MULTIPLE CONGENITAL ANOMALIES GENE PANEL DGD20062014

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM ID
A4GALT	121,2	100%	100%	[Blood group, P1Pk system, p phenotype], 111400 [Blood group, P1Pk system, P(2) phenotype], 111400 NOR polyagglutination syndrome, 111400
AAAS	95	100%	100%	Achalasia-addisonianism-alacrimia syndrome, 231550
AAGAB	109,9	100%	95%	Keratoderma, palmoplantar, punctate type IA, 148600
AARS	91,8	97%	94%	Charcot-Marie-Tooth disease, axonal, type 2N, 613287
AARS2	87,9	100%	98%	Combined oxidative phosphorylation deficiency 8, 614096
AASS	100	100%	98%	Hyperlysinemia, 238700 Saccharopinuria, 268700 (1)
ABAT	64,9	98%	90%	GABA-transaminase deficiency, 613163
ABCA1	88,1	100%	98%	Tangier disease, 205400 HDL deficiency, type 2, 604091 {Coronary artery disease in familial hypercholesterolemia, protection against}, 143890
ABCA12	101	100%	99%	Ichthyosis, congenital, autosomal recessive 4A, 601277 Ichthyosis, autosomal recessive 4B (harlequin), 242500
ABCA3	90,8	100%	96%	Surfactant metabolism dysfunction, pulmonary, 3, 610921
ABCA4	86,5	99%	94%	Stargardt disease 1, 248200 Retinitis pigmentosa 19, 601718 Cone-rod dystrophy 3, 604116 Macular degeneration, age-related, 2, 153800 Fundus flavimaculatus, 248200 Retinal dystrophy, early-onset severe, 248200
ABCB11	93,3	100%	98%	Cholestasis, progressive familial intrahepatic 2, 601847 Cholestasis, benign recurrent intrahepatic, 2, 605479

ABCB4	91,3	97%	94%	Cholestasis, progressive familial intrahepatic 3, 602347 Cholestasis, intrahepatic, of pregnancy, 3, 614972 Gallbladder disease 1, 600803
ABCB6	115,8	100%	100%	Microphthalmia, isolated, with coloboma 7, 614497 [Blood group, Langereis system], 111600 Dyschromatosis universalis hereditaria 3, 615402
ABCB7	121,9	100%	100%	Anemia, sideroblastic, with ataxia, 301310
ABCC2	98,9	100%	99%	Dubin-Johnson syndrome, 237500
ABCC6	50,4	70%	67%	Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850 Arterial calcification, generalized, of infancy, 2, 614473
ABCC8	86,2	99%	96%	Hyperinsulinemic hypoglycemia, familial, 1, 256450 Hypoglycemia of infancy, leucine-sensitive, 240800 Diabetes mellitus, transient neonatal 2, 610374 Diabetes mellitus, noninsulin-dependent, 125853 Diabetes mellitus, permanent neonatal, 606176
ABCC9	109,8	100%	99%	Cardiomyopathy, dilated, 1O, 608569 Atrial fibrillation, familial, 12, 614050 Hypertrichotic osteochondrodysplasia, 239850
ABCD1	58,2	73%	73%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABCD4	100,8	100%	99%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	117,1	98%	94%	Sitosterolemia, 210250
ABCG8	90,3	96%	95%	Sitosterolemia, 210250 Gallbladder disease 4, 611465
ABHD12	60,9	100%	87%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ABHD5	116,3	100%	95%	Chanarin-Dorfman syndrome, 275630
ABL1	100,6	99%	95%	Leukemia, Philadelphia chromosome-positive, resistant to imatinib
ACAD8	103,7	100%	100%	Isobutyryl-CoA dehydrogenase deficiency, 611283

ACAD9	90,8	100%	100%	ACAD9 deficiency, 611126
ACADM	144,9	100%	100%	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	101,2	100%	98%	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	78,8	98%	95%	2-methylbutyrylglycinuria, 610006
ACADVL	88,3	100%	95%	VLCAD deficiency, 201475
ACAN	106,7	94%	90%	Spondyloepiphyseal dysplasia, Kimberley type, 608361 Spondyloepimetaphyseal dysplasia, aggrecan type, 612813 Osteochondritis dissecans, short stature, and early-onset osteoarthritis, 165800
ACAT1	106,8	100%	98%	Alpha-methylacetoacetic aciduria, 203750
ACE	91,1	98%	93%	{Myocardial infarction, susceptibility to} {Alzheimer disease, susceptibility to}, 104300 {Microvascular complications of diabetes 3}, 612624 [Angiotensin I-converting enzyme, benign serum increase] {SARS, progression of} Renal tubular dysgenesis, 267430 {Stroke, hemorrhagic}, 614519
ACO2	77,5	89%	82%	Infantile cerebellar-retinal degeneration, 614559
ACOX1	77,7	97%	94%	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACP5	98,7	100%	100%	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACSF3	82,9	100%	100%	Combined malonic and methylmalonic aciduria, 614265
ACSL4	129,4	100%	98%	Mental retardation, X-linked 63, 300387
ACSL6	81,8	99%	99%	Myelodysplastic syndrome Myelogenous leukemia, acute
ACTA1	69,7	100%	87%	Nemaline myopathy 3, autosomal dominant or recessive, 161800 Myopathy, actin, congenital, with excess of thin myofilaments, 161800 Myopathy, actin, congenital, with cores, 161800 Myopathy, congenital, with fiber-type disproportion 1, 255310

ACTA2	82,9	100%	99%	Aortic aneurysm, familial thoracic 6, 611788 Multisystemic smooth muscle dysfunction syndrome, 613834 Moyamoya disease 5, 614042
ACTB	61,8	100%	95%	Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACTC1	72,9	100%	93%	Cardiomyopathy, dilated, 1R, 613424 Cardiomyopathy, familial hypertrophic, 11, 612098 Atrial septal defect 5, 612794 Left ventricular noncompaction 4, 613424
ACTG1	60,2	97%	87%	Deafness, autosomal dominant 20/26, 604717 Baraitser-Winter syndrome 2, 614583
ACTN1	99,2	100%	98%	Bleeding disorder, platelet-type, 15, 615193
ACTN4	92,4	98%	92%	Glomerulosclerosis, focal segmental, 1, 603278
ACVR1	92,6	100%	98%	Fibrodysplasia ossificans progressiva, 135100
ACVR1B	94	94%	91%	Pancreatic cancer, somatic
ACVR2B	77,9	96%	96%	Heterotaxy, visceral, 4, autosomal, 613751
ACVRL1	52,9	95%	89%	Telangiectasia, hereditary hemorrhagic, type 2, 600376
ACY1	85,1	100%	96%	Aminoacylase 1 deficiency, 609924
ADA	72,3	100%	96%	Severe combined immunodeficiency due to ADA deficiency, 102700 Adenosine deaminase deficiency, partial, 102700
ADAM10	116,9	100%	100%	Reticulate acropigmentation of Kitamura, 615537 {Alzheimer disease 18, susceptibility to}
ADAM17	111,7	99%	97%	Inflammatory skin and bowel disease, neonatal, 614328
ADAM9	107,5	100%	99%	Cone-rod dystrophy 9, 612775
ADAMTS10	74,1	97%	92%	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS13	52,9	94%	81%	Thrombotic thrombocytopenic purpura, familial, 274150

ADAMTS17	73,9	92%	81%	Weill-Marchesani-like syndrome, 613195
ADAMTS18	92,3	100%	96%	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
ADAMTS2	95,5	98%	92%	Ehlers-Danlos syndrome, type VIIC, 225410
ADAMTSL2	68	96%	86%	Geleophysic dysplasia 1, 231050
ADAMTSL4	95,3	100%	98%	Ectopia lentis, isolated, autosomal recessive, 225100 Ectopia lentis et pupillae, 225200
ADAR	131	99%	98%	Dyschromatosis symmetrica hereditaria, 127400 Aicardi-Goutieres syndrome 6, 615010
ADAT3	54,1	100%	94%	Mental retardation, autosomal recessive 36, 615286
ADCY5	83,9	99%	94%	Dyskinesia, familial, with facial myokymia, 606703
ADIPOQ	154,3	100%	100%	Adiponectin deficiency, 612556
ADK	113,5	94%	94%	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADRB2	134,5	100%	100%	{Asthma, nocturnal, susceptibility to}, 600807 {Obesity, susceptibility to}, 601665 Beta-2-adrenoreceptor agonist, reduced response to
ADSL	126,2	100%	99%	ade(-)I bifunctional Adenylosuccinase deficiency, 103050
AFF2	135	99%	99%	Mental retardation, X-linked, FRAXE type, 309548
AFG3L2	77,3	95%	92%	Spinocerebellar ataxia 28, 610246 Ataxia, spastic, 5, autosomal recessive, 614487
AGA	111,6	100%	91%	Aspartylglucosaminuria
AGK	104,1	100%	100%	Sengers syndrome, 212350 Cataract 38, autosomal recessive, 614691
AGL	137,7	100%	100%	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGPAT2	65,7	92%	88%	Lipodystrophy, congenital generalized, type 1, 608594

AGPS	104,3	100%	100%	Rhizomelic chondrodysplasia punctata, type 3, 600121
AGRN	85,3	98%	90%	Myasthenia, limb-girdle, familial, 254300
AGT	130,9	100%	100%	{Hypertension, essential, susceptibility to}, 145500 {Preeclampsia, susceptibility to} Renal tubular dysgenesis, 267430
AGTR1	155,7	97%	97%	Hypertension, essential, 145500 Renal tubular dysgenesis, 267430
AGXT	84,9	98%	92%	Hyperoxaluria, primary, type 1, 259900
AHCY	76,9	91%	69%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHI1	105,7	99%	98%	Joubert syndrome-3, 608629
AICDA	80,4	100%	96%	Immunodeficiency with hyper-IgM, type 2, 605258
AIFM1	109,8	100%	99%	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490
AIMP1	117,9	100%	100%	Leukodystrophy, hypomyelinating, 3, 260600
AIP	101,7	96%	93%	Pituitary adenoma, growth hormone-secreting, 102200 Pituitary adenoma, prolactin-secreting, 600634 Pituitary adenoma, ACTH-secreting, 219090
AIPL1	90	100%	100%	Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393 Cone-rod dystrophy, 604393
AIRE	71,1	98%	87%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AK1	91,4	100%	99%	Hemolytic anemia due to adenylate kinase deficiency, 612631
AK2	70,8	78%	74%	Reticular dysgenesis, 267500
AKAP9	120,7	100%	99%	Long QT syndrome-11, 611820
AKR1C2	73,9	92%	82%	Obesity, hyperphagia, and developmental delay 46XY sex reversal 8, 614279

AKR1D1	96,2	100%	100%	Bile acid synthesis defect, congenital, 2, 235555
AKT1	118,5	99%	98%	IGH Breast cancer, somatic, 114480 Colorectal cancer, somatic, 114500 Ovarian cancer, somatic, 167000 {Schizophrenia, susceptibility to}, 181500 (2) Proteus syndrome, somatic, 176920 Cowden syndrome 6, 615109
AKT2	124,5	98%	95%	Diabetes mellitus, type II, 125853 Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900
AKT3	109,1	100%	100%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome, 603387
ALAD	91,9	96%	90%	Porphyria, acute hepatic, 612740 {Lead poisoning, susceptibility to}, 612740
ALAS2	84,4	92%	88%	Anemia, sideroblastic, X-linked, 300751 Protoporphyria, erythropoietic, X-linked, 300752
ALB	103,7	100%	99%	Analbuminemia [Dysalbuminemic hyperthyroxinemia] [Dysalbuminemic hyperzincemia], 194470 (1)
ALDH18A1	97,7	99%	95%	Cutis laxa, autosomal recessive, type IIIA, 219150
ALDH1A3	83,5	94%	93%	Microphthalmia, isolated 8, 615113
ALDH2	85,1	98%	93%	Alcohol sensitivity, acute, 610251 {Hangover, susceptibility to}, 610251 {Sublingual nitroglycerin, susceptibility to poor response to} {Esophageal cancer, alcohol-related, susceptibility to}
ALDH3A2	92,1	100%	100%	Sjogren-Larsson syndrome, 270200
ALDH4A1	70,7	93%	90%	Hyperprolinemia, type II, 239510
ALDH5A1	61,9	97%	90%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	96,6	100%	99%	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	63,3	91%	86%	Epilepsy, pyridoxine-dependent, 266100

ALDOA	105,2	98%	94%	Glycogen storage disease XII, 611881
ALDOB	117,9	100%	99%	Fructose intolerance, 229600
ALG1	45,6	45%	45%	Congenital disorder of glycosylation, type Ik, 608540
ALG11	140,7	100%	100%	Congenital disorder of glycosylation, type Ip, 613661
ALG12	93,4	100%	97%	Congenital disorder of glycosylation, type Ig, 607143
ALG13	113	96%	95%	Congenital disorder of glycosylation, type Is, 300884
ALG2	103,1	99%	89%	Congenital disorder of glycosylation, type Ii, 607906
ALG3	81,3	98%	89%	Congenital disorder of glycosylation, type Id, 601110
ALG6	92,5	100%	99%	Congenital disorder of glycosylation, type Ic, 603147
ALG8	84,7	97%	95%	Congenital disorder of glycosylation, type Ih, 608104
ALG9	80,7	99%	98%	Congenital disorder of glycosylation, type Il, 608776
ALMS1	190,1	98%	98%	Alstrom syndrome, 203800
ALOX12B	96	100%	99%	Ichthyosis, congenital, autosomal recessive 2, 242100
ALOXE3	83,6	100%	98%	Ichthyosis, congenital, autosomal recessive 3, 606545
ALPL	81,6	100%	97%	Hypophosphatasia, infantile, 241500 Hypophosphatasia, childhood, 241510 Odontohypophosphatasia, 146300 Hypophosphatasia, adult, 146300
ALS2	130,1	99%	97%	Amyotrophic lateral sclerosis 2, juvenile, 205100 Primary lateral sclerosis, juvenile, 606353 Spastic paralysis, infantile onset ascending, 607225
ALX1	151,9	100%	100%	Frontonasal dysplasia 3, 613456
ALX3	77,1	86%	77%	Frontonasal dysplasia 1, 136760

ALX4	72	100%	99%	Parietal foramina 2, 609597 Frontonasal dysplasia 2, 613451
AMACR	80,9	100%	100%	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
AMELX	118,9	100%	100%	Amelogenesis imperfecta, hypoplastic/hypomaturation type 1E, 301200 -3
AMH	27,9	77%	69%	Persistent Mullerian duct syndrome, type I, 261550
AMHR2	125	100%	100%	Persistent Mullerian duct syndrome, type II, 261550
AMN	67,1	92%	84%	Megaloblastic anemia-1, Norwegian type, 261100
AMPD1	100,1	100%	98%	Myoadenylate deaminase deficiency
AMT	127,9	100%	100%	Glycine encephalopathy, 605899
ANG	162,6	100%	96%	Amyotrophic lateral sclerosis 9, 611895
ANGPTL3	108,6	100%	100%	Hypobetalipoproteinemia, familial, 2, 605019
ANK1	93,9	99%	95%	Spherocytosis, type 1, 182900
ANK2	125,1	100%	99%	Long QT syndrome-4, 600919 Cardiac arrhythmia, ankyrin-B-related, 600919
ANKH	105,9	100%	100%	Craniometaphyseal dysplasia, 123000 Chondrocalcinosis 2, 118600
ANKK1	94,5	100%	98%	Dopamine receptor D2, reduced brain density of
ANKRD11	105,7	91%	86%	KBG syndrome, 148050
ANKRD26	110,9	99%	98%	Thrombocytopenia 2, 188000
ANKS6	59,2	91%	82%	Nephronophthisis 16, 615382
ANO10	99,7	100%	98%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANO3	107,2	99%	98%	Dystonia 24, 615034

ANO5	98,2	100%	99%	Gnathodiaphyseal dysplasia, 166260 Muscular dystrophy, limb-girdle, type 2L, 611307 Miyoshi muscular dystrophy 3, 613319
ANO6	93,5	98%	95%	Scott syndrome, 262890
ANTXR1	75,6	96%	90%	{Hemangioma, capillary infantile, susceptibility to}, 602089 GAPO syndrome, 230740
ANTXR2	115,4	100%	100%	Hyaline fibromatosis syndrome, 228600
AP1S1	65,2	99%	88%	MEDNIK syndrome, 609313
AP1S2	171,4	100%	100%	Mental retardation, X-linked syndromic, Fried type, 300630
AP2S1	89,2	100%	100%	Hypocalciuric hypercalcemia, familial, type III, 600740
AP3B1	104,8	100%	99%	Hermansky-Pudlak syndrome 2, 608233
AP4B1	101,9	100%	100%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	121,8	100%	99%	Spastic paraplegia 51, autosomal recessive, 613744
AP4M1	103,3	100%	99%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	67,4	94%	87%	Spastic paraplegia 52, autosomal recessive, 614067
AP5Z1	75,7	97%	87%	Spastic paraplegia 48, autosomal recessive, 613647
APC	149,1	100%	99%	Adenomatous polyposis coli, 175100 Gastric cancer, somatic, 613659 Adenoma, periampullary, somatic Hepatoblastoma, somatic, 114550 Desmoid disease, hereditary, 135290 Colorectal cancer, somatic, 114500 Brain tumor-polyposis syndrome 2, 175100 Gardner syndrome, 175100
APCDD1	119,5	100%	100%	Hypotrichosis simplex, 605389

APOA1	71,6	100%	93%	ApoA-I and apoC-III deficiency, combined Hypoalphalipoproteinemia, 604091 Corneal clouding, autosomal recessive Amyloidosis, 3 or more types, 105200
APOA2	86	100%	100%	Apolipoprotein A-II deficiency {Hypercholesterolemia, familial, modification of}, 143890
APOA5	127,8	100%	100%	{Hypertriglyceridemia, susceptibility to}, 145750 Hyperchylomicronemia, late-onset, 144650
APOB	154,2	99%	99%	Ag linked Hypobetalipoproteinemia Hypobetalipoproteinemia, normotriglyceridemic Hypercholesterolemia, due to ligand-defective apo B, 144010
APOC2	168,8	100%	100%	Hyperlipoproteinemia, type Ib, 207750
APOC3	108	100%	100%	Hyperalphalipoproteinemia 2, 614028
APOE	39,2	82%	68%	Hyperlipoproteinemia, type III {Myocardial infarction susceptibility} Sea-blue histiocyte disease, 269600 Alzheimer disease-2, 104310 {?Macular degeneration, age-related}, 603075 Lipoprotein glomerulopathy, 611771
APOE	39,2	82%	68%	{Myocardial infarction, susceptibility to}, 608446
APP	85,4	100%	98%	Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants, 605714 Alzheimer disease 1, familial, 104300
APRT	49,4	100%	94%	Adenine phosphoribosyltransferase deficiency, 614723
APTX	115,6	100%	94%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
AQP2	85,3	96%	90%	Diabetes insipidus, nephrogenic, 125800
AQP5	94,1	100%	94%	Palmoplantar keratoderma, Bothnian type, 600231

AR	99,2	100%	99%	Androgen insensitivity, 300068 Spinal and bulbar muscular atrophy of Kennedy, 313200 Androgen insensitivity, partial, with or without breast cancer, 312300 {Prostate cancer, susceptibility to}, 176807 Hypospadias 1, X-linked, 300633
ARFGEF2	104,1	100%	99%	Periventricular heterotopia with microcephaly, 608097
ARG1	121,9	98%	90%	Argininemia, 207800
ARHGAP26	117,4	100%	100%	Leukemia, juvenile myelomonocytic, 607785
ARHGAP31	135,6	100%	98%	Adams-Oliver syndrome 1, 100300
ARHGEF10	85	98%	92%	Slowed nerve conduction velocity, AD, 608236
ARHGEF12	118,5	100%	99%	Leukemia, acute myeloid, 601626
ARHGEF6	109,6	99%	98%	Mental retardation, X-linked 46, 300436
ARHGEF9	91,3	99%	95%	Epileptic encephalopathy, early infantile, 8, 300607
ARID1A	102,4	99%	96%	Mental retardation, autosomal dominant 14, 614607
ARID1B	115,7	99%	97%	Mental retardation, autosomal dominant 12, 614562
ARL13B	127,7	100%	98%	Joubert syndrome 8, 612291
ARL2BP	85	100%	96%	Retinitis pigmentosa with or without situs inversus, 615434
ARL6	134,1	100%	100%	Bardet-Biedl syndrome 3, 209900 {Bardet-Biedl syndrome 1, modifier of}, 209900 Retinitis pigmentosa 55, 613575
ARMC4	87,7	87%	86%	Ciliary dyskinesia, primary, 23, 615451
ARNT	76,4	97%	92%	Leukemia, acute myeloblastic
ARSA	78,2	97%	94%	Metachromatic leukodystrophy, 250100

ARSB	90,7	100%	97%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARSE	84,7	95%	86%	Chondrodysplasia punctata, X-linked recessive, 302950
ARX	60,5	80%	73%	Epileptic encephalopathy, early infantile, 1, 308350 Lissencephaly, X-linked 2, 300215 Mental retardation, X-linked 29 and others, 300419 Proud syndrome, 300004 Partington syndrome, 309510 Hydranencephaly with abnormal genitalia, 300215
ASAH1	93,5	100%	100%	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASB10	69,6	95%	87%	Glaucoma 1, open angle, F, 603383
ASCC1	95,4	99%	92%	Barrett esophagus/esophageal adenocarcinoma, 614266
ASCL1	144,8	100%	100%	Central hypoventilation syndrome, congenital, 209880 Haddad syndrome, 209880
ASL	72,9	97%	93%	Argininosuccinic aciduria, 207900
ASNS	55,1	93%	81%	temperature sensitive G1 mutant
ASPA	96	100%	100%	Canavan disease, 271900
ASPM	134,6	100%	99%	Microcephaly 5, primary, autosomal recessive, 608716
ASPSCR1	79,4	98%	89%	Alveolar soft-part sarcoma, 606243
ASS1	41,5	86%	54%	Citrullinemia, 215700
ASXL1	142	98%	97%	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ASXL3	148,6	100%	99%	No OMIM phenotype
ATCAY	94,1	100%	98%	Ataxia, cerebellar, Cayman type, 601238
ATIC	109,3	100%	97%	AICA-ribosiduria due to ATIC deficiency, 608688

ATL1	105	100%	100%	Spastic paraplegia 3A, autosomal dominant, 182600 Neuropathy, hereditary sensory, type ID, 613708
ATL3	97,2	99%	98%	Bainbridge-Ropers syndrome, 615485
ATM	111,1	99%	99%	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic {Breast cancer, susceptibility to}, 114480 Lymphoma, mantle cell T-cell prolymphocytic leukemia, somatic
ATN1	119,5	97%	96%	Dentatorubro-pallidoluysian atrophy, 125370
ATP13A2	78,8	99%	94%	Parkinson disease 9, 606693
ATP1A2	100,9	100%	98%	Migraine, familial hemiplegic, 2, 602481 Alternating hemiplegia of childhood, 104290 Migraine, familial basilar, 602481
ATP1A3	108,3	100%	98%	Dystonia-12, 128235 Alternating hemiplegia of childhood 2, 614820
ATP2A1	119,9	100%	98%	Brody myopathy, 601003
ATP2A2	115,4	100%	100%	Darier disease, 124200 Acrokeratosis verruciformis, 101900
ATP2C1	113,4	100%	99%	Hailey-Hailey disease, 169600
ATP5E	149	100%	100%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053
ATP6V0A2	97,9	100%	99%	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250
ATP6V0A4	79,5	97%	91%	Renal tubular acidosis, distal, autosomal recessive, 602722
ATP7A	122,6	100%	100%	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATP7B	122,5	99%	97%	Wilson disease, 277900

ATP8B1	104,8	98%	97%	Cholestasis, progressive familial intrahepatic 1, 211600 Cholestasis, benign recurrent intrahepatic, 243300 Cholestasis, intrahepatic, of pregnancy, 1, 147480
ATPAF2	66,4	100%	100%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
ATR	113,1	100%	99%	Seckel syndrome 1, 210600 Cutaneous telangiectasia and cancer syndrome, familial, 614564
ATRX	136,1	100%	100%	Alpha-thalassemia/mental retardation syndrome, 301040 Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Mental retardation-hypotonic facies syndrome, X-linked, 309580
ATXN1	103,8	100%	100%	Spinocerebellar ataxia 1, 164400
ATXN10	116,9	100%	100%	Spinocerebellar ataxia 10, 603516
ATXN2	88,5	87%	81%	Spinocerebellar ataxia 2, 183090 {Amyotrophic lateral sclerosis, susceptibility to, 13}, 183090
ATXN3	122,8	100%	99%	Machado-Joseph disease, 109150
ATXN7	131,4	98%	93%	Spinocerebellar ataxia 7, 164500
AUH	111,3	100%	100%	3-methylglutaconic aciduria, type I, 250950
AVP	44,4	94%	78%	Diabetes insipidus, neurohypophyseal, 125700
AVPR2	96,4	99%	95%	Diabetes insipidus, nephrogenic, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539
AXIN1	120,1	98%	91%	Hepatocellular carcinoma, somatic, 114550 Caudal duplication anomaly, 607864
AXIN2	95,6	97%	91%	Oligodontia-colorectal cancer syndrome, 608615 Colorectal cancer, somatic, 114500
B2M	166,1	100%	100%	Hypoproteinemia, hypercatabolic, 241600
B3GALT6	56,2	79%	73%	Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640 Ehlers-Danlos syndrome, progeroid type, 2, 615349
B3GALTL	108,3	100%	96%	Peters-plus syndrome, 261540

B3GAT3	55,2	91%	82%	Multiple joint dislocations, short stature, craniofacial dysmorphism, and congenital heart defects, 245600
B3GNT1	95,8	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
B4GALNT1	82	89%	82%	Spastic paraplegia 26, autosomal recessive, 609195
B4GALT1	80,7	100%	100%	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	77,9	100%	96%	Ehlers-Danlos syndrome, progeroid type, 1, 130070
B9D1	91,4	100%	93%	Meckel syndrome 9, 614209
B9D2	50,8	100%	100%	Meckel syndrome 10, 614175
BAAT	121	99%	96%	Hypercholanemia, familial, 607748
BAG3	151,4	100%	100%	Myopathy, myofibrillar, 6, 612954 Cardiomyopathy, dilated, 1HH, 613881
BANF1	44,6	55%	54%	Nestor-Guillermo progeria syndrome, 614008
BAP1	89,6	99%	97%	Tumor predisposition syndrome, 614327
BAX	84,2	97%	95%	Colorectal cancer T-cell acute lymphoblastic leukemia
BBS1	119,3	100%	99%	Bardet-Biedl syndrome 1, 209900
BBS10	121	100%	100%	Bardet-Biedl syndrome 10, 209900
BBS12	143,1	100%	100%	Bardet-Biedl syndrome 12, 209900
BBS2	113,4	100%	100%	Bardet-Biedl syndrome 2, 209900
BBS4	85,8	100%	96%	Bardet-Biedl syndrome 4, 209900
BBS5	130,3	100%	100%	Bardet-Biedl syndrome 5, 209900
BBS7	119,3	100%	99%	Bardet-Biedl syndrome 7, 209900

BCHE	141,8	100%	100%	Apnea, postanesthetic
BCKDHA	100,9	100%	99%	Maple syrup urine disease, type Ia, 248600
BCKDHB	97,7	99%	85%	Maple syrup urine disease, type Ib, 248600
BCKDK	130,9	100%	100%	Branched-chain ketoacid dehydrogenase kinase deficiency, 614923
BCL10	90	99%	92%	Lymphoma, MALT, somatic, 137245 {Lymphoma, follicular, somatic}, 613024 {Male germ cell tumor, somatic}, 273300, {Sezary syndrome, somatic}, {Mesothelioma, somatic}, 156240
BCL2	150,4	99%	97%	Leukemia/lymphoma, B-cell, 2
BCL7A	67,3	99%	86%	B-cell non-Hodgkin lymphoma, high-grade
BCMO1	120,4	100%	98%	Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300
BCOR	124,5	100%	99%	Microphthalmia, syndromic 2, 300166
BCR	81,6	84%	82%	Leukemia, chronic myeloid, 608232 Leukemia, acute lymphocytic, 613065
BCS1L	149,3	100%	100%	Mitochondrial complex III deficiency, nuclear type 1, 124000 Leigh syndrome, 256000 Bjornstad syndrome, 262000 GRACILE syndrome, 603358
BDNF	164,4	99%	96%	{Memory impairment, susceptibility to} Central hypoventilation syndrome, congenital, 209880 {Obsessive-compulsive disorder, protection against}, 164230 {Bulimia nervosa, age of onset of weight loss in},607499 {Anorexia nervosa, susceptibility to}, 610269

BEST1	116,1	100%	97%	Best macular dystrophy, 153700 Maculopathy, bull's-eye Vitelliform macular dystrophy, adult-onset, 608161 Bestrophinopathy, 611809 Vitreoretinochoroidopathy, 193220 Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma, 193220 Retinitis pigmentosa-50, 613194 Retinitis pigmentosa, concentric, 613194
BFSP1	124,5	100%	100%	Cataract 33, 611391
BFSP2	54,7	98%	91%	Cataract 12, multiple types, 611597
BICD2	89,6	98%	95%	Spinal muscular atrophy, lower extremity-predominant, 2, AD, 615290 -3
BIN1	54	92%	72%	Myopathy, centronuclear, autosomal recessive, 255200
BLK	119,5	100%	100%	Maturity-onset diabetes of the young, type 11, 613375
BLNK	102,4	100%	100%	Agammaglobulinemia 4, 613502
BLOC1S3	33,9	83%	73%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	114,4	96%	86%	Hermansky-pudlak syndrome 9, 614171
BLVRA	87,9	100%	100%	Hyperbiliverdinemia, 614156
BMP1	93,1	98%	95%	Osteogenesis imperfecta, type XIII, 614856
BMP15	148,6	100%	100%	Ovarian dysgenesis 2, 300510 Premature ovarian failure 4, 300510
BMP2	116,2	100%	99%	{HFE hemochromatosis, modifier of}, 235200 Brachydactyly, type A2, 112600
BMP4	115,4	100%	97%	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625 -3
BMPER	113,7	99%	97%	Diaphanospondylodysostosis, 608022

BMPR1A	59,2	79%	64%	Polyposis, juvenile intestinal, 174900 Polyposis syndrome, hereditary mixed, 2, 610069 Juvenile polyposis syndrome, infantile form, 174900
BMPR1B	109,7	100%	97%	Brachydactyly, type A2, 112600 Chrondrodysplasia, acromesomelic, with genital anomalies, 609441
BMPR2	145,6	100%	99%	Pulmonary hypertension, familial primary, 1, with or without HHT, 178600 Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600 Pulmonary venoocclusive disease, 265450
BOLA3	51,3	100%	99%	Multiple mitochondrial dysfunctions syndrome 2, 614299
BPGM	140,6	100%	100%	Erythrocytosis due to bisphosphoglycerate mutase deficiency, 222800
BRAF	72,6	100%	97%	Melanoma, malignant, somatic Colorectal cancer, somatic Adenocarcinoma of lung, somatic, 211980 Nonsmall cell lung cancer, somatic Cardiofaciocutaneous syndrome, 115150 Noonan syndrome 7, 613706 LEOPARD syndrome 3, 613707
BRAT1	70,2	100%	94%	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BRCA2	142,7	99%	99%	{Breast-ovarian cancer, familial, 2}, 612555 Fanconi anemia, complementation group D1, 605724 Prostate cancer, 176807 {Breast cancer, male, susceptibility to}, 114480 Wilms tumor, 194070 {Medulloblastoma}, 155255 {Glioblastoma 3}, 613029 {Pre-B-cell acute lymphoblastic leukemia} Pancreatic cancer, 613347
BRWD3	115,6	99%	99%	Mental retardation, X-linked 93, 300659
BSCL2	104,1	100%	100%	Lipodystrophy, congenital generalized, type 2, 269700 Silver spastic paraplegia syndrome, 270685 Neuropathy, distal hereditary motor, type V, 600794

BSND	105,3	100%	97%	Bartter syndrome, type 4a, 602522 Sensorineural deafness with mild renal dysfunction, 602522
BTD	132	100%	100%	Biotinidase deficiency, 253260
BTK	98,6	100%	99%	Agammaglobulinemia, X-linked 1, 300755 Agammaglobulinemia and isolated hormone deficiency, 307200
BUB1	103,1	98%	98%	Colorectal cancer with chromosomal instability
BUB1B	111,3	100%	99%	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430
C10orf11	90,2	100%	100%	Albinism, oculocutaneous, type VII, 615179
C10orf2	137,8	100%	100%	Progressive external ophthalmoplegia, autosomal dominant, 3, 609286 Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245
C12orf57	68,3	100%	96%	Temtamy syndrome, 218340
C12orf65	174,3	100%	100%	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035
C19orf12	78,9	100%	94%	Neurodegeneration with brain iron accumulation 4, 614298
C1GALT1C1	164,6	100%	100%	Tn polyagglutination syndrome, somatic, 300622
C1QA	119,1	98%	91%	C1q deficiency, 613652
C1QB	90,3	94%	87%	C1q deficiency, 613652
C1QC	121,9	84%	69%	C1q deficiency, 613652
C1QTNF5	90,4	88%	76%	Retinal degeneration, late-onset, autosomal dominant, 605670
C1S	95,5	99%	99%	C1s deficiency, 613783
C2	15,6	76%	27%	C2 deficiency, 217000 {Macular degeneration, age-related, reduced risk of}, 603075
C2orf71	103,7	98%	93%	Retinitis pigmentosa 54, 613428

C3	99,1	97%	93%	C3 deficiency, 613779 {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 {Macular degeneration, age-related, 9}, 611378
C4A	1,6	3%	2%	C4a deficiency, 614380 [Blood group, Rodgers], 614374 ?Systemic lupus erythematosus, susceptibility to or protection against}, 152700 (2)
C4B	1,6	4%	2%	C4B deficiency, 614379
C4orf26	124,7	100%	100%	Amelogenesis imperfecta, hypomaturation type, IIA4, 614832
C5	97	100%	99%	C5 deficiency, 609536
C5orf42	122,1	100%	99%	Joubert syndrome 17, 614615
C6	111,6	100%	99%	C6 deficiency, 612446 Combined C6/C7 deficiency
C7	90,4	99%	95%	C7 deficiency, 610102
C8A	77,2	100%	98%	C8 deficiency, type I, 613790
C8B	92,3	100%	96%	C8 deficiency, type II, 613789
C8orf37	92	100%	100%	Retinitis pigmentosa 64, 614500 Cone-rod dystrophy 16, 614500 -3
C9	109,2	100%	100%	C9 deficiency Macular degeneration, age-related, 15, susceptibility to
C9orf72	81,6	100%	100%	Amyotrophic lateral sclerosis and/or frontotemporal dementia, 105550 -3
CA12	81,6	100%	100%	Hyperchlorhidrosis, isolated, 143860
CA4	78,8	100%	97%	Retinitis pigmentosa 17, 600852
CA8	79,5	100%	100%	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CABP2	50,4	87%	70%	Deafness, autosomal recessive 93, 614899
CABP4	68,6	100%	99%	Night blindness, congenital stationary (incomplete), 2B, autosomal recessive, 610427

CACNA1A	80	98%	91%	Migraine, familial hemiplegic, 1, 141500 Episodic ataxia, type 2, 108500 Spinocerebellar ataxia 6, 183086 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500
CACNA1C	93,7	98%	96%	Timothy syndrome, 601005 Brugada syndrome 3, 611875
CACNA1D	113,9	100%	98%	Sinoatrial node dysfunction and deafness, 614896
CACNA1F	88,8	98%	96%	Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071 Cone-rod dystropy, X-linked, 3, 300476 Aland Island eye disease, 300600
CACNA1S	93,8	99%	98%	Hypokalemic periodic paralysis, type 1, 170400 {Malignant hyperthermia susceptibility 5}, 601887 {Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580
CACNA2D4	76,8	98%	92%	Retinal cone dystrophy 4, 610478
CACNB2	127,4	100%	99%	Brugada syndrome 4, 611876
CACNB4	91,3	100%	94%	{Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682 {Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 Episodic ataxia, type 5, 613855
CACNG2	106,8	100%	100%	Mental retardation, autosomal dominant 10, 614256
CALM1	103,3	100%	100%	Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916 -3
CALR	136,9	96%	92%	distal to C3, near LDLR
CALR3	89,8	100%	98%	Cardiomyopathy, familial hypertrophic, 19, 613875
CAMTA1	125,2	97%	97%	Cerebellar ataxia, nonprogressive, with mental retardation, 614756 -3
CANT1	101,7	100%	97%	Desbuquois dysplasia, 251450
CAPN3	112,2	100%	96%	Muscular dystrophy, limb-girdle, type 2A, 253600
CAPN5	80,1	98%	94%	Vitreoretinopathy, neovascular inflammatory, 193235

CARD11	93	100%	98%	Persistent polyclonal B-cell lymphocytosis, 606445 Immunodeficiency 11, 615206
CARD14	64,3	97%	88%	{Psoriasis susceptibility 2}, 602723 Pityriasis rubra pilaris, 173200
CARD9	64,5	99%	98%	Candidiasis, familial, 2, autosomal recessive, 212050
CASC5	143,5	98%	97%	Microcephaly 4, primary, autosomal recessive, 604321
CASK	100,9	100%	100%	Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 FG syndrome 4, 300422 Mental retardation, with or without nystagmus, 300422
CASP10	107,1	100%	100%	Autoimmune lymphoproliferative syndrome, type II, 603909 Non-Hodgkin lymphoma, somatic, 605027 Gastric cancer, somatic, 613659
CASP8	117,9	100%	97%	Immunodeficiency due to CASP8 deficiency, 607271 Hepatocellular carcinoma, somatic, 114550 {Breast cancer, protection against}, 114480 {Lung cancer, protection against}, 211980
CASQ2	86,7	100%	98%	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CASR	111,5	100%	99%	Hypocalciuric hypercalcemia, type I, 145980 Hyperparathyroidism, neonatal, 239200 Hypocalcemia, autosomal dominant, 601198 Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 {Epilepsy idiopathic generalized, susceptibility to, 8}, 612899 Hypercalciuric hypercalcemia {Calcium, serum level of}
CAT	83,6	99%	91%	Acatalasemia, 614097
CATSPER1	107,9	99%	98%	Spermatogenic failure 7, 612997
CAV1	131,6	100%	100%	Lipodystrophy, congenital generalized, type 3, 612526 Pulmonary hypertension, primary, 3, 615343

CAV3	160	100%	100%	Muscular dystrophy, limb-girdle, type IC, 607801 Rippling muscle disease, 606072 Creatine phosphokinase, elevated serum, 123320 Myopathy, distal, Tateyama type, 614321 Cardiomyopathy, familial hypertrophic, 192600 Long QT syndrome-9, 611818
CBL	121,9	100%	100%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563
CBS	73,5	99%	81%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CBX2	131,5	100%	99%	46XY sex reversal 5, 613080
CC2D1A	91	100%	98%	Mental retardation, autosomal recessive 3, 608443
CC2D2A	90,5	98%	97%	Joubert syndrome 9, 612285 Meckel syndrome 6, 612284 COACH syndrome, 216360
CCBE1	83,8	95%	88%	Hennekam lymphangiectasia-lymphedema syndrome, 235510
CCDC103	106,6	100%	99%	Ciliary dyskinesia, primary, 17, 614679
CCDC11	161,7	100%	98%	Heterotaxy, visceral, 6, autosomal recessive, 614779
CCDC114	75,9	100%	98%	Ciliary dyskinesia, primary, 20, 615067
CCDC39	101,1	100%	99%	Ciliary dyskinesia, primary, 14, 613807
CCDC40	86,8	97%	94%	Ciliary dyskinesia, primary, 15, 613808
CCDC50	121,1	99%	96%	Deafness, autosomal dominant 44, 607453
CCDC65	74,1	100%	97%	Ciliary dyskinesia, primary, 27, 615504
CCDC78	93	100%	100%	Myopathy, centronuclear, 4, 614807
CCDC8	138,2	100%	100%	Three M syndrome 3, 614205

CCDC88C	89,5	100%	97%	Hydrocephalus, nonsyndromic, autosomal recessive, 236600
CCT5	76,2	93%	83%	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CD151	82,8	100%	96%	Nephropathy with pretibial epidermolysis bullosa and deafness, 609057 [Blood group, Raph], 179620
CD19	71,7	100%	98%	Immunodeficiency, common variable, 3, 613493
CD247	86,5	100%	100%	Immunodeficiency due to defect in CD3-zeta, 610163
CD2AP	110,9	100%	100%	Glomerulosclerosis, focal segmental, 3, 607832
CD320	74,6	94%	80%	Methylmalonic aciduria due to transcobalamin receptor defect, 613646
CD36	133,9	100%	100%	[Macrothrombocytopenia] (1) Platelet glycoprotein IV deficiency, 608404 {Malaria, cerebral, susceptibility to}, 611162 {Malaria, cerebral, reduced risk of}, 611162 {Coronary heart disease, susceptibility to, 7}, 610938
CD3D	84,2	100%	91%	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971
CD3E	93	99%	84%	Immunodeficiency due to defect in CD3-epsilon Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971
CD3G	96,3	100%	100%	Immunodeficiency due to defect in CD3-gamma
CD4	83,7	99%	97%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
CD4	83,7	99%	97%	OKT4 epitope deficiency, 613949
CD40	105	95%	92%	Immunodeficiency with hyper-IgM, type 3, 606843
CD59	113,5	100%	100%	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD79A	79,1	95%	87%	Agammaglobulinemia 3, 613501
CD79B	113,8	100%	100%	Agammaglobulinemia 6, 612692

CD81	59,4	99%	89%	Immunodeficiency, common variable, 6, 613496
CD8A	82,6	100%	99%	CD8 deficiency, familial, 608957
CD96	114,9	100%	99%	C syndrome, 211750
CDAN1	89,4	100%	97%	Anemia, congenital dyserythropoietic, type I, 224120
CDC6	94,2	99%	95%	Meier-Gorlin syndrome 5, 613805
CDC73	138,9	100%	100%	Hyperparathyroidism, familial primary, 145000 Hyperparathyroidism-jaw tumor syndrome, 145001 Parathyroid adenoma with cystic changes, 145001 Parathyroid carcinoma, 608266
CDH1	105,2	100%	100%	Endometrial carcinoma, somatic, 608089 Ovarian carcinoma, somatic, 167000 {Breast cancer, lobular}, 114480 Gastric cancer, familial diffuse, with or without cleft lip and/or palate, 137215 {Prostate cancer, susceptibility to}, 176807
CDH15	80,3	99%	95%	Mental retardation, autosomal dominant 3, 612580
CDH23	94,4	100%	98%	Usher syndrome, type 1D, 601067 Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D/F digenic, 601067
CDH3	91	99%	95%	i Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280
CDHR1	109,9	98%	96%	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660 -3
CDK5RAP2	98,7	99%	95%	Microcephaly 3, primary, autosomal recessive, 604804
CDKL5	126,9	100%	99%	Epileptic encephalopathy, early infantile, 2, 300672 Angelman syndrome-like, 105830
CDKN1B	117,8	100%	100%	Multiple endocrine neoplasia, type IV, 610755
CDKN1C	41,6	94%	84%	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732

CDKN2A	137,5	100%	100%	{Melanoma, cutaneous malignant, 2}, 155601 Melanoma and neural system tumor syndrome, 155755 Pancreatic cancer/melanoma syndrome, 606719 Orolaryngeal cancer, multiple, -3
CDON	109,8	99%	97%	Holoprosencephaly 11, 614226
CDSN	18,3	87%	34%	Hypotrichosis simplex of scalp 1, 146520 Peeling skin syndrome, 270300
CDT1	48,1	91%	77%	Meier-Gorlin syndrome 4, 613804
CEACAM16	92,9	99%	93%	Deafness, autosomal dominant 4B, 614614
CEBPA	36,7	95%	67%	Leukemia, acute myeloid, 601626
CEBPE	102,2	100%	100%	Specific granule deficiency, 245480
CEL	56,4	62%	59%	Maturity-onset diabetes of the young, type VIII, 609812
CENPJ	127,2	100%	100%	Microcephaly 6, primary, autosomal recessive, 608393 Seckel syndrome 4, 613676
CEP135	118,3	99%	98%	Microcephaly 8, primary, autosomal recessive, 614673
CEP152	125,4	99%	99%	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823
CEP164	75,5	98%	92%	Nephronophthisis 15, 614845
CEP290	93,6	99%	98%	Joubert syndrome 5, 610188 Senior-Loken syndrome 6, 610189 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Bardet-Biedl syndrome 14, 209900
CEP41	85,5	100%	100%	Joubert syndrome 15, 614464
CEP57	87,1	100%	97%	Mosaic variegated aneuploidy syndrome 2, 614114
CERKL	127,5	100%	99%	Retinitis pigmentosa 26, 608380

CERS3	80,2	100%	99%	Ichthyosis, congenital, autosomal recessive 9, 615023
CES1	44	55%	51%	Carboxylesterase 1 deficiency
CETP	97,4	100%	100%	Hyperalphalipoproteinemia, 143470 [High density lipoprotein cholesterol level QTL 10], 143470
CFC1	1,7	0%	0%	Heterotaxy, visceral, 2, autosomal, 605376 Double-outlet right ventricle, 217095 Transposition of the great arteries, dextro-looped 2, 613853
CFD	49,4	96%	77%	Complement factor D deficiency, 613912
CFHR5	95,7	94%	86%	Nephropathy due to CFHR5 deficiency, 614809
CFI	131,3	100%	100%	Complement factor I deficiency, 610984 {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 {Macular degeneration, age-related, 13, susceptibility to}, 615439
CFL2	117,6	100%	100%	Nemaline myopathy 7, autosomal recessive, 610687
CFTR	113,1	95%	93%	Cystic fibrosis, 219700 Congenital bilateral absence of vas deferens, 277180 Sweat chloride elevation without CF {Pancreatitis, idiopathic}, 167800 {Hypertrypsinemia, neonatal} {Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400
CHAT	64,7	87%	79%	Myasthenic syndrome, congenital, associated with episodic apnea, 254210
CHD2	116,6	99%	98%	Epileptic encephalopathy, childhood-onset, 615369
CHD7	117,2	100%	99%	CHARGE syndrome, 214800 {Scoliosis, idiopathic 3}, 608765 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CHEK2	53,4	66%	61%	Li-Fraumeni syndrome, 609265 Osteosarcoma, somatic, 259500 {Breast cancer, susceptibility to}, 114480 {Prostate cancer, familial, susceptibility to}, 176807 {Breast and colorectal cancer, susceptibility to}

CHKB	77	91%	90%	Muscular dystrophy, congenital, megaconial type, 602541
CHM	90	99%	99%	Choroideremia, 303100
CHMP1A	93,9	97%	94%	Pontocerebellar hypoplasia, type 8, 614961
CHMP2B	123,4	100%	100%	Dementia, familial, nonspecific, 600795 Amyotrophic lateral sclerosis 17, 614696
CHMP4B	103,6	100%	100%	Cataract 31, multiple types, 605387
CHN1	113,5	100%	97%	Duane retraction syndrome 2, 604356
CHRDL1	113,9	100%	98%	Megalocornea 1, X-linked 309300
CHRM3	160,3	100%	100%	Eagle-Barrett syndrome, 100100
CHRNA1	103,3	100%	97%	Myasthenic syndrome, slow-channel congenital, 601462 Myasthenic syndrome, fast-channel congenital, 608930 Multiple pterygium syndrome, lethal type, 253290
CHRNA2	119,8	100%	98%	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA4	98,9	98%	95%	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction, susceptibility to}, 188890
CHRNB1	97,7	99%	94%	Myasthenic syndrome, slow-channel congenital, 601462 Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931
CHRNB2	144,2	95%	93%	Epilepsy, nocturnal frontal lobe, 3, 605375
CHRND	98,3	99%	92%	Myasthenic syndrome, slow-channel congenital, 601462 Myasthenic syndrome, fast-channel congenital, 608930 Multiple pterygium syndrome, lethal type, 253290
CHRNE	160,8	100%	100%	Myasthenic syndrome, slow-channel congenital, 601462 Myasthenic syndrome, fast-channel congenital, 608930 Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931
CHRNG	101,6	100%	99%	Myasthenia gravis, neonatal transient (2) Escobar syndrome, 265000 Multiple pterygium syndrome, lethal type, 253290

CHST14	118,4	100%	97%	Ehlers-Danlos syndrome, musculocontractural type , 601776
CHST3	63,9	100%	98%	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHST6	123,3	100%	100%	Macular corneal dystrophy, 217800
CHSY1	142,4	96%	94%	Temtamy preaxial brachydactyly syndrome, 605282
CHUK	86,2	100%	97%	Cocoon syndrome, 613630
CIB2	115,1	100%	100%	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CIRH1A	105,2	100%	100%	Cirrhosis, North American Indian childhood type, 604901
CISD2	163,2	77%	77%	Wolfram syndrome 2, 604928
CITED2	105	99%	96%	Ventricular septal defect 2, 614431 Atrial septal defect 8, 614433
CLCF1	40,6	82%	67%	Cold-induced sweating syndrome 1, 610313
CLCN1	88,3	100%	98%	Myotonia congenita, recessive, 255700 Myotonia congenita, dominant, 160800 Myotonia levior, recessive
CLCN2	102,5	100%	99%	{Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628 {Epilepsy, juvenile absence, susceptibility to, 2}, 607628 {Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628
CLCN5	160,5	100%	99%	Dent disease, 300009 Nephrolithiasis, type I, 310468 Hypophosphatemic rickets, 300554 Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990
CLCN7	82,1	100%	97%	Osteopetrosis, autosomal recessive 4, 611490 Osteopetrosis, autosomal dominant 2, 166600
CLCNKA	83,1	84%	77%	Bartter syndrome, type 4b, digenic, 613090
CLCNKB	72,7	86%	82%	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090

CLDN1	106,6	100%	100%	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626
CLDN14	69,2	100%	94%	Deafness, autosomal recessive 29, 614035
CLDN16	112,8	96%	91%	Hypomagnesemia 3, renal, 248250
CLDN19	78,3	100%	95%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLEC7A	103,3	100%	100%	Candidiasis, familial, 4, autosomal recessive, 613108 {Aspergillosis, susceptibility to}, 614079
CLIC2	70,9	98%	88%	Mental retardation, X-linked, syndromic 32, 300886
CLMP	104,7	97%	96%	Congenital short bowel syndrome, 615237
CLN3	84,1	100%	98%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	125	97%	93%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	72,1	98%	85%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	124,6	100%	100%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLPP	76,1	93%	88%	Perrault syndrome 3, 614129
CLRN1	157,6	100%	99%	Usher syndrome, type 3A, 276902 Retinitis pigmentosa 61, 614180 -3
CNGA1	115,4	91%	90%	Retinitis pigmentosa 49, 613756
CNGA3	143	99%	96%	Achromatopsia-2, 216900
CNGB1	85,9	97%	92%	Retinitis pigmentosa 45, 613767
CNGB3	97	99%	94%	Achromatopsia-3, 262300 Macular degeneration, juvenile, 248200 -3
CNNM2	134,3	100%	100%	Hypomagnesemia 6, renal, 613882
CNNM4	166,1	100%	96%	Jalili syndrome, 217080

CNTN1	100,2	100%	99%	Myopathy, congenital, Compton-North, 612540
CNTNAP2	100	100%	98%	Cortical dysplasia-focal epilepsy syndrome, 610042 {Autism susceptibility 15}, 612100 Pitt-Hopkins like syndrome 1, 610042
COA5	84,2	99%	99%	Mitochondrial complex IV deficiency, 220110
COASY	117,6	100%	100%	Neurodegeneration with brain iron accumulation 6, 615643
COCH	104,4	99%	97%	Deafness, autosomal dominant 9, 601369
COG1	120,4	99%	97%	Congenital disorder of glycosylation, type IIg, 611209
COG4	85,3	97%	95%	Congenital disorder of glycosylation, type IIj, 613489
COG5	99,2	100%	96%	Congenital disorder of glycosylation, type IIi, 613612
COG6	94,9	100%	96%	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328
COG7	80,3	99%	94%	Congenital disorder of glycosylation, type IIe, 608779
COG8	108,6	100%	100%	Congenital disorder of glycosylation, type IIh, 611182
COL10A1	107,9	99%	98%	Metaphyseal chondrodysplasia, Schmid type, 156500
COL11A1	93,4	98%	97%	Stickler syndrome, type II, 604841 Marshall syndrome, 154780 {Lumbar disc herniation, susceptibility to}, 603932 Fibrochondrogenesis, 228520
COL11A2	13,4	55%	16%	Stickler syndrome, type III, 184840 Otospondylomegaepiphyseal dysplasia, 215150 Weissenbacher-Zweymuller syndrome, 277610 Deafness, autosomal dominant 13, 601868 Deafness, autosomal recessive 53, 609706 Fibrochondrogenesis 2, 614524
COL17A1	85,1	97%	92%	Epidermolysis bullosa, junctional, non-Herlitz type, 226650

COL18A1	86,2	98%	87%	Knobloch syndrome, type 1, 267750
COL1A1	109,9	98%	97%	Osteogenesis imperfecta, type I, 166200 OI type II, 166210 OI type III, 259420 OI type IV, 166220 Ehlers-Danlos syndrome, type I, 130000 Ehlers-Danlos syndrome, type VIIA, 130060 {Osteoporosis}, 166710 Caffey disease, 114000 [Bone mineral density variation QTL], 166710
COL1A2	88,3	97%	91%	Ehlers-Danlos syndrome, type VIIB, 130060 Osteogenesis imperfecta, type IV, 166220 Osteogenesis imperfecta, type III, 259420 Osteogenesis imperfecta, type II, 166210 {Osteoporosis, postmenopausal}, 166710 Ehlers-Danlos syndrome, cardiac valvular form, 225320
COL2A1	81,1	99%	95%	Stickler syndrome, type I, 108300 Kniest dysplasia, 156550 Achondrogenesis, type II or hypochondrogenesis, 200610 SED congenita, 183900 SMED Strudwick type, 184250 Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 Spondyloperipheral dysplasia, 271700 SED, Namaqualand type Osteoarthritis with mild chondrodysplasia, 604864 Vitreoretinopathy with phalangeal epiphyseal dysplasia Platyspondylic skeletal dysplasia, Torrance type, 151210 Otospondylomegaepiphyseal dysplasia, 215150 Avascular necrosis of the femoral head, 608805 Legg-Calve-Perthes disease, 150600 Stickler sydrome, type I, nonsyndromic ocular, 609508 Czech dysplasia, 609162
COL3A1	67,6	97%	91%	Ehlers-Danlos syndrome, type IV, 130050 Ehlers-Danlos syndrome, type III, 130020

COL4A1	82	98%	95%	Porencephaly 1, 175780 Brain small vessel disease with hemorrhage, 607595 Angiopathy, hereditary, with nephropathy, aneurysms, and muscle, 611773 Brain small vessel disease with Axenfeld-Rieger anomaly, 607595 {Hemorrhage, intracerebral, susceptibility to}, 614519
COL4A2	80,5	100%	97%	Porencephaly 2, 614483 {Hemorrhage, intracerebral, susceptibility to}, 614519
COL4A3	70	97%	93%	Alport syndrome, autosomal recessive, 203780 Hematuria, benign familial, 141200 Alport syndrome, autosomal dominant, 104200
COL4A4	83,8	99%	97%	Alport syndrome, autosomal recessive, 203780 Hematuria,familial benign
COL4A5	69,4	100%	96%	Alport syndrome, 301050
COL5A1	97,3	97%	96%	Ehlers-Danlos syndrome, type II, 130010 Ehlers-Danlos syndrome, type I, 130000
COL5A2	82,1	98%	94%	Ehlers-Danlos syndrome, type I, 130000
COL6A1	84,9	99%	96%	Bethlem myopathy, 158810 Ullrich congenital muscular dystrophy, 254090 {Ossification of the posterior longitudinal spinal ligaments}, 602475 (2)
COL6A2	82,3	100%	95%	Bethlem myopathy, 158810 Ullrich congenital muscular dystrophy, 254090 Myosclerosis, congenital, 255600
COL6A3	116	99%	99%	Bethlem myopathy, 158810 Ullrich congenital muscular dystrophy, 254090
COL7A1	100,6	100%	99%	Epidermolysis bullosa dystrophica, AD, 131750 Epidermolysis bullosa dystrophica, AR, 226600 Epidermolysis bullosa, pretibial, 131850 EBD, Bart type, 132000 EBD, localisata variant Transient bullous of the newborn, 131705 Epidermolysis bullosa pruriginosa, 604129 Toenail dystrophy, isolated, 607523 EBD inversa, 226600

COL8A2	60,8	98%	95%	Corneal dystrophy, Fuchs endothelial, 1, 136800 Corneal dystrophy polymorphous posterior, 2, 609140
COL9A1	96,9	99%	93%	Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	75,1	95%	93%	Epiphyseal dysplasia, multiple, 2, 600204 {Intervertebral disc disease, susceptibility to}, 603932 Stickler syndrome, type V, 614284
COL9A3	65,6	97%	89%	Epiphyseal dysplasia, multiple, 3, 600969 Epiphyseal dysplasia, multiple, with myopathy {Intervertebral disc disease, susceptibility to}, 603932
COLQ	77,5	100%	97%	Endplate acetylcholinesterase deficiency, 603034
COMP	91,9	100%	99%	Pseudoachondroplasia, 177170 Epiphyseal dysplasia, multiple 1, 132400
COQ2	71,8	100%	88%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ6	107,3	100%	96%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ9	82,4	99%	87%	Coenzyme Q10 deficiency, primary, 5, 614654
CORIN	114,2	100%	99%	Preeclampsia/eclampsia 5, 614595
CORO1A	91,8	85%	85%	Immunodeficiency 8, 615401
COX10	129,5	100%	93%	Encephalopathy, progressive mitochondrial, with proximal renal tubulopathy due to cytochrome c oxidase deficiency
COX14	156,4	100%	100%	Mitochondrial complex IV deficiency, 220110
COX15	74,1	100%	98%	Leigh syndrome due to cytochrome c oxidase deficiency, 256000 Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119
COX20	53,5	88%	73%	Mitochondrial complex IV deficiency, 220110
COX4I2	49,5	99%	90%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX7B	56,8	99%	98%	Aplasia cutis congenita, reticulolinear, with mmicrocephaly, facial dysmorphism and other congenital anomalies, 300887

CP	85,2	98%	93%	[Hypoceruloplasminemia, hereditary], 604290 Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290
CPA6	124,5	100%	100%	Epilepsy, familial temporal lobe, 5, 614417 Febrile seizures, familial, 11, 614418
CPN1	79,8	100%	95%	Carboxypeptidase N deficiency, 212070
CPOX	79,2	100%	98%	Coproporphyria, 121300 Harderoporphyria, 121300
CPS1	101,6	100%	100%	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}
CPT1A	96,5	100%	98%	CPT deficiency, hepatic, type IA, 255120
CPT2	94	95%	91%	Myopathy due to CPT II deficiency, 255110 CPT deficiency, hepatic, type II, 600649 CPT II deficiency, lethal neonatal, 608836 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212
CR2	108,9	100%	100%	{Systemic lupus erythematosus, susceptibility to, 9}, 610927 Immunodeficiency, common variable, 7, 614699
CRADD	121,8	99%	93%	Mental retardation, autosomal recessive 34, 614499
CRB1	150,9	100%	99%	Retinitis pigmentosa-12, autosomal recessive, 600105 Leber congenital amaurosis 8, 613835 Pigmented paravenous chorioretinal atrophy, 172870
CRBN	134	100%	100%	Mental retardation, autosomal recessive 2, 607417
CREB1	84,6	100%	100%	Histiocytoma, angiomatoid fibrous, somatic, 612160
CREBBP	77,5	99%	97%	Rubinstein-Taybi syndrome, 180849
CRELD1	80,6	100%	95%	{Atrioventricular septal defect, susceptibility to, 2}, 606217 Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217
CRLF1	66,4	89%	84%	Cold-induced sweating syndrome, 272430
CRTAP	98,4	100%	99%	Osteogenesis imperfecta, type VII, 610682

CRTC1	85,8	98%	91%	Mucoepidermoid salivary gland carcinoma
CRX	140,6	100%	100%	Cone-rod retinal dystrophy-2, 120970 Leber congenital amaurosis 7, 613829
CRYAA	107,4	100%	100%	Cataract 9, multiple types, 604219
CRYAB	124,7	100%	100%	Myopathy, myofibrillar, 2, 608810 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, fatal infantile hypertrophy, alpha-B crystallin-related, 613869 Cardiomyopathy, dilated, 1II, 615184
CRYBA1	92,7	100%	98%	Cataract 10, multiple types, 600881
CRYBA4	72,3	100%	100%	Cataract 23, 610425
CRYBB1	59,1	100%	88%	Cataract 17, multiple types, 611544
CRYBB2	101,6	100%	100%	Cataract 3, multiple types, 601547
CRYBB3	103,4	100%	100%	Cataract 22, autosomal recessive, 609741
CRYGB	64,6	100%	95%	Cataract 39, multiple types, autosomal dominant, 615188
CRYGC	86,3	99%	93%	Cataract 2, multiple types, 604307
CRYGD	72,9	82%	76%	Cataract 4, multiple types, 115700
CRYGS	102,6	99%	91%	Cataract 20, multiple types, 116100
CRYM	70	100%	99%	Deafness, autosomal dominant 40
CSF1R	74,8	100%	94%	Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
CSF2RA	0	0%	0%	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF2RB	106,2	98%	93%	Surfactant metabolism dysfunction, pulmonary, 5, 614370
CSF3R	82,8	100%	98%	Neutrophilia, hereditary, 162830

CSNK1D	90,9	94%	87%	Advanced sleep-phase syndrome, familial, 2, 615224
CSPP1	117,4	100%	99%	Joubert syndrome 21, 615636
CSRP3	110,7	100%	100%	Cardiomyopathy, dilated, 1M, 607482 Cardiomyopathy, familial hypertrophic, 12, 612124
CST3	52,4	100%	88%	Cerebral amyloid angiopathy, 105150 Macular degeneration, age-related, 11, 611953
CSTA	97,4	100%	100%	Exfoliative ichthyosis, autosomal recessive, ichthyosis bullosa of Siemens-like, 607936
CSTB	163,5	100%	99%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTC1	96,7	99%	96%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTCF	111,4	100%	99%	Mental retardation, autosomal dominant 21, 615502
CTDP1	70,7	89%	86%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTH	117,2	100%	100%	Cystathioninuria, 219500 Homocysteine, total plasma, elevated -3
CTHRC1	87,1	100%	99%	Barrett esophagus/esophageal adenocarcinoma, 614266
CTNNA3	112,3	98%	97%	Arrhythmogenic right ventricular dysplasia, familial, 13, 615616
CTNNB1	108	99%	95%	Mental retardation, autosomal dominant 19, 615075 Colorectal cancer, somatic, 114500 Pilomatricoma, somatic, 132600 Ovarian cancer, somatic, 167000 Hepatocellular carcinoma, somatic, 114550
CTNS	109,2	93%	86%	? Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, atypical nephropathic, 219800 -3
CTSA	97	100%	99%	Galactosialidosis, 256540

CTSC	94,7	100%	99%	Papillon-Lefevre syndrome, 245000 Haim-Munk syndrome, 245010 Periodontitis 1, juvenile, 170650
CTSD	93,6	100%	97%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	103,7	98%	83%	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362
CTSK	121	100%	98%	Pycnodysostosis, 265800
CUBN	83,2	98%	94%	Megaloblastic anemia-1, Finnish type, 261100
CUL3	107,2	99%	97%	Pseudohypoaldosteronism, type IIE, 614496
CUL4B	109,7	100%	99%	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
CUL7	98,9	99%	98%	3-M syndrome 1, 273750
CXCR4	193,2	100%	100%	WHIM syndrome, 193670 Myelokathexis, isolated
CYB5A	52,8	100%	94%	Methemoglobinemia, type IV, 250790
CYB5R3	81,6	97%	90%	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYBA	38,2	92%	71%	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690
CYBB	94,4	92%	88%	Chronic granulomatous disease, X-linked, 306400 Atypical mycobacteriosis, familial, X-linked 2, 300645
CYC1	85,1	93%	76%	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYCS	61	100%	93%	Thrombocytopenia 4, 612004
CYLD	110,7	100%	98%	Cylindromatosis, familial, 132700 Brooke-Spiegler syndrome, 605041 Trichoepithelioma, multiple familial, 1, 601606
CYP11B1	137,4	97%	93%	anti-Lepore-like Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 Aldosteronism, glucocorticoid-remediable, 103900

CYP11B2	101,3	98%	93%	Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 {Low renin hypertension, susceptibility to} Aldosterone to renin ratio raised
CYP17A1	102,8	100%	98%	17-alpha-hydroxylase/17,20-lyase deficiency, 202110 17,20-lyase deficiency, isolated, 202110
CYP19A1	125,2	100%	100%	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300 -3
CYP1B1	108,4	100%	99%	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 Peters anomaly, 604229
CYP21A2	7,4	21%	14%	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910
CYP24A1	93,3	99%	98%	Hypercalcemia, infantile, 143880
CYP26B1	74,2	100%	96%	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
CYP26C1	55,3	100%	91%	Focal facial dermal dysplasia 4, 614974
CYP27A1	106,5	100%	96%	Cerebrotendinous xanthomatosis, 213700
CYP27B1	96,1	100%	95%	Vitamin D-dependent rickets, type I, 264700
CYP2A6	26,8	56%	45%	Coumarin resistance, 122700 {Nicotine addiction, protection from}, 188890 {Lung cancer, resistance to}, 211980
CYP2B6	100,6	88%	85%	Efavirenz, poor metabolism of, 614546 {Efavirenz central nervous system toxicity, susceptibility to}, 614546
CYP2C8	124,3	100%	100%	Rhabdomyolysis, cerivastatin-induced
CYP2C9	101,1	98%	94%	Tolbutamide poor metabolizer Warfarin sensitivity, 122700
CYP2R1	102,6	97%	95%	Rickets due to defect in vitamin D 25-hydroxylation, 600081
CYP2U1	104,8	98%	93%	Spastic paraplegia 56, autosomal recessive, 615030
CYP4F22	91,1	99%	97%	Ichthyosis, congenital, autosomal recessive 5, 604777

CYP4V2	113,7	100%	100%	Bietti crystalline corneoretinal dystrophy, 210370
CYP7B1	91,6	99%	94%	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, autosomal recessive, 270800
D2HGDH	61	98%	86%	D-2-hydroxyglutaric aciduria, 600721
DAG1	134,7	100%	100%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818
DARS	117,8	100%	100%	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2	112,6	100%	99%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBH	100,7	99%	96%	[Dopamine-beta-hydroxylase activity levels, plasma] Dopamine beta-hydroxylase deficiency, 223360
DBT	104,9	100%	100%	Maple syrup urine disease, type II, 248600
DCAF17	101	100%	95%	Woodhouse-Sakati syndrome, 241080
DCC	110,5	99%	98%	Mirror movements 1, 157600 Colorectal cancer, somatic, 114500 Esophageal carcinoma, somatic 133239
DCLRE1C	104,2	97%	97%	Severe combined immunodeficiency, Athabascan type, 602450 Omenn syndrome, 603554
DCN	120,7	100%	100%	Corneal dystrophy, congenital stromal, 610048
DCTN1	112,8	99%	96%	Neuropathy, distal hereditary motor, type VIIB, 607641 {Amyotrophic lateral sclerosis, susceptibility to}, 105400 Perry syndrome, 168605
DCX	114,4	100%	100%	Lissencephaly, X-linked, 300067 Subcortical laminal heteropia, X-linked, 300067
DDB2	90,8	100%	98%	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DDC	90,3	100%	98%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	136,3	99%	93%	Spastic paraplegia 28, autosomal recessive, 609340
DDHD2	97,2	100%	99%	Spastic paraplegia 54, autosomal recessive, 615033

DDOST	103,2	100%	98%	Congenital disorder of glycosylation, type Ir, 614507
DDR2	125,7	100%	100%	Spondylometaepiphyseal dysplasia, short limb-hand type, 271665
DDX11	10,1	16%	12%	Warsaw breakage syndrome, 613398
DDX59	144	100%	100%	Orofaciodigital syndrome V, 174300
DEPDC5	109,9	99%	99%	Epilepsy, familial focal, with variable foci, 604364
DES	88,1	94%	91%	Myopathy, myofibrillar, 1, 601419 Cardiomyopathy, dilated, 1I, 604765 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 ?Muscular dystrophy, limb-girdle, type 2R, 615325
DFNA5	96,6	97%	93%	Deafness, autosomal dominant 5, 600994
DGKE	110,2	98%	97%	Nephrotic syndrome, type 7, 615008
DGUOK	99,6	100%	99%	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHCR24	81,2	99%	97%	Desmosterolosis, 602398
DHCR7	107	99%	98%	Smith-Lemli-Opitz syndrome, 270400
DHDDS	68,2	100%	94%	Retinitis pigmentosa 59, 613861
DHFR	50,6	79%	63%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHH	75,6	100%	100%	46XY partial gonadal dysgenesis, with minifascicular neuropathy, 607080 46XY sex reversal 7, 233420
DHODH	93,2	100%	97%	Miller syndrome, 263750
DHTKD1	104,7	100%	99%	2-aminoadipic 2-oxoadipic aciduria, 204750 Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DIAPH1	80	99%	90%	Deafness, autosomal dominant 1, 124900
DIAPH2	110,5	98%	95%	Premature ovarian failure, 300511

DIAPH3	101	99%	94%	Auditory neuropathy, autosomal dominant, 1, 609129
DICER1	116,1	100%	99%	Pleuropulmonary blastoma, 601200 Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800
DIP2B	100,2	99%	97%	Mental retardation, FRA12A type, 136630
DIS3L2	127,7	98%	92%	Perlman syndrome, 267000
DKC1	91,1	100%	98%	Dyskeratosis congenita, X-linked, 305000
DLAT	98,3	100%	100%	Pyruvate dehydrogenase E2 deficiency, 245348
DLC1	144,6	100%	99%	Colorectal cancer, somatic
DLD	131,2	100%	100%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLG3	76,9	97%	92%	Mental retardation, X-linked 90, 300850
DLL3	76,8	96%	81%	Spondylocostal dysostosis, autosomal recessive, 1, 277300
DLX3	71,8	98%	93%	Trichodontoosseous syndrome, 190320 Amelogenesis imperfecta, hypomaturation-hypoplastic type, with taurodontism, 104510
DMD	108,2	100%	99%	Duchenne muscular dystrophy, 310200 Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045
DMGDH	106	98%	96%	Dimethylglycine dehydrogenase deficiency, 605850
DMP1	110,4	100%	100%	Hypophosphatemic rickets, AR, 241520
DMPK	100	100%	96%	Myotonic dystrophy 1, 160900
DNA2	107,8	100%	98%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 6, 615156
DNAAF3	67,6	96%	81%	Ciliary dyskinesia, primary, 2, 606763
DNAH11	109,4	99%	99%	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884

DNAH5	90,9	99%	98%	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAI1	125,2	100%	100%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	115,3	97%	93%	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJB2	101,9	100%	98%	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881
DNAJB6	39,3	84%	68%	Muscular dystrophy, limb-girdle, type 1E, 603511
DNAJC19	55,5	79%	78%	3-methylglutaconic aciduria, type V, 610198
DNAJC5	71	92%	80%	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350
DNAJC6	95,1	98%	95%	Parkinson disease 19, juvenile-onset, 615528
DNAL1	125,2	100%	100%	Ciliary dyskinesia, primary, 16, 614017
DNASE1L3	83,8	100%	99%	Systemic lupus erythematosus 16, 614420
DNM1L	94,4	100%	100%	Encephalopahty, lethal, due to defective mitochondrial peroxisomal fission, 614388
DNM2	78,1	100%	96%	Charcot-Marie-Tooth disease, dominant intermediate B, 606482 Myopathy, centronuclear, 160150 Charcot-Marie-Tooth disease, axonal, type 2M, 606482 Lethal congenital contracture syndrome 5, 615368
DNMT1	102,1	99%	96%	Neuropathy, hereditary sensory, type IE, 614116 Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121
DNMT3B	93,3	100%	97%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK6	86,7	99%	94%	Adams-Oliver syndrome 2, 614219
DOCK8	84	100%	98%	Mental retardation, autosomal dominant 2, 614113 Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
DOK7	54,3	91%	83%	Myasthenia, limb-girdle, familial, 254300 Fetal akinesia deformation sequence, 208150
DPAGT1	87,5	100%	94%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, with tubular aggregates 2, 614750

DPM1	145,4	100%	100%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	79,4	98%	98%	Congenital disorder of glycosylation, type Iu, 615042
DPM3	100,1	100%	100%	Congenital disorder of glycosylation, type Io, 612937
DPP6	94,6	95%	89%	Ventricular fibrillation, paroxysmal familial, 2, 612956
DPY19L2	21,1	32%	21%	Spermatogenic failure 9, 613958
DPYD	112,6	99%	97%	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270
DPYS	59,1	100%	98%	Dihydropyrimidinuria, 222748
DRC1	70,5	100%	96%	Ciliary dyskinesia, primary, 21, 615294
DRD2	101,5	99%	97%	Dystonia, myoclonic, 159900
DRD4	39	85%	63%	Autonomic nervous system dysfunction [Novelty seeking personality], 601696 (1) {Attention deficit-hyperactivity disorder}, 143465
DRD5	18	58%	46%	{Blepharospasm, primary benign}, 606798 Dystonia, primary cervical {Attention deficit-hyperactivity disorder, susceptibility to}, 143465
DSC2	96,1	99%	97%	Arrhythmogenic right ventricular dysplasia 11, 610476 Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476
DSC3	94,2	99%	99%	Hypotrichosis and recurrent skin vesicles, 613102
DSG1	147,8	100%	100%	pemphigus foliaceous antigen Keratosis palmoplantaris striata I, 148700
DSG2	122,9	99%	98%	Arrhythmogenic right ventricular dysplasia 10, 610193 Cardiomyopathy, dilated, 1BB, 612877
DSG4	123,8	100%	100%	Hypotrichosis, localized, autosomal recessive, 607903

DSP	129,2	99%	98%	Keratosis palmoplantaris striata II, 612908 Dilated cardiomyopathy with woolly hair and keratoderma, 605676 Arrhythmogenic right ventricular dysplasia 8, 607450 Skin fragility-woolly hair syndrome, 607655 Epidermolysis bullosa, lethal acantholytic, 609638
DSPP	138,4	98%	96%	Dentinogenesis imperfecta, Shields type II, 125490 Deafness, autosomal dominant 36, with dentinogenesis, 605594 Dentinogenesis imperfecta, Shields type III, 125500 Dentin dysplasia, type II, 125420 -3
DST	134,3	99%	98%	Neuropathy, hereditary sensory and autonomic, type VI, 614653 Epidermolysis bullosa simplex, sutosomal recessive 2, 615425
DTNA	98,2	98%	96%	Left ventricular noncompaction 1, with or without congenital heart defects, 604169
DTNBP1	112,5	100%	100%	{Schizophrenia}, 181500 (2) Hermansky-Pudlak syndrome 7, 614076
DUOX2	93,2	94%	90%	Thryoid dyshormonogenesis 6, 607200
DUOXA2	90,8	99%	95%	Thyroid dyshormonogenesis 5, 274900
DUSP6	140,4	100%	98%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
DYM	93,4	100%	100%	Dyggve-Melchior-Clausen disease, 223800 Smith-McCort dysplasia, 607326
DYNC1H1	112,5	99%	97%	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant, AD, 158600
DYNC2H1	111,2	99%	99%	Asphyxiating thoracic dystrophy 3, 613091 Short rib-polydactyly syndrome, type III, 263510 Short rib-polydactyly syndrome, type IIB, 615087
DYRK1A	150,8	99%	98%	Mental retardation, autosomal dominant 7, 614104
DYSF	93,1	100%	98%	Muscular dystrophy, limb-girdle, type 2B, 253601 Myopathy, distal, with anterior tibial onset, 606768 Miyoshi muscular dystrophy 1, 254130
DYX1C1	87,4	100%	100%	{Dyslexia, susceptibility to, 1}, 127700

EARS2	69,5	93%	90%	Combined oxidative phosphorylation deficiency 12, 614924
EBP	95,6	99%	95%	Chondrodysplasia punctata, X-linked dominant, 302960
ECE1	93,2	97%	97%	Hirschsprung disease, cardiac defects, and autonomic dysfunction, 613870 {Hypertension, essential, susceptibility to}, 145500
ECEL1	65,1	97%	84%	Arthrogryposis, distal, type 5D, 615065
ECM1	105,9	100%	98%	Urbach-Wiethe disease, 247100
EDAR	76,3	100%	98%	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490 Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900 [Hair morphology 1, hair thickness], 612630 -3
EDARADD	114,5	100%	98%	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941 Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, 614940
EDN1	138,6	100%	100%	[High density lipoprotein cholesterol level QTL 7]
EDN3	91,7	100%	100%	Waardenburg syndrome, type 4B, 613265 Central hypoventilation syndrome, congenital, 209880 {Hirschsprung disease, susceptibility to, 4}, 613712
EDNRA	118	100%	100%	Migraine, resistance to, 157300
EDNRB	137,1	100%	99%	{Hirschsprung disease, susceptibility to, 2}, 600155 ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580
EFEMP1	117,3	100%	99%	Doyne honeycomb degeneration of retina, 126600
EFEMP2	95,4	100%	100%	Cutis laxa, autosomal recessive, type IB, 614437
EFNB1	106	100%	100%	? Craniofrontonasal dysplasia, 304110
EFTUD2	86,7	98%	98%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EGF	110,2	100%	98%	Hypomagnesemia 4, renal, 611718

EGFR	95,2	100%	99%	Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980 Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980 {Nonsmall cell lung cancer, susceptibility to}, 211980
EGLN1	91,2	77%	74%	Erythrocytosis, familial, 3, 609820
EGR2	70	100%	97%	Neuropathy, congenital hypomyelinating, 1, 605253 Charcot-Marie-Tooth disease, type 1D, 607678 Dejerine-Sottas disease, 145900
EHMT1	93,1	96%	93%	Kleefstra syndrome, 610253
EIF2AK3	106,5	92%	91%	Wolcott-Rallison syndrome, 226980
EIF2B1	98,1	100%	97%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B2	84,9	100%	97%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B3	79,6	100%	98%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B4	114,3	100%	100%	Leukoencephaly with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B5	90,4	100%	97%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF4G1	104,3	100%	98%	Parkinson disease 18, 614251
ELAC2	85,3	100%	100%	{Prostate cancer, hereditary, 2, susceptibility to}, 614731 Combined oxidative phosphorylation deficiency 17, 615440
ELANE	109,7	99%	92%	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
ELN	67,9	100%	98%	Supravalvar aortic stenosis, 185500 Cutis laxa, AD, 123700
ELOVL4	107,3	100%	100%	Stargardt disease 3, 600110 Macular dystrophy, autosomal dominant, chromosome 6-linked, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
EMD	177,1	100%	99%	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
EMG1	91,5	100%	98%	Bowen-Conradi syndrome, 211180

EMX2	104,4	100%	100%	Schizencephaly, 269160
ENAM	125,1	100%	100%	Amelogenesis imperfecta, type IB, 104500 Amelogenesis imperfecta, type IC, 204650
ENG	68,5	97%	85%	Telangiectasia, hereditary hemorrhagic, type 1, 187300
ENO3	100,3	99%	94%	Glycogen storage disease XIII, 612932
ENPP1	106,7	96%	93%	Ossification of posterior longitudinal ligament of spine, 602475 {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853 {Obesity, susceptibility to}, 601665 Arterial calcification, generalized, of infancy, 1, 208000 Hypophosphatemic rickets, autosomal recessive, 2, 613312
ENTPD1	120,7	100%	99%	Spastic paraplegia 64, 615683
EOGT	101,9	100%	100%	Adams-Oliver syndrome 4, 615297
EP300	130,9	100%	97%	Rubinstein-Taybi syndrome 2, 613684 Colorectal cancer, somatic, 114500
EPAS1	91,1	98%	94%	Erythrocytosis, familial, 4, 611783
EPB41	116,6	100%	100%	Elliptocytosis-1, 611804
EPB42	89,9	100%	97%	Spherocytosis, hereditary, type 5, 612690
EPCAM	105,2	100%	95%	Diarrhea 5, with tufting enteropathy, congenital, 613217 Colorectal cancer, hereditary nonpolyposis, type 8, 613244
EPG5	87,2	100%	99%	Vici syndrome, 242840
EPHA2	87,1	97%	92%	Cataract 6, multiple types, 116600
EPHB2	113,9	97%	97%	Prostate cancer, progression and metastasis of, 603688
EPHX1	92,1	95%	84%	?Fetal hydantoin syndrome (1) Diphenylhydantoin toxicity (1) Hypercholanemia, familial, 607748 {Preeclampsia, susceptibility to}, 189800

EPM2A	60	81%	77%	Epilepsy, progressive myoclonic 2A (Lafora), 254780
EPX	105,8	97%	93%	Eosinophil peroxidase deficiency, 261500
ERBB2	98,5	100%	98%	Adenocarcinoma of lung, somatic, 211980 Glioblastoma, somatic, 137800 Gastric cancer, somatic, 613659 Ovarian cancer, somatic,
ERBB3	113,1	100%	99%	Lethal congenital contractural syndrome 2, 607598
ERBB4	119	100%	100%	Amyotrophic lateral sclerosis 19, 615515
ERCC1	79,1	98%	94%	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	87,2	99%	93%	Xeroderma pigmentosum, group D, 278730 Trichothiodystrophy, 601675 Cerebrooculofacioskeletal syndrome 2, 610756
ERCC3	121,5	100%	100%	Xeroderma pigmentosum, group B, 610651 Trichothiodystrophy, 601675
ERCC4	141,5	99%	94%	Xeroderma pigmentosum, group F, 278760 XFE progeroid syndrome, 610965 Fanconi anemia, complementation group Q, 615272 Xeroderma pigmentosum, type F/Cockayne syndrome, 278760
ERCC5	114,7	98%	98%	Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	141,3	98%	97%	Cockayne syndrome, type B, 133540 Cerebrooculofacioskeletal syndrome 1, 214150 De Sanctis-Cacchione syndrome, 278800 {Macular degeneration, age-related, susceptibility to 5}, 613761 UV-sensitive syndrome 1, 600630 {Lung cancer, susceptibility to}, 211980
ERCC8	88,4	100%	98%	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621
ERF	103	100%	100%	Craniosynostosis 4, 600775
ERLIN2	116,7	100%	98%	Spastic paraplegia 18, autosomal recessive, 611225

ESCO2	78	100%	99%	Roberts syndrome, 268300 SC phocomelia syndrome, 269000
ESPN	41,3	78%	57%	Deafness, autosomal recessive 36, 609006 Deafness, neurosensory, without vestibular involvement, autosomal dominant
ESR1	107,2	100%	100%	Estrogen resistance, 615363 {HDL response to hormone replacement, augmented} {Migraine, susceptibility to}, 157300 {Atherosclerosis, susceptibility to} {Myocardial infarction, susceptibility to}, 608446 {Breast cancer}, 114480 (1)
ESRRB	54,1	87%	73%	Deafness, autosomal recessive 35, 608565
ETFA	108,5	100%	100%	Glutaric acidemia IIA, 231680
ETFB	105,7	100%	100%	Glutaric acidemia IIB, 231680
ETFDH	120,2	100%	100%	Glutaric acidemia IIC, 231680
ETHE1	57,6	100%	98%	Ethylmalonic encephalopathy, 602473
ETV6	116	100%	100%	Leukemia, acute myeloid, somatic, 601626
EVC	74	92%	88%	Ellis-van Creveld syndrome, 225500 Weyers acrodental dysostosis, 193530
EWSR1	52,1	84%	72%	Ewing sarcoma, 612219 Neuroepithelioma, 612219
EXOSC3	61,7	93%	75%	Pontocerebellar hypoplasia, type 1B, 614678
EXPH5	143,1	99%	99%	Epidermolysis bullosa, nonspecific, autosomal recessive, 615028
EXT1	103,7	97%	95%	Exostoses, multiple, type 1, 133700 Chondrosarcoma, 215300
EXT2	105,1	100%	95%	Exostoses, multiple, type 2, 133701

EYA1	104,9	100%	99%	Branchiootorenal syndrome 1, with or without cataracts, 113650 Anterior segment anomalies with or without cataract, 113650 Branchiootic syndrome 1, 602588 Otofaciocervical syndrome, 166780
EYA4	114,2	100%	100%	Deafness, autosomal dominant 10, 601316 Cardiomyopathy, dilated, 1J, 605362
EYS	120,8	100%	100%	Retinitis pigmentosa 25, 602772
EZH2	83,1	99%	94%	Weaver syndrome, 277590
F10	96,7	100%	99%	Factor X deficiency, 227600
F11	99	98%	92%	Factor XI deficiency, autosomal recessive, 612416 Factor XI deficiency, autosomal dominant, 612416
F12	96,7	100%	99%	Factor XII deficiency, 234000 Angioedema, hereditary, type III, 610618
F13A1	96,2	98%	96%	Factor XIIIA deficiency, 613225 {Myocardial infarction, protection against}, 608446 {Venous thrombosis, protection against}, 188050
F13B	89,1	100%	100%	Factor XIIIB deficiency, 613235
F2	85	96%	89%	Hypoprothrombinemia, 613679 Dysprothrombinemia, 613679 Thrombophilia due to thrombin defect, 188050 {Stroke, ischemic, susceptibility to}, 601367 {Pregnancy loss, recurrent, susceptibility to, 2}, 614390
F5	128,2	99%	98%	Factor V deficiency, 227400 {Thrombophilia, susceptibility to, due to factor V Leiden}, 188055 {Stroke, ischemic, susceptibility to}, 601367 {Budd-Chiari syndrome}, 600880 Thrombophilia due to activated protein C resistance, 188055 {Pregnancy loss, recurrent, susceptibility to, 1}, 614389
F7	89,1	100%	100%	Factor VII deficiency, 227500 {Myocardial infarction, decreased susceptibility to}, 608446
F8	128,6	99%	98%	Hemophilia A, 306700

F9	149,9	100%	100%	Hemophilia B, 306900 {Warfarin sensitivity}, 122700 Thrombophilia, X-linked, due to factor IX defect, 300807 {Deep venous thrombosis, protection against}, 300807
FADD	100,9	100%	98%	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovasuclar malformations, 613759
FAH	111,8	100%	100%	Tyrosinemia, type I, 276700
FAM111A	164,4	100%	100%	Kenny-Caffey syndrome, type 2, 127000 Gracile bone dysplasia, 602361
FAM126A	130,2	100%	100%	Leukodystrophy, hypomyelinating, 5, 610532
FAM134B	85,2	100%	95%	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
FAM161A	134,5	100%	100%	Retinitis pigmentosa 28, 606068
FAM20A	68,4	99%	85%	Amelogenesis imperfecta and gingival fibromatosis syndrome, 614253
FAM20C	77,6	92%	87%	Raine syndrome, 259775
FAM58A	40,7	78%	53%	STAR syndrome, 300707
FAM83H	73,2	99%	97%	Amelogenesis imperfecta, type 3, 130900
FAN1	112,7	100%	99%	Interstitial nephritis, karyomegalic, 614817
FANCA	86,8	99%	97%	Fanconi anemia, complementation group A, 227650
FANCC	74,6	100%	93%	Fanconi anemia, complementation group C, 227645
FANCD2	92,4	87%	86%	Fanconi anemia, complementation group D2, 227646
FANCE	86,4	97%	92%	Fanconi anemia, complementation group E, 600901
FANCF	143,4	100%	100%	Fanconi anemia, complementation group F, 603467
FANCI	118,8	100%	100%	Fanconi anemia, complementation group I, 609053

FANCM	117,3	100%	99%	Fanconi anemia, complementation group M, 614087
FARS2	101,4	100%	95%	Combined oxidative phosphorylation deficiency 14, 614946
FAT4	157,9	100%	100%	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 Van Maldergem syndrome 2, 615546
FBLN1	104,9	97%	95%	Synpolydactyly, 3/3'4, associated with metacarpal and metatarsal synostoses, 608180
FBLN5	77,6	100%	97%	Cutis laxa, autosomal recessive, type IA, 219100 Cutis laxa, autosomal dominant 2, 614434 Macular degeneration, age-related, 3, 608895
FBN1	95,8	100%	98%	Marfan syndrome, 154700 Ectopia lentis, familial, 129600 MASS syndrome, 604308 Weill-Marchesani syndrome 2, dominant, 608328 Aortic aneurysm, ascending, and dissection Stiff skin syndrome, 184900 Acromicric dysplasia, 102370 Geleophysic dysplasia 2, 614185
FBN2	103,9	99%	99%	Contractural arachnodactyly, congenital, 121050
FBP1	100,5	100%	97%	Fructose-1,6-bidphosphatase deficiency, 229700
FBXL4	152,8	100%	100%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FBXO38	110,8	98%	94%	Neuronopathy, distal hereditary motor, type IID, 65575
FBXO7	150,9	100%	100%	Parkinson disease 15, autosomal recessive, 260300
FCGR3A	54	56%	50%	{Viral infections, recurrent}
FCGR3B	49,5	57%	55%	Neutropenia, alloimmune neonatal
FCN3	99,7	98%	96%	Immunodeficiency due to ficolin 3 deficiency, 613860
FECH	104,2	100%	100%	Protoporphyria, erythropoietic, autosomal recessive, 177000

FGA	171,1	100%	99%	Dysfibrinogenemia, alpha type, causing bleeding diathesis Dysfibrinogenemia, alpha type, causing recurrent thrombosis Amyloidosis, hereditary renal, 105200 Afibrinogenemia, congenital, 202400
FGB	103,6	100%	98%	Dysfibrinogenemia, beta type Afibrinogenemia, congenital, 202400 Thrombophilia, dysfibrinogenemic
FGD1	88,6	99%	95%	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400
FGD4	121,6	100%	100%	Charcot-Marie-Tooth disease, type 4H, 609311
FGF10	110,7	100%	100%	Aplasia of lacrimal and salivary glands, 180920 LADD syndrome, 149730
FGF14	105,1	100%	100%	Spinocerebellar ataxia 27, 609307
FGF16	118,5	100%	99%	Metacarpal 4-5 fusion, 609630
FGF17	95,9	100%	100%	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
FGF23	82,6	96%	91%	Hypophosphatemic rickets, autosomal dominant, 193100 Osteomalacia, tumor-induced (1) Tumoral calcinosis, hyperphosphatemic, familial, 211900
FGF3	88,2	100%	98%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGF8	50,6	80%	66%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGF9	149,2	100%	100%	Multiple synostoses syndrome 3, 612961
FGFR1	113,9	99%	96%	Pfeiffer syndrome, 101600 Jackson-Weiss syndrome, 123150 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Osteoglophonic dysplasia, 166250 Trigonocephaly 1, 190440 Hartsfield syndrome, 615465

FGFR2	121,7	100%	100%	Crouzon syndrome, 123500 Jackson-Weiss syndrome, 123150 Beare-Stevenson cutis gyrata syndrome, 123790 Pfeiffer syndrome, 101600 Apert syndrome, 101200 Saethre-Chotzen syndrome, 101400 Craniosynostosis, nonspecific Gastric cancer, somatic, 613659 Craniofacial-skeletal-dermatologic dysplasia, 101600 Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Scaphocephaly and Axenfeld-Rieger anomaly LADD syndrome, 149730 Scaphocephaly, maxillary retrusion, and mental retardation, 609579 Bent bone dysplasia syndrome, 614592
FGFR3	73,2	95%	88%	Achondroplasia, 100800 Hypochondroplasia, 146000 Thanatophoric dysplasia, type I, 187600 Crouzon syndrome with acanthosis nigricans, 612247 Muenke syndrome, 602849 Bladder cancer, somatic, 109800 Colorectal cancer, somatic, 114500 Cervical cancer, somatic, 603956 LADD syndrome, 149730 CATSHL syndrome, 610474 Nevus, epidermal, somatic, 162900 Thanatophoric dysplasia, type II, 187601 Spermatocytic seminoma, somatic, 273300
FGG	114,1	100%	98%	Dysfibrinogenemia, gamma type Hypofibrinogenemia, gamma type Thrombophilia, dysfibrinogenemic
FH	85,3	96%	89%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FHL1	79,5	97%	91%	Hemophagocytic lymphohistiocytosis, familial, 1 (2)

FIG4	124	98%	96%	Charcot-Marie-Tooth disease, type 4J, 611228 Amyotrophic lateral sclerosis 11, 612577 Yunis-Varon syndrome, 216340
FIGLA	81,3	93%	84%	Premature ovarian failure 6, 612310
FKBP10	79,3	100%	99%	Osteogenesis imperfecta, type XI, 610968
FKBP14	124,6	100%	100%	Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss, 614557
FKRP	80,2	99%	98%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155
FKTN	112,3	100%	99%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588
FLCN	108,9	100%	97%	Birt-Hogg-Dube syndrome, 135150 Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700 Colorectal cancer, somatic, 114500
FLG	44,1	98%	81%	Ichthyosis vulgaris, 146700 {Dermatitis, atopic, susceptibility to, 2}, 605803
FLNA	116,1	100%	99%	Heterotopia, periventricular, 300049 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Frontometaphyseal dysplasia, 305620 Heterotopia, periventricular, ED variant, 300537 FG syndrome 2, 300321 Cardiac valvular dysplasia, X-linked, 314400 Terminal osseous dysplasia, 300244 Congenital short bowel syndrome, 300048

FLNB	86,5	99%	97%	Spondylocarpotarsal synostosis syndrome, 272460 Larsen syndrome, 150250 Atelosteogenesis, type I, 108720 Atelosteogenesis, type III, 108721 Boomerang dysplasia, 112310
FLNC	93,4	96%	94%	Myopathy, myofibrillar, 5, 609524 Myopathy, distal, 4, 614065 -3
FLRT3	195,1	100%	100%	Hypogonadotropic hypogonadism 21 with anosmia, 615271
FLT3	98,9	98%	94%	Leukemia, acute myeloid, reduced survival in Leukemia, acute myeloid, 601626 Leukemia, acute lymphoblastic
FLT4	92,1	99%	99%	Lymphedema, hereditary I, 153100 Hemangioma, capillary infantile, somatic, 602089
FLVCR1	88,9	100%	100%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FLVCR2	137,4	100%	100%	Proliferative vasculopathy and hydraencephaly-hydrocephaly syndrome, 225790
FMO3	105,4	98%	96%	Trimethylaminuria, 602079
FMR1	109,5	100%	100%	Fragile X syndrome, 300624 Fragile X tremor/ataxia syndrome, 300623 Premature ovarian failure 1, 311360
FN1	89	99%	95%	Glomerulopathy with fibronectin deposits 2, 601894 Plasma fibronectin deficiency, 614101 (1)
FOLR1	79,3	96%	89%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXC1	44,2	100%	88%	Iridogoniodysgenesis, type 1, 601631 Rieger or Axenfeld anomalies, 602482 Axenfeld-Rieger syndrome, type 3, 602482 Iris hypoplasia and glaucoma, 601631
FOXC2	83,3	100%	98%	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXE3	17,1	61%	38%	Anterior segment mesenchymal dysgenesis, 107250 Aphakia, congenital primary, 610256

FOXF1	108,5	100%	97%	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380
FOXG1	86,2	84%	78%	Rett syndrome, congenital variant, 613454
FOXI1	101,7	100%	100%	Enlarged vestibular aqueduct, 600791
FOXL2	82,2	100%	100%	Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 Premature ovarian failure 3, 608996
FOXN1	113,5	100%	96%	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXP1	99,9	99%	96%	Mental retardation with language impairment and autistic features, 613670
FOXP2	111,1	100%	100%	Speech-language disorder-1, 602081
FOXP3	69,7	98%	91%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790 {Diabetes mellitus, type I, susceptibility to}, 222100
FOXRED1	95,4	100%	92%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
FRAS1	96,2	98%	95%	Fraser syndrome, 219000
FREM1	107,3	99%	99%	Bifid nose with or without anorectal and renal anomalies, 608980 Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485
FREM2	126,5	100%	98%	Fraser syndrome, 219000
FRMD7	117,6	100%	100%	Nystagmus 1, congenital, X-linked, 310700 Nystagmus, infantile periodic alternating, X-linked, 310700
FSCN2	78,2	100%	99%	Retinitis pigmentosa 30, 607921
FSHB	96	100%	100%	Follicle-stimulating hormone deficiency, isolated, 229070
FSHR	89,9	100%	97%	Ovarian dysgenesis 1, 233300 Ovarian response to FSH stimulation, 276400 Ovarian hyperstimulation syndrome, 608115
FTCD	57,3	87%	80%	Glutamate formiminotransferase deficiency, 229100

FTL	75,6	96%	91%	Hyperferritinemia-cataract syndrome, 600886 Neurodegeneration with brain iron accumulation 3, 606159
FTO	109,4	97%	97%	Growth retardation, developmental delay, coarse facies, and early death, 612938
FTSJ1	89,4	92%	88%	Mental retardation, X-linked 9, 309549
FUCA1	80,4	100%	99%	Fucosidosis, 230000
FUS	70,7	98%	93%	Amyotrophic lateral sclerosis 6, autosomal recessive, with or without frontotemporal dementia, 608030 Tremor, hereditary essential, 4, 614782
FUT6	74	84%	68%	Fucosyltransferase 6 deficiency, 613852
FUZ	84,9	100%	99%	Neural tube defects, 182940
FXN	83,2	88%	76%	Friedreich ataxia, 229300 Friedreich ataxia with retained reflexes, 229300
FXYD2	61,4	93%	88%	Hypomagnesemia-2, renal, 154020
FYCO1	87,2	100%	98%	Cataract 18, autosomal recessive, 610019
FZD4	146,7	100%	100%	Exudative vitreoretinopathy, 133780 Retinopathy of prematurity, 133780
FZD6	146,9	100%	100%	Nail disorder, nonsyndromic congenital, 10, (claw-shaped nails), 614157
G6PC	139,6	100%	100%	Glycogen storage disease Ia, 232200
G6PC3	115,8	100%	99%	Neutropenia, severe congenital 4, autosomal recessive, 612541 Dursun syndrome, 612541
G6PD	100,2	95%	95%	Hemolytic anemia due to G6PD deficiency Favism, 134700 {Resistance to malaria due to G6PD deficiency}, 611162
GAA	97,3	100%	98%	Glycogen storage disease II, 232300
GABRA1	139,7	99%	97%	{Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136 {Epilepsy, childhood absence, susceptibility to, 4}, 611136
GABRB3	116,3	100%	94%	Insomnia {Epilepsy, childhood absence, susceptibility to, 5}, 612269

GABRG2	134,8	99%	93%	Epilepsy, generalized, with febrile seizures plus, type 3, 611277 {Epilepsy, childhood absence, susceptibility to, 2}, 607681 Febrile seizures, familial, 8, 611277
GAD1	91,4	100%	98%	Cerebral palsy, spastic quadriplegic, 1, 603513
GALC	99,5	100%	98%	Krabbe disease, 245200
GALE	116,4	100%	100%	Galactose epimerase deficiency, 230350
GALK1	93,7	100%	97%	Galactokinase deficiency with cataracts, 230200
GALNS	65,5	92%	91%	Mucopolysaccharidosis IVA, 253000
GALNT3	106,2	100%	100%	Tumoral calcinosis, hyperphosphatemic, familial, 211900
GALT	113,7	100%	100%	Galactosemia, 230400
GAMT	89,1	96%	89%	Cerebral creatine deficiency syndrome 2, 612736
GAN	128,2	100%	99%	Giant axonal neuropathy-1, 256850
GATA1	118,2	99%	97%	Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367 Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 Thrombocytopenia with beta-thalassemia, X-linked, 314050 Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835
GATA2	96,6	96%	91%	Dendritic cell, monocyte, B lymphocyte, and natural killer lymphocyte deficiency, 614172 Emberger syndrome, 614038 {Myelodysplastic syndrome, susceptibility to}, 614286 {Leukemia, acute myeloid, susceptibility to}, 601626
GATA3	124,5	100%	97%	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GATA4	55	82%	67%	Atrial septal defect 2, 607941 Ventricular septal defect 1, 614429 Atrioventricular septal defect 4, 614430

GATA6	62,5	89%	80%	Atrioventricular septal defect 5, 614474 Atrial septal defect 9, 614475 Pancreatic agenesis and congenital heart defects, 600001 Persistent truncus arteriosus, 217095 Tetralogy of Fallot, 187500
GATAD1	73,5	91%	88%	Cardiomyopathy, dilated, 2B, 614672
GATAD2B	109,9	100%	97%	Mental retardation, autosomal dominant 18, 615074
GATM	81,7	98%	89%	Cerebral creatine deficiency syndrome 3, 612718
GBA	65,2	65%	62%	Gaucher disease, type I, 230800 Gaucher disease, type II, 230900 Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 Gaucher disease, perinatal lethal, 608013 {Parkinson disease, late-onset, susceptibility to}, 168600 {Lewy body dementia, susceptibility to}, 127750
GBA2	119,1	100%	100%	Spastic paraplegia 46, autosomal recessive, 614409
GBE1	104,1	99%	94%	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GCDH	78,5	92%	87%	Glutaricaciduria, type I, 231670
GCH1	86,3	100%	100%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GCK	85,3	100%	99%	MODY, type II, 125851 Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, gestational, 125851 Hyperinsulinemic hypoglycemia, familial, 3, 602485 Diabetes mellitus, permanent neonatal, 606176
GCLC	127,1	100%	100%	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 {Myocardial infarction, susceptibility to}, 608446
GCNT2	152,6	100%	100%	[Blood group, Ii], 110800 Cataract 13 with adult i phenotype, 110800 Adult i phenotype without cataract, 110800

GCSH	14,5	52%	37%	Glycine encephalopathy, 605899
GDAP1	102,1	100%	100%	Charcot-Marie-Tooth disease, type 4A, 214400 Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706 Charcot-Marie-Tooth disease, axonal, type 2K, 607831 Charcot-Marie-Tooth disease, recessive intermediate, A, 608340
GDF1	25,2	79%	60%	Double-outlet right ventricle, 217095 Tetralogy of Fallot, 187500 Transposition of great arteries, dextro-looped 3, 613854 Right atrial isomerism, 208530
GDF2	135,9	100%	100%	Telangiectasia, hereditary hemorraghic, type 5, 615506
GDF3	122,9	100%	100%	Klippel-Feil syndrome 3, autosomal dominant, 613702 Microphthalmia with coloboma 6, 613703 Microphthalmia, isolated 7, 613704
GDF5	96	100%	100%	Acromesomelic dysplasia, Hunter-Thompson type, 201250 Brachydactyly, type C, 113100 Chondrodysplasia, Grebe type, 200700 Du Pan syndrome, 228900 Brachydactyly, type A2, 112600 Symphalangism, proximal, 1B, 615298 Multiple synostoses syndrome 2, 610017 {Osteoarthritis-5}, 612400 Brachydactyly, type A1, C, 615072
GDF6	143,2	100%	100%	Klippel-Feil syndrome 1, autosomal dominant, 118100 Microphthalmia, isolated 4, 613094 Microphthalmia with coloboma 6, digenic, 613703 Leber congenital amaurosis 17, 615360
GDI1	139,8	100%	100%	Mental retardation, X-linked 41, 300849
GDNF	146,5	100%	97%	Central hypoventilation syndrome, 209880 {Pheochromocytoma, modifier of}, 171300 {Hirschsprung disease, susceptibility to, 3}, 613711
GFAP	77,8	100%	96%	Alexander disease, 203450

GFER	63,7	99%	93%	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076
GFI1	71,2	100%	98%	Neutropenia, severe congenital 2, autosomal dominant, 613107 Neutropenia, nonimmune chronic idiopathic, of adults, 607847
GFI1B	103	100%	95%	Bleeding disorder, platelet-type,17, 187900
GFM1	110,2	100%	100%	Combined oxidative phosphorylation deficiency 1, 609060
GFPT1	99,5	99%	96%	Myasthenia, congenital, with tubular aggregates 1, 610542
GGCX	92,6	100%	97%	Vitamin K-dependent coagulation defect, 277450 Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842
GH1	59,2	67%	67%	Growth hormone deficiency, isolated, type IA, 262400 Growth hormone deficiency, isolated, type IB, 612781 Growth hormone deficiency, isolated, type II, 173100 Kowarski syndrome, 262650
GHR	122,3	99%	99%	Laron dwarfism, 262500 Short stature, 604271 {Hypercholesterolemia, familial, modification of}, 143890 Increased responsiveness to growth hormone
GHRHR	93,1	100%	99%	Growth hormone deficiency, isolated, type IB, 612781
GHSR	122,1	100%	100%	Short stature, 604271
GIF	102	100%	99%	Intrinsic factor deficiency, 261000
GIGYF2	108,3	100%	98%	Parkinson disease 11, 607688
GIPC3	111,9	98%	91%	Deafness, autosomal recessive 15, 601869
GJA1	66	85%	73%	Oculodentodigital dysplasia, 164200 Syndactyly, type III, 186100 Hypoplastic left heart syndrome 1, 241550 Atrioventricular septal defect 3, 600309 Oculodentodigital dysplasia, autosomal recessive, 257850 Craniometaphyseal dysplasia, autosomal recessive, 218400

GJA3	90,2	100%	93%	Cataract 14, multiple types, 601885
GJA5	123,4	100%	100%	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic, 108770
GJA8	110,4	100%	86%	Cataract 1, multiple types, 116200
GJB1	172,8	100%	100%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GJB2	174,6	100%	100%	Deafness, autosomal recessive 1A, 220290 Deafness, autosomal dominant 3A, 601544 Vohwinkel syndrome, 124500 Keratoderma, palmoplantar, with deafness, 148350 Keratitis-ichthyosis-deafness syndrome, 148210 Hystrix-like ichthyosis with deafness, 602540 Bart-Pumphrey syndrome, 149200
GJB3	128,3	100%	100%	Erythrokeratodermia variabilis et progressiva, 133200 Deafness, autosomal dominant 2B, 612644 Deafness, autosomal recessive Deafness, autosomal dominant, with peripheral neuropathy Deafness, digenic, GJB2/GJB3, 220290
GJB4	137,7	100%	100%	Erythrokeratodermia variabilis with erythema gyratum repens, 133200
GJB6	141,5	100%	100%	Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290 Ectodermal dysplasia 2, Clouston type, 129500

GJC2	52,1	92%	82%	Leukodystrophy, hypomyelinating, 2, 608804 Spastic paraplegia 44, autosomal recessive, 613206 Lymphedema, hereditary, IC, 613480
GK	48	89%	79%	Glycerol kinase deficiency, 307030
GLA	92,4	100%	98%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	79,6	98%	94%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLDC	57,6	97%	85%	Glycine encephalopathy, 605899
GLE1	101,6	96%	94%	Lethal congenital contracture syndrome 1, 253310 Arthrogryposis, lethal, with anterior horn cell disease, 611890
GLI2	104,9	100%	95%	Holoprosencephaly-9, 610829
GLI3	118	100%	100%	Greig cephalopolysyndactyly syndrome, 175700 Pallister-Hall syndrome, 146510 Polydactyly, preaxial, type IV, 174700 Polydactyly, postaxial, types A1 and B, 174200 {Hypothalamic hamartomas, somatic}, 241800
GLIS2	92,8	100%	96%	Nephronophthisis 7, 611498
GLIS3	88,1	100%	99%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199 -3
GLRA1	112,6	100%	99%	Hyperekplexia, hereditary 1, autosomal dominant or recessive, 149400
GLRB	122,5	100%	98%	Hyperekplexia 2, autosomal recessive, 614619
GLRX5	39,1	81%	62%	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive, 205950
GLUD1	110,4	88%	88%	Hyperinsulinism-hyperammonemia syndrome, 606762
GLUL	30,2	70%	55%	Glutamine deficiency, congenital, 610015

GLYCTK	82,9	100%	97%	D-glyceric aciduria, 220120
GM2A	125,2	100%	100%	GM2-gangliosidosis, AB variant, 272750
GMPPB	118,4	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351
GMPS	107,3	100%	97%	Leukemia, acute myelogenous, 601626
GNA11	86,6	100%	99%	Hypocalciuric hypercalcemia, type II, 145981 Hypocalcemia, autosomal dominant 2, 615361
GNAI2	83,5	100%	99%	GNAI2L Pituitary ACTH-secreting adenoma Ventricular tachycardia, idiopathic, 192605
GNAI3	105,3	100%	99%	Auriculocondylar syndrome 1, 602483
GNAL	94,8	100%	100%	Dystonia 25, 615073
GNAO1	112,3	100%	98%	Epileptic encephalopathy, early infantile, 17, 615473
GNAQ	63,2	96%	93%	Sturge-Weber syndrome, somatic, mosaic, 185300 Capillary malformations, congenital, 1, somatic, mosaic, 163000
GNAS	126,8	100%	98%	Pseudohypoparathyroidism Ia, 103580 McCune-Albright syndrome, 174800 Pseudohypoparathyroidism Ic, 612462 Osseous heteroplasia, progressive, 166350 Pseudohypoparathyroidism Ib, 603233 Prolonged bleeding time, brachydactyly and mental retardation Acromegaly, 102200 Pseudopseudohypoparathyroidism, 612463 Prolonged bleeding time, brachydactyly, and mental retardation ACTH-independent macronodular adrenal hyperplasia, 219080
GNAT1	84	98%	93%	Night blindness, congenital stationary, autosomal dominant 3, 610444
GNAT2	118,1	100%	97%	Achromatopsia-4, 613856
GNB4	136,3	100%	99%	Charcot-Marie-Tooth disease, dominant intermediate F, 615185

GNE	99,1	100%	99%	Sialuria, 269921 Inclusion body myopathy, autosomal recessive, 600737 Nonaka myopathy, 605820
GNMT	85,6	100%	99%	Glycine N-methyltransferase deficiency, 606664
GNPAT	122	100%	100%	Chondrodysplasia punctata, rhizomelic, type 2, 222765
GNPTAB	129,1	100%	100%	Mucolipidosis III alpha/beta, 252600 Mucolipidosis II alpha/beta, 252500
GNRH1	37,6	100%	96%	Hypogonadotropic hypogonadism 12 with or without anosmia, 614841
GNRHR	157,4	100%	100%	Hypogonadotropic hypogonadism 7 with or without anosmia, 146110 Fertile eunuch syndrome, 228300
GNS	80,1	97%	88%	Mucopolysaccharidosis type IIID, 252940
GOLGA5	120,2	100%	98%	Thyroid carcinoma, papillary, 188550
GORAB	154,2	100%	98%	Geroderma osteodysplasticum, 231070
GOSR2	107,8	100%	100%	Epilepsy, progressive myoclonic 6, 614018
GOT1	104,1	100%	100%	Aspartate aminotransferase, serum level of, QTL1, 614419
GP1BA	113,3	97%	95%	Bernard-Soulier syndrome, type A1 (recessive), 231200 {Nonarteritic anterior ischemic optic neuropathy, susceptibility to}, 258660 Bernard-Soulier syndrome, type A2 (dominant), 153670 von Willebrand disease, platelet-type, 177820
GP1BB	25,6	81%	58%	Bernard-Soulier syndrome, type B, 231200 Giant platelet disorder, isolated, 231200
GP6	106,4	99%	92%	Bleeding disorder, platelet-type, 11, 614201
GP9	44,5	95%	84%	Bernard-Soulier syndrome, type C, 231200

GPC3	93,2	100%	100%	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPC6	97	100%	100%	Omodysplasia 1, 258315
GPD1	72,9	100%	97%	Hypertriglyceridemia, transient infantile, 614480
GPD1L	100,8	100%	100%	Brugada syndrome 2, 611777
GPHN	119,9	100%	100%	Molybdenum cofactor deficiency, type C, 252150
GPI	91,2	100%	97%	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470
GPR143	47,3	90%	79%	Ocular albinism, type I, Nettleship-Falls type, 300500 Nystagmus 6, congenital, X-linked, 300814
GPR179	141,5	100%	99%	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
GPR56	88,5	100%	98%	Polymicrogyria, bilateral frontoparietal, 606854
GPR98	108,3	100%	98%	Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472
GPSM2	131,1	100%	100%	Chudley-McCullough syndrome, 604213
GRHL2	102	100%	99%	Deafness, autosomal dominant 28, 608641
GRHPR	90,4	91%	85%	Hyperoxaluria, primary, type II, 260000
GRIA3	98,2	100%	96%	Mental retardation, X-linked 94, 300699
GRIK2	110,1	100%	100%	Mental retardation, autosomal recessive, 6, 611092
GRIN1	83,3	99%	96%	Mental retardation, autosomal dominant 8, 614254
GRIN2A	131,9	100%	99%	Epilepsy with neurodevelopmental defects, 613971
GRIN2B	135,5	99%	98%	Mental retardation, autosomal dominant 6, 613970
GRIP1	91,7	99%	97%	Fraser syndrome, 219000

GRK1	96,8	100%	100%	Oguchi disease-2, 613411
GRM1	138,5	100%	97%	Spinocerebellar ataxia, autosomal recessive 13, 614831
GRM6	92,4	96%	92%	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270
GRN	114,2	100%	99%	Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706
GRXCR1	182,9	100%	100%	Deafness, autosomal recessive 25, 613285
GSC	52,5	100%	86%	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471
GSN	71,1	93%	86%	Amyloidosis, Finnish type, 105120
GSS	83,5	98%	97%	Hemolytic anemia due to glutathione synthetase deficiency, 231900 Glutathione synthetase deficiency, 266130
GTF2H5	89,9	100%	100%	Trichothiodystrophy, complementation group A, 601675
GUCA1A	64,6	65%	59%	Cone dystrophy-3, 602093 Cone-rod dystrophy 14, 602093
GUCA1B	118	100%	100%	Retinitis pigmentosa 48, 613827
GUCY1A3	152,2	100%	100%	Moyamoya 6 with achalasia, 615750
GUCY2C	91,8	100%	99%	Diarrhea 6, 614616 Meconium ileus, 614665
GUCY2D	79,4	99%	95%	Leber congenital amaurosis 1, 204000 Cone-rod dystrophy 6, 601777
GUSB	67,7	89%	80%	Mucopolysaccharidosis VII, 253220
GYG1	45,8	77%	54%	Glycogen storage disease XV, 613507
GYS1	67,4	96%	77%	Glycogen storage disease 0, muscle, 611556
GYS2	90,3	100%	100%	Glycogen storage disease, type 0, 240600

H6PD	120,6	100%	100%	Cortisone reductase deficiency 1, 604931
HADHA	92,9	93%	89%	LCHAD deficiency, 609016 Trifunctional protein deficiency, 609015 HELLP syndrome, maternal, of pregnancy, 609016 Fatty liver, acute, of pregnancy, 609016
HADHB	86,6	100%	100%	Trifunctional protein deficiency, 609015
HAMP	112	100%	99%	Hemochromatosis, type 2B, 613313
HARS	116	100%	98%	Usher syndrome type 3B, 614504
HARS2	131,7	100%	100%	Perrault syndrome 2, 614926
HAX1	129,3	100%	100%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HBA1	54,2	61%	43%	5'-zeta-pseudozeta-pseudoalpha-alpha-2-alpha-1-3' Thalassemias, alpha-, 604131 Methemoglobinemias, alpha- Erythremias, alpha- Heinz body anemias, alpha-, 140700 Hemoglobin H disease, nondeletional, 613978
HBA2	62,8	65%	53%	Thalassemia, alpha-, 604131 Heinz body anemia, 140700 Erythrocytosis Hypochromic microcytic anemia Hemoglobin H disease, nondeletional, 613978

HBB	131,5	100%	100%	Sickle cell anemia, 603903 Thalassemias, beta-, 613985 Erythremias, beta- Methemoglobinemias, beta- Heinz body anemias, beta-, 140700 Thalassemia-beta, dominant inclusion-body, 603902 Hereditary persistence of fetal hemoglobin, 141749 Delta-beta thalassemia, 141749 {Malaria, resistance to}, 611162
HBD	188,7	100%	100%	Thalassemia, delta- Thalassemia due to Hb Lepore
HBG1	13,9	30%	26%	Fetal hemoglobin quantitative trait locus 1, 141749
HBG2	36,1	79%	74%	Fetal hemoglobin quantitative trait locus 1, 141749 Cyanosis, transient neonatal, 613977
HCCS	107,1	100%	99%	Microphthalmia, syndromic 7, 309801
HCFC1	72,6	99%	94%	Mental retardation, X-linked 3, 309541
HCN4	61,7	100%	97%	Sick sinus syndrome 2, 163800 Brugada syndrome 8, 613123
HCRT	55,8	85%	76%	Narcolepsy 1, 161400
HDAC4	65,3	93%	90%	Brachydactyly-mental retardation syndrome, 600430
HDAC6	113,1	95%	94%	Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863
HDAC8	96,8	100%	99%	Wilson-Turner syndrome, 309585 Cornelia de Lange syndrome 5, 300882
HEATR2	69,8	91%	80%	Ciliary dyskinesia, primary, 18, 614874
HEPACAM	65	83%	78%	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926

HERC2	62,1	63%	59%	[Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220
HES7	39,6	96%	77%	Spondylocostal dysostosis 4, autosomal recessive, 613686
HESX1	94,7	100%	95%	Septooptic dysplasia, 182230 Pituitary hormone deficiency, combined, 5, 182230 Growth hormone deficiency with pituitary anomalies, 182230
HEXA	91,9	100%	100%	Tay-Sachs disease, 272800 GM2-gangliosidosis, several forms, 272800 [Hex A pseudodeficiency], 272800
HEXB	105,5	100%	100%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HFE	93,1	100%	94%	Hemochromatosis, 235200 {Microvascular complications of diabetes 7}, 612635 {Porphyria variegata, susceptibility to}, 176200 {Porphyria cutanea tarda, susceptibility to}, 176100 {Alzheimer disease, susceptibility to}, 104300 [Transferrin serum level QTL2], 614193
HFM1	101,2	99%	97%	Split hand/foot malformation 1 (4)
HGD	85,6	100%	100%	Alkaptonuria, 203500
HGF	103,3	100%	100%	Deafness, autosomal recessive 39, 608265
HGSNAT	99,2	93%	93%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930
HIBCH	61,5	100%	98%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HINT1	73,9	95%	81%	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200
HK1	106,9	100%	98%	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285
HLCS	140,4	100%	100%	Holocarboxylase synthetase deficiency, 253270
HMBS	88,3	100%	96%	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000

HMGCL	102,4	100%	98%	HMG-CoA lyase deficiency, 246450
HMGCS2	112,5	100%	100%	HMG-CoA synthase-2 deficiency, 605911
HMOX1	64	100%	93%	Heme oxygenase-1 deficiency, 614034 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963
HMX1	32,5	97%	80%	Oculoauricular syndrome, 612109
HNF1A	85,2	100%	95%	MODY, type III, 600496 {Diabetes mellitus, noninsulin-dependent, 2}, 125853 {Diabetes mellitus, insulin-dependent}, 222100 Hepatic adenoma, somatic, 142330 Renal cell carcinoma, 144700 Diabetes mellitus, insulin-dependent, 20, 612520
HNF1B	77,9	98%	94%	Renal cysts and diabetes syndrome, 137920 Diabetes mellitus, noninsulin-dependent, 125853 {Renal cell carcinoma}, 144700
HNF4A	73,2	100%	96%	MODY, type I, 125850 {Diabetes mellitus, noninsulin-dependent}, 125853
HNRNPA1	41,7	94%	79%	?Inclusion body myopathy wtih early-onset Paget disease without frontotemporal dementia 3, 615424 Amyotrophic lateral sclerosis 19, 615426
HOGA1	71,3	97%	93%	Hyperoxaluria, primary, type III, 613616
HOXA1	126	100%	100%	Bosley-Salih-Alorainy syndrome, 601536 Athabaskan brainstem dysgenesis syndrome, 601536
HOXA11	115,9	100%	100%	Radioulnar synostosis with amegakaryocytic thrombocytopenia, 605432 -3
HOXA13	54,1	92%	69%	Hand-foot-uterus syndrome, 140000 Guttmacher syndrome, 176305 -3
HOXB1	96,4	100%	100%	Facial paresis, hereditary congenital, 3
HOXC13	83,2	100%	98%	Ectodermal dysplasia 9, hair/nail type, 614931
HOXD10	153,2	100%	100%	Vertical talus, congenital, 192950 Charcot-Marie-Tooth disease, foot deformity of, 192950

HOXD13	103	100%	94%	Synpolydactyly, type II, 186000 Brachydactyly, type E, 113300 Brachydactyly, type D, 113200 Synpolydactyly with foot anomalies, 186000 Syndactyly, type V, 186300 Brachydactyly-syndactyly syndrome, 610713 VACTERL association, 192350
HPD	101,1	100%	98%	Tyrosinemia, type III, 276710 Hawkinsinuria, 140350
HPGD	71,6	99%	90%	Cranioosteoarthropathy, 259100 Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100 Digital clubbing, isolated congenital, 119900
HPRT1	96,2	100%	98%	Lesch-Nyhan syndrome, 300322 HPRT-related gout, 300323
HPS1	73,3	99%	90%	Hermansky-Pudlak syndrome 1, 203300
HPS3	109,9	100%	98%	Hermansky-Pudlak syndrome 3, 614072
HPS4	109,1	100%	98%	Hermansky-Pudlak syndrome 4, 614073
HPS5	97,5	100%	99%	Hermansky-Pudlak syndrome 5, 614074
HPS6	88,1	100%	82%	Hermansky-Pudlak syndrome 6, 614075
HPSE2	75,1	100%	99%	Urofacial syndrome 1, 236730
HR	81,7	99%	94%	Alopecia universalis, 203655 Atrichia with papular lesions, 209500 Hypotrichosis, hereditary, Marie Unna type, 1, 146550
HRAS	88,1	100%	100%	{Bladder cancer, somatic}, 109800 Costello syndrome, 218040 {Thyroid carcinoma, follicular, somatic}, 188470 Congenital myopathy with excess of muscle spindles, 218040 {Nevus sebaceous, somatic}, 162900 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200

HRG	136,6	96%	93%	Thrombophilia due to HRG deficiency, 613116 Thrombophilia due to elevated HRG, 613116 (1)
HSD11B1	107,1	100%	98%	Cortisone reductase deficiency 2, 614662
HSD11B2	110,6	77%	75%	Apparent mineralocorticoid excess, 218030
HSD17B10	105,3	99%	92%	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 Mental retardation, X-linked syndromic 10, 300220 Mental retardation, X-linked 17/31, microduplication, 300705
HSD17B3	97,4	100%	100%	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	91,8	100%	98%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B2	41	88%	72%	3-beta-hydroxysteroid dehydrogenase, type II, deficiency, 201810
HSD3B7	71,9	86%	80%	Bile acid synthesis defect, congenital, 1, 607765
HSF4	99	99%	96%	Cataract 5, multiple types, 116800
HSPB1	44,6	94%	82%	Neuropathy, distal hereditary motor, type IIB, 608634 Charcot-Marie-Tooth disease, axonal, type 2F, 606595
HSPB3	168	100%	100%	Neuronopathy, distal hereditary motor, type IIC, 613376
HSPB8	103,6	100%	100%	Neuropathy, distal hereditary motor, type IIA, 158590 Charcot-Marie-Tooth disease, axonal, type 2L, 608673
HSPD1	16,8	59%	40%	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233
HSPG2	75,4	99%	95%	Schwartz-Jampel syndrome, type 1, 255800 Dyssegmental dysplasia, Silverman-Handmaker type, 224410
HTR1A	118,2	100%	100%	Periodic fever, menstrual cycle dependent, 614674
HTRA1	72,3	85%	80%	{Macular degeneration, age-related, 7}, 610149 {Macular degeneration, age-related, neovascular type}, 610149 CARASIL syndrome, 600142
HTRA2	133,9	100%	98%	Parkinson disease 13, 610297

HTT	96,3	98%	96%	Huntington disease, 143100
HUWE1	95,7	99%	97%	Mental retardation, X-linked syndromic, Turner type, 300706
HYAL1	87,5	99%	96%	Mucopolysaccharidosis type IX, 601492
HYDIN	92,7	88%	85%	Ciliary dyskinesia, primary, 5, 608647
HYLS1	153,3	100%	100%	Hydrolethalus syndrome, 236680
ICK	102,1	100%	99%	Endocrine-cerebroosteodysplasia, 612651
ICOS	125,5	100%	100%	Immunodeficiency, common variable, 1, 607594
IDH2	107,5	100%	99%	D-2-hydroxyglutaric aciduria 2, 613657
IDH3B	124	95%	95%	Retinitis pigmentosa 46, 612572
IDS	101,7	90%	86%	Mucopolysaccharidosis II, 309900
IDUA	81	95%	89%	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Is, 607016 Mucopolysaccharidosis Ih/s, 607015
IER3IP1	58,5	100%	93%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFITM5	89,6	100%	92%	Osteogenesis imperfecta, type V, 610967
IFNGR1	141,9	100%	100%	Mycobacterial infection, atypical, familial disseminated, 209950 BCG infection, generalized familial, 209950 {H. pylori infection, susceptibility to}, 600263 {Tuberculosis, susceptibility to}, 607948 {Mycobacterium tuberculosis infection, protection against}, 607948 {Hepatitis B virus infection, susceptibility to}, 610424
IFT122	77,2	96%	95%	Cranioectodermal dysplasia 1, 218330
IFT140	83,8	99%	95%	Mainzer-Saldino syndrome, 266920

IFT172	97,4	100%	97%	Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 Mainzer-Saldino syndrome (Halbritter (2013) Am J Hum Genet 93, 915) Asphyxiating thoracic dystrophy with or without Joubert Syndrome (Halbritter (2013) Am J Hum Genet 93, 915)
IFT43	81,8	100%	100%	Cranioectodermal dysplasia 3, 614099
IFT80	79,9	98%	93%	Asphyxiating thoracic dystrophy 2, 611263
IGBP1	93,4	94%	88%	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472
IGF1	117,8	100%	96%	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IGF1R	102,5	100%	99%	Insulin-like growth factor I, resistance to, 270450
IGF2R	93,1	98%	94%	Hepatocellular carcinoma
IGFALS	68,8	100%	96%	Acid-labile subunit, deficiency of
IGFBP7	33,3	70%	44%	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis, 614224
IGHMBP2	74,3	99%	93%	Neuronopathy, distal hereditary motor, type VI, 604320
IGLL1	20,7	72%	35%	Agammaglobulinemia 2, 613500
IGSF1	119,6	99%	97%	Hypothyroidism, central, and testicular enlargement, 300888
IHH	98,8	100%	99%	Acrocapitofemoral dysplasia, 607778 Brachydactyly, type A1, 112500
IKBKAP	103,4	100%	99%	Dysautonomia, familial, 223900
IKBKG	18,8	20%	20%	Incontinentia pigmenti, type II, 308300 Ectodermal dysplasia, hypohidrotic, with immune deficiency, 300291 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 Immunodeficiency, isolated, 300584 {Atypical mycobacteriosis, familial}, 300636 Invasive pneumococcal disease, recurrent isolated, 2, 300640
IKZF1	104	100%	99%	Leukemia, acute lymphoblastic

IL10RA	97	100%	98%	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL11RA	90,3	98%	93%	Craniosynostosis and dental anomalies, 614188
IL17F	91,9	100%	95%	Candidiasis, familial, 6, autosomal dominant, 613956
IL17RA	87,2	98%	89%	Candidiasis, familial, 5, autosomal recessive, 613953
IL17RD	101,4	100%	97%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
IL1RAPL1	135,7	100%	100%	Mental retardation, X-linked 21/34, 300143
IL1RN	106	100%	100%	{Gastric cancer risk after H. pylori infection}, 137215 {Microvascular complications of diabetes 4}, 612628 Interleukin 1 receptor antagonist deficiency, 612852
IL21R	110,7	100%	99%	[IgE, elevated level of], 147050 Immunodeficiency, primary, autosomal recessive, IL21R-related, 615207
IL2RA	98,1	100%	100%	Interleukin-2 receptor, alpha chain, deficiency of, 606367 {Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942
IL2RG	93,2	100%	95%	Severe combined immunodeficiency, X-linked, 300400 Combined immunodeficiency, X-linked, moderate, 312863
IL31RA	117	100%	97%	Amyloidosis, primary localized cutaneous, 2, 613955
IL36RN	89	100%	100%	Psoriasis, generalized pustular, 614204
IL7R	88,9	100%	95%	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
ILDR1	54	100%	99%	Deafness, autosomal recessive 42, 609646
IMPAD1	124,9	100%	100%	Chondrodysplasia with joint dislocations, GRAPP type, 614078
IMPDH1	36,7	73%	61%	Retinitis pigmentosa 10, 180105 Leber congenital amaurosis 11, 613837
IMPG2	119,9	99%	96%	Retinitis pigmentosa 56, 613581 Maculopathy, IMPG2-related, 613581
INF2	67	93%	87%	Glomerulosclerosis, focal segmental, 5, 613237 Charcot-Marie-Tooth disease, dominant intermediate E, 614455

ING1	118,6	100%	98%	Squamous cell carcinoma, head and neck, somatic, 275355
INPP5E	68,4	98%	94%	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300
INPPL1	92,4	97%	95%	Opsismodysplasia, 258480
INS	40,3	100%	92%	Hyperproinsulinemia, familial, with or without diabetes Maturity-onset diabetes of the young, type 10, 613370 Diabetes mellitus, permanent neonatal, 606176 Diabetes mellitus, type 1, 125852 Diabetes mellitus, insulin-dependent, 2, 125852
INSL3	48,5	89%	80%	Cryptorchidism, 219050
INSR	117,8	96%	92%	Leprechaunism, 246200 Rabson-Mendenhall syndrome, 262190 Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968
INVS	115,2	100%	98%	Nephronophthisis 2, infantile, 602088
IQCB1	86,2	95%	91%	Senior-Loken syndrome 5, 609254
IQSEC2	77,3	94%	85%	Mental retardation, X-linked 1, 309530
IRAK4	109,6	100%	100%	IRAK4 deficiency, 607676 Invasive pneumococcal disease, recurrent isolated, 1, 610799
IRF1	105,1	100%	100%	Myelodysplastic syndrome, preleukemic Myelogenous leukemia, acute Gastric cancer, somatic, 613659 Nonsmall cell lung cancer, somatic, 211980
IRF4	112,9	100%	99%	Multiple myeloma, 254500
IRF6	91	96%	93%	van der Woude syndrome, 119300 Popliteal pterygium syndrome 1, 119500 Orofacial cleft 6, 608864
IRF8	66,2	100%	98%	Monocyte and dendritic cell deficiency, recessive, 614894 CD11C+/CD1C+ dendritic cell deficiency, dominant, 614893

IRGM	148,4	100%	100%	{Mycobacterium tuberculosis, protection against}, 607948 Inflammatory bowel disease 19, 612278
IRX5	63,3	93%	84%	Hamamy syndrome, 611174
ISCU	91,6	100%	99%	Myopathy with lactic acidosis, hereditary, 255125
ISPD	75,3	96%	91%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
ITCH	101,5	95%	95%	Autoimmune disease, syndromic multisystem, 613385
ITGA2B	72,1	97%	91%	BAK platelet antigen Glanzmann thrombasthenia, 273800 Thrombocytopenia, neonatal alloimmune, BAK antigen related Bleeding disorder, platelet-type, 16, autosomal dominant, 187800
ITGA3	107,2	100%	92%	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748
ITGA6	124,4	99%	98%	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730 -3
ITGA7	85,2	98%	91%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
ITGB2	87,2	100%	99%	Leukocyte adhesion deficiency, 116920
ITGB3	90,6	100%	99%	PL(A) platelet antigen Glanzmann thrombasthenia, 273800 Thrombocytopenia, neonatal alloimmune {Myocardial infarction, susceptibility to}, 608446 Purpura, posttransfusion Bleeding disorder, platelet-type, 16, autosomal dominant, 187800
ITGB4	78,8	97%	94%	Epidermolysis bullosa, junctional, with pyloric atresia, 226730 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa of hands and feet, 131800
ITK	100,4	100%	100%	Lymphoproliferative syndrome 1, 613011
ITM2B	86	100%	100%	Dementia, familial British, 176500 Dementia, familial Danish, 117300
ITPR1	103,3	99%	98%	Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360
IVD	96,3	100%	98%	Isovaleric acidemia, 243500

IYD	82,1	100%	99%	Thyroid dyshormonogenesis 4, 274800
JAG1	104,8	97%	97%	Alagille syndrome, 118450 Tetralogy of Fallot, 187500 Deafness, congenital heart defects, and posterior embryotoxon
JAK2	109,7	100%	99%	Polycythemia vera, 263300 Thrombocythemia 3, 614521 Myelofibrosis, somatic, 254450 {Budd-Chiari syndrome}, 600880 Leukemia, acute myelogenous, 601626 Erythrocytosis, somatic, 133100
JAK3	84,9	98%	94%	SCID, autosomal recessive, T-negative/B-positive type, 600802
JAM3	71	95%	90%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
JPH2	73,4	99%	86%	Cardiomyopathy, familial hypertrophic 17, 613873
JPH3	105,5	97%	96%	Huntington disease-like 2, 606438
JUP	69,6	97%	91%	Naxos disease, 601214 Arrhythmogenic right ventricular dysplasia 12, 611528
KAL1	78,4	96%	91%	hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
KANK1	133,7	100%	99%	Cerebral palsy, spastic quadriplegic, 2, 612900
KANSL1	35,1	79%	64%	Koolen-De Vries syndrome, 610443
KARS	112,2	100%	100%	Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness, autosomal recessive 89, 613916
KAT6B	140,2	100%	100%	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KBTBD13	50,1	100%	91%	Nemaline myopathy 6, autosomal dominant, 609273
KCNA1	102,4	100%	100%	Episodic ataxia/myokymia syndrome, 160120
KCNA5	124,4	99%	93%	Atrial fibrillation, familial, 7, 612240

KCNC3	86	74%	66%	Spinocerebellar ataxia 13, 605259
KCND3	135	98%	96%	Spinocerebellar ataxia 19, 607346
KCNE1	203,1	100%	100%	Jervell and Lange-Nielsen syndrome 2, 612347 Long QT syndrome-5, 613695
KCNE2	140,8	100%	100%	Long QT syndrome-6, 613693 Atrial fibrillation, familial, 4, 611493
KCNE3	106,6	100%	100%	Brugada syndrome 6, 613119
KCNH2	63,9	97%	87%	Long QT syndrome-2, 613688 {Long QT syndrome-2, acquired, susceptibility to}, 613688 Short QT syndrome-1, 609620
KCNJ1	132,8	97%	97%	Bartter syndrome, type 2, 241200
KCNJ10	148,5	100%	100%	SESAME syndrome, 612780 Enlarged vestibular aqueduct, digenic, 600791
KCNJ11	127,2	100%	100%	Hyperinsulinemic hypoglycemia, familial, 2, 601820 Diabetes, permanent neonatal, 606176 Diabetes mellitus, permanent neonatal, with neurologic features, 606176 {Diabetes mellitus, type 2, susceptibility to}, 125853 Diabetes mellitus, transient neonatal, 3, 610582
KCNJ13	205,2	100%	100%	Snowflake vitreoretinal degeneration, 193230 Leber congenital amaurosis 16, 614186
KCNJ2	113,3	94%	91%	Andersen syndrome, 170390 Short QT syndrome-3, 609622 Atrial fibrillation, familial, 9, 613980
KCNJ5	171,7	100%	100%	Long QT syndrome 13, 613485 Hyperaldosteronism, familial, type III, 613677
KCNK3	106,2	97%	92%	Pulmonary hypertension, primary, 4, 615344
KCNMA1	72,9	94%	89%	Generalized epilepsy and paroxysmal dyskinesia, 609446
KCNQ1	64	91%	82%	Long QT syndrome-1, 192500 Jervell and Lange-Nielsen syndrome, 220400 Atrial fibrillation, familial, 3, 607554

				Short QT syndrome-2, 609621 {Long QT syndrome 1, acquired, susceptibility to}, 192500
KCNQ2	72,8	100%	98%	Seizures, benign neonatal, 1, 121200 Myokymia, 121200 Epileptic encephalopathy, early infantile, 7, 613720
KCNQ3	101,8	100%	96%	Seizures, benign neonatal, type 2, 121201
KCNQ4	106,8	90%	84%	Deafness, autosomal dominant 2A, 600101
KCNT1	74,8	100%	96%	Epileptic encephalopathy, early infantile, 14, 614959 Epilepsy, nocturnal frontal lobe, 5, 615005
KCNV2	78	100%	99%	Retinal cone dystrophy 3B, 610356
KCTD1	112,8	99%	96%	Scalp-ear-nipple syndrome, 181270
KCTD7	123	91%	84%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM5C	111,4	100%	100%	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534 -3
KDM6A	120	100%	99%	Kabuki syndrome 2, 300867
KDR	101,5	100%	99%	Hemangioma, capillary infantile, somatic, 602089 {Hemangioma, capillary infantile, susceptibility to}, 602089
KERA	134,6	100%	100%	Cornea plana congenita, recessive, 217300
KHDC3L	130,6	100%	100%	Hydatidiform mole, recurrent, 2, 614293
KIAA0196	97,1	98%	98%	Spastic paraplegia 8, autosomal dominant, 603563
KIAA1279	97,7	99%	97%	Goldberg-Shprintzen megacolon syndrome, 609460
KIAA2022	156,3	100%	100%	?Mental retardation, nonsyndromic, X-linked (2)
KIF11	95,9	100%	98%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF1A	69	99%	92%	Spastic paraplegia 30, autosomal recessive, 610357 Neuropathy, hereditary sensory, type IIC, 614213 Mental retardation, autosomal dominant 9, 614255

KIF1B	118,8	100%	100%	Charcot-Marie-Tooth disease, type 2A1, 118210 Pheochromocytoma, 171300 {Neuroblastoma, susceptibility to, 1}, 256700
KIF1C	108,8	100%	98%	Spastic ataxia 2, autosomal recessive, 611302
KIF21A	104,2	100%	99%	Fibrosis of extraocular muscles, congenital, 1, 135700 Fibrosis of extraocular muscles, congenital, 3B, 135700
KIF22	107,6	100%	100%	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546
KIF2A	111,6	100%	95%	Cortical dysplasia, complex, with other brain malformations 3, 615411
KIF5A	89,4	98%	95%	Spastic paraplegia 10, autosomal dominant, 604187
KIF7	65	93%	82%	Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990
KIRREL3	81,3	99%	96%	Mental retardation, autosomal dominant 4, 612581
KISS1	35,5	88%	74%	Hypogonadotropic hypogonadism 13 with or without anosmia, 614842
KISS1	35,5	88%	74%	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 Precocious puberty, central, 1, 176400
KISS1R	43,7	98%	87%	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 Precocious puberty, central, 1, 176400
KIT	106,6	98%	96%	Piebaldism, 172800 Gastrointestinal stromal tumor, familial, 606764 Mast cell disease, 154800 Leukemia, acute myeloid, 601626 Germ cell tumors, 273300
KITLG	73,9	100%	98%	[Skin/hair/eye pigmentation 7, blond/brown hair], 611664 Hyperpigmentation, familial progressive, 2, 145250
KL	132,5	99%	96%	{Coronary artery disease, susceptibility to} Tumoral calcinosis, hyperphosphatemic, 211900

KLF1	49,9	100%	95%	Blood group--Lutheran inhibitor, 111150 [Hereditary persistence of fetal hemoglobin], 613566 Anemia, dyserythropoietic congenital, type IV, 613673
KLF11	165,6	97%	97%	Maturity-onset diabetes of the young, type VII, 610508
KLF6	128,8	100%	100%	Prostate cancer, somatic, 176807 Gastric cancer, somatic, 613659
KLHDC8B	60,6	83%	73%	Hodgkin lymphoma, 236000
KLHL10	142,3	100%	98%	Spermatogenic failure 11, 615081
KLHL3	81,9	95%	92%	Pseudohypoaldosteronism, type IID, 614495
KLHL40	88,7	100%	100%	Nemaline myopathy 8, autosomal recessive, 615348
KLHL7	117,4	100%	100%	Retinitis pigmentosa 42, 612943
KLK4	143,3	100%	100%	Amelogenesis imperfecta, type IIA1, 204700
KLKB1	131,2	100%	100%	Fletcher factor deficiency, 612423
KLLN	109,6	100%	100%	Cowden syndrome 4, 615107

KRAS	63,6	98%	93%	Lung cancer, somatic, 211980 Bladder cancer, somatic, 109800 Pancreatic carcinoma, somatic, 260350 Gastric cancer, somatic, 137215 Leukemia, acute myelogenous Noonan syndrome 3, 609942 Cardiofaciocutaneous syndrome 2, 615278 Breast cancer, somatic, 114480 SFM syndrome, somatic mosaic, 163200

KRT1	100	100%	98%	Epidermolytic hyperkeratosis, 113800 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602 Ichthyosis histrix, Curth-Macklin type, 146590 Palmoplantar keratoderma, nonepidermolytic, 600962 Palmoplantar keratoderma, epidermolytic, 144200 Keratosis palmoplantaris striata III, 607654
KRT10	95,3	97%	88%	Epidermolytic hyperkeratosis, 113800 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602 Ichthyosis with confetti, 609165
KRT12	108	98%	94%	Meesmann corneal dystrophy, 122100
KRT13	86,9	100%	99%	White sponge nevus, 193900
KRT14	25,9	70%	50%	Epidermolysis bullosa simplex, Dowling-Meara type, 131760 Epidermolysis bullosa simplex, Koebner type, 131900 Epidermolysis bullosa simplex, recessive 1, 601001 Naegeli-Franceschetti-Jadassohn syndrome, 161000 Dermatopathia pigmentosa reticularis, 125595 Epidermolysis bullosa simplex, Weber-Cockayne type, 131800
KRT16	7,5	24%	3%	Pachyonychia congenita, Jadassohn-Lewandowsky type, 167200 Palmoplantar keratoderma, nonepidermolytic, focal, 613000
KRT17	11,2	48%	15%	Pachyonychia congenita, Jackson-Lawler type, 167210 Steatocystoma multiplex, 184500
KRT18	20,4	78%	35%	Cirrhosis, cryptogenic {Cirrhosis, noncryptogenic, susceptibility to}, 215600
KRT2	112,4	99%	97%	Ichthyosis bullosa of Siemens, 146800
KRT3	79,9	100%	99%	Meesmann corneal dystrophy, 122100
KRT4	83,7	100%	99%	White sponge nevus, 193900

KRT5	73,4	97%	92%	Epidermolysis bullosa simplex, Dowling-Meara type, 131760 Epidermolysis bullosa simplex, Koebner type, 131900 Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 Epidermolysis bullosa simplex with mottled pigmentation, 131960 Dowling-Degos disease 1, 179850 Epidermolysis bullosa simplex with migratory circinate erythema, 609352
KRT6A	32,6	68%	45%	Pachyonychia congenita, Jadassohn-Lewandowsky type, 167200
KRT6B	32,2	76%	52%	Pachyonychia congenita, Jackson-Lawler type, 167210
KRT6C	22	52%	36%	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735
KRT74	83,3	95%	88%	Woolly hair, autosomal dominant, 194300 Hypotrichosis simplex of the scalp 2, 613981
KRT8	29,9	87%	57%	Cirrhosis, cryptogenic {Cirrhosis, noncryptogenic, susceptibility to}, 215600
KRT81	19,9	61%	37%	Monilethrix, 158000
KRT83	23,6	63%	39%	Monilethrix, 158000
KRT85	34,1	78%	56%	Ectodermal dysplasia 4, hair/nail type, 602032
KRT86	24,2	59%	45%	Monilethrix, 158000
KRT9	106,7	98%	94%	Palmoplantar keratoderma, epidermolytic, 144200
L1CAM	120,7	100%	99%	Hydrocephalus due to aqueductal stenosis, 307000 MASA syndrome, 303350 CRASH syndrome, 303350 Hydrocephalus with Hirschsprung disease, 307000 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 Corpus callosum, partial agenesis of, 304100
L2HGDH	78,2	100%	96%	L-2-hydroxyglutaric aciduria, 236792
LAMA2	97,9	99%	97%	Muscular dystrophy, congenital merosin-deficient, 607855 Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855

LAMA3	97,9	99%	98%	Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, generalized atrophic benign, 226650 Laryngoonychocutaneous syndrome, 245660
LAMB1	112,1	100%	98%	Lissencephaly 5, 615191
LAMB2	107,7	100%	100%	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 Pierson syndrome, 609049
LAMB3	72,7	99%	95%	Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LAMC2	101,9	99%	97%	Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LAMC3	93,4	100%	93%	Cortical malformations, occipital, 614115
LAMP2	109,4	100%	99%	Danon disease, 300257
LAMTOR2	73,8	100%	100%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798 -3
LARGE	100,5	97%	92%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840
LARP7	99,6	100%	100%	Alazami syndrome, 615071
LARS2	108,5	100%	100%	Perrault syndrome 4, 615300
LBR	90,6	100%	99%	Pelger-Huet anomaly, 169400 HEM skeletal dysplasia, 215140 Reynolds syndrome, 613471
LCA5	142,4	100%	98%	Leber congenital amaurosis 5, 604537
LCAT	105	94%	88%	Norum disease, 245900 Fish-eye disease, 136120
LCT	128,5	99%	98%	Lactase deficiency, congenital, 223000
LDB3	85,9	95%	91%	Myopathy, myofibrillar, 4, 609452 Cardiomyopathy, dilated 1C, 601493 Left ventricular noncompaction 3, with or without dilated cardiomyopathy, 601493
LDHA	38,1	65%	57%	Glycogen storage disease XI, 612933

LDHB	76,8	100%	100%	Lactate dehydrogenase-B deficiency, 614128
LDLR	119,4	100%	98%	C3 Hypercholesterolemia, familial, 143890 LDL cholesterol level QTL2, 143890
LDLRAP1	82,9	96%	90%	Hypercholesterolemia, familial, autosomal recessive, 603813
LEF1	90,6	100%	100%	Sebaceous tumors, somatic
LEMD3	109,8	100%	99%	Osteopoikilosis, 166700 Buschke-Ollendorff syndrome, 166700 Melorheostosis with osteopoikilosis, 155950
LEP	103,8	100%	100%	Obesity, morbid, due to leptin deficiency, 614962
LEPR	121,6	94%	93%	Obesity, morbid, due to leptin receptor deficiency, 614963
LEPRE1	92	100%	99%	Osteogenesis imperfecta, type VIII, 610915
LEPREL1	65,6	93%	84%	Myopia, high, with cataract and vitreoretinal degeneration, 614292 -3
LFNG	56	80%	75%	Spondylocostal dysostosis, autosomal recessive 3, 609813
LGI1	133,6	100%	100%	Epilepsy, familial temporal lobe, 1, 600512
LHB	18,8	57%	53%	Hypogonadism, hypergonadotropic ?Male pseudohermaphroditism due to defective LH (1)
LHCGR	135,4	100%	93%	Precocious puberty, male, 176410 Leydig cell hypoplasia with pseudohermaphroditism, 238320 Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320 Luteinizing hormone resistance, female, 238320 Leydig cell adenoma, somatic, with precocious puberty, 176410
LHFPL5	160,3	100%	100%	Deafness, autosomal recessive 67, 610265
LHX3	46,1	98%	89%	Pituitary hormone deficiency, combined, 3, 221750
LHX4	83,9	100%	100%	Pituitary hormone deficiency, combined, 4, 262700

LIAS	104,3	100%	100%	Pyruvate dehydrogenase lipoic acid synthetase deficiency, 614462
LIFR	102,1	100%	97%	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LIG1	74,5	98%	91%	DNA ligase I deficiency
LIG4	177,1	100%	100%	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500 Severe combined immunodeficiency with sensitivity to ionizing radiation, 602450
LIM2	66,4	83%	81%	Cataract 19, 615277
LIPA	112,8	100%	100%	? Wolman disease, 278000 Cholesteryl ester storage disease, 278000 -3
LIPC	89	97%	94%	[High density lipoprotein cholesterol level QTL 12], 612797 {Diabetes mellitus, noninsulin-dependent}, 125853 Hepatic lipase deficiency, 614025
LIPH	118	100%	98%	Hypotrichosis, localized, autosomal recessive 2, 604379 Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379
LIPN	108,4	100%	99%	Ichthyosis, congenital, autosomal recessive 8, 613943
LITAF	71,8	100%	97%	Charcot-Marie-Tooth disease, type 1C, 601098
LMAN1	115,2	100%	100%	Combined factor V and VIII deficiency, 227300
LMBR1	93,1	100%	100%	Acheiropody, 200500 Polydactyly, preaxial type II, 174500 Triphalangeal thumb, type I, 174500 Triphalangeal thumb-polysyndactyly syndrome, 174500 Syndactyly, type IV, 186200
LMBRD1	111,9	100%	100%	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LMF1	90,3	100%	96%	Lipase deficiency, combined, 246650

LMNA	68,5	98%	88%	Emery-Dreifuss muscular dystrophy 2, AD, 181350 Cardiomyopathy, dilated, 1A, 115200 Lipodystrophy, familial partial, 2, 151660 Emery-Dreifuss muscular dystrophy 3, AR, 181350 Charcot-Marie-Tooth disease, type 2B1, 605588 Muscular dystrophy, congenital, 613205 Muscular dystrophy, limb-girdle, type 1B, 159001 Mandibuloacral dysplasia, 248370 Hutchinson-Gilford progeria, 176670 Restrictive dermopathy, lethal, 275210 Heart-hand syndrome, Slovenian type, 610140 Malouf syndrome, 212112
LMNB1	79,1	95%	89%	Leukodystrophy, adult-onset, autosomal dominant, 169500
LMX1B	82,4	98%	94%	Nail-patella syndrome, 161200
LOR	37,3	99%	85%	Vohwinkel syndrome with ichthyosis, 604117
LOXHD1	99,6	100%	99%	Deafness, autosomal recessive 77, 613079
LPAR6	114,3	100%	100%	Hypotrichosis 8, 278150 Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150
LPIN1	101	100%	97%	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	75,8	99%	95%	Majeed syndrome, 609628
LPL	110,7	100%	99%	Lipoprotein lipase deficiency, 238600 Combined hyperlipidemia, familial, 144250 [High density lipoprotein cholesterol level QTL 11]
LPP	130,1	100%	98%	Leukemia, acute myeloid, 601626
LRAT	187,7	100%	100%	Retinal dystrophy, early-onset severe, 613341 Leber congenital amaurosis 14, 613341 Retinitis pigmentosa, juvenile, 613341
LRBA	103,8	100%	97%	Immunodeficiency, common variable, 8, with autoimmunity, 614700

LRIG2	120,3	100%	97%	Urofacial syndrome 2, 615112
LRIT3	138,1	94%	93%	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRP2	104,6	100%	98%	Donnai-Barrow syndrome, 222448
LRP4	93,9	99%	96%	Cenani-Lenz syndactyly syndrome, 212780 Sclerosteosis 2, 614305
LRP5	92,3	98%	93%	Osteoporosis-pseudoglioma syndrome, 259770 [Bone mineral density variability 1], 601884 Hyperostosis, endosteal, 144750 van Buchem disease, type 2, 607636 Osteosclerosis, 144750 {Osteoporosis}, 166710 Exudative vitreoretinopathy 4, 601813 Osteopetrosis, autosomal dominant 1, 607634
LRPAP1	81,6	98%	92%	Myopia 23, autosomal recessive, 615431
LRPPRC	92,4	100%	97%	Leigh syndrome, French-Canadian type, 220111
LRRC6	113,8	100%	100%	Ciliary dyskinesia, primary, 19, 614935
LRRC8A	122,9	100%	100%	Agammaglobulinemia 5, 613506
LRRK2	111,5	99%	99%	Parkinson disease 8, 607060
LRSAM1	80	100%	97%	Charcot-Marie-Toothe disease, axonal, type 2P, 614436
LRTOMT	100	94%	92%	Deafness, autosomal recessive 63, 611451
LTBP2	72,4	99%	94%	Glaucoma 3, primary congenital, D, 613086 Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750 Weill-Marchesani syndrome 3, recessive, 614819
LTBP3	74,5	99%	95%	Tooth agenesis, selective, 6, 613097
LTBP4	83,8	97%	89%	Cutis laxa, autosomal recessive, type IC, 613177

LYZ	98,2	100%	100%	Amyloidosis, renal, 105200
LZTFL1	83,2	100%	99%	Bardet-Biedl syndrome 17, 615994
MAD1L1	72	97%	88%	Lymphoma, somatic Prostate cancer, somatic, 176807
MAF	76,1	77%	74%	Cataract, pulverulent or cerulean, with or without microcornea, 610202
MAFB	82,5	100%	100%	Multicentric carpotarsal osteolysis syndrome, 166300
MAGEL2	89	96%	89%	Prader-Willi-like syndrome, 615547
MAGT1	97,6	100%	100%	Mental retardation, X-linked 95, 300716 Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853
MAK	74,5	96%	94%	REtinitis pigmentosa 62, 614181
MAML2	105,9	100%	99%	Mucoepidermoid salivary gland carcinoma
MAMLD1	121,9	100%	100%	Hypospadias 2, X-linked, 300758
MAN1B1	96,6	100%	99%	Mental retardation, autosomal recessive 15, 614202
MAN2B1	81,3	99%	92%	Mannosidosis, alpha-, types I and II, 248500
MANBA	87,6	100%	99%	Mannosidosis, beta, 248510
MAOA	101,9	100%	100%	Brunner syndrome, 300615
MAP2K1	95,9	99%	84%	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	111,8	100%	98%	Cardiofaciocutaneous syndrome 4, 615280
MAP3K1	110,7	98%	96%	46XY sex reversal 6, 613762
MAP3K8	117,3	100%	99%	Lung cancer, somatic, 211980
MAPT	21,3	51%	36%	Dementia, frontotemporal, with or without parkinsonism, 600274 Pick disease, 172700 Supranuclear palsy, progressive, 601104

				Supranuclear palsy, progressive atypical, 260540 {Parkinson disease, susceptibility to}, 168600 Tauopathy and respiratory failure
MARS2	149,3	100%	100%	Spastic ataxia 3, autosomal recessive, 611390
MARVELD2	150	98%	96%	Deafness, autosomal recessive 49, 610153
MASP1	116,8	100%	98%	3MC syndrome 1, 257920
MASP2	114,4	99%	96%	MASP2 deficiency, 613791
MASTL	117,3	100%	99%	Thrombocytopenia-2, 188000
MAT1A	90	100%	96%	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MATN3	98,4	90%	84%	Epiphyseal dysplasia, multiple, 5, 607078 {Osteoarthritis susceptibility 2}, 140600 Spondyloepimetaphyseal dysplasia, 608728
MATR3	105	96%	93%	Myopathy, distal 2, 606070
MBD5	140	100%	99%	Mental retardation, autosomal dominant 1, 156200
MBTPS2	126,7	100%	100%	IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800
MC2R	126,8	100%	99%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MC4R	172,4	100%	100%	Obesity, autosomal dominant, 601665
MCC	80,2	99%	96%	Colorectal cancer
MCCC1	94,6	100%	98%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	105,2	95%	89%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	85,8	100%	100%	Methylmalonyl-CoA epimerase deficiency, 251120

MCFD2	64,1	100%	93%	Factor V and factor VIII, combined deficiency of, 613625
MCM4	97,8	100%	97%	Natural killer cell and glucocorticoid deficiency with DNA repair defect, 609981
MCM6	104,1	100%	99%	Lactase persistance/nonpersistance, 223100
MCOLN1	101,3	97%	93%	Mucolipidosis IV, 252650
MCPH1	118,2	100%	100%	Microcephaly 1, primary, autosomal recessive, 251200

MECP2	163,2	100%	98%	Rett syndrome, 312750 Mental retardation, X-linked, syndromic 13, 300055 Rett syndrome, preserved speech variant, 312750 Encephalopathy, neonatal severe, 300673 {Autism susceptibility, X-linked 3}, 300496 Angelman syndrome, 105830 Mental retardation, X-linked syndromic, Lubs type, 300260
MED12	122,4	97%	94%	Opitz-Kaveggia syndrome, 305450 Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895
MED13L	110,6	99%	98%	Transposition of the great arteries, dextro-looped 1, 608808
MED17	138,3	99%	97%	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	108,5	100%	99%	Mental retardation, autosomal recessive 18, 614249
MED25	92,7	97%	89%	Charcot-Marie-Tooth disease, type 2B2, 605589
MEF2C	106,5	100%	99%	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443 Chromosome 5q14.3 deletion syndrome, 613443 (4)
MEFV	117,3	100%	99%	Familial Mediterranean fever, AR, 249100 Familial Mediterranean fever, AD, 134610
MEGF10	103,3	100%	98%	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399
MEGF8	87,2	99%	93%	Carpenter syndrome 2, 614976

MEN1	110,2	100%	97%	Multiple endocrine neoplasia 1, 131100 Carcinoid tumor of lung Parathyroid adenoma, somatic Lipoma, somatic Angiofibroma, somatic Adrenal adenoma, somatic
MEOX1	75,6	100%	100%	Klippel-Feil syndrome 2, 214300
MERTK	112,8	100%	98%	Retinitis pigmentosa 38, 613862
MESP2	71,7	100%	99%	Spondylocostal dysostosis, autosomal recessive 2, 608681
MET	125,1	100%	99%	Renal cell carcinoma, papillary, familial and somatic, 605074 Hepatocellular carcinoma, childhood type, 114550 {Autism suseptibility 9}, 611015
MFN2	100,3	100%	99%	Charcot-Marie-Tooth disease, type 2A2, 609260 Hereditary motor and sensory neuropathy VI, 601152
MFRP	85,1	97%	94%	Microphthalmia, isolated 5, 611040 Nanophthalmos 2, 609549
MFSD8	105,4	100%	100%	Ceroid lipofuscinosis, neuronal, 7, 610951
MGAT2	194,6	100%	100%	Congenital disorder of glycosylation, type IIa, 212066
MGME1	138,2	100%	100%	Mitochondrial DNA depletion syndrome 11, 615084
MGP	66,8	100%	99%	Keutel syndrome, 245150 {Natural teeth remaining intact} (2)
MIB1	103,6	100%	100%	Left ventricular noncompaction 7, 615092
MID1	176,2	100%	99%	Opitz GBBB syndrome, type I, 300000
MINPP1	151,3	100%	100%	Thyroid carcinoma, follicular, 188470
MIP	74,5	100%	96%	Cataract 15, multiple types, 615274

MITF	145,9	100%	100%	Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 Tietz albinism-deafness syndrome, 103500 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456
MKKS	137,9	100%	100%	McKusick-Kaufman syndrome, 236700 Bardet-Biedl syndrome 6, 209900
MKL1	70,8	97%	91%	Megakaryoblastic leukemia, acute
MKRN3	104,4	100%	100%	Precocious puberty, central, 2, 615346
MKS1	104,1	98%	95%	Meckel syndrome 1, 249000 Bardet-Biedl syndrome 13, 209900
MLC1	96,4	100%	100%	Megalencephalic leukoencephalopathy with subcortical cysts, 604004 -3
MLH1	98,5	100%	100%	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MLH3	156,5	100%	98%	Colorectal cancer, somatic, 114500 Colorectal cancer, hereditary nonpolyposis, type 7, 614385 Endometrial cancer, 608089
MLLT11	120,7	100%	100%	Leukemia, acute myelomonocytic
MLPH	81,9	100%	94%	Griscelli syndrome, type 3, 609227
MLYCD	69,5	89%	84%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	160,5	100%	100%	Methylmalonic aciduria, vitamin B12-responsive, 251100
MMAB	75,3	99%	92%	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110
MMACHC	175,4	100%	100%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMP1	107,8	100%	100%	COPD, rate of decline of lung function in, 606963 {Epidermolysis bullosa dystrophica, autosomal recessive, modifier of}, 226600
MMP13	132,7	100%	100%	Spondyloepimetaphyseal dysplasia, Missouri type, 602111 Metaphyseal anadysplasia 1, 602111

MMP2	98,9	100%	99%	Torg-Winchester syndrome, 259600
MMP20	94	100%	98%	Amelogenesis imperfecta, type IIA2, 612529
MMP9	87,6	96%	91%	Metaphyseal anadysplasia 2, 613073
MN1	78,9	100%	100%	Meningioma, 607174
MNX1	42,1	70%	61%	Currarino syndrome, 176450
MOCS1	71,4	99%	93%	Molybdenum cofactor deficiency, type A, 252150
MOCS2	112,6	100%	100%	Molybdenum cofactor deficiency, type B, 252150
MOG	14,2	58%	15%	Narcolepsy 7, 614250
MPDU1	117,6	100%	100%	Congenital disorder of glycosylation, type If, 609180
MPDZ	99,9	98%	96%	Hydrocephalus, nonsyndromic, autosomal recessive 2, 615219
MPI	96,7	100%	95%	Congenital disorder of glycosylation, type Ib, 602579
MPL	113	100%	99%	Thrombocytopenia, congenital amegakaryocytic, 604498 Thrombocythemia 2, 601977 Myelofibrosis with myeloid metaplasia, somatic, 254450
MPLKIP	66	100%	100%	Trichothiodystrophy, nonphotosensitive 1, 234050
MPO	88,6	100%	98%	Myeloperoxidase deficiency, 254600 {Alzheimer disease, susceptibility to}, 104300 {Lung cancer, protection against, in smokers}
MPV17	105,5	100%	100%	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810 -3

MPZ	103,6	100%	99%	Charcot-Marie-Tooth disease, type 1B, 118200 Dejerine-Sottas disease, 145900 Neuropathy, congenital hypomyelinating, 605253 Charcot-Marie-Tooth disease, type 2J, 607736 Roussy-Levy syndrome, 180800 Charcot-Marie-Tooth disease, type 2I, 607677 Charcot-Marie-Tooth disease, dominant intermediate D, 607791
MRE11A	90,5	100%	100%	Ataxia-telangiectasia-like disorder, 604391
MRPL3	65,1	98%	87%	Combined oxidative phosphorylation deficiency 9, 614582
MRPS16	123,3	100%	100%	Combined oxidative phosphorylation deficiency 2, 610498
MRPS22	96,7	100%	100%	Combined oxidative phosphorylation deficiency 5, 611719
MS4A1	127,7	100%	100%	Immunodeficiency, common variable, 5, 613495
MSH2	99	98%	97%	Colorectal cancer, hereditary nonpolyposis, type 1, 120435 Muir-Torre syndrome, 158320 Mismatch repair cancer syndrome, 276300
MSH3	107,9	99%	98%	Endometrial carcinoma
MSH6	144,7	100%	100%	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 Endometrial cancer, familial, 608089 Mismatch repair cancer syndrome, 276300
MSR1	113,6	100%	98%	Prostate cancer, hereditary, 176807 Barrett esophagus/esophageal adenocarcinoma, 614266
MSRB3	117,1	100%	100%	Deafness, autosomal recessive 74, 613718
MSX1	55,6	100%	82%	Tooth agenesis, selective, 1, with or without orofacial cleft, 106600 Orofacial cleft 5, 608874 Ectodermal dysplasia 3, Witkop type, 189500
MSX2	41,1	82%	63%	Craniosynostosis, type 2, 604757 Parietal foramina 1, 168500 Parietal foramina with cleidocranial dysplasia, 168550

MTAP	88,6	100%	100%	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250
MTFMT	92,2	100%	100%	Combined oxidative phosphorylation deficiency 15, 614947
MTHFR	97,7	100%	97%	Homocystinuria due to MTHFR deficiency, 236250 {Schizophrenia, susceptibility to}, 181500 {Vascular disease, susceptibility to} {Neural tube defects, susceptibility to}, 601634 {Thromboembolism, susceptibility to}, 188050
MTM1	103,6	100%	100%	Myotubular myopathy, X-linked, 310400
MTMR2	96,5	100%	100%	Charcot-Marie-Tooth disease, type 4B1, 601382
MTO1	118,3	99%	95%	Combined oxidative phosphorylation deficiency 10, 614702
MTPAP	106,7	91%	91%	Ataxia, spastic, 4, 613672
MTR	100,3	99%	98%	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTRR	102,8	100%	98%	Homocystinuria-megaloblastic anemia, cbl E type, 236270 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MUC1	77,7	98%	88%	Medullary cystic kidney disease 1, 174000
MUSK	124,5	100%	98%	Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931
MUT	115,5	100%	100%	Methylmalonic aciduria, mut(0) type, 251000
MUTYH	110,4	100%	99%	Adenomas, multiple colorectal, 608456 Gastric cancer, somatic, 613659 Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600
MVK	86,1	100%	99%	Mevalonic aciduria, 610377 Hyper-IgD syndrome, 260920 Porokeratosis 3, disseminated superficial actinic, 175900
MXI1	105,8	100%	96%	Neurofibrosarcoma {Prostate cancer, susceptibility to}, 176807
MYBPC1	89,5	100%	98%	Arthrogryposis, distal, type 1B, 614335 Lethal congenital contracture syndrome 4, 614915

MYBPC3	91	97%	94%	Cardiomyopathy, familial hypertrophic, 4, 115197 Cardiomyopathy, dilated, 1MM, 615396 Left ventricular noncompaction 10, 615396
MYC	161,9	100%	100%	Burkitt lymphoma, 113970
MYCN	95	99%	97%	Feingold syndrome, 164280
MYD88	165,1	100%	99%	Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260 Macroglobulinemia, Waldenstrom, somatic, 153600
MYF6	140,5	100%	100%	Myopathy, centronuclear, 3, 614408
MYH11	117,7	99%	96%	Aortic aneurysm, familial thoracic 4, 132900
MYH14	61,5	92%	81%	Deafness, autosomal dominant 4A, 600652 Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369
MYH2	99,7	95%	92%	Inclusion body myopathy-3, 605637
MYH3	110,2	98%	93%	Arthrogryposis, distal, type 2A, 193700 Arthrogryposis, distal, type 2B, 601680
MYH6	95,5	95%	89%	Cardiomyopathy, familial hypertrophic, 14, 613251 Atrial septal defect 3, 614089 Cardiomyopathy, dilated, 1EE, 613252 {Sick sinus syndrome 3}, 614090
MYH7	90,3	94%	87%	Cardiomyopathy, familial hypertrophic, 1, 192600 Cardiomyopathy, dilated, 1S, 613426 Myopathy, myosin storage, 608358 Laing distal myopathy, 160500 Scapuloperoneal syndrome, myopathic type, 181430 Left ventricular noncompaction 5, 613426
MYH8	101,5	97%	88%	Carney complex variant, 608837 Trismus-pseudocamptodactyly syndrome, 158300

MYH9	93,7	99%	97%	May-Hegglin anomaly, 155100 Fechtner syndrome, 153640 Sebastian syndrome, 605249 Deafness, autosomal dominant 17, 603622 Epstein syndrome, 153650 Macrothrombocytopenia and progressive sensorineural deafness, 600208
MYL2	107,4	100%	100%	Cardiomyopathy, familial hypertrophic, 10, 608758
MYL3	83,7	100%	99%	Cardiomyopathy, familial hypertrophic, 8, 608751
MYLK	106,6	99%	94%	Aortic aneurysm, familial thoracic 7, 613780
MYLK2	84,4	99%	98%	Cardiomyopathy, hypertrophic, midventricular, digenic, 192600
MYO15A	89,4	97%	92%	Deafness, autosomal recessive 3, 600316
MYO1A	102,1	100%	99%	Deafness, autosomal dominant 48, 607841
MYO3A	104,4	99%	96%	Deafness, autosomal recessive 30, 607101
MYO5A	91,2	99%	97%	Griscelli syndrome, type 1, 214450
MYO5B	84,3	97%	91%	Microvillus inclusion disease, 251850
MYO6	99,5	99%	98%	Deafness, autosomal dominant 22, 606346 Deafness, autosomal recessive 37, 607821 Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346
MYO7A	76,4	97%	91%	Usher syndrome, type 1B, 276900 Deafness, autosomal recessive 2, 600060 Deafness, autosomal dominant 11, 601317
MYOC	184	100%	100%	Glaucoma 1A, primary open angle, 137750
MYOT	124,5	100%	99%	Muscular dystrophy, limb-girdle, type 1A, 159000 Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
MYOZ2	102,6	100%	100%	Cardiomyopathy, familial hypertrophic, 16, 613838

MYPN	110,7	100%	99%	Cardiomyopathy, dilated, 1KK, 615248 Cardiomypathy, familial hypertrophic, 22, 615248 Cardiomyopaty, familial restrictive 4, 615248
NAA10	92,7	94%	94%	N-terminal acetyltransferase deficiency, 300855
NAGA	82,1	100%	95%	Schindler disease, type I, 609241 Kanzaki disease, 609242 Schindler disease, type III, 609241
NAGLU	67	94%	84%	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NAGS	56,5	94%	76%	N-acetylglutamate synthase deficiency, 237310
NALCN	105,1	99%	96%	?Neuroaxonal neurodegeneration, infantile, with facial dysmophism, 615419
NANOS1	42	95%	85%	Spermatogenic failure 12, 615413
NBAS	97,1	100%	98%	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NBEAL2	109,8	99%	97%	Gray platelet syndrome, 139090
NCF1	0,5	0%	0%	Chronic granulomatous disease due to deficiency of NCF-1, 233700
NCF2	95,2	100%	98%	Chronic granulomatous disease due to deficiency of NCF-2, 233710
NCF4	90,1	98%	97%	Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III, 613960
NCOA4	33,9	72%	63%	Thyroid carcinoma, papillary, 188550
NCSTN	78,4	97%	92%	Acne inversa, familial, 1, 142690
NDE1	99	100%	97%	Lissencephaly 4 (with microcephaly), 614019
NDN	36,2	100%	99%	Prader-Willi syndrome, 176270
NDP	79,4	99%	94%	Norrie disease, 310600 Exudative vitreoretinopathy, X-linked, 305390
NDRG1	78,9	98%	92%	Charcot-Marie-Tooth disease, type 4D, 601455

NDUFA1	189,6	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFA10	89,6	99%	96%	previous assignment to chr. 12 Leigh syndrome, 256000
NDUFA11	97,4	98%	85%	Mitochondrial complex I deficiency, 252010
NDUFA12	96,6	100%	100%	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000
NDUFA2	151,5	100%	100%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFA9	95,8	99%	95%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 -3
NDUFAF1	100,4	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF2	53,6	100%	95%	Mitochondrial complex I deficiency, 252010 Leigh syndrome, 256000
NDUFAF3	127,9	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF4	66	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF5	128,7	100%	100%	Mitochondrial complex 1 deficiency, 252010
NDUFAF6	102,3	100%	98%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFB3	0,3	0%	0%	Mitochondrial complex I deficiency, 252010
NDUFS1	76,8	100%	97%	Mitochondrial complex I deficiency, 252010
NDUFS2	121,8	100%	96%	Mitochondrial complex I deficiency, 252010
NDUFS3	149,1	100%	100%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
NDUFS4	126,6	100%	100%	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFS6	108,8	95%	87%	Complex I, mitochondrial respiratory chain, deficiency of, 252010
NDUFS7	112,5	100%	100%	Leigh syndrome, 256000

NDUFS8	105,4	100%	95%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFV1	62,3	97%	91%	Mitochondrial complex I deficiency, 252010
NDUFV2	106,1	100%	100%	Mitochondrial complex I deficiency, 252010
NEB	85,6	82%	79%	Nemaline myopathy 2, autosomal recessive, 256030
NEFL	126,4	100%	100%	Charcot-Marie-Tooth disease, type 2E, 607684 Charcot-Marie-Tooth disease, type 1F, 607734
NEK1	115,8	100%	99%	Short rib-polydactyly syndrome, type IIA, 263520
NEU1	17,2	61%	35%	Sialidosis, type I, 256550 Sialidosis, type II, 256550
NEUROD1	131,4	100%	100%	{Diabetes mellitus, noninsulin-dependent}, 125853 Maturity-onset diabetes of the young 6, 606394
NEUROG3	118,2	100%	100%	Diarrhea 4, malabsorptive, congenital, 610370
NEXN	125,6	100%	100%	Cardiomyopathy, dilated, 1CC, 613122 Cardiomyopathy, familial hypertrophic, 20, 613876
NF1	79	82%	81%	Neurofibromatosis, type 1, 162200 Leukemia, juvenile myelomonocytic, 607785 Melanoma, desmoplastic neurotrophic (2) Neurofibromatosis, familial spinal, 162210 Neurofibromatosis-Noonan syndrome, 601321 Watson syndrome, 193520
NF2	81,6	100%	98%	loss of heterozygosity Neurofibromatosis, type 2, 101000 Meningioma, NF2-related, somatic, 607174 Schwannomatosis, 162091
NFIX	124,8	98%	98%	Sotos syndrome 2, 614753 Marshall-Smith syndrome, 602535
NFKB2	85,3	100%	97%	Immunodeficiency, common variable, 10, 615577
NFKBIA	95,9	100%	99%	Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency, 612132

NFU1	79,1	97%	91%	Multiple mitochondrial dysfunctions syndrome 1, 605711
NHEJ1	80,9	100%	94%	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
NHLRC1	107,4	100%	100%	Epilepsy, progressive myoclonic 2B (Lafora), 254780
NHS	131,1	96%	93%	Nance-Horan syndrome, 302350 Cataract 40, X-linked, 302200
NIN	129,3	99%	99%	Seckel syndrome 7, 614851
NIPA1	95,6	90%	82%	Spastic paraplegia 6, autosomal dominant, 600363
NIPAL4	125,7	100%	99%	Ichthyosis, congenital, autosomal recessive 6, 612281
NIPBL	113,4	99%	98%	Cornelia de Lange syndrome 1, 122470
NKX2-1	98,4	100%	98%	Goiter, familial, due to TTF-1 defect (1) Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978
NKX2-5	125,3	100%	99%	Atrial septal defect 7, with or without AV conduction defects, 108900 Tetrology of Fallot, 187500 Hypothyroidism, congenital nongoitrous, 5, 225250 Ventricular septal defect 3, 614432 Hypoplastic left heart syndrome 2, 614435 Conotruncal heart malformations, variable, 217095
NKX2-6	87,5	98%	95%	Persistent truncus arteriosus, 217095
NKX3-2	63,9	100%	100%	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330
NLRP12	100,4	100%	98%	Familial cold autoinflammatory syndrome 2, 611762
NLRP3	117,3	100%	99%	Cold-induced autoinflammatory syndrome, familial, 120100 Muckle-Wells syndrome, 191900 CINCA syndrome, 607115
NME1	138,7	100%	100%	Neuroblastoma, 256700

NME8	93,7	100%	99%	Ciliary dyskinesia, primary, 6, 610852
NMNAT1	93,1	100%	100%	Leber congenital amaurosis 9, 608553
NNT	95,1	100%	100%	Glucocorticoid deficiency 4, 614736
NOBOX	80,7	98%	91%	Premature ovarian failure 5, 611548
NOD2	91,7	100%	98%	{Inflammatory bowel disease 1}, 266600 Blau syndrome, 186580 {Psoriatic arthritis, susceptibility to}, 607507 Sarcoidosis, early-onset, 609464
NODAL	125,9	98%	81%	Heterotaxy, visceral, 5, 270100
NOG	108	100%	100%	Symphalangism, proximal, 185800 Multiple synostosis syndrome 1, 186500 Tarsal-carpal coalition syndrome, 186570 Stapes ankylosis with broad thumb and toes, 184460 Brachydactyly, type B2, 611377
NOP56	102,8	98%	96%	Spinocerebellar ataxia 36, 614153
NOTCH1	69,6	98%	87%	Aortic valve disease, 109730 Leukemia, T-cell acute lymphoblastic (2)
NOTCH2	90,7	91%	89%	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NOTCH3	65,6	91%	83%	Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy, 125310 ?Myofibromatosis, infantile 2, 615293
NPC1	88,3	100%	96%	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220
NPC2	68,6	100%	100%	Niemann-pick disease, type C2, 607625
NPHP1	109	100%	100%	Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900 Joubert syndrome 4, 609583

NPHP3	102,5	100%	100%	Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540 Meckel syndrome 7, 267010
NPHP4	91,8	99%	95%	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
NPHS1	82,3	99%	96%	Nephrotic syndrome, type 1, 256300
NPM1	39,2	85%	75%	Leukemia, acute promyelocytic, NPM/RARA type Leukemia, acute myeloid, 601626
NPPA	147,3	100%	99%	Atrial fibrillation, familial, 6, 612201
NPR2	145	100%	99%	Acromesomelic dysplasia, Maroteaux type, 602875
NR0B2	68,7	100%	98%	Obesity, mild, early-onset, 601665
NR2E3	79,1	99%	92%	Enhanced S-cone syndrome, 268100 Retinitis pigmentosa 37, 611131
NR2F1	157	100%	100%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NR3C1	95,4	98%	95%	Cortisol resistance
NR3C2	139,5	100%	98%	Pseudohypoaldosteronism type I, autosomal dominant, 177735 Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115
NRAS	135	100%	100%	Autoimmune lymphoproliferative syndrome type IV, 614470 Noonan syndrome 6, 613224 Epidermal nevus, somatic, 162900 Thyroid carcinoma, follicular, somatic, 188470 Colorectal cancer, somatic, 114500
NRL	45,1	100%	100%	Retinitis pigmentosa 27, 613750 Retinal degeneration, autosomal recessive, clumped pigment type
NRXN1	111,6	99%	97%	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332
NSD1	119,7	100%	99%	Sotos syndrome 1, 117550 Leukemia, acute myeloid, 601626 (1) Beckwith-Wiedemann syndrome, 130650

NSDHL	112	99%	97%	CHILD syndrome, 308050 CK syndrome, 300831
NSMF	84,3	94%	93%	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
NSUN2	118,1	100%	88%	Mental retardation, autosomal recessive 5, 611091
NT5C2	113,9	100%	100%	Spastic paraplegia 45, 613162
NT5E	107,3	100%	99%	Calcification of joints and arteries, 211800
NTF4	33,3	94%	84%	Glaucoma 1, open angle, 1O, 613100
NTRK1	64,5	98%	90%	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma, familial, 155240
NTRK2	98,7	97%	95%	Obesity, hyperphagia, and developmental delay, 613886
NUBPL	85,8	100%	100%	Mitochondrial complex I deficiency, 252010
NUMA1	89,6	98%	97%	Leukemia, acute promyelocytic, NUMA/RARA type
NUP214	122,5	99%	98%	Leukemia, acute myeloid, 601626 Leukemia, T-cell acute lymphoblastic
NUP62	87,1	100%	97%	Striatonigral degeneration, infantile, 271930
NYX	87	98%	95%	Night blindness, congenital stationary (complete), 1A, X-linked, 310500
OAT	42,2	82%	62%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OBSL1	75,6	98%	89%	3-M syndrome 2, 612921
OCA2	90,9	100%	98%	?hypopigmentation in PWS and AS Albinism, oculocutaneous, type II, 203200 Albinism, brown oculocutaneous, 203200 [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220
OCLN	87,3	74%	72%	Band-like calcification with simplified gyration and polymicrogyria, 251290
OCRL	112,6	98%	97%	Lowe syndrome, 309000 Dent disease 2, 300555

OFD1	67,8	93%	88%	Oral-facial-digital syndrome 1, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 Joubert syndrome 10, 300804
OGG1	92,9	99%	98%	Renal cell carcinoma, clear cell, somatic, 144700
OPA1	123,7	100%	99%	Optic atrophy 1, 165500 {Glaucoma, normal tension, susceptibility to}, 606657 Optic atrophy plus syndrome, 125250
OPA3	107,3	100%	100%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPHN1	98,8	99%	98%	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
OPLAH	97,5	99%	97%	5-oxoprolinase deficiency, 260005
OPN1LW	1,5	4%	2%	Colorblindness, protan, 303900 Blue cone monochromacy, 303700 -3
OPN1MW	1,3	0%	0%	Colorblindness, deutan, 303800 Blue cone monochromacy, 303700 -3
OPN1SW	93,3	99%	97%	Colorblindness, tritan, 190900
OPTN	95,2	100%	99%	Glaucoma 1, open angle, E, 137760 {Glaucoma, normal tension, susceptibility to}, 606657 Amyotrophic lateral sclerosis 12, 613435
ORAI1	77,2	92%	88%	Immunodeficiency 9, 612782
ORC1	103,5	100%	97%	Meier-Gorlin syndrome 1, 224690
ORC4	102,5	100%	100%	Meier-Gorlin syndrome 2, 613800
ORC6	88,2	100%	99%	Meier-Gorlin syndrome 3, 613803
OSMR	134,1	100%	100%	Amyloidosis, primary localized cutaneous, 1, 105250
OSTM1	111,2	100%	100%	Osteopetrosis, autosomal recessive 5, 259720
OTC	103,3	100%	99%	Ornithine transcarbamylase deficiency, 311250

OTOA	69,9	67%	66%	Deafness, autosomal recessive 22, 607039
OTOF	92	99%	97%	Deafness, autosomal recessive 9, 601071 Auditory neuropathy, autosomal recessive, 1, 601071
OTOG	88,5	97%	94%	Deafness, autosomal recessive 18B, 614945
OTOGL	111,3	100%	99%	Deafness, autosomal recessive 84B, 614944
OTX2	154,7	100%	100%	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, and pituitary dysfunction, 610125
OXCT1	89,9	100%	99%	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050
P2RX1	71,3	100%	90%	Bleeding disorder due to P2RX1 defect
P2RX2	104,1	100%	99%	Deafness, autosomal dominant 41, 608224
P2RY12	157,3	100%	100%	Bleeding disorder, platelet-type, 8, 609821
PABPN1	61,9	69%	59%	Oculopharyngeal muscular dystrophy, 164300
PACS1	99,9	97%	95%	Mental retardation, autosomal dominant 17, 615009
PAFAH1B1	74,6	89%	77%	Lissencephaly 1, 607432 Subcortical laminar heterotopia, 607432 -3
PAH	82,4	100%	95%	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600
PAK3	102,3	100%	100%	Mental retardation, X-linked 30/47, 300558
PALB2	122,1	99%	97%	Fanconi anemia, complementation group N, 610832 {Breast cancer, susceptibility to}, 114480 {Pancreatic cancer, susceptibility to, 3}, 613348
PANK2	113,2	99%	93%	Neurodegeneration with brain iron accumulation 1, 234200 HARP syndrome, 607236
PAPSS2	92,3	100%	100%	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847 -3

PAX2	92,1	92%	86%	Papillorenal syndrome, 120330 Renal hypoplasia, isolated, 191830 -3
PAX3	105,6	99%	97%	Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820 Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220
PAX4	75,2	100%	98%	Maturity-onset diabetes of the young, type IX, 612225 Diabetes mellitus, type 2, 125853 Diabetes mellitus, ketosis-prone, 612227
PAX6	91,2	100%	98%	Aniridia, 106210 Peters anomaly, 604229 Cataract with late-onset corneal dystrophy, 106210 Keratitis, 148190 Foveal hyperplasia, 136520 Morning glory disc anomaly, 120430 Optic nerve hypoplasia, 165550 Coloboma, ocular, 120200 Coloboma of optic nerve, 120430 Gillespie syndrome, 206700
PAX8	67,7	100%	90%	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700
PAX9	209,9	99%	99%	Tooth agenesis, selective, 3, 604625
PC	94	97%	91%	Pyruvate carboxylase deficiency, 266150
PCCA	95,3	98%	95%	Propionicacidemia, 606054
PCCB	106,8	100%	100%	Propionicacidemia, 606054
PCDH15	123,5	100%	100%	Usher syndrome, type 1F, 602083 Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1D/F digenic, 601067
PCDH19	148,5	99%	99%	Epileptic encephalopathy, early infantile, 9, 300088
PCM1	116,6	100%	99%	Thyroid carcinoma, papillary, 188550

PCNT	92	97%	90%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720 -3
PCSK1	103,5	100%	97%	Obesity with impaired prohormone processing, 600955 {Obesity, susceptibility to, BMIQ12}, 612362
PCSK9	68,3	98%	90%	Hypercholesterolemia, familial, 3, 603776 {Low density lipoprotein cholesterol level QTL 1}, 603776
PCYT1A	83,7	100%	98%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDCD10	68,1	83%	67%	Cerebral cavernous malformations 3, 603285
PDE11A	105,5	100%	96%	Pigmented nodular adrenocortical disease, primary, 2, 610475
PDE4D	102,1	96%	88%	{Stroke, susceptibility to, 1}, 606799 Acrodysostosis 2, with or without hormone resistance, 614613
PDE6A	90,5	98%	96%	Retinitis pigmentosa 43, 613810
PDE6B	99,5	100%	98%	Night blindness, congenital stationary, autosomal dominant 2, 163500 Retinitis pigmentosa-40, 613801
PDE6C	103,5	100%	100%	Cone dystrophy 4, 613093
PDE6G	89,3	100%	100%	Retinitis pigmentosa 57, 613582
PDE6H	32,3	94%	75%	Retinal cone dystrophy 3, 610024 Achromatopsia 6, 610024
PDE8B	95,4	100%	99%	Pigmented nodular adrenocortical disease, primary, 3, 614190 Striatal degeneration, autosomal dominant, 609161
PDGFB	66,9	100%	97%	Meningioma, SIS-related, 607174 Dermatofibrosarcoma protuberans, 607907
PDGFRA	123,1	99%	98%	Gastrointestinal stromal tumor, somatic, 606764 Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685
PDGFRB	82,4	100%	98%	Myeloproliferative disorder with eosinophilia, 131440 (4) Basal ganglia calcification, idiopathic, 4, 615007 Myofibromatosis, infantile, 1, 228550
PDGFRL	105,3	100%	96%	Hepatocellular cancer, somatic, 114550 Colorectal cancer, somatic, 114500

PDHA1	112,9	100%	99%	Pyruvate dehydrogenase E1-alpha deficiency, 312170 Leigh syndrome, X-linked, 308930
PDHB	98,2	100%	100%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDP1	157,7	100%	100%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	94,9	88%	87%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	92,3	100%	98%	Coenzyme Q10 deficiency, primary, 3, 614652
PDX1	39,6	100%	94%	Lacticacidemia due to PDX1 deficiency, 245349
PDYN	139,5	100%	99%	Spinocerebellar ataxia 23, 610245
PDZD7	75,3	95%	87%	{Retinal disease in Usher syndrome type IIA, modifier of}, 276901 Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472
PEPD	63,3	100%	89%	Prolidase deficiency, 170100
PER2	76,7	100%	97%	Advanced sleep phase syndrome, familial, 1, 604348
PET100	66,2	100%	99%	Mitochondrial complex IV deficiency, 220110
PEX1	118,6	100%	100%	Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	73,1	89%	85%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX11B	167,9	100%	100%	Peroxisome biogenesis disorder 14B, 614920
PEX12	114	100%	100%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	134,7	98%	96%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	90,9	100%	99%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	86	92%	83%	Peroxisome biogenesis disorder 8A, (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	107,5	100%	100%	Peroxisome biogenesis disorder 12A (Zellweger), 614886

PEX26	116,8	100%	100%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	130,7	100%	100%	Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	91,1	98%	96%	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370
PEX6	89,8	94%	85%	Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863
PEX7	98,2	99%	89%	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PFKM	98,7	100%	98%	Glycogen storage disease VII, 232800
PFN1	56,9	99%	84%	Amyotrophic lateral sclerosis 18, 614808
PGAM2	101,7	100%	100%	Glycogen storage disease X, 261670
PGAP2	119,4	100%	99%	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGK1	76,4	85%	77%	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	103,5	99%	96%	Glycogen storage disease XIV, 612934 Congenital disorder of glycosylation, type It, 614921
PHEX	121	100%	100%	Hypophosphatemic rickets, X-linked dominant, 307800
PHF6	147,5	100%	100%	Borjeson-Forssman-Lehmann syndrome, 301900
PHF8	100,9	100%	99%	Mental retardation syndrome, X-linked, Siderius type, 300263
PHGDH	85,1	100%	99%	Phosphoglycerate dehydrogenase deficiency, 601815
PHKA1	88,2	95%	94%	Muscle glycogenosis, 300559
PHKA2	97	100%	98%	Glycogen storage disease, type IXa1, 306000 Glycogen storage disease, type IXa2, 306000
PHKB	113,9	97%	97%	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750
PHKG2	146,8	100%	100%	Glycogen storage disease IXc, 613027 Cirrhosis due to liver phosphorylase kinase deficiency

PHOX2A	30	78%	63%	Fibrosis of extraocular muscles, congenital, 2, 602078
PHYH	85,5	100%	98%	Refsum disease, 266500
PICALM	102,8	95%	93%	Leukemia, acute myeloid, 601626 Leukemia, acute T-cell lymphoblastic
PIEZO1	85,9	98%	94%	Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380
PIEZO2	95,1	99%	98%	?Marden-Walker syndrome, 248700 Arthrogryposis, distal, type 3, 114300 Arthrogryposis, distal, type 5, 108145
PIGA	134,6	100%	99%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818 Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868
PIGL	92,4	100%	99%	CHIME syndrome, 280000
PIGM	108,4	100%	100%	Glycosylphosphatidylinositol deficiency, 610293
PIGN	102,9	100%	99%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	104,9	100%	99%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGV	165	100%	100%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIK3CA	112,1	93%	91%	Ovarian cancer, somatic, 167000 Breast cancer, somatic, 114480 Colorectal cancer, somatic, 114500 Gastric cancer, somatic, 613659 Hepatocellular carcinoma, somatic, 114550 Nonsmall cell lung cancer, somatic, 211980 Keratosis, seborrheic, somatic, 182000 Nevus, epidermal, somatic, 162900 CLOVE syndrome, somatic, 612918 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome, somatic, 603387 Cowden syndrome 5, 615108
PIK3CD	89	99%	93%	Immunodeficiency 14, 615513

PIK3R1	144,8	100%	100%	Agammaglobulinemia 7, autosomal recessive, 615214 SHORT syndrome, 269880
PIK3R2	76,5	90%	83%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome, 603387
PIK3R5	79,5	100%	100%	Ataxia-oculomotor apraxia 3, 615217
PIKFYVE	132,7	100%	99%	Corneal fleck dystrophy, 121850
PINK1	81,7	92%	88%	Parkinson disease 6, early onset, 605909
PIP5K1C	64	88%	84%	Lethal congenital contractural syndrome 3, 611369
PITPNM3	76,8	98%	93%	Cone-rod dystrophy 5, 600977
PITX1	67,5	100%	86%	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800 Liebenberg syndrome, 186550 (4)
PITX2	109,7	94%	86%	Axenfeld-Rieger syndrome, type 1, 180500 Iridogoniodysgenesis, type 2, 137600 Ring dermoid of cornea, 180550 Peters anomaly, 604229
PITX3	38,3	100%	93%	Anterior segment mesenchymal dysgenesis, 107250 Cataract 11, multiple types, 610623 Cataract 11, syndromic, 610623
PKD1	12,4	19%	17%	Polycystic kidney disease, adult type I, 173900
PKD2	92,4	97%	90%	Polycystic kidney disease 2, 613095
PKLR	121,9	100%	97%	Pyruvate kinase deficiency, 266200 Adenosine triphosphate, elevated, of erythrocytes, 102900
PKP1	86,7	97%	89%	Ectodermal dysplasia/skin fragility syndrome, 604536
PKP2	64,5	79%	74%	Arrhythmogenic right ventricular dysplasia 9, 609040
PLA2G4A	124,8	100%	100%	Phospholipase A2, group IV A, deficiency of
PLA2G5	102,8	100%	100%	Fleck retina, familial benign, 228980

PLA2G6	77,8	100%	93%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, 612953
PLA2G7	112,1	100%	100%	Platelet-activating factor acetylhydrolase deficiency, 614278 {Asthma, susceptibility to}, 600807 {Atopy, susceptibility to}, 147050
PLAG1	149,4	100%	98%	Adenomas, salivary gland pleomorphic, 181030
PLAU	95,8	100%	94%	{Alzheimer disease, late-onset, susceptibility to}, 104300 Quebec platelet disorder, 601709
PLCB1	107,8	99%	98%	Epileptic encephalopathy, early infantile, 12, 613722
PLCB4	89	100%	97%	Auriculocondylar syndrome 2, 614669
PLCD1	105,5	99%	95%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCE1	118	98%	96%	Nephrotic syndrome, type 3, 610725
PLCG2	105,4	100%	99%	Familial cold autoinflammatory syndrome 3, 614468 Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878
PLEKHG5	79,8	97%	94%	Spinal muscular atrophy, distal, autosomal recessive, 4, 611067 Charcot-Marie-Tooth disease, recessive intermediate C, 615376
PLEKHM1	6,1	28%	18%	Osteopetrosis, autosomal recessive 6, 611497
PLG	64,4	75%	68%	Plasminogen Tochigi disease Thrombophilia, dysplasminogenemic (1) Plasminogen deficiency, types I and II (1) Conjunctivitis, ligneous, 217090
PLIN1	44,2	92%	67%	Lipodystrophy, familial partial, type 4, 613877
PLN	155,2	100%	100%	Cardiomyopathy, dilated, 1P, 609909 Cardiomyopathy, familial hypertrophic, 18, 613874
PLOD1	80,5	100%	97%	Ehlers-Danlos syndrome, type VI, 225400
PLOD2	108,2	100%	100%	Bruck syndrome 2, 609220

PLOD3	82,8	99%	88%	Lysyl hydroxylase 3 deficiency, 612394
PLP1	86,2	100%	95%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PLS3	124,4	100%	100%	Bone mineral density QTL18, osteoporosis, 300910
PML	107	99%	98%	Leukemia, acute promyelocytic, PML/RARA type
PMM2	85,6	100%	100%	Congenital disorder of glycosylation, type Ia, 212065
PMP22	91,7	100%	98%	Charcot-Marie-Tooth disease, type 1A, 118220 Dejerine-Sottas disease, 145900 Neuropathy, recurrent, with pressure palsies, 162500 Charcot-Marie-Tooth disease, type 1E, 118300 Roussy-Levy syndrome, 180800 Neuropathy, inflammatory demyelinating, 139393
PMS2	68,3	56%	55%	Mismatch repair cancer syndrome, 276300 Colorectal cancer, hereditary nonpolyposis, type 4, 614337
PNKP	67,7	99%	95%	Epileptic encephalopathy, early infantile, 10, 613402
PNP	117,2	100%	100%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA1	121,9	100%	100%	Ichthyosis, congenital, autosomal recessive 10, 615024
PNPLA2	76,4	97%	93%	Neutral lipid storage disease with myopathy, 610717
PNPLA6	82,6	99%	96%	Spastic paraplegia 39, autosomal recessive, 612020
PNPO	69,7	100%	90%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
PNPT1	99,1	100%	99%	Combined oxidative phosphorylation deficiency 13, 614932 Deafness, autosomal recessive 70, 614934
POC1A	100,2	97%	92%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POFUT1	110	100%	98%	Dowling-Degos disease 2, 615327

POLD1	78,7	94%	91%	{Colorectal cancer, susceptibility to, 10}, 612591 Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381
POLG	86,5	99%	91%	Progressive external ophthalmoplegia, autosomal recessive, 258450 Progressive external ophthalmoplegia, autosomal dominant, 157640 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459
POLG2	113,8	100%	99%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131
POLH	141	98%	96%	Xeroderma pigmentosum, variant type, 278750
POLR1C	116,4	99%	95%	Treacher Collins syndrome 3, 248390
POLR1D	167,4	100%	100%	Treacher Collins syndrome 2, 613717
POLR3A	85,6	99%	95%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	100,3	99%	98%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMC	43	79%	66%	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734 {Obesity, early-onset, susceptibility to}, 601665
POMGNT1	97,4	100%	98%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157
POMP	153,1	100%	100%	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952
POMT1	94,7	100%	97%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308
POMT2	69,5	98%	92%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158
POR	101,6	100%	100%	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571
PORCN	105,8	86%	83%	Focal dermal hypoplasia, 305600

POU1F1	104,3	100%	100%	Pituitary hormone deficiency, combined, 1, 613038
POU3F4	137,5	100%	100%	Deafness, X-linked 2, 304400
POU4F3	160,4	100%	100%	Deafness, autosomal dominant 15, 602459

PPARG	104,9	98%	93%	Obesity, severe, 601665 [Obesity, resistance to] Insulin resistance, severe, digenic, 604367 Lipodystrophy, familial partial, type 3, 604367 Carotid intimal medial thickness 1, 609338 {Diabetes, type 2}, 125853
PPIB	86,3	100%	100%	Osteogenesis imperfecta, type IX, 259440
PPM1D	138,6	100%	99%	Breast cancer, 114480
PPM1K	109,9	100%	92%	Maple syrup urine disease, mild variant, 615135
PPOX	92,4	100%	97%	Porphyria variegata, 176200
PPP1R3A	190,1	100%	100%	Insulin resistance, severe, digenic, 604367
PPP2R1B	105,5	100%	100%	Lung cancer, 211980
PPP2R2B	95,7	95%	88%	Spinocerebellar ataxia 12, 604326
PPT1	63	100%	90%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	125,1	100%	100%	Renpenning syndrome, 309500
PRCC	94,7	100%	99%	Renal cell carcinoma, papillary, 605074
PRCD	80,9	100%	100%	Retinitis pigmentosa 36, 610599
PRDM16	110,3	97%	95%	Left ventricular noncompaction 8, 615373 Cardiomyopathy, dilated, 1LL, 615373
PRDM5	108,8	100%	100%	Brittle cornea syndrome 2, 614170

PRF1	84,5	100%	97%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027
PRG4	106,8	95%	80%	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250
PRICKLE1	105,4	100%	98%	Epilepsy, progressive myoclonic 1B, 612437
PRICKLE2	112,4	100%	100%	Epilepsy, progressive myoclonic 5, 613832
PRKAG2	87,4	100%	100%	Wolff-Parkinson-White syndrome, 194200 Cardiomyopathy, familial hypertrophic 6, 600858 Glycogen storage disease of heart, lethal congenital, 261740
PRKAR1A	94,7	91%	83%	Carney complex, type 1, 160980 Myxoma, intracardiac, 255960 Thyroid carcinoma, papillary, somatic, 188550 Pigmented nodular adrenocortical disease, primary, 1, 610489 Adrenocortical tumor, somatic, Acrodysostosis 1, with or without hormone resistance, 101800
PRKCA	108	100%	98%	Pituitary tumor, invasive
PRKCG	98	98%	94%	Spinocerebellar ataxia 14, 605361
PRKCSH	86	100%	93%	Polycystic liver disease, 174050
PRKG1	89,9	99%	94%	Aortic aneurysm, familial thoracic 8, 615436
PRLR	91,7	100%	100%	?Hyperprolactinemia, 615555 Multiple fibroadenomas of the breast, 615554
PRNP	91,4	100%	100%	Creutzfeldt-Jakob disease, 123400 Gerstmann-Straussler disease, 137440 Insomnia, fatal familial, 600072 Prion disease with protracted course, 606688 Huntington disease-like 1, 603218 {Kuru, susceptibility to}, 245300
PROC	86,8	100%	96%	Thrombophilia due to protein C deficiency, autosomal dominant, 176860 Thrombophilia due to protein C deficiency, autosomal recessive, 612304
PRODH	52	78%	63%	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850

PROK2	81,2	100%	86%	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	174,5	100%	100%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200

PROM1	88,1	98%	94%	Retinitis pigmentosa 41, 612095 Cone-rod dystrophy 12, 612657 Stargardt disease 4, 603786 Macular dystrophy, retinal, 2, 608051
PROP1	75,7	100%	85%	Pituitary hormone deficiency, combined, 2, 262600
PROS1	52,1	80%	67%	Thrombophilia due to protein S deficiency, autosomal dominant, 612336 Thrombophilia due to protein S deficiency, autosomal recessive, 614514
PRPF31	91	92%	84%	Retinitis pigmentosa 11, 600138
PRPF6	80,9	100%	99%	Retinitis pigmentosa 60, 613983
PRPF8	116,7	99%	98%	Retinitis pigmentosa 13, 600059
PRPH2	157,3	100%	99%	Retinitis pigmentosa 7, 608133 Retinitis punctata albescens, 136880 Macular dystrophy, patterned, 169150 Macular dystrophy, vitelliform, 608161 Foveomacular dystrophy, adult-onset, with choroidal neovascularization, 608161 Macular dystrophy Retinitis pigmentosa, digenic, 608133 Choriodal dystrophy, central areolar 2, 613105
PRPS1	131,2	100%	100%	Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Arts syndrome, 301835 Deafness, X-linked 1, 304500
PRRT2	80,5	100%	100%	Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751 Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066
PRRX1	81,7	99%	95%	Agnathia-otocephaly complex, 202650

PRSS1	113,8	78%	76%	Preeclampsia/eclampsia 5, 614595
PRSS1	113,8	78%	76%	Pancreatitis, hereditary, 167800 Trypsinogen deficiency, 614044 (1)
PRSS12	97,6	100%	95%	Mental retardation, autosomal recessive 1, 249500
PRSS56	66,5	96%	86%	Microphthalmia, isolated 6, 613517
PRX	110,1	99%	99%	Dejerine-Sottas disease, autosomal recessive, 145900 Charcot-Marie-Tooth disease, type 4F, 614895
PSAP	82,2	100%	99%	Metachromatic leukodystrophy due to SAP-b deficiency, 249900 Gaucher disease, atypical, 610539 Combined SAP deficiency, 611721 Krabbe disease, atypical, 611722
PSAT1	38,9	69%	49%	Phosphoserine aminotransferase deficiency, 610992
PSEN1	92,2	100%	95%	Alzheimer disease, type 3, 607822 Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 Dementia, frontotemporal, 600274 Pick disease, 172700 Cardiomyopathy, dilated, 1U, 613694 Acne inversa, familial, 3, 613737
PSEN2	86,4	100%	98%	Alzheimer disease-4, 606889 Cardiomyopathy, dilated, 1V, 613697 -3
PSENEN	119,6	100%	100%	Acne inversa, familial, 2, 613736
PSMB8	9,4	40%	3%	Autoinflammation, lipodystrophy, and dermatosis syndrome, 256040
PSMC3IP	140,5	96%	94%	Ovarian dysgenesis 3, 614324
PSTPIP1	58	96%	89%	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416 -3
PTCH1	86,9	99%	95%	Basal cell nevus syndrome, 109400 Basal cell carcinoma, somatic, 605462 Holoprosencephaly-7, 610828

PTCH2	80,5	99%	96%	Medulloblastoma, 155255 Basal cell carcinoma, somatic, 605462 -3
PTDSS1	117,2	100%	100%	Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	122,4	99%	94%	Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Bannayan-Riley-Ruvalcaba syndrome, 153480 {Meningioma}, 607174 {Glioma susceptibility 2}, 613028 Macrocephaly/autism syndrome, 605309 PTEN hamartoma tumor syndrome VATER association with macrocephaly and ventriculomegaly, 276950 {Prostate cancer, somatic}, 176807 Thyroid carcinoma, follicular, somatic, 188470 Malignant melanoma, somatic, 155600 Endometrial carcinoma, somatic, 608089 Squamous cell carcinoma, head and neck, somatic, 275355
PTF1A	29,7	86%	57%	Diabetes mellitus, permanent neonatal, with cerebellar agenesis, 609069
PTGIS	57	98%	89%	Hypertension, essential, 145500
PTH	169,3	100%	100%	Hypoparathyroidism, autosomal dominant, 146200 Hypoparathyroidism, autosomal recessive, 146200
PTHLH	144,6	100%	100%	Humoral hypercalcemia of malignancy (1) Brachydactyly, type E2, 613382
PTPN11	41,7	83%	68%	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, 607785 Metachondromatosis, 156250
PTPN12	113,3	100%	99%	Colon cancer
PTPN14	118,4	100%	99%	Choanal atresia and lymphedema, 613611
PTPRC	102,3	97%	94%	{Hepatitic C virus, susceptibility to}, 609532 Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971

PTPRJ	105,8	97%	97%	Colon cancer, somatic, 114500
PTPRO	96,6	98%	97%	Nephrotic syndrome, type 6, 614196
PTPRQ	110,9	99%	98%	Deafness, autosomal recessive 84A, 613391
PTRF	146,8	100%	100%	Lipodystrophy, congenital generalized, type 4, 613327
PTS	103	100%	100%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUS1	65,6	99%	96%	Mitochondrial myopathy and sideroblastic anemia 1, 600462
PYCR1	81,9	100%	98%	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
PYGL	96,3	99%	98%	Glycogen storage disease VI, 232700
PYGM	95,8	100%	99%	McArdle disease, 232600
QARS	117	99%	99%	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	75,9	100%	96%	Hyperphenylalaninemia, BH4-deficient, C, 261630
RAB18	101,1	100%	100%	Warburg micro syndrome 3, 614222
RAB23	127,8	100%	100%	Carpenter syndrome, 201000
RAB27A	114,6	100%	100%	Griscelli syndrome, type 2, 607624
RAB28	71,1	99%	94%	Cone-rod dystrophy 18, 615374
RAB39B	166,5	100%	100%	Mental retardation, X-linked 72, 300271
RAB3GAP1	123,8	100%	99%	Warburg micro syndrome 1, 600118
RAB3GAP2	106,7	100%	98%	Martsolf syndrome, 212720 Warburg micro syndrome 2, 614225
RAB40AL	29,8	99%	82%	Mental retardation, X-linked, syndromic, Martin-Probst type, 300519 -3

RAC2	59,1	99%	95%	Neutrophil immunodeficiency syndrome, 608203
RAD21	86,4	98%	90%	Cornelia de Lange syndrome 4, 614701
RAD50	113,2	100%	99%	Nijmegen breakage syndrome-like disorder, 613078
RAD51C	93,2	100%	100%	Fanconi anemia, complementation group O, 613390 {Breast-ovarian cancer, familial, susceptibility to, 3}, 613399
RAD54B	120,2	100%	99%	Lymphoma, non-Hodgkin Colon adenocarcinoma
RAD54L	88,6	100%	96%	{Breast cancer, invasive ductal}, 114480 Lymphoma, non-Hodgkin,somatic, 605027 Adenocarcinoma, colonic, somatic
RAF1	89,7	100%	98%	Noonan syndrome 5, 611553 LEOPARD syndrome 2, 611554
RAG1	132,2	100%	100%	Severe combined immunodeficiency, B cell-negative, 601457 Omenn syndrome, 603554 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	187,2	100%	100%	Severe combined immunodeficiency, B cell-negative, 601457 Omenn syndrome, 603554 Combined cellular and humoral immune defects with granulomas, 233650
RAI1	133,7	99%	98%	Smith-Magenis syndrome, 182290
RAP1GDS1	80,2	99%	96%	Lymphocytic leukemia, acute T-cell
RAPSN	95,3	96%	86%	Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931 Myasthenic syndrome, congenital, associated with facial dysmorphism and acetylcholine receptor deficiency, 608931 Fetal akinesia deformation sequence, 208150
RARB	142,9	98%	98%	Microphthalmia, syndromic 12, 615524
RARS2	82,3	100%	98%	Pontocerebellar hypoplasia, type 6, 611523

RASA1	90,5	100%	99%	Parkes Weber syndrome, 608355 Capillary malformation-arteriovenous malformation, 608354 Basal cell carcinoma, somatic, 605462
RAX	88,4	83%	71%	Mental retardation, X-linked, FRAXE type, 309548 Microphthalmia, isolated 3, 611038
RB1	103,6	98%	98%	Retinoblastoma, 180200 Osteosarcoma, somatic, 259500 Bladder cancer, somatic, 109800 Small cell cancer of the lung, somatic, 182280 Retinoblastoma, trilateral, 180200
RB1CC1	120	100%	100%	Breast cancer, somatic, 114480
RBBP8	111,4	100%	100%	Pancreatic carcinoma, somatic Seckel syndrome 2, 606744 Jawad syndrome, 251255
RBM10	98,2	99%	97%	TARP syndrome, 311900
RBM20	108,9	100%	97%	Cardiomyopathy, dilated, 1DD, 613172
RBM28	101,2	100%	97%	Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RBM8A	75,8	100%	95%	Thrombocytopenia-absent radius syndrome, 274000
RBP4	71,1	91%	85%	Retinol dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RBPJ	68,5	99%	95%	Adams-Oliver syndrome 3, 614814
RD3	59,4	100%	100%	Leber congenital amaurosis 12, 610612
RDH12	66,3	93%	88%	Leber congenital amaurosis 13, 612712
RDH5	99	100%	96%	Fundus albipunctatus, 136880
RDX	48,9	84%	72%	Deafness, autosomal recessive 24, 611022

RECQL4	91,5	98%	96%	Rothmund-Thomson syndrome, 268400 RAPADILINO syndrome, 266280 Baller-Gerold syndrome, 218600
REEP1	89,1	100%	95%	Spastic paraplegia 31, autosomal dominant, 610250 Neuronopathy, distal hereditary motor, type VB, 614751
RELN	102	99%	97%	Lissencephaly 2 (Norman-Roberts type), 257320
REN	98,9	100%	100%	[Hyperproreninemia] Renal tubular dysgenesis, 267430 Hyperuricemic nephropathy, familial juvenile 2, 613092
RET	88,5	97%	94%	Multiple endocrine neoplasia IIA, 171400 Medullary thyroid carcinoma, 155240 Multiple endocrine neoplasia IIB, 162300 Central hypoventilation syndrome, congenital, 209880 Pheochromocytoma, 171300 Renal agenesis, 191830 {Hirschsprung disease, susceptibility to, 1}, 142623
RFT1	73,9	100%	96%	Congenital disorder of glycosylation, type In, 612015
RFX5	106,5	99%	99%	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFX6	128,3	100%	100%	Martinez-Frias syndrome, 601346
RFXANK	89,6	99%	96%	MHC class II deficiency, complementation group B, 209920
RFXAP	87,4	92%	86%	Bare lymphocyte syndrome, type II, complementation group D, 209920
RGR	76,2	96%	78%	Retinitis pigmentosa 44, 613769
RGS9	106	97%	93%	Bradyopsia, 608415
RGS9BP	46,1	100%	99%	Bradyopsia, 608415
RHAG	84,7	99%	97%	Anemia, hemolytic, Rh-null, regulator type, 268150 Rh-mod syndrome
RHBDF2	60,1	95%	87%	Tylosis with esophageal cancer, 148500

RHCE	112,6	73%	71%	[Blood group, Rhesus], 111690 Rh-null disease, amorph type -3
RHO	105,7	100%	98%	Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis punctata albescens, 136880
RIMS1	99,2	100%	99%	Cone-rod dystrophy 7, 603649
RIN2	109,5	99%	97%	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075
RIPK4	98,7	100%	96%	Popliteal pterygium syndrome 2, lethal type, 263650
RIT1	130,2	100%	100%	Noonan syndrome 8, 615355
RLBP1	94,9	100%	99%	Fundus albipunctatus, 136880 Retinitis punctata albescens, 136880 Newfoundland rod-cone dystrophy, 607476 Bothnia retinal dystrophy, 607475
RMND1	69,7	91%	89%	Combined oxidative phosphorylation deficiency 11, 614922
RNASEH2A	96,1	99%	94%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	102,3	99%	97%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	133	100%	100%	Aicardi-Goutieres syndrome 3, 610329
RNASEL	146,3	100%	97%	Prostate cancer 1, 601518
RNASET2	85	100%	99%	Leukoencephalopathy, cystic, without megalencephaly, 612951
RNF135	87,1	96%	84%	Macrocephaly, macrosomia, facial dysmorphism syndrome, 614192
RNF139	150,6	100%	100%	Renal cell carcinoma, 144700
RNF168	193,1	100%	100%	RIDDLE syndrome, 611943
RNF170	110,4	100%	100%	Ataxia, sensory, 1, autosomal dominant, 608984

RNF212	91,6	100%	98%	Recombination rate QTL 1, 612042
RNF216	89,9	96%	95%	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840
RNF6	146,7	100%	99%	Esophageal carcinoma, somatic, 133239
ROBO2	110,4	100%	99%	Vesicoureteral reflux 2, 610878
ROBO3	79,9	94%	85%	Gaze palsy, horizontal, with progressive scoliosis, 607313
ROGDI	94,2	97%	95%	Kohlschutter-Tonz syndrome, 226750
ROM1	98,3	100%	100%	Retinitis pigmentosa 7, digenic, 608133
ROR2	110,5	100%	97%	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RP1	169,8	100%	100%	Retinitis pigmentosa 1, 180100 {Hypertriglyceridemia, susceptibility to}, 145750
RP1L1	120,6	100%	100%	Occult macular dystrophy, 613587
RP2	105,6	100%	100%	Retinitis pigmentosa 2, 312600
RPE65	113,6	99%	97%	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794
RPGR	96,3	86%	83%	Retinitis pigmentosa 3, 300029 Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 Macular degeneration, X-linked atrophic, 300834 Cone-rod dystrophy, X-linked, 1, 304020
RPGRIP1	116,1	100%	99%	Leber congenital amaurosis 6, 613826 Cone-rod dystrophy 13, 608194
RPGRIP1L	94,3	98%	96%	Joubert syndrome 7, 611560 Meckel syndrome 5, 611561 COACH syndrome, 216360
RPIA	66,9	100%	99%	Ribose 5-phosphate isomerase deficiency, 608611
RPL11	69,4	94%	87%	Diamond-Blackfan anemia 7, 612562

RPL35A	26,5	75%	50%	Diamond-Blackfan anemia 5, 612528
RPL5	30,4	79%	60%	Diamond-Blackfan anemia 6, 612561
RPS10	29	89%	71%	Diamond-Blackfan anemia 9, 613308
RPS14	36,3	77%	61%	Macrocytic anemia, refractory, due to 5q deletion, somatic, 153550 -3
RPS17	0	0%	0%	Diamond-Blackfan anemia 4, 612527
RPS19	39,8	65%	45%	Diamond-Blackfan anemia 1, 105650
RPS24	89,6	96%	94%	Diamond-blackfan anemia 3, 610629
RPS26	34,5	64%	62%	Diamond-Blackfan anemia 10, 613309
RPS6KA3	97,9	100%	99%	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844
RPS7	18,4	79%	41%	Diamond-Blackfan anemia 8, 612563
RPSA	21,2	72%	45%	Asplenia, isolated congenital
RRAS2	93,4	100%	90%	Ovarian carcinoma
RRM2B	109,6	100%	100%	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 5, 613077 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075
RS1	75,4	99%	95%	25cM from XG Retinoschisis, 312700
RSPH1	105	100%	100%	Ciliary dyskinesia, primary, 24, 615481
RSPH4A	136,8	100%	99%	Ciliary dyskinesia, primary, 11, 612649
RSPH9	80,2	100%	97%	Ciliary dyskinesia, primary, 12, 612650
RSPO1	45,8	88%	81%	Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644 Palmoplantar hyperkeratosis and true hermaphroditism, 610644

RSPO4	89,7	100%	100%	Anonychia congenita, 206800
RTEL1	82,1	99%	95%	Dyskeratosis congenita, autosomal recessive 5, 615190 Dyskeratosis congenita, autosomal dominant 4, 615190
RTN2	67,3	97%	92%	Spastic paraplegia 12, autosomal dominant, 604805
RTTN	89,3	99%	98%	Polymicrogyria with seizures, 614833
RUNX1	65,3	97%	89%	Leukemia, acute myeloid, 601626 Platelet disorder, familial, with associated myeloid malignancy, 601399
RUNX2	132,1	100%	100%	Cleidocranial dysplasia, 119600 Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600 Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510
RYR1	77,5	97%	91%	{Malignant hyperthermia susceptibility 1}, 145600 Central core disease, 117000 Minicore myopathy with external ophthalmoplegia, 255320 Neuromuscular disease, congenital, with uniform type 1 fiber, 117000 King-Denborough syndrome, 145600
RYR2	108,1	100%	99%	Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772 Arrhythmogenic right ventricular dysplasia 2, 600996
SACS	150,7	100%	100%	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAG	108,6	100%	100%	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758
SALL1	137,1	100%	99%	Townes-Brocks syndrome, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480
SALL4	103,7	97%	96%	Duane-radial ray syndrome, 607323 IVIC syndrome, 147750
SAMD9	178,1	100%	100%	Tumoral calcinosis, familial, normophosphatemic, 610455
SAMHD1	112,7	100%	98%	Aicardi-Goutieres syndrome 5, 612952 Chilblain lupus 2, 614415 -3
SAR1B	93,9	100%	100%	Chylomicron retention disease, 246700

SARS2	71,2	98%	91%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SART3	85,8	100%	96%	Porokeratosis, disseminated superficial actinic, 1, 175900
SAT1	127,1	100%	100%	Keratosis follicularis spinulosa decalvans, 308800
SATB2	95,9	100%	95%	Cleft palate and mental retardation, 119540
SBF2	101	99%	98%	Charcot-Marie-Tooth disease, type 4B2, 604563
SCARB2	96,9	100%	98%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCARF2	51,2	93%	83%	Van den Ende-Gupta syndrome, 600920
SCN10A	117,2	99%	98%	Episodic pain syndrome, familial, 2, 615551
SCN11A	122,4	99%	99%	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN1A	115,6	100%	99%	Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Dravet syndrome, 607208 Migraine, familial hemiplegic, 3, 609634 Febrile seizures, familial, 3A, 604403
SCN1B	109,1	99%	96%	Epilepsy, generalized, with febrile seizures plus, type 1, 604233 Brugada syndrome 5, 612838 Cardiac conduction defect, nonspecific, 612838 Atrial fibrillation, familial, 13, 615377
SCN2A	118,6	99%	99%	Seizures, benign familial infantile, 3, 607745 Epileptic encephalopathy, early infantile, 11, 613721
SCN2B	98,5	100%	95%	Atrial fibrillation, familial, 14, 615378
SCN3B	93,7	100%	100%	Brugada syndrome 7, 613120
SCN4A	123,9	100%	98%	Hyperkalemic periodic paralysis, type 2, 170500 Paramyotonia congenita, 168300 Myotonia congenita, atypical, acetazolamide-responsive, 608390 Myasthenic syndrome, acetazolamide-responsive, 614198 Hypokalemic periodic paralysis, type 2, 613345

SCN4B	89	100%	99%	Long QT syndrome-10, 611819

SCN5A	109,9	100%	99%	Long QT syndrome-3, 603830 Brugada syndrome 1, 601144 Heart block, progressive, type IA, 113900 Heart block, nonprogressive, 113900 Ventricular fibrillation, familial, 1, 603829 Sick sinus syndrome 1, 608567 Cardiomyopathy, dilated, 1E, 601154 {Sudden infant death syndrome, susceptibility to}, 272120 Atrial fibrillation, familial, 10, 614022
SCN8A	141,3	100%	99%	Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy, early infantile, 13, 614558
SCN9A	111,4	100%	99%	Erythermalgia, primary, 133020 Insensitivity to pain, channelopathy-associated, 243000 Paroxysmal extreme pain disorder, 167400 Febrile seizures, familial, 3B, 613863 Epilepsy, generalized, with febrile seizures plus, type 7, 613863 Small fiber neuropathy, 133020 {Dravet syndrome, modifier of}, 607208
SCNN1A	91	99%	93%	Pseudohypoaldosteronism, type I, 264350 Bronchiectasis with or without elevated sweat chloride 2, 613021
SCNN1B	90,6	100%	96%	Liddle syndrome, 177200 Pseudohypoaldosteronism, type I, 264350 Bronchiectasis with or without elevated sweat chloride 1, 211400
SCNN1G	127,3	100%	100%	Liddle syndrome, 177200 Pseudohypoaldosteronism, type I, 264350 Bronchiectasis with or without elevated sweat chloride 3, 613071
SCO2	82,5	100%	100%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908
SCP2	91,9	100%	97%	Leukoencephalopathy with dystonia and motor neuropathy, 613724
SDCCAG8	92,6	100%	98%	Senior-Loken syndrome 7, 613615

SDHA	8,6	34%	12%	Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Cardiomyopathy, dilated, 1GG, 613642 Paragangliomas 5, 614165
SDHAF1	50,7	100%	91%	Mitochondrial complex II deficiency, 252011
SDHAF2	106	100%	100%	Paragangliomas 2, 601650
SDHB	94,2	100%	100%	Pheochromocytoma, 171300 Paraganglioma and gastric stromal sarcoma, 606864 Cowden syndrome 2, 612359 Gastrointestinal stromal tumor, 606764
SDHC	23,8	41%	36%	Paragangliomas 3, 605373 Paraganglioma and gastric stromal sarcoma, 606864 Gastrointestinal stromal tumor, 606764
SDHD	8,9	24%	17%	Paragangliomas 1, with or without deafness, 168000 Pheochromocytoma, 171300 Carcinoid tumors, intestinal, 114900 Merkel cell carcinoma, somatic Paraganglioma and gastric stromal sarcoma, 606864 Cowden syndrome 3, 615106
SEC23A	113	100%	98%	Craniolenticulosutural dysplasia, 607812
SEC23B	109	100%	100%	Anemia, dyserythropoietic congenital, type II, 224100
SEC63	81	93%	92%	Polycystic liver disease, 174050
SECISBP2	95,1	100%	98%	Thyroid hormone metabolism, abnormal, 609698
SEMA3E	104,9	100%	100%	CHARGE syndrome, 214800
SEMA4A	120,6	99%	97%	Retinitis pigmentosa 35, 610282 Cone-rod dystrophy 10, 610283 -3
SEPN1	73,4	88%	80%	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310
SERAC1	88,8	100%	100%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739

SERPINA1	130	100%	100%	Emphysema due to AAT deficiency, 613490 Emphysema-cirrhosis, due to AAT deficiency, 613490 Hemorrhagic diathesis due to \'antithrombin\' Pittsburgh, 613490 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963 (1)
SERPINA3	150	99%	97%	Cerebrovascular disease, occlusive
SERPINB6	122,2	93%	93%	Deafness, autosomal recessive 91, 613453
SERPINB7	103,3	100%	100%	Palmoplantar keratoderma, Nagashima type, 615598
SERPINC1	128,8	100%	100%	Thrombophilia due to antithrombin III deficiency, 613118
SERPINF1	111,4	96%	86%	Osteogenesis imperfecta, type VI, 613982
SERPINH1	141,1	100%	100%	{Preterm premature rupture of the membranes, susceptibility to}, 610504 Osteogenesis imperfecta, type X, 613848
SERPINI1	80,7	98%	95%	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218 -3
SETBP1	142	98%	96%	Schinzel-Giedion midface retraction syndrome, 269150
SETX	139	100%	100%	Ataxia-ocular apraxia-2, 606002 Amyotrophic lateral sclerosis 4, juvenile, 602433
SF3B1	106,9	100%	98%	Myelodysplastic syndrome, somatic, 614286
SF3B4	77,3	100%	97%	Acrofacial dysostosis 1, Nager type, 154400
SFTPA2	23,2	50%	43%	contiguous with SFTPA1 Pulmonary fibrosis, idiopathic, 178500
SFTPB	56,6	97%	91%	Surfactant metabolism dysfunction, pulmonary, 1, 265120
SFTPC	65,9	100%	98%	Surfactant metabolism dysfunction, pulmonary, 2, 610913
SGCA	84,7	89%	81%	Muscular dystrophy, limb-girdle, type 2D, 608099
SGCB	131,3	96%	96%	Muscular dystrophy, limb-girdle, type 2E, 604286
SGCD	103,6	100%	99%	Muscular dystrophy, limb-girdle, type 2F, 601287 Cardiomyopathy, dilated, 1L, 606685

SGCE	82,5	92%	87%	Dystonia-11, myoclonic, 159900
SGCG	89,1	100%	100%	Muscular dystrophy, limb-girdle, type 2C, 253700
SGSH	74,9	94%	93%	Mucopolysaccharidisis type IIIA (Sanfilippo A), 252900
SH2B3	99	99%	94%	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950 Erythrocytosis, somatic, 133100
SH2D1A	87	99%	99%	Lymphoproliferative syndrome, X-linked, 308240
SH3BP2	85,8	91%	88%	Cherubism, 118400
SH3PXD2B	116,1	99%	96%	Frank-ter Haar syndrome, 249420
SH3TC2	96,3	98%	96%	Charcot-Marie-Tooth disease, type 4C, 601596 Mononeuropathy of the median nerve, mild, 613353
SHANK3	68,6	90%	80%	Phelan-McDermid syndrome, 606232 {Schizophrenia 15}, 613950
SHH	94	100%	91%	Holoprosencephaly-3, 142945 Single median maxillary central incisor, 147250 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160
SHOC2	117,9	100%	98%	Noonan-like syndrome with loose anagen hair, 607721
SHOX	0,6	0%	0%	Short stature, idiopathic familial, 300582 Leri-Weill dyschondrosteosis, 127300 Langer mesomelic dysplasia, 249700
SHROOM4	129,2	100%	99%	Stocco dos Santos X-linked mental retardation syndrome, 300434
SI	105	100%	99%	Sucrase-isomaltase deficiency, congenital, 222900
SIGMAR1	88,5	100%	98%	Amyotrophic lateral sclerosis 16, juvenile, 614373
SIL1	93,9	100%	100%	Marinesco-Sjogren syndrome, 248800
SIM1	113,6	100%	98%	Obesity, severe, 601665

SIX1	84,4	100%	100%	Brachiootic syndrome 3, 608389 Deafness, autosomal dominant 23, 605192
SIX3	115,4	100%	100%	Holoprosencephaly-2, 157170 Schizensephaly, 269160
SIX5	39	95%	79%	Branchiootorenal syndrome 2, 610896
SIX6	128,5	98%	87%	Microphthalmia with cataract 2, 212550
SKI	63,3	89%	79%	Shprintzen-Goldberg syndrome, 182212
SKIV2L	17,7	68%	36%	Trichohepatoenteric syndrome 2, 614602
SLC10A2	137,7	100%	100%	Bile acid malabsorption, primary, 613291
SLC12A1	135,9	100%	99%	Bartter syndrome, type 1, 601678
SLC12A3	88,2	100%	99%	Gitelman syndrome, 263800
SLC12A6	91	100%	99%	Agenesis of the corpus callosum with peripheral neuropathy, 218000 -3
SLC16A1	142,9	100%	100%	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia, familial, 7, 610021
SLC16A12	112,2	100%	97%	Cataract, juvenile, with microcornea and glucosuria, 612018
SLC16A2	93,9	100%	99%	Allan-Herndon-Dudley syndrome, 300523
SLC17A5	95,4	100%	99%	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC17A8	126,4	100%	100%	Deafness, autosomal dominant 25, 605583
SLC19A2	89,2	100%	99%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	110,6	100%	99%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A3	116,5	100%	100%	Episodic ataxia, type 6, 612656
SLC20A2	87,9	100%	96%	Basal ganglia cancification, idiopathic, 3, 614540

SLC22A12	80,9	98%	93%	Hypouricemia, renal, 220150
SLC22A5	108,5	100%	98%	Carnitine deficiency, systemic primary, 212140
SLC24A1	134,9	100%	99%	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
SLC24A5	103,9	100%	97%	[Skin/hair/eye pigmentation 4, fair/dark skin], 113750 Albinism, oculocutaneous, type VI, 113750
SLC25A1	69,1	84%	79%	No OMIM phenotype
SLC25A12	114,4	100%	99%	Hypomyelination, global cerebral, 612949
SLC25A13	92,1	100%	98%	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A15	102,9	95%	80%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 -3
SLC25A19	81	100%	99%	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A20	73,5	100%	96%	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A22	73,5	98%	93%	Epileptic encephalopathy, early infantile, 3, 609304
SLC25A3	90,6	100%	100%	Mitochondrial phosphate carrier deficiency, 610773
SLC25A38	69,5	100%	95%	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive, 205950
SLC25A4	111,2	100%	95%	Progressive external ophthalmoplegia with mitochondrial DNA deletions 3, 609283 Mitochondrial DNA depletion syndrome 12 (cardiomyopathic type), 615418
SLC26A2	127,1	100%	100%	Diastrophic dysplasia, 222600 Atelosteogenesis II, 256050 Achondrogenesis Ib, 600972 Epiphyseal dysplasia, multiple, 4, 226900 Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 De la Chapelle dysplasia, 256050
SLC26A3	106,4	100%	96%	?Colon cancer (1) Chloride diarrhea, congenital, Finnish type, 214700

SLC26A4	94,1	99%	97%	Pendred syndrome, 274600 Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791
SLC26A5	89,1	100%	97%	Deafness, autosomal recessive 61, 613865
SLC26A8	100,2	100%	95%	Spermatogenic failure 3, 606766
SLC27A4	78,8	89%	87%	Ichthyosis prematurity syndrome, 608649
SLC29A3	147,2	100%	99%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC2A1	88,3	100%	100%	GLUT1 deficiency syndrome 1, 606777 GLUT1 deficiency syndrome 2, 612126 {Epilepsy, idiopathic generalized, suscpetibility to, 12}, 614847 Dystonia 9, 601042
SLC2A10	89,4	100%	97%	Arterial tortuosity syndrome, 208050
SLC2A2	115,5	100%	100%	{Diabetes mellitus, noninsulin-dependent} Fanconi-Bickel syndrome, 227810
SLC2A9	62,7	100%	94%	{Uric acid concentration, serum, QTL 2}, 612076 Hypouricemia, renal, 2, 612076
SLC30A10	131,8	100%	100%	Hypermanganesemia with dystonia, polycythemia, and cirrhosis, 613280
SLC30A2	79,9	100%	97%	Zinc deficiency, transient neonatal, 608118
SLC33A1	96,9	100%	99%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC34A1	85	98%	95%	Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286 Fanconi renotubular syndrome 2, 613388
SLC34A2	119,8	100%	100%	Pulmonary alveolar microlithiasis, 265100 ?Testicular microlithiasis, 610441
SLC34A3	72,4	99%	89%	Hypophosphatemic rickets with hypercalciuria, 241530
SLC35A1	103,6	100%	99%	Congenital disorder of glycosylation, type IIf, 603585
SLC35A2	103	100%	98%	Congenital disorder of glycosylation, type IIm, 300896

SLC35C1	96,6	100%	100%	Congenital disorder of glycosylation, type IIc, 266265
SLC35D1	112,5	100%	100%	Schneckenbecken dysplasia, 269250
SLC36A2	129,3	100%	100%	Iminoglycinuria, digenic, 242600 Hyperglycinuria, 138500
SLC37A4	84,9	100%	97%	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240
SLC39A13	104,8	100%	99%	Spondylocheirodysplasia, Ehlers-Danlos syndrome-like, 612350
SLC39A4	69,9	100%	97%	Acrodermatitis enteropathica, 201100
SLC3A1	124,5	100%	100%	Cystinuria, 220100
SLC40A1	117,6	99%	96%	Hemochromatosis, type 4, 606069
SLC45A2	95,3	98%	95%	Albinism, oculocutaneous, type IV, 606574 [Skin/hair/eye pigmentation 5, black/nonblack hair], 227240 [Skin/hair/eye pigmentation 5, dark/fair skin], 227240 [Skin/hair/eye pigmentation 5, dark/light eyes], 227240
SLC46A1	80,3	100%	98%	Folate malabsorption, hereditary, 229050
SLC4A1	90,5	100%	95%	Ovalocytosis Spherocytosis, type 4, 612653 [Malaria, resistance to], 611162 Renal tubular acidosis, distal, AD, 179800 Renal tubular acidosis, distal, AR, 611590 [Blood group, Diego], 110500 [Blood group, Waldner], 112010 [Blood group, Wright], 112050 [Blood group, Froese], 601551 [Blood group, Swann], 601550
SLC4A11	112,1	99%	98%	Corneal endothelial dystrophy 2, autosomal recessive, 217700 Corneal endothelial dystrophy and perceptive deafness, 217400 Corneal dystrophy, Fuchs endothelial, 4, 613268
SLC4A4	103,4	100%	100%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278

SLC52A1	127,6	100%	100%	Riboflavin deficiency, 615026
SLC52A2	110,6	100%	100%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	73,8	99%	96%	Brown-Vialetto-Van Laere syndrome 1, 211530 Fazio-Londe disease, 211500
SLC5A1	98,2	100%	98%	Glucose/galactose malabsorption, 606824
SLC5A2	73,7	98%	94%	Renal glucosuria, 233100
SLC5A5	54,6	99%	91%	Thyroid dyshormonogenesis 1, 274400
SLC5A7	101,7	100%	100%	Neuronopathy, distal hereditary motor, type VIIA, 158580
SLC6A19	81,3	97%	94%	Hartnup disorder, 234500 Iminoglycinuria, digenic, 242600 Hyperglycinuria, 138500
SLC6A20	97,4	92%	91%	Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC6A3	74	100%	96%	{Nicotine dependence, protection against}, 188890 Parkinsonism-dystonia, infantile, 613135
SLC6A5	110,4	100%	98%	Hyperekplexia 3, 614618
SLC6A8	7,8	20%	11%	Cerebral creatine deficiency syndrome 1, 300352
SLC7A7	95	100%	99%	Lysinuric protein intolerance, 222700
SLC7A9	70	100%	100%	Cystinuria, 220100
SLC9A3R1	99,9	100%	96%	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
SLC9A6	119	100%	97%	Mental retardation, X-linked syndromic, Christianson type, 300243
SLCO1B1	107,7	100%	97%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO1B3	110,5	100%	95%	Hyperbilirubinemia, Rotor type, digenic, 237450

SLCO2A1	65,8	100%	97%	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
SLITRK1	128,2	100%	100%	Tourette syndrome, 137580 Trichotillomania, 613229
SLITRK6	156,2	100%	100%	Deafness and myopia, 221200
SLURP1	35,7	96%	84%	Meleda disease, 248300
SLX4	128,5	97%	95%	Fanconi anemia, complementation group P, 613951
SMAD3	74,9	97%	81%	Loeys-Dietz syndrome, type 3, 613795
SMAD6	84	94%	81%	Aortic valve disease 2, 614823
SMARCA2	85,8	97%	93%	Nicolaides-Baraitser syndrome, 601358
SMARCA4	82,9	98%	92%	Rhabdoid tumor predisposition syndrome 2, 613325 Mental retardation, autosomal dominant 16, 614609
SMARCAD1	119,7	100%	100%	Adermatoglyphia, 136000
SMARCAL1	118,1	99%	97%	Schimke immunoosseous dysplasia, 242900
SMARCB1	116	100%	100%	Rhabdoid tumors, somatic, 609322 Rhabdoid predisposition syndrome 1, 609322 Mental retardation, autosomal dominant 15, 614608
SMCHD1	105,5	100%	100%	Fascioscapulohumeral muscular dystrophy 2, digenic, 158901
SMN1	1,5	7%	0%	Spinal muscular atrophy-1, 253300 Spinal muscular atrophy-2, 253550 Spinal muscular atrophy-3, 253400 Spinal muscular atrophy-4, 271150
SMOC1	73,7	100%	94%	Microphthalmia with limb anomalies, 206920
SMOC2	76,5	98%	88%	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SMPD1	103,9	97%	90%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616

SMPX	106,6	100%	99%	Deafness, X-linked 4, 300066
SMS	25,5	84%	60%	Mental retardation, X-linked, Snyder-Robinson type, 309583
SNAI2	79,3	100%	100%	Waardenburg syndrome, type 2D, 608890 Piebaldism, 172800
SNAP29	118	100%	100%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNCA	106,1	100%	100%	Parkinson disease 4, 605543 Dementia, Lewy body, 127750 Parkinson disease 1, 168601
SNCB	66,2	100%	100%	Dementia, Lewy body, 127750
SNIP1	132	99%	97%	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501
SNRNP200	110,9	100%	99%	Retinitis pigmentosa 33, 610359
SNRPE	48,6	79%	70%	Hypotrichosis 11, 615059
SNRPN	80,2	100%	85%	Prader-Willi syndrome, 176270
SNX10	104,1	100%	98%	Osteopetrosis, autosomal recessive 8, 615085
SOBP	106	98%	92%	Mental retardation, anterior maxillary protrusion, and strabismus, 613671
SOD1	79,1	100%	100%	Amyotrophic lateral sclerosis 1, 105400
SOS1	113,2	100%	99%	Fibromatosis, gingival, 135300 Noonan syndrome 4, 610733
SOST	103,7	100%	100%	Sclerosteosis, 269500 Van Buchem disease, 239100 Craniodiaphyseal dysplasia, autosomal dominant, 122860
SOX10	67,6	100%	100%	Waardenburg syndrome, type 4C, 613266 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 PCWH syndrome, 609136
SOX17	73,5	100%	100%	Vesicoureteral reflux 3, 613674

SOX18	22,6	76%	44%	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823
SOX2	146,3	100%	100%	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SOX3	83,3	98%	95%	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000
SOX9	114,7	100%	99%	Campomelic dysplasia with autosomal sex reversal, 114290 Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290
SP110	96,2	100%	100%	Hepatic venoocclusive disease with immunodeficiency, 235550 {Mycobacterium tuberculosis, susceptibility to}, 607948
SP7	74,1	100%	100%	Osteogenesis imperfecta, type XII, 613849
SPAG1	114	100%	95%	Ciliary dyskinesia, primary, 28, 615505
SPAST	107,9	100%	100%	Spastic paraplegia 4, autosomal dominant, 182601
SPATA16	122,6	99%	95%	Spermatogenic failure 6, 102530
SPATA7	127,3	100%	99%	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, autosomal recessive, 604232
SPECC1L	119,8	100%	97%	Facial clefting, oblique, 1, 600251
SPG11	104,1	99%	97%	Spastic paraplegia 11, autosomal recessive, 604360
SPG20	108	100%	100%	Troyer syndrome, 275900
SPINK1	108,3	100%	87%	Pancreatitis, hereditary, 167800 {Fibrocalculous pancreatic diabetes, susceptibility to}, 608189 Tropical calcific pancreatitis, 608189
SPINK5	97,3	100%	99%	Netherton syndrome, 256500 Atopy, 147050
SPINT2	55,1	91%	69%	Diarrhea 3, secretory sodium, congenital, syndromic, 270420
SPR	56,2	100%	99%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716

SPRED1	129,3	100%	100%	Legius syndrome, 611431
SPRY4	93,8	100%	100%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
SPTA1	98,7	100%	98%	Elliptocytosis-2, 130600 Pyropoikilocytosis, 266140 Spherocytosis, type 3, 270970
SPTAN1	95,4	99%	97%	Epileptic encephalopathy, early infantile, 5, 613477
SPTB	100,3	99%	98%	Elliptocytosis-3 Spherocytosis, type 2 Anemia, neonatal hemolytic, fatal and near-fatal
SPTBN2	94,6	99%	97%	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia, autosomal recessive 14, 615386
SPTLC1	79,2	94%	88%	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	102,2	100%	98%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SQSTM1	84,7	99%	97%	Paget disease of bone, 602080
SRC	79,5	93%	90%	?Colon cancer, advanced
SRCAP	133,4	100%	99%	Floating-Harbor syndrome, 136140
SRD5A2	57	100%	100%	Pseudovaginal perineoscrotal hypospadias, 264600
SRD5A3	122,5	100%	100%	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713
SRP72	89,9	100%	97%	Bone marrow failure, familial, 614675
SRPX2	81,6	100%	97%	Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643 -3
SRY	1,5	1%	1%	46XY sex reversal 1, 400044 46XX sex reversal 1, 400045
SSTR5	104,2	94%	92%	Somatostatin analog, resistance to, 102200
ST14	85,6	98%	91%	Ichthyosis with hypotrichosis, 610765

ST3GAL3	120,2	100%	100%	Mental retardation, autosomal recessive 12, 611090 Epileptic encephalopathy, early infantile, 15, 615006
STAMBP	122,4	100%	100%	Microcephaly-capillary malformation syndrome, 614261
STAR	106,9	100%	100%	Lipoid adrenal hyperplasia, 201710
STAT1	85,9	100%	98%	Mycobacterial infection, atypical, familial disseminated, 209950 Mycobacterial and viral infections, susceptibility to, autosomal recessive, 613796 Candidiasis, familial, 7, 614162
STAT3	85	100%	96%	Hyper-IgE recurrent infection syndrome, 147060
STAT5B	72,5	83%	75%	Leukemia, acute promyelocytic, STAT5B/RARA type Growth hormone insensitivity with immunodeficiency, 245590
STIL	136,7	100%	99%	Microcephaly 7, primary, autosomal recessive, 612703
STIM1	73,4	94%	89%	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 160565
STK11	73,3	99%	95%	Peutz-Jeghers syndrome, 175200 Melanoma, malignant, somatic Pancreatic cancer, 260350 Testicular tumor, somatic, 273300
STK4	99,9	100%	99%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STOX1	121,2	90%	89%	Preeclampsia/eclampsia 4, 609404
STRA6	72,5	99%	95%	Microphthalmia, syndromic 9, 601186 Microphthalmia, isolated, with coloboma 8, 601186
STRADA	75,6	99%	94%	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087
STRC	17,5	18%	16%	Deafness, autosomal recessive 16, 603720
STS	140,6	100%	99%	nonlyonizing Ichthyosis, X-linked, 308100
STX11	169,7	100%	100%	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STX16	116,4	100%	98%	Pseudohypoparathyroidism, type IB, 603233

STXBP1	91	100%	97%	Epileptic encephalopathy, early infantile, 4, 612164 (2)
STXBP2	84,4	100%	95%	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
SUCLA2	78,4	94%	84%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	89,4	95%	87%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUFU	84,3	98%	89%	Medulloblastoma, desmoplastic, 155255 {Meningioma, familial, susceptibility to}, 607174
SUMF1	64,8	98%	88%	Multiple sulfatase deficiency, 272200
SUMO1	25,5	69%	53%	Orofacial cleft 10, 613705
SUOX	176,2	100%	100%	Sulfite oxidase deficiency, 272300
SURF1	86,7	88%	88%	Leigh syndrome, due to COX deficiency, 256000
SYCP3	126	100%	100%	Spermatogenic failure 4, 270960 {Pregnancy loss, susceptibility to}
SYN1	69,1	96%	75%	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNE1	107,4	99%	97%	Spinocerebellar ataxia, autosomal recessive 8, 610743 Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998
SYNE2	102,3	99%	98%	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999
SYNGAP1	52,9	93%	81%	Mental retardation, autosomal dominant 5, 612621
SYNJ1	100,2	99%	97%	Parkinson disease 20, early-onset, 615530
SYP	93,4	100%	99%	Mental retardation, X-linked 96, 300802
SYT14	132,2	95%	92%	Spinocerebellar ataxia, autosomal recessive 11, 614229
SZT2	100,3	99%	96%	Epileptic encephalopathy, early infantile, 18, 615476
T	117,4	99%	97%	{Neural tube defects, susceptibility to}, 182940

TAB2	160,1	100%	98%	Congenital heart defects, nonsyndromic, 2, 614980
TAC3	71,7	100%	100%	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACR3	154,7	100%	100%	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
TACSTD2	151,9	100%	100%	Corneal dystrophy, gelatinous drop-like, 204870
TAF1	143,7	100%	100%	SVA retrotransposon insertion Dystonia-Parkinsonism, X-linked, 314250
TAF2	101,6	100%	100%	Mental retardation, autosomal recessive 40
TAL1	29,3	93%	70%	Leukemia-1, T-cell acute lymphocytic
TAL2	154	100%	100%	Leukemia-2, T-cell acute lymphoblastic
TALDO1	95,3	100%	100%	Transaldolase deficiency, 606003
TAP1	11,1	47%	9%	Bare lymphocyte syndrome, type I, 604571
TAP2	10,3	29%	13%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571 Wegener-like granulomatosis
TAPBP	19	67%	31%	Bare lymphocyte syndrome, type I, 604571
TARDBP	29,2	44%	33%	Amyotrophic lateral sclerosis 10, with or without FTD, 612069 Frontotemporal lobar degeneration, TARDBP-related, 612069
TAT	92	100%	98%	Tyrosinemia, type II, 276600
TAZ	102,3	100%	100%	Barth syndrome, 302060
TBC1D20	78,1	94%	92%	Warburg micro syndrome 4, 615663
TBC1D24	112,3	100%	100%	Myoclonic epilepsy, infantile, familial, 605021 Epileptic encephalopathy, early infantile, 16, 615338
TBCE	117,6	100%	100%	Kenny-Caffey syndrome-1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410
TBP	100	100%	98%	Spinocerebellar ataxia 17, 607136 {Parkinson disease, susceptibility to}, 168600

TBX1	66,3	72%	66%	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400 Velocardiofacial syndrome, 192430 Tetralogy of Fallot, 187500
TBX15	91,1	100%	98%	Cousin syndrome, 260660
TBX19	141,1	100%	99%	Adrenocorticotropic hormone deficiency, 201400
TBX20	44,9	74%	67%	Atrial septal defect 4, 611363
TBX21	95	89%	82%	{Asthma, aspirin-induced, susceptibility to}, 208550 Asthma and nasal polyps, 208550
TBX22	171,8	100%	99%	Cleft palate with ankyloglossia, 303400 ?Abruzzo-Erickson syndrome, 302905
TBX3	76,1	98%	93%	Ulnar-mammary syndrome, 181450
TBX4	108	92%	84%	Small patella syndrome, 147891
TBX5	83	99%	93%	Holt-Oram syndrome, 142900
TBXAS1	94,6	100%	98%	Ghosal hematodiaphyseal syndrome, 231095 ?Thromboxane synthase deficiency, 614158 (1)
TCAP	43,3	69%	51%	Muscular dystrophy, limb-girdle, type 2G, 601954 Cardiomyopathy, dilated, 1N, 607487
TCF12	116,9	100%	100%	Craniosynostosis 3, 615314
TCF4	90,8	97%	97%	Pitt-Hopkins syndrome, 610954
TCIRG1	74,8	92%	84%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	104,1	100%	97%	Transcobalamin II deficiency, 275350
TCOF1	95,3	99%	97%	Treacher Collins syndrome 1, 154500
TCTN2	92	100%	98%	Meckel syndrome 8, 613885
TCTN3	99,3	100%	100%	Orofaciodigital syndrome IV, 258860 Joubert syndrome 18, 614815

TDGF1	75,7	96%	88%	Forebrain defects
TDP1	106,4	100%	99%	Spinocerebellar ataxia, autosomal recessive with axonal neuropathy, 607250
TDRD7	112,1	100%	100%	Cataract 36, 613887
TEAD1	82,1	100%	98%	Sveinsson choreoretinal atrophy, 108985
TECPR2	108,4	100%	99%	Spastic paraplegia 49, autosomal recessive, 615031
TECR	83,1	100%	91%	Mental retardation, autosomal recessive 14, 614020
TECTA	114,6	99%	97%	Deafness, autosomal dominant 8/12, 601543 Deafness, autosomal recessive 21, 603629
TEK	108	100%	98%	Venous malformations, multiple cutaneous and mucosal, 600195
TET2	127	100%	99%	Myelodysplastic syndrome, somatic, 614286
TEX28	0	0%	0%	No OMIM phenotype Mental retardation, x-linked 99 Blue cone monochromacy Achromatopsia Colorblindness
TF	99,8	98%	95%	Atransferrinemia, 209300
TFAP2A	72,3	100%	90%	Branchiooculofacial syndrome, 113620
TFAP2B	102,2	100%	100%	Char syndrome, 169100
TFE3	77,9	97%	92%	Renal cell carcinoma, papillary, 1, 300854
TFG	116,8	100%	95%	Hereditary motor and sensory neuropathy, proximal type, 604484 Chondrosarcoma, extraskeletal myxoid, 612237 (1)
TFR2	74,2	96%	86%	Hemochromatosis, type 3, 604250
TG	100	100%	95%	Thyroid dyshormonogenesis 3, 274700 {Autoimmune thyroid disease, susceptibility to, 3}, 608175

TGFB1	53,4	98%	87%	Camurati-Engelmann disease, 131300 {Cystic fibrosis lung disease, modifier of}, 219700
TGFB2	124,1	97%	94%	Loeys-Dietz syndrome, type 4, 614816
TGFB3	100,6	100%	98%	Arrhythmogenic right ventricular dysplasia 1, 107970

TGFBI	101,6	100%	100%	Corneal dystrophy, Groenouw type I, 121900 Corneal dystrophy, lattice type I, 122200 Corneal dystrophy, Reis-Bucklers type, 608470 Corneal dystrophy, Avellino type, 607541 Corneal dystrophy, lattice type IIIA, 608471 Corneal dystrophy, Thiel-Behnke type, 602082 Corneal dystrophy, epithelial basement membrane, 121820
TGFBR1	116,9	96%	94%	Loeys-Dietz syndrome, type 1A, 609192 Loeys-Dietz syndrome, type 2A, 608967 {Multiple self-healing squamous epithelioma, susceptiblity to}, 132800
TGFBR2	83,1	100%	95%	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome, type 1B, 610168 Loeys-Dietz syndrome, type 2B, 610380
TGM1	101,1	100%	96%	Ichthyosis, congenital, autosomal recessive 1, 242300
TGM5	98	100%	97%	Peeling skin syndrome, acral type, 609796
TGM6	63,1	92%	84%	Spinocerebellar ataxia 35, 613908
TH	77,8	95%	87%	Segawa syndrome, recessive, 605407
THAP1	121,8	100%	100%	Dystonia 6, torsion, 602629
THBD	74,2	100%	100%	Thrombophilia due to thrombomodulin defect, 614486 {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
THOC6	157,1	100%	97%	Beaulieu-Boycott-Innes syndrome, 613680
THPO	100,2	98%	92%	Thrombocythemia 1, 187950

THRA	122,4	100%	99%	Hypothyroidism, congenital, nongoitrous, 6, 614450

THRB	114,3	100%	100%	Thyroid hormone resistance, 188570 Thyroid hormone resistance, autosomal recessive, 274300 Thyroid hormone resistance, selective pituitary, 145650
TIA1	107,3	100%	100%	Welander distal myopathy, 604454
TIMM8A	57,3	94%	85%	Deafness, X-linked 1, progressive Mohr-Tranebjaerg syndrome, 304700 Jensen syndrome, 311150
TIMP3	118,3	100%	100%	Sorsby fundus dystrophy, 136900
TINF2	170,7	100%	100%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TJP2	85,9	100%	97%	Hypercholanemia, familial, 607748
TK2	82,9	100%	95%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560
TLL1	107,4	100%	99%	Atrial septal defect 6, 613087
TLR4	150,9	100%	99%	Endotoxin hyporesponsiveness {Macular degeneration, age-related, 10}, 611488 {Colorectal cancer, susceptibility to}, 114500
TMC1	106,6	100%	100%	Deafness, autosomal recessive 7, 600974 Deafness, autosomal dominant 36, 606705
TMC6	57,9	99%	94%	Epidermodysplasia verruciformis, 226400
TMC8	76,1	99%	94%	Epidermodysplasia verruciformis, 226400
TMCO1	71,3	100%	93%	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 614132
TMEM126A	75	100%	95%	Optic atrophy-7, 612989
TMEM138	90,6	100%	100%	Joubert syndrome 16, 614465
TMEM165	86,6	100%	100%	Congenital disorder of glycosylation, type IIk, 614727

TMEM216	53,4	87%	78%	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	73,4	96%	92%	Joubert syndrome 20, 614970 Meckel syndrome, type 11, 615397 -3
TMEM237	91,2	100%	99%	Joubert syndrome 14, 614424
TMEM38B	111,9	100%	100%	Osteogenesis imperfecta, type XIV, 615066
TMEM43	79,8	100%	97%	Arrhythmogenic right ventricular dysplasia 5, 604400 Emery-Dreifuss muscular dystrophy 7, AD, 614302
TMEM5	141,3	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
TMEM67	106,4	100%	99%	Meckel syndrome 3, 607361 Joubert syndrome 6, 610688 {Bardet-Biedl syndrome 14, modifier of}, 209900 COACH syndrome, 216360 Nephronophthisis 11, 613550
TMEM70	163,9	100%	100%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMIE	58,1	99%	91%	Deafness, autosomal recessive 6, 600971
TMLHE	68,4	80%	78%	Epsilon-trimethyllysine hydroxylase deficiency, 300872
TMPRSS3	91,1	100%	96%	Deafness, autosomal recessive 8/10, 601072
TMPRSS6	70,8	98%	92%	Iron-refractory iron deficiency anemia, 206200
TNC	126,5	100%	98%	Deafness, autosomal dominant 56, 615629
TNFRSF10B	99,5	100%	100%	Squamous cell carcinoma, head and neck, 275355
TNFRSF11A	96,3	96%	94%	Osteolysis, familial expansile, 174810 Paget disease of bone, 602080 Osteopetrosis, autosomal recessive 7, 612301
TNFRSF11B	166,1	100%	100%	Paget disease, juvenile, 239000

TNFRSF13B	59	99%	93%	Immunoglobulin A deficiency 2, 609529 Immunodeficiency, common variable, 2, 240500
TNFRSF13C	54,1	100%	81%	Immunodeficiency, common variable, 4, 613494
TNFRSF1A	72	96%	91%	Periodic fever, familial, 142680 {Multiple sclerosis, susceptibility to, 5}, 614810
TNFSF11	145,7	100%	100%	Osteopetrosis, autosomal recessive 2, 259710
TNNC1	108,5	100%	100%	Cardiomyopathy, dilated, 1Z, 611879 Cardiomyopathy, familial hypertrophic, 13, 613243
TNNI2	78,4	100%	97%	Arthrogryposis multiplex congenita, distal, type 2B, 601680
TNNI3	77,5	100%	87%	Cardiomyopathy, familial hypertrophic, 7, 613690 Cardiomyopathy, familial restrictive, 115210 Cardiomyopathy, dilated, 2A, 611880 Cardiomyopathy, dilated, 1FF, 613286
TNNT1	83,9	93%	93%	Nemaline myopathy 5, Amish type, 605355
TNNT2	94,7	99%	95%	Cardiomyopathy, familial hypertrophic, 2, 115195 Cardiomyopathy, dilated, 1D, 601494 Cardiomyopathy, familial restrictive, 3, 612422 Left ventricular noncompaction 6, 601494
TNNT3	77,1	96%	83%	Arthyrgryposis, distal, type 2B, 601680
TNXB	8,5	24%	8%	Ehlers-Danlos syndrome, autosomal recessive, due to tenascin X deficiency, 606408 Ehlers-Danlos syndrome, autosomal dominant, hypermobility type, 130020
TOP1	100,7	100%	97%	DNA topoisomerase I, camptothecin-resistant
TOP2A	116,3	99%	97%	DNA topoisomerase II, resistance to inhibition of, by amsacrine
TOPORS	148,1	100%	100%	Retinitis pigmentosa 31, 609923

TP53	82,4	100%	100%	Colorectal cancer, 114500 Li-Fraumeni syndrome, 151623 Hepatocellular carcinoma, 114550 Osteosarcoma, 259500 Choroid plexus papilloma, 260500 Nasopharyngeal carcinoma, 607107 Pancreatic cancer, 260350 Adrenal cortical carcinoma, 202300 Breast cancer, 114480 {Basal cell carcinoma 7}, 614740 {Glioma susceptibility 1}, 137800
TP63	128,7	100%	100%	Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 Split-hand/foot malformation 4, 605289 Hay-Wells syndrome, 106260 ADULT syndrome, 103285 Limb-mammary syndrome, 603543 Rapp-Hodgkin syndrome, 129400 Orofacial cleft 8, 129400
TPI1	63,7	96%	94%	Hemolytic anemia due to triosephosphate isomerase deficiency
TPK1	80,4	100%	100%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TPM1	80,8	98%	94%	Cardiomyopathy, familial hypertrophic, 3, 115196 Cardiomyopathy, dilated, 1Y, 611878 Left ventricular noncompaction 9, 611878
TPM2	93,8	100%	96%	Arthrogryposis multiplex congenita, distal, type 1, 108120 Arthrogryposis, distal, type 2B, 601680 Nemaline myopathy 4, autosomal dominant, 609285 CAP myopathy 2, 609285
TPM3	61,5	82%	74%	Nemaline myopathy 1, autosomal dominant or recessive, 609284 CAP myopathy 1, 609284 Myopathy congenital, with fiber-type disproportion, 255310
TPMT	93,7	100%	100%	6-mercaptopurine sensitivity, 610460
TPO	76,9	98%	93%	Thyroid dyshormonogenesis 2A, 274500

TPP1	119,2	100%	96%	Ceroid lipofuscinosis, neuronal, 2, 204500
TPRN	42,6	82%	74%	Deafness, autosomal recessive 79, 613307
TRAPPC11	109,7	100%	99%	Muscular dystrophy, limb-girdle, type 2S, 615356
TRAPPC2	52,4	97%	74%	Spondyloepiphyseal dysplasia tarda, 313400
TRAPPC9	67,4	98%	94%	Mental retardation, autosomal recessive 13, 613192
TRDN	75,2	99%	92%	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441
TREM2	98,1	100%	99%	Nasu-Hakola disease, 221770
TREX1	135,6	100%	100%	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700
TRHR	129,8	100%	98%	Thyrotropin-releasing hormone resistance, generalized
TRIM24	98	100%	97%	Thyroid carcinoma, papillary, 188550
TRIM32	103,2	100%	100%	Muscular dystrophy, limb-girdle, type 2H, 254110 Bardet-Biedl syndrome 11, 209900
TRIM33	90,7	100%	84%	Thyroid carcinoma, papillary, 188550
TRIM37	95,2	100%	98%	Mulibrey nanism, 253250
TRIOBP	95	96%	92%	Deafness, autosomal recessive 28, 609823
TRIP11	115,1	99%	97%	Achondrogenesis, type IA, 200600
TRMU	75,9	100%	95%	{Deafness, mitochondrial, modifier of}, 580000 Liver failure, transient infantile, 613070
TRPA1	63,6	83%	79%	Episodic pain syndrome, familial, 615040
TRPC6	81,4	95%	90%	Glomerulosclerosis, focal segmental, 2, 603965

TRPM1	130,7	98%	97%	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TRPM4	83,7	99%	98%	Progressive familial heart block, type IB, 604559
TRPM6	113,9	100%	98%	Hypomagnesemia 1, intestinal, 602014
TRPS1	131,4	100%	100%	Trichorhinophalangeal syndrome, type I, 190350 Trichorhinophalangeal syndrome, type III, 190351
TRPV3	107,2	100%	94%	Olmsted syndrome, 614594
TRPV4	96,7	100%	99%	Brachyolmia type 3, 113500 Spondylometaphyseal dysplasia, Kozlowski type, 184252 Metatropic dysplasia, 156530 Hereditary motor and sensory neuropathy, type IIc, 606071 Scapuloperoneal spinal muscular atrophy, 181405 [Sodium serum level QTL 1], 613508 Parastremmatic dwarfism, 168400 SED, Maroteaux type, 184095 Spinal muscular atrophy, distal, congenital nonprogressive, 600175 Digital arthropathy-brachydactyly, familial, 606835
TSC1	91,1	99%	97%	Otosclerosis 1 (2)
TSC1	91,1	99%	97%	Tuberous sclerosis-1, 191100 Lymphangioleiomyomatosis, 606690 Focal cortical dysplasia, Taylor balloon cell type, 607341
TSC2	83,9	98%	95%	distal to PKD1 Tuberous sclerosis-2, 613254 Lymphangioleiomyomatosis, somatic, 606690
TSEN2	113,6	100%	100%	Pontocerebellar hypoplasia type 2B, 612389
TSEN34	56,9	100%	91%	Pontocerebellar hypoplasia type 2C, 612390
TSEN54	102,3	96%	96%	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753
TSFM	106	99%	94%	Combined oxidative phosphorylation deficiency 3, 610505
TSG101	102,7	100%	100%	Breast cancer, somatic, 114480

TSHB	165,6	100%	100%	Hypothryoidism, congenital, nongoitrous 4, 275100
TSHR	153,2	99%	97%	Hypothyroidism, congenital, nongoitrous, 1 275200 Thyroid adenoma, hyperfunctioning, somatic Hyperthyroidism, nonautoimmune, 609152 Thyroid carcinoma with thyrotoxicosis Hyperthyroidism, familial gestational, 603373
TSHZ1	114,1	100%	99%	Aural atresia, congenital, 607842
TSPAN12	104,9	100%	100%	Exudative vitreoretinopathy 5, 613310
TSPAN7	86,4	99%	95%	Mental retardation, X-linked 58, 300210
TSPEAR	108,5	100%	99%	Deafness, autosomal recessive 98, 614861
TSPYL1	181,6	100%	100%	Sudden infant death with dysgenesis of the testes syndrome, 608800 -3
TTBK2	117,5	100%	99%	Spinocerebellar ataxia 11, 604432
TTC19	63,6	80%	72%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTC21B	105,2	99%	98%	Nephronophthisis 12, 613820 Asphyxiating thoracic dystrophy 4, 613819
TTC37	111,4	100%	100%	Trichohepatoenteric syndrome 1, 222470
TTC7A	62,8	97%	94%	Intestinal atresia, multiple, 243150
TTC8	99,5	100%	99%	Bardet-Biedl syndrome 8, 209900 Retinitis pigmentosa 51, 613464
TTI2	109,9	99%	98%	Mental retardation, autosomal recessive 39, 615541
TTN	139,6	98%	97%	Cardiomyopathy, familial hypertrophic, 9, 613765 Cardiomyopathy, dilated, 1G, 604145 Tibial muscular dystrophy, tardive, 600334 Muscular dystrophy, limb-girdle, type 2J, 608807 Myopathy, proximal, with early respiratory muscle involvement, 603689 Myopathy, early-onset, with fatal cardiomyopathy, 611705

TTPA	84,8	100%	90%	Ataxia with isolated vitamin E deficiency, 277460
TTR	81,4	100%	98%	Amyloidosis, hereditary, transthyretin-related, 105210 [Dystransthyretinemic hyperthyroxinemia], 145680 Carpal tunnel syndrome, familial, 115430
TUBA1A	23,6	90%	51%	Lissencephaly 3, 611603
TUBA8	95	99%	97%	Polymicrogyria with optic nerve hypoplasia, 613180
TUBB1	135,1	100%	100%	Macrothrombocytopenia, autosomal dominant, TUBB1-related, 613112
TUBB2A	47,6	100%	93%	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB2B	54,7	100%	97%	Polymicrogyria, symmetric or asymmetric, 610031
TUBB3	76,4	82%	79%	Fibrosis of extraocular muscles, congenital, 3A, 600638 Cortical dysplasia, complex, with other brain malformations 1, 614039
TUBB4A	57,7	76%	71%	?Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438
TUBG1	104,1	86%	78%	Cortical dysplasia, complex, with other brain malformations 4, 615412
TUBGCP6	113,9	99%	98%	Microcephaly and chorioretinopathy with or without mental retardation, 251270
TUFM	107,4	100%	95%	Combined oxidative phosphorylation deficiency 4, 610678
TULP1	90,5	98%	91%	Retinitis pigmentosa 14, 600132 Leber congenital amaurosis 15, 613843
TUSC3	116,9	100%	100%	Mental retardation, autosomal recessive 7, 611093
TWIST1	116,4	100%	99%	Saethre-Chotzen syndrome, 101400 Saethre-Chotzen syndrome with eyelid anomalies, 101400 Craniosynostosis, type 1, 123100 Robinow-Sorauf syndrome, 180750
TWIST2	79,7	100%	99%	Focal facial dermal dysplasia 3, Setleis type, 227260
TYK2	86,4	99%	95%	Tyrosine kinase 2 deficiency, 611521

TYMP	79,2	100%	93%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYR	138	74%	74%	Albinism, oculocutaneous, type IA, 203100 Waardenburg syndrome/albinism, digenic, 103470 Albinism, oculocutaneous, type IB, 606952 [Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 {Melanoma, cutaneous malignant, susceptibility to, 8}, 601800 [Skin/hair/eye pigmentation 3, blue/green eyes], 601800
TYROBP	60,5	100%	100%	Nasu-Hakola disease, 221770
TYRP1	107,1	100%	99%	Albinism, oculocutaneous, type III, 203290 Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair), 612271
UBA1	127,2	100%	100%	Spinal muscular atrophy, X-linked 2, infantile, 301830
UBE2A	103,8	100%	100%	Mental retardation, X-linked syndromic, Nascimento-type, 300860
UBE3A	101,5	100%	99%	Angelman syndrome, 105830
UBE3B	99,8	100%	98%	Blepharophimosis-ptosis-intellectual disability syndrome, 615057 -3
UBIAD1	94,2	100%	100%	Corneal dystrophy, Schnyder type, 121800
UBQLN2	145,7	100%	100%	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857
UBR1	101	100%	100%	Johanson-Blizzard syndrome, 243800
UGT1A1	137,1	100%	96%	Crigler-Najjar syndrome, type I, 218800 [Gilbert syndrome], 143500 Crigler-Najjar syndrome, type II, 606785 Hyperbilirubinemia, familial transcient neonatal, 237900 [Bilirubin, serum level of, QTL1], 601816
UMOD	78	100%	97%	Hyperuricemic nephropathy, familial juvenile 1, 162000 Medullary cystic kidney disease 2, 603860 Glomerulocystic kidney disease with hyperuricemia and isosthenuria, 609886
UMPS	106,7	100%	100%	Orotic aciduria, 258900
UNC93B1	40,3	54%	53%	Herpes simplex encephalitis, susceptibility to, 1, 610551

UNG	67,5	92%	85%	Immunodeficiency with hyper IgM, type 5, 608106
UPB1	114,7	100%	97%	Beta-ureidopropionase deficiency, 613161
UPF3B	105,4	100%	96%	Mental retardation, X-linked, syndromic 14, 300676
UQCRB	103,9	100%	100%	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC2	84,1	98%	96%	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRQ	55,8	100%	90%	Mitochondrial complex III deficiency, nuclear type 4, 615159
UROC1	72,7	99%	95%	Urocanase deficiency, 276880
UROD	84,4	99%	91%	Porphyria cutanea tarda, 176100 Porphyria, hepatoerythropoietic, 176100
UROS	79	95%	82%	Porphyria, congenital erythropoietic, 263700
USH1C	75,6	98%	93%	Acadian and Samaritan variety Usher syndrome, type 1C, 276904 Deafness, autosomal recessive 18A, 602092
USH2A	109,7	99%	98%	Usher syndrome, type 2A, 276901 Retinitis pigmentosa 39, 613809 -3
USP9Y	0,2	0%	0%	Spermatogenic failure, Y-linked, 2, 415000
UVSSA	65,5	100%	95%	UV-sensitive syndrome 3, 614640
VANGL1	146,4	100%	100%	Caudal regression syndrome, 600145 Neural tube defects, 182940 -3
VANGL2	111	99%	95%	Neural tube defects, 182940
VAPB	145,2	99%	93%	Amyotrophic lateral sclerosis 8, 608627 Spinal muscular atrophy, late-onset, Finkel type, 182980
VAX1	82	98%	92%	Microphthalmia, syndromic 11, 614402
VCAN	142,1	100%	99%	Wagner syndrome 1, 143200
VCL	100	97%	93%	Cardiomyopathy, dilated, 1W, 611407 Cardiomyopathy, familial hypertrophic, 15, 613255

VCP	110,5	100%	97%	Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320 Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954
VDR	76,9	100%	99%	Rickets, vitamin D-resistant, type IIA, 277440 ?Osteoporosis, involutional, 166710 (1)
VHL	121,9	100%	100%	von Hippel-Lindau syndrome, 193300 Renal cell carcinoma, somatic, 144700 Pheochromocytoma, 171300 Hemangioblastoma, cerebellar, somatic Erythrocytosis, familial, 2, 263400
VIM	104,9	100%	100%	Cataract 30, pulverulent, 116300
VIPAS39	110,2	100%	97%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VKORC1	128,1	100%	100%	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473 Warfarin resistance, 122700
VLDLR	106,5	100%	99%	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VPS13A	113,6	100%	98%	Choreoacanthocytosis, 200150
VPS13B	104,4	99%	98%	Cohen syndrome, 216550
VPS33B	101,6	100%	98%	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
VPS35	73,9	96%	89%	Parkinson disease 17, 614203
VPS37A	69,1	100%	95%	Spastic paraplegia 53, autosomal recessive, 614898
VPS45	105,3	95%	95%	Neutropenia, severe congenital, 5, autosomal recessive, 615285
VRK1	124,6	100%	100%	Pontocerebellar hypoplasia type 1A, 607596
VSX1	54	100%	91%	Keratoconus 1, 148300 Corneal dystrophy, hereditary polymorphous posterior, 122000 Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195
VWF	59,7	79%	72%	von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 von Willebrand disease, type 1, 193400 von Willibrand disease, type 3, 277480

WAS	60,1	100%	90%	Wiskott-Aldrich syndrome, 301000 Thrombocytopenia, X-linked, 313900 Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, intermittent, 313900
WDR11	92,3	100%	99%	Hypogonadotropic hypogonadism 14 with or without anosmia, 614858
WDR19	115,8	100%	99%	Asphyxiating thoracic dystrophy 5, 614376 Nephronophthisis 13, 614377 Cranioectodermal dysplasia 4, 614378
WDR34	88,3	100%	98%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
WDR35	109,2	100%	98%	Cranioectodermal dysplasia 2, 613610 Short rib-polydactyly syndrome, type V, 614091
WDR36	109,1	100%	93%	Glaucoma 1, open angle, G, 609887
WDR45	91,1	100%	99%	Neurodegeneration with brain iron acculation 5, 300894
WDR60	101,9	99%	97%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WDR62	103,8	99%	94%	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
WDR72	111,5	96%	96%	Amelogenesis imperfecta, hypomaturation type, IIA3, 613211
WDR81	112,6	100%	97%	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185
WFS1	146,8	100%	99%	Wolfram syndrome, 222300 Deafness, autosomal dominant 6/14/38, 600965 Wolfram-like syndrome, autosomal dominant, 614296 {Diabetes mellitus, noninsulin-dependent, association with}, 125853
WHSC1L1	116,3	99%	97%	Leukemia, acute myeloid, 601626
WIPF1	92,2	97%	96%	Wiskott-Aldrich syndrome 2, 614493
WISP3	125,3	100%	100%	Arthropathy, progressive pseudorheumatoid, of childhood, 208230 Spondyloepiphyseal dysplasia tarda with progressive arthropathy, 208230
WNK1	135,6	99%	99%	Pseudohypoaldosteronism, type IIC, 614492 Neuropathy, hereditary sensory and autonomic, type II, 201300

WNK4	111,8	100%	99%	Pseudohypoaldosteronism, type IIB, 614491
WNT1	134,4	98%	90%	Osteogenesis imperfecta, type XV, 615220 {Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221
WNT10A	68,9	94%	88%	Odontoonychodermal dysplasia, 257980 Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400
WNT10B	90,7	100%	93%	Split-hand/foot malformation 6, 225300
WNT3	138,7	98%	88%	Tetra-amelia, autosomal recessive, 273395
WNT4	137	92%	92%	SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330
WNT5A	111,3	100%	98%	Robinow syndrome, autosomal dominant, 180700
WNT7A	129,9	100%	100%	Ulna and fibula, absence of, with sever limb deficiency, 276820 Fuhrmann syndrome, 228930
WRAP53	133,9	100%	99%	Dyskeratosis congenita, autosomal recessive 3, 613988
WT1	60,5	100%	99%	Wilms tumor, type 1, 194070 Denys-Drash syndrome, 194080 Nephrotic syndrome, type 4, 256370 Frasier syndrome, 136680 Meacham syndrome, 608978 Mesothelioma, somatic, 156240
WWOX	107,1	100%	99%	Esophageal squamous cell carcinoma, 133239
XDH	90	100%	99%	Xanthinuria, type I, 278300
XIAP	130,3	90%	82%	Lymphoproliferative syndrome, X-linked, 2, 300635
XK	134,6	100%	100%	McLeod syndrome with or without chronic granulomatous disease, 300842
XPA	80,9	100%	93%	Xeroderma pigmentosum, group A, 278700
XPC	114,6	99%	97%	Xeroderma pigmentosum, group C, 278720

XPNPEP3	113,5	100%	98%	Nephronophthisis-like nephropathy 1, 613159
YAP1	71,5	96%	86%	Coloboma, ocular, 120433 Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433
YARS	94,4	100%	96%	Charcot-Marie-Tooth disease, dominant intermediate C, 608323
YARS2	99,4	100%	100%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
ZAP70	86,7	96%	87%	Selective T-cell defect, 269840
ZBTB16	115,8	100%	99%	Leukemia, acute promyelocytic, PL2F/RARA type Skeletal defects, genital hypoplasia, and mental retardation, 612447
ZBTB24	162,4	100%	99%	Immunodeficiency-centromeric instability-facial anomalies syndrome-2, 614069
ZC4H2	98,2	100%	99%	Wieacker-Wolf syndrome, 314580
ZDHHC9	91,3	100%	99%	Mental retardation, X-linked syndromic, Raymond type, 300799
ZEB1	140,1	98%	97%	Corneal dystrophy, posterior polymorphous, 3, 609141 Corneal dystrophy, Fuchs endothelial, 6, 613270
ZEB2	151,6	100%	99%	Mowat-Wilson syndrome, 235730
ZFP57	18,2	82%	40%	Diabetes mellitus, transient neonatal, 1, 601410
ZFPM2	178,9	98%	98%	Tetralogy of Fallot, 187500 Diaphragmatic hernia 3, 610187
ZFYVE26	87,2	97%	91%	Spastic paraplegia 15, autosomal recessive, 270700
ZFYVE27	77,6	98%	96%	Spastic paraplegia 33, autosomal dominant, 610244
ZIC2	62,3	93%	87%	Holoprosencephaly-5, 609637
ZIC3	95,6	100%	100%	Heterotaxy, visceral, 1, X-linked 306955 Congenital heart defects, nonsyndromic, 1, X-linked, 306955 VACTERL association, X-linked, 314390
ZMPSTE24	137,9	100%	100%	Mandibuloacral dysplasia with type B lipodystrophy, 608612 Restrictive dermopathy, lethal, 275210

ZMYND10	90,6	99%	94%	Ciliary dyskinesia, primary, 22, 615444
ZNF335	79,7	98%	93%	Microcephaly 10, primary, autosomal recessive, 615095
ZNF423	128,4	100%	99%	Nephronophthisis 14, 614844 Joubert syndrome 19, 614844
ZNF469	91,7	100%	99%	Brittle cornea syndrome, 229200
ZNF513	110,6	100%	96%	Retinitis pigmentosa 58, 613617
ZNF592	107,3	93%	91%	Spinocerebellar ataxia, autosomal recessive 5, 606937
ZNF644	149,8	100%	99%	Myopia 21, autosomal dominant, 614167
ZNF711	134,6	100%	100%	Mental retardation, X-linked 97, 300803
ZNF750	120,6	100%	100%	Seborrhea-like dermatitis with psoriasiform elements, 610227
ZNF81	108,5	100%	99%	Mental retardation, X-linked 45, 300498

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated October 2013
Median Coverage describes the average number of reads seen across 50 exomes
% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x
% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x
OMIM release used for OMIM disease identifiers and descriptions : 15 october 2013
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors