aortic aneurysm, familial thoracic 8, 615436
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
PRLR	99,90%	99,80%	100%	100%	Hyperprolactinemia, 615555 Multiple fibroadenomas of the breast, 615554
PRMT7	100%	99,90%	100%	100%	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157
PRNP	100%	100%	100%	100%	Insomnia, fatal familial, 600072 Huntington disease-like 1, 603218 Prion disease with protracted course, 606688 Cerebral amyloid angiopathy, PRNP-related, 137440 Creutzfeldt-Jakob disease, 123400 Gerstmann-Straussler disease, 137440
PROC	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
PROK2	99,90%	98,50%	100%	100%	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	100%	100%	100%	100%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
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
PRPF3	98,80%	95,30%	100%	100%	Retinitis pigmentosa 18, 601414
PRPF31	100%	98,70%	100%	100%	Retinitis pigmentosa 11, 600138
PRPF4	100%	99,80%	100%	100%	Retinitis pigmentosa 70, 615922
PRPF6	100%	99,80%	100%	100%	Retinitis pigmentosa 60, 613983

PRPF8	100%	99,30%	100%	100%	Retinitis pigmentosa 13, 600059
PRPH2	100%	100%	100%	100%	Macular dystrophy, patterned, 1, 169150 Retinitis punctata albescens, 136880 Choroidal dystrophy, central areolar 2, 613105 Retinitis pigmentosa 7 and digenic form, 608133 Leber congenital amaurosis 18, 608133 Macular dystrophy, vitelliform, 3, 608161
PRPS1	100%	100%	100%	100%	Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Phosphoribosylpyrophosphate synthetase superactivity, 300661 Deafness, X-linked 1, 304500 Arts syndrome, 301835 Gout, PRPS-related, 300661
PRRT2	100%	99,60%	100%	100%	Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751 Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066
PRRX1	100%	99,70%	100%	100%	Agnathia-otocephaly complex, 202650
PRSS1	100%	100%	100%	100%	Pancreatitis, hereditary, 167800
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
PSEN1	100%	100%	100%	100%	Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 ?Acne inversa, familial, 3, 613737 Alzheimer disease, type 3, 607822 Dementia, frontotemporal, 600274 Pick disease, 172700 Cardiomyopathy, dilated, 1U, 613694
PSEN2	100%	100%	100%	100%	Alzheimer disease-4, 606889 Cardiomyopathy, dilated, 1V, 613697
PSENEN	100%	100%	100%	100%	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736

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
PSMD12	98,60%	92,90%	100%	100%	Stankiewicz-Isidor syndrome, 617516
PSPH	100%	100%	100%	100%	Phosphoserine phosphatase deficiency, 614023
PSTPIP1	100%	99,10%	100%	99,90%	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416
PTCH1	99,20%	97,60%	99,90%	99,80%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly 7, 610828
PTCH2	99,90%	99,00%	100%	100%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Medulloblastoma, somatic, 155255
PTDSS1	100%	100%	100%	100%	Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	99,50%	97,00%	100%	100%	Prostate cancer, somatic, 176807 Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309
PTF1A	95,80%	85,60%	98,60%	93,30%	Pancreatic and cerebellar agenesis, 609069 Pancreatic agenesis 2, 615935
PTGIS	98,20%	95,10%	100%	100%	Hypertension, essential, 145500
PTH	99,70%	97,00%	100%	100%	Hypoparathyroidism, autosomal dominant, 146200 Hypoparathyroidism, autosomal recessive, 146200
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
PTHLH	99,70%	98,40%	100%	100%	Brachydactyly, type E2, 613382
PTPN11	99,10%	93,70%	100%	100%	LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Noonan syndrome 1, 163950 Leukemia, juvenile myelomonocytic, somatic, 607785
PTPN12	99,10%	96,80%	100%	100%	Colon cancer, somatic, 114500
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
PTPRF	100%	99,70%	100%	100%	?Breasts and/or nipples, aplasia or hypoplasia of, 2, 616001
PTPRJ	97,70%	97,20%	99,90%	99,60%	Colon cancer, somatic, 114500
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
PUF60	100%	99,30%	100%	100%	Verheij syndrome, 615583
PUM1	100%	99,90%	100%	100%	Spinocerebellar ataxia 47, 617931
PURA	99,00%	95,20%	100%	99,80%	Mental retardation, autosomal dominant 31, 616158
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
QRICH1	100%	99,90%	100%	100%	Ververi-Brady syndrome, 617982
QRICH2	94,30%	93,50%	100%	100%	Spermatogenic failure 35, 618341
RAB11B	100%	100%	100%	100%	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807
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
RAB39B	100%	100%	100%	100%	Waisman syndrome, 311510 Mental retardation, X-linked 72, 300271
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
RAB7A	100%	99,90%	100%	100%	Charcot-Marie-Tooth disease, type 2B, 600882
RAC1	99,90%	96,20%	100%	99,90%	Mental retardation, autosomal dominant 48, 617751
RAC2	99,90%	98,30%	100%	100%	Neutrophil immunodeficiency syndrome, 608203
RAC3	97,30%	94,40%	99,70%	98,20%	Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577

RAD21	99,20%	96,60%	100%	100%	?Mungan syndrome, 611376 Cornelia de Lange syndrome 4, 614701
RAD50	97,50%	91,60%	100%	100%	Nijmegen breakage syndrome-like disorder, 613078
RAD51	89,40%	89,40%	89,40%	89,40%	?Fanconi anemia, complementation group R, 617244 Mirror movements 2, 614508
RAD51C	100%	99,80%	100%	100%	Fanconi anemia, complementation group O, 613390
RAD54B	99,70%	97,30%	100%	100%	Colon cancer, somatic, 114500 Lymphoma, non-Hodgkin, somatic, 605027
RAD54L	100%	98,90%	100%	100%	Lymphoma, non-Hodgkin, somatic, 605027 Adenocarcinoma, colonic, somatic, 0
RAF1	100%	100%	100%	100%	LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553 Cardiomyopathy, dilated, 1NN, 615916
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
RAI1	100%	100%	100%	100%	Smith-Magenis syndrome, 182290
RAP1GDS1	99,80%	96,80%	100%	100%	Lymphocytic leukemia, acute T-cell, 0
RAPGEF2	99,80%	99,10%	100%	100%	?Epilepsy, familial adult myoclonic, 7, 618075
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
RASA1	98,80%	96,30%	100%	100%	Capillary malformation-arteriovenous malformation 1, 608354 Basal cell carcinoma, somatic, 605462
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
RAX2	100%	92,30%	100%	100%	?Macular degeneration, age-related, 6, 613757 Cone-rod dystrophy 11, 610381
RB1	98,50%	93,70%	100%	100%	Small cell cancer of the lung, somatic, 182280 Bladder cancer, somatic, 109800

					Retinoblastoma, trilateral, 180200 Osteosarcoma, somatic, 259500 Retinoblastoma, 180200
RB1CC1	99,60%	96,70%	100%	100%	Breast cancer, somatic, 114480
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
RBM10	99,50%	97,10%	100%	100%	TARP syndrome, 311900
RBM20	100%	99,90%	100%	100%	Cardiomyopathy, dilated, 1DD, 613172
RBM28	100%	100%	100%	100%	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RBM8A	99,80%	97,90%	100%	100%	Thrombocytopenia-absent radius syndrome, 274000
RBMX	94,60%	84,00%	100%	100%	?Mental retardation, X-linked, syndromic 11, Shashi type, 300238
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
RBPJ	98,40%	92,80%	100%	100%	Adams-Oliver syndrome 3, 614814
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
REEP1	78,70%	76,10%	100%	100%	Spastic paraplegia 31, autosomal dominant, 610250 ?Neuronopathy, distal hereditary motor, type VB, 614751
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
RELA	99,60%	98,80%	100%	100%	?Mucocutaneous ulceration, chronic, 618287
RELB	98,80%	88,70%	100%	100%	?Immunodeficiency 53, 617585
RELN	100%	99,80%	100%	100%	Lissencephaly 2 (Norman-Roberts type), 257320
RELT	100%	99,90%	100%	100%	Amelogenesis imperfecta, type IIIC, 618386
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
RERE	96,30%	91,40%	100%	100%	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975
REST	98,50%	98,20%	98,60%	98,60%	Fibromatosis, gingival, 5, 617626 ?Deafness, autosomal dominant 27, 612431
RET	99,90%	99,10%	100%	100%	Multiple endocrine neoplasia IIB, 162300 Pheochromocytoma, 171300 Multiple endocrine neoplasia IIA, 171400 Medullary thyroid carcinoma, 155240 Central hypoventilation syndrome, congenital, 209880
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
RGS9	98,50%	97,10%	100%	100%	Bradyopsia, 608415
RGS9BP	99,60%	95,00%	100%	100%	Bradyopsia, 608415
RHAG	100%	99,70%	100%	100%	Overhydrated hereditary stomatocytosis, 185000 Anemia, hemolytic, Rh-null, regulator type, 268150
RHBDF2	99,90%	98,60%	100%	100%	Tylosis with esophageal cancer, 148500
RHCE	98,10%	98,10%	97,60%	97,50%	Rh-null disease, amorph type, 617970
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
RHOBTB2	100%	100%	100%	100%	Epileptic encephalopathy, early infantile, 64, 618004
RIMS1	99,80%	97,70%	100%	100%	Cone-rod dystrophy 7, 603649
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
RIT1	100%	100%	100%	100%	Noonan syndrome 8, 615355
RLBP1	100%	99,90%	100%	100%	Fundus albipunctatus, 136880 Retinitis punctata albescens, 136880 Bothnia retinal dystrophy, 607475 Newfoundland rod-cone dystrophy, 607476
RLIM	100%	99,00%	100%	100%	Tonne-Kalscheuer syndrome, 300978
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
RNASEL	100%	99,80%	100%	100%	Prostate cancer 1, 601518
RNASET2	97,40%	93,10%	100%	100%	Leukoencephalopathy, cystic, without megalencephaly, 612951
RNF113A	100%	100%	100%	100%	?Trichothiodystrophy 5, nonphotosensitive, 300953
RNF125	99,90%	99,20%	100%	100%	Tenorio syndrome, 616260
RNF13	95,20%	81,60%	100%	100%	Epileptic encephalopathy, early infantile, 73, 618379
RNF139	100%	100%	100%	100%	Renal cell carcinoma, 144700
RNF168	100%	99,80%	100%	100%	RIDDLE syndrome, 611943
RNF170	99,60%	97,60%	100%	100%	Ataxia, sensory, 1, autosomal dominant, 608984
RNF212	100%	99,60%	100%	100%	Recombination rate QTL 1, 612042
RNF216	99,80%	98,70%	100%	100%	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840
RNF43	99,90%	99,10%	100%	100%	Sessile serrated polyposis cancer syndrome, 617108
RNF6	100%	99,70%	100%	100%	Esophageal carcinoma, somatic, 133239
RNPC3	91,50%	70,70%	100%	100%	?Growth hormone deficiency, isolated, type V, 618160
RNU4ATAC	NC	NC	NC	NC	Microcephalic osteodysplastic primordial dwarfism, type I, 210710 Roifman syndrome, 616651
ROBO2	99,40%	97,80%	100%	100%	Vesicoureteral reflux 2, 610878
ROBO3	98,90%	96,10%	100%	100%	Gaze palsy, familial horizontal, with progressive scoliosis, 1, 607313
ROBO4	99,90%	98,60%	100%	100%	Aortic valve disease 8, 618496
ROGDI	98,40%	95,20%	99,90%	99,10%	Kohlschutter-Tonz syndrome, 226750
ROM1	100%	99,90%	100%	100%	Retinitis pigmentosa 7, digenic form, 608133
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
RORA	96,70%	90,20%	100%	100%	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060
RORC	100%	100%	100%	100%	Immunodeficiency 42, 616622
RP1	91,50%	90,60%	100%	100%	Retinitis pigmentosa 1, 180100
RP1L1	100%	100%	100%	100%	Retinitis pigmentosa 88, 618826 Occult macular dystrophy, 613587
RP2	100%	99,80%	100%	100%	Retinitis pigmentosa 2, 312600
RP9	80,80%	75,90%	100%	99,30%	?Retinitis pigmentosa 9, 180104
RPE65	99,80%	97,80%	100%	100%	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 87 with choroidal involvement, 618697 Retinitis pigmentosa 20, 613794
RPGR	77,30%	72,70%	100%	99,90%	Cone-rod dystrophy, X-linked, 1, 304020 Retinitis pigmentosa 3, 300029 Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 Macular degeneration, X-linked atrophic, 300834
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
RPL10	97,40%	89,10%	100%	100%	Mental retardation, X-linked, syndromic, 35, 300998
RPL11	100%	100%	100%	100%	Diamond-Blackfan anemia 7, 612562
RPL15	86,80%	78,00%	100%	100%	?Diamond-Blackfan anemia 12, 615550
RPL18	100%	100%	100%	100%	?Diamond-Blackfan anemia 18, 618310
RPL21	88,80%	71,70%	100%	100%	Hypotrichosis 12, 615885
RPL26	97,20%	84,40%	100%	100%	?Diamond-Blackfan anemia 11, 614900
RPL27	73,60%	56,50%	100%	100%	?Diamond-Blackfan anemia 16, 617408
RPL35	86,40%	75,00%	100%	100%	?Diamond-Blackfan anemia 19, 618312
RPL35A	97,10%	88,70%	100%	100%	Diamond-Blackfan anemia 5, 612528
RPL5	86,20%	70,00%	100%	100%	Diamond-Blackfan anemia 6, 612561
RPS10	97,70%	91,70%	100%	100%	Diamond-Blackfan anemia 9, 613308
RPS14	98,80%	95,00%	100%	100%	Macrocytic anemia, refractory, due to 5q deletion, somatic, 153550
RPS15A	96,90%	86,70%	80,50%	80,40%	?Diamond-Blackfan anemia 20, 618313
RPS17	84,20%	69,80%	100%	100%	Diamond-Blackfan anemia 4, 612527
RPS19	100%	99,60%	100%	100%	Diamond-Blackfan anemia 1, 105650

RPS23	87,80%	80,10%	100%	100%	Brachycephaly, trichomegaly, and developmental delay, 617412
RPS24	98,50%	93,40%	100%	100%	Diamond-blackfan anemia 3, 610629
RPS26	95,70%	84,90%	100%	100%	Diamond-Blackfan anemia 10, 613309
RPS27	89,30%	60,90%	100%	99,80%	?Diamond-Blackfan anemia 17, 617409
RPS28	100%	94,80%	100%	100%	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164
RPS29	100%	98,20%	100%	100%	Diamond-Blackfan anemia 13, 615909
RPS6KA3	98,40%	94,40%	99,90%	98,80%	Mental retardation, X-linked 19, 300844 Coffin-Lowry syndrome, 303600
RPS7	80,00%	68,70%	100%	100%	Diamond-Blackfan anemia 8, 612563
RPSA	100%	99,80%	100%	100%	Asplenia, isolated congenital, 271400
RRAS2	96,80%	88,70%	100%	100%	Noonan syndrome 12, 618624 Ovarian carcinoma, 0
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
RS1	99,80%	93,90%	100%	100%	Retinoschisis, 312700
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
RTN2	100%	99,20%	100%	100%	Spastic paraplegia 12, autosomal dominant, 604805
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
RUNX1	99,30%	94,90%	100%	100%	Leukemia, acute myeloid, 601626 Platelet disorder, familial, with associated myeloid malignancy, 601399

RUNX2	72,20%	72,20%	100%	100%	Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510 Cleidocranial dysplasia, 119600 Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600
RUSC2	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
RYR2	99,90%	99,00%	100%	100%	Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772 Arrhythmogenic right ventricular dysplasia 2, 600996
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
SALL1	99,90%	99,00%	100%	100%	Townes-Brocks syndrome 1, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480
SALL2	100%	100%	100%	100%	?Coloboma, ocular, autosomal recessive, 216820
SALL4	98,60%	96,70%	100%	100%	Duane-radial ray syndrome, 607323 IVIC syndrome, 147750
SAMD12	100%	100%	100%	100%	Epilepsy, familial adult myoclonic, 1, 601068
SAMD9	100%	99,80%	100%	100%	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455
SAMD9L	100%	100%	100%	100%	Ataxia-pancytopenia syndrome, 159550
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
SATB2	99,70%	97,40%	100%	100%	Glass syndrome, 612313
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
SCN10A	100%	99,70%	100%	100%	Episodic pain syndrome, familial, 2, 615551
SCN11A	99,80%	98,30%	100%	100%	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN1A	99,90%	99,50%	100%	100%	Febrile seizures, familial, 3A, 604403 Migraine, familial hemiplegic, 3, 609634 Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Epileptic encephalopathy, early infantile, 6 (Dravet syndrome), 607208
SCN1B	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
SCN2A	99,60%	97,60%	100%	100%	Epileptic encephalopathy, early infantile, 11, 613721 Seizures, benign familial infantile, 3, 607745
SCN2B	100%	100%	100%	100%	Atrial fibrillation, familial, 14, 615378
SCN3A	99,90%	99,30%	100%	100%	Epilepsy, familial focal, with variable foci 4, 617935 Epileptic encephalopathy, early infantile, 62, 617938
SCN3B	100%	100%	100%	100%	Brugada syndrome 7, 613120 Atrial fibrillation, familial, 16, 613120
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
SCN4B	100%	99,60%	100%	100%	Atrial fibrillation, familial, 17, 611819 Long QT syndrome 10, 611819
SCN5A	99,00%	99,00%	100%	100%	Atrial fibrillation, familial, 10, 614022 Sick sinus syndrome 1, 608567 Ventricular fibrillation, familial, 1, 603829 Long QT syndrome 3, 603830 Heart block, nonprogressive, 113900 Cardiomyopathy, dilated, 1E, 601154 Brugada syndrome 1, 601144 Heart block, progressive, type IA, 113900
SCN8A	100%	99,80%	100%	100%	Seizures, benign familial infantile, 5, 617080 Cognitive impairment with or without cerebellar ataxia, 614306

					?Myoclonus, familial, 2, 618364 Epileptic encephalopathy, early infantile, 13, 614558
SCN9A	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
SDHAF2	94,60%	94,20%	98,90%	95,40%	Paragangliomas 2, 601650
SDHB	100%	100%	100%	100%	Gastrointestinal stromal tumor, 606764 Pheochromocytoma, 171300 Paragangliomas 4, 115310 Paraganglioma and gastric stromal sarcoma, 606864
SDHC	100%	99,30%	100%	100%	Paragangliomas 3, 605373 Paraganglioma and gastric stromal sarcoma, 606864 Gastrointestinal stromal tumor, 606764

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
SEC61A1	100%	100%	100%	100%	Hyperuricemic nephropathy, familial juvenile, 4, 617056
SEC63	91,20%	83,30%	100%	100%	Polycystic liver disease 2, 617004
SECISBP2	99,80%	97,40%	100%	100%	Thyroid hormone metabolism, abnormal, 609698
SELENBP1	100%	100%	100%	100%	Extraoral halitosis due to MTO deficiency, 618148
SELENON	84,50%	84,00%	87,70%	85,10%	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310
SEMA3E	100%	99,60%	100%	100%	?CHARGE syndrome, 214800
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
SEPT12	100%	98,80%	100%	100%	Spermatogenic failure 10, 614822
SEPT9	100%	99,90%	100%	100%	Amyotrophy, hereditary neuralgic, 162100
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
SERPINA3	100%	100%	100%	100%	Cerebrovascular disease, occlusive, 0 Alpha-1-antichymotrypsin deficiency, 0
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
SERPIND1	100%	100%	100%	100%	Thrombophilia due to heparin cofactor II deficiency, 612356
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
SERPINI1	99,90%	99,00%	100%	100%	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218
SET	98,30%	90,50%	98,90%	97,00%	Mental retardation, autosomal dominant 58, 618106
SETBP1	99,90%	98,70%	100%	100%	Mental retardation, autosomal dominant 29, 616078 Schinzel-Giedion midface retraction syndrome, 269150
SETD2	100%	99,90%	100%	100%	Luscan-Lumish syndrome, 616831
SETD5	100%	99,80%	100%	100%	Mental retardation, autosomal dominant 23, 615761
SETX	100%	99,80%	100%	100%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 Amyotrophic lateral sclerosis 4, juvenile, 602433
SF3B1	99,70%	98,60%	100%	100%	Myelodysplastic syndrome, somatic, 614286
SF3B4	99,90%	97,30%	100%	100%	Acrofacial dysostosis 1, Nager type, 154400
SFRP4	100%	99,80%	100%	100%	Pyle disease, 265900
SFTPA2	100%	100%	100%	100%	Pulmonary fibrosis, idiopathic, 178500
SFTPB	99,50%	96,80%	100%	100%	Surfactant metabolism dysfunction, pulmonary, 1, 265120
SFTPC	99,20%	95,70%	100%	100%	Surfactant metabolism dysfunction, pulmonary, 2, 610913
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
SGCE	98,70%	94,00%	95,20%	95,20%	Dystonia-11, myoclonic, 159900
SGCG	100%	99,20%	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700
SGMS2	100%	100%	100%	100%	Calvarial doughnut lesions with bone fragility with or without spondylometaphyseal dysplasia, 126550
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
SH2B3	99,40%	95,10%	100%	99,90%	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950 Erythrocytosis, somatic, 133100
SH2D1A	97,20%	94,00%	100%	100%	Lymphoproliferative syndrome, X-linked, 1, 308240
SH3BP2	91,40%	91,20%	97,00%	95,30%	Cherubism, 118400
SH3KBP1	99,70%	95,90%	100%	100%	?Immunodeficiency 61, 300310
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
SHANK3	92,20%	82,10%	98,00%	94,40%	Phelan-McDermid syndrome, 606232
SHH	100%	99,50%	100%	100%	Schizencephaly, 269160 Microphthalmia with coloboma 5, 611638 Single median maxillary central incisor, 147250 Holoprosencephaly 3, 142945
SHOC2	99,90%	99,40%	100%	100%	Noonan syndrome-like with loose anagen hair, 607721
SHOX	70,00%	59,70%	95,10%	95,10%	Langer mesomelic dysplasia, 249700 Short stature, idiopathic familial, 300582 Leri-Weill dyschondrosteosis, 127300
SHROOM4	100%	99,00%	100%	100%	Stocco dos Santos X-linked mental retardation syndrome, 300434
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
SIK1	98,70%	94,40%	100%	100%	Epileptic encephalopathy, early infantile, 30, 616341
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
SIN3A	100%	99,00%	100%	100%	Witteveen-Kolk syndrome, 613406
SIPA1L3	99,90%	98,90%	100%	100%	?Cataract 45, 616851
SIX1	100%	99,20%	100%	100%	Deafness, autosomal dominant 23, 605192 Branchiootic syndrome 3, 608389
SIX3	99,90%	98,60%	100%	100%	Holoprosencephaly 2, 157170 Schizencephaly, 269160
SIX5	95,40%	88,20%	100%	100%	Branchiootorenal syndrome 2, 610896
SIX6	100%	100%	100%	100%	Optic disc anomalies with retinal and/or macular dystrophy, 212550
SKI	99,30%	94,90%	100%	99,40%	Shprintzen-Goldberg syndrome, 182212
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
SLC16A12	100%	99,90%	100%	100%	Cataract 47, juvenile, with microcornea, 612018
SLC16A2	99,20%	93,70%	100%	100%	Allan-Herndon-Dudley syndrome, 300523
SLC17A5	99,60%	97,00%	100%	100%	Sialic acid storage disorder, infantile, 269920 Salla disease, 604369
SLC17A8	100%	100%	100%	100%	Deafness, autosomal dominant 25, 605583
SLC17A9	96,30%	95,40%	100%	100%	Porokeratosis 8, disseminated superficial actinic type, 616063
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
SLC1A2	100%	99,30%	100%	100%	Epileptic encephalopathy, early infantile, 41, 617105
SLC1A3	100%	99,90%	100%	100%	Episodic ataxia, type 6, 612656
SLC1A4	99,00%	95,80%	100%	100%	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC20A2	100%	99,20%	100%	100%	Basal ganglia calcification, idiopathic, 1, 213600
SLC22A12	100%	99,80%	100%	100%	Hypouricemia, renal, 220150
SLC22A18	100%	98,10%	100%	100%	Lung cancer, somatic, 211980 Breast cancer, somatic, 114480 Rhabdomyosarcoma, somatic, 268210
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
SLC25A11	100%	100%	100%	100%	Paragangliomas 6, 618464
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
SLC25A24	99,40%	99,30%	99,80%	99,80%	Fontaine progeroid syndrome, 612289
SLC25A26	100%	99,50%	100%	100%	Combined oxidative phosphorylation deficiency 28, 616794
SLC25A3	99,80%	98,00%	100%	100%	Mitochondrial phosphate carrier deficiency, 610773
SLC25A32	100%	100%	100%	100%	?Exercise intolerance, riboflavin-responsive, 616839
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
SLC26A8	100%	99,60%	100%	100%	Spermatogenic failure 3, 606766
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
SLC30A2	100%	99,00%	100%	100%	Zinc deficiency, transient neonatal, 608118
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
SLC35A2	99,90%	98,40%	100%	100%	Congenital disorder of glycosylation, type IIm, 300896
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
SLC36A2	100%	100%	100%	100%	Iminoglycinuria, digenic, 242600 Hyperglycinuria, 138500
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
SLC39A5	99,90%	99,00%	100%	100%	Myopia 24, autosomal dominant, 615946
SLC39A8	100%	99,70%	100%	100%	Congenital disorder of glycosylation, type IIn, 616721
SLC3A1	100%	99,80%	96,60%	96,60%	Cystinuria, 220100
SLC40A1	100%	99,50%	100%	100%	Hemochromatosis, type 4, 606069
SLC44A4	100%	99,50%	100%	100%	?Deafness, autosomal dominant 72, 617606
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
SLC52A1	100%	100%	100%	100%	Riboflavin deficiency, 615026

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
SLC6A1	100%	100%	100%	100%	Myoclonic-atonic epilepsy, 616421
SLC6A17	100%	100%	100%	100%	Mental retardation, autosomal recessive 48, 616269
SLC6A19	100%	100%	100%	100%	Iminoglycinuria, digenic, 242600 HEARTnup disorder, 234500 Hyperglycinuria, 138500
SLC6A2	100%	99,90%	100%	100%	?Orthostatic intolerance, 604715
SLC6A20	100%	99,90%	100%	100%	Iminoglycinuria, digenic, 242600 Hyperglycinuria, 138500
SLC6A3	100%	100%	100%	100%	Parkinsonism-dystonia, infantile, 1, 613135
SLC6A5	100%	100%	100%	100%	Hyperekplexia 3, 614618
SLC6A8	93,50%	81,60%	100%	99,80%	Cerebral creatine deficiency syndrome 1, 300352
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
SLC9A3R1	100%	98,70%	100%	100%	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
SLC9A6	99,30%	95,50%	100%	98,30%	Mental retardation, X-linked syndromic, Christianson type, 300243
SLC9A7	97,60%	90,30%	99,90%	99,50%	Intellectual developmental disorder, X-linked 108, 301024
SLCO1B1	99,20%	93,70%	100%	100%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO1B3	98,80%	90,80%	100%	100%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO2A1	100%	99,40%	100%	100%	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
SLFN14	100%	100%	100%	100%	Bleeding disorder, platelet-type, 20, 616913
SLITRK1	100%	100%	100%	100%	?Trichotillomania, 613229 Tourette syndrome, 137580
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

SMAD3	99,90%	99,00%	100%	100%	Loeys-Dietz syndrome 3, 613795
SMAD4	100%	99,90%	100%	100%	Polyposis, juvenile intestinal, 174900 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050
SMAD6	90,90%	81,00%	100%	99,60%	Aortic valve disease 2, 614823
SMAD9	100%	99,90%	100%	100%	Pulmonary hypertension, primary, 2, 615342
SMARCA2	96,70%	96,20%	97,40%	96,80%	Nicolaides-Baraitser syndrome, 601358
SMARCA4	99,90%	99,00%	100%	100%	Coffin-Siris syndrome 4, 614609
SMARCAD1	99,30%	95,80%	100%	100%	Huriez syndrome, 181600 Basan syndrome, 129200 Adermatoglyphia, 136000
SMARCAL1	100%	99,90%	100%	100%	Schimke immunoosseous dysplasia, 242900
SMARCB1	100%	100%	100%	100%	Rhabdoid tumors, somatic, 609322 Coffin-Siris syndrome 3, 614608
SMARCC2	99,00%	96,60%	100%	100%	Coffin-Siris syndrome 8, 618362
SMARCD2	87,00%	85,90%	99,60%	97,00%	Specific granule deficiency 2, 617475
SMARCE1	96,10%	88,10%	100%	100%	Coffin-Siris syndrome 5, 616938
SMC1A	100%	98,70%	100%	99,80%	Cornelia de Lange syndrome 2, 300590 Epileptic encephalopathy, early infantile, 85, with or without midline brain defects, 301044
SMC3	95,20%	91,00%	100%	100%	Cornelia de Lange syndrome 3, 610759
SMCHD1	99,50%	96,30%	100%	100%	Fascioscapulohumeral muscular dystrophy 2, digenic, 158901 Bosma arhinia microphthalmia syndrome, 603457
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
SMO	97,80%	94,70%	100%	100%	Curry-Jones syndrome, somatic mosaic, 601707 Basal cell carcinoma, somatic, 605462
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
SMPX	100%	97,60%	100%	100%	Deafness, X-linked 4, 300066
SMS	91,50%	78,50%	100%	99,90%	Mental retardation, X-linked, Snyder-Robinson type, 309583

SNAI2	100%	99,10%	100%	100%	Waardenburg syndrome, type 2D, 608890 Piebaldism, 172800
SNAP25	100%	99,90%	100%	100%	?Myasthenic syndrome, congenital, 18, 616330
SNAP29	100%	100%	100%	100%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNCA	100%	100%	100%	100%	Dementia, Lewy body, 127750 Parkinson disease 1, 168601 Parkinson disease 4, 605543
SNCB	100%	99,90%	100%	100%	Dementia, Lewy body, 127750
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
SNRNP200	99,90%	99,10%	100%	100%	Retinitis pigmentosa 33, 610359
SNRPB	100%	99,30%	100%	100%	Cerebrocostomandibular syndrome, 117650
SNRPE	99,50%	92,60%	100%	100%	Hypotrichosis 11, 615059
SNRPN	100%	97,00%	100%	100%	Prader-Willi syndrome, 176270
SNTA1	87,00%	78,80%	99,30%	97,20%	Long QT syndrome 12, 612955
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
SOHLH1	99,70%	96,50%	100%	100%	Ovarian dysgenesis 5, 617690 Spermatogenic failure 32, 618115
SON	98,80%	94,90%	100%	100%	ZTTK syndrome, 617140
SOS1	99,80%	98,40%	100%	100%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOS2	100%	99,20%	100%	100%	Noonan syndrome 9, 616559
SOST	100%	99,50%	100%	100%	Sclerosteosis 1, 269500 Van Buchem disease, 239100 Craniodiaphyseal dysplasia, autosomal dominant, 122860
SOX10	99,90%	97,90%	100%	100%	Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 PCWH syndrome, 609136 Waardenburg syndrome, type 4C, 613266
SOX11	100%	100%	100%	100%	Coffin-Siris syndrome 9, 615866
SOX17	100%	99,50%	100%	100%	Vesicoureteral reflux 3, 613674
SOX18	70,70%	55,20%	96,10%	92,60%	Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940 Hypotrichosis-lymphedema-telangiectasia syndrome, 607823
SOX2	100%	100%	100%	100%	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900

SOX3	91,40%	75,20%	100%	99,50%	Panhypopituitarism, X-linked, 312000 Mental retardation, X-linked, with isolated growth hormone deficiency, 300123
SOX4	97,30%	90,90%	99,90%	98,20%	Coffin-Siris syndrome 10, 618506
SOX5	99,90%	98,90%	100%	100%	Lamb-Shaffer syndrome, 616803
SOX9	100%	98,60%	100%	100%	Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290 Campomelic dysplasia with autosomal sex reversal, 114290
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
SPAST	99,80%	98,70%	100%	100%	Spastic paraplegia 4, autosomal dominant, 182601
SPATA16	100%	99,50%	100%	100%	?Spermatogenic failure 6, 102530
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
SPECC1L	100%	99,60%	100%	100%	Hypertelorism, Teebi type, 145420 ?Facial clefting, oblique, 1, 600251 Opitz GBBB syndrome, type II, 145410
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
SPINK1	100%	99,30%	100%	100%	Pancreatitis, hereditary, 167800 Tropical calcific pancreatitis, 608189
SPINK2	99,30%	99,10%	99,30%	99,30%	?Spermatogenic failure 29, 618091
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
SPRED1	100%	98,90%	100%	100%	Legius syndrome, 611431
SPRTN	100%	100%	100%	100%	Ruijs-Aalfs syndrome, 616200
SPRY4	100%	100%	100%	100%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266

SPTA1	99,90%	99,20%	100%	100%	Pyropoikilocytosis, 266140 Elliptocytosis-2, 130600 Spherocytosis, type 3, 270970
SPTAN1	99,10%	98,60%	100%	100%	Epileptic encephalopathy, early infantile, 5, 613477
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
SPTLC1	99,20%	95,40%	100%	100%	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	100%	100%	100%	100%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
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
SRC	100%	99,80%	100%	100%	Colon cancer, advanced, somatic, 114500 ?Thrombocytopenia 6, 616937
SRCAP	99,40%	98,90%	100%	100%	Floating-Harbor syndrome, 136140
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
SRP72	97,60%	89,70%	100%	100%	Bone marrow failure syndrome 1, 614675
SRPX2	99,80%	96,50%	100%	100%	?Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643
SRY	50,00%	50,00%	60,00%	60,00%	46XY sex reversal 1, 400044
SSR4	100%	99,70%	100%	100%	Congenital disorder of glycosylation, type Iy, 300934
SSTR5	100%	99,90%	100%	100%	Somatostatin analog, resistance to, 0
SSX1	82,20%	82,00%	100%	100%	?Sarcoma, synovial, 300813
SSX2	65,30%	62,60%	100%	100%	?Sarcoma, synovial, 300813
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
STAG1	99,60%	97,30%	100%	100%	Mental retardation, autosomal dominant 47, 617635
STAG2	97,60%	89,40%	99,90%	98,70%	Mullegama-Klein-Martinez syndrome, 301022 Holoprosencephaly 13, X-linked, 301043
STAG3	93,50%	93,20%	100%	100%	Premature ovarian failure 8, 615723

STAMBP	100%	99,40%	100%	100%	Microcephaly-capillary malformation syndrome, 614261
STAR	100%	100%	100%	100%	Lipoid adrenal hyperplasia, 201710
STARD7	98,20%	93,10%	100%	100%	Epilepsy, familial adult myoclonic, 2, 607876
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
STAT3	100%	99,80%	100%	100%	Hyper-IgE recurrent infection syndrome, 147060 Autoimmune disease, multisystem, infantile-onset, 1, 615952
STAT5B	100%	98,50%	100%	100%	Leukemia, acute promyelocytic, somatic, 102578 Growth hormone insensitivity with immunodeficiency, 245590
STEAP3	100%	99,70%	100%	100%	?Anemia, hypochromic microcytic, with iron overload 2, 615234
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
STK11	100%	99,30%	100%	100%	Testicular tumor, somatic, 273300 Peutz-Jeghers syndrome, 175200 Pancreatic cancer, somatic, 260350 Melanoma, malignant, somatic, 0
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
STOX1	89,60%	89,60%	97,70%	95,60%	Preeclampsia/eclampsia 4, 609404
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
STS	99,70%	98,10%	100%	99,90%	Ichthyosis, X-linked, 308100
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
STX16	100%	98,60%	100%	100%	Pseudohypoparathyroidism, type IB, 603233
STX1B	100%	100%	100%	100%	Generalized epilepsy with febrile seizures plus, type 9, 616172
STXBP1	96,80%	96,50%	100%	100%	Epileptic encephalopathy, early infantile, 4, 612164
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
SUGCT	99,90%	98,50%	100%	100%	Glutaric aciduria III, 231690
SULT2B1	100%	100%	100%	100%	Ichthyosis, congenital, autosomal recessive 14, 617571
SUMF1	97,50%	90,80%	100%	100%	Multiple sulfatase deficiency, 272200
SUMO1	67,20%	49,90%	69,40%	69,40%	?Orofacial cleft 10, 613705
SUN5	100%	99,80%	100%	100%	Spermatogenic failure 16, 617187
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
SYCE1	100%	98,60%	100%	100%	?Premature ovarian failure 12, 616947 ?Spermatogenic failure 15, 616950
SYCP3	99,70%	98,20%	100%	100%	Spermatogenic failure 4, 270960 Pregnancy loss, recurrent, 4, 270960
SYN1	81,90%	73,20%	100%	99,60%	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
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
SYNE2	99,70%	98,20%	100%	100%	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999
SYNE4	99,70%	97,00%	100%	100%	Deafness, autosomal recessive 76, 615540
SYNGAP1	99,40%	98,10%	100%	100%	Mental retardation, autosomal dominant 5, 612621
SYNJ1	99,90%	99,40%	100%	100%	Epileptic encephalopathy, early infantile, 53, 617389 Parkinson disease 20, early-onset, 615530
SYP	99,90%	96,70%	100%	100%	Mental retardation, X-linked 96, 300802
SYT1	99,80%	98,50%	100%	100%	Baker-Gordon syndrome, 618218
SYT14	61,90%	58,90%	95,90%	95,90%	?Spinocerebellar ataxia, autosomal recessive 11, 614229
SYT2	99,90%	99,00%	100%	100%	Myasthenic syndrome, congenital, 7, presynaptic, 616040
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
TBC1D8B	98,50%	93,20%	100%	100%	Nephrotic syndrome, type 20, 301028
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
TBK1	99,70%	97,20%	100%	100%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439
TBL1X	97,50%	92,10%	100%	100%	Hypothyroidism, congenital, nongoitrous, 8, 301033
TBL1XR1	96,50%	84,90%	100%	100%	Pierpont syndrome, 602342 Mental retardation, autosomal dominant 41, 616944
TBL1Y	49,40%	45,30%	60,00%	59,90%	?Deafness, Y-linked 2, 400047
TBP	100%	99,90%	100%	100%	Spinocerebellar ataxia 17, 607136
TBR1	99,90%	97,90%	100%	100%	Intellectual developmental disorder with autism and speech delay, 606053
TBX1	86,90%	79,50%	94,10%	90,80%	Velocardiofacial syndrome, 192430 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Conotruncal anomaly face syndrome, 217095
TBX15	100%	99,90%	100%	100%	Cousin syndrome, 260660
TBX18	99,50%	97,10%	100%	100%	Congenital anomalies of kidney and urinary tract 2, 143400
TBX19	100%	100%	100%	100%	Adrenocorticotropic hormone deficiency, 201400
TBX2	99,90%	97,50%	99,00%	96,90%	Vertebral anomalies and variable endocrine and T-cell dysfunction, 618223
TBX20	100%	99,70%	100%	100%	Atrial septal defect 4, 611363
TBX21	95,40%	86,60%	100%	100%	Asthma and nasal polyps, 208550
TBX22	99,20%	95,70%	100%	100%	?Abruzzo-Erickson syndrome, 302905 Cleft palate with ankyloglossia, 303400
TBX3	99,20%	96,80%	100%	100%	Ulnar-mammary syndrome, 181450
TBX4	97,60%	95,10%	100%	100%	Amelia, posterior, with pelvic and pulmonary hypoplasia syndrome, 601360 Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension, 147891
TBX5	100%	100%	100%	100%	Holt-Oram syndrome, 142900
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

TCF12	100%	99,90%	100%	100%	Craniosynostosis 3, 615314
TCF20	100%	100%	100%	100%	Developmental delay with variable intellectual impairment and behavioral abnormalities, 618430
TCF3	97,10%	94,00%	100%	100%	Agammaglobulinemia 8, autosomal dominant, 616941
TCF4	100%	99,80%	100%	100%	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954
TCHH	100%	98,80%	100%	100%	?Uncombable hair syndrome 3, 617252
TCIRG1	97,60%	90,10%	100%	100%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	100%	100%	100%	100%	Transcobalamin II deficiency, 275350
TCOF1	99,70%	98,60%	100%	100%	Treacher Collins syndrome 1, 154500
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
TDGF1	99,90%	96,70%	100%	100%	Forebrain defects, 0
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
TDRD9	99,30%	98,20%	100%	99,90%	?Spermatogenic failure 30, 618110
TEAD1	100%	99,90%	100%	100%	Sveinsson chorioretinal atrophy, 108985
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
TEK	100%	100%	100%	100%	Glaucoma 3, primary congenital, E, 617272 Venous malformations, multiple cutaneous and mucosal, 600195
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
TENM4	100%	99,60%	100%	100%	Essential tremor, hereditary, 5, 616736
TERC	NC	NC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550
TET2	100%	100%	100%	100%	Myelodysplastic syndrome, somatic, 614286
TEX11	93,80%	88,20%	97,10%	97,00%	Spermatogenic failure, X-linked, 2, 309120
TEX14	99,90%	98,90%	100%	100%	Spermatogenic failure 23, 617707
TEX15	100%	99,70%	100%	100%	Spermatogenic failure 25, 617960

TF	100%	100%	100%	100%	Atransferrinemia, 209300
TFAM	97,50%	83,50%	100%	100%	?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156
TFAP2A	99,40%	94,30%	100%	100%	Branchiooculofacial syndrome, 113620
TFAP2B	99,90%	98,60%	100%	100%	Char syndrome, 169100 Patent ductus arteriosus 2, 617035
TFE3	99,30%	94,20%	100%	100%	Renal cell carcinoma, papillary, 1, 300854
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
TGFB2	100%	100%	100%	100%	Loeys-Dietz syndrome 4, 614816
TGFB3	100%	100%	100%	100%	Loeys-Dietz syndrome 5, 615582 Arrhythmogenic right ventricular dysplasia 1, 107970
TGFBI	99,50%	94,60%	100%	100%	Corneal dystrophy, lattice type IIIA, 608471 Corneal dystrophy, Groenouw type I, 121900 Corneal dystrophy, lattice type I, 122200 Corneal dystrophy, Reis-Bucklers type, 608470 Corneal dystrophy, Thiel-Behnke type, 602082 Corneal dystrophy, epithelial basement membrane, 121820 Corneal dystrophy, Avellino type, 607541
TGFBR1	93,70%	93,60%	99,00%	96,30%	Loeys-Dietz syndrome 1, 609192
TGFBR2	100%	100%	100%	100%	Esophageal cancer, somatic, 133239 Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Loeys-Dietz syndrome 2, 610168
TGIF1	100%	100%	100%	100%	Holoprosencephaly 4, 142946
TGM1	100%	99,90%	100%	100%	Ichthyosis, congenital, autosomal recessive 1, 242300
TGM3	100%	99,70%	100%	100%	?Uncombable hair syndrome 2, 617251
TGM5	100%	99,70%	100%	100%	Peeling skin syndrome 2, 609796
TGM6	99,70%	97,30%	100%	100%	Spinocerebellar ataxia 35, 613908
TH	99,30%	96,10%	100%	100%	Segawa syndrome, recessive, 605407
THAP1	100%	100%	100%	100%	Dystonia 6, torsion, 602629
THBD	100%	99,70%	100%	100%	Thrombophilia due to thrombomodulin defect, 614486
THOC2	98,80%	93,70%	100%	100%	Mental retardation, X-linked 12/35, 300957
THOC6	100%	100%	100%	100%	Beaulieu-Boycott-Innes syndrome, 613680

THPO	100%	99,50%	100%	100%	Thrombocythemia 1, 187950
THRA	100%	99,60%	100%	100%	Hypothyroidism, congenital, nongoitrous, 6, 614450
THRB	100%	99,70%	100%	100%	Thyroid hormone resistance, 188570 Thyroid hormone resistance, selective pituitary, 145650 Thyroid hormone resistance, autosomal recessive, 274300
TIA1	99,70%	97,80%	100%	100%	Welander distal myopathy, 604454
TIMM50	98,30%	94,40%	100%	100%	3-methylglutaconic aciduria, type IX, 617698
TIMM8A	98,00%	90,10%	100%	100%	Mohr-Tranebjaerg syndrome, 304700
TIMMDC1	100%	100%	100%	100%	Mitochondrial complex I deficiency, nuclear type 31, 618251
TIMP3	100%	100%	100%	100%	Sorsby fundus dystrophy, 136900
TINF2	100%	100%	100%	100%	Revesz syndrome, 268130 Dyskeratosis congenita, autosomal dominant 3, 613990
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
TLK2	99,10%	95,10%	100%	100%	Mental retardation, autosomal dominant 57, 618050
TLL1	100%	100%	100%	100%	Atrial septal defect 6, 613087
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
TMEM106B	99,90%	98,80%	100%	100%	Leukodystrophy, hypomyelinating, 16, 617964
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
TMEM173	99,70%	95,30%	100%	100%	STING-associated vasculopathy, infantile-onset, 615934
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
TMEM240	100%	100%	100%	100%	Spinocerebellar ataxia 21, 607454
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
TMEM43	99,90%	98,90%	100%	100%	Emery-Dreifuss muscular dystrophy 7, AD, 614302 Arrhythmogenic right ventricular dysplasia 5, 604400
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
TMEM98	99,30%	97,80%	100%	100%	Nanophthalmos 4, 615972
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
TNC	100%	99,80%	100%	100%	Deafness, autosomal dominant 56, 615629
TNFAIP3	100%	100%	100%	100%	Autoinflammatory syndrome, familial, Behcet-like, 616744
TNFRSF10B	100%	100%	100%	100%	Squamous cell carcinoma, head and neck, 275355
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
TNFRSF1A	90,60%	87,60%	92,80%	92,80%	Periodic fever, familial, 142680
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

TNNC1	100%	100%	100%	100%	Cardiomyopathy, hypertrophic, 13, 613243 Cardiomyopathy, dilated, 1Z, 611879
TNNI2	100%	99,70%	100%	100%	Arthrogryposis, distal, type 2B1, 601680
TNNI3	99,70%	95,20%	100%	100%	Cardiomyopathy, hypertrophic, 7, 613690 ?Cardiomyopathy, dilated, 2A, 611880 Cardiomyopathy, dilated, 1FF, 613286 Cardiomyopathy, familial restrictive, 1, 115210
TNNI3K	100%	99,40%	100%	100%	Cardiac conduction disease with or without dilated cardiomyopathy, 616117
TNNT1	99,90%	97,60%	100%	100%	Nemaline myopathy 5, Amish type, 605355
TNNT2	100%	100%	100%	100%	Cardiomyopathy, familial restrictive, 3, 612422 Cardiomyopathy, hypertrophic, 2, 115195 Left ventricular noncompaction 6, 601494 Cardiomyopathy, dilated, 1D, 601494
TNNT3	100%	99,70%	100%	100%	Arthrogryposis, distal, type 2B2, 618435
TNPO3	100%	99,90%	100%	100%	Muscular dystrophy, limb-girdle, autosomal dominant 2, 608423
TNRC6A	99,90%	99,30%	100%	100%	?Epilepsy, familial adult myoclonic, 6, 618074
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
TOP1	99,90%	98,60%	100%	100%	DNA topoisomerase I, camptothecin-resistant, 0
TOP2A	100%	99,30%	100%	100%	DNA topoisomerase II, resistance to inhibition of, by amsacrine, 0
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
TOPORS	100%	100%	100%	100%	Retinitis pigmentosa 31, 609923
TOR1A	100%	99,90%	100%	100%	Dystonia-1, torsion, 128100
TOR1AIP1	99,90%	98,00%	100%	100%	?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072
TP53	99,90%	97,70%	91,70%	91,70%	Breast cancer, somatic, 114480 Li-Fraumeni syndrome, 151623 Pancreatic cancer, somatic, 260350 Bone marrow failure syndrome 5, 618165 Nasopharyngeal carcinoma, somatic, 607107 Hepatocellular carcinoma, somatic, 114550
TP53RK	92,50%	79,60%	100%	100%	Galloway-Mowat syndrome 4, 617730
TP63	100%	100%	100%	100%	Limb-mammary syndrome, 603543 Orofacial cleft 8, 618149 Split-hand/foot malformation 4, 605289 Hay-Wells syndrome, 106260

					Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 Rapp-Hodgkin syndrome, 129400 ADULT syndrome, 103285
TPI1	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
TPM1	100%	99,40%	100%	99,90%	Left ventricular noncompaction 9, 611878 Cardiomyopathy, hypertrophic, 3, 115196 Cardiomyopathy, dilated, 1Y, 611878
TPM2	100%	100%	84,00%	83,70%	Nemaline myopathy 4, autosomal dominant, 609285 Arthrogryposis, distal, type 2B4, 108120 Arthrogryposis, distal, type 1A, 108120 CAP myopathy 2, 609285
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
TRAF3IP2	100%	99,30%	100%	100%	?Candidiasis, familial, 8, 615527
TRAF7	100%	99,80%	100%	100%	Cardiac, facial, and digital anomalies with developmental delay, 618164
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
TRAPPC2	89,70%	69,60%	100%	100%	Spondyloepiphyseal dysplasia tarda, 313400
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
TREH	96,90%	92,10%	100%	100%	Trehalase deficiency, 612119
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

TRHR	100%	99,20%	100%	100%	Hypothyroidism, congenital, nongoitrous, 7, 618573
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
TRIM44	99,80%	96,30%	100%	100%	?Aniridia 3, 617142
TRIO	99,20%	97,50%	99,30%	98,40%	Intellectual developmental disorder, autosomal dominant 44, with microcephaly, 617061 Intellectual developmental disorder, autosomal dominant 63, with macrocephaly, 618825
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
TRIP12	99,90%	99,20%	100%	100%	Mental retardation, autosomal dominant 49, 617752
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
TRPA1	96,10%	89,80%	100%	100%	?Episodic pain syndrome, familial, 1, 615040
TRPC3	99,70%	98,00%	100%	100%	?Spinocerebellar ataxia 41, 616410
TRPC6	98,20%	96,10%	100%	100%	Glomerulosclerosis, focal segmental, 2, 603965
TRPM1	100%	99,80%	100%	100%	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TRPM4	100%	99,50%	100%	100%	Erythrokeratodermia veriabilis et progressiva 6, 618531 Progressive familial heart block, type IB, 604559
TRPM6	99,90%	99,50%	100%	100%	Hypomagnesemia 1, intestinal, 602014
TRPS1	100%	99,90%	100%	100%	Trichorhinophalangeal syndrome, type I, 190350 Trichorhinophalangeal syndrome, type III, 190351
TRPV3	99,80%	98,50%	97,10%	97,10%	?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400 Olmsted syndrome, 614594
TRPV4	100%	99,90%	100%	100%	Spondylometaphyseal dysplasia, Kozlowski type, 184252 Parastremmatic dwarfism, 168400

					SED, Maroteaux type, 184095 Neuronopathy, distal hereditary motor, type VIII, 600175 Scapuloperoneal spinal muscular atrophy, 181405 Metatropic dysplasia, 156530 Digital arthropathy-brachydactyly, familial, 606835 Hereditary motor and sensory neuropathy, type IIc, 606071 Brachyolmia type 3, 113500 ?Avascular necrosis of femoral head, primary, 2, 617383
TRPV6	100%	99,50%	99,90%	98,90%	Hyperparathyroidism, transient neonatal, 618188
TRRAP	99,90%	99,50%	100%	100%	Developmental delay with or without dysmorphic facies and autism, 618454 ?Deafness, autosomal dominant 75, 618778
TSC1	99,80%	98,70%	100%	100%	Tuberous sclerosis-1, 191100 Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690
TSC2	100%	99,60%	100%	100%	Tuberous sclerosis-2, 613254 ?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690
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
TSGA10	99,90%	99,10%	100%	100%	?Spermatogenic failure 26, 617961
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
TSHZ1	98,80%	98,80%	100%	100%	Aural atresia, congenital, 607842
TSPAN12	100%	99,80%	100%	100%	Exudative vitreoretinopathy 5, 613310
TSPAN7	100%	100%	100%	100%	Mental retardation, X-linked 58, 300210
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
TSR2	100%	100%	100%	99,90%	?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946

TTBK2	99,80%	97,60%	100%	100%	Spinocerebellar ataxia 11, 604432
TTC19	81,50%	73,80%	100%	99,20%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTC21A	100%	100%	100%	100%	Spermatogenic failure 37, 618429
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
TTN	98,60%	98,10%	100%	100%	Cardiomyopathy, dilated, 1G, 604145 Tibial muscular dystrophy, tardive, 600334 Salih myopathy, 611705 Cardiomyopathy, familial hypertrophic, 9, 613765 Myopathy, myofibrillar, 9, with early respiratory failure, 603689 Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807
TTPA	94,70%	87,10%	100%	100%	Ataxia with isolated vitamin E deficiency, 277460
TTR	94,60%	94,60%	94,60%	94,60%	Amyloidosis, hereditary, transthyretin-related, 105210 Carpal tunnel syndrome, familial, 115430
TUB	99,40%	97,10%	100%	100%	?Retinal dystrophy and obesity, 616188
TUBA1A	99,90%	97,00%	100%	100%	Lissencephaly 3, 611603
TUBA3D	100%	99,20%	100%	100%	Keratoconus 9, 617928
TUBA4A	100%	100%	100%	100%	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia, 616208
TUBA8	99,90%	99,50%	100%	100%	Cortical dysplasia, complex, with other brain malformations 8, 613180
TUBB	97,30%	93,90%	99,80%	99,80%	Symmetric circumferential skin creases, congenital, 1, 156610 Cortical dysplasia, complex, with other brain malformations 6, 615771
TUBB1	100%	100%	100%	100%	Macrothrombocytopenia, autosomal dominant, TUBB1-related, 613112
TUBB2A	97,00%	95,70%	100%	100%	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB2B	100%	99,50%	100%	100%	Cortical dysplasia, complex, with other brain malformations 7, 610031
TUBB3	98,30%	96,90%	100%	100%	Fibrosis of extraocular muscles, congenital, 3A, 600638 Cortical dysplasia, complex, with other brain malformations 1, 614039
TUBB4A	95,90%	94,00%	97,10%	96,00%	Leukodystrophy, hypomyelinating, 6, 612438 Dystonia 4, torsion, autosomal dominant, 128101
TUBB4B	99,90%	96,90%	100%	100%	Leber congenital amaurosis with early-onset deafness, 617879
TUBB6	90,60%	90,10%	100%	100%	?Facial palsy, congenitla, with ptosis and velopharyngeal dysfunction, 617732

TUBB8	83,00%	55,30%	100%	100%	Oocyte maturation defect 2, 616780
TUBG1	100%	100%	100%	100%	Cortical dysplasia, complex, with other brain malformations 4, 615412
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
TWIST1	100%	98,90%	97,20%	92,30%	Robinow-Sorauf syndrome, 180750 Craniosynostosis 1, 123100 Sweeney-Cox syndrome, 617746 Saethre-Chotzen syndrome with or without eyelid anomalies, 101400
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
TXNRD2	96,80%	95,90%	100%	100%	?Glucocorticoid deficiency 5, 617825
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
UBA1	99,40%	98,20%	99,80%	99,00%	Spinal muscular atrophy, X-linked 2, infantile, 301830
UBA5	97,80%	86,80%	100%	100%	?Spinocerebellar ataxia, autosomal recessive 24, 617133 Epileptic encephalopathy, early infantile, 44, 617132
UBAP1	98,80%	93,40%	100%	100%	Spastic paraplegia 80, autosomal dominant, 618418
UBE2A	99,70%	96,00%	100%	99,70%	Mental retardation, X-linked syndromic, Nascimento-type, 300860
UBE2T	100%	99,90%	100%	100%	Fanconi anemia, complementation group T, 616435
UBE3A	99,10%	94,80%	100%	100%	Angelman syndrome, 105830
UBE3B	100%	99,90%	100%	100%	Kaufman oculocerebrofacial syndrome, 244450
UBIAD1	99,50%	96,00%	100%	100%	Corneal dystrophy, Schnyder type, 121800

UBQLN2	100%	99,40%	100%	100%	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857
UBR1	99,90%	99,10%	98,00%	98,00%	Johanson-Blizzard syndrome, 243800
UBTF	100%	99,40%	100%	100%	Neurodegeneration, childhood-onset, with brain atrophy, 617672
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
UFSP2	100%	99,60%	100%	100%	?Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974 ?Hip dysplasia, Beukes type, 142669
UGT1A1	100%	100%	100%	100%	Hyperbilirubinemia, familial transient neonatal, 237900 Crigler-Najjar syndrome, type I, 218800 Crigler-Najjar syndrome, type II, 606785
UMOD	97,70%	96,20%	100%	100%	Glomerulocystic kidney disease with hyperuricemia and isosthenuria, 609886 Medullary cystic kidney disease 2, 603860 Hyperuricemic nephropathy, familial juvenile 1, 162000
UMPS	100%	99,40%	97,00%	97,00%	Orotic aciduria, 258900
UNC119	100%	99,70%	100%	100%	?Immunodeficiency 13, 615518 ?Cone-rod dystrophy, 0
UNC13D	99,70%	98,10%	100%	100%	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC45B	99,30%	98,00%	100%	100%	?Cataract 43, 616279
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
UPF3B	92,20%	84,10%	100%	100%	Mental retardation, X-linked, syndromic 14, 300676
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
USP27X	100%	100%	100%	100%	Mental retardation, X-linked 105, 300984
USP45	99,60%	98,10%	100%	100%	?Leber congenital amaurosis 19, 618513
USP8	96,90%	87,80%	100%	100%	Pituitary adenoma 4, ACTH-secreting, somatic, 219090
USP9X	98,20%	92,90%	100%	100%	Mental retardation, X-linked 99, 300919 Mental retardation, X-linked 99, syndromic, female-restricted, 300968
USP9Y	48,60%	43,20%	60,00%	60,00%	Spermatogenic failure, Y-linked, 2, 415000
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
VANGL1	100%	100%	100%	100%	Caudal regression syndrome, 600145
VANGL2	99,90%	99,00%	100%	100%	Neural tube defects, 182940
VAPB	100%	99,90%	100%	100%	Spinal muscular atrophy, late-onset, Finkel type, 182980 Amyotrophic lateral sclerosis 8, 608627
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
VCAN	100%	100%	100%	100%	Wagner syndrome 1, 143200
VCL	99,90%	99,00%	100%	100%	Cardiomyopathy, dilated, 1W, 611407 Cardiomyopathy, hypertrophic, 15, 613255
VCP	100%	99,20%	100%	100%	Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320 Charcot-Marie-Tooth disease, type 2Y, 616687 Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954
VDR	99,90%	99,00%	100%	100%	Rickets, vitamin D-resistant, type IIA, 277440
VEGFC	100%	100%	100%	100%	Lymphatic malformation 4, 615907
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
VIM	99,30%	97,00%	100%	100%	Cataract 30, pulverulent, 116300
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

VMA21	99,00%	94,60%	100%	98,60%	Myopathy, X-linked, with excessive autophagy, 310440
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
VSX1	84,70%	80,50%	100%	100%	Keratoconus 1, 148300 ?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195
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
WAC	100%	99,70%	100%	100%	Desanto-Shinawi syndrome, 616708
WARS	99,80%	98,30%	100%	100%	Neuronopathy, distal hereditary motor, type IX, 617721
WARS2	100%	99,40%	100%	100%	Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710
WAS	95,90%	85,30%	100%	99,80%	Thrombocytopenia, X-linked, intermittent, 313900 Thrombocytopenia, X-linked, 313900 Wiskott-Aldrich syndrome, 301000 Neutropenia, severe congenital, X-linked, 300299
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
WDFY3	100%	99,60%	100%	100%	?Microcephaly 18, primary, autosomal dominant, 617520
WDPCP	98,20%	94,40%	98,10%	98,10%	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR11	98,00%	96,50%	100%	100%	Hypogonadotropic hypogonadism 14 with or without anosmia, 614858

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
WDR26	99,00%	96,60%	100%	100%	Skraban-Deardorff syndrome, 617616
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
WDR36	99,70%	97,60%	100%	100%	Glaucoma 1, open angle, G, 609887
WDR37	100%	99,60%	100%	100%	Neurooculocardiogenitourinary syndrome, 618652
WDR4	100%	100%	100%	100%	Microcephaly, growth deficiency, seizures, and brain malformations, 618346 Galloway-Mowat syndrome 6, 618347
WDR45	96,40%	89,70%	100%	100%	Neurodegeneration with brain iron accumulation 5, 300894
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
WDR66	100%	100%	100%	100%	Spermatogenic failure 33, 618152
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
WNK4	99,90%	99,30%	100%	100%	Pseudohypoaldosteronism, type IIB, 614491
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
WNT2B	98,00%	91,30%	100%	100%	Diarrhea 9, 618168
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
WNT5A	100%	100%	100%	100%	Robinow syndrome, autosomal dominant 1, 180700
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
WT1	99,90%	98,30%	100%	100%	Mesothelioma, somatic, 156240 Wilms tumor, type 1, 194070 Frasier syndrome, 136680 Denys-Drash syndrome, 194080 Meacham syndrome, 608978 Nephrotic syndrome, type 4, 256370
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
XIAP	93,00%	88,80%	100%	100%	Lymphoproliferative syndrome, X-linked, 2, 300635
XIST	NC	NC	NC	NC	X-inactivation, familial skewed, 300087
XK	99,80%	98,10%	100%	100%	McLeod syndrome with or without chronic granulomatous disease, 300842
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
XPR1	100%	99,90%	100%	100%	Basal ganglia calcification, idiopathic, 6, 616413
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
YAP1	96,40%	89,40%	100%	100%	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433
YARS	100%	99,90%	100%	100%	Charcot-Marie-Tooth disease, dominant intermediate C, 608323

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
YWHAG	100%	100%	100%	100%	Epileptic encephalopathy, early infantile, 56, 617665
YY1	100%	99,80%	100%	100%	Gabriele-de Vries syndrome, 617557
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
ZBTB18	100%	99,90%	100%	99,80%	Mental retardation, autosomal dominant 22, 612337
ZBTB20	100%	100%	100%	100%	PriINTELLECTUAL DISABILITYose syndrome, 259050
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
ZC4H2	100%	99,00%	100%	100%	Wieacker-Wolff syndrome, 314580 Wieacker-Wolff syndrome, female-restricted, 301041
ZDHHC9	99,90%	93,80%	100%	100%	Mental retardation, X-linked syndromic, Raymond type, 300799
ZEB1	100%	99,40%	100%	100%	Corneal dystrophy, posterior polymorphous, 3, 609141 Corneal dystrophy, Fuchs endothelial, 6, 613270
ZEB2	99,90%	99,10%	97,40%	97,40%	Mowat-Wilson syndrome, 235730
ZFHX2	100%	99,60%	100%	100%	?Marsili syndrome, 147430
ZFHX3	100%	99,60%	100%	100%	Prostate cancer, somatic, 176807
ZFP57	100%	99,80%	100%	100%	Diabetes mellitus, transient neonatal, 1, 601410
ZFPM2	100%	100%	100%	100%	46XY sex reversal 9, 616067 Diaphragmatic hernia 3, 610187 Tetralogy of Fallot, 187500
ZFYVE26	100%	99,10%	100%	100%	Spastic paraplegia 15, autosomal recessive, 270700
ZFYVE27	100%	100%	100%	100%	Spastic paraplegia 33, autosomal dominant, 610244
ZIC1	100%	100%	100%	100%	Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736 ?Craniosynostosis 6, 616602
ZIC2	100%	98,70%	98,50%	95,70%	Holoprosencephaly 5, 609637
ZIC3	100%	99,90%	100%	100%	Congenital heart defects, nonsyndromic, 1, X-linked, 306955 Heterotaxy, visceral, 1, X-linked, 306955 VACTERL association, X-linked, 314390
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
ZMYND11	100%	99,60%	100%	100%	Mental retardation, autosomal dominant 30, 616083
ZMYND15	100%	99,40%	100%	100%	?Spermatogenic failure 14, 615842
ZNF141	100%	100%	100%	100%	?Polydactyly, postaxial, type A6, 615226
ZNF142	100%	99,90%	100%	100%	Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425
ZNF148	99,90%	99,60%	100%	100%	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260
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
ZNF462	100%	99,90%	100%	100%	Weiss-Kruszka syndrome, 618619
ZNF469	100%	100%	100%	100%	Brittle cornea syndrome 1, 229200
ZNF513	100%	100%	100%	100%	?Retinitis pigmentosa 58, 613617
ZNF644	100%	100%	100%	100%	Myopia 21, autosomal dominant, 614167
ZNF687	100%	100%	100%	100%	Paget disease of bone 6, 616833
ZNF711	99,80%	98,20%	100%	100%	Mental retardation, X-linked 97, 300803
ZNF750	100%	100%	100%	100%	Seborrhea-like dermatitis with psoriasiform elements, 610227
ZNHIT3	74,40%	74,40%	74,60%	74,40%	PEHO syndrome, 260565
ZP1	100%	100%	100%	100%	Oocyte maturation defect 1, 615774
ZP2	99,80%	98,40%	100%	100%	Oocyte maturation defect 6, 618353
ZP3	100%	100%	100%	100%	Oocyte maturation defect 3, 617712
ZSWIM6	95,50%	91,90%	94,90%	92,10%	Acromelic frontonasal dysostosis, 603671 Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.
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