

# MULTIPLE CONGENITAL ANOMALIES GENE PANEL DG 2.5/2.6

<i>Gene</i>	<i>Median coverage</i>	<i>% covered &gt; 10x</i>	<i>% covered &gt; 20x</i>	<i>Associated Phenotype description and OMIM disease ID</i>
A4GALT	115.9	100%	100%	NOR polyagglutination syndrome,111400
AAAS	87.2	100%	99%	Achalasia-addisonianism-alacrimia syndrome,231550
AAGAB	153.2	97%	92%	Keratoderma palmoplantar punctate type IA,148600
AARS	116.4	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 Epileptic encephalopathy,early infantile,29,616339
AARS2	111	99%	97%	Combined oxidative phosphorylation deficiency 8, 614096
AASS	131.8	100%	95%	Hyperlysinemia, 238700 Saccharopinuria, 268700
ABAT	86.7	100%	97%	GABA-transaminase deficiency, 613163
ABCA1	119.2	99%	97%	HDL deficiency,type 2,604091 Tangier disease,205400 {Coronary artery disease in familial hypercholesterolemia,protection against},143890
ABCA12	145.7	98%	96%	Ichthyosis, autosomal recessive 4B (harlequin),242500 Ichthyosis, congenital, autosomal recessive 4A,601277
ABCA3	111.3	99%	98%	Surfactant metabolism dysfunction,pulmonary,3,610921
ABCA4	112.8	100%	98%	Cone-rod dystrophy 3, 604116 Fundus flavimaculatus, 248200 Retinal dystrophy, early-onset severe, 248200 Retinitis pigmentosa 19, 601718 Stargardt disease 1, 248200 {Macular degeneration, age-related, 2,} 153800
ABCB11	161.6	100%	97%	Cholestasis,benign recurrent intrahepatic,2,605479 Cholestasis,progressive familial intrahepatic 2,601847
ABCB4	126.3	99%	96%	Cholestasis,intrahepatic,of pregnancy,3,614972 Cholestasis,progressive familial intrahepatic 3,602347 Gallbladder disease 1,600803
ABCB6	111.6	100%	99%	Dyschromatosis universalis hereditaria 3,615402 Microphthalmia,isolated, with coloboma 7,614497 [Blood group, Langereis system],111600

ABCB7	85.3	96%	93%	Anemia, sideroblastic, with ataxia, 301310
ABCC2	142.4	100%	100%	Dubin-Johnson syndrome,237500
ABCC6	94.8	93%	91%	Arterial calcification generalized of infancy 2,614473 Pseudoxanthoma elasticum,264800 Pseudoxanthoma elasticum, forme fruste,177850
ABCC8	139.6	100%	100%	Hyperinsulinemic hypoglycemia, familial, 1, 256450
ABCC9	156.9	100%	99%	Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 10, 608569 Hypertrichotic osteochondrodysplasia, 239850
ABCD1	49.6	69%	64%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABCD4	117.2	98%	93%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	121.1	100%	100%	Sitosterolemia, 210250
ABCG8	137.9	99%	96%	Gallbladder disease 4, 611465 Sitosterolemia, 210250
ABHD12	93.7	89%	78%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 614857
ABHD5	244.7	100%	100%	Chanarin-Dorfman syndrome, 275630
ABL1	115.5	100%	99%	Leukemia,Philadelphia chromosome-positive,resistant to imatinib
ACAD8	123.5	100%	100%	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	137.4	99%	97%	ACAD9 deficiency, 611126
ACADM	96.1	94%	92%	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	123.8	99%	96%	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	123.6	98%	89%	2-methylbutyrylglycinuria, 610006
ACADVL	107.6	99%	96%	VLCAD deficiency, 201475
ACAN	99.5	90%	85%	Osteochondritis dissecans, short stature, and early-onset osteoarthritis, 165800 Spondyloepimetaphyseal dysplasia, aggrecan type, 612813 Spondyloepiphyseal dysplasia, Kimberley type, 608361
ACAT1	120.3	98%	93%	Alpha-methylacetoacetic aciduria, 203750
ACE	106.4	93%	91%	Renal tubular dysgenesis,267430 [Angiotensin I-converting enzyme,benign serum increase] {Alzheimer disease,susceptibility to},104300 {Microvascular complications of diabetes 3},612624 {Myocardial infarction,susceptibility to} {SARS,progression of}

				{Stroke,hemorrhagic},614519
ACO2	111.4	95%	89%	Infantile cerebellar-retinal degeneration, 614559
ACOX1	163.5	100%	100%	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACP5	190.6	100%	100%	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACSF3	122.6	100%	97%	Combined malonic and methylmalonic aciduria, 614265
ACSL4	70.3	92%	81%	Mental retardation, X-linked 63, 300387
ACSL6	118.2	99%	97%	Myelodysplastic syndrome Myelogenous leukemia,acute
ACTA1	92.7	99%	92%	?Myopathy,scapulohumeroperoneal,616852 Myopathy, actin, congenital, with cores, 161800 Myopathy, actin, congenital, with excess of thin myofilaments, 161800 Myopathy, congenital, with fiber-type disproportion 1, 255310 Nemaline myopathy 3, autosomal dominant or recessive, 161800
ACTA2	150.7	100%	100%	Aortic aneurysm familial thoracic 6,611788 Moyamoya disease 5,614042 Multisystemic smooth muscle dysfunction syndrome,613834
ACTB	106.7	100%	93%	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACTC1	154.8	100%	100%	Atrial septal defect 5, 612794 Cardiomyopathy, dilated, 1R, 613424 Cardiomyopathy, familial hypertrophic, 11, 612098 Left ventricular noncompaction 4, 613424
ACTG1	110	100%	100%	Baraitser-Winter syndrome 2, 614583 Deafness, autosomal dominant 20/26, 604717
ACTN1	132.9	100%	99%	Bleeding disorder,platelet-type,15,615193
ACTN4	130	100%	100%	Glomerulosclerosis, focal segmental, 1, 603278
ACVR1	169	100%	100%	Fibrodysplasia ossificans progressiva, 135100
ACVR1B	170.5	95%	94%	Pancreatic cancer,somatic
ACVR2B	144.8	96%	90%	Heterotaxy, visceral, 4, autosomal, 613751

ACVRL1	117.6	100%	99%	Telangiectasia hereditary hemorrhagic type 2,600376
ACY1	127.7	100%	97%	Aminoacylase 1 deficiency, 609924
ADA	107.3	97%	96%	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADAM10	141.1	99%	97%	Reticulate acropigmentation of Kitamura,615537 {Alzheimer disease 18, susceptibility to},615590
ADAM17	125.2	98%	91%	?Inflammatory skin and bowel disease,neonatal,1,614328
ADAM9	149.9	98%	93%	Cone-rod dystrophy 9, 612775
ADAMTS10	97.1	99%	99%	Weill-Marchesani syndrome 1 recessive,277600
ADAMTS13	89.7	95%	89%	Thrombotic thrombocytopenic purpura, familial, 274150
ADAMTS17	112.2	88%	84%	Weill-Marchesani-like syndrome,613195
ADAMTS18	147.5	98%	97%	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
ADAMTS2	112.9	100%	96%	Ehlers-Danlos syndrome type VIIC,225410
ADAMTSL2	95.7	96%	84%	Geleophysic dysplasia 1,231050
ADAMTSL4	78.4	100%	97%	Ectopia lentis et pupillae,225200 Ectopia lentis,isolated,autosomal recessive,225100
ADAR	107.2	99%	98%	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
ADAT3	68.3	97%	84%	Mental retardation, autosomal recessive 36, 615286
ADCK3	114.9	99%	97%	Coenzyme Q10 deficiency, primary, 4, 612016
ADCK4	90.9	100%	100%	Nephrotic syndrome type 9, 615573
ADCY5	115.2	91%	88%	Dyskinesia, familial, with facial myokymia, 606703
ADIPOQ	109.6	100%	99%	Adiponectin deficiency,612556
ADK	95.5	99%	95%	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADRB2	115.7	100%	100%	Beta-2-adrenoreceptor agonist, reduced response to {Asthma, nocturnal, susceptibility to}, 600807 {Obesity, susceptibility to}, 601665
ADSL	175.6	100%	100%	ade(-)I bifunctional Adenylosuccinase deficiency, 103050
AFF2	87.3	100%	98%	Mental retardation, X-linked, FRAXE type, 309548
AFG3L2	108.2	90%	85%	Ataxia, spastic, 5, autosomal recessive, 614487 Spinocerebellar ataxia 28, 610246
AGA	134	100%	100%	Aspartylglucosaminuria, 208400
AGBL1	126.5	100%	100%	Corneal dystrophy, Fuchs endothelial, 8, 615523
AGK	115.1	99%	94%	Hyperoxaluria, primary, type 1, 259900

AGL	147.4	100%	99%	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGPAT2	101.5	100%	94%	Lipodystrophy, congenital generalized, type 1, 608594
AGPS	51.1	96%	81%	Lipodystrophy, congenital generalized, type 1, 608594
AGRN	99	93%	89%	Myasthenia, limb-girdle, familial, 254300
AGT	182	100%	100%	Renal tubular dysgenesis, 267430 {Hypertension, essential, susceptibility to}, 145500 {Preeclampsia, susceptibility to}
AGTR1	137.5	100%	100%	Hypertension, essential, 145500
AGXT	118.7	100%	100%	Hyperoxaluria, primary, type 1, 259900
AHCY	107.5	100%	99%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHI1	133	99%	93%	Joubert syndrome-3, 608629
AICDA	128.9	98%	96%	Immunodeficiency with hyper-IgM, type 2, 605258
AIFM1	76.8	100%	100%	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490
AIMP1	82.2	95%	86%	Leukodystrophy, hypomyelinating, 3, 260600
AIP	150.2	100%	97%	Pituitary adenoma,ACTH-secreting,219090 Pituitary adenoma, growth hormone-secreting,102200 Pituitary adenoma,prolactin-secreting,600634
AIPL1	111.6	100%	100%	Cone-rod dystrophy, 604393 Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393
AIRE	66.7	100%	92%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AK1	104	100%	99%	Hemolytic anemia due to adenylate kinase deficiency, 612631
AK2	94.4	91%	84%	Reticular dysgenesis, 267500
AKAP9	89.9	97%	92%	Long QT syndrome-11, 611820
AKR1C2	177.3	94%	90%	46XY sex reversal 8,614279 Obesity,hyperphagia and developmental delay
AKR1D1	104.3	96%	91%	Bile acid synthesis defect, congenital, 2, 235555

AKT1	146.7	99%	95%	Breast cancer somatic,114480 Colorectal cancer, somatic,114500 Cowden syndrome 6,615109 Ovarian cancer, somatic,167000 Proteus syndrome, somatic,176920 {Schizophrenia, susceptibility to},181500
AKT2	134.3	100%	98%	Diabetes mellitus,type II,125853 Hypoinsulinemic hypoglycemia with hemihypertrophy,240900
AKT3	73.2	99%	86%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome, 603387
ALAD	95.2	100%	98%	Porphyria, acute hepatic, 612740 {Lead poisoning, susceptibility to,} 612740
ALAS2	63.7	97%	91%	Anemia, sideroblastic, X-linked, 300751 Protoporphyrinemia, erythropoietic, X-linked, 300752
ALB	159.6	100%	99%	Analbuminemia,616000 [Dysalbuminemic hyperthyroxinemia],615999
ALDH18A1	125.3	100%	100%	Cutis laxa, autosomal recessive, type IIIA, 219150
ALDH1A3	103.2	91%	87%	Microphthalmia, isolated 8, 615113
ALDH2	105.9	99%	97%	Alcohol sensitivity, acute, 610251 {Esophageal cancer, alcohol-related, susceptibility to} {Hangover, susceptibility to, 610251} {Sublingual nitroglycerin, susceptibility to poor response to}
ALDH3A2	133.4	100%	100%	Sjogren-Larsson syndrome, 270200
ALDH4A1	107.5	100%	99%	Hyperprolinemia, type II, 239510
ALDH5A1	76.6	84%	78%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	126.8	100%	99%	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	67.2	95%	85%	Epilepsy, pyridoxine-dependent, 266100
ALDOA	151	100%	95%	Glycogen storage disease XII, 611881
ALDOB	152.7	100%	100%	Fructose intolerance, 229600
ALG1	49.2	50%	45%	ngenital disorder of glycosylation, type Ik, 608540
ALG11	154.1	100%	100%	Congenital disorder of glycosylation, type Ip, 613661
ALG12	145.3	100%	100%	Congenital disorder of glycosylation, type Ig, 607143
ALG13	57.3	97%	89%	Congenital disorder of glycosylation, type Is, 300884
ALG2	98.1	100%	98%	Congenital disorder of glycosylation, type Ii, 607906
ALG3	101.1	100%	99%	Congenital disorder of glycosylation, type Id, 601110

ALG6	91.8	95%	94%	Congenital disorder, type Ic, 603147
ALG8	127.7	96%	87%	Congenital disorder of glycosylation, type Ih, 608104
ALG9	118.5	98%	95%	Congenital disorder of glycosylation, type II, 608776
ALMS1	167.8	99%	99%	Alstrom syndrome, 203800
ALOX12B	124.4	99%	98%	Ichthyosis, congenital, autosomal recessive 2, 242100
ALOXE3	114.8	100%	99%	Ichthyosis congenital autosomal recessive 3,606545
ALPL	134.9	100%	100%	Hypophosphatasia, adult, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500 Odontohypophosphatasia, 146300
ALS2	166.3	100%	99%	Amyotrophic lateral sclerosis 2,juvenile,205100 Primary lateral sclerosis, juvenile, 606353 Spastic paraplegia, infantile onset ascending, 607225
ALX1	147.3	99%	95%	Frontonasal dysplasia 3, 613456
ALX3	92.5	73%	69%	Frontonasal dysplasia 1, 136760
ALX4	112.6	97%	85%	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597 {Craniosynostosis 5,susceptibility to},615529
AMACR	132.1	100%	99%	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
AMELX	47.2	100%	90%	Amelogenesis imperfecta, type 1E,301200
AMER1	55.7	99%	95%	Osteopathia striata with cranial sclerosis,300373
AMH	27.4	92%	59%	Persistent Mullerian duct syndrome, type I,261550
AMHR2	127.5	99%	97%	Persistent Mullerian duct syndrome, type II,261550
AMN	50.1	77%	62%	Megaloblastic anemia-1, Norwegian type, 261100
AMPD1	132.4	100%	99%	Myopathy due to myoadenylate deaminase deficiency,615511
AMT	144.4	100%	100%	Glycine encephalopathy, 605899
ANG	147.9	100%	100%	Amyotrophic lateral sclerosis 9, 611895
ANGPTL3	93.5	100%	99%	Hypobetalipoproteinemia,familial,2,605019
ANK1	126.7	100%	99%	Spherocytosis,type 1,182900
ANK2	141.4	99%	99%	Cardiac arrhythmia, ankyrin-B-related, 600919 Long QT syndrome-4, 600919
ANKH	104.1	100%	100%	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000

ANKK1	140.6	100%	98%	Dopamine receptor D2,reduced brain density of
ANKRD11	85.4	96%	91%	KBG syndrome, 148050
ANKRD26	80.6	91%	80%	Thrombocytopenia 2,188000
ANKS6	77.2	93%	88%	Nephronophthisis 16, 615382
ANO10	108.7	96%	95%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANO3	159.2	99%	98%	Dystonia 24, 615034
ANO5	142.2	97%	94%	Gnathodiaphyseal dysplasia, 166260 Miyoshi muscular dystrophy 3, 613319 Muscular dystrophy, limb-girdle, type 2L, 611307
ANO6	125.7	97%	94%	Scott syndrome,262890
ANTXR1	127.4	97%	96%	GAPO syndrome, 230740 {Hemangioma, capillary infantile, susceptibility to}, 602089
ANTXR2	84.7	99%	88%	Hyaline fibromatosis syndrome,228600
AP1S1	104.6	100%	99%	MEDNIK syndrome, 609313
AP1S2	53.2	66%	56%	Mental retardation, X-linked syndromic, Fried type, 300630
AP2S1	117.6	90%	87%	Hypocalciuric hypercalcemia, familial, type III, 600740
AP3B1	93.9	99%	92%	Hermansky-Pudlak syndrome 2, 608233
AP4B1	143	100%	100%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	103.9	99%	96%	Spastic paraplegia 51, autosomal recessive, 613744
AP4M1	93.2	100%	93%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	72.8	73%	68%	Spastic paraplegia 52, autosomal recessive, 614067
AP5Z1	78	100%	97%	Spastic paraplegia 48, autosomal recessive, 613647
APC	152.5	99%	97%	Adenoma, periampullary, somatic Adenomatous polyposis coli, 175100 Brain tumor-polyposis syndrome 2, 175100 Colorectal cancer, somatic, 114500 Desmoid disease, hereditary, 135290 Gardner syndrome,175100 Gastric cancer, somatic, 613659 Hepatoblastoma, somatic, 114550
APCDD1	162.2	100%	97%	Hypotrichosis 1,605389
APOA1	90	100%	100%	Amyloidosis,3 or more types,105200 ApoA-I and ApoC-III deficiency,combined Corneal clouding,autosomal recessive



				Hypoalphalipoproteinemia,604091
APOA2	91.3	81%	81%	Apolipoprotein A-II deficiency {Hypercholesterolemia,familial,modifier of},143890
APOA5	120.8	100%	100%	Hyperchylomicronemia,late-onset,144650 {Hypertriglyceridemia,susceptibility to},145750
APOB	181.6	99%	99%	Hypercholesterolemia,due to ligand-defective apo B,144010 Hypobetalipoproteinemia,615558
APOC2	87	100%	100%	Hyperlipoproteinemia, type Ib, 207750
APOC3	96.8	100%	100%	Apolipoprotein C-III deficiency,614028
APOE	42.5	93%	82%	Alzheimer disease-2,104310 Hyperlipoproteinemia,type III Lipoprotein glomerulopathy,611771 Sea-blue histiocyte disease,269600 {?Macular degeneration,age-related},603075 {Myocardial infarction susceptibility}
APP	149.5	100%	100%	Alzheimer disease 1,familial,104300 Cerebral amyloid angiopathy,Dutch,Italian,Iowa,Flemish,Arctic variants,605714
APRT	45.3	100%	99%	Adenine phosphoribosyltransferase deficiency, 614723
APTX	122.4	92%	86%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
AQP2	90.1	100%	89%	Diabetes insipidus, nephrogenic, 125800
AQP5	106.9	100%	97%	Palmoplantar keratoderma, Bothnian type,600231
AR	53.9	91%	83%	Androgen insensitivity,300068 Androgen insensitivity,partial,with/without breast cancer,312300 Hypospadias 1,X-linked,300633 Spinal and bulbar muscular atrophy of Kennedy,313200 {Prostate cancer,susceptibility to},176807
ARFGEF2	154.8	98%	97%	Periventricular heterotopia with microcephaly, 608097
ARG1	149.5	100%	100%	Argininemia, 207800
ARHGAP26	146.6	100%	100%	Leukemia,juvenile myelomonocytic,somatic,607785
ARHGAP31	103.4	99%	98%	Adams-Oliver syndrome 1,100300
ARHGEF10	127.2	99%	97%	?Slowed nerve conduction velocity,AD,608236

ARHGEF12	143.5	98%	97%	No OMIM phenotype Increased insulin sensitivity,association with (Kovacs (2006) Diabetes 55,1497)
ARHGEF6	99.8	96%	89%	Mental retardation, X-linked 46, 300436
ARHGEF9	64.2	100%	99%	Epileptic encephalopathy, early infantile, 8, 300607
ARID1A	135.8	91%	87%	Mental retardation, autosomal dominant 14, 614607
ARID1B	131.2	92%	84%	Mental retardation, autosomal dominant 12, 614562
ARL13B	78.8	99%	81%	Joubert syndrome 8, 612291
ARL2BP	72.8	78%	71%	Retinitis pigmentosa with or without situs inversus, 615434
ARL6	94.9	99%	96%	Bardet-Biedl syndrome 3, 209900 Retinitis pigmentosa 55, 613575 {Bardet-Biedl syndrome 1, modifier of}, 209900
ARMC4	123.8	90%	89%	Ciliary dyskinesia, primary, 23, 615451
ARNT	121	97%	95%	No OMIM phenotype Myocardial infarction,association with (Wang (2011) Ann Hum Genet 75,475)
ARSA	84.7	99%	97%	Metachromatic leukodystrophy, 250100
ARSB	116.2	91%	90%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARSE	58.9	96%	86%	Chondrodysplasia punctata, X-linked recessive, 302950
ARX	18.6	62%	41%	Epileptic encephalopathy, early infantile, 1, 308350 Hydranencephaly with abnormal genitalia,300215 Lissencephaly, X-linked 2, 300215 Mental retardation, X-linked 29 and others, 300419 Partington syndrome, 309510 Proud syndrome, 300004
ASAH1	118.4	96%	87%	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASB10	88.4	100%	97%	Glaucoma 1,open angle,F,603383
ASCC1	151	95%	92%	Barrett esophagus/esophageal adenocarcinoma,614266
ASCL1	161.1	87%	71%	Central hypoventilation syndrome,congenital,209880 Haddad syndrome,209880
ASL	88.5	100%	96%	Argininosuccinic aciduria, 207900
ASNS	80.6	96%	91%	Asparagine synthetase deficiency, 615574
ASPA	148.4	100%	96%	Canavan disease, 271900
ASPM	95.3	96%	91%	Microcephaly 5, primary, autosomal recessive, 608716
ASPSCR1	74.8	99%	93%	Alveolar soft-part sarcoma,606243

ASS1	86.8	95%	88%	Citrullinemia, 215700
ASXL1	145	99%	98%	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ASXL3	150.2	98%	97%	Bainbridge-Ropers syndrome, 615485
ATCAY	137.9	100%	100%	Ataxia, cerebellar, Cayman type, 601238
ATIC	114.3	100%	98%	AICA-ribosiduria due to ATIC deficiency, 608688
ATL1	165.2	97%	91%	Neuropathy,hereditary sensory,type 1D,613708 Spastic paraplegia 3A, autosomal dominant, 182600
ATL3	117.6	97%	92%	Neuropathy,hereditary sensory,type IF,615632
ATM	112	98%	95%	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic Lymphoma, mantle cell T-cell prolymphocytic leukemia, somatic {Breast cancer, susceptibility to}, 114480
ATN1	104.8	97%	96%	Dentatorubro-pallidoluysian atrophy,125370
ATP13A2	104	99%	95%	Parkinson disease 9, 606693
ATP1A2	176.3	100%	100%	Alternating hemiplegia of childhood, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481
ATP1A3	169.4	100%	100%	Alternating hemiplegia of childhood 2,614820 CAPOS syndrome,601338 Dystonia-12,128235
ATP2A1	150.9	100%	100%	Brody myopathy, 601003
ATP2A2	166.4	100%	99%	Acrokeratosis verruciformis, 101900 Darier disease, 124200
ATP2C1	120	99%	98%	Hailey-Hailey disease,169600
ATP5E	167.3	100%	100%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053
ATP6V0A2	141.9	100%	99%	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250
ATP6V0A4	112.4	100%	97%	Renal tubular acidosis, distal, autosomal recessive, 602722
ATP6V1B1	164.5	100%	100%	Renal tubular acidosis with deafness, 267300
ATP7A	88.3	99%	93%	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489

ATP7B	147.1	100%	99%	Wilson disease, 277900
ATP8B1	149.6	95%	93%	Cholestasis, benign recurrent intrahepatic, 243300 Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, progressive familial intrahepatic 1, 211600
ATPAF2	83.3	100%	98%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
ATR	137.4	97%	95%	?Cutaneous telangiectasia and cancer syndrome, familial, 614564 Seckel syndrome 1, 210600
ATRX	54.3	96%	88%	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Alpha-thalassemia/mental retardation syndrome, 301040 Mental retardation-hypotonic facies syndrome, X-linked, 309580
ATXN1	144.5	100%	100%	Spinocerebellar ataxia 1,164400
ATXN10	152.3	96%	95%	Spinocerebellar ataxia 10, 603516
ATXN2	94.9	89%	80%	Spinocerebellar ataxia 2,183090 {Amyotrophic lateral sclerosis,susceptibility to,13},183090 {Parkinson disease,late-onset,susceptibility to},168600
ATXN3	105.9	94%	85%	Machado-Joseph disease,109150
ATXN7	122.2	96%	93%	Spinocerebellar ataxia 7,164500
ATXN8OS				Spinocerebellar ataxia 8,608768
AUH	64.3	93%	89%	3-methylglutaconic aciduria, type I, 250950
AURKC	72.8	100%	98%	Spermatogenic failure 5,243060
AVP	46.1	70%	51%	Diabetes insipidus,neurohypophyseal,125700
AVPR2	77.2	91%	87%	Diabetes insipidus, nephrogenic, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539
AXIN1	128.3	98%	98%	?Caudal duplication anomaly,607864 Hepatocellular carcinoma,somatic,114550
AXIN2	111.4	100%	99%	Colorectal cancer somatic,114500 Oligodontia-colorectal cancer syndrome,608615
B2M	319.4	100%	100%	?Amyloidosis,familial visceral,105200 Immunodeficiency 43,241600
B3GALNT2	115.4	90%	88%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies,type A,11,615181
B3GALT6	47.2	70%	64%	Ehlers-Danlos syndrome progeroid type 2,615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1,with or without fractures,271640
B3GALTL	100.3	95%	94%	Peters-plus syndrome, 261540
B3GAT3	79.5	100%	98%	Multiple joint dislocations, short stature, craniofacial dysmorphism, and congenital heart defects, 245600

B3GNT1	101.9	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
B4GALNT1	139.8	99%	92%	Spastic paraplegia 26, autosomal recessive, 609195
B4GALT1	110.9	100%	100%	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	96.5	95%	94%	Ehlers-Danlos syndrome, progeroid type, 1, 130070
B9D1	101.1	92%	91%	Meckel syndrome 9, 614209
B9D2	110.8	100%	100%	Meckel syndrome 10, 614175
BAAT	121.8	98%	96%	Hypercholanemia, familial, 607748
BAG3	94.9	100%	99%	Myopathy, myofibrillar, 6, 612954 Cardiomyopathy, dilated, 1HH, 613881
BANF1	57.3	99%	91%	Nestor-Guillermo progeria syndrome,614008
BAP1	121.5	98%	97%	Tumor predisposition syndrome, 614327
BAX	83.7	84%	84%	Colorectal cancer,somatic,114500 T-cell acute lymphoblastic leukemia,somatic,613065
BBS1	133.8	100%	100%	Bardet-Biedl syndrome 1, 209900
BBS10	160.2	100%	99%	Bardet-Biedl syndrome 10, 209900
BBS12	196.3	100%	100%	Bardet-Biedl syndrome 12, 209900
BBS2	173	100%	98%	Bardet-Biedl syndrome 2, 209900
BBS4	136.1	99%	95%	Bardet-Biedl syndrome 4, 209900
BBS5	103.3	97%	91%	Bardet-Biedl syndrome 5, 209900
BBS7	121.3	96%	90%	Bardet-Biedl syndrome 7, 209900
BBS9	105.1	94%	93%	Bardet-Biedl syndrome 9, 209900
BCAP31	46.6	94%	68%	Deafness, dystonia and cerebellar hypomyelination, 300475
BCHE	154.5	100%	99%	Apnea,postanesthetic
BCKDHA	147.8	100%	100%	Maple syrup urine disease, type Ia, 248600
BCKDHB	120.7	85%	81%	Maple syrup urine disease, type Ib, 248600
BCKDK	151.4	100%	99%	Branched-chain ketoacid dehydrogenase kinase deficiency,614923
BCL10	91.7	100%	100%	?Immunodeficiency 37,616098 Lymphoma,MALT,somatic,137245 {Lymphoma,follicular,somatic},605027 {Male germ cell tumor,somatic},273300 {Mesothelioma,somatic},156240 {Sezary syndrome,somatic}
BCL2	122.8	100%	98%	Leukemia/lymphoma,B-cell,2
BCL7A	132.7	100%	93%	B-cell non-Hodgkin lymphoma,high-grade

BCMO1	159.2	100%	100%	Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300
BCOR	71	98%	95%	Microphthalmia, syndromic 2, 300166
BCR	100.1	87%	79%	Leukemia,acute lymphocytic,somatic,613065 Leukemia,chronic myeloid,somatic,608232
BCS1L	147	100%	100%	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BDNF	174.6	100%	100%	Central hypoventilation syndrome,congenital,209880 {Anorexia nervosa,susceptibility to},610269 {Bulimia nervosa,age of onset of weight loss in},607499 {Memory impairment,susceptibility to} {Obsessive-compulsive disorder,protection against},164230
BEAN1	117.9	94%	85%	Spinocerebellar ataxia 31,117210
BEST1	111.7	99%	96%	Bestrophinopathy, 611809 Macular dystrophy,vitelliform,2,153700 Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma, 193220 Retinitis pigmentosa,concentric,613194 Retinitis pigmentosa 50,613194 Vitreoretinchoroidopathy,193220
BFSP1	90.6	94%	89%	Cataract 33, 611391
BFSP2	80.3	98%	94%	Cataract 12, multiple types, 611597
BICD2	126.8	100%	99%	Spinal muscular atrophy, lower extremity-predominant, 2, AD, 615290 -3
BIN1	92.7	99%	93%	Myopathy, centronuclear, autosomal recessive, 255200
BLK	100.3	100%	100%	Maturity-onset diabetes of the young,type 11,613375
BLM	115.5	98%	92%	Bloom syndrome, 210900
BLNK	102.3	93%	91%	Agammaglobulinemia 4, 613502
BLOC1S3	38.5	99%	84%	Hermansky-Pudlak syndrome 8,614077
BLOC1S6	89.9	99%	84%	Hermansky-pudlak syndrome 9, 614171
BLVRA	119	100%	100%	Hyperbiliverdinemia, 614156
BMP1	131.7	100%	99%	Osteogenesis imperfecta,type XIII,614856
BMP15	76.8	100%	97%	Ovarian dysgenesis 2,300510 Premature ovarian failure 4,300510
BMP2	166.2	100%	100%	Brachydactyly, type A2, 112600

				{HFE hemochromatosis, modifier of}, 235200
BMP4	120.8	100%	98%	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625
BMPER	144.9	100%	100%	Diaphanospondylodysostosis,608022
BMPR1A	98.6	99%	89%	Juvenile polyposis syndrome, infantile form, 174900 Polyposis syndrome, hereditary mixed, 2, 610069 Polyposis, juvenile intestinal, 174900
BMPR1B	191.5	100%	99%	Acromesomelic dysplasia, Demirhan type, 609441 Brachydactyly, type A2, 112600
BMPR2	195.7	100%	100%	Pulmonary hypertension, familial primary, 1, with or without HHT, 178600 Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600 Pulmonary venoocclusive disease 1, 265450
BOLA3	55.4	91%	83%	Multiple mitochondrial dysfunctions syndrome 2, 614299
BPGM	101	100%	100%	Erythrocytosis due to bisphosphoglycerate mutase deficiency, 222800
BRAF	68.9	89%	83%	Adenocarcinoma of lung, somatic, 211980 Cardiofaciocutaneous syndrome, 115150 Colorectal cancer, somatic LEOPARD syndrome 3, 613707 Melanoma, malignant, somatic Non-small cell lung cancer, somatic Noonan syndrome 7, 613706
BRAT1	88.8	98%	92%	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BRCA2	96.1	98%	95%	Fanconi anemia, complementation group D1, 605724 Wilms tumor, 194070 {Breast cancer, male, susceptibility to}, 114480 {Breast-ovarian cancer, familial, 2}, 612555 {Glioblastoma 3}, 613029 {Medulloblastoma}, 155255 {Pancreatic cancer 2}, 613347 {Prostate cancer}, 176807
BRIP1	120.7	99%	97%	?Breast cancer, early-onset, 114480 Fanconi anemia, complementation group J, 609054
BRWD3	65.9	96%	90%	Mental retardation, X-linked 93, 300659

BSCL2	107.4	100%	98%	Encephalopathy,progressive,with or without lipodystrophy,615924 Lipodystrophy, congenital generalized, type 2, 269700 Neuropathy, distal hereditary motor, type V, 600794 Silver spastic paraplegia syndrome, 270685
BSND	131.6	100%	100%	Bartter syndrome, type 4a, 602522 Sensorineural deafness with mild renal dysfunction, 602522
BTD	122.5	100%	99%	Biotinidase deficiency, 253260
BTK	87.3	100%	98%	Agammaglobulinemia, X-linked 1, 300755
BUB1	134.3	99%	97%	Colorectal cancer with chromosomal instability,somatic
BUB1B	143.9	98%	97%	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430
C10orf11	135.8	99%	99%	Albinism, oculocutaneous type VII,615179
C10orf2	154	100%	100%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type),271245 Perrault syndrome 5,616138 Progressive external ophthalmoplegia with mitochondrial DNA depletions, autosomal dominant,609286
C12orf57	124.3	100%	100%	Temtamy syndrome, 218340
C12orf65	74.3	99%	97%	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55,autosomal recessive, 615035
C15orf41	124.3	100%	100%	Dyserythropoietic anemia, congenital, type Ib, 615631
C19orf12	78.4	100%	96%	?Spastic paraplegia 43, autosomal recessive, 615043 Neurodegeneration with brain iron accumulation 4, 614298
C1GALT1C1	77.1	100%	99%	Tn polyagglutination syndrome, somatic, 300622
C1QA	106.9	100%	88%	C1q deficiency, 613652
C1QB	157.3	100%	100%	C1q deficiency, 613652
C1QC	180.5	100%	98%	C1q deficiency, 613652
C1QTNF5	141.5	78%	60%	Retinal degeneration, late-onset, autosomal dominant, 605670
C1S	127.5	100%	99%	C1s deficiency, 613783
C2	15	67%	18%	C2 deficiency, 217000
C21orf59	152.5	100%	91%	Ciliary dyskinesia, primary, 26, 615500
C2orf71	116.9	100%	99%	Retinitis pigmentosa 54, 613428
C3	143.3	100%	99%	C3 deficiency, 613779
C4A	22.4	77%	49%	C4a deficiency, 614380
C4B	18.9	71%	40%	C4B deficiency, 614379



C4orf26	183.7	100%	100%	Amelogenesis imperfecta, type IIA4,614832
C5	132.3	97%	94%	C5 deficiency, 609536
C5orf42	119.9	95%	91%	Joubert syndrome 17, 614615
C6	159.9	100%	100%	C6 deficiency, 612446
C7	135.2	97%	93%	C7 deficiency, 610102
C8A	125.1	100%	99%	C8 deficiency, type I, 613790
C8B	136.9	100%	99%	C8 deficiency, type II, 613789
C8orf37	116.7	100%	99%	Cone-rod dystrophy 16, 614500 Retinitis pigmentosa 64, 614500
C9	137	100%	99%	C9 deficiency, 613825
C9orf72	114.8	100%	97%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1,105550
CA12	103.8	100%	100%	Hyperchlorhidrosis,isolated,143860
CA2	130.8	90%	85%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA4	133.6	100%	97%	Retinitis pigmentosa 17, 600852
CA5A	110.7	100%	97%	Hyperammonemia due to carbonic anhydrase VA deficiency,615751
CA8	98.2	87%	87%	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CABP2	73.1	97%	91%	Deafness, autosomal recessive 93, 614899
CABP4	90.2	96%	92%	Night blindness, congenital stationary (incomplete), 2B, autosomal recessive, 610427
CACNA1A	84.9	93%	87%	Episodic ataxia,type 2,108500 Migraine, familial hemiplegic, 1, 141500 Migraine, familial hemiplegic,1,with progressive cerebellar ataxia,141500 Spinocerebellar ataxia 6,183086
CACNA1C	145.4	99%	98%	Brugada syndrome 3, 611875 Timothy syndrome, 601005
CACNA1D	143.9	100%	99%	Sinoatrial node dysfunction and deafness, 614896
CACNA1F	62.7	98%	91%	Aland Island eye disease, 300600 Cone-rod dystrophy, X-linked, 3, 300476 Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071
CACNA1S	132.8	100%	99%	Hypokalemic periodic paralysis, type 1, 170400 {Malignant hyperthermia susceptibility 5}, 601887 {Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580
CACNA2D4	100.4	98%	97%	Retinal cone dystrophy 4, 610478
CACNB2	133.6	100%	97%	Brugada syndrome 4, 611876

CACNB4	110.2	95%	94%	Episodic ataxia, type 5, 613855 {Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682 {Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682
CACNG2	118.1	100%	100%	Mental retardation, autosomal dominant 10, 614256
CALM1	129.9	100%	100%	Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916
CALR	96.4	99%	95%	Myelofibrosis,somatic,254450 Thrombocythemia,somatic,187950
CALR3	138.3	100%	100%	Cardiomyopathy, familial hypertrophic, 19, 613875
CAMTA1	155.6	99%	97%	Cerebellar ataxia, nonprogressive, with mental retardation, 614756
CANT1	134.1	100%	100%	[Glutaric aciduria III], 231690
CAPN3	113.1	98%	93%	Muscular dystrophy, limb-girdle, type 2A, 253600
CAPN5	142.6	100%	99%	Vitreoretinopathy, neovascular inflammatory, 193235
CARD11	145.9	100%	96%	Persistent polyclonal B-cell lymphocytosis, 606445
CARD14	93.1	99%	96%	Pityriasis rubra pilaris,173200 Psoriasis 2,602723
CARD9	100.1	95%	95%	Candidiasis, familial, 2, autosomal recessive, 212050
CASC5	108.1	97%	94%	Microcephaly 4,primary,autosomal recessive,604321
CASK	63.1	99%	90%	FG syndrome 4, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 Mental retardation, with or without nystagmus, 300422
CASP10	107.4	99%	95%	Autoimmune lymphoproliferative syndrome, type II, 603909
CASP8	159.1	100%	100%	Immunodeficiency due to CASP8 deficiency, 607271
CASQ2	148.7	100%	99%	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CASR	147.9	99%	99%	Hyperparathyroidism,neonatal,239200 Hypocalcemia,autosomal dominant,601198 Hypocalciuric hypercalcemia,type I,145980 {Epilepsy idiopathic generalized,susceptibility to,8},612899
CAT	156.1	100%	100%	Desbuquois dysplasia, 251450
CATSPER1	166.2	100%	99%	Spermatogenic failure 7,612997
CAV1	238.9	100%	100%	?Lipodystrophy,congenital generalized,type 3,612526 ?Partial lipodystrophy, congenital cataracts and neurodegeneration syndrome,606721 Pulmonary hypertension, primary, 3,615343

CAV3	269.3	100%	100%	Cardiomyopathy,familial hypertrophic,192600 Creatine phosphokinase,elevated serum,123320 Long QT syndrome 9, 611818 Muscular dystrophy,limb-girdle,type IC,607801 Myopathy,distal,Tateyama type,614321 Rippling muscle disease,606072
CBL	118.2	98%	96%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563
CBS	106.9	99%	91%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CBX2	81.5	98%	94%	46XY sex reversal 5,613080
CC2D1A	103.6	100%	98%	Mental retardation, autosomal recessive 3, 608443
CC2D2A	121.8	99%	94%	COACH syndrome,216360 Joubert syndrome 9,612285 Meckel syndrome 6,612284
CCBE1	69.9	100%	95%	Hennekam lymphangiectasia-lymphedema syndrome, 235510
CCDC103	96.6	100%	98%	Ciliary dyskinesia, primary, 17, 614679
CCDC11	146.9	96%	92%	Heterotaxy,visceral,6,autosomal recessive,614779
CCDC114	105.7	100%	99%	Ciliary dyskinesia, primary, 20, 615067
CCDC39	81.5	95%	90%	Ciliary dyskinesia, primary, 14, 613807
CCDC40	106.5	99%	97%	Ciliary dyskinesia, primary, 15, 613808
CCDC50	128.4	100%	99%	Deafness, autosomal dominant 44, 607453
CCDC65	89.6	98%	87%	Ciliary dyskinesia, primary, 27, 615504
CCDC78	92.3	100%	98%	Myopathy, centronuclear, 4, 614807
CCDC8	75.4	100%	100%	3-M syndrome 3,614205
CCDC88C	98.7	99%	94%	?Spinocerebellar ataxia 40,616053 Hydrocephalus,nonsyndromic,autosomal recessive,236600
CCM2	143.1	97%	97%	Cerebral cavernous malformations-2,603284
CCT5	145.8	100%	97%	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CD151	122.1	100%	100%	Nephropathy with pretibial epidermolysis bullosa and deafness,609057 [Blood group, Raph],179620
CD19	76.9	98%	92%	Immunodeficiency, common variable, 3, 613493
CD247	92.9	100%	100%	Immunodeficiency due to defect in CD3-zeta, 610163
CD27	114	100%	99%	Lymphoproliferative syndrome 2, 615122
CD2AP	84.6	98%	91%	Glomerulosclerosis, focal segmental, 3, 607832

CD320	87.4	100%	97%	Methylmalonic aciduria due to transcobalamin receptor defect,613646
CD36	124.4	100%	99%	Platelet glycoprotein IV deficiency,608404 {Coronary heart disease,susceptibility to,7},610938 {Malaria,cerebral,reduced risk of},61162
CD3D	178.9	100%	100%	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971
CD3E	142.2	100%	100%	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971
CD3G	143.1	100%	100%	Immunodeficiency 17, CD3 gamma deficient, 615607
CD4	92.4	100%	100%	OKT4 epitope deficiency,613949
CD40	167	100%	99%	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	93.1	95%	80%	Immunodeficiency, X-linked, with hyper-IgM, 308230
CD59	207.5	90%	82%	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD79A	121.1	99%	93%	Agammaglobulinemia 3, 613501
CD79B	219.4	100%	100%	Agammaglobulinemia 6, 612692
CD81	127.2	99%	99%	Immunodeficiency, common variable, 6, 613496
CD8A	83.7	100%	97%	CD8 deficiency, familial, 608957
CD96	161.8	100%	100%	C syndrome,211750
CDAN1	87.3	97%	95%	Dyserythropoietic anemia, congenital, type Ia,224120
CDC6	147.9	99%	97%	Meier-Gorlin syndrome 5,613805
CDC73	96.6	100%	95%	Hyperparathyroidism, familial primary, 145000 Hyperparathyroidism-jaw tumor syndrome, 145001 Parathyroid adenoma with cystic changes, 145001 Parathyroid carcinoma, 608266
CDH1	107.7	99%	98%	Endometrial carcinoma, somatic, 608089 Gastric cancer, familial diffuse, with or without cleft lip and/or palate, 137215 Ovarian carcinoma, somatic, 167000 {Breast cancer, lobular}, 114480 {Prostate cancer, susceptibility to}, 176807
CDH15	111.1	100%	95%	Mental retardation, autosomal dominant 3, 612580
CDH23	183	100%	99%	Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D, 601067 Usher syndrome, type 1D/F digenic, 601067
CDH3	132.2	100%	97%	Ectodermal dysplasia,ectrodactyly and macular dystrophy,225280 Hypotrichosis, congenital, with juvenile macular dystrophy,601553
CDHR1	141.1	100%	99%	Cone-rod dystrophy 15, 613660

				Retinitis pigmentosa 65, 613660
CDK5RAP2	127.9	99%	96%	Microcephaly 3, primary, autosomal recessive, 604804
CDKL5	83	95%	91%	Epileptic encephalopathy, early infantile, 2, 300672
CDKN1B	63.1	100%	94%	Multiple endocrine neoplasia,type IV,610755
CDKN1C	26.9	59%	53%	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732
CDKN2A	56.4	93%	91%	Melanoma and neural system tumor syndrome, 155755 Orolaryngeal cancer, multiple Pancreatic cancer/melanoma syndrome, 606719 {Melanoma, cutaneous malignant, 2}, 155601
CDON	132.5	100%	98%	Holoprosencephaly 11, 614226
CDSN	16.5	59%	32%	Hypotrichosis 2,146520 Peeling skin syndrome 1,270300
CDT1	86	97%	90%	Meier-Gorlin syndrome 4,613804
CEACAM16	131.5	100%	100%	Deafness, autosomal dominant 4B, 614614
CEBPA	44.2	86%	59%	Leukemia,acute myeloid,601626
CEBPE	56.1	100%	94%	Specific granule deficiency, 245480
CECR1	95.6	100%	95%	?Sneddon syndrome,182410 Polyarteritis nodosa,childhood-onset,615688
CEL	110	84%	79%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CENPJ	144.5	99%	96%	Microcephaly 6, primary, autosomal recessive, 608393 Seckel syndrome 4, 613676
CEP135	73.6	94%	85%	Microcephaly 8, primary, autosomal recessive, 614673
CEP152	181.5	97%	94%	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823
CEP164	75.2	99%	94%	Nephronophthisis 15, 614845
CEP19	173.9	100%	100%	Morbid obesity and spermatogenic failure,615703
CEP290	69.5	89%	76%	?Bardet-Biedl syndrome 14,615991 Joubert syndrome 5,610188 Leber congenital amaurosis 10,611775 Meckel syndrome 4,611134 Senior-Loken syndrome 6,610189
CEP41	83.4	97%	91%	Joubert syndrome 15, 614464

CEP57	110.5	96%	89%	Mosaic variegated aneuploidy syndrome 2,614114
CERKL	108.2	99%	95%	Maturity-onset diabetes of the young, type VIII, 609812
CERS3	118.7	100%	98%	Ichthyosis, congenital, autosomal recessive 9, 615023
CES1	138	97%	93%	Carboxylesterase 1 deficiency
CETP	132.3	100%	100%	Hyperalphalipoproteinemia,143470 [High density lipoprotein cholesterol level QTL 10],143470
CFC1	47.8	77%	56%	Cardiofaciocutaneous syndrome,115150
CFD	64.1	95%	80%	Complement factor D deficiency, 613912
CFH	176.8	98%	95%	{Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400
CFHR5	107.9	97%	95%	Nephropathy due to CFHR5 deficiency, 614809
CFI	143.7	98%	97%	Complement factor I deficiency, 610984
CFL2	116.8	85%	84%	Nemaline myopathy 7, autosomal recessive, 610687
CFP	55.4	96%	88%	Properdin deficiency,X-linked, 312060
CFTR	129.9	97%	95%	Congenital bilateral absence of vas deference, 277180 Cystic fibrosis, 219700 Sweat chloride elevation without CF {Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400 {Hypertrypsinemia, neonatal} {Pancreatitis, idiopathic},
CHAT	123	87%	86%	Myasthenic syndrome, congenital, associated with episodic apnea, 254210
CHD2	124.9	99%	98%	Epileptic encephalopathy, childhood-onset, 615369
CHD7	136.1	99%	96%	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CHEK2	99	82%	80%	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	82.4	98%	90%	Muscular dystrophy, congenital, megaconial type, 602541
CHM	66.7	90%	77%	Choroideremia, 303100
CHMP1A	98.6	100%	100%	Pontocerebellar hypoplasia,type 8,614961
CHMP2B	87	99%	88%	Amyotrophic lateral sclerosis 17, 614696 Dementia, familial, nonspecific, 600795
CHMP4B	125.3	99%	96%	Cataract 31, multiple types, 605387

CHN1	143.6	97%	96%	Duane retraction syndrome 2,604356
CHRD1	85.8	98%	96%	Megalocornea 1,X-linked,309300
CHRM3	130.5	100%	100%	?Prune belly syndrome,100100
CHRNA1	114.8	100%	98%	Myasthenic syndrome, slow-channel congenital, 601462 Myasthenic syndrome, fast-channel congenital, 60893 Multiple pterygium syndrome, lethal type, 253290
CHRNA2	225.5	100%	100%	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA4	135.6	96%	96%	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction,susceptibility to},188890
CHRN1	130.2	98%	93%	Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931 Myasthenic syndrome, slow-channel congenital, 601462
CHRN2	249.4	100%	100%	Epilepsy, nocturnal frontal lobe, 3, 605375
CHRN	144.5	99%	96%	Multiple pterygium syndrome, lethal type, 253290 Myasthenic syndrome, slow-channel congenital, 601462 Myasthenic syndrome, fast-channel congenital, 608930
CHRE	123.5	100%	95%	Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931 Myasthenic syndrome, fast-channel congenital, 608930 Myasthenic syndrome, slow-channel congenital, 601462
CHRG	147	100%	100%	Escobar syndrome,26500 Multiple pterygium syndrome,lethal type,253290
CHST14	155.8	94%	92%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	75.8	97%	90%	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHST6	280	100%	100%	Macular corneal dystrophy, 217800
CHSY1	121.1	94%	93%	Temtamy preaxial brachydactyly syndrome, 605282
CHUK	135.4	99%	95%	Cocoon syndrome,613630
CIB2	188.6	100%	100%	Deafness, autosomal recessive 48, 609439 Usher syndrome, type II, 614869
CIITA	104.5	100%	100%	Bare lymphocyte syndrome type II, complementation group A, 209920 {Rheumatoid arthritis, susceptibility to}, 180300
CIRH1A	189	100%	99%	Cirrhosis,North American Indian childhood type,604901
CISD2	112.3	83%	83%	Wolfram syndrome 2,604928
CITED2	108.7	100%	100%	Ventricular septal defect 2, 614431 Atrial septal defect 8, 614433
CLCF1	84.2	97%	97%	Cold-induced sweating syndrome 2,610313

CLCN1	136.3	100%	99%	Myotonia congenita, dominant, 160800 Myotonia congenita, recessive, 255700 Myotonia levior, recessive
CLCN2	96.1	98%	98%	Leukoencephalopathy with ataxia,615651 {Epilepsy,idiopathic generalized,susceptibility to,11},607628
CLCN5	89.5	95%	89%	Dent disease, 300009 Hypophosphatemic rickets,300554 Nephrolithiasis,type I,310468 Proteinuria,low molecular weight,with hypercalciuric nephrocalcinosis,308990
CLCN7	124.4	100%	97%	Osteopetrosis,autosomal dominant 2,166600 Osteopetrosis,autosomal recessive 4,611490
CLCNKA	95.7	98%	90%	Bartter syndrome,type 4b,digenic,613090
CLCNKB	87.7	95%	89%	Bartter syndrome, type 3, 607364 Bartter syndrome,type 4b,digenic,613090
CLDN1	138.4	100%	100%	Ichthyosis,leukocyte vacuoles,alopecia and sclerosing cholangitis,607626
CLDN14	120.7	100%	99%	Deafness, autosomal recessive 29, 614035
CLDN16	140	100%	98%	Hypomagnesemia 3, renal, 248250
CLDN19	102.6	100%	94%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLEC7A	156.3	100%	100%	Candidiasis, familial, 4, autosomal recessive, 613108
CLIC2	50.1	90%	81%	Mental retardation, X-linked, syndromic 32, 300886
CLMP	126	100%	99%	Congenital short bowel syndrome,615237
CLN3	107.2	99%	97%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	144.8	97%	91%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	119.2	100%	92%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	206.6	100%	100%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLPP	106.7	98%	92%	Perrault syndrome 3, 614129
CLRN1	141.9	100%	100%	Retinitis pigmentosa 61, 614180 Usher syndrome type 3A, 276902
CNBP	118.3	100%	100%	Myotonic dystrophy 2,602668
CNGA1	128.2	84%	84%	Retinitis pigmentosa 49, 613756
CNGA3	149.8	99%	98%	Achromatopsia-2, 216900
CNGB1	91.5	95%	92%	Retinitis pigmentosa 45, 613767



CNGB3	104.7	91%	86%	Achromatopsia-3, 262300 Macular degeneration, juvenile, 248200
CNNM2	175.1	100%	98%	Hypomagnesemia 6, renal, 613882
CNNM4	174.3	98%	97%	Jalili syndrome,217080
CNTN1	158.4	100%	97%	Myopathy, congenital, Compton-North, 612540
CNTNAP2	130	100%	99%	Cortical dysplasia-focal epilepsy syndrome, 610042 Pitt-Hopkins like syndrome 1, 610042 {Autism susceptibility 15}, 612100
COA5	42.1	85%	85%	Mitochondrial complex IV deficiency, 220110
COASY	126.7	100%	100%	Neurodegeneration with brain iron accumulation 6, 615643
COCH	197.6	100%	99%	Deafness, autosomal dominant 9, 601369
COG1	114.1	100%	99%	Congenital disorder of glycosylation, type IIg, 611209
COG4	113.2	100%	100%	Congenital disorder of glycosylation, type 2j, 613189
COG5	104.4	97%	90%	Congenital disorder of glycosylation, type 2i, 613612
COG6	78.4	89%	79%	Congenital disorder of glycosylation, type 2l, 614576 Shaheen syndrome, 615328
COG7	120.1	100%	100%	Congenital disorder of glycosylation, type IIe, 608779
COG8	92.3	100%	90%	Congenital disorder of glycosylation, type IIh, 611182
COL10A1	85.1	100%	91%	Metaphyseal chondrodysplasia,Schmid type,156500
COL11A1	85.5	92%	83%	Fibrochondrogenesis, 228520 Marshall syndrome, 154780 Stickler syndrome, type II, 604841 {Lumbar disc herniation, susceptibility to}, 603932
COL11A2	11	46%	13%	Deafness, autosomal dominant 13, 601868 Deafness, autosomal recessive 53, 609706 Fibrochondrogenesis 2, 614524 Otospondylomegaepiphyseal dysplasia, 215150 Stickler syndrome, type III, 184840 Weissenbacher-Zweymuller syndrome, 277610
COL17A1	99.3	96%	92%	Epidermolysis bullosa,junctional,non-Herlitz type,226650
COL18A1	66.8	90%	83%	Knobloch syndrome,type 1,267750

COL1A1	116.9	97%	94%	Caffey disease,114000 Ehlers-Danlos syndrome,classis,130000 Ehlers-Danlos syndrome,type VIIA,130060 Osteogenesis imperfecta,type I,166200 Osteogenesis imperfecta,type II,166210 Osteogenesis imperfecta,type III,259420
COL1A2	96.1	96%	92%	Ehlers-Danlos syndrome,cardiac valvular form,225320 Ehlers-Danlos syndrome, type VIIB,130060 Osteogenesis imperfecta, type II,166210 Osteogenesis imperfecta, type III,259420 Osteogenesis imperfecta, type IV,166220 {Osteoporosis, postmenopausal},16671
COL2A1	93.5	99%	96%	Achondrogenesis, type II or hypochondrogenesis,200610 Avascular necrosis of the femoral head,608805 Czech dysplasia,609162 Epiphyseal dysplasia,multiple,myopia and deafness,132450 Kniest dysplasia,156550 Legg-Calve-Perthes disease,150600 Osteoarthritis with mild chondrodysplasia,604864 Otospondylomegaloeipiphyseal dysplasia, 215150 Platylospondylic skeletal dysplasia,Torrance type,151210 SED congenita,183900 SMED Strudwick type,184250 Spondyloepiphyseal dysplasia,Stanescu type,616583 Spondyloperipheral dysplasia,271700 Stickler syndrome,type 1,nonsyndromic ocular,609508 Stickler syndrome,type I,108300 Vitreoretinopathy with phalangeal epiphyseal dysplasia
COL3A1	100.5	89%	84%	Ehlers-Danlos syndrome, type IV,130050
COL4A1	83	96%	89%	?Retinal arteries,tortuosity of,180000 Angiopathy,hereditary,with nephropathy,aneurysms and muscle cramps,611773 Brain small vessel disease with or without ocular anomalies,607595 Porencephaly 1,175780 {Hemorrhage,intracerebral,susceptibility to},614519

COL4A2	83.6	98%	92%	Porencephaly 2, 614483 {Hemorrhage, intracerebral, susceptibility to}, 614519
COL4A3	81.2	97%	90%	Alport syndrome, autosomal dominant, 104200 Alport syndrome, autosomal recessive, 203780 Hematuria,benign familial, 141200
COL4A4	78.4	98%	93%	Alport syndrome, autosomal recessive, 203780
COL4A5	32.2	76%	50%	Alport syndrome, 301050
COL5A1	96.6	95%	92%	Ehlers-Danlos syndrome, classic type I,130000
COL5A2	78.4	99%	96%	Ehlers-Danlos syndrome, classic type I,130000
COL6A1	123.5	100%	98%	Bethlem myopathy, 158810 Ullrich congenital muscular dystrophy, 254090 {Ossification of the posterior longitudinal spinal ligaments}, 602475 (2)
COL6A2	132.6	100%	99%	Bethlem myopathy, 158810 Myosclerosis, congenital, 255600 Ullrich congenital muscular dystrophy, 254090
COL6A3	154.9	100%	99%	Bethlem myopathy, 158810 Ullrich congenital muscular dystrophy, 254090
COL7A1	111.7	99%	95%	EBD inversa,226600 EBD, Bart type,132000 Epidermolysis bullosa dystrophica, AD,131750 Epidermolysis bullosa dystrophica, AR,226600 Epidermolysis bullosa,pretibial,131850 Epidermolysis bullosa pruriginosa,604129 Toenail dystrophy,isolated,607523
COL8A2	21.8	76%	53%	Corneal dystrophy, Fuchs endothelial, 1, 136800 Corneal dystrophy, posterior polymorphous 2, 609140
COL9A1	107.6	99%	95%	Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	55.5	98%	86%	Epiphyseal dysplasia, multiple, 2, 600204 Stickler syndrome, type V, 614284 {Intervertebral disc disease, susceptibility to}, 603932
COL9A3	53.1	94%	81%	Epiphyseal dysplasia,multiple,3,600969 {Intervertebral disc disease,susceptibility to},603932
COLEC11	179.7	100%	100%	3MC syndrome 2, 265050
COLQ	98.1	99%	97%	Endplate acetylcholinesterase deficiency, 603034

COMP	117.6	94%	92%	Epiphyseal dysplasia,multiple,1,132400 Pseudoachondroplasia,177170
COQ2	71.1	97%	91%	Coenzyme Q10 deficiency, primary, 1, 607426
COQ6	127.8	98%	96%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ9	90.2	100%	97%	Coenzyme Q10 deficiency, primary, 5, 614654
CORIN	170.2	99%	98%	Preeclampsia/eclampsia 5,614595
CORO1A	144.8	99%	95%	Immunodeficiency 8, 615401
COX10	202	100%	98%	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 Mitochondrial complex IV deficiency, 220110
COX14	151.3	100%	100%	Mitochondrial complex IV deficiency, 220110
COX15	94.5	100%	99%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000
COX20	44.1	95%	79%	Mitochondrial complex IV deficiency, 220110
COX4I2	93.5	100%	100%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis,612714
COX7B	41.8	79%	54%	Linear skin defects with multiple congenital anomalies,300887
CP	127.7	94%	91%	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 [Hypoceruloplasminemia, hereditary], 604290
CPA6	110.9	100%	100%	Epilepsy, familial temporal lobe, 5, 614417 Febrile seizures,familial,11,614418
CPN1	107.1	97%	94%	Carboxypeptidase N deficiency,212070
CPOX	96.6	84%	79%	Coproporphyrria, 121300 Harderoporphyria, 121300
CPS1	148.8	100%	99%	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}
CPT1A	164.9	100%	97%	CPT deficiency, hepatic, type IA, 255120
CPT2	142.8	96%	93%	CPT deficiency, hepatic, type II, 600649 CPT II deficiency, lethal neonatal, 608836 Myopathy due to CPT II deficiency, 255110 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212
CR2	152.3	100%	98%	{Systemic lupus erythematosus, susceptibility to, 9}, 610927
CRADD	108.1	100%	93%	Mental retardation, autosomal recessive 34, 614499

CRB1	199.9	100%	99%	Leber congenital amaurosis 8, 613835 Pigmented paravenous chorioretinal atrophy, 172870 Retinitis pigmentosa-12, autosomal recessive, 600105
CRBN	135.5	100%	97%	Mental retardation, autosomal recessive 2, 607417
CREB1	136	100%	93%	Histiocytoma,angiomaoid fibrous,somatic,612160
CREBBP	119.9	99%	96%	Rubinstein-Taybi syndrome, 180849
CRELD1	94	98%	96%	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217 {Atrioventricular septal defect, susceptibility to, 2}, 606217
CRLF1	107.6	90%	87%	Cold-induced sweating syndrome 1,272430
CRTAP	108.2	100%	85%	Osteogenesis imperfecta,type VII,610682
CRTC1	119.6	99%	93%	Mucoepidermoid salivary gland carcinoma
CRX	91.4	100%	98%	Cone-rod retinal dystrophy-2, 120970 Leber congenital amaurosis 7, 613829
CRYAA	121.6	84%	82%	Cataract 9, multiple types, 604219
CRYAB	120.2	97%	95%	Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, 2, 608810 Myopathy, myofibrillar, fatal infantile hypertrophy, alpha-B crystallin-related, 613869
CRYBA1	130.8	100%	100%	Cataract 10, multiple types, 600881
CRYBA4	98.4	100%	97%	Cataract 23, 610425
CRYBB1	108.3	100%	99%	Cataract 17, multiple types, 611544
CRYBB2	152	100%	100%	Cataract 3, multiple types, 601547
CRYBB3	134.1	100%	98%	Cataract 22, autosomal recessive, 609741
CRYGB	95	100%	95%	Cataract 39, multiple types, autosomal dominant, 615188
CRYGC	114.1	100%	100%	Cataract 2, multiple types, 604307
CRYGD	89	100%	100%	Cataract 4, multiple types, 115700
CRYGS	130.9	95%	91%	Cataract 20, multiple types, 116100
CRYM	88	99%	92%	Deafness, autosomal dominant 40
CSF1R	122.5	98%	96%	Leukoencephalopathy,diffuse hereditary,with spheroids,221820
CSF2RA	49.6	89%	84%	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF2RB	81.2	99%	98%	Surfactant metabolism dysfunction,pulmonary,5,614370
CSF3R	82.6	97%	90%	Neutrophilia, hereditary, 162830
CSNK1D	163.6	94%	88%	Advanced sleep-phase syndrome,familial,2,615224
CSPP1	99.3	99%	92%	Joubert syndrome 21, 615636

CSRP3	101	100%	100%	Cardiomyopathy, dilated, 1M, 607482 Cardiomyopathy, familial hypertrophic, 12, 612124
CST3	106.8	78%	46%	Cerebral amyloid angiopathy,105150 Macular degeneration,age-related,11,611953
CSTA	99.9	100%	99%	Exfoliative ichthyosis,autosomal recessive,ichthyosis bullosa of Siemens-like,607936
CSTB	103.4	100%	100%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTC1	93.8	100%	99%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTCF	143.8	98%	95%	Mental retardation, autosomal dominant 21, 615502
CTDP1	93.2	90%	85%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTH	166	100%	98%	Cystathioninuria, 219500 Homocysteine, total plasma, elevated
CTHRC1	97.6	89%	77%	Barrett esophagus/esophageal adenocarcinoma,614266
CTNNA3	151	100%	100%	Arrhythmogenic right ventricular dysplasia, familial, 13, 615616
CTNNB1	163.2	100%	100%	Colorectal cancer, somatic, 114500 Hepatocellular carcinoma, somatic, 114550 Mental retardation, autosomal dominant 19, 615075 Ovarian cancer, somatic, 167000 Pilomatricoma, somatic, 132600
CTNS	117.3	100%	100%	Cystinosis, atypical nephropathic, 219800 Cystinosis, late-onset juvenile or adolescent nephropathic,219900 Cystinosis,ocular nonnephropathic,219750
CTSA	126.3	99%	96%	Galactosialidosis, 256540
CTSC	128.6	100%	100%	Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000 Periodontitis 1, juvenile, 170650
CTSD	152.8	98%	94%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	91.5	84%	80%	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362
CTSK	105.1	100%	100%	Pycnodysostosis, 265800
CUBN	124.5	99%	97%	Megaloblastic anemia-1, Finnish type, 261100
CUL3	117.2	100%	95%	Pseudohypoaldosteronism,type IIE,614496
CUL4B	47.4	93%	84%	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
CUL7	137.6	99%	98%	3-M syndrome 1,273750
CXCR4	195.6	100%	100%	WHIM syndrome, 193670
CYB5A	109.8	100%	100%	Methemoglobinemia, type IV,250790

CYB5R3	152.2	98%	98%	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYBA	82	78%	72%	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690
CYBB	82.2	100%	96%	Chronic granulomatous disease, X-linked, 306400
CYC1	164.2	97%	86%	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYCS	68.2	99%	98%	Thrombocytopenia 4, 612004
CYLD	116.2	98%	93%	Brooke-Spiegler syndrome, 605041 Cylindromatosis, familial, 132700 Trichoepithelioma, multiple familial, 1, 601606
CYP11A1	127.5	100%	97%	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	140	100%	98%	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 Aldosteronism, glucocorticoid-remediable, 103900
CYP11B2	153.5	100%	99%	Aldosterone to renin ratio raised Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 Low renin hypertension, susceptibility to
CYP17A1	132.3	100%	99%	17-alpha-hydroxylase/17,20-lyase deficiency, 202110 17,20-lyase deficiency, isolated, 202110
CYP19A1	175.4	100%	100%	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300
CYP1B1	106.3	100%	99%	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 Peters anomaly, 604229
CYP21A2	14.5	54%	23%	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910
CYP24A1	159	100%	97%	Hypercalcemia, infantile, 143880
CYP26B1	132.8	100%	99%	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
CYP26C1	72.2	100%	95%	Focal facial dermal dysplasia 4, 614974
CYP27A1	155	95%	94%	Cerebrotendinous xanthomatosis, 213700
CYP27B1	110.9	100%	98%	Vitamin D-dependent rickets, type I, 264700
CYP2A6	151.4	100%	99%	Coumarin resistance, 122700 {Lung cancer, resistance to}, 211980 {Nicotine addiction, protection from}, 188890
CYP2B6	92.7	99%	94%	Efavirenz, poor metabolism of, 614546 {Efavirenz central nervous system toxicity, susceptibility to}, 614546

CYP2C19	182.4	99%	96%	Clopidogrel,impaired responsiveness to,609535 Mephenytoin poor metabolizer,609535 Omeprazole poor metabolizer,609535 Proguanil poor metabolizer,609535
CYP2C8	112.2	97%	96%	Rhabdomyolysis,cerivastatin-induced
CYP2C9	188.1	100%	98%	Tolbutamide poor metabolizer Warfarin sensitivity,122700
CYP2R1	136.4	93%	87%	Rickets due to defect in vitamin D 25-hydroxylation, 600081
CYP2U1	115.2	93%	90%	Spastic paraplegia 56, autosomal recessive, 615030
CYP4F22	112.6	100%	99%	Ichthyosis,congenital,autosomal recessive 5,604777
CYP4V2	157	100%	98%	Bietti crystalline corneoretinal dystrophy, 210370
CYP7B1	94.3	95%	88%	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, autosomal recessive, 270800
D2HGDH	116.5	97%	93%	D-2-hydroxyglutaric aciduria, 600721
DAG1	180.9	100%	100%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818
DARS	90.5	99%	93%	Hypomyelination with brainstem and spinal cord involvement and leg spasticity,615281
DARS2	117.7	100%	100%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBH	126.2	100%	98%	Dopamine beta-hydroxylase deficiency, 223360 [Dopamine-beta-hydroxylase activity levels, plasma]
DBT	106.5	99%	87%	Maple syrup urine disease, type II, 248600
DCAF17	92.2	99%	92%	Woodhouse-Sakati syndrome, 241080
DCC	142.6	100%	99%	Colorectal cancer,somatic,114500 Esophageal carcinoma,somatic,133239 Mirror movements 1,157600
DCHS1	138.6	99%	98%	Mitral valve prolapse 2,607829 Van Maldergem syndrome 1,601390
DCLRE1C	120.5	95%	92%	Severe combined immunodeficiency, Athabaskan type, 602450
DCN	141.1	95%	94%	Corneal dystrophy, congenital stromal, 610048
DCTN1	119.2	100%	98%	Neuropathy, distal hereditary motor, type VIIB, 607641 Perry syndrome,168605 {Amyotrophic lateral sclerosis,susceptibility to},105400
DCX	76.6	100%	100%	Lissencephaly, X-linked, 300067 Subcortical laminar heteropia, X-linked, 300067
DDB2	148.5	100%	98%	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740



DDC	109.6	98%	92%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	145	96%	93%	Spastic paraplegia 28, autosomal recessive,609340
DDHD2	160.8	99%	94%	Spastic paraplegia 54, autosomal recessive, 615033
DDOST	105.5	99%	98%	Congenital disorder of glycosylation, type 1r, 614507
DDR2	152.8	100%	100%	Spondylometaepiphyseal dysplasia,short limb-hand type,271665
DDX11	83.4	79%	70%	Warsaw breakage syndrome, 613398
DDX59	157.1	100%	98%	Orofaciodigital syndrome V, 174300
DEPDC5	136.8	99%	99%	Epilepsy, familial focal, with variable foci, 604364
DES	108.8	100%	99%	?Muscular dystrophy, limb-girdle, type 2R, 615325 Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400
DFNA5	102.2	99%	98%	Deafness, autosomal dominant 5, 600994
DFNB31	102.8	100%	97%	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
DFNB59	116.4	100%	98%	Deafness, autosomal recessive 59, 610220
DGKE	137.4	97%	89%	Nephrotic syndrome, type 7, 615008
DGUOK	103.6	100%	97%	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHCR24	159.3	100%	100%	Desmosterolosis, 602398
DHCR7	149.9	100%	100%	Smith-Lemli-Opitz syndrome, 270400
DHDDS	84.7	95%	93%	Retinitis pigmentosa 59, 613861
DHFR	65.1	94%	89%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHH	88.3	100%	100%	46XY sex reversal 7,233420 46XY partial gonadal dysgenesis, with minifascicular neuropathy,607080
DHODH	94.9	100%	100%	Miller syndrome, 263750
DHTKD1	139.3	98%	97%	2-aminoadipic 2-oxoadipic aciduria, 204750 Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DIABLO	244.1	100%	100%	Deafness, autosomal dominant 64, 614152
DIAPH1	114.3	99%	97%	Deafness,autosomal dominant 1,124900 Seizures,cortical blindness,microcephaly syndrome,616632
DIAPH2	39.8	85%	68%	Premature ovarian failure,300511
DIAPH3	77.4	97%	88%	Auditory neuropathy, autosomal dominant, 1, 609129
DICER1	147.8	99%	97%	Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800 Pleuropulmonary blastoma, 601200

				Rhabdomyosarcoma,embryonal,2,180295
DIP2B	165.3	98%	96%	Mental retardation, FRA12A type, 136630
DIS3L2	150.7	99%	97%	Perlman syndrome,267000
DKC1	77.4	99%	92%	Dyskeratosis congenita, X-linked, 305000
DLAT	85.7	99%	96%	Pyruvate dehydrogenase E2 deficiency, 245348
DLC1	171.1	99%	98%	Colorectal cancer,somatic,114500
DLD	129.1	98%	98%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLG3	60.4	99%	86%	Mental retardation, X-linked 90, 300850
DLL3	62.5	84%	79%	Spondylocostal dysostosis 1,autosomal recessive,277300
DLX3	112.7	100%	96%	Amelogenesis imperfecta,type IV,104510 Trichodontoosseous syndrome,190320
DMD	83.9	99%	94%	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200
DMGDH	147.3	97%	96%	Dimethylglycine dehydrogenase deficiency, 605850
DMP1	131.1	100%	98%	Hypophosphatemic rickets,AR,241520
DMPK	98.4	100%	93%	Myotonic dystrophy 1, 160900
DNA2	128.6	99%	96%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 6, 615156
DNAAF1	102.1	100%	98%	Ciliary dyskinesia, primary, 13, 613193
DNAAF2	82.9	94%	90%	Ciliary dyskinesia, primary, 10, 612518
DNAAF3	82.3	97%	89%	Ciliary dyskinesia, primary, 2, 606763
DNAH11	130.3	99%	97%	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH5	123.9	99%	96%	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAI1	107.4	98%	96%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	138.5	95%	93%	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJB2	104.9	100%	100%	?Charcot-Marie-Tooth disease,axonal, type 2T,616233 Spinal muscular atrophy, distal, autosomal recessive,5, 614881
DNAJB6	55.5	89%	75%	Muscular dystrophy, limb-girdle, type 1E, 603511
DNAJC19	96.9	99%	88%	3-methylglutaconic aciduria, type V, 610198
DNAJC5	163.5	100%	99%	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350
DNAJC6	165.6	100%	99%	Parkinson disease 19,juvenile-onset,615528
DNAL1	103.6	93%	79%	Ciliary dyskinesia, primary, 16, 614017

DNASE1L3	146.1	100%	100%	Systemic lupus erythematosus 16,614420
DNM1L	109.2	100%	94%	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission, 614388
DNM2	116	96%	93%	Charcot-Marie-Tooth disease, axonal, type 2M, 606482 Charcot-Marie-Tooth disease, dominant intermediate B, 606482 Lethal congenital contracture syndrome 5, 615368 Myopathy, centronuclear, 160150
DNMT1	104.1	99%	98%	Neuropathy, hereditary sensory, type IE, 614116
DNMT3B	122.9	100%	99%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK6	108.9	99%	95%	Adams-Oliver syndrome 2,614219
DOCK8	129.4	100%	99%	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
DOK7	66	94%	90%	?Fetal akinesia deformation sequence, 208150 Myasthenic syndrome, congenital, 10, 254300
DOLK	170.5	100%	100%	Congenital disorder of glycosylation, type Im, 610768
DPAGT1	112.3	100%	100%	Congenital disorder of glycosylation, type lj, 608093 Myasthenic syndrome, congenital, with tubular aggregates 2, 614750
DPM1	112.1	89%	84%	Congenital disorder of glycosylation, type le, 608799
DPM2	98.3	100%	100%	Congenital disorder of glycosylation, type lu, 615042
DPM3	136	100%	100%	Congenital disorder of glycosylation, type lo, 612937
DPP6	132.1	98%	94%	Mental retardation, autosomal dominant 33, 616311 {Ventricular fibrillation, paroxysmal familial, 2}
DPY19L2	84.6	71%	57%	Spermatogenic failure,613958
DPYD	166.8	97%	93%	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270
DPYS	119.3	100%	98%	Dihydropyrimidinuria, 222748
DRC1	91.7	100%	98%	Ciliary dyskinesia, primary, 21, 615294
DRD2	195.6	100%	98%	No OMIM phenotype Myoclonus dystonia (Klein (1999) Proc Natl Acad Sci USA 96,5173 Schizophrenia, association with (Arinami (1994) Lancet 343,703)
DRD4	68.6	80%	76%	Autonomic nervous system dysfunction [Novelty seeking personality],601696 {Attention deficit-hyperactivity disorder},143465
DRD5	84.5	100%	100%	Dystonia,primary cervical {Attention deficit-hyperactivity disorder,susceptibility to},143465 {Blepharospasm,primary benign},606798

DSC2	145.9	97%	95%	Arrhythmogenic right ventricular dysplasia 11 without/with mild palmoplantar keratoderma and woolly hair,610476
DSC3	87.8	91%	81%	?Hypotrichosis and recurrent skin vesicles,613102
DSG1	169.4	98%	95%	Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis and hyper IgE,615508 Keratosis palmoplantaris striata I,AD,148700
DSG2	128.9	100%	99%	Arrhythmogenic right ventricular dysplasia 10, 610193 Cardiomyopathy, dilated, 1BB, 612877
DSG4	214.6	98%	96%	Hypotrichosis 6,607903
DSP	131.6	99%	97%	Arrhythmogenic right ventricular dysplasia 8,607450 Cardiomyopathy, dilated, with woolly hair and keratoderma,605676 Dilated cardiomyopathy with woolly hair, keratoderma and tooth agenesis,615821 Epidermolysis bullosa,lethal acantholytic,609638
DSPP	182.5	100%	98%	Deafness,autosomal dominant 36,with dentinogenesis,605594 Dentin dysplasia,type II,125420 Dentinogenesis imperfecta, Shields type II,125490 Dentinogenesis imperfecta, Shields type III, 125500
DST	159.5	99%	97%	?Neuropathy, hereditary sensory and autonomic, type VI, 614653 Epidermolysis bullosa simplex, sutosomal recessive 2, 615425
DTNA	160.9	100%	100%	Left ventricular noncompaction 1, with or without congenital heart defects, 604169
DTNBP1	100.5	100%	95%	Hermansky-Pudlak syndrome 7,614076 {Schizophrenia},181500
DUOX2	131.9	95%	94%	Thyroid dysmorphogenesis 6,607200
DUOXA2	106.9	100%	98%	Thyroid dysmorphogenesis 5,274900
DUSP6	144.6	100%	100%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
DYM	105	97%	95%	Dyggve-Melchior-Clausen disease, 223800 Smith-McCort dysplasia, 607326
DYNC1H1	166.1	99%	98%	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant, AD, 158600
DYNC2H1	91.6	95%	82%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYRK1A	153.5	100%	99%	Mental retardation, autosomal dominant 7, 614104
DYSF	124	99%	99%	Miyoshi muscular dystrophy 1, 254130 Muscular dystrophy, limb-girdle, type 2B, 253601 Myopathy, distal, with anterior tibial onset, 606768

DYX1C1	78	96%	84%	Ciliary dyskinesia, primary, 25, 615482 {Dyslexia, susceptibility to, 1}, 127700
EARS2	89.3	99%	94%	Combined oxidative phosphorylation deficiency 12, 614924
EBP	71	97%	89%	Chondrodysplasia punctata, X-linked dominant, 302960
ECE1	142.1	97%	97%	Hirschprung disease,cardiac defects, and autonomic dysfunction,613870 {Hypertension,essential,susceptibility to},145500
ECEL1	80.9	87%	78%	Arthrogryposis,distal,type 5D,615065
ECM1	152.7	100%	100%	Urbach-Wiethe disease,247100
EDA	47.4	80%	73%	Ectodermal dysplasia 1,hypohidrotic,X-linked,305100 Tooth agenesis,selective,X-linked 1,313500
EDAR	118.2	100%	99%	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
EDARADD	75.6	91%	91%	Ectodermal dysplasia 11A,hypohidrotic/hair/tooth type, autosomal dominant,614940 Ectodermal dysplasia 11B,hypohidrotic/hair/tooth type, autosomal recessive,614941
EDN1	138.6	100%	100%	auriculocondylar syndrome 3,615706 Question mark ears,isolated,612798 {High density lipoprotein cholesterol level QTL 7}
EDN3	106.9	100%	95%	Central hypoventilation syndrome congenital,209880 Waardenburg syndrome, type 4B,613265 {Hirshprung disease,susceptibility to,4},613712
EDNRA	206.4	100%	100%	mandibulofacial dysostosis with alopecia, 616367 {Migraine, resistance to},157300
EDNRB	124.7	95%	89%	?{Hirschsprung disease, susceptibility to}, 600155 ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580
EFEMP1	176.8	100%	98%	Doyne honeycomb degeneration of retina, 126600
EFEMP2	110	100%	100%	Cutis laxa,autosomal recessive,type 1B,614437
EFNB1	82.8	100%	98%	Craniofrontonasal dysplasia,304110
EFTUD2	114.2	99%	98%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EGF	138.4	100%	100%	Hypomagnesemia 4, renal, 611718
EGFR	143.2	97%	97%	?Inflammatory skin and bowel disease,neonatal,2,616069 Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980 Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980

				{Nonsmall cell lung cancer, susceptibility to}, 211980
EGLN1	42.9	81%	66%	Erythrocytosis,familial,3,609820 [Hemoglobin,high altitude adaptation],609070
EGR2	110	100%	100%	Charcot-Marie-Tooth disease,type 1D,607678 Dejerine-Sottas disease,145900 Neuropathy, congenital hypomyelinating, 1, 605253
EHMT1	130.5	99%	97%	Kleefstra syndrome, 610253
EIF2AK3	152.8	97%	89%	Wolcott-Rallison syndrome, 226980
EIF2AK4	147.1	99%	98%	Pulmonary venoocclusive disease 2,234810
EIF2B1	135.4	100%	100%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B2	127.5	100%	100%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B3	150.2	100%	100%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B4	140.8	99%	96%	Leukoencephaly with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B5	113.7	100%	99%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF4A3	106.7	100%	100%	Robin sequence with cleft mandible and limb abnormalities,268305
EIF4G1	119.8	100%	99%	Parkinsons disease 18, 614251
ELAC2	113.9	100%	97%	Combined oxidative phosphorylation deficiency 17, 615440 {Prostate cancer, hereditary, 2, susceptibility to}, 614731
ELANE	93.3	100%	93%	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
ELN	91.5	99%	96%	Cutis laxa AD,123700 Supravalvar aortic stenosis,185500
ELOVL4	77.5	100%	98%	?Spinocerebellar ataxia 34,133190 Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Stargardt disease 3, 600110
EMD	60	97%	84%	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
EMG1	122.6	100%	100%	Bowen-Conradi syndrome,211180
EMX2	105	100%	99%	Schizencephaly, 269160

ENAM	124.6	100%	100%	Amelogenesis imperfecta type IB,104500 Amelogenesis imperfecta type IC,204650
ENG	122.7	98%	95%	Telangiectasia,hereditary hemorrhagic,type 1,187300
ENO3	170.5	100%	100%	Glycogen storage disease XIII, 612932
ENPP1	134.4	88%	82%	Arterial calcification,generalized,of infancy,1,208000 Cole disease,615522 Hypophosphatemic rickets,autosomal recessive,2,613312 {Diabetes mellitus,non-insulin-dependent,susceptibility to},125853 {Obesity,susceptibility to},601665
ENTPD1	163.2	100%	98%	Spastic paraplegia 64,autosomal recessive,615683
EOGT	155.3	100%	97%	Adams-Oliver syndrome 4,615297
EP300	175.7	100%	99%	Colorectal cancer, somatic, 114500 Rubinstein-Taybi syndrome 2, 613684
EPAS1	127.4	98%	95%	Erythrocytosis,familial,4,611783
EPB41	122.7	99%	96%	Elliptocytosis-1,611804
EPB42	148.8	100%	98%	Spherocytosis,type 5,612690
EPCAM	64.6	93%	80%	Colorectal cancer,hereditary nonpolyposis, type 8,613244 Diarrhea 5,with tufting enteropathy,congenital,613217
EPG5	123.6	99%	94%	Vici syndrome, 242840
EPHA2	170.6	97%	97%	Cataract 6, multiple types, 116600
EPHB2	203.7	98%	98%	{Prostate cancer/brain cancer susceptibility,somatic},603688
EPHX1	116.9	98%	96%	?Fetal hydantoin syndrome Diphenylhydantoin toxicity Hypercholanemia, familial, 607748 Preeclampsia, susceptibility to, 189800
EPM2A	111.5	85%	82%	Epilepsy, progressive myoclonic 2A (Lafora), 254780
EPX	126.6	100%	100%	[Eosinophil peroxidase deficiency],261500
ERBB2	117.4	100%	97%	Adenocarcinoma of lung,somatic,211980 Gastric cancer,somatic,613659 Glioblastoma,somatic,137800 Ovarian cancer,somatic
ERBB3	119	99%	98%	Lethal congenital contractural syndrome 2, 607598
ERBB4	137.9	99%	96%	Amyotrophic lateral sclerosis 19,615515
ERCC1	74.9	100%	99%	Cerebrooculofacioskeletal syndrome 4, 610758

ERCC2	120	100%	99%	Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy, 601675 Xeroderma pigmentosum, group D, 278730
ERCC3	94.3	100%	99%	Trichothiodystrophy, 601675 Xeroderma pigmentosum, group B, 610651
ERCC4	144.3	100%	99%	?XFE progeroid syndrome, 610965 Fanconi anemia, complementation group Q, 615272 Xeroderma pigmentosum, group F, 278760 Xeroderma pigmentosum, type F/Cockayne syndrome, 278760
ERCC5	133.9	100%	96%	Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	165.9	100%	100%	Cerebrooculofacioskeletal syndrome 1, 214150 Cockayne syndrome, type B, 133540 De Sanctis-Cacchione syndrome, 278800 Premature ovarian failure 11,616946 UV-sensitive syndrome 1, 600630 {Lung cancer, susceptibility to}, 211980 {Macular degeneration, age-related, susceptibility to 5}, 613761
ERCC6L2	112.1	98%	97%	Bone marrow failure syndrome 2,615715
ERCC8	86.8	90%	83%	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621
ERF	93.8	100%	95%	Craniosynostosis 4, 600775
ERLIN2	151.4	100%	99%	Spastic paraplegia 18, autosomal recessive, 611225
ESCO2	100	91%	86%	Roberts syndrome, 268300 SC phocomelia syndrome, 269000
ESPN	39.8	70%	56%	Deafness, autosomal recessive 36, 609006 Deafness, neurosensory, without vestibular involvement, autosomal dominant
ESR1	109.7	99%	98%	Estrogen resistance,615363 {Breast cancer},114480 {Migraine,susceptibility to},157300 {Myocardial infarction,susceptibility to},608446
ESRRB	107.8	98%	94%	Deafness, autosomal recessive 35, 608565
ETFA	149.8	100%	100%	Glutaric acidemia IIA, 231680
ETFB	106.2	100%	100%	Glutaric acidemia 2B, 231680
ETFDH	102.8	100%	98%	Glutaric acidemia IIC, 231680



ETHE1	64.8	99%	88%	Ethylmalonic encephalopathy, 602473
ETV6	114.9	100%	100%	Leukemia,acute myeloid,somatic,601626 Thrombocytopenia 5,616216
EVC	95.9	94%	89%	Ellis-van Creveld syndrome,225500 Weyers acrodental dysostosis,193530
EVC2	105.5	95%	91%	Ellis-van Creveld syndrome,225500 Weyers acrodental dysostosis,193530
EWSR1	65.8	93%	79%	Ewing sarcoma,612219 Neuroepithelioma,612219
EXOSC3	62.6	97%	88%	Pontocerebellar hypoplasia, type 1B, 614678
EXPH5	196.7	100%	100%	Epidermolysis bullosa,nonspecific,autosomal recessive,615028
EXT1	94.5	100%	98%	Chondrosarcoma, 215300 Exostoses, multiple, type 1, 133700
EXT2	170.1	99%	97%	?Seizures,scoliosis and macrocephaly syndrome,616682 Exostoses, multiple, type 2, 133701
EYA1	139.1	100%	99%	Branchiootorenal syndrome 1, with or without cataracts, 113650
EYA4	153.1	99%	97%	Cardiomyopathy, dilated, 1J, 605362 Deafness, autosomal dominant 10, 601316
EYS	143.4	98%	94%	Retinitis pigmentosa 25, 602772
EZH2	131.8	99%	96%	Weaver syndrome, 277590
F10	153.9	98%	96%	Factor X deficiency,227600
F11	140.7	100%	97%	Factor XI deficiency,612416
F12	96.2	100%	98%	Factor XII deficiency, 234000
F13A1	164.5	100%	99%	Factor XIII deficiency,613225 {Myocardial infarction,protection against},608446 {Venous thrombosis,protection against},188050
F13B	141.4	95%	85%	Factor XIII B deficiency,613235
F2	106.3	100%	97%	Dysprothrombinemia,613679 Hypoprothrombinemia,613679 Thrombophilia due to thrombin defect,188050 {Pregnancy loss,recurrent,susceptibility to,2},614390 {Stroke,ischemic,susceptibility to},601367

F5	176.1	99%	97%	Factor V deficiency,227400 Thrombophilia due to activated prtein C resistance,188055 {Budd-Chiari syndrome},600880 {Pregnancy loss,recurrent,susceptibility to,1},614389 {Stroke,ischemic,susceptibility to},601367
F7	138.6	100%	96%	Factor VII deficiency,227500 {Myocardial infarction,decreased susceptibility to},608446
F8	79.5	98%	96%	Hemophilia A,306700
F9	102.4	95%	86%	Hemophilia B,306900 Thrombophilia,X-linked,due to factor IX defect},300807 {Warfarin sensitivity},122700
FA2H	87.6	93%	81%	Spastic paraplegia 35, autosomal recessive, 612319
FADD	105.6	99%	95%	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovasuclar malformations, 613759
FAH	143.4	100%	100%	Tyrosinemia, type I, 276700
FAM111A	272.7	100%	100%	Gracile bone dysplasia,602361 Kenny-Caffey syndrome,type 2,127000
FAM111B	159.6	100%	100%	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy and pulmonary fibrosis, 615704
FAM126A	119.4	99%	94%	Leukodystrophy, hypomyelinating, 5, 610532
FAM134B	104.6	93%	81%	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
FAM161A	110.4	99%	93%	Retinitis pigmentosa 28, 606068
FAM20A	101.9	95%	91%	Amelogenesis imperfecta,type IG (enamel-renal syndrome),204690
FAM20C	79.6	95%	85%	Raine syndrome,259775
FAM58A	46.7	79%	74%	STAR syndrome, 300707
FAM83H	60.9	92%	86%	Amelogenesis imperfecta type 3,130900
FAN1	162.2	100%	100%	Interstitial nephritis,karyomegalic,614817
FANCA	107	98%	95%	Fanconi anemia, complementation group A, 227650
FANCB	49.3	89%	75%	Fanconi anemia, complementation group B, 300514
FANCC	108.3	100%	94%	Fanconi anemia, complementation group C, 227645
FANCD2	125.5	98%	96%	Fanconi anemia, complementation group D2, 227646
FANCE	92.2	84%	84%	Fanconi anemia, complementation group E, 600901
FANCF	120.8	100%	100%	Fanconi anemia, complementation group F, 603467
FANCG	116.2	100%	98%	Fanconi anemia, complementation group G, 614082
FANCI	158.1	98%	96%	Fanconi anemia, complementation group I, 609053

FANCL	73.4	100%	92%	Fanconi anemia, complementation group L, 614083
FANCM	98.4	98%	94%	Fanconi anemia, complementation group M, 614087
FARS2	190	100%	99%	Combined oxidative phosphorylation deficiency 14, 614946
FAS	254	100%	100%	{Autoimmune lymphoproliferative syndrome}, 601859
FASLG	83.8	100%	98%	Autoimmune lymphoproliferative syndrome, type IB, 601859 {Lung cancer, susceptibility to}, 211980
FAT4	220.5	100%	100%	Hennekam lymphangiectasia-lymphedema syndrome 2,616006 Van Maldergem syndrome 2,615546
FBLN1	145.8	99%	96%	Synpolydactyly,3/3'4,associated with metacarpal and metatarsal synostoses,608180
FBLN5	100.8	91%	89%	Cutis laxa,autosomal dominant 2,614434 Cutis laxa,autosomal recessive,type IA,219100 Macular degeneration,age-related,3,608895
FBN1	161.3	99%	98%	Acromicric dysplasia, 102370 Aortic aneurysm, ascending, and dissection Ectopia lentis, familial, 129600 Geleophysic dysplasia 2,614185 Marfan lipodystrophy syndrome,616914 Marfan syndrome, 154700 MASS syndrome, 604308 Stiff skin syndrome, 184900 Weill-Marchesani syndrome 2, dominant, 608328
FBN2	154.3	99%	99%	Contractural arachnodactyly, congenital, 121050
FBP1	99.6	100%	98%	Fructose-1,6-bidphosphatase deficiency, 229700
FBXL4	204	100%	100%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FBXO38	159.7	100%	99%	Neuronopathy,distal hereditary motor,type IID,615575
FBXO7	209.5	100%	96%	Parkinson disease 15, autosomal recessive, 260300
FCGR3A	209.1	100%	100%	Immunodeficiency 20, 615707
FCGR3B	125.4	99%	99%	Neutropenia,alloimmune neonatal
FCN3	125.3	100%	98%	Immunodeficiency due to ficolin 3 deficiency, 613860
FECH	122.4	100%	100%	Protoporphyrin, erythropoietic, autosomal recessive, 177000
FERMT1	97.3	98%	94%	Kindler syndrome,173650
FERMT3	103.7	100%	97%	Leukocyte adhesion deficiency, type III, 612840

FGA	149.6	97%	95%	Afibrinogenemia,congenital,202400 Amyloidosis,familial visceral,105200 Dysfibrinogenemia,congenital,616004 Hypodysfibrinogenemia,congenital,616004
FGB	168	100%	98%	Afibrinogenemia,congenital,202400 Dysfibrinogenemia,congenital,616004 Hypofibrinogenemia,congenital,202400
FGD1	53.8	89%	77%	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400
FGD4	108.6	96%	92%	Charcot-Marie-Tooth disease, type 4H, 609311
FGF10	140.5	100%	100%	Aplasia of lacrimal and salivary glands,180920 LADD syndrome,149730
FGF14	197	100%	98%	Spinocerebellar ataxia 27, 609307
FGF16	86	98%	94%	Metacarpal 4-5 fusion,309630
FGF17	125.4	100%	100%	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
FGF23	115.8	100%	97%	Hypophosphatemic rickets, autosomal dominant, 193100 Osteomalacia,tumor-induced Tumoral calcinosis,hyperphosphatemic,familial,211900
FGF3	49.5	88%	75%	Deafness,congenital with inner ear agenesis,microtia and microdontia,610706
FGF8	90.6	81%	74%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGF9	185.2	100%	100%	?Multiple synostoses syndrome 3,612961
FGFR1	135.5	100%	95%	Encephalocraniocutaneous lipomatosis,613001 Hartsfield syndrome, 615465 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Jackson-Weiss syndrome, 123150 Osteoglophonic dysplasia, 166250 Pfeiffer syndrome, 101600 Trigonocephaly 1, 190440

FGFR2	133.9	96%	95%	<p>Antley-Bixler syndrome wo genital anomalies or disordered steroidogenesis,207410</p> <p>Apert syndrome, 101200</p> <p>Beare-Stevenson cutis gyrata syndrome,123790</p> <p>Bent bone dysplasia syndrome,614592</p> <p>Craniofacial-skeletal-dermatologic dysplasia,101600</p> <p>Craniosynostosis,nonspecific</p> <p>Crouzon syndrome,123500</p> <p>Gastric cancer,somatic,613659</p> <p>Jackson-Weiss syndrome,123150</p> <p>LADD syndrome,149730</p> <p>Pfeiffer syndrome,101600</p> <p>Saethre-Chotzen syndrome,101400</p> <p>Scaphocephaly and Axenfeld-Rieger anomaly</p> <p>Scaphocephaly,maxillary retrusion,mental retardation,609579</p>
FGFR3	91.3	100%	98%	<p>Achondrodysplasia,100800</p> <p>Bladder cancer,somatic,109800</p> <p>CATSHL syndrome,610474</p> <p>Cervical cancer,somatic,603956</p> <p>Colorectal cancer,somatic,114500</p> <p>Crouzon syndrome with acanthosis nigricans,612247</p> <p>Hypochondroplasia,146000</p> <p>LADD syndrome,149730</p> <p>Muenke syndrome,602849</p> <p>Nevus,epidermal,somatic,162900</p> <p>SADDAN,616482</p> <p>Spermatocytic seminoma, somatic,273300</p> <p>Thanatophoric dysplasia,type I,187600</p> <p>Thanatophoric dysplasia,type II,187601</p>
FGG	137.8	100%	98%	<p>Afibrinogenemia,congenital,202400</p> <p>Dysfibrinogenemia,congenital,616004</p>
FH	160.3	90%	87%	<p>Fumarase deficiency, 606812</p> <p>Leiomyomatosis and renal cell cancer, 150800</p>

FHL1	58.5	96%	75%	Emery-Dreifuss muscular dystrophy 6,X-linked,300696 Myopathy,X-linked,with postural muscle atrophy,300696 Reducing body myopathy,X-linked 1a,severe,infantile or early childhood onset,300717 Reducing body myopathy,X-linked 1b,with late childhood or adult onset,300718 Scapuloperoneal myopathy,X-linked dominant,300695
FIG4	155.7	100%	99%	?Polymicrogyria,bilateral temporooccipital,612691 Amyotrophic lateral sclerosis 11,612577 Charcot-Marie-Tooth disease, type 4J, 611228 Yunis-Varon syndrome,216340
FIGLA	84.3	88%	84%	Premature ovarian failure,612310
FKBP10	141.6	97%	88%	Bruck syndrome 1,259450 Osteogenesis imperfecta type XI,610968
FKBP14	65.4	100%	100%	Ehlers-Danlos syndrome with progressive kyphoscoliosis myopathy and hearing loss,614557
FKRP	70.4	100%	96%	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
FKTN	137.2	97%	89%	Cardiomyopathy,dilated,1X,611615 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies),type A,4,253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation),type B,4,613152 Muscular dystrophy-dystroglycanopathy (limb-girdle),type C,4,611588
FLCN	142.8	100%	98%	Birt-Hogg-Dube syndrome, 135150 Colorectal cancer, somatic, 114500 Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700
FLG	212.1	100%	99%	Ichthyosis vulgaris,146700 {Dermatitis,atopic,susceptibility to,2},605803

FLNA	85.1	99%	97%	Cardiac valvular dysplasia,X-linked,314400 Congenital short bowel syndrome,300048 FG syndrome 2,300321 Frontometaphyseal dysplasia,305620 Heterotopia,periventricular,300049 Heterotopia,periventricular,ED variant,300537 Intestinal pseudoobstruction,neuronal,300048 Melnick-Needles syndrome,309350 Otopalatodigital syndrome,type I,311300 Otopalatodigital syndrome,type II,304120 Terminal osseous dysplasia,300244
FLNB	139.4	100%	99%	Atelosteogenesis,type I,108720 Atelosteogenesis,type III,108721 Boomerang dysplasia,112310 Larsen syndrome,150250 Spondylocarpotarsal synostosis syndrome,272460
FLNC	149.8	100%	99%	Myopathy, distal, 4, 614065 Myopathy, myofibrillar, 5, 609524
FLRT3	234.3	100%	100%	Hypogonadotropic hypogonadism 21 with or without anosmia, 615271
FLT3	142.6	98%	96%	Leukemia,acute lymphoblastic,somatic,613065 Leukemia,acute myeloid,reduced survival in,somatic,601626
FLT4	148	99%	95%	Hemangioma,capillary infantile,somatic,602089 Lymphedema,hereditary,IA,153100
FLVCR1	121.4	99%	97%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FLVCR2	169.4	100%	100%	Proliferative vasculopathy and hydraencephaly-hydrocephaly syndrome,225790
FMO3	158.1	100%	99%	Trimethylaminuria, 602079
FMR1	51.7	86%	75%	Fragile X syndrome, 300624 Fragile X tremor/ataxia syndrome, 300623 Premature ovarian failure 1, 311360
FN1	136.1	99%	98%	Glomerulopathy with fibronectin deposits 2, 601894
FOLR1	139.5	100%	100%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXC1	31.1	95%	72%	Axenfeld-Rieger syndrome, type 3, 602482 Iridogoniodysgenesis, type 1, 601631 Iris hypoplasia and glaucoma, 601631 Rieger or Axenfeld anomalies, 602482

FOXC2	42.2	92%	74%	Lymphedema-distichiasis syndrome with/without renal disease and diabetes mellitus,153400
FOXE1	27.1	71%	57%	Bamforth-Lazarus syndrome,241850
FOXE3	9.3	49%	33%	Anterior segment mesenchymal dysgenesis, 107250 Aphakia, congenital primary, 610256
FOXF1	65.5	94%	86%	Alveolar capillary dysplasia with misalignment of pulmonary veins,265380
FOXG1	88.7	85%	81%	Rett syndrome, congenital variant, 613454
FOXI1	131.3	100%	100%	Enlarged vestibular aqueduct, 600791
FOXL2	22.4	84%	54%	Blepharophimosis,epicanthus inversus and ptosis,type 1 and 2,110100 Premature ovarian failure 3,608996
FOXN1	90.7	100%	99%	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXO1	135.7	92%	89%	Rhabdomyosarcoma,alveolar,268220
FOXP1	123.6	100%	99%	Mental retardation with language impairment and autistic features, 613670
FOXP2	156.4	98%	94%	Speech-language disorder-1, 602081
FOXP3	82.4	83%	74%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790
FOXRED1	131.8	100%	98%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency,252010
FRAS1	142.2	100%	99%	Fraser syndrome, 219000
FREM1	134.9	99%	98%	Bifid nose with or without anorectal and renal anomalies, 608980
FREM2	164.1	99%	98%	Fraser syndrome, 219000
FRMD7	79.4	99%	93%	Nystagmus 1,congenital,X-linked,310700 Nystagmus,infantile periodic alternating X-linked,310700
FSCN2	124.5	100%	100%	Retinitis pigmentosa 30,607921
FSHB	146.2	100%	100%	Hypogonadotropic hypogonadism 24 without anosmia,229070
FSHR	144.7	100%	98%	Ovarian dysgenesis 1,233300 Ovarian hyperstimulation syndrome,608115 Ovarian response to FSH stimulation,276400
FTCD	79.6	88%	80%	Glutamate formiminotransferase deficiency, 229100
FTL	104.5	99%	82%	Hyperferritinemia-cataract syndrome, 600886 Neurodegeneration with brain iron accumulation 3, 606159
FTO	154.4	99%	98%	Growth retardation, developmental delay, coarse facies, and early death, 612938
FTSJ1	78.7	96%	92%	Mental retardation, X-linked 9, 309549
FUCA1	120.8	99%	98%	Fucosidosis, 230000
FUS	134.1	98%	96%	Amyotrophic lateral sclerosis 6, autosomal recessive, with or without frontotemporal dementia, 608030 Tremor, hereditary essential, 4, 614782



FUT6	151.4	100%	100%	Fucosyltransferase 6 deficiency, 613852
FUZ	87.5	99%	97%	Neural tube defects, 182940
FXN	82	75%	75%	Friedreich ataxia, 229300 Friedreich ataxia with retained reflexes, 229300
FXD2	81.7	100%	100%	Hypomagnesemia-2, renal, 154020
FYCO1	114.3	100%	99%	Cataract 18, autosomal recessive, 610019
FZD4	207.1	100%	98%	Exudative vitreoretinopathy, 133780 Retinopathy of prematurity, 133780
FZD6	181	100%	100%	Nail disorder,nonsyndromic,congenital 10 (claw-shaped nails),614157
G6PC	191.7	100%	100%	Glycogen storage disease Ia, 232200
G6PC3	107.7	100%	100%	Dursun syndrome,612541 Neutropenia,severe congenital 4,autosomal recessive,612541
G6PD	69.8	98%	96%	Favism, 134700 Hemolytic anemia due to G6PD deficiency,300908 Resistance to malaria due to G6PD deficiency, 611162
GAA	90.9	100%	99%	Glycogen storage disease II, 232300
GABRA1	176.6	100%	100%	Epileptic encephalopathy, early infantile, 19, 615744 {Epilepsy, childhood absence, susceptibility to, 4} {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136
GABRB3	143.2	95%	95%	{Epilepsy,childhood absence, susceptibility to, 5},612269 Epileptic encephalopathy (Epi4K consortium, Nature. 2013 Sep 12;501(7466):217-21)
GABRG2	155.9	92%	92%	Epilepsy, generalized, with febrile seizures plus, type 3, 611277 Febrile seizures,familial,8,611277 {Epilepsy,childhood absence,susceptibility to,2},607681
GAD1	116.9	99%	93%	Cerebral palsy, spastic quadriplegic, 1, 603513
GALC	94.8	95%	87%	Krabbe disease, 245200
GALE	141.6	100%	100%	Galactose epimerase deficiency, 230350
GALK1	88.7	96%	91%	Galactokinase deficiency with cataracts, 230200
GALNS	85.3	99%	93%	Mucopolysaccharidosis IVA, 253000
GALNT3	125.7	97%	87%	Tumoral calcinosis, hyperphosphatemic, familial,211900
GALT	141.5	100%	99%	Galactosemia, 230400
GAMT	94.4	98%	91%	Cerebral creatine deficiency syndrome 2, 612736
GAN	179.2	99%	98%	Giant axonal neuropathy-1, 256850

GARS	127.7	100%	99%	Charcot-Marie-Tooth disease, type 2D, 601472 Neuropathy,distal hereditary motor,type VA,600794
GATA1	49.6	97%	90%	Anemia,X-linked,with/without neutropenia and/or platelet abnormalities,300835 Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 Thrombocytopenia with beta-thalassemia, X-linked, 314050 Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367
GATA2	117.2	100%	98%	Dendritic cell, monocyte, B lymphocyte, and natural killer lymphocyte deficiency, 614172
GATA3	150	100%	100%	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GATA4	72.6	63%	58%	?Testicular anomalies with or without congenital heart disease,615542 Atrial septal defect 2,607941 Atrioventricular septal defect 4,614430 Tetralogy of Fallot,187500 Ventricular septal defect 1,614429
GATA6	49.7	83%	66%	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	139.4	99%	92%	Cardiomyopathy, dilated, 2B, 614672
GATAD2B	122.4	100%	99%	Mental retardation, autosomal dominant 18, 615074
GATM	152.2	100%	100%	Cerebral creatine deficiency syndrome 3, 612718
GBA	210.6	100%	100%	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, 16860
GBA2	152.3	100%	98%	Spastic paraplegia 46, autosomal recessive
GBE1	148.6	100%	93%	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GCDH	121.1	92%	90%	Glutaricaciduria, type I, 231670
GCH1	68.7	87%	79%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910

GCK	125.7	100%	100%	Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, permanent neonatal, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485 MODY, type II, 125851
GCLC	131.6	100%	98%	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 Myocardial infarction, susceptibility to, 608446
GCM2	143	100%	100%	Hypoparathyroidism,familial isolated,146200
GCNT2	164.9	100%	100%	Adult i phenotype without cataract, 110800 Cataract 13 with adult i phenotype, 110800 [Blood group, Ii], 110800
GCSH	38.9	93%	61%	Glycine encephalopathy, 605899
GDAP1	170.7	100%	99%	Charcot-Marie-Tooth disease,axonal,type 2K,607831 Charcot-Marie-Tooth disease,axonal,with vocal cord paresis,607706 Charcot-Marie-Tooth disease,recessive intermediate,A,608340 Charcot-Marie-Tooth disease, type 4A, 214400
GDF1	15.6	80%	41%	Double-outlet right ventricle, 217095 Right atrial isomerism, 208530 Tetralogy of Fallot, 187500 Transposition of great arteries, dextro-looped 3, 613854
GDF2	139.3	100%	99%	Telangiectasia, hereditary hemorrhagic, type 5,615506
GDF3	119.6	100%	100%	Klippel-Feil syndrome 3, autosomal dominant, 613702 Microphthalmia with coloboma 6, 613703 Microphthalmia, isolated 7, 613704
GDF5	106.2	100%	100%	Acromesomelic dysplasia,Hunter-Thompson type,201250 Brachydactyly,type A1,C,615072 Brachydactyly,type A2,112600 Brachydactyly,type C,113100 Chondrodysplasia,Grebe type,200700 Du Pan syndrome,228900 Multiple synostoses syndrome 2,610017
GDF6	56.8	93%	82%	Klippel-Feil syndrome 1, autosomal dominant, 118100 Leber congenital amaurosis 17, 615360 Microphthalmia, isolated 4, 613094 Microphthalmia with coloboma 6, digenic, 613703
GDI1	103.7	100%	100%	Mental retardation, X-linked 41, 300849

GDNF	138.3	98%	94%	Central hypoventilation syndrome, 209880 {Pheochromocytoma, modifier of}, 171300 {Hirschsprung disease, susceptibility to, 3}, 613711
GFAP	87.2	100%	97%	Alexander disease, 203450
GFER	69.7	98%	71%	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076
GFI1	88.8	97%	91%	Neutropenia, nonimmune chronic idiopathic, of adults, 607847 Neutropenia, severe congenital 2, autosomal dominant, 613107
GFI1B	141.8	100%	100%	Bleeding disorder, platelet-type, 17,187900
GFM1	100.4	99%	95%	Combined oxidative phosphorylation deficiency 1, 609060
GFPT1	145.8	100%	96%	Myasthenia, congenital, with tubular aggregates 1, 610542
GGCX	104.8	99%	96%	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency,610842 Vitamin K-dependent clotting factors,combined deficiency of,1,277450
GH1	180.2	100%	100%	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	208.7	100%	100%	Growth hormone insensitivity,partial,604271 Laron dwarfism,262500 {Hypercholesterolemia,familial,modifier of},143890
GHRHR	114.6	100%	100%	Growth hormone deficiency,isolated,type IB,612781
GHSR	183.1	98%	97%	Growth hormone deficiency,isolated partial,615925
GIF	160.6	100%	100%	Intrinsic factor deficiency,261000
GIGYF2	113.6	96%	94%	{Parkinson disease 11},607688
GIPC3	89.3	88%	84%	Deafness, autosomal recessive 15, 601869
GJA1	205.3	100%	100%	Atrioventricular septal defect 3,600309 Craniometaphyseal dysplasia, autosomal recessive,218400 Erythrokeratoderma variabilis et progressiva,133200 Hypoplastic left heart syndrome 1,241550 Oculodentodigital dysplasia,164200 Oculodentodigital dysplasia,autosomal recessive,257850 Palmoplantar keratoderma with congenital alopecia,104100 Syndactyly,type III,186100
GJA3	152.3	100%	100%	Cataract 14, multiple types, 601885

GJA5	225.4	100%	100%	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic, 108770
GJA8	136.5	100%	100%	Cataract 1, multiple types, 116200
GJB1	114.6	100%	100%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GJB2	182.1	100%	100%	Bart-Pumphrey syndrome,149200 Deafness,autosomal recessive 1A,220290 Hystrix-like ichthyosis with deafness,602540 Keratitis,ichthyosis-deafness syndrome,148210 Keratoderma,palmoplantar,with deafness,148350 Vohwinkel syndrome,124500
GJB3	300.8	100%	100%	Deafness autosomal dominant 2B,612644 Deafness,digenic,GJB2/GJB3,220290 Erythrokeratoderma variabilis et progressiva,133200
GJB4	363.8	100%	100%	Erythrokeratoderma variabilis with erythema gyratum repens,133200
GJB6	191.1	100%	100%	Deafness,autosomal dominant 3B,612643 Deafness,autosomal recessive 1B,612645 Deafness,digenic GJB2/GJB6,220290 Ectodermal dysplasia 2,Clouston type,129500
GJC2	34.3	81%	62%	Leukodystrophy, hypomyelinating, 2, 608804 Lymphedema, hereditary, IC, 613480 Spastic paraplegia 44, autosomal recessive, 613206
GK	29.1	66%	50%	Glycerol kinase deficiency, 307030
GLA	47.8	99%	90%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	73.9	98%	90%	GM1-gangliosidosis, type I, 230500
GLDC	77.2	87%	79%	Glycine encephalopathy, 605899
GLE1	112.2	100%	96%	Arthrogryposis,lethal,with anterior horn cell disease,611890 Lethal congenital contracture syndrome 1,253310
GLI2	110.3	97%	93%	Holoprosencephaly-9, 610829
GLI3	138.5	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	75.5	99%	90%	Nephronophthisis 7, 611498

GLIS3	127.9	100%	96%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GLMN	81.4	93%	79%	Glomuvenous malformations,138000
GLRA1	120.6	100%	100%	Hyperekplexia, hereditary 1, autosomal dominant or recessive,149400
GLRB	87.7	97%	90%	Hyperekplexia 2, autosomal recessive, 614619
GLRX5	99.2	90%	84%	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive, 205950
GLUD1	71.2	88%	81%	Hyperinsulinism-hyperammonemia syndrome, 606762
GLUL	80.3	96%	92%	Glutamine deficiency, congenital, 610015
GLYCTK	213.4	99%	97%	D-glyceric aciduria, 220120
GM2A	128.7	100%	100%	GM2-gangliosidosis, AB variant, 272750
GMPPA	124.4	100%	100%	Alacrima, achalasia and mental retardation syndrome, 615510
GMPPB	206.3	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A,14, 6135350 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (limb-girdle),type C,14,615352
GMPS	102.2	96%	90%	Leukemia, acute myelogenous, 601626
GNA11	134.5	100%	100%	Hypocalcemia,autosomal dominant 2,615361 Hypocalciuric hypercalcemia, type II, 145981
GNAI2	144.5	100%	100%	Pituitary ACTH-secreting adenoma Ventricular tachycardia,idiopathic,192605
GNAI3	124.4	100%	98%	Auriculocondylar syndrome 1,602483
GNAL	132	95%	92%	Dystonia 25, 615073
GNAO1	173.4	100%	100%	Epileptic encephalopathy, early infantile, 17, 615473
GNAQ	88.5	82%	58%	Capillary malformations,congenital,1, somatic,mosaic,163000 Sturge-Weber syndrome, somatic, mosaic,185300
GNAS	110.5	95%	94%	Acromegaly,somatic,102200 ACTH-independent macronodular adrenal hyperplasia,219080 McCune-Albright syndrome,somatic,mosaic 174800 Osseous heteroplasia, progressive, 166350 Pseudohypoparathyroidism Ia, 103580 Pseudohypoparathyroidism Ib, 603233 Pseudohypoparathyroidism Ic, 612462 Pseudopseudohypoparathyroidism,612463
GNAT1	153.1	100%	100%	Night blindness, congenital stationary, autosomal dominant 3, 610444
GNAT2	142.9	100%	98%	Achromatopsia-4, 613856
GNB4	140.3	100%	98%	Charcot-Marie-Tooth disease, dominant intermediate F, 615185

GNE	150.9	100%	100%	Nonaka myopathy, 605820 Sialuria, 269921
GNMT	141.1	98%	96%	Glycine N-methyltransferase deficiency, 606664
GNPAT	138.4	99%	96%	Chondrodysplasia punctata, rhizomelic, type 2, 222765
GNPTAB	172.1	97%	97%	Mucopolysaccharidosis III alpha/beta, 252600 Mucopolysaccharidosis II alpha/beta, 252500
GNPTG	125.2	90%	87%	Mucopolysaccharidosis III gamma, 252605
GNRH1	78	99%	88%	Hypogonadotropic hypogonadism 12 with or without anosmia, 614841
GNRHR	177.5	100%	100%	Hypogonadotropic hypogonadism 7 with or without anosmia, 138850
GNS	104.7	92%	88%	Mucopolysaccharidosis type IIID, 252940
GOLGA5	139.5	100%	100%	No OMIM phenotype Colorectal cancer,increased risk,association with (Webb (2006) Hum Mol Genet 15,3263)
GORAB	166.6	100%	99%	Geroderma osteodysplasticum,231070
GOSR2	118.6	96%	93%	Epilepsy, progressive myoclonic 6
GOT1	113.4	100%	100%	Aspartate aminotransferase, serum level of, QTL1, 614419
GP1BA	122.6	99%	95%	Bernard-Soulier syndrome, type A1 (recessive),231200 Bernard-Soulier syndrome, type A2 (dominant),153670 von Willebrand disease,platelet-type,177820 {Nonarteric anterior ischemic optic neuropathy,susceptibility to},258660
GP1BB	37	86%	65%	Bernard-Soulier syndrome,type B,231200 Giant platelet disorder,isolated,231200
GP6	101	100%	99%	Bleeding disorder,platelet-type,11,614201
GP9	66.9	99%	95%	Bernard-Soulier syndrome,type C,231200
GPC3	72	93%	85%	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPC6	133.6	100%	100%	Omodysplasia 1,258315
GPD1	94.4	96%	90%	Hypertriglyceridemia, transient infantile, 614480
GPD1L	150.3	100%	98%	Brugada syndrome 2, 611777
GPHN	172.9	96%	96%	Molybdenum cofactor deficiency, type C, 252150
GPI	123	100%	100%	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470
GPR143	39.9	90%	67%	Nystagmus 6,congenital,X-linked,300814 Ocular albinism, type I, Nettleship-Falls type,300500
GPR179	119.3	100%	97%	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
GPR56	140.4	100%	100%	Polymicrogyria, bilateral frontoparietal, 606854

GPR98	145.8	99%	94%	Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472
GPSM2	109.2	99%	94%	Chudley-McCullough syndrome, 604213
GRHL2	127.4	100%	100%	Deafness,autosomal dominant 28,608641 Ectodermal dysplasia/short stature syndrome,616029
GRHL3	127.8	100%	100%	Van der Woude syndrome 2, 606713
GRHPR	104.1	82%	78%	Hyperoxaluria, primary, type II, 260000
GRIA3	65.5	96%	85%	Mental retardation, X-linked 94, 300699
GRIK2	130.6	95%	89%	Mental retardation, autosomal recessive, 6, 611092
GRIN1	132.6	100%	98%	Mental retardation, autosomal dominant 8, 614254
GRIN2A	157.9	100%	100%	Epilepsy with neurodevelopmental defects, 613971
GRIN2B	184.8	99%	98%	Mental retardation, autosomal dominant 6, 613970
GRIP1	128.9	100%	99%	Fraser syndrome,219000
GRK1	100.1	100%	100%	Oguchi disease-2, 613411
GRM1	169.3	100%	99%	Spinocerebellar ataxia, autosomal recessive 13, 614831
GRM6	139.7	93%	83%	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270
GRN	171.5	100%	100%	Aphasia,primary progressive,607485 Ceroid lipofuscinosis,neuronal,11,614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
GRXCR1	189.1	100%	100%	Deafness, autosomal recessive 25, 613285
GSC	82.5	79%	72%	Short stature,auditory canal atresia,mandibular hypoplasia,skeletal abnormalities,602471
GSN	109	91%	87%	Amyloidosis, Finnish type, 105120 Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930
GSS	92.9	100%	100%	Hemolytic anemia due to glutathione synthetase deficiency, 231900
GTF2H5	160.4	100%	99%	Trichothiodystrophy, complementation group A, 601675
GUCA1A	147.4	100%	100%	Cone dystrophy-3, 602093 Cone-rod dystrophy 14, 602093
GUCA1B	130.1	100%	98%	Retinitis pigmentosa 48, 613827
GUCY1A3	157.3	99%	97%	Moyamoya 6 with achalasia,615750
GUCY2C	137.3	100%	99%	Diarrhea 6,614616 Meconium ileus,614665
GUCY2D	79.9	96%	84%	Cone-rod dystrophy 6, 601777 Leber congenital amaurosis 1, 204000



GUSB	105.3	89%	85%	Mucopolysaccharidosis VII, 253220
GYG1	128.6	100%	97%	Glycogen storage disease XV, 613507
GYS1	104	100%	97%	Glycogen storage disease 0, muscle, 611556
GYS2	165.9	100%	95%	Glycogen storage disease, type 0, 240600
H19				Beckwith-Wiedemann syndrome,130650 Silver-Russell syndrome,180860 Wilms tumor 2,194071
H6PD	153.5	99%	99%	Cortisone reductase deficiency 1, 604931
HADH	109.6	97%	94%	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HADHA	70.5	93%	86%	Fatty liver, acute, of pregnancy, 609016 HELLP syndrome, maternal, of pregnancy, 609016 LCHAD deficiency, 609016 Trifunctional protein deficiency, 609015
HADHB	87	92%	84%	Trifunctional protein deficiency, 609015
HAMP	162	100%	100%	Hemochromatosis, type 2B, 613313
HARS	143.6	100%	100%	Usher syndrome type 3B, 614504
HARS2	166.6	100%	100%	Perrault syndrome 2, 614926
HAX1	123.9	100%	99%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HBA1	67.7	100%	100%	Erythremias,alpha- Heinz body anemias,alpha-,140700 Hemoglobin H disease,nondeletional,613978 Methemoglobinemias,alpha- Thalassemias,alpha-,604131
HBA2	65.1	94%	84%	Erythrocytosis Heinz body anemia,140700 Hemoglobin H disease,nondeletional,613978 Hypochromic microcytic anemia Thalassemia,alpha-,604131

HBB	133.5	100%	100%	Delta-beta thalassemia,141749 Erythremias,beta- Heinz body anemias,beta-,140700 Hereditary persistence of fetal hemoglobin,141749 Thalassemia-beta,dominant inclusion body,603902 Sickle cell anemia,603903 Thalassemsias,beta-,613985
HBD	201.9	100%	100%	Thalassemia due to Hb Lepore Thalassemia,delta-
HBG1	123.4	98%	93%	Fetal hemoglobin quantitative trait locus 1,141749
HBG2	235.1	100%	100%	Cyanosis,transient neonatal,613977 Fetal hemoglobin quantitative trait locus 1,141749
HCCS	63.8	99%	92%	Microphthalmia, syndromic 7, 309801
HCFC1	64.6	97%	91%	Mental retardation, X-linked 3, 309541
HCN4	80.9	98%	90%	Sick sinus syndrome 2, 163800 Brugada syndrome 8, 613123
HCRT	31.4	79%	57%	?Narcolepsy 1,161400
HDAC4	94.5	100%	99%	Brachydactyly-mental retardation syndrome, 600430
HDAC6	65.9	96%	87%	Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863
HDAC8	90.5	100%	100%	Cornelia de Lange syndrome 5, 300882 Wilson-Turner syndrome, 309585
HEATR2	107.9	84%	77%	Ciliary dyskinesia, primary, 18, 614874
HEPACAM	124.5	76%	75%	Megalencephalic leukoencephalopathy with subcortical cysts 2A,613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B,remitting,with or without mental retardation,613926
HERC2	98.8	77%	72%	Mental retardation, autosomal recessive 38, 615516 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220
HES7	32.5	72%	65%	Spondylocostal dysostosis 4,autosomal recessive,613686
HESX1	65.2	99%	85%	Growth hormone deficiency with pituitary anomalies, 182230 Pituitary hormone deficiency, combined, 5, 182230 Septo-optic dysplasia, 182230

HEXA	125.4	100%	99%	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800 [Hex A pseudodeficiency], 272800
HEXB	134.4	97%	91%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HFE	126.5	100%	97%	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
HFE2	106.1	100%	100%	Hemochromatosis type 2A
HFM1	45	89%	79%	Premature ovarian failure 9,615724
HGD	121	100%	94%	Alkaptonuria, 203500
HGF	139.5	98%	97%	Deafness, autosomal recessive 39, 608265
HGSNAT	95	81%	81%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930
HIBCH	75.9	93%	70%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HINT1	49.4	96%	81%	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200
HK1	134.8	100%	99%	Hemolytic anemia due to hexokinase deficiency, 235700
HLCS	160.7	100%	100%	Holocarboxylase synthetase deficiency, 253270
HMBS	90	100%	96%	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000
HMGCL	125.7	100%	100%	HMG-CoA lyase deficiency, 246450
HMGCS2	127.1	100%	100%	HMG-CoA synthase-2 deficiency, 605911
HMOX1	123.9	98%	88%	Heme oxygenase-1 deficiency, 614034 Pulmonary disease, chronic obstructive, susceptibility to, 606963
HMX1	19.6	63%	39%	Oculoauricular syndrome, 612109
HNF1A	124.6	99%	97%	Diabetes mellitus, insulin-dependent, 20, 612520 Hepatic adenoma, somatic, 142330 MODY, type III, 600496 Renal cell carcinoma, 144700 {Diabetes mellitus, noninsulin-dependent, 2}, 125853 {Diabetes mellitus, insulin-dependent}, 222100
HNF1B	103	100%	97%	Diabetes mellitus,noninsulin-dependent,125853 Renal cysts and diabetes syndrome, 137920

				{Renal cell carcinoma},144700
HNF4A	134	100%	98%	Fanconi renotubular syndrome 4,with maturity-onset diabetes of the young,616026 MODY,type I,125850 {Diabetes mellitus,noninsulin-dependent},125853
HNRNPA1	81.4	86%	78%	?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3,615424 Amyotrophic lateral sclerosis 20,615426
HOGA1	123.1	100%	94%	Hyperoxaluria, primary, type III, 613616
HOXA1	125.4	100%	100%	Athabaskan brainstem dysgenesis syndrome, 601536 Bosley-Salih-Alorainy syndrome, 601536
HOXA11	85.6	82%	79%	Radioulnar synostosis with amegakaryocytic thrombocytopenia,605432
HOXA13	38	65%	60%	Guttmacher syndrome,176305 Hand-foot-uterus syndrome,140000
HOXB1	94.8	100%	100%	Facial paresis,hereditary congenital 3,614744
HOXC13	87.2	92%	87%	Ectodermal dysplasia 9 hair/nail type,614931
HOXD10	101.5	100%	99%	Charcot-Marie-Tooth disease,foot deformity of,192950 Vertical talus,congenital,192950
HOXD13	95.9	93%	88%	?Brachydactyly-syndactyly syndrome,610713 Brachydactyly type D,113200 Brachydactyly,type E,113300 Syndactyly,type V,186300 Synpolydactyly with foot anomalies,286000
HPD	131.5	100%	100%	Hawkinsinuria, 140350 Tyrosinemia, type III, 276710
HPGD	94.2	97%	89%	Cranioosteoarthropathy,259100 Digital clubbing,isolated congenital,119900 Hypertrophic osteoarthropathy,primary,autosomal recessive 1,259100
HPRT1	45.8	95%	84%	HPRT-related gout,300323 Lesch-Nyhan syndrome, 300322
HPS1	102.9	100%	98%	Hermansky-Pudlak syndrome 1,203300
HPS3	138	100%	95%	Hermansky-Pudlak syndrome 3,614072
HPS4	130.4	100%	98%	Hermansky-Pudlak syndrome 4,614073
HPS5	137.7	99%	98%	Hermansky-Pudlak syndrome 5,614074
HPS6	100.4	100%	92%	Hermansky-Pudlak syndrome 6,614075

HPSE2	108.3	97%	92%	Urofacial syndrome 1,236730
HR	76.4	96%	91%	Alopecia universalis,203655 Atrichia with papular lesions,209500 Hypotrichosis 4,146550
HRAS	165.7	100%	98%	Congenital myopathy with excess of muscle spindles, 218040 Costello syndrome, 218040 Schimmelpenning-Feuerstein-Mims syndrome,somatic mosaic,163200 {Bladder cancer, somatic}, 109800 {Nevus sebaceous, somatic}, 162900 {Spitz nevus or nevus spilus,somatic},137550 {Thyroid carcinoma, follicular, somatic}, 188470
HRG	153.1	97%	94%	Thrombophilia due to elevated HRG,613116 Thrombophilia due to HRG deficiency,613116
HSD11B1	163.5	100%	100%	Cortisone reductase deficiency 2, 614662
HSD11B2	143.8	87%	84%	Apparent mineralocorticoid excess, 218030
HSD17B10	71.2	100%	98%	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 Mental retardation, X-linked syndromic 10, 300220 Mental retardation, X-linked 17/31, microduplication, 300705
HSD17B3	139.5	100%	100%	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	100.2	93%	92%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B2	152	100%	100%	3-beta-hydroxysteroid dehydrogenase, type II, deficiency, 201810
HSD3B7	127.2	98%	92%	Bile acid synthesis defect, congenital, 1, 607765
HSF4	89.4	96%	87%	Cataract 5, multiple types, 116800
HSPB1	28.6	95%	63%	Charcot-Marie-Tooth disease,axonal,type 2F,606595 Neuropathy, distal hereditary motor, type IIB, 608634
HSPB3	305.4	100%	100%	?Neuronopathy, distal hereditary motor, type IIC, 613376
HSPB8	126.7	100%	100%	Charcot-Marie-Tooth disease,axonal,type 2L,608673 Neuropathy, distal hereditary motor, type IIA, 158590
HSPD1	84.8	97%	89%	Leukodystrophy, hypomyelinating, 4, 612233 Spastic paraplegia 13, autosomal dominant, 605280
HSPG2	108	99%	97%	Dysegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
HTR1A	200.6	100%	100%	Periodic fever,menstrual cycle dependent,614674

HTRA1	98.5	78%	75%	CARASIL syndrome,600142 {Macular degeneration,age-related,7},610149 {Macular degeneration,age-related,neovascular type},610149
HTRA2	106	100%	96%	{Parkinson disease 13},610297
HTT	139.8	98%	97%	Huntington disease,143100
HUWE1	66	98%	94%	Mental retardation, X-linked syndromic, Turner type, 300706
HYAL1	101	100%	100%	Mucopolysaccharidosis type IX, 601492
HYDIN	130.8	99%	99%	Ciliary dyskinesia, primary, 5, 608647
HYLS1	148.9	100%	100%	Hydrolethalus syndrome, 236680
ICK	130	100%	99%	Endocrine-cerebroosteodysplasia,612651
ICOS	146.6	100%	100%	Immunodeficiency, common variable, 1, 607594
IDH2	95.2	100%	99%	D-2-hydrosylglutaric aciduria 2, 613657
IDH3B	166.6	100%	100%	Retinitis pigmentosa 46, 612572
IDS	62.5	98%	98%	Mucopolysaccharidosis II, 309900
IDUA	94.1	89%	85%	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Is, 607016 Mucopolysaccharidosis Ih/s, 607015
IER3IP1	55.9	98%	85%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFITM5	63.9	99%	89%	Osteogenesis imperfecta,type V,610967
IFNGR1	135.2	99%	96%	Mycobacterial infection, atypical, familial disseminated, 209950
IFT122	145.6	100%	99%	Cranioectodermal dysplasia 1, 218330
IFT140	105.2	99%	96%	Mainzer-Saldino syndrome, 266920
IFT172	108.6	99%	97%	Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT43	110.9	100%	100%	Cranioectodermal dysplasia 3, 614099
IFT80	55.9	79%	58%	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IGBP1	75.4	96%	90%	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472
IGF1	124.4	100%	100%	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IGF1R	130.1	100%	99%	Insulin-like growth factor I,resistance to,270450
IGF2R	135.2	98%	94%	Hepatocellular carcinoma,somatic,114550
IGFALS	62.3	100%	99%	Acid-labile subunit,deficiency of,615961
IGFBP7	54.9	92%	75%	Retinal arterial macroaneurysm with supraaortic pulmonic stenosis,614224
IGHMBP2	94.6	100%	87%	Neuronopathy, distal hereditary motor, type VI, 604320
IGLL1	68.7	98%	91%	Agammaglobulinemia 2, 613500
IGSF1	53.5	96%	86%	Hypothyroidism,central,and testicular enlargement,300888

IHH	112.6	100%	100%	Acrocaptofemoral dysplasia,607778 Brachydactyly,type A1,112500
IKBKAP	140.6	100%	99%	Dysautonomia, familial, 223900
IKBKB	112.8	98%	96%	Immunodeficiency 15,615592
IKBKG	25.6	68%	55%	Ectodermal dysplasia,hypohidrotic,with immune deficiency,300291 Ectodermal,dysplasia,anhidrotic,lymphedema and immunodeficiency,300301 Immunodeficiency 33,300636 Immunodeficiency,isolated,300584 Uncontinentia pigmenti,308300 Invasive pneumococcal disease,recurrent isolated,2,300640
IKZF1	162.4	100%	99%	Leukemia,acute lymphoblastic Systemic lupus erythematosus, association with (Han (2009) Nat Genet 41,1234)
IL10RA	129.6	100%	98%	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	185.5	100%	95%	Inflammatory bowel disease 25, early onset, autosomal recessive, 612567 {Hepatitis B virus, susceptibility to}, 610424
IL11RA	126.7	100%	97%	Craniosynostosis and dental anomalies, 614188
IL17F	76.1	100%	91%	Candidiasis, familial, 6, autosomal dominant, 613956
IL17RA	115.9	96%	94%	Candidiasis, familial, 5, autosomal recessive, 613953
IL17RD	119.3	99%	96%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
IL1RAPL1	77.5	100%	92%	Mental retardation, X-linked 21/34, 300143
IL1RN	164.4	100%	100%	{Gastric cancer risk after H. pylori infection}, 137215
IL21R	116.3	100%	100%	Immunodeficiency, primary, autosomal recessive, IL21R-related, 615207 [IgE, elevated level of], 147050
IL2RA	125.4	100%	97%	Interleukin-2 receptor, alpha chain, deficiency of, 606367
IL2RG	41.1	100%	92%	Severe combined immunodeficiency, X-linked, 300400
IL31RA	126.8	100%	99%	Amyloidosis,primary localized cutaneous 2,613955
IL36RN	84.2	100%	98%	Psoriasis, generalized pustular, 614204
IL7R	129.2	100%	98%	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
ILD1R	93.5	100%	99%	Deafness, autosomal recessive 42, 609646
IMPAD1	144.3	100%	98%	Chondrodysplasia with joint dislocations, GRAPP type, 614078
IMPDH1	47.6	91%	74%	Leber congenital amaurosis 11, 613837 Retinitis pigmentosa 10, 180105
IMPG2	166.8	100%	97%	Macular dystrophy,vitelliform,5,616152 Retinitis pigmentosa 56, 613581

INF2	76.5	92%	86%	Charcot-Marie-Tooth disease,dominant intermediate E,614455 Glomerulosclerosis, focal segmental, 5, 613237
ING1	96.3	97%	85%	Squamous cell carcinoma,head and neck,somatic,275355
INPP5E	83.9	96%	88%	Joubert syndrome 1,213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INPPL1	109.3	99%	93%	Opsismodysplasia, 258480
INS	89.6	100%	100%	Diabetes mellitus,insulin-dependent 2,125852 Diabetes mellitus,permanent neonatal,606176 Hyperproinsulinemia,616214 Maturity-onset diabetes of the young,type 10,613370
INSL3	82.1	80%	80%	Cryptorchidism,219050
INSR	123.7	97%	93%	Leprechaunism, 246200
INVS	153.4	100%	99%	Nephronophthisis 2, infantile, 602088
IQCB1	96.9	78%	74%	Senior-Loken syndrome 5, 609254
IQSEC2	37.5	87%	76%	Mental retardation, X-linked 1, 309530
IRAK4	96.9	99%	91%	IRAK4 deficiency, 607676
IRF1	147	100%	100%	Gastric cancer,somatic,613659 Myelodysplastic syndrome,preleukemic Myelogenous leukemia,acute Nonsmall cell lung cancer,somatic,211980
IRF4	153.5	99%	97%	Multiple myeloma,254500 [Skin/hair/eye pigmentation, variation in,8],611724
IRF6	121.7	99%	93%	Orofacial cleft 6,608864 Popliteal pterygium syndrome 1,119500 van der Woude syndrome,119300
IRF8	88.1	100%	95%	Monocyte and dendritic cell deficiency, recessive, 614894
IRGM	160.9	100%	100%	Inflammatory bowel disease 19,612278 {Mycobacterium tuberculosis,protection against},607948
IRX5	57.1	93%	82%	Hamamy syndrome,611174
ISCU	123.5	100%	99%	Myopathy with lactic acidosis, hereditary, 255125
ISPD	107.2	96%	82%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
ITCH	115.6	95%	95%	Autoimmune disease, syndromic multisystem, 613385
ITGA2B	99	98%	92%	Bleeding disorder,platelet-type,16,autosomal dominant,187800 Glanzmann thrombasthenia,273800



				Thrombocytopenia,neonatal alloimmune,BAK antigen related
ITGA3	123.9	99%	96%	Interstitial lung disease, nephrotic syndrome and epidermolysis bullosa, congenital,614748
ITGA6	145.9	99%	98%	Epidermolysis bullosa,junctional, with pyloric stenosis,226730
ITGA7	115.7	98%	94%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
ITGA8	121.2	99%	98%	Renal hypodysplasia/aplasia 1, 191830
ITGB2	140.4	100%	99%	Leukocyte adhesion deficiency, 116920
ITGB3	124.8	100%	97%	Bleeding disorder,platelet-type 16,autosomal dominant,187800 Glanzmann thrombasthenia,273800 Purpura,posttransfusion Thrombocytopenia,neonatal alloimmune {Myocardial infarction,susceptibility to},608446
ITGB4	132.4	96%	93%	Epidermolysis bullosa of hands and feet,131800 Epidermolysis bullosa,junctional,non-Herlitz type,226650 Epidermolysis bullosa,junctional,with pyloric atresia,226730
ITK	118.5	100%	98%	Lymphoproliferative syndrome 1, 613011
ITM2B	115.5	100%	100%	?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities,616079 Dementia,familial British,176500 Dementia,familial Danish,117300
ITPR1	157.5	100%	99%	Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360
IVD	106.8	100%	100%	Isovaleric acidemia, 243500
IYD	115.4	100%	97%	Thyroid dysmorphogenesis 4,274800
JAG1	139.8	100%	97%	Alagille syndrome, 118450
JAK2	89.8	95%	93%	Erythrocytosis,somatic,133100 Leukemia,acute myelogenous,601626 Myelofibrosis,somatic,254450 Polycythemia vera,263300 Thrombocythemia 3,614521 {Budd-Chiari syndrome},600880
JAK3	95.9	97%	93%	SCID, autosomal recessive, T-negative/B-positive type, 600802
JAM3	136.4	100%	99%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
JPH2	74.8	96%	82%	Cardiomyopathy, familial hypertrophic 17, 613873
JPH3	118.2	100%	96%	Huntington disease-like 2,606438

JUP	134.9	100%	99%	Arrhythmogenic right ventricular dysplasia 12,611528 Naxos disease,601214
KAL1	65.1	89%	85%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
KANK1	155.8	100%	100%	Cerebral palsy, spastic quadriplegic, 2, 612900
KANSL1	70.8	96%	89%	Koolen-De Vries syndrome, 610443
KARS	116.5	100%	98%	?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness,autosomal recessive 89,613916
KAT6B	167.3	98%	97%	Genitopatellar syndrome, 606170 SBBYSS syndrome, 603736
KBTBD13	80.5	100%	94%	Nemaline myopathy 6, autosomal dominant, 609273
KCNA1	170.1	100%	99%	Episodic ataxia/myokymia syndrome, 160120
KCNA5	123.4	99%	96%	Atrial fibrillation, familial, 7, 612240
KCNC3	140.8	64%	53%	Spinocerebellar ataxia 13,605259
KCND3	172.2	100%	100%	Spinocerebellar ataxia 19, 607346
KCNE1	443.9	100%	100%	Jervell and Lange-Nielsen syndrome 2, 612347 Long QT syndrome-5, 613695
KCNE2	113.9	100%	100%	Atrial fibrillation, familial, 4, 611493 Long QT syndrome-6, 613693
KCNE3	171.2	100%	100%	Brugada syndrome 6, 613119
KCNH2	89.2	93%	83%	Long QT syndrome-2, 613688 {Long QT syndrome-2, acquired, susceptibility to}, 613688 Short QT syndrome-1, 609620
KCNJ1	230.7	100%	100%	Bartter syndrome, type 2, 241200
KCNJ10	199.6	100%	100%	SESAME syndrome, 612780
KCNJ11	257	100%	100%	Diabetes mellitus,permanent neonatal,with neurologic features,606176 Diabetes mellitus,transient neonatal,3,610582 Hyperinsulinemic hypoglycemia,familial,2,601820 Maturity-onset diabetes of the young,type 13,616329 {Diabetes mellitus,type 2,susceptibility to},125853
KCNJ13	194	100%	98%	Leber congenital amaurosis 16, 614186 Snowflake vitreoretinal degeneration, 193230
KCNJ2	212.3	100%	100%	Andersen syndrome, 170390 Atrial fibrillation, familial, 9, 613980 Short QT syndrome-3, 609622

KCNJ5	179.7	100%	99%	Hyperaldosteronism, familial, type III, 613677 Long QT syndrome 13, 613485
KCNK3	140.3	100%	98%	Pulmonary hypertension, primary 4, 615344
KCNK9	164.3	100%	100%	Birk-Barel mental retardation dysmorphism syndrome, 612292
KCNMA1	133.7	100%	99%	Generalized epilepsy and paroxysmal dyskinesia, 609446
KCNQ1	96.7	88%	85%	Atrial fibrillation, familial, 3, 607554 Jervell and Lange-Nielsen syndrome, 220400 Long QT syndrome-1, 192500 Short QT syndrome-2, 609621 {Long QT syndrome 1, acquired, susceptibility to}, 192500
KCNQ1OT1				Beckwith-Wiedemann syndrome, 130650
KCNQ2	80.5	98%	92%	Epileptic encephalopathy, early infantile, 7, 613720 Myokymia, 121200 Seizures, benign neonatal, 1, 121200
KCNQ3	98.8	100%	94%	Seizures, benign neonatal, type 2, 121201
KCNQ4	118.2	93%	91%	Deafness, autosomal dominant 2A, 600101
KCNT1	106.6	94%	91%	Epileptic encephalopathy, early infantile, 14, 614959 Epilepsy, nocturnal frontal lobe, 5, 615005
KCNV2	120	100%	100%	Retinal cone dystrophy 3B, 610356
KCTD1	130	92%	86%	Scalp-ear-nipple syndrome, 181270
KCTD7	113.5	95%	93%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM5C	68.6	95%	90%	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
KDM6A	74.7	88%	75%	Kabuki syndrome 2, 300867
KDR	146.3	100%	99%	Hemangioma, capillary infantile, somatic, 602089
KERA	185.9	100%	100%	Cornea plana congenita, recessive, 217300
KHDC3L	87.4	99%	87%	Hydatidiform mole, recurrent, 2, 614293
KIAA0196	141.3	100%	97%	Spastic paraplegia 8, autosomal dominant, 603563
KIAA1279	165.3	100%	100%	Goldberg-Shprintzen megacolon syndrome, 609460
KIAA2022	99.9	100%	98%	Mental retardation, X-linked 98, 300912
KIF11	76.6	97%	95%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF1A	107.9	98%	94%	Mental retardation, autosomal dominant 9, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal recessive, 610357

KIF1B	149.8	99%	98%	Charcot-Marie-Tooth disease, type 2A1, 118210 Pheochromocytoma, 171300 {Neuroblastoma, susceptibility to, 1}, 256700
KIF1C	101.7	100%	98%	Spastic ataxia 2,autosomal recessive, 611302
KIF21A	123.4	98%	93%	Fibrosis of extraocular muscles,congenital,1,135700 Fibrosis of extraocular muscles,congenital,3B,135700
KIF22	135	100%	99%	Spondyloepimetaphyseal dysplasia with joint laxity,type 2,603546
KIF2A	98	100%	84%	Cortical dysplasia,complex,with other brain malformations 3,615411
KIF5A	126.5	100%	99%	Spastic paraplegia 10, autosomal dominant, 604187
KIF7	70	91%	84%	?Al-Gazali-Bakalinova syndrome,607131 ?Hydroletharus syndrome 2,614120 Acrocallosal syndrome,200990 Joubert syndrome 12,200990
KIRREL3	132.8	100%	98%	Mental retardation, autosomal dominant 4, 612581
KISS1	35.9	99%	95%	Hypogonadotropic hypogonadism 13 with or without anosmia, 614842
KISS1R	74.7	97%	88%	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 ?Precocious puberty,central,1,176400
KIT	153.8	99%	98%	Gastrointestinal stromal tumor, familial, 606764 Germ cell tumors, 273300 Leukemia, acute myeloid, 601626 Mast cell disease, 154800 Piebaldism, 172800
KITLG	76.3	91%	88%	Hyperpigmentation familial progressive 2,145250 [Skin/hair/eye pigmentation 7],611664
KL	162	98%	95%	Tumoral calcinosis, hyperphosphatemic,211900
KLF1	50.7	90%	83%	Blood group--Lutheran inhibitor, 111150 Dyserthropoietic anemia,congenital,type IV,613673 [Hereditary persistence of fetal hemoglobin], 613566
KLF11	143.9	100%	100%	Maturity-onset diabetes of the young,type VII,610508
KLF6	117.4	100%	97%	Gastric cancer,somatic,613659 Prostate cancer,somatic,176807
KLHDC8B	92.2	100%	100%	{Hodgkin lymphoma,susceptibility to},236000
KLHL10	196.7	100%	99%	Spermatogenic failure 11,615081
KLHL3	132.3	99%	98%	Pseudohypoaldosteronism,type IID,614495

KLHL40	118.2	100%	100%	Nemaline myopathy 8,autosomal recessive,615348
KLHL41	198.1	100%	98%	Nemaline myopathy 9, 615731
KLHL7	139.1	99%	98%	Retinitis pigmentosa 42, 612943
KLK4	180	100%	94%	Amelogenesis imperfecta type IIA1,204700
KLKB1	152.3	97%	92%	Fletcher factor (prekallikrein) deficiency,612423
KLLN	81.4	100%	100%	Cowden syndrome 4, 615107
KMT2A	142.8	99%	98%	Leukemia, myeloid/lymphoid or mixed-lineage, 159555 Wiedemann-Steiner syndrome, 605130
KMT2D	133.1	99%	99%	Kabuki syndrome 1, 147920
KPTN	88.6	100%	99%	Mental retardation, autosomal recessive 41, 615637
KRAS	57.8	100%	99%	Bladder cancer,somatic,109800 Breast cancer,somatic,114480 Cardiofaciocutaneous syndrome 2,615278 Gastric cancer,somatic,137215 Leukemia,acute myeloid,601626 Lung cancer,somatic,211980 Noonan syndrome 3,609942 Pancreatic carcinoma, somatic,260350 RAS-associated autoimmune leukoproliferative disorder,614470 Schimmelpenning-Feuerstein-Mins syndrome,somatic mosaic,163200
KRIT1	85.9	100%	99%	Cavernous malformations of CNS and retina,116860 Cerebral cavernous malformations-1,116860 Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations,116860
KRT1	134.3	99%	91%	Epidermolytic hyperkeratosis,113800 Ichthyosis histrix,Curth-Macklin type,146590 Ichthyosis, cyclic, with epidermolytic hyperkeratosis,607602 Keratosis palmoplantaris striata III,607654 Palmoplantar keratoderma,epidermolytic,144200
KRT10	90.8	94%	91%	Epidermolytic hyperkeratosis,113800 Ichthyosis with confetti,609165 Ichthyosis,cyclic,with epidermolytic hyperkeratosis,607602
KRT12	88	95%	92%	Meesmann corneal dystrophy, 122100
KRT13	111.4	100%	94%	White sponge nevus 2,615785

KRT14	53.6	86%	78%	Dermatopathia pigmentosa reticularis,125595 Epidermolysis bullosa simplex,Dowling-Meara type,131760 Epidermolysis bullosa simplex,Koebner type,131900 Epidermolysis bullosa simplex,recessive 1,601001 Epidermolysis bullosa simplex,Weber-Cockayne type,13
KRT16	28.8	71%	47%	Pachyonychia congenita 1,167200 Palmoplantar keratoderma,nonepidermolytic,focal,613000
KRT17	19.3	44%	34%	Pachyonychia congenita 2,167210 Steatocystoma multiplex,184500
KRT18	30.7	92%	53%	Cirrhosis, cryptogenic,215600 {Cirrhosis,noncryptogenic,susceptibility to},215600
KRT2	121.2	99%	97%	Ichthyosis bullosa of Siemens,146800
KRT3	90.9	100%	96%	Meesmann corneal dystrophy, 122100
KRT4	108.2	100%	98%	White sponge nevus 1,193900
KRT5	109	100%	99%	Dowling-Degos disease 1,179850 Epidermolysis bullosa simplex,Dowling-Meara type,131760 Epidermolysis bullosa simplex,Koebner type,131900 Epidermolysis bullosa simplex,recessive 1,601001 Epidermolysis bullosa simplex,Weber-Cockayne type,131800
KRT6A	164	90%	85%	Pachyonychia congenita 3,167200
KRT6B	169.4	90%	85%	Pachyonychia congenita Jackson-Lawler type,615726
KRT6C	158.9	89%	82%	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse,615735
KRT74	126.9	100%	100%	?Ectodermal dysplasia 7, hair/nail type,614929 ?Hypotrichosis 3,613981 Woolly hair, autosomal dominant,194300
KRT8	37.3	85%	67%	Cirrhosis, cryptogenic,215600 {Cirrhosis,noncryptogenic,susceptibility to},215600
KRT81	73.5	93%	88%	Monilethrix,158000
KRT83	70.6	98%	86%	Monilethrix,158000
KRT85	98.4	98%	92%	Ectodermal dysplasia 4 hair/nail type,602032
KRT86	86.6	100%	87%	Monilethrix,158000
KRT9	71.4	95%	93%	Epidermolytic palmoplantar keratoderma,144200

L1CAM	87.3	98%	95%	Corpus callosum,partial agenesis of,304100 CRASH syndrome, 303350 Hydrocephalus due to aqueductal stenosis, 307000 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 Hydrocephalus with Hirschsprung disease, 307000 MASA syndrome, 303350
L2HGDH	120.8	97%	95%	L-2-hydroxyglutaric aciduria, 236792
LAMA2	144.2	99%	98%	Muscular dystrophy, congenital merosin-deficient, 607855 Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855
LAMA3	149.1	99%	99%	Epidermolysis bullosa,generalized atrophic benign,226650 Epidermolysis bullosa,junctional,Herlitz type,226700 Laryngoonychocutaneous syndrome,245660
LAMA4	129	100%	99%	Cardiomyopathy, dilated, 1JJ, 615235
LAMB1	156.3	100%	99%	Lissencephaly 5,615191
LAMB2	175.6	100%	99%	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 Pierson syndrome,609049
LAMB3	107.8	100%	99%	Amelogenesis imperfecta,type IA,104530 Epidermolysis bullosa,junctional,Herlitz type,226700 Epidermolysis bullosa,junctional,non-Herlitz type,226650
LAMC2	108.7	100%	97%	Epidermolysis bullosa,junctional,Herlitz type,226700 Epidermolysis bullosa,junctional,non-Herlitz type,226650
LAMC3	112.2	98%	92%	Cortical malformations, occipital, 614115
LAMP2	76.4	92%	91%	Danon disease, 300257
LAMTOR2	139	100%	100%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LARGE	122.9	100%	97%	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	48.3	71%	58%	Alazami syndrome, 615071
LARS2	126.7	100%	100%	Perrault syndrome 4, 615300
LBR	71.6	93%	79%	?Reynolds syndrome,613471 Greenberg skeletal dysplasia,215140 Pelger-Huet anomaly,169400
LCA5	130	95%	94%	Leber congenital amaurosis 5, 604537
LCAT	133	99%	94%	Norum disease, 245900
LCT	124.4	99%	96%	Lactase deficiency, congenital, 223000

LDB3	108.4	94%	92%	Cardiomyopathy, dilated 1C, 601493 Left ventricular noncompaction 3, with or without dilated cardiomyopathy, 601493 Myopathy, myofibrillar, 4, 609452
LDHA	51.8	89%	81%	Glycogen storage disease XI, 612933
LDHB	103	96%	91%	Lactate dehydrogenase-B deficiency, 614128
LDLR	158.9	99%	97%	Hypercholesterolemia,familial,143890 LDL cholesterol level QTL2,143890
LDLRAP1	136.5	97%	90%	Hypercholesterolemia,familial,autosomal recessive,603813
LEF1	121.2	100%	100%	Sebaceous tumors,somatic
LEFTY2	36.5	75%	65%	Left-right axis malformations (Koasaki (1999) Am J Hum Genet 64, 712)
LEMD3	92	94%	86%	Buschke-Ollendorff syndrome,166700 Melorheostosis with osteopoikilosis,155950 Osteopoikilosis,166700
LEP	163.8	100%	95%	Obesity,morbid,due to leptin deficiency,614962
LEPR	107.3	93%	88%	Obesity,morbid,due to leptin receptor deficiency,614963
LEPRE1	124.9	100%	100%	Osteogenesis imperfecta,type VIII,610915
LEPREL1	87.8	93%	87%	Myopia,high,with cataract and vitreoretinal degeneration,614292
LFNG	69.2	82%	79%	Spondylocostal dysostosis, autosomal recessive 3, 609813
LGI1	177.2	95%	91%	Epilepsy, familial temporal lobe, 1, 600512
LHB	29.3	99%	84%	Hypogonadotropic hypogonadism 23 with or without anosmia,228300
LHCGR	149	94%	92%	Leydig cell adenoma,somatic,with precocious puberty,176410 Leydig cell hypoplasia with hypergonadotropic hypogonadism,238320 Leydig cell hypoplasia with pseudohermaphroditism,238320 Luteinizing hormone resistance,female,238320
LHFPL5	289.3	100%	100%	Deafness, autosomal recessive 67, 610265
LHX3	85.7	97%	80%	Pituitary hormone deficiency,combined,3,221750
LHX4	121.4	100%	96%	Pituitary hormone deficiency,combined,4,262700
LIAS	149.8	100%	98%	Pyruvate dehydrogenase lipoic acid synthetase deficiency, 614462
LIFR	125.5	94%	87%	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome,601559
LIG1	81.9	99%	97%	DNA ligase I deficiency
LIG4	163.2	100%	98%	LIG4 syndrome, 606593 Severe combined immunodeficiency with sensitivity to ionizing radiation, 602450 {Multiple myeloma, resistance to}, 254500
LIM2	88.4	100%	99%	Cataract 19, 615277



LINS	131.6	99%	96%	Mental retardation, autosomal recessive 27, 614340
LIPA	109.7	95%	95%	Cholesteryl ester storage disease, 278000 Wolman disease, 278000
LIPC	116.6	99%	98%	Diabetes mellitus, noninsulin-dependent, 125853 Hepatic lipase deficiency, 614025 [High density lipoprotein cholesterol level QTL 12], 612797
LIPH	116.4	100%	99%	Hypotrichosis 7,604379 Woolly hair,autosomal recessive 2,with or without hypotrichosis
LIPN	141.2	98%	96%	Ichthyosis,congenital,autosomal recessive 8,613943
LITAF	115.7	99%	93%	Charcot-Marie-Tooth disease, type 1C, 601098
LMAN1	128.7	99%	87%	Combined factor V and VIII deficiency,227300
LMBR1	94.8	92%	87%	Acheiropody,200500 Hypoplastic or aplastic tibia with polydactyly,188740 Laurin-Sandrow syndrome,135750 Polydactyly,preaxial type II,174500 Syndactyly,type IV,186200 Triphalangeal thumb type I,174500
LMBRD1	73.2	87%	82%	Methylmalonic aciduria and homocystinuria, cb1F type, 277380
LMF1	125.9	99%	97%	Lipase deficiency,combined,246650
LMNA	72	95%	87%	Cardiomyopathy,dilated,1A,115200 Charcot-Marie-Tooth disease,type 2B1,605588 Emery-Dreifuss muscular dystrophy 2,AD,181350 Emery-Dreifuss muscular dystrophy 3,AR,616516 Heart-hand syndrome,Slovenian type,610140 Hutchinson-Gilford progeria,176670 Lipodystrophy,familial partial,2,151660 Malouf syndrome,212112 Mandibuloacral dysplasia,248370 Muscular dystrophy,congenital,613205 Muscular dystrophy,limb-girdle,type 1B,159001 Restrictive dermopathy,lethal,275210
LMNB1	112.2	98%	96%	Leukodystrophy,adult-onset,autosomal dominant,169500
LMX1B	108.5	100%	88%	Nail-patella syndrome, 161200
LOR	16.1	82%	32%	Vohwinkel syndrome with ichthyosis,604117
LOXHD1	128.4	99%	98%	Deafness, autosomal recessive 77, 613079

LPAR6	108.2	100%	96%	Hypotrichosis 8,278150 Woolly hair,autosomal recessive 1,with or without hypotrichosis,278150
LPIN1	123.7	97%	92%	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	119.1	100%	99%	Majeed syndrome, 609628
LPL	156.4	100%	100%	Combined hyperlipidemia, familial, 144250 Lipoprotein lipase deficiency, 238600 [High density lipoprotein cholesterol level QTL 11]
LPP	118.5	100%	99%	Leukemia,acute myeloid,601626 Lipoma
LRAT	285.3	100%	100%	Leber congenital amaurosis 14, 613341 Retinal dystrophy, early-onset severe, 613341 Retinitis pigmentosa, juvenile, 613341
LRBA	126.4	99%	96%	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRIG2	158.2	100%	97%	Urofacial syndrome 2
LRIT3	148.4	94%	93%	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRP2	176.4	100%	99%	Donnai-Barrow syndrome, 222448
LRP4	157.9	99%	98%	Cenani-Lenz syndactyly syndrome, 212780 Sclerosteosis 2, 614305
LRP5	165.5	98%	96%	Exudative vitreoretinopathy 4, 601810 Hyperostosis, endosteal, 144750 Osteoporosis-pseudoglioma syndrome, 259770 Osteosclerosis, 144750 van Buchem disease, type 2, 607636 [Bone mineral density variability 1], 601884 {Osteoporosis}, 166710
LRPAP1	129.2	99%	96%	Myopia 23,autosomal recessive,615431
LRPPRC	130	98%	94%	Leigh syndrome, French-Canadian type, 220111
LRRC6	163.6	96%	92%	Ciliary dyskinesia, primary, 19, 614935
LRRC8A	245	100%	98%	Agammaglobulinemia 5, 613506
LRRK2	117	99%	92%	{Parkinson disease 8},607060
LRSAM1	122.7	100%	99%	Charcot-Marie-Tooth disease, axonal, type 2P, 614436
LRTOMT	105.8	98%	90%	Deafness, autosomal recessive 63, 611451
LTBP2	98.5	95%	93%	Glaucoma 3,primary congenital,D,613086 Microspherophakia and/or megalocornea,with ectopia lentis and with or without secondary

				glaucoma,251750 Weill-Marchesani syndrome 3,recessive,614819
LTBP3	99.2	98%	96%	Dental anomalies and short stature,601216
LTBP4	100.8	99%	93%	Cutis laxa autosomal recessive type IC,613177
LYST	129.6	97%	92%	Chediak-Higashi syndrome,214500
LYZ	171.9	100%	100%	Amyloidosis, renal, 105200
LZTFL1	121.2	98%	90%	Bardet-Biedl syndrome 17, 615994
LZTS1	84.6	100%	100%	Esophageal squamous cell carcinoma,133239
MAD1L1	81.6	99%	93%	Lymphoma,somatic Prostate cancer,somatic,176807
MAF	51.8	75%	72%	Cataract, pulverulent or cerulean, with or without microcornea, 610202
MAFB	104.3	100%	99%	Multicentric carpotarsal osteolysis syndrome, 166300
MAGEL2	114.5	100%	100%	Prader-Willi-like syndrome, 615547
MAGT1	71	97%	84%	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853 Mental retardation, X-linked 95, 300716
MAK	146.6	94%	93%	Retinitis pigmentosa 62, 614181
MAML2	113.1	100%	99%	Mucoepidermoid salivary gland carcinoma
MAMLD1	76.8	100%	96%	Hypospadias 2,X-linked,300758
MAN1B1	125.7	100%	100%	Mental retardation, autosomal recessive 15, 614202
MAN2B1	103.9	97%	91%	Mannosidosis, alpha-, types I and II, 248500
MANBA	120.6	99%	93%	Mannosidosis, beta, 248510
MAOA	81.3	98%	96%	Brunner syndrome, 300615
MAP2K1	89.5	100%	98%	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	103.9	94%	90%	Cardiofaciocutaneous syndrome 4, 615280
MAP3K1	152.7	92%	85%	46XY sex reversal 6,613762
MAP3K8	151.4	100%	100%	Lung cancer,somatic,211980
MAPT	88.8	99%	93%	Dementia,frontotemporal,with or without parkinsonism,600274 Pick disease,172700 Supranuclear palsy,progressive,601104 Supranuclear palsy,progressive atypical,260540 {Parkinson disease,susceptibility to},168600
MARS2	122.6	100%	100%	Spastic ataxia 3, autosomal recessive, 611390
MARVELD2	146	93%	92%	Deafness, autosomal recessive 49, 610153
MASP1	139.7	100%	99%	3MC syndrome 1,257920

MASP2	149.4	100%	96%	MASP2 deficiency, 613791
MASTL	121.6	99%	98%	?Thrombocytopenia 2,188000
MAT1A	176.9	97%	96%	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MATN3	110.1	84%	83%	Epiphyseal dysplasia,multiple,5,607078
MATR3	82.2	96%	87%	Myopathy, distal 2, 606070
MBD5	179.2	100%	100%	Mental retardation, autosomal dominant 1, 156200
MBTPS2	84.9	99%	91%	IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosi follicularis spinulosa decalvans, X-linked, 308800
MC2R	192.2	100%	100%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MC4R	291.6	100%	100%	Obesity,autosomal dominant,601665
MCC	132.1	100%	99%	Colorectal cancer,somatic,114500
MCCC1	147.3	100%	100%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	120.8	99%	96%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	116.2	100%	100%	Methylmalonyl-CoA epimerase deficiency, 251120
MCFD2	104.9	100%	100%	Factor V and factor VIII,combined deficiency of,613625
MCM4	158.4	100%	97%	Natural killer cell and glucocorticoid deficiency with DNA repair defect, 609981
MCM6	160.2	100%	99%	Lactase persistence/nonpersistence,223100
MCOLN1	130.6	97%	95%	Mucopolidosis IV, 252650
MCPH1	133.9	99%	94%	Microcephaly 1, primary, autosomal recessive, 251200
MECP2	48.4	97%	75%	Encephalopathy, neonatal severe, 300673 Mental retardation,X-linked,syndromic,Lubs type,300260 Mental retardation, X-linked, syndromic 13, 300055 Rett syndrome, 312750 Rett syndrome,atypical,312750 Rett syndrome,preserved speech variant,312750 {Autism susceptibility, X-linked 3}, 300496
MED12	64.3	95%	89%	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Opitz-Kaveggia syndrome, 305450
MED13L	131.9	100%	98%	Mental retardation and distinctive facial features with or without cardiac defects,616789 Transposition of the great arteries, dextro-looped 1, 608808
MED17	129.9	99%	97%	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668

MED23	131	99%	97%	Mental retardation, autosomal recessive 18, 614249
MED25	82.6	97%	85%	?Charcot-Marie-Tooth disease, type 2B2, 605589 Basel-Vanagait-Smirin-Yosef syndrome,616449
MEF2C	112.8	99%	88%	Chromosome 5q14.3 deletion syndrome, 613443 Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
MEFV	105.6	94%	89%	Familial Mediterranean fever, AR, 249100
MEGF10	140.6	100%	99%	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399
MEGF8	109	99%	96%	Carpenter syndrome 2, 614976
MEN1	110.9	96%	89%	Adrenal adenoma, somatic Angiofibroma, somatic Carcinoid tumor of lung Lipoma, somatic Multiple endocrine neoplasia 1, 131100 Parathyroid adenoma, somatic
MEOX1	85.4	95%	93%	Klippel-Feil syndrome 2,214300
MERTK	162.2	100%	99%	Retinitis pigmentosa 38, 613862
MESP2	58	86%	79%	Spondylocostal dysostosis 2,autosomal recessive,608681
MET	192.4	100%	99%	papillary renal cell cancer
MFN2	138	100%	100%	Charcot-Marie-Tooth disease, type 2A2, 609260 Hereditary motor and sensory neuropathy VIA,601152
MFRP	117.5	100%	100%	Microphthalmia, isolated 5, 611040 Nanophthalmos 2, 609549
MFSD8	120.9	100%	98%	Ceroid lipofuscinosis, neuronal, 7, 610951
MGAT2	137.6	100%	99%	Congenital disorder of glycosylation, type IIa, 212066
MGME1	168.8	100%	100%	Mitochondrial DNA depletion syndrome 11, 615084
MGP	132.8	91%	91%	Keutel syndrome,245150
MIB1	144.8	99%	99%	Left ventricular noncompaction 7, 615092
MICU1	113.1	94%	89%	Myopathy with extrapyramidal signs, 615673
MID1	111.1	100%	99%	Opitz GBBB syndrome, type I, 300000
MINPP1	138.1	99%	98%	Thyroid carcinoma, follicular, 188470
MIP	104.3	94%	87%	Cataract 15, multiple types, 615274
MIR17HG				Feingold syndrome 2,614326
MIR184				EDICT syndrome,614303

MIR96				Deafness,autosomal dominant 50,613074
MITF	134.3	100%	100%	Tietz albinism-deafness syndrome,103500 Waardenburg syndrome, type 2A,193510 Waardenburg syndrome/ocular albinism, digenic,103470 {Melanoma,cutaneous malignant,susceptibility to 8},614456
MKKS	222.1	89%	89%	Bardet-Biedl syndrome 6,605231 McKusick-Kaufman syndrome, 236700
MKL1	89.8	98%	93%	Megakaryoblastic leukemia,acute
MKRN3	113.6	100%	98%	Precocious puberty,central,2,615346
MKS1	95.9	99%	96%	Bardet-Biedl syndrome 13,615990 Meckel syndrome 1, 249000
MLC1	90.6	100%	100%	Megalencephalic leukoencephalopathy with subcortical cysts,604004
MLH1	162.3	100%	99%	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MLH3	157	100%	100%	Colorectal cancer,hereditary nonpolyposis,type 7,614385 Colorectal cancer,somatic,114500 {Endometrial cancer,susceptibility to},608089
MLLT10	129.9	95%	92%	Leukemia,acute myeloid,601626
MLLT11	83.2	100%	100%	Leukemia,acute myelomonocytic,somatic,607785
MLPH	83.1	97%	94%	Griscelli syndrome type 3,609227
MLYCD	71	93%	90%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	177.2	100%	99%	Methylmalonic aciduria, vitamin B12-responsive, 251100
MMAB	97.1	100%	100%	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cbIB complementation type, 251110
MMACHC	173.1	100%	100%	Methylmalonic aciduria and homocystinuria, cbIC type, 277400
MMADHC	79.1	83%	55%	Homocystinuria, cbID type, 277410
MMP1	174.8	100%	100%	COPD,rate of decline of lung function in,606963 {Epidermolysis bullosa dystrophica,autosomal recessive,modifier of},226600
MMP13	136.6	92%	91%	Metaphyseal anadysplasia 1,602111 Spondyloepimetaphyseal dysplasia,Missouri type,602111
MMP2	148.6	100%	100%	Torg-Winchester syndrome,259600
MMP20	101.1	99%	97%	Amelogenesis imperfecta type IIA2,612529
MMP9	100.1	100%	97%	Metaphyseal anadysplasia 2,613073

MN1	77	95%	91%	Meningioma,607174
MNX1	24.9	56%	51%	Currarino syndrome,176450
MOCS1	73	96%	91%	Molybdenum cofactor deficiency, type A, 252150
MOCS2	145.7	99%	99%	Molybdenum cofactor deficiency, type B, 252150
MOG	9.8	38%	11%	?Narcolepsy 7,614250
MOGS	99.7	99%	98%	Congenital disorder of glycosylation, type 2b, 606056
MPC1	110.7	100%	98%	Mitochondrial pyruvate carrier deficiency,614741
MPDU1	113.6	100%	100%	Congenital disorder of glycosylation, type If, 609180
MPDZ	156.9	98%	96%	Hydrocephalus, nonsyndromic, autosomal recessive 2, 615219
MPI	112.9	100%	100%	Congenital disorder of glycosylation, type Ib, 602579
MPL	127.5	98%	92%	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498
MPLKIP	76.3	88%	52%	Trichothiodystrophy, nonphotosensitive 1, 234050
MPO	142.8	100%	99%	Cardiomyopathy, dilated, 1T, 613740 Myeloperoxidase deficiency, 254600 {Alzheimer disease, susceptibility to}, 104300 {Lung cancer, protection against, in smokers}
MPV17	103	100%	100%	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810 -3
MPZ	88.5	100%	97%	Charcot-Marie-Tooth disease,dominant intermediate D,607791 Charcot-Marie-Tooth disease, type 1B, 118200 Charcot-Marie-Tooth disease, type 2I, 607677 Charcot-Marie-Tooth disease, type 2J, 607736 Dejerine-Sottas disease,145900
MR1	120.7	99%	97%	Paroxysmal nonkinesigenic dyskinesia,118800
MRAP	158.5	100%	100%	Glucocorticoid deficiency 2,607398
MRE11A	53.1	95%	84%	Ataxia-telangiectasia-like disorder, 604391
MRPL3	61.9	88%	72%	Combined oxidative phosphorylation deficiency 9, 614582
MRPS16	119.4	100%	100%	Combined oxidative phosphorylation deficiency 2, 610498
MRPS22	129.9	93%	90%	Combined oxidative phosphorylation deficiency 5, 611719
MS4A1	119.2	98%	93%	Immunodeficiency, common variable, 5, 613495
MSH2	109.1	96%	88%	Colorectal cancer, hereditary nonpolyposis, type 1, 120435 Muir-Torre syndrome, 158320 Mismatch repair cancer syndrome, 276300

MSH3	109	96%	92%	Endometrial carcinoma,somatic,608089
MSH6	159.6	100%	99%	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 Endometrial cancer, familial, 608089 Mismatch repair cancer syndrome, 276300
MSR1	181.2	100%	100%	Barett esophagus/esophageal adenocarcinoma,614266 Prostate cancer,hereditary,176807
MSRB3	147.3	96%	96%	Deafness, autosomal recessive 74, 613718
MSTN	176.6	100%	99%	Muscle hypertrophy, 614160
MSX1	75.7	94%	87%	Ectodermal dysplasia 3,Witkop type,189500 Orofacial cleft 5,608874 Tooth agenesis,selective,1,with or without orofacial cleft,106600
MSX2	82.3	100%	92%	Craniosynostosis, type 2, 604757 Parietal foramina 1, 168500 Parietal foramina with cleidocranial dysplasia, 168550
MTAP	104.9	92%	84%	Diaphyseal medullary stenosis with malignant fibrous histiocytoma,112250
MTFMT	130.8	99%	95%	Combined oxidative phosphorylation deficiency 15, 614947
MTHFR	134.9	100%	100%	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}
MTM1	72.4	96%	81%	Myotubular myopathy, X-linked, 310400
MTMR2	115	100%	98%	Charcot-Marie-Tooth disease, type 4B1, 601382
MTO1	155.5	90%	88%	Combined oxidative phosphorylation deficiency 10, 614702
MTPAP	115.1	99%	90%	Ataxia, spastic, 4, 613672
MTR	132.8	99%	99%	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTRR	118.7	100%	97%	Homocystinuria-megaloblastic anemia, cbl E type, 236270
MTTP	142.3	99%	97%	Abetalipoproteinemia, 200100; {Metabolic syndrome, protection against}, 605552
MUC1	87	96%	92%	Medullary cystic kidney disease 1,174000
MUSK	158.2	100%	100%	Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931
MUT	113.3	97%	91%	Methylmalonic aciduria, mut(0) type, 251000
MUTYH	135	100%	99%	Adenomas, multiple colorectal, 608456 Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600



				Gastric cancer, somatic, 613659
MVK	126.3	100%	100%	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, disseminated superficial actinic, 175900
MXI1	102.6	89%	85%	Neurofibrosarcoma {Prostate cancer,susceptibility to},176807
MYBPC1	159.7	99%	98%	Arthrogryposis,distal,type 1B,614335 Lethal congenital contracture syndrome 4,614915
MYBPC3	129.6	97%	93%	Cardiomyopathy, dilated, 1MM, 615396 Cardiomyopathy, familial hypertrophic, 4, 115197 Left ventricular noncompaction 10, 615396
MYC	149.6	100%	100%	Burkitt lymphoma,113970
MYCN	68.2	90%	78%	Feingold syndrome, 164280
MYD88	172.2	100%	99%	Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260
MYF6	81.4	100%	100%	Myopathy, centronuclear, 3, 614408
MYH11	124.4	99%	98%	Aortic aneurysm, familial thoracic 4, 132900
MYH14	92.2	95%	84%	Deafness, autosomal dominant 4A, 600652 Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369
MYH2	126	100%	99%	Inclusion body myopathy-3, 605637
MYH3	104.2	99%	97%	Arthrogryposis, distal, type 2A, 193700 Arthrogryposis, distal, type 2B, 601680
MYH6	107.5	98%	93%	Atrial septal defect 3, 614089 Cardiomyopathy, dilated, 1EE, 613252 Cardiomyopathy, familial hypertrophic, 14, 613251 {Sick sinus syndrome 3}, 614090
MYH7	106.3	99%	95%	Cardiomyopathy,dilated, 1S,613426 Cardiomyopathy,hypertrophic,1, 192600 Left ventricular noncompaction 5, 613426 Liang distal myopathy,160500 Myopathy,myosin storage,autosomal dominant,608358 Myopathy,myosin storage,autosomal recessive,255160 Scapuloperoneal syndrome,myopathic type,181430

MYH8	128.8	100%	99%	Carney complex variant,608837 Trismus-pseudocamptodactyly syndrome,158300
MYH9	123.8	99%	95%	Deafness,autosomal dominant 17,603622 Epstein syndrome,153650 Fechtner syndrome,153640 Macrothrombocytopenia and progressive sensorineural deafness,600208 May-Hegglin anomaly, 155100 Sebastian syndrome,605249
MYL2	117.5	98%	83%	Cardiomyopathy, familial hypertrophic, 10, 608758
MYL3	119.4	100%	100%	Cardiomyopathy, familial hypertrophic, 8, 608751
MYLK	144	99%	99%	Aortic aneurysm, familial thoracic 7, 613780
MYLK2	95.4	99%	99%	Cardiomyopathy, hypertrophic, midventricular, digenic, 192600
MYO15A	102.3	94%	90%	Deafness, autosomal recessive 3, 600316
MYO1A	121.6	100%	99%	?deafness,autosomal dominant 48,607841
MYO1E	122.9	97%	93%	Glomerulosclerosis, focal segmental, 6, 614131
MYO3A	116.8	98%	87%	Deafness, autosomal recessive 30, 607101
MYO5A	122.5	98%	95%	Griscelli syndrome, type 1, 214450
MYO5B	130.8	97%	95%	Microvillus inclusion disease,251850
MYO6	86.8	96%	86%	Deafness,autosomal dominant 22,606346 Deafness,autosomal dominant 22,with hypertrophic cardiomyopathy,606346 Deafness,autosomal recessive 37,607821
MYO7A	127	98%	95%	Deafness, autosomal dominant 11, 601317 Deafness, autosomal recessive 2, 600060 Usher syndrome, type 1B, 276900
MYOC	171.9	100%	100%	Glaucoma 1A, primary open angle, 137750
MYOT	144.1	100%	96%	Muscular dystrophy, limb-girdle, type 1A, 159000 Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
MYOZ2	145.5	100%	100%	Cardiomyopathy, familial hypertrophic, 16, 613838
MYPN	147	99%	97%	Cardiomyopathy, dilated, 1KK, 615248 Cardiomyopathy, familial hypertrophic, 22, 615248 Cardiomyopathy, familial restrictive 4, 615248
NAA10	58	100%	91%	N-terminal acetyltransferase deficiency, 300855

NAGA	147.9	100%	100%	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241
NAGLU	99.9	91%	88%	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NAGS	58.3	90%	84%	N-acetylglutamate synthase deficiency, 237310
NALCN	136.2	99%	96%	?Neuroaxonal neurodegeneration, infantile, with facial dysmorphism, 615419
NANOS1	36.2	86%	70%	Spermatogenic failure 12,615413
NBAS	134.1	99%	97%	Infantile liver failure syndrome 2,616483 Short stature,optic nerve atrophy and Pelger-Huet anomaly,614800
NBEAL2	147.3	99%	97%	Gray platelet syndrome,139090
NBN	74.1	99%	96%	Aplastic anemia,609135 Leukemia,acute lymphoblastic,613065 Nijmegen breakage syndrome,251260
NCF1	16.5	23%	19%	Chronic granulomatous disease due to deficiency of NCF-1, 233700
NCF2	114.9	100%	98%	Chronic granulomatous disease due to deficiency of NCF-2, 233710
NCF4	148.2	100%	100%	Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III, 613960
NCOA4	107	92%	90%	?Thyroid cancer,nonmedullary,1},188550
NCSTN	111.4	100%	99%	Acne inversa, familial, 1, 142690
NDE1	77	100%	100%	Lissencephaly 4 (with microcephaly), 614019
NDN	77.2	89%	76%	Prader-Willi syndrome,176270
NDP	80.3	100%	100%	Exudative vitreoretinopathy, X-linked, 305390 Norrie disease, 310600
NDRG1	119.4	100%	97%	Charcot-Marie-Tooth disease, type 4D, 601455
NDUFA1	138.7	100%	97%	Mitochondrial complex I deficiency, 252010
NDUFA10	137.9	97%	95%	previous assignment to chr. 12 Leigh syndrome, 256000
NDUFA11	74.8	98%	91%	Mitochondrial complex I deficiency, 252010
NDUFA12	138	100%	100%	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000
NDUFA2	110.5	100%	100%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFA9	133	98%	96%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 -3
NDUFAF1	95.1	100%	99%	Mitochondrial complex I deficiency, 252010
NDUFAF2	43	85%	47%	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFAF3	106.1	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF4	80.8	99%	94%	Mitochondrial complex I deficiency, 252010

NDUFAF5	86.5	99%	91%	Mitochondrial complex I deficiency, 252010
NDUFAF6	83.1	100%	95%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFB3	26	96%	56%	Mitochondrial complex I deficiency, 252010
NDUFS1	134.3	97%	97%	Mitochondrial complex I deficiency, 252010
NDUFS2	102.5	100%	99%	Mitochondrial complex I deficiency, 252010
NDUFS3	121.8	90%	90%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
NDUFS4	155.3	100%	99%	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFS6	124.5	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFS7	106.9	100%	99%	Leigh syndrome, 256000
NDUFS8	125.5	100%	99%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFV1	132.1	100%	97%	Mitochondrial complex I deficiency, 252010
NDUFV2	60.6	83%	54%	Mitochondrial complex I deficiency, 252010
NEB	130.7	83%	81%	Nemaline myopathy 2, autosomal recessive, 256030
NEFL	131.5	99%	97%	Charcot-Marie-Tooth disease, type 1F,607734 Charcot-Marie-Tooth disease, type 2E, 607684
NEK1	113.4	98%	91%	Short rib-polydactyly syndrome, type IIA, 263520
NEU1	16.3	66%	31%	Sialidosis, type I, 256550 Sialidosis, type II, 256550
NEUROD1	153.6	100%	100%	Maturity-onset diabetes of the young 6, 606394 {Diabetes mellitus, noninsulin-dependent}, 125853
NEUROG3	105.7	100%	99%	Diarrhea 4,malabsorptive,congenital,610370
NEXN	66.2	87%	71%	Cardiomyopathy, dilated, 1CC, 613122 Cardiomyopathy, familial hypertrophic, 20, 613876
NF1	122.7	91%	86%	Neurofibromatosis,type 1,162200
NF2	96.7	100%	100%	Neurofibromatosis, type 2, 101000 Meningioma, NF2-related, somatic, 607174 Schwannomatosis, 162091
NFIX	139.2	98%	94%	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753
NFKB2	113.1	98%	93%	Immunodeficiency, common variable, 10, 615577
NFKBIA	87.2	96%	89%	Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency, 612132
NFU1	43.4	90%	75%	Multiple mitochondrial dysfunctions syndrome 1, 605711

NGF	227.1	100%	100%	Neuropathy, hereditary sensory and autonomic, type V, 608654
NHEJ1	74.4	100%	99%	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
NHLRC1	138.8	100%	100%	Epilepsy, progressive myoclonic 2B (Lafora), 254780
NHP2	66.3	100%	99%	Dyskeratosis congenita, autosomal recessive 2, 613987
NHS	83.7	93%	88%	Cataract 40, X-linked, 302200 Nance-Horan syndrome, 302350
NIN	150.7	99%	97%	Seckel syndrome 7,614851
NIPA1	134.2	100%	100%	Spastic paraplegia 6, autosomal dominant, 600363
NIPAL4	126.7	100%	93%	Ichthyosis,congenital,autosomal recessive 6,612281
NIPBL	111.4	96%	93%	Cornelia de Lange syndrome 1, 122470
NKX2-1	47.7	98%	80%	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 {Thyroid cancer,nonmedullary,1},188550
NKX2-5	70.2	100%	94%	Atrial septal defect 7, with or without AV conduction defects, 108900 Conotruncal heart malformations, variable, 217095 Hypoplastic left heart syndrome 2, 614435 Hypothyroidism, congenital nongoitrous, 5, 225250 Tetrology of Fallot, 187500 Ventricular septal defect 3, 614432
NKX2-6	96.8	100%	98%	Persistent truncus arteriosus, 217095
NKX3-2	31	77%	52%	Spondylo-megaepiphyseal-metaphyseal dysplasia,613330
NLGN4X	113.8	100%	95%	Mental retardation, X-linked, 300495 {Asperger syndrome susceptibility, X-linked 2}, 300497 {Autism susceptibility,X-linked 2},300495
NLRP12	142.7	99%	99%	Familial cold autoinflammatory syndrome 2, 611762
NLRP3	130.4	100%	100%	CINCA syndrome, 607115 Cold-induced autoinflammatory syndrome, familial, 120100 Muckle-Wells syndrome, 191900
NLRP7	116	99%	98%	Hydatidiform mole,recurrent, 1,231090
NME1	93.2	100%	98%	Neuroblastoma,256700
NME8	107.5	97%	89%	Ciliary dyskinesia, primary, 6, 610852
NMNAT1	128.7	100%	99%	Leber congenital amaurosis 9, 608553
NNT	131.9	98%	96%	Glucocorticoid deficiency 4, 614736

NOBOX	69.5	100%	100%	Premature ovarian failure 5,611548
NOD2	118.2	100%	99%	{Inflammatory bowel disease 1}, 266600
NODAL	130.9	100%	100%	Heterotaxy, visceral, 5, 270100
NOG	145.3	100%	99%	Brachydactyly, type B2, 611377 Multiple synostosis syndrome 1, 186500 Stapes ankylosis with broad thumb and toes, 184460 Symphalangism, proximal, 185800 Tarsal-carpal coalition syndrome, 186570
NOL3	79.9	95%	86%	Myoclonus, familial cortical, 614937
NOP10	159.4	100%	100%	Dyskeratosis congenita, autosomal recessive 1, 224230
NOP56	123	99%	96%	Spinocerebellar ataxia 36,614153
NOTCH1	121.3	99%	97%	Adams-Oliver syndrome 5,616028 Aortic valve disease,109730
NOTCH2	166.5	100%	98%	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome,102500
NOTCH3	99.6	92%	85%	?Myofibromatosis,infantile 2,615293 Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy,125310
NPC1	139.1	100%	97%	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220
NPC2	123.5	100%	100%	Niemann-pick disease, type C2, 607625
NPHP1	122	99%	94%	Joubert syndrome 4,609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1,266900
NPHP3	111.4	98%	91%	Meckel syndrome 7,267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1,208540
NPHP4	127.5	99%	97%	Nephronophthisis 4, 606966
NPHS1	92	100%	93%	Nephrotic syndrome, type 1, 256300
NPHS2	97.9	95%	76%	Nephrotic syndrome, type 2, 600995
NPM1	75	92%	78%	Lateral meningocele syndrome,130720
NPPA	78.9	100%	99%	Atrial fibrillation, familial, 6, 612201
NPR2	151.3	100%	99%	Acromesomelic dysplasia,Maroteaux type,602875 Epiphyseal chondrodysplasia,Miura type,615923 Short stature with nonspecific skeletal abnormalities,616255

NROB1	63.6	96%	91%	46XY sex reversal 2,dosage-sensitive,300018 Adrenal hypoplasia, congenital, with hypogonadotropic hypogonadism,300200
NROB2	82.5	100%	100%	Obesity,mild,early-onset,601665
NR2E3	95.9	100%	100%	Enhanced S-cone syndrome,268100 Retinitis pigmentosa 37,611131
NR2F1	182.2	99%	92%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NR3C1	134.7	100%	98%	Glucocorticoid resistance,615962
NR3C2	165.4	100%	98%	Pseudohypoaldosteronism type I, autosomal dominant, 177735
NR4A3	103.7	100%	92%	Chondrosarcoma,extraskeletal myxoid,612237
NR5A1	76.5	100%	100%	46XY sex reversal 3,612965 Aderenocortical insufficiency Premature ovarian failure 7,612964 Spermatogenic failure 8,613957
NRAS	179.8	100%	100%	?RAS-associated autoimmune lymphoproliferative syndrome type IV,somatic,611470 Colorectal cancer,somatic,114500 Epidermal nevus,somatic,162900 Melanocytic nevus syndrome,congenital,somatic,137550 Neurocutaneous melanosis,somatic,249400 Noonan syndrome 6,613224 Schimmelpenning-Feuerstein-Mims syndrome,somatic mosaic,163200 Thyroid carcinoma,follicular,somatic,188470
NRL	61.1	98%	86%	Retinal degeneration, autosomal recessive, clumped pigment type Retinitis pigmentosa 27, 613750
NRXN1	160	99%	96%	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332
NSD1	142.2	100%	100%	Beckwith-Wiedemann syndrome, 130650 Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550
NSDHL	119.2	99%	93%	CHILD syndrome, 308050 CK syndrome, 300831
NSMF	86.6	96%	96%	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
NSUN2	108.9	95%	90%	Mental retardation, autosomal recessive 5, 611091
NT5C2	129.2	97%	96%	Spastic paraplegia 45,autosomal recessive,613162
NT5C3A	58.9	83%	73%	Anemia, hemolytic, due to UMPH1 deficiency, 266120

NT5E	174.7	100%	99%	Calcification of joints and arteries, 211800
NTF4	42.2	90%	77%	Glaucoma 1,open angle, 10,613100
NTRK1	115.4	97%	93%	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma, familial, 155240
NTRK2	173.4	100%	99%	?Obesity,hyperphagia,and developmental delay,613886
NUBPL	90.3	82%	82%	Mitochondrial complex I deficiency, 252010
NUMA1	104.9	100%	99%	Leukemia,acute promyelocytic,somatic,612376
NUP214	150.8	99%	99%	Leukemia,acute myeloid,somatic,601626 Leukemia,T-cell acute lymphoblastic,somatic,613065
NUP62	98.5	100%	100%	Striatonigral degeneration, infantile, 271930
NYX	50.7	96%	95%	Night blindness, congenital stationary (complete), 1A, X-linked, 310500
OAT	73.6	68%	61%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OBSL1	117.5	98%	95%	3-M syndrome 2,612921
OCA2	126.1	97%	95%	Albinism brown oculocutaneous,203200 [Skin/hair/eye pigmentation 1],227220
OCLN	164.9	100%	99%	Band-like calcification with simplified gyration and polymicrogyria, 251290
OCRL	87	97%	88%	Dent disease 2,300555 Lowe syndrome, 309000
OFD1	29.6	74%	53%	?Retinitis pigmentosa 23,300424 Joubert syndrome 10,300804 Oral-facial-digital syndrome 1, 311200 Simpson-Golabi-Behmel syndrome type 2,300209
OGG1	117.5	100%	98%	Renal cell carcinoma, clear cell, somatic, 144700
OPA1	115.4	98%	89%	Optic atrophy 1, 165500
OPA3	89.2	99%	93%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPHN1	63.9	98%	94%	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
OPLAH	86.1	100%	97%	5-oxoprolinase deficiency, 260005
OPN1LW	48.9	66%	44%	Blue cone monochromacy, 303700 Colorblindness, protan, 303900
OPN1MW	64.3	65%	50%	Blue cone monochromacy, 303700 Colorblindness, deutan, 303800
OPN1SW	120.3	100%	100%	Colorblindness,tritan,190900



OPTN	107.8	100%	99%	Amyotrophic lateral sclerosis 12, 613435 Glaucoma 1, open angle, E, 137760 {Glaucoma, normal tension, susceptibility to}, 606657
ORAI1	177.8	90%	88%	Immunodeficiency 9,612782 Myopathy,tubular aggregate, 2,615883
ORC1	104.1	100%	97%	Meier-Gorlin syndrome 1, 224690
ORC4	61.5	87%	77%	Meier-Gorlin syndrome 2,613800
ORC6	131.2	100%	100%	Meier-Gorlin syndrome 3,613803
OSMR	135	99%	99%	Amyloidosis primary localized cutaneous 1,105250
OSTM1	73.9	90%	88%	Osteopetrosis,autosomal recessive 5,259720
OTC	87	100%	99%	CGD Ornithine transcarbamyase deficiency, 311250
OTOA	112	98%	95%	Deafness, autosomal recessive 22, 607039
OTOF	121.5	100%	99%	Deafness, autosomal recessive 9, 601071
OTOG	124.6	100%	98%	Deafness, autosomal recessive 18B, 614945
OTOGL	123.3	96%	93%	Deafness, autosomal recessive 84B, 614944
OTX2	116.2	100%	97%	Microphthalmia, syndromic 5
OXCT1	105.7	100%	96%	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050
P2RX1	114.5	100%	98%	Bleeding disorder due to P2RX1 defect,somatic,609821
P2RX2	110.6	98%	95%	Deafness, autosomal dominant 41, 608224
P2RY12	168	100%	100%	Bleeding disorder, platelet-type 8,609821
PABPN1	70.5	63%	60%	Oculopharyngeal muscular dystrophy, 164300
PACS1	108.7	95%	94%	Mental retardation, autosomal dominant 17, 615009
PAFAH1B1	97.6	89%	80%	Lissencephaly, 607432 Subcortical laminar heterotopia, 607432
PAH	170.6	100%	100%	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600
PAK3	53	96%	80%	Mental retardation, X-linked 30/47, 300558
PALB2	148.7	100%	100%	Fanconi anemia, complementation group N, 610832 {Breast cancer, susceptibility to}, 114480 {Pancreatic cancer, susceptibility to, 3}, 613348
PANK2	142.9	95%	86%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PAPSS2	94.7	99%	97%	Bracyolmia 4 with mild epiphyseal and metaphyseal changes,612847
PARK2	116	98%	96%	Lung cancer

PARK7	80.1	100%	95%	Parkinson disease 7,autosomal recessive early-onset,606324
PAX2	148.6	100%	99%	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330 Renal hypoplasia, isolated, 191830
PAX3	108.8	100%	99%	Craniofacial-deafness-hand syndrome,122880 Rhabdomyosarcoma 2,alveolar,268220 Waardenburg syndrome,type 1,193500 Waardenburg syndrome,type 3,148820
PAX4	84.5	100%	97%	Diabetes mellitus,type 2,125853 Maturity-onset diabetes of the young,type IX,612225 {Diabetes mellitus,ketosis-prone,susceptibility to},612227
PAX6	129.3	100%	100%	?Morning glory disc anomaly,120430 Aniridia,106210 Cataract with late-onset corneal dystrophy,106210 Coloboma of optic nerve,120430 Coloboma, ocular,120200 Foveal hypoplasia 1,136520 Keratitis,148190 Optic nerve hypoplasia,165550 Peters Anomaly,604229
PAX8	74.5	100%	99%	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700
PAX9	228.6	99%	93%	Tooth agenesis selective 3,604625
PC	134.1	98%	94%	Pyruvate carboxylase deficiency, 266150
PCBD1	115.9	100%	99%	Hyperphenylalaninemia, BH4-deficient, D,264070
PCCA	106.9	99%	90%	Propionicacidemia, 606054
PCCB	141.5	96%	95%	Propionicacidemia, 606054
PCDH15	164.2	99%	98%	Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1F, 602083
PCDH19	133.3	100%	95%	Epileptic encephalopathy, early infantile, 9, 300088
PCM1	126	98%	94%	No OMIM phenotype Schizophrenia (Kamiya (2008) Arch Gen Psychiatry 65,996) Breast cancer (Guan (2015) Fam Cancer 14,9)
PCNT	108.1	97%	93%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720

PCSK1	148.2	100%	99%	Obesity with impaired prohormone processing,60955 {Obesity,susceptibility to,BMIQ12},612362
PCSK9	85.5	97%	88%	Hypercholesterolemia,familial,3,603776 {Low density lipoprotein cholesterol level QTL 1},603776
PCYT1A	123.1	97%	94%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDCD10	94	98%	82%	Cerebral cavernous malformations 3,603285
PDE11A	143.3	100%	100%	Pigmented nodular adrenocortical disease,primary,2,610475
PDE4D	112.7	97%	95%	Acrocydostosis 2 with or without hormone resistance, 614613 {Stroke, susceptibility to, 1}, 606799
PDE6A	120.3	100%	99%	Retinitis pigmentosa 43, 613810
PDE6B	136.8	100%	99%	Night blindness, congenital stationary, autosomal dominant 2, 163500 Retinitis pigmentosa-40, 613801
PDE6C	131.5	96%	94%	Cone dystrophy 4, 613093
PDE6G	85.3	99%	94%	Retinitis pigmentosa 57, 613582
PDE6H	58.1	51%	48%	Achromatopsia 6, 610024 Retinal cone dystrophy 3, 610024
PDE8B	104.1	100%	91%	Pigmented nodular adrenocortical disease, primary, 3, 614190 Striatal degeneration, autosomal dominant, 609161
PDGFB	87	100%	100%	Basal ganglia calcification,idiopathic,5,615483 Dermatofibrosarcoma protuberans,607907 Meningioma, SIS-related,607174
PDGFRA	145.2	100%	99%	Gastrointestinal stromal tumor,somatic,606764 Hypereosinophilic syndrome,idiopathic,resistant to imatinib,607685
PDGFRB	135.4	98%	96%	Basal ganglia calcification idiopathic 4,615007 Myofibromatosis, infantile, 1, 228550 Myeloproliferative disorder with eosinophilia, 131440
PDGFRL	130	100%	99%	Colorectal cancer,somatic,114500 Hepatocellular cancer,somatic,114550
PDHA1	72.7	94%	82%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	127.9	97%	94%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDP1	173.3	100%	100%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	118.2	92%	83%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	109	97%	92%	Coenzyme Q10 deficiency, primary, 3, 614652

PDX1	29.1	82%	68%	MODY,type IV,606392 Pancreatic agenesis 1,260370 {Diabetes mellitus,type II,susceptibility to},125853
PDYN	99.6	100%	100%	Spinocerebellar ataxia 23, 610245
PDZD7	77.7	100%	89%	{Retinal disease in Usher syndrome type IIA, modifier of}, 276901 Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472
PEPD	99.8	100%	97%	Prolidase deficiency, 170100
PER2	85.3	100%	98%	Advanced sleep phase syndrome,familial,1,604348
PET100	110.5	89%	72%	Mitochondrial complex IV deficiency, 220110
PEX1	106.9	97%	97%	Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	100.3	97%	87%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX11B	107.2	100%	100%	Peroxisome biogenesis disorder 14B, 614920
PEX12	138.3	100%	100%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	194.6	100%	100%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	109.8	100%	95%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	114	93%	91%	Peroxisome biogenesis disorder 8A, (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	100	100%	100%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	147.6	100%	100%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	73.3	100%	98%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	97	100%	99%	Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	100.1	98%	94%	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370
PEX6	75.8	86%	82%	Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863
PEX7	121.4	91%	82%	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PFKM	149.2	100%	100%	Glycogen storage disease VII, 232800

PFN1	149.2	100%	100%	Amyotrophic lateral sclerosis 18, 614808
PGAM2	138.7	100%	99%	Glycogen storage disease X, 261670
PGAP2	156.2	100%	100%	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGAP3	113.5	97%	94%	Hyperphosphatasia with mental retardation syndrome 4, 615716
PGK1	35.1	80%	65%	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	120.8	100%	99%	Glycogen storage disease XIV, 612934 Congenital disorder of glycosylation, type It, 614921
PHEX	78.8	95%	93%	Hypophosphatemic rickets, X-linked dominant, 307800
PHF6	46.3	87%	81%	Borjeson-Forssman-Lehmann syndrome, 301900
PHF8	65.5	98%	92%	Mental retardation syndrome, X-linked, Siderius type, 300263
PHGDH	119.4	100%	99%	Phosphoglycerate dehydrogenase deficiency, 601815
PHKA1	72.8	94%	90%	? Muscle glycogenosis, 300559
PHKA2	71	100%	98%	Glycogen storage disease, type IXa1, 306000 Glycogen storage disease, type IXa2, 306000
PHKB	133.9	100%	99%	Phosphorylase kinase deficiency of liver and muscle,autosomal recessive,261750
PHKG2	138.7	100%	100%	Cirrhosis due to liver phosphorylase kinase deficiency Glycogen storage disease Ixc,613027
PHOX2A	28.6	53%	22%	Fibrosis of extraocular muscles,congenital,2,602078
PHOX2B	85.6	91%	87%	Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, 209880 {Neuroblastoma, susceptibility to, 2}, 613013 Neuroblastoma with Hirschsprung disease, 613013
PHYH	87.5	97%	92%	Refsum disease, 266500
PICALM	108.5	99%	91%	Leukemia,acute myeloid,somatic,601626
PIEZO1	125.8	98%	96%	Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema ,194380
PIEZO2	121.7	99%	98%	?Marden-Walker syndrome,248700 Arthrogyrosis,distal,type 3,114300 Arthrogyrosis,distal,type 5,108145
PIGA	56.5	76%	73%	Multiple congenital anomalies-hypotonia-seizures syndrome 2,300868 Paroxysmal nocturnal hemoglobinuria,somatic,300818
PIGL	111.9	100%	99%	CHIME syndrome, 280000
PIGM	138.4	100%	100%	Glycosylphosphatidylinositol deficiency, 610293
PIGN	116.9	96%	90%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	113.4	100%	98%	Hyperphosphatasia with mental retardation syndrome 2, 614749

PIGV	149.2	100%	100%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIK3CA	125.3	99%	98%	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
PIK3CD	119	98%	96%	Immunodeficiency 14, 615513
PIK3R1	123.8	100%	99%	Agammaglobulinemia 7, autosomal recessive, 615214
PIK3R2	74.1	87%	86%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome, 603387
PIK3R5	95.4	100%	99%	Ataxia-oculomotor apraxia 3, 615217
PIKFYVE	137.7	99%	98%	Corneal fleck dystrophy, 121850
PINK1	75.6	93%	86%	Parkinson disease 6, early onset, 605909
PIP5K1C	96.9	96%	95%	Lethal congenital contractural syndrome 3, 611369
PITPNM3	94.2	99%	97%	Cone-rod dystrophy 5,600977
PITX1	119	92%	83%	Clubfoot,congenital,with or without deficiency of long bones and/or mirror-image polydactyly,119800 Liebenberg syndrome,186550
PITX2	128.1	96%	93%	Axenfeld-Rieger syndrome type 1,180500 Iridogoniodysgenesis,type 2,137600 Peters anomaly,604229 Ring dermoid of cornea,180550
PITX3	56	100%	92%	Anterior segment mesenchymal dysgenesis, 107250
PKD1	23.2	39%	30%	Polycystic kidney disease, adult type I, 173900
PKD2	95.3	87%	85%	Polycystic kidney disease 2, 613095
PKHD1	154	100%	99%	Polycystic kidney and hepatic disease, 263200
PKLR	152.5	100%	99%	Adenosine triphosphate, elevated, of erythrocytes, 102900 Pyruvate kinase deficiency, 266200
PKP1	108.1	100%	99%	Ectodermal dysplasia/skin fragility syndrome,604536
PKP2	90.4	97%	86%	Arrhythmogenic right ventricular dysplasia 9, 609040
PLA2G4A	135.8	100%	99%	Phospholipase A2,group IV A,deficiency of
PLA2G5	117.5	100%	100%	Fleck retina, familial benign, 228980
PLA2G6	108.7	100%	98%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, 612953

PLA2G7	123.9	100%	98%	Asthma, susceptibility to, 600807 Atopy, susceptibility to, 147050 Platelet-activating factor acetylhydrolase deficiency, 614278
PLAG1	224.4	100%	100%	Adenomas, salivary gland pleomorphic, somatic, 181030
PLAU	104.4	100%	98%	Quebec platelet disorder, 601709 {Alzheimer disease, late-onset, susceptibility to}, 104300
PLCB1	153.4	100%	100%	Epileptic encephalopathy, early infantile, 12, 613722
PLCB4	125.6	100%	97%	Auriculocondylar syndrome 2, 614669
PLCD1	101.4	98%	94%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCE1	145.3	98%	98%	Nephrotic syndrome, type 3, 610725
PLCG2	116.1	99%	98%	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468
PLEC	87.5	98%	95%	?Epidermolysis bullosa simplex with nail dystrophy, 616487 Epidermolysis bullosa simplex, Ogna type, 131950 Epidermolysis bullosa simplex with muscular dystrophy, 226670 Epidermolysis bullosa simplex with pyloric atresia, 612138 Muscular dystrophy, limb-girdle, type 2Q, 613723
PLEKHG5	79.2	92%	85%	Charcot-Marie-Tooth disease, recessive intermediate C, 615376 Spinal muscular atrophy, distal, autosomal recessive, 4, 611067
PLEKHM1	51.9	84%	75%	Osteopetrosis, autosomal recessive 6, 611497
PLG	114	87%	87%	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PLIN1	74.5	92%	81%	Lipodystrophy, familial partial, type 4, 613877
PLN	163.8	100%	100%	Cardiomyopathy, dilated, 1P, 609909 Cardiomyopathy, familial hypertrophic, 18, 613874
PLOD1	128.3	100%	98%	Ehlers-Danlos syndrome, type VI, 225400
PLOD2	113.1	95%	89%	Bruck syndrome 2, 609220
PLOD3	103.3	100%	99%	Lysyl hydroxylase 3 deficiency, 612394
PLP1	91.3	100%	99%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PLS3	102.3	98%	93%	Bone mineral density QTL18, osteoporosis, 300910
PML	105.1	100%	96%	Leukemia, acute promyelocytic, PML/RARA type
PMM2	163.1	100%	98%	Congenital disorder of glycosylation, type Ia, 212065

PMP22	93.2	87%	84%	Charcot-Marie-Tooth disease, type 1A, 118220 Charcot-Marie-Tooth disease, type 1E, 118300 Dejerine-Sottas disease, 145900 Neuropathy, inflammatory demyelinating, 139393 Neuropathy, recurrent, with pressure palsies, 162500 Roussy-Levy syndrome, 180800
PMS2	79.5	82%	79%	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 Mismatch repair cancer syndrome, 276300
PNKP	78.6	100%	95%	Epileptic encephalopathy, early infantile, 10, 613402
PNP	125.9	100%	100%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA1	163.2	99%	97%	Ichthyosis congenital autosomal recessive 10, 615024
PNPLA2	104.4	100%	96%	Neutral lipid storage disease with myopathy, 610717
PNPLA6	111.2	99%	97%	Spastic paraplegia 39, autosomal recessive, 612020
PNPO	63.8	100%	97%	Pyridoxamine 5-phosphate oxidase deficiency, 610090
PNPT1	48.8	94%	77%	Combined oxidative phosphorylation deficiency 13, 614932
POC1A	132.6	100%	100%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POF1B	55	78%	70%	Premature ovarian failure 2B, 300604
POFUT1	130.1	98%	92%	Dowling-Degos disease 2, 615327
POGLUT1	124.6	100%	98%	Dowling-Degos disease 4, 615696
POLD1	89	94%	92%	{Colorectal cancer, susceptibility to, 10}, 612591 Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381
POLE	130.6	99%	98%	{Colorectal cancer, susceptibility to, 12}, 615083 FILS syndrome, 615139
POLG	107.7	100%	98%	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Progressive external ophthalmoplegia, autosomal dominant, 1, 157640 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
POLG2	143.9	99%	97%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131
POLH	149	100%	99%	Xeroderma pigmentosum variant type, 278750
POLR1C	107.8	97%	94%	Leukodystrophy, hypomyelinating, 11, 616494 Treacher Collins syndrome 3, 248390
POLR1D	171.4	100%	100%	Treacher Collins syndrome 2, 613717
POLR3A	140.2	100%	99%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic



				hypogonadism, 607694
POLR3B	143.9	100%	99%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMC	69.2	100%	99%	Obesity adrenal insufficiency and red hair due to POMC deficiency,609734 {Obesity,early-onset,susceptibility to},601665
POMGNT1	107	100%	95%	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
POMGNT2	259.3	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies,type A,8),614830
POMP	138.6	86%	85%	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma,601952
POMT1	152.8	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	101.6	97%	95%	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	139.2	100%	97%	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis,201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase,613571
PORCN	79.8	100%	96%	Focal dermal hypoplasia, 305600
POU1F1	118.2	99%	92%	Pituitary hormone deficiency, combined, 1, 613038
POU3F4	90.3	100%	100%	Deafness, X-linked 2, 304400
POU4F3	239.8	100%	100%	Deafness, autosomal dominant 15, 602459
PPARG	147.9	100%	99%	Carotid intimal medial thickness 1,609338 Insulin resistance,severe,digenic,604367 Lipodystrophy,familial partial,type 3,604367 Obesity,severe,601665 [Obesity,resistance to] {Diabetes,type 2},125853
PPIB	121.9	100%	100%	Osteogenesis imperfecta, type IX,259440
PPM1D	157.1	100%	99%	Breast cancer,114480
PPM1K	163.9	100%	99%	Maple syrup urine disease, mild variant, 615135
PPOX	92.1	100%	97%	Porphyria variegata, 176200
PPP1R3A	160.1	99%	97%	Insulin resistance,severe,digenic,604367
PPP2R1B	157.6	100%	99%	Lung cancer,211980

PPP2R2B	149.7	99%	98%	Spinocerebellar ataxia 12,604326
PPT1	150.6	100%	100%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	97	97%	94%	Renpenning syndrome, 309500
PRCC	110.4	100%	80%	Renal cell carcinoma,papillary,605074
PRCD	89.1	100%	100%	Retinitis pigmentosa 36, 610599
PRDM16	131.7	99%	97%	Left ventricular noncompaction 8, 615373 Cardiomyopathy, dilated, 1LL, 615373
PRDM5	134.3	100%	96%	Brittle cornea syndrome 2,614170
PRF1	113.4	100%	99%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027
PRG4	124.7	98%	87%	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome,208250
PRICKLE1	118.1	100%	100%	Epilepsy, progressive myoclonic 1B, 612437
PRICKLE2	114.2	100%	100%	Epilepsy, progressive myoclonic 5,613832
PRIMPOL	102.1	96%	92%	Myopia 22,autosomal dominant,615420
PRKAG2	118.2	96%	89%	Cardiomyopathy, familial hypertrophic 6, 600858 Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200
PRKAR1A	90.8	98%	90%	Acrodysostosis 1, with or without hormone resistance,101800 Adrenocortical tumor, somatic Carney complex, type 1, 160980 Myxoma, intracardiac, 255960 Pigmented nodular adrenocortical disease, primary, 1, 610489
PRKCA	170.8	100%	100%	Pituitary tumor,invasive
PRKCG	103.6	96%	91%	Spinocerebellar ataxia 14, 605361
PRKCSH	111	95%	90%	Polycystic liver disease,174050
PRKG1	115.9	96%	90%	Aortic aneurysm, familial thoracic 8, 615436
PRKRA	140.7	96%	95%	Dystonia 16, 612067
PRLR	139	100%	100%	?Hyperprolactinemia,615555 Multiple fibroadenomas of the breast,615554
PRNP	170.2	100%	100%	Cerebral amyloid angiopathy,PRNP related,137440 Creutzfeldt-Jakob disease,123400 Gerstmann-Straussler disease,137440 Huntington disease-like 1,603218 Insomnia,fatal familial,600072

				Prion disease with protracted course,606688
PROC	116.9	99%	95%	Thrombophilia due to protein C deficiency, autosomal dominant, 176860 Thrombophilia due to protein C deficiency, autosomal recessive, 612304
PRODH	68.5	84%	77%	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850
PROK2	86.9	99%	99%	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	329.1	100%	100%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROM1	112.9	96%	93%	Cone-rod dystrophy 12, 612657 Macular dystrophy, retinal, 2, 608051 Retinitis pigmentosa 41, 612095 Stargardt disease 4, 603786
PROP1	78.7	90%	86%	Pituitary hormone deficiency, combined, 2,262600
PROS1	77.7	93%	89%	Thrombophilia due to protein S deficiency, autosomal dominant, 612336 Thrombophilia due to protein S deficiency, autosomal recessive,614514
PRPF3	73.3	98%	91%	Retinitis pigmentosa 18, 601414
PRPF31	99.1	100%	93%	Retinitis pigmentosa 11, 600138
PRPF6	114.1	100%	99%	Retinitis pigmentosa 60, 613983
PRPF8	132.4	100%	99%	Retinitis pigmentosa 13, 600059
PRPH2	209.9	100%	98%	Foveomacular dystrophy, adult-onset, with choroidal neovascularization, 608161 Retinitis pigmentosa 7, 608133 Retinitis punctata albescens, 136880 Macular dystrophy, patterned, 169150 Macular dystrophy, vitelliform, 608161
PRPS1	113.7	100%	100%	Arts syndrome, 301835 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Deafness, X-linked 1, 304500 Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661
PRRT2	62.2	100%	99%	Convulsions,familial infantile,with paroxysmal choreoathetosis,602066 Episodic kinesigenic dyskinesia 1, 128200

				Seizures,benign familial infantile, 2,605751
PRRX1	94.8	100%	100%	Agnathia-otocephaly complex,202650
PRSS1	191	100%	100%	Pancreatitis,hereditary,167800 Trypsinogen deficiency,614044
PRSS12	154.4	98%	96%	Mental retardation, autosomal recessive 1, 249500
PRSS56	38.9	93%	73%	Microphthalmia, isolated 6, 613517
PRX	92	100%	99%	Charcot-Marie-Tooth disease,type 4F,614895 Dejerine-Sottas disease, autosomal recessive, 145900
PSAP	111.2	100%	97%	Combined SAP deficiency, 611721 Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PSAT1	44.3	90%	70%	Phosphoserine aminotransferase deficiency, 610992
PSEN1	140.9	98%	94%	Acne inversa, familial, 3, 613737 Alzheimer disease, type 3, 607822 Cardiomyopathy, dilated, 1U, 613694 Dementia,frontotemporal,600274 Pick disease,172700
PSEN2	123.4	100%	100%	Alzheimer disease-4,606889 Cardiomyopathy,dilated,1V,613697
PSENE1	83.2	100%	99%	Acne inversa, familial, 2, 613736
PSMB8	13.2	50%	16%	Autoinflammation, lipodystrophy, and dermatosis syndrome, 256040
PSMC3IP	104.3	100%	99%	Ovarian dysgenesis 3,614324
PSPH	122.1	100%	96%	Phosphoserine phosphatase deficiency, 614023
PSTPIP1	76.9	96%	86%	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416
PTCH1	116.3	98%	96%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly-7, 610828
PTCH2	108.1	99%	97%	Basal cell carcinoma somatic,605462 Basal cell nevus syndrome,109400 Medulloblastoma,155255
PTDSS1	130.4	100%	99%	Lenz-Majewski hyperostotic dwarfism, 151050

PTEN	142.2	100%	100%	Bannayan-Riley-Ruvalcaba syndrome,153480 Cowden syndrome 1,158350 Endometrial carcinoma, somatic,608089 Lhermitte-Duclos syndrome,158350 Macrocephaly/Autism syndrome,605309 Malignant melanoma,somatic,155600 PTEN hamartoma tumor syndrome Squamous cell carcinoma,head and neck,somatic,275355 VATER association with macrocephaly and ventriculomegaly,276950 {Glioma susceptibility 2},613028 {Meningioma},607174 {Prostate cancer,somatic},176807
PTF1A	61.3	79%	72%	Pancreatic agenesis 2,615935 Pancreatic and cerebellar agenesis,609069
PTGIS	115.4	94%	93%	Hypertension, essential, 145500
PTH	109	100%	95%	Hypoparathyroidism,146200
PTH1R	92.9	99%	97%	Chondrodysplasia, Blomstrand type, 215045 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Metaphyseal chondrodysplasia, Murk-Jansen type, 156400
PTHLH	126.8	99%	83%	Brachydactyly type E2,613382 Humoral hypercalcemia of malignancy
PTPN11	93	96%	89%	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
PTPN12	145	96%	95%	Colon cancer,somatic,114500
PTPN14	161.1	99%	97%	Choanal atresia and lymphedema,613611
PTPRC	99.2	94%	84%	{Hepatitis C virus, susceptibility to}, 609532 Severe combined immunodeficiency,T cell-negative,B-cell/natural killer-cell positive,608971
PTPRJ	170.5	97%	95%	Colon cancer, somatic, 114500
PTPRO	160.4	100%	99%	Nephrotic syndrome, type 6, 614196
PTPRQ	105	92%	87%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
PTRF	115.2	100%	97%	Lipodystrophy, congenital generalized, type 4, 613327
PTS	113.3	96%	86%	Hyperphenylalaninemia, BH4-deficient, A, 261640

PUF60	140.1	99%	97%	Verheij syndrome, 615583
PUS1	130	99%	97%	Mitochondrial myopathy and sideroblastic anemia 1, 600462
PVRL1	134.6	100%	99%	Cleft lip/palate-ectodermal dysplasia syndrome,225060 Orofacial cleft 7,225060
PVRL4	120	100%	100%	Ectodermal dysplasia-syndactyly syndrome 1,613573
PYCR1	80.4	100%	91%	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
PYGL	160.7	100%	100%	Glycogen storage disease VI, 232700
PYGM	135.5	100%	99%	McArdle disease, 232600
QARS	145.6	100%	99%	Microcephaly, progressive,seizures, and cerebral and cerebellar atrophy, 615760
QDPR	73.4	100%	100%	Hyperphenylalaninemia, BH4-deficient, C, 261630
RAB18	88.4	97%	78%	Warburg micro syndrome 3, 614222
RAB23	103.9	100%	99%	Carpenter syndrome,201000
RAB27A	177.8	100%	100%	Griscelli syndrome, type 2, 607624
RAB28	50.6	93%	77%	Cone-rod dystrophy 18, 615374
RAB33B	229.3	100%	100%	Smith-McCort dysplasia 2,615222
RAB39B	89.2	99%	95%	Mental retardation, X-linked 72, 300271
RAB3GAP1	127.3	99%	99%	Warburg micro syndrome 1, 600118
RAB3GAP2	94	98%	93%	Martsolf syndrome, 212720 Warburg micro syndrome 2, 614225
RAB40AL	100	100%	100%	Mental retardation, X-linked, syndromic, Martin-Probst type, 300519
RAB7A	151.6	100%	100%	Charcot-Marie-Tooth disease,type 2B, 600882
RAC2	116.7	100%	100%	Neutrophil immunodeficiency syndrome, 608203
RAD21	89.1	99%	96%	Cornelia de Lange syndrome 4, 614701
RAD50	91.4	93%	86%	Nijmegen breakage syndrome-like disorder, 613078
RAD51	130.1	89%	89%	Mirror movements 2,614508 {Breast cancer,susceptibility to},114480
RAD51C	148.2	100%	100%	Fanconi anemia, complementation group O, 613390 {Breast-ovarian cancer, familial, susceptibility to, 3}, 613399
RAD54B	97.1	96%	88%	Colon cancer,somatic,114500 Lymphoma,non-Hodgkin,somatic,605027
RAD54L	112.6	100%	99%	Adenocarcinoma,colonic,somatic Lymphoma,non-Hodgkin,somatic,605027 {Breast cancer,invasive ductal},114480

RAF1	118.6	100%	99%	Cardiomyopathy,dilated,1NN,615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553
RAG1	203.2	100%	100%	Severe combined immunodeficiency, B cell-negative, 601457
RAG2	250.1	100%	99%	Severe combined immunodeficiency, B cell-negative, 601457
RAI1	113.3	100%	99%	Immunodeficiency 9, 612782 Smith-Magenis syndrome, 182290
RAP1GDS1	92.2	98%	92%	Lymphocytic leukemia,acute T-cell
RAPSN	116.4	94%	93%	Fetal akinesia deformation sequence, 208150 Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931 Myasthenic syndrome, congenital, associated with facial dysmorphism and acetylcholine receptor deficiency, 608931
RARB	124.5	100%	100%	Microphthalmia, syndromic 12, 615524
RARS2	111.7	100%	99%	Pontocerebellar hypoplasia, type 6, 611523
RASA1	98.9	96%	86%	Basal cell carcinoma,somatic,605462 Capillary malformation-arteriovenous malformation,608354 Parkes Weber syndrome,608355
RAX	64	94%	72%	Microphthalmia,isolated 3,611038
RAX2	45.5	90%	69%	Cone-rod dystrophy 11, 610381 Macular degeneration, age-related, 6,613757
RB1	77.5	91%	76%	Bladder cancer, somatic, 109800 Osteosarcoma, somatic, 259500 Retinoblastoma, 180200 Retinoblastoma, trilateral, 180200 Small cell cancer of the lung, somatic, 182280
RB1CC1	107.9	96%	90%	Breast cancer,somatic,114480
RBBP8	93.1	100%	95%	Jawad syndrome,251255 Pancreatic carcinoma,somatic Seckel syndrome 2,606744
RBM10	67.6	99%	94%	TARP syndrome, 311900
RBM20	161.8	99%	95%	Cardiomyopathy, dilated, 1DD, 613172
RBM28	128.8	100%	100%	Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RBM8A	96.4	98%	94%	Thrombocytopenia-absent radius syndrome,274000
RBP4	87	93%	91%	Microphthalmia,isolated,with coloboma 10,616428

				Retinol dystrophy iris coloboma and comedogenic acne syndrome,615147
RBPJ	75.6	91%	83%	Adams-Oliver syndrome 3,614814
RD3	136.9	100%	100%	Leber congenital amaurosis 12, 610612
RDH12	87.6	97%	83%	Leber congenital amaurosis 13, 612712
RDH5	144	100%	100%	Fundus albipunctatus, 136880
RDX	36.8	74%	58%	Deafness, autosomal recessive 24, 611022
RECQL4	121.3	97%	95%	Baller-Gerold syndrome, 218600 RAPADILINO syndrome, 266280 Rothmund-Thomson syndrome, 268400
REEP1	96.5	95%	95%	Neuronopathy, distal hereditary motor, type VB, 614751 Spastic paraplegia 31, autosomal dominant, 610250
RELN	158.4	100%	99%	Lissencephaly 2 (Norman-Roberts type), 257320
REN	140.2	100%	99%	Hyperuricemic nephropathy,familial juvenile 2,613092 Renal tubular dysgenesis,267430 [Hyperproreninemia]
RET	137.7	99%	97%	Central hypoventilation syndrome, congenital, 209880 Medullary thyroid carcinoma, 155240 Multiple endocrine neoplasia IIA, 171400 Multiple endocrine neoplasia IIB, 162300 Pheochromocytoma, 171300 {Hirschsprung disease, susceptibility to},142623
RFT1	93.3	100%	91%	Congenital disorder of glycosylation, type In, 612015
RFX5	108.6	96%	94%	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFX6	161.1	100%	100%	Mitchell-Riley syndrome,615710
RFXANK	95.6	100%	100%	MHC class II deficiency, complementation group B, 209920
RFXAP	95.3	92%	90%	Bare lymphocyte syndrome, type II, complementation group D, 209920
RGR	123.5	100%	100%	Retinitis pigmentosa 44, 613769
RGS9	94.3	100%	99%	Bradyopsia, 608415
RGS9BP	59	100%	97%	Bradyopsia, 608415
RHAG	153.2	100%	100%	Anemia,hemolytic,Rh-null,regulator type,268150 Overhydrated hereditary stomatocytosis,185000 Rh-mod syndrome
RHBDF2	95.5	98%	95%	Tylosis with esophageal cancer,148500



RHCE	169.9	99%	99%	Rh-null disease,amorph type [Blood group,Rhesus],111690
RHO	224.6	100%	100%	Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 Retinitis punctata albescens, 136880
RIMS1	113.5	96%	93%	Cone-rod dystrophy 7, 603649
RIN2	110.7	99%	98%	Macrocephaly alopecia cutis laxa and scoliosis,613075
RIPK4	127.7	100%	98%	Popliteal pterygium syndrome 2, lethal type,263650
RIT1	154.3	100%	100%	Noonan syndrome 8, 615355
RLBP1	127.9	100%	100%	Bothnia retinal dystrophy, 607475 Fundus albipunctatus, 136880 Newfoundland rod-cone dystrophy, 607476 Retinitis punctata albescens, 136880
RMND1	131.5	99%	90%	Combined oxidative phosphorylation deficiency 11, 614922
RMRP				Anauxetic dysplasia,607095 Cartilage-hair hypoplasia,250250 Metaphyseal dysplasia without hypotrichosis,250460
RNASEH2A	124.1	100%	95%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	106.7	88%	76%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	175.9	100%	99%	Aicardi-Goutieres syndrome 3, 610329
RNASEL	136.4	100%	98%	Prostate cancer 1,601518
RNASET2	74.6	83%	80%	Leukoencephalopathy, cystic, without megalencephaly, 612951
RNF135	75.6	93%	89%	Macrocephaly,macrosomia,facial dysmorphism syndrome,614192
RNF139	227.1	100%	100%	Renal cell carcinoma,144700
RNF168	212.8	100%	100%	RIDDLE syndrome, 611943
RNF170	145.9	95%	89%	taxia, sensory, 1, autosomal dominant, 608984
RNF212	98.4	100%	95%	Recombination rate QTL 1,612042
RNF216	134.7	100%	98%	Cerebellar ataxia and hypogonadotropic hypogonadism,212840
RNF6	225.9	100%	99%	Esophageal carcinoma,somatic,133239
RNU4ATAC				Microcephalic osteodysplastic primordial dwarfism,type I,210710 Roifman syndrome,616651
ROBO2	138.7	98%	96%	Vesicoureteral reflux 2, 610878
ROBO3	85.2	96%	91%	Gaze palsy, horizontal, with progressive scoliosis, 607313
ROGDI	110.8	96%	94%	Kohlschutter-Tonz syndrome, 226750

ROM1	97.7	100%	98%	Retinitis pigmentosa 7, digenic, 608133
ROR2	155	99%	97%	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RP1	123.7	99%	96%	Retinitis pigmentosa 1, 180100 {Hypertriglyceridemia, susceptibility to}, 145750
RP1L1	81.7	100%	98%	Occult macular dystrophy, 613587
RP2	117.2	100%	98%	Retinitis pigmentosa 2, 312600
RPE65	138.6	100%	99%	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794
RPGR	59.2	81%	66%	Cone-rod dystrophy, X-linked, 1, 304020 Macular degeneration, X-linked atrophic, 300834 Retinitis pigmentosa 3, 300029 Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455
RPGRIP1	145.2	100%	99%	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826
RPGRIP1L	133.2	95%	93%	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561
RPIA	110.3	91%	87%	Ribose 5-phosphate isomerase deficiency, 608611
RPL11	89.3	100%	95%	Diamond-Blackfan anemia 7, 612562
RPL35A	64.6	95%	80%	Diamond-Blackfan anemia 5, 612528
RPL5	31	71%	50%	Diamond-Blackfan anemia 6, 612561
RPS10	112.9	93%	89%	Diamond-Blackfan anemia 9, 613308
RPS14	115.5	98%	91%	Macrocytic anemia, refractory, due to 5q deletion, somatic, 153550
RPS17	40.8	78%	68%	Diamond-Blackfan anemia 4, 612527
RPS19	78.4	100%	97%	Diamond-Blackfan anemia 1, 105650
RPS24	105.6	99%	87%	Diamond-blackfan anemia 3, 610629
RPS26	71.8	79%	69%	Diamond-Blackfan anemia 10, 613309
RPS6KA3	55.2	88%	80%	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844
RPS7	91.1	79%	70%	Diamond-Blackfan anemia 8, 612563
RPSA	82.1	100%	100%	Asplenia, isolated congenital, 271400
RRAS2	70.1	82%	75%	Ovarian carcinoma

RRM2B	133	98%	96%	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy),612075 Mitochondrial DNA depletion syndrome 8B (MNGIE type),612075 Progressive external ophthalmoplegia with mitochondrial DNA deletions,autosomal dominant,613077
RS1	32.2	83%	72%	Retinoschisis, 312700
RSPH1	151.3	100%	100%	Ciliary dyskinesia, primary, 24, 615481
RSPH4A	128.9	98%	96%	Ciliary dyskinesia, primary, 11, 612649
RSPH9	123.6	100%	97%	Ciliary dyskinesia, primary, 12, 612650
RSPO1	95.6	100%	97%	Palmoplantar hyperkeratosis and true hermaphroditism,610644 Palmoplantar hyperkeratosis with squamous cell carcinoma and sex reversal,610644
RSPO4	92.8	100%	96%	Anonychia congenita,206800
RTEL1	100	98%	92%	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190
RTN2	90.8	98%	94%	Spastic paraplegia 12, autosomal dominant, 604805
RTTN	132.4	98%	95%	Polymicrogyria with seizures,614833
RUNX1	106.5	96%	92%	Leukemia, acute myeloid, 601626 Platelet disorder, familial, with associated myeloid malignancy, 601399
RUNX2	94.4	74%	74%	Cleidocranial dysplasia, 119600 Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600 Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 Metaphyseal dysplasia with maxillary hypoplasia with/without brachydactyly, 156510
RXFP2	136.7	98%	94%	?Cryptorchidism,219050
RYR1	107.3	95%	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	134.1	99%	97%	Arrhythmogenic right ventricular dysplasia 2, 600996 Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772
SACS	148.8	99%	99%	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAG	126.2	100%	100%	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758
SALL1	115.7	100%	98%	Townes-Brocks syndrome, 107480
SALL4	122	97%	96%	Duane-radial ray syndrome, 607323
SAMD9	169.1	100%	96%	Tumoral calcinosis familial normophosphatemic,610455

SAMHD1	124.9	97%	94%	Aicardi-Goutieres syndrome 5, 612952
SAR1B	121.5	89%	88%	Chylomicron retention disease,246700
SARS2	92	96%	94%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SART3	122.1	100%	97%	No OMIM phenotype Disseminated superficial actinic porokeratosis (Zhang (2005) Br J Dermatol 152,658)
SAT1	77.6	93%	92%	Keratosis follicularis spinulosa decalvans, 308800
SATB2	98	98%	91%	Cleft palate and mental retardation, 119540
SBDS	170.6	100%	99%	Shwachman-Bodian-Diamond syndrome, 260400
SBF2	117	99%	95%	Charcot-Marie-Tooth disease, type 4B2, 604563
SC5D	199.5	100%	100%	Lathosterolosis, 607330
SCARB2	126.6	100%	97%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCARF2	66.3	89%	75%	Van den Ende-Gupta syndrome,600920
SCN10A	164	99%	98%	Episodic pain syndrome,familial 2,615551
SCN11A	131.8	98%	97%	Episodic pain syndrome, familial, 3, 615552 Neuropathy,hereditary sensory and autonomic,type VIII,615548
SCN1A	151.4	99%	98%	Dravet syndrome, 607208 Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Febrile seizures, familial, 3A, 604403 Migraine, familial hemiplegic, 3, 609634
SCN1B	151.7	100%	96%	Atrial fibrillation, familial, 13, 615377 Brugada syndrome 5, 612838 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233
SCN2A	153.2	99%	97%	Epileptic encephalopathy, early infantile, 11, 613721 Seizures, benign familial infantile, 3, 607745
SCN2B	176.9	100%	100%	Atrial fibrillation, familial, 14, 615378
SCN3B	136.4	100%	100%	Brugada syndrome 7, 613120
SCN4A	209.6	100%	99%	Hyperkalemic periodic paralysis, type 2, 170500 Hypokalemic periodic paralysis, type 2, 613345 Myasthenic syndrome, acetazolamide-responsive, 614198 Myotonia congenita, atypical, acetazolamide-responsive, 608390 Paramyotonia congenita, 168300
SCN4B	67.8	100%	99%	Long QT syndrome-10, 611819

SCN5A	169.5	100%	99%	Atrial fibrillation, familial, 10, 614022 Brugada syndrome 1, 601144 Cardiomyopathy,dilated, 1E,601154 Heart block,nonprogressive,113900 Heart block,progressive,type 1A,113900 Long QT syndrome-3,603830 Sick sinus syndrome 1, 608567 Ventricular fibrillation,familial,1,603829 {Sudden infant death syndrome, susceptibility},272120
SCN8A	192.7	99%	97%	Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy, early infantile, 13, 614558
SCN9A	146.4	96%	94%	Epilepsy,generalized,with febrile seizures plus,type 7,613863 Erythralgia, primary, 133020 Febrile seizures,familial,3B,613863 HSAN2D,autosomal recessive,243000 Insensitivity to pain,congenital,243000 Paroxysmal extreme pain disorder,167400
SCNN1A	117.7	94%	92%	Bronchiectasis with or without elevated sweat chloride 2,613021 Pseudohypoaldosteronism, type I, 264350
SCNN1B	133.8	100%	100%	Bronchiectasis with or without elevated sweat chloride 1,211400 Liddle syndrome, 177200 Pseudohypoaldosteronism,type I,264350
SCNN1G	140.6	98%	96%	Bronchiectasis with or without elevated sweat chloride 3,613071 Liddle syndrome, 177200 Pseudohypoaldosteronism, type I,264350
SCO1	110.2	98%	94%	Mitochondrial complex IV deficiency,220110
SCO2	94.1	100%	100%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908
SCP2	106.4	100%	99%	Leukoencephalopathy with dystonia and motor neuropathy, 613724
SDCCAG8	116.7	100%	96%	Senior-Loken syndrome 7, 613615
SDHA	96.8	81%	73%	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165
SDHAF1	36.9	100%	94%	Mitochondrial complex II deficiency, 252011

SDHAF2	142.1	94%	94%	Paragangliomas 2, 601650
SDHB	124.7	100%	100%	Cowden syndrome 2, 612359 Gastrointestinal stromal tumor, 606764 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 4, 115310 Pheochromocytoma, 171300
SDHC	94	99%	92%	Gastrointestinal stromal tumor, 606764 Paragangliomas 3, 605373 Paraganglioma and gastric stromal sarcoma, 606864
SDHD	51.2	59%	59%	Carcinoid tumors, intestinal, 114900 Cowden syndrome 3, 615106 Merkel cell carcinoma, somatic Mitochondrial complex II deficiency,252011 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 1, with or without deafness, 168000 Pheochromocytoma, 171300
SEC23A	133.1	100%	95%	Craniolenticulosutural dysplasia,607812
SEC23B	157.3	97%	96%	Anemia dyserythropoietic congenital type II,224100
SEC63	63.6	83%	77%	Polycystic liver disease,174050
SECISBP2	119.2	99%	95%	Thyroid hormone metabolism,abnormal,609698
SEMA3E	143.2	100%	98%	CHARGE syndrome,214800
SEMA4A	118.9	100%	97%	Cone-rod dystrophy 10, 610283 Retinitis pigmentosa 35, 610282
SEPN1	95.8	83%	79%	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310
SEPSECS	173.1	100%	100%	Pontocerebellar hypoplasia type 2D, 613811
SEPT12				Spermatogenic failure 10,614822
SEPT9				Amyotrophy,hereditary neuralgic,162100 Leukemia,acute myeloid,therapy-related Ovarian carcinoma
SERAC1	112.7	99%	94%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPINA1	151.5	100%	100%	Emphysema due to AAT deficiency,613490 {Pulmonary disease,chronic obstructive,susceptibility to},606963
SERPINA3	129	100%	100%	Alpha-1-antichymotrypsin deficiency

				Cerebrovascular disease,occlusive
SERPINA6	178.1	100%	100%	Corticosteroid-binding globulin deficiency,611489
SERPINA7	122	100%	99%	Thyroxine-binding globulin deficiency
SERPINB6	167.2	100%	100%	Deafness, autosomal recessive 91, 613453
SERPINB7	130.6	100%	100%	Palmoplantar keratoderma, Nagashima type, 615598 ,615598
SERPINC1	128.2	100%	100%	Thrombophilia due to antithrombin III deficiency,613118
SERPIND1	151.3	100%	100%	Thrombophilia due to heparin cofactor II deficiency,612356
SERPINE1	118.8	100%	100%	Plasminogen activator inhibitor-1 deficiency,613329 {Transcription of plasminogen activator inhibitor,modulator of}
SERPINF1	87	99%	97%	Osteogenesis imperfecta,type VI,613982
SERPINF2	115.5	100%	98%	Alpha-2-plasmin inhibitor deficiency,262850
SERPING1	80.6	96%	89%	Angioedema, hereditary, types I and II, 106100
SERPINH1	145.9	100%	100%	Osteogenesis imperfecta type X,613848 {Preterm premature rupture of the membranes, susceptibility to},610504
SERPINI1	110.5	100%	99%	Encephalopathy,familial,with neuroserpin inclusion bodies,604218
SETBP1	115.9	97%	96%	Schinzel-Giedion midface retraction syndrome, 269150
SETD5	176.3	100%	99%	Mental retardation,autosomal dominant 24,615761
SETX	165.1	99%	98%	Amyotrophic lateral sclerosis 4, juvenile, 602433 Ataxia-ocular apraxia-2, 606002
SF3B1	130	99%	98%	Myelodysplastic syndrome,somatic,614286
SF3B4	72.3	98%	84%	Acrofacial dysostosis 1,Nager type,154400
SFTPA2	134.2	100%	100%	Pulmonary fibrosis, idiopathic, 178500
SFTPB	99.3	100%	98%	Surfactant metabolism dysfunction,pulmonary 1,265120
SFTPC	76.8	98%	87%	Surfactant metabolism dysfunction, pulmonary 2,610913
SFXN4	132	100%	99%	Combined oxidative phosphorylation deficiency 18, 615578
SGCA	136.9	100%	100%	Muscular dystrophy, limb-girdle, type 2D, 608099
SGCB	168.1	96%	95%	Muscular dystrophy, limb-girdle, type 2E, 604286
SGCD	81.4	99%	94%	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, type 2F, 601287
SGCE	84.1	95%	90%	maternally imprinted Dystonia-11, myoclonic, 159900
SGCG	119.5	100%	100%	Muscular dystrophy, limb-girdle, type 2C, 253700
SGSH	120.8	94%	92%	Mucopolysaccharidosis type 3A (Sanfilippo A), 252900

SH2B3	79.5	84%	70%	Erythrocytosis,somatic,133100 Myelofibrosis,somatic,254450 Thrombocythemia,somatic,187950
SH2D1A	82.1	89%	89%	Lymphoproliferative syndrome, X-linked, 308240
SH3BP2	100.1	91%	90%	Cherubism, 118400
SH3PXD2B	124.1	100%	100%	Frank-ter Haar syndrome,249420
SH3TC2	103.3	100%	99%	Charcot-Marie-Tooth disease, type 4C, 601596 Mononeuropathy of the median nerve,mild,613353
SHANK3	65.5	79%	70%	Phelan-McDermid syndrome, 606232 {Schizophrenia 15}, 613950
SHH	84.8	95%	91%	Holoprosencephaly-3, 142945 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250
SHOC2	124.9	100%	99%	Noonan-like syndrome with loose anagen hair, 607721
SHOX	21.6	66%	54%	Langer mesomelic dysplasia,249700 Leri-Weill dyschondrosteosis,127300 Short stature,idiopathic familial,300582
SHROOM4	58.9	99%	90%	Stocco dos Santos X-linked mental retardation syndrome, 300434
SI	99.9	96%	85%	Sucrase-isomaltase deficiency, congenital, 222900
SIGMAR1	110.6	100%	99%	Amyotrophic lateral sclerosis 16, juvenile, 614373
SIL1	149.3	97%	96%	Marinesco-Sjogren syndrome, 248800
SIM1	128.5	100%	99%	Obesity,severe,601665
SIX1	100.8	95%	94%	Brachiootic syndrome 3, 608389 Deafness,autosomal dominant 23,605192
SIX3	122.8	97%	89%	Holoprosencephaly-2, 157170 Schizensephaly, 269160
SIX5	32.9	82%	61%	Branchiootorenal syndrome 2, 610896
SIX6	180.4	100%	100%	Microphthalmia with cataract 2, 212550
SKI	68.1	96%	95%	Shprintzen-Goldberg syndrome, 182212
SKIV2L	21.4	75%	47%	Trichohepatoenteric syndrome 2, 614602
SLC10A2	135.5	100%	99%	Bile acid malabsorption,primary,613291
SLC11A2	130.1	100%	100%	Anemia, hypochromic microcytic,206100
SLC12A1	167.2	100%	98%	Bartter syndrome, type 1, 601678



SLC12A3	126.1	100%	100%	Gitelman syndrome, 263800
SLC12A6	144.7	100%	100%	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC16A1	140.7	99%	96%	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia, familial, 7, 610021 (3)
SLC16A12	167.6	100%	100%	Cataract, juvenile, with microcornea and glucosuria, 612018
SLC16A2	34.9	90%	77%	Allan-Herndon-Dudley syndrome, 300523
SLC17A5	104.2	99%	92%	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC17A8	128.8	100%	97%	Deafness, autosomal dominant 25, 605583
SLC19A2	115.7	97%	92%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	168.9	100%	100%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A3	141.2	100%	100%	Episodic ataxia, type 6, 612656
SLC20A2	99.8	98%	91%	Basal ganglia calcification, idiopathic, 1, 213600
SLC22A12	101.6	100%	100%	Hypouricemia, renal, 220150
SLC22A18	74.4	88%	87%	Breast cancer,somatic,114480 Lung cancer,somatic,211980 Rhabdomyosarcoma,somatic,268210
SLC22A5	137.2	100%	100%	Carnitine deficiency, systemic primary, 212140
SLC24A1	199.4	100%	99%	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
SLC24A5	119.9	95%	93%	Albinism, oculocutaneous, type VI,113750 [skin/hair/eye pigmentation 4],113750
SLC25A1	64.1	97%	88%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A12	149.7	99%	95%	Hypomyelination, global cerebral, 612949
SLC25A13	103.6	93%	90%	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A15	212.9	98%	97%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A19	60.5	100%	95%	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A20	101.4	100%	100%	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A22	91.8	100%	97%	Epileptic encephalopathy, early infantile, 3, 609304
SLC25A3	137.6	97%	88%	Mitochondrial phosphate carrier deficiency, 610773
SLC25A38	109.8	100%	98%	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive, 205950

SLC25A4	125.7	100%	100%	Progressive external ophthalmoplegia with mitochondrial DNA deletions 3, 609283 Mitochondrial DNA depletion syndrome 12 (cardiomyopathic type), 615418
SLC26A2	223	100%	100%	Achondrogenesis Ib,600972 Atelosteogenesis II,256050 De la Chapelle dysplasia,256050 Diastrophic dysplasia,222600 Diastrophic dysplasia,broad bone-platyspondylic variant,222600 Epiphyseal dysplasia,multiple,4,226900
SLC26A3	161.1	100%	97%	Diarrhea 1,secretory chloride,congenital,214700
SLC26A4	121.4	99%	97%	Deafness,autosomal recessive 4,with enlarged vestibular aqueduct,600791 Pendred syndrome, 274600
SLC26A5	151.2	100%	99%	Deafness, autosomal recessive 61, 613865
SLC26A8	140	100%	100%	Spermatogenic failure 3,606766
SLC27A4	128	100%	97%	Ichthyosis prematurity syndrome,608649
SLC29A3	190.4	99%	99%	Histiocytosis-lymphadenopathy plus syndrome,602782
SLC2A1	161.8	100%	98%	Dystonia 9,601042 GLUT1 deficiency syndrome 1, 606777 GLUT1 deficiency syndrome 2, 612126 Stomatin-deficient cryohydrocytosis with neurologic defects,608885 {Epilepsy, idiopathic generalized,susceptibility,12},614847
SLC2A10	138.4	100%	99%	Arterial tortuosity syndrome,208050
SLC2A2	154.8	100%	100%	Fanconi-Bickel syndrome,227810 {Diabetes mellitus, noninsulin-dependent},125853
SLC2A9	127.5	100%	93%	Hypouricemia,renal,2,612076 {Uric acid concentration, serum, QTL 2}, 612076
SLC30A10	156.5	100%	100%	Hypermanganesemia with dystonia, polycythemia, and cirrhosis, 613280
SLC30A2	128.4	100%	99%	Zinc deficiency,transient neonatal,608118
SLC33A1	133.8	98%	89%	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539
SLC34A1	131.8	100%	99%	Fanconi renotubular syndrome 2,613388 Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286
SLC34A2	162.5	100%	99%	?Testicular microlithiasis,610441 Pulmonary alveolar microlithiasis,265100
SLC34A3	104.9	95%	94%	Hypophosphatemic rickets with hypercalciuria, 241530

SLC35A1	127	100%	97%	Congenital disorder of glycosylation, type 2f, 603585
SLC35A2	66.2	97%	93%	Congenital disorder of glycosylation, type 2m, 300896
SLC35C1	198.2	99%	96%	Congenital disorder of glycosylation, type IIc, 266265
SLC35D1	121.1	92%	91%	Schneckenbecken dysplasia,269250
SLC36A2	112.2	100%	100%	Hyperglycinuria,138500 Iminoglycinuria,digenic,242600
SLC37A4	119.5	100%	100%	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240
SLC38A8	74	100%	93%	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218
SLC39A13	97	100%	94%	Spondylocheirodysplasia Ehlers-Danlos syndrome-like,612350
SLC39A4	67.6	99%	94%	Acrodermatitis enteropathica, 201100
SLC3A1	171.4	100%	97%	Cystinuria, 220100
SLC40A1	137.7	99%	98%	Hemochromatosis, type 4, 606069
SLC45A2	128.1	100%	98%	Oculocutaneous albinism type IV,606574 [skin/hair/eye pigmentation 5],227240
SLC46A1	90.7	97%	88%	Folate malabsorption, hereditary, 229050
SLC4A1	115.7	100%	99%	Ovalocytosis Renal tubular acidosis,distal,AD,179800 Renal tubular acidosis,distal,AR,611590 Spherocytosis,type 4,612653
SLC4A11	133.3	100%	99%	Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy 2, autosomal recessive, 217700 Corneal endothelial dystrophy and perceptive deafness, 217400
SLC4A4	132.4	100%	99%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC52A1	194.6	100%	100%	Riboflavin deficiency, 615026
SLC52A2	168	100%	100%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	104.7	100%	98%	Brown-Vialetto-Van Laere syndrome 1,211530 Fazio-Londe disease,211500
SLC5A1	131.9	100%	100%	Glucose/galactose malabsorption, 606824
SLC5A2	112.7	100%	100%	Renal glucosuria, 233100
SLC5A5	85.2	96%	91%	Thyroid dyshormonogenesis 1,274400
SLC5A7	121	100%	100%	Neuronopathy, distal hereditary motor, type VIIA, 158580
SLC6A19	147.7	98%	95%	Hartnup disorder, 234500 Hyperglycinuria,138500

				Iminoglycinuria,digenic,242600
SLC6A2	129	98%	96%	Orthostatic intolerance,604715
SLC6A20	159.4	100%	97%	Hyperglycinuria, 138500
SLC6A3	122.6	100%	99%	Parkinsonism -dystonia, infantile, 613135 {Nicotine dependence, protection against}, 188890
SLC6A5	133.8	99%	95%	Hyperekplexia 3,614618
SLC6A8	32.5	79%	66%	Cerebral creatine deficiency syndrome 1, 300352
SLC7A14	179.5	100%	100%	Retinitis pigmentosa 68, 615725
SLC7A7	106.3	100%	100%	Lysinuric protein intolerance, 222700
SLC7A9	125.6	100%	94%	Cystinuria, 220100
SLC9A3R1	96.8	99%	97%	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
SLC9A6	73.9	93%	83%	Mental retardation, X-linked syndromic, Christianson type, 300243
SLCO1B1	49.9	93%	84%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO1B3	55.9	96%	81%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO2A1	94.9	100%	97%	Hypertrophic osteoarthropathy primary autosomal recessive 2,614441
SLITRK1	122.1	100%	100%	?Trichotillomania,613229 Tourette syndrome,137580
SLITRK6	232.1	100%	100%	Deafness and myopia, 221200
SLURP1	89.6	99%	89%	Meleda disease,248300
SLX4	101.3	100%	99%	Fanconi anemia, complementation group P, 613951
SMAD3	112.5	99%	97%	Loeys-Dietz syndrome type 3,613795
SMAD4	114.6	99%	99%	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer,somatic,260350 Polyposis, juvenile intestinal, 174900
SMAD6	78.5	82%	63%	Aortic valve disease 2, 614823
SMAD9	140.8	100%	99%	Pulmonary hypertension,primary,615342
SMARCA2	109.5	96%	94%	Nicolaides-Baraitser syndrome, 601358
SMARCA4	131.9	99%	94%	Mental retardation, autosomal dominant 16, 614609 Rhabdoid tumor predisposition syndrome 2, 613325
SMARCAD1	83	99%	94%	Adermatoglyphia,136000
SMARCAL1	121.1	100%	98%	Schimke immunosseous dysplasia, 242900

SMARCB1	202.6	100%	100%	Coffin-Siris syndrome 3,614608 Rhabdoid tumors,somatic,609322 {Rhabdoid predisposition syndrome 1},609322 {Schwannomatosis-1,susceptibility to},162091
SMC1A	67.1	99%	95%	Cornelia de Lange syndrome 2, 300590
SMC3	77.3	94%	77%	Cornelia de Lange syndrome 3, 610759
SMCHD1	94.9	95%	90%	Fascioscapulohumeral muscular dystrophy 2,digenic,158901
SMN1	73.3	99%	91%	Spinal muscular atrophy-1, 253300 Spinal muscular atrophy-2, 253550 Spinal muscular atrophy-3, 253400 Spinal muscular atrophy-4, 271150
SMO	147.3	96%	89%	Basal cell carcinoma, somatic
SMOC1	107.8	100%	96%	Microphthalmia with limb anomalies, 206920
SMOC2	98.8	97%	92%	Dentin dysplasia type I with microdontia and misshapen teeth,125400
SMPD1	110.9	100%	97%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SMPX	50.2	100%	97%	Deafness, X-linked 4, 300066
SMS	40.3	85%	61%	Mental retardation, X-linked, Snyder-Robinson type, 309583
SNAI2	125.1	100%	98%	Piebaldism,172800 Waardenburg syndrome, type 2D,608890
SNAP29	134.3	100%	100%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNCA	129.3	100%	100%	Dementia, Lewy body, 127750 Parkinson disease 1, 168601 Parkinson disease 4, 605543
SNCB	70.3	100%	100%	Dementia,Lewy body,127750
SNIP1	125.3	97%	95%	Psychomotor retardation, epilepsy and craniofacial dysmorphism, 614501
SNRNP200	144.9	100%	98%	Retinitis pigmentosa 33, 610359
SNRPE	92.7	99%	99%	Hypotrichosis 11,615059
SNRPN	112.6	100%	82%	Prader-Willi syndrome,176270
SNTA1	81	80%	73%	Long QT syndrome 12, 612955
SNX10	95.8	100%	98%	Osteopetrosis autosomal recessive 8,615085
SOBP	93	90%	77%	Mental retardation, anterior maxillary protrusion, and strabismus, 613671
SOD1	156.1	100%	100%	Amyotrophic lateral sclerosis 1, 105400

SOS1	87.5	93%	88%	Fibromatosis, gingival, 135300 Noonan syndrome 4, 610733
SOST	107.6	100%	97%	Craniodiaphyseal dysplasia,autosomal dominant,122860 Sclerosteosis 1,269500 Van Buchem disease,239100
SOX10	65.7	92%	88%	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266
SOX17	59.6	99%	83%	Vesicoureteral reflux 3, 613674
SOX18	13.9	56%	44%	Hypotrichosis-lymphedema-telangiectasia syndrome,607823 Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome,137940
SOX2	80.8	99%	96%	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SOX3	17.5	81%	38%	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000
SOX9	96.4	94%	87%	Acampomelic campomelic dysplasia,114290 Campomelic dysplasia,114290 Campomelic dysplasia with autosomal sex reversal,114290
SP110	129.9	100%	100%	Hepatic venoocclusive disease with immunodeficiency, 235550
SP7	159.5	100%	100%	Osteogenesis imperfecta type XII,613849
SPAG1	71.3	91%	82%	Ciliary dyskinesia, primary, 28, 615505
SPAST	59.8	84%	72%	Spastic paraplegia 4, autosomal dominant, 182601
SPATA16	156.1	100%	98%	?Spermatogenic failure 6,102530
SPATA7	128	96%	86%	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, autosomal recessive, 604232
SPECC1L	141.1	100%	100%	Facial clefting, oblique, 1, 600251
SPG11	130.1	98%	95%	Spastic paraplegia 11, autosomal recessive, 604360
SPG20	137.6	98%	95%	Troyer syndrome, 275900
SPG21	120.9	98%	91%	Mast syndrome, 248900
SPG7	109	93%	89%	Spastic paraplegia 7, autosomal recessive, 607259
SPINK1	78.3	100%	100%	Pancreatitis,hereditary,167800 Tropical calcific pancreatitis,608189
SPINK5	154.4	100%	95%	Netherton syndrome, 256500
SPINT2	54.1	94%	77%	Diarrhea 3 secretory sodium congenital syndromic,270420

SPR	148	92%	78%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRED1	165.6	96%	96%	Legius syndrome, 611431
SPRY4	128.5	100%	100%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
SPTA1	121.6	99%	97%	Elliptocytosis-2,130600 Pyropoikilocytosis,266140 Spherocytosis,type 3,270970
SPTAN1	116.4	99%	97%	Epileptic encephalopathy, early infantile, 5
SPTB	131.3	100%	99%	Anemia,neonatal hemolytic,fatal and near-fatal Elliptocytosis-3 Spherocytosis,type 2,616649
SPTBN2	103	99%	98%	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia, autosomal recessive 14, 615386
SPTLC1	107.7	100%	92%	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	159.1	100%	100%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SQSTM1	97.5	97%	92%	Paget disease of bone, 602080
SRC	90.8	99%	95%	Colon cancer,advanced,somatic
SRCAP	131.2	99%	98%	Floating-Harbor syndrome, 136140
SRD5A2	82.2	100%	89%	Pseudovaginal perineoscrotal hypospadias,264600
SRD5A3	140.5	99%	96%	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713
SRP72	69.7	97%	86%	Bone marrow failure syndrome 1,614675
SRPX2	51.4	98%	95%	Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643
SRY	99.8	97%	97%	46XX sex reversal 1,400045 46XY sex reversal 1,400044
SSTR5	143.6	99%	92%	Somatostatin analog, resistance to
ST14	151.4	100%	98%	Ichthyosis with hypotrichosis,610765
ST3GAL3	163.4	100%	100%	Epileptic encephalopathy, early infantile, 15, 615006 Mental retardation, autosomal recessive 12, 611090
ST3GAL5	118.3	94%	93%	Amish infantile epilepsy syndrome, 609056
STAC3	117.7	100%	100%	Native American myopathy,255995
STAMBP	118	100%	96%	Microcephaly-capillary malformation syndrome,614261
STAR	114.1	100%	100%	Lipoid adrenal hyperplasia, 201710
STAT1	134.1	96%	94%	Mycobacterial infection, atypical, familial disseminated, 209950
STAT3	122.2	100%	98%	Hyper-IgE recurrent infection syndrome, 147060

STAT5B	116.6	100%	95%	Growth hormone insensitivity with immunodeficiency, 245590 Leukemia, acute promyelocytic, STAT5B/RARA type
STIL	135.4	100%	99%	Microcephaly 7, primary, autosomal recessive, 612703
STIM1	125.1	99%	95%	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070
STK11	112.3	100%	97%	Melanoma, malignant, somatic Peutz-Jeghers syndrome, 175200 Pancreatic cancer, 260350 Testicular tumor, somatic, 273300
STK4	132.2	100%	99%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STOX1	188.5	89%	89%	Preeclampsia/eclampsia 4,609404
STRA6	105.6	100%	100%	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186
STRADA	123.4	99%	94%	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087
STRC	88	99%	95%	Deafness, autosomal recessive 16, 603720
STS	61.2	97%	92%	Ichthyosis, X-linked, 308100
STX11	245.7	100%	100%	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STX16	149.1	98%	98%	Pseudohypoparathyroidism, type 1B, 603233
STXBP1	124.2	100%	100%	Epileptic encephalopathy, early infantile, 4, 612164
STXBP2	107.6	99%	94%	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
SUCLA2	58	94%	76%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with/without methylmalonic aciduria), 612073
SUCLG1	94.7	100%	98%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUFU	125.3	97%	96%	Medulloblastoma, desmoplastic, 155255 {Meningioma, familial, susceptibility to}, 607174
SUMF1	122.6	91%	86%	Multiple sulfatase deficiency, 272200
SUMO1	22.5	66%	45%	Orofacial cleft 10, 613705
SUOX	189.8	100%	100%	Sulfite oxidase deficiency, 272300
SURF1	87.3	88%	88%	Leigh syndrome, due to COX deficiency, 256000
SYCP3	93.7	97%	77%	Spermatogenic failure 4, 270960 {Pregnancy loss, susceptibility to}
SYN1	44.3	70%	56%	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491



SYNE1	137.1	99%	98%	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 Spinocerebellar ataxia, autosomal recessive 8, 610743
SYNE2	114.9	98%	95%	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999
SYNE4	58.9	96%	90%	Deafness, autosomal recessive 76, 615540
SYNGAP1	60.9	92%	79%	Mental retardation, autosomal dominant 5, 612621
SYNJ1	123.1	98%	95%	Parkinson disease 20,early-onset,615530
SYP	40	93%	74%	Mental retardation, X-linked 96, 300802
SYT14	174.6	93%	83%	Spinocerebellar ataxia, autosomal recessive 11, 614229
SZT2	128.5	99%	99%	Epileptic encephalopathy, early infantile, 18, 615476
T	127.8	100%	98%	Sacral agenesis with vertebral anomalies,615709
TAB2	189.8	99%	95%	Congenital heart defects, nonsyndromic, 2, 614980
TAC3	74.2	95%	75%	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACR3	164.3	100%	99%	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
TACSTD2	176.2	99%	97%	Corneal dystrophy, gelatinous drop-like, 204870
TAF1	80.2	96%	91%	Dystonia-Parkinsonism, X-linked, 314250
TAF2	121	99%	95%	Mental retardation, autosomal recessive 40, 615599
TAL1	39.8	67%	58%	Leukemia,T-cell acute lymphocytic,somatic,613065
TAL2	80.6	100%	100%	Leukemia,T-cell acute lymphocytic,somatic,613065
TALDO1	127.8	100%	100%	Transaldolase deficiency, 606003
TAP1	10.3	39%	7%	Bare lymphocyte syndrome, type I, 604571
TAP2	10	37%	15%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	18	72%	40%	Bare lymphocyte syndrome, type I, 604571
TARDBP	149.6	100%	99%	Amyotrophic lateral sclerosis 10, with or without FTD, 612069 Frontotemporal lobar degeneration, TARDBP-related, 612069
TAT	117.2	100%	100%	Tyrosinemia, type II, 276600
TAZ	74.7	100%	99%	Barth syndrome, 302060
TBC1D20	135.5	94%	93%	Warburg micro syndrome 4,615663
TBC1D24	149.2	100%	100%	Deafness,autosomal recessive 86,614617 Deafness,autosomal dominant 65,616044 DOOR syndrome,220500 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021
TBCE	147.1	99%	98%	Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Kenny-Caffey syndrome-1, 244460

TBP	118.9	100%	94%	Spinocerebellar ataxia 17,607136 {Parkinson disease,susceptibility to},168600
TBX1	70.9	73%	59%	Conotruncal anomaly face syndrome, 217095
TBX15	103.1	100%	96%	Cousin syndrome,260660
TBX19	161.9	100%	99%	Adrenocorticotropic hormone deficiency,201400
TBX20	125.5	100%	99%	Atrial septal defect 4, 611363
TBX21	77.6	91%	87%	Asthma and nasal polyps,208550
TBX22	66.2	96%	90%	Cleft palate with ankyloglossia, 303400 ?Abruzzo-Erickson syndrome, 302905
TBX3	74.3	99%	94%	Ulnar-mammary syndrome,181450
TBX4	145.9	92%	91%	Small patella syndrome,147891
TBX5	131.6	100%	100%	Holt-Oram syndrome, 142900
TBXAS1	133.8	100%	100%	Ghosal hematodiaphyseal syndrome, 231095 ?Thromboxane synthase deficiency, 614158
TCAP	59.8	100%	100%	Cardiomyopathy, dilated, 1N, 607487 Muscular dystrophy, limb-girdle, type 2G, 601954
TCF12	147.6	100%	99%	Craniosynostosis 3, 615314
TCF4	137.6	100%	100%	Pitt-Hopkins syndrome, 610954
TCIRG1	107.1	95%	84%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	155.6	100%	100%	Transcobalamin II deficiency, 275350
TCOF1	75.9	98%	93%	Treacher Collins syndrome 1, 154500
TCTN1	98.8	94%	92%	Joubert syndrome 13, 614173
TCTN2	143.3	98%	95%	?Meckel syndrome 8, 613885
TCTN3	116.3	99%	98%	Joubert syndrome 18,614815 Orofaciodigital syndrome IV, 258860
TDGF1	137.6	99%	85%	Forebrain defects Congenital heart defects (Roessler (2008) Am J Hum Genet 83, 18)
TDP1	109.2	98%	95%	Spinocerebellar ataxia, autosomal recessive with axonal neuropathy, 607250
TDRD7	170.6	98%	97%	Cataract 36, 613887
TEAD1	161.4	100%	96%	Sveinsson choreoretinal atrophy, 108985
TECPR2	146.5	100%	99%	Spastic paraplegia 49, autosomal recessive, 615031
TECR	80.7	98%	91%	Mental retardation, autosomal recessive 14, 614020
TECTA	192.7	100%	100%	Deafness, autosomal dominant 8/12, 601543 Deafness,autosomal recessive 21,603629

TEK	193	100%	99%	Venous malformations multiple cutaneous and mucosal,600195
TENM3	185.5	99%	98%	Microphthalmia, isolated, with coloboma 9, 61545
TERC				Dyskeratosis congenita,autosomal dominant 1,127550 {Aplastic anemia},614743
TET2	191.7	99%	99%	Myelodysplastic syndrome,somatic,614286
TEX28	1	0%	0%	No OMIM phenotype
TF	125.8	100%	99%	Atransferrinemia, 209300
TFAP2A	109.5	100%	98%	Branchiooculofacial syndrome, 113620
TFAP2B	117	98%	95%	Char syndrome, 169100
TFE3	48.6	100%	76%	Renal cell carcinoma,300854
TFG	118.8	95%	90%	?Spastic paraplegia 57,autosomal recessive,615658 Hereditary motor and sensory neuropathy,Okinawa type,604484
TFR2	86.8	98%	95%	Hemochromatosis, type 3, 604250
TG	119	100%	98%	Thyroid dysmorphogenesis 3,274700 {Autoimmune thyroid disease,susceptibility to},608175
TGFB1	63.6	100%	99%	Camurati-Engelmann disease, 131300 {Cystic fibrosis lung disease, modifier of}, 219700
TGFB2	153	100%	100%	Loeys-Dietz syndrome type 4,614816
TGFB3	147.1	100%	100%	Arrhythmogenic right ventricular dysplasia 1, 107970
TGFBI	121.8	100%	92%	Corneal dystrophy, Avellino type, 607541 Corneal dystrophy, epithelial basement membrane, 121820 Corneal dystrophy, Groenouw type I, 121900 Corneal dystrophy, lattice type I, 122200 Corneal dystrophy, lattice type IIIA, 608471
TGFBR1	196.2	93%	93%	Loeys-Dietz syndrome, type 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800
TGFBR2	184.5	100%	100%	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome, type 2, 610168
TGIF1	126.4	100%	97%	Holoprosencephaly-4, 142946
TGM1	157.1	100%	99%	Ichthyosis congenital autosomal recessive 1,242300
TGM5	161.4	100%	99%	Peeling skin syndrome acral type,609796
TGM6	130.5	98%	97%	Spinocerebellar ataxia 35, 613908
TH	66.4	95%	87%	Segawa syndrome,recessive,605407

THAP1	139.3	100%	100%	Dystonia 6, torsion, 602629
THBD	85.5	100%	91%	Thrombophilia due to thrombomodulin defect, 614486 {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
THOC6	227.3	100%	100%	Beaulieu-Boycott-Innes syndrome, 613680
THPO	74.3	100%	100%	Thrombocythemia 1,187950
THRA	153	100%	97%	Hypothyroidism,congenital,nongoitrous,6,614450
THRB	165.9	100%	98%	Thyroid hormone resistance, 188570 Thyroid hormone resistance, autosomal recessive, 274300 Thyroid hormone resistance, selective pituitary, 145650
TIA1	123.5	96%	83%	Welander distal myopathy,604454
TIMM8A	17.5	64%	30%	Mohr-Tranebjaerg syndrome, 304700
TIMP3	160.3	100%	100%	Sorsby fundus dystrophy, 136900
TINF2	156.3	100%	100%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TJP2	108.5	99%	99%	Cholestasis, progressive familial intrahepatic 4, 615878 Hypercholanemia, familial, 607748
TK2	99.5	93%	90%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560
TLL1	146.7	100%	99%	Atrial septal defect 6, 613087
TLR4	131	99%	91%	Endotoxin hyporesponsiveness {Colorectal cancer,susceptibility to},114500 {Macular degeneration,age-related,10},611488
TMC1	136.6	95%	94%	Deafness,autosomal dominant 36,606705 Deafness, autosomal recessive 7, 600974
TMC6	66	97%	92%	Epidermodysplasia verruciformis, 226400
TMC8	102.2	96%	88%	Epidermodysplasia verruciformis, 226400
TMCO1	81	100%	98%	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 614132
TMEM126A	108.6	99%	91%	Optic atrophy-7, 612989
TMEM138	102.7	99%	97%	Joubert syndrome 16, 614465
TMEM165	107.3	99%	96%	Congenital disorder of glycosylation, type IIk, 614727
TMEM216	129.4	100%	97%	Joubert syndrome 2, 608091 Meckel syndrome 2,603194
TMEM231	86.1	99%	95%	Joubert syndrome 20, 614970 Meckel syndrome 11,615397
TMEM237	96.1	99%	97%	Joubert syndrome 14, 614424

TMEM38B	120.6	100%	100%	Osteogenesis imperfecta,type XIV,615066
TMEM43	116.8	100%	98%	Arrhythmogenic right ventricular dysplasia 5, 604400 Emery-Dreifuss muscular dystrophy 7, AD, 614302
TMEM5	111.8	93%	89%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
TMEM67	70.3	91%	84%	COACH syndrome,216360 Joubert syndrome 6,610688 Meckel syndrome 3,607361 Nephronophthisis 11,613550 {Bardet-Biedl syndrome 14,modifier of},209900
TMEM70	121.2	94%	90%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMIE	100.5	96%	87%	Deafness, autosomal recessive 6, 600971
TMLHE	63.4	100%	86%	Epsilon-trimethyllysine hydroxylase deficiency, 300872
TMPRSS15	125.6	94%	89%	Enterokinase deficiency,226200
TMPRSS3	111.4	99%	97%	Deafness, autosomal recessive 8/10, 601072
TMPRSS6	93.9	100%	98%	Iron-refractory iron deficiency anemia, 206200
TNC	166.6	99%	99%	Deafness, autosomal dominant 56, 615629
TNFRSF10B	128.2	100%	100%	Squamous cell carcinoma,head and neck,275355
TNFRSF11A	127.3	92%	89%	Osteolysis, familial expansile, 174810 Paget disease of bone, 602080 Osteopetrosis, autosomal recessive 7, 612301
TNFRSF11B	240.7	100%	100%	Paget disease of bone 5, juvenile-onset,239000
TNFRSF13B	95	100%	100%	Immunoglobulin A deficiency 2, 609529
TNFRSF13C	47.4	80%	59%	Immunodeficiency, common variable, 4, 613494
TNFRSF1A	80.4	91%	88%	Periodic fever, familial, 142680
TNFSF11	172.9	100%	90%	Osteopetrosis,autosomal recessive 2,259710
TNNC1	187.5	100%	100%	Cardiomyopathy, dilated, 1Z, 611879 Cardiomyopathy, familial hypertrophic, 13, 613243
TNNI2	99.5	100%	99%	Arthrogyrosis multiplex congenita, distal, type 2B, 601680
TNNI3	89.6	100%	84%	Cardiomyopathy, dilated, 1FF, 613286 Cardiomyopathy, dilated, 2A, 611880 Cardiomyopathy, familial hypertrophic, 7, 613690 Cardiomyopathy, familial restrictive, 115210
TNNT1	86	97%	93%	Nemaline myopathy 5, Amish type, 605355

TNNT2	100.7	100%	100%	Cardiomyopathy, dilated, 1D, 601494 Cardiomyopathy, familial hypertrophic, 2, 115195 Cardiomyopathy, familial restrictive, 3, 612422 Left ventricular noncompaction 6, 601494
TNNT3	95.2	99%	92%	Arthrogyryposis,distal,type 2B,601680
TNXB	13.3	48%	20%	Ehlers-Danlos syndrome due to tenascin X deficiency,606408 Vesicoureteral reflux 8,615963
TOP1	87.3	100%	97%	DNA topoisomerase I,camptothecin-resistant
TOP2A	131.3	98%	91%	DNA topoisomerase II,resistance to inhibition of,by amsacrine
TOPORS	234.8	100%	100%	Retinitis pigmentosa 31, 609923
TOR1A	211.4	100%	100%	Dystonia-1, torsion, 128100 Dystonia, early-onset atypical, with myoclonic features {Dystonia-1, modifier of}
TP53	86.8	98%	91%	Adrenal cortical carcinoma, 202300 Breast cancer,114480 Choroid plexus papilloma, 260500 Colorectal cancer, 114500 Hepatocellular carcinoma, 114550 Li-Fraumeni syndrome, 151623 Nasopharyngeal carcinoma,607107 Osteosarcoma, 259500 Pancreatic cancer,260350 {Basal cell carcinoma 7},614740 {Glioma susceptibility 1},137800  Nasopharyngeal carcinoma, 607107 Pancreatic cancer, 260350
TP63	186.5	100%	100%	ADULT syndrome,103285 Ectrodactyly,ectodermal dysplasia,cleft lip/palate syndrome 3,604292 Hay-Wells syndrome,106260 Limb-mammary syndrome,603543 Orofacial cleft 8,129400 Rapp-Hodgkin syndrome,129400 Split-hand/foot malformation 4,605289
TPI1	85.5	98%	95%	Hemolytic anemia due to triosephosphate isomerase deficiency

TPK1	117	100%	95%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TPM1	135.4	100%	97%	Cardiomyopathy, dilated, 1Y, 611878 Cardiomyopathy, familial hypertrophic, 3, 115196 Left ventricular noncompaction 9, 611878
TPM2	104	100%	97%	Arthrogryposis, distal, type 2B, 601680 Arthrogryposis multiplex congenita, distal, type 1, 108120 CAP myopathy 2, 609285 Nemaline myopathy 4, autosomal dominant, 609285
TPM3	91.3	89%	88%	CAP myopathy 1, 609284 Myopathy congenital, with fiber-type disproportion, 255310 Nemaline myopathy 1, autosomal dominant or recessive, 609284
TPMT	44.5	93%	80%	6-mercaptopurine sensitivity, 610460
TPO	126.1	100%	95%	Thyroid dyshormonogenesis 2A,274500
TPP1	124	100%	100%	Ceroid lipofuscinosis, neuronal, 2, 204500
TPRN	55.4	81%	74%	Deafness, autosomal recessive 79, 613307
TRAPPC11	121	96%	95%	Muscular dystrophy, limb-girdle, type 2S, 615356
TRAPPC2	71.5	68%	57%	Spondyloepiphyseal dysplasia tarda,313400
TRAPPC9	124	100%	99%	Mental retardation, autosomal recessive 13, 613192
TRDN	68	80%	64%	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441
TREM2	100.2	99%	91%	Nasu-Hakola disease,221770
TREX1	214.3	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	241	100%	99%	Thyrotropin-releasing hormone resistance,generalized
TRIM24	118.8	94%	84%	No OMIM phenotype
TRIM32	134.2	100%	100%	Bardet-Biedl syndrome 11, 209900 Muscular dystrophy, limb-girdle, type 2H, 254110
TRIM33	120.7	94%	90%	No OMIM phenotype
TRIM37	112.2	99%	97%	Mulibrey nanism,253250
TRIOBP	104.5	96%	94%	Deafness, autosomal recessive 28, 609823
TRIP11	84.4	94%	88%	Achondrogenesis,type IA,200600
TRMU	91.6	100%	97%	{Deafness, mitochondrial, modifier of}, 580000 Liver failure, transient infantile, 613070

TRPA1	88.6	90%	83%	Episodic pain syndrome,familial,615040
TRPC6	114.3	96%	90%	Glomerulosclerosis, focal segmental, 2, 603965
TRPM1	161.4	100%	98%	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TRPM4	94.9	99%	94%	Progressive familial heart block, type IB, 604559
TRPM6	145.2	100%	99%	Hypomagnesemia 1, intestinal,602014
TRPS1	171.7	100%	100%	Trichorhinophalangeal syndrome,type I,190350 Trichorhinophalangeal syndrome,type III,190351
TRPV3	157.8	100%	99%	?Palmoplantar keratoderma,nonepidermolytic,focal 2,616400 Olmsted syndrome,614594
TRPV4	173.3	99%	98%	Brachyolmia type 3, 113500 Hereditary motor and sensory neuropathy, type IIc, 606071 Metatropic dysplasia, 156530 Scapuloperoneal spinal muscular atrophy, 181405 Spondylometaphyseal dysplasia, Kozlowski type, 184252 [Sodium serum level QTL 1], 613508
TSC1	127.2	99%	97%	Focal cortical dysplasia, Taylor balloon cell type, 607341 Lymphangioliomyomatosis, 606690 Tuberous sclerosis-1, 191100
TSC2	111.2	99%	98%	Lymphangioliomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254
TSEN2	112.6	100%	100%	Pontocerebellar hypoplasia type 2B,612389
TSEN34	47.3	84%	82%	Pontocerebellar hypoplasia type 2C,612390
TSEN54	70.9	90%	77%	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753
TSFM	117.4	100%	100%	Combined oxidative phosphorylation deficiency 3, 610505
TSG101	105.7	100%	97%	Breast cancer,somatic,114480
TSHB	337.5	100%	100%	Hypothyroidism,congenital,nongoitrous 4,275100
TSHR	208.2	100%	99%	Hyperthyroidism,congenital,nongoitrous,1,275200 Hyperthyroidism,familial gestational,603373 Hyperthyroidism,nonautoimmune,609152 Thyroid adenoma,hyperfunctioning,somatic Thyroid carcinoma with thyrotoxicosis
TSHZ1	150.3	98%	98%	Aural atresia,congenital,607842
TSPAN12	135.9	100%	98%	Exudative vitreoretinopathy 5, 613310



TSPAN7	84.2	100%	100%	Mental retardation, X-linked 58, 300210
TSPEAR	128.2	100%	99%	Deafness, autosomal recessive 98, 614861
TSPYL1	111.7	100%	100%	Sudden infant death with dysgenesis of the testes syndrome,608800
TTBK2	144.9	100%	99%	Spinocerebellar ataxia 11, 604432
TTC19	90.6	89%	82%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTC21B	94.2	98%	94%	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly,613819
TTC37	127.9	99%	97%	Trichohepatoenteric syndrome 1, 222470
TTC7A	101.9	99%	95%	Intestinal atresia, multiple, 243150
TTC8	88.4	98%	88%	?Retinitis pigmentosa 51,613464 Bardet-Biedl syndrome 8, 615985
TTI2	92.8	99%	97%	Mental retardation, autosomal recessive 39, 615541
TTN	196	98%	97%	Cardiomyopathy,dilated,1G,604145 Cardiomyopathy,familial hypertrophic,9,613765 Muscular dystrophy,limb-girdle,type 2J,608807 Myopathy,early-onset,with fatal cardiomyopathy,611705 Myopathy,proximal,with early respiratory muscular involvement,603689 Tibial muscular dystrophy,tardive,600334
TTPA	104.9	85%	67%	Ataxia with isolated vitamin E deficiency, 277460
TTR	138.6	100%	100%	Amyloidosis,hereditary,transthyretin-related,105210 Carpal tunnel syndrome,familial,115430 [Dystransthyretinemic hyperthyroxinemia],145680
TUBA1A	105.3	99%	93%	Lissencephaly 3, 611603
TUBA8	151.5	100%	99%	Polymicrogyria with optic nerve hypoplasia, 613180
TUBB1	152.4	100%	100%	Macrothrombocytopenia,autosomal dominant,TUBB1-related,613112
TUBB2A	102	99%	95%	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB2B	88.7	100%	100%	Polymicrogyria, symmetric or asymmetric, 610031
TUBB3	181	97%	97%	Cortical dysplasia,complex,with other brain malformations,614039 Fibrosis of extraocular muscles,congenital,3A,600638
TUBB4A	99.9	96%	92%	Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438
TUBG1	155.1	100%	100%	Cortical dysplasia,complex,with other brain malformations 4,615412
TUBGCP6	124.7	100%	97%	Microcephaly and chorioretinopathy, autosomal recessive 1, 251270
TUFM	119	100%	96%	Combined oxidative phosphorylation deficiency 4, 610678

TULP1	88.6	98%	93%	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132
TUSC3	130	100%	98%	Mental retardation, autosomal recessive 7, 611093
TWIST1	104.6	97%	87%	Craniosynostosis, type 1, 123100 Robinow-Sorauf syndrome, 180750 Saethre-Chatzen syndrome, 101400 Saethre-Chatzen syndrome with eyelid anomalies, 101400
TWIST2	122.6	99%	91%	Ablepharon-macrostomia syndrome,200110 Barber-Say syndrome,209885 Focal facial dermal dysplasia 3,Setleis type,227260
TYK2	105.4	99%	98%	Tyrosine kinase 2 deficiency, 611521
TYMP	74.9	92%	69%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYR	189.7	100%	100%	Albinism, oculocutaneous, type IA, 203100 Albinism, oculocutaneous, type IB, 606952 Waardenburg syndrome/albinism, digenic, 103470 [Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800
TYROBP	79.5	100%	100%	Nasu-Hakola disease,221770
TYRP1	190.1	100%	100%	Albinism, oculocutaneous, type III, 203290 Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair), 612271
UBA1	97.2	97%	96%	Spinal muscular atrophy, X-linked 2, infantile, 301830
UBE2A	62.6	96%	91%	Mental retardation, X-linked syndromic, Nascimento-type, 300860
UBE3A	92.8	97%	92%	Angelman syndrome, 105830
UBE3B	121.1	100%	99%	Blepharophimosis-ptosis-intellectual disability syndrome, 615057
UBIAD1	225.9	98%	93%	Corneal dystrophy, Schnyder type, 121800
UBQLN2	80.9	100%	93%	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857
UBR1	130.1	99%	95%	Johanson-Blizzard syndrome, 243800
UGT1A1	209.5	100%	100%	Crigler-Najjar syndrome, type I, 218800 Crigler-Najjar syndrome, type II, 606785 Hyperbilirubinemia, familial transient neonatal, 237900 [Bilirubin, serum level of, QTL1], 601816 [Gilbert syndrome], 143500
UMOD	109.9	97%	95%	Glomerulocystic kidney disease with hyperuricemia and isotheruria,609886 Hyperuricemic nephropathy, familial juvenile 1, 162000 Medullary cystic kidney disease 2,603860

UMPS	162.2	100%	97%	Orotic aciduria, 258900
UNC13D	87.8	97%	95%	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC93B1	59	55%	53%	s simplex encephalitis, susceptibility to, 1, 610551
UNG	61.1	100%	96%	Immunodeficiency with hyper IgM, type 5, 608106
UPB1	151.8	100%	100%	Beta-ureidopropionase deficiency, 613161
UPF3B	35.5	88%	64%	Mental retardation, X-linked, syndromic 14, 300676
UQCRB	101.6	99%	96%	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC2	137.1	100%	99%	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRQ	121.5	100%	100%	Mitochondrial complex III deficiency, nuclear type 4, 615159
UROC1	133.1	100%	100%	Urocanase deficiency, 276880
UROD	143.9	100%	99%	Porphyria cutanea tarda, 176100 Porphyria, hepatoerythropoietic, 176100
UROS	97.3	100%	100%	Porphyria, congenital erythropoietic, 263700
USB1	120.7	100%	98%	Poikiloderma with neutropenia, 604173
USH1C	99.1	98%	95%	Acadian and Samaritan variety Usher syndrome, type 1C, 276904 Deafness, autosomal recessive 18A, 602092
USH1G	153.6	95%	93%	Usher syndrome, type 1G, 606943
USH2A	156.8	99%	98%	Retinitis pigmentosa 39, 613809 Usher syndrome, type 2A, 276901
USP9Y	63.6	95%	82%	Spermatogenic failure,Y-linked,415000
UVSSA	106.4	100%	98%	UV-sensitive syndrome 3,614640
VANGL1	149.6	100%	100%	Caudal regression syndrome,600145 {Neural tube defects,susceptibility to},182940
VANGL2	131.5	100%	98%	Neural tube defects,182940
VAPB	111.4	99%	99%	Amyotrophic lateral sclerosis 8, 608627 Spinal muscular atrophy, late-onset, Finkel type, 182980
VAX1	44.7	87%	70%	Microphthalmia, syndromic 11, 614402
VCAN	185.9	100%	100%	Wagner syndrome 1, 143200
VCL	101.4	99%	97%	Cardiomyopathy, dilated, 1W, 611407 Cardiomyopathy, familial hypertrophic, 15, 613255
VCP	147	100%	99%	Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954 Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320
VDR	114.9	97%	94%	Rickets,vitamin D-resistant,type IIA,277440 ?Osteoporosis,involutional,166710

VHL	91.1	98%	75%	Erythrocytosis, familial, 2, 263400 Hemangioblastoma, cerebellar, somatic Pheochromocytoma, 171300 Renal cell carcinoma, somatic, 144700 von Hippel-Lindau syndrome, 193300
VIM	117	100%	93%	Cataract 30, pulverulent, 116300
VIPAS39	135.4	100%	100%	Arthrogyryposis, renal dysfunction, and cholestasis 2, 613404
VKORC1	132.8	100%	100%	Vitamin K-dependent clotting factors,combined deficiency of,2,607473 Warfarin resistance,122700
VLDLR	205.3	100%	98%	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VPS13A	72.3	92%	85%	Choreoacanthocytosis, 200150
VPS13B	140	98%	97%	Cohen syndrome, 216550
VPS33B	132.2	100%	99%	Arthrogyryposis, renal dysfunction, and cholestasis 1, 208085
VPS35	89.8	97%	89%	{Parkinson disease 17},614203
VPS37A	73.7	87%	67%	Spastic paraplegia 53, autosomal recessive, 614898
VPS45	128.3	95%	95%	Neutropenia, severe congenital, 5, autosomal recessive, 615285
VRK1	127.4	100%	97%	Pontocerebellar hypoplasia type 1A,607596
VSX1	54.9	91%	73%	Corneal dystrophy, posterior polymorphous, 1, 122000 Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195 Keratoconus 1, 148300
VSX2	67.6	98%	88%	Microphthalmia, isolated 2, 610093 Microphthalmia with coloboma 3, 610092
VWF	107.4	99%	98%	von Willebrand disease, type 1,193400 von Willebrand disease,type 2A,2B,2M and 2N,613554 von Willebrand disease,type 3,277480
WAS	35.9	81%	62%	Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, 313900 Thrombocytopenia, X-linked, intermittent, 313900 Wiskott-Aldrich syndrome, 301000
WDPCP	107.1	94%	93%	?Bardet-Biedl syndrome 15, 615992
WDR11	129.2	96%	96%	Hypogonadotropic hypogonadism 14 with or without anosmia, 614858
WDR19	133.5	100%	98%	?Cranioectodermal dysplasia 4,614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly,614376 Nephronophtthisis 13,614377

				Senior-Loken syndrome 8,616307
WDR34	91.1	94%	89%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
WDR35	143.9	97%	94%	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly,614091
WDR36	110.9	99%	90%	Glaucoma 1,open angle,G,609887
WDR45	50.2	94%	89%	Neurodegeneration with brain iron accululation 5, 300894
WDR60	103.3	98%	94%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WDR62	142.7	99%	97%	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
WDR72	145.6	99%	98%	Amelogenesis imperfecta hypomaturation type IIA3,613211
WDR81	135.5	99%	99%	Cerebellar ataxia, mental retardation and dysequilibrium syndrome 2, 610185
WFS1	218.6	98%	97%	?Cataract 41,116400 Deafness,autosomal dominant 6/14/38,600965 Wolfram syndrome,222300 Wolfram-like syndrome,autosomal dominant,614296 {Diabetes mellitus,noninsulin-dependent,association with},125853
WHSC1L1	122	100%	99%	Leukemia,acute myeloid,601626
WIPF1	69.4	100%	97%	Wiskott-Aldrich syndrome 2, 614493
WISP3	100.9	100%	100%	Arthropathy,progressive pseudorheumatoid,of childhood,208230 Spondyloepiphyseal dysplasia tarda with progressive arthropathy,208230
WNK1	159.4	98%	97%	Neuropathy,hereditary sensory and autonomic type II,201300 Pseudohypoaldosteronism, type IIC, 614492
WNK4	107.1	97%	92%	Pseudohypoaldosteronism, type IIB, 614491
WNT1	159.8	100%	100%	Osteogenesis imperfecta,type XV,615220 {Osteoporosis,early-onset,susceptibility to,autosomal dominant,615221
WNT10A	90.5	100%	96%	Odontoonychodermal dysplasia,257980 Schopf-Schulz-Passarge syndrome,224750 Tooth agenesis,selective,4,150400
WNT10B	106.1	100%	100%	Split-hand/foot malformation 6,225300
WNT3	144	97%	92%	?Tetra-amelia syndrome,273395
WNT4	243.3	92%	92%	Mullerian aplasia and hyperandrogenism,158330 SERKAL syndrome, 611812
WNT5A	138.5	100%	100%	Robinow syndrome autosomal dominant,180700

WNT7A	198.8	100%	100%	Fuhrmann syndrome,228930 Ulna and fibula,absence of,with severe limb deficiency,276820
WRAP53	127.4	100%	100%	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	128.3	97%	94%	Werner syndrome, 277700
WT1	84.4	92%	85%	Denys-Drash syndrome, 194080 Frasier syndrome, 136680 Meacham syndrome, 608978 Mesothelioma, somatic, 156240 Nephrotic syndrome, type 4, 256370 Wilms tumor, type 1, 194070
WWOX	122.5	100%	99%	Epileptic encephalopathy, early infantile, 28, 616211 Esophageal squamous cell carcinoma, somatic, 133239 Spinocerebellar ataxia, autosomal recessive, 12, 614322
XDH	100.5	100%	99%	Xanthinuria, type I, 278300
XIAP	67.6	86%	79%	Lymphoproliferative syndrome, X-linked, 2, 300635
XIST				X-inactivation,familial skewed,300087
XK	60.4	100%	97%	McLeod syndrome with or without chronic granulomatous disease,300842
XPA	46.6	95%	84%	Xeroderma pigmentosum, group A, 278700
XPC	148.7	100%	100%	Xeroderma pigmentosum, group C, 278720
XPNPEP3	128.2	97%	97%	Nephronophthisis-like nephropathy 1, 613159
YAP1	95.5	85%	79%	Coloboma, ocular with or without hearing impairment, cleft lip/palate and mental retardation, 120433
YARS	119.6	100%	100%	Charcot-Marie-Tooth disease, dominant intermediate C, 608323
YARS2	157.3	99%	98%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
ZAP70	185.8	100%	99%	Selective T-cell defect, 269840
ZBTB16	154.8	100%	100%	Leukemia, acute promyelocytic, PL2F/RARA type Skeletal defects, genital hypoplasia, and mental retardation, 612447
ZBTB24	151.4	100%	100%	Immunodeficiency-centromeric instability-facial anomalies syndrome-2, 614069
ZC4H2	56.3	100%	92%	Wieacker-Wolff syndrome,314580
ZDHH9	32.9	93%	74%	Mental retardation, X-linked syndromic, Raymond type, 300799
ZEB1	188	100%	99%	Corneal dystrophy, Fuchs endothelial, 6, 613270 Corneal dystrophy, posterior polymorphous, 3, 609141
ZEB2	149.6	100%	98%	Mowat-Wilson syndrome, 235730
ZFP57	12.1	60%	21%	Diabetes mellitus,transient neonatal,1,601410

ZFPM2	188.7	100%	100%	Diaphragmatic hernia 3, 610187 Tetralogy of Fallot, 187500
ZFYVE26	110.3	99%	98%	Spastic paraplegia 15, autosomal recessive, 270700
ZFYVE27	114.7	100%	100%	Spastic paraplegia 33, autosomal dominant, 610244
ZIC2	82.9	88%	76%	Holoprosencephaly-5, 609637
ZIC3	71.1	100%	99%	Congenital heart defects, nonsyndromic, 1, X-linked, 306955 Heterotaxy, visceral, 1, X-linked 306955 VACTERL association, X-linked, 314390
ZMPSTE24	125.2	100%	97%	Mandibuloacral dysplasia with type B lipodystrophy,608612 Restrictive dermopathy,lethal,275210
ZMYND10	135	100%	100%	Ciliary dyskinesia, primary, 22, 615444
ZNF335	132.1	99%	98%	?Microcephaly 10,primary,autosomal recessive,615095
ZNF423	226.6	100%	100%	Joubert syndrome 19,614844 Nephronophthisis 14, 614844
ZNF469	62.1	98%	94%	Brittle cornea syndrome 1,229200
ZNF513	90.9	100%	97%	Retinitis pigmentosa 58, 613617
ZNF592	109.2	99%	98%	Spinocerebellar ataxia, autosomal recessive 5, 606937
ZNF644	163.1	100%	100%	Myopia 21, autosomal dominant, 614167
ZNF711	107.2	100%	96%	Mental retardation, X-linked 97, 300803
ZNF750	130.7	100%	100%	Seborrhea-like dermatitis with psoriasiform elements,610227
ZNF81	58.1	95%	89%	Mental retardation, X-linked 45, 300498

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

Median Coverage describes the average number of reads seen across 50 exomes.

% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.

% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.

Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.

OMIM release used for OMIM disease identifiers and descriptions : April 10th, 2016.

This list is accurate for panel versions DG 2.5 and DG 2.6 No changes were made to the composition of the gene panel between these versions.

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors