# MENDELIOME GENE PANEL DG 2.5/2.6

| Gene | Median coverage | % covered > 10x | % covered > 20x | Associated Phenotype description and OMIM disease ID |
|---|---|---|---|---|
| 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<br>Epileptic encephalopathy,early infantile,29,616339 |
| AARS2 | 111 | 99% | 97% | Combined oxidative phosphorylation deficiency 8, 614096 |
| AASS | 131.8 | 100% | 95% | Hyperlysinemia, 238700<br>Saccharopinuria, 268700 |
| ABAT | 86.7 | 100% | 97% | GABA-transaminase deficiency, 613163 |
| ABCA1 | 119.2 | 99% | 97% | HDL deficiency,type 2,604091<br>Tangier disease,205400<br>{Coronary artery disease in familial hypercholesterolemia,protection against},143890 |
| ABCA12 | 145.7 | 98% | 96% | Ichthyosis, autosomal recessive 4B (harlequin),242500<br>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<br>Fundus flavimaculatus, 248200<br>Retinal dystrophy, early-onset severe, 248200<br>Retinitis pigmentosa 19, 601718<br>Stargardt disease 1, 248200<br>{Macular degeneration, age-related, 2,} 153800 |
| ABCB11 | 161.6 | 100% | 97% | Cholestasis,benign recurrent intrahepatic,2,605479<br>Cholestasis,progressive familial intrahepatic 2,601847 |
| ABCB4 | 126.3 | 99% | 96% | Cholestasis,intrahepatic,of pregnancy,3,614972<br>Cholestasis,progressive familial intrahepatic 3,602347<br>Gallbladder disease 1,600803 |
| ABCB6 | 111.6 | 100% | 99% | Dyschromatosis universalis hereditaria 3,615402<br>Microphthalmia,isolated, with coloboma 7,614497<br>[Blood group, Langereis system],111600 |

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| 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<br>Pseudoxanthoma elasticum,264800<br>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<br>Cardiomyopathy, dilated, 1O, 608569<br>Hypertrichotic osteochondrodysplasia, 239850 |
| ABCD1 | 49.6 | 69% | 64% | Adrenoleukodystrophy, 300100<br>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<br>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 imatrinib |
| 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<br>Spondyloepimetaphyseal dysplasia, aggrecan type, 612813<br>Spondyloepiphyseal dysplasia, Kimberley type, 608361 |
| ACAT1 | 120.3 | 98% | 93% | Alpha-methylacetoacetic aciduria, 203750 |
| ACE | 106.4 | 93% | 91% | Renal tubular dysgenesis,267430<br>[Angiotensin I-converting enzyme,benign serum increase]<br>{Alzheimer disease,susceptibility to},104300<br>{Microvascular complications of diabetes 3},612624<br>{Myocardial infarction,susceptibility to}<br>{SARS,progression of} |

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| | | | | {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<br>Myelogenous leukemia,acute |
| ACTA1 | 92.7 | 99% | 92% | ?Myopathy,scapulohumeroperoneal,616852<br>Myopathy, actin, congenital, with cores, 161800<br>Myopathy, actin, congenital, with excess of thin myofilaments, 161800<br>Myopathy, congenital, with fiber-type disproportion 1, 255310<br>Nemaline myopathy 3, autosomal dominant or recessive, 161800 |
| ACTA2 | 150.7 | 100% | 100% | Aortic aneurysm familial thoracic 6,611788<br>Moyamoya disease 5,614042<br>Multisystemic smooth muscle dysfunction syndrome,613834 |
| ACTB | 106.7 | 100% | 93% | ?Dystonia, juvenile-onset, 607371<br>Baraitser-Winter syndrome 1, 243310 |
| ACTC1 | 154.8 | 100% | 100% | Atrial septal defect 5, 612794<br>Cardiomyopathy, dilated, 1R, 613424<br>Cardiomyopathy, familial hypertrophic, 11, 612098<br>Left ventricular noncompaction 4, 613424 |
| ACTG1 | 110 | 100% | 100% | Baraitser-Winter syndrome 2, 614583<br>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 |

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| 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<br>Severe combined immunodeficiency due to ADA deficiency, 102700 |
| ADAM10 | 141.1 | 99% | 97% | Reticulate acropigmentation of Kitamura,615537<br>{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<br>Ectopia lentis,isolated,autosomal recessive,225100 |
| ADAR | 107.2 | 99% | 98% | Aicardi-Goutieres syndrome 6, 615010<br>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<br>{Asthma, nocturnal, susceptibility to}, 600807<br>{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<br>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 |

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| AGL | 147.4 | 100% | 99% | Glycogen storage disease IIIa, 232400<br>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<br>{Hypertension, essential, susceptibility to}, 145500<br>{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<br>Cowchock syndrome, 310490 |
| AIMP1 | 82.2 | 95% | 86% | Leukodystrophy, hypomyelinating, 3, 260600 |
| AIP | 150.2 | 100% | 97% | Pituitary adenoma,ACTH-secreting,219090<br>Pituitary adenoma, growth hormone-secreting,102200<br>Pituitary adenoma,prolactin-secreting,600634 |
| AIPL1 | 111.6 | 100% | 100% | Cone-rod dystrophy, 604393<br>Leber congenital amaurosis 4, 604393<br>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<br>Obesity,hyperphagia and developmental delay |
| AKR1D1 | 104.3 | 96% | 91% | Bile acid synthesis defect, congenital, 2, 235555 |

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| AKT1 | 146.7 | 99% | 95% | Breast cancer somatic,114480<br>Colorectal cancer, somatic,114500<br>Cowden syndrome 6,615109<br>Ovarian cancer, somatic,167000<br>Proteus syndrome, somatic,176920<br>{Schizophrenia, susceptibility to},181500 |
| AKT2 | 134.3 | 100% | 98% | Diabetes mellitus,type II,125853<br>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<br>{Lead poisoning, susceptibility to,} 612740 |
| ALAS2 | 63.7 | 97% | 91% | Anemia, sideroblastic, X-linked, 300751<br>Protoporphyria, erythropoietic, X-linked, 300752 |
| ALB | 159.6 | 100% | 99% | Analbuminemia,616000<br>[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<br>{Esophageal cancer, alcohol-related, susceptibility to}<br>{Hangover, susceptibility to, 610251}<br>{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 |

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| 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 Il, 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<br>Hypophosphatasia, childhood, 241510<br>Hypophosphatasia, infantile, 241500<br>Odontohypophosphatasia, 146300 |
| ALS2 | 166.3 | 100% | 99% | Amyotrophic lateral sclerosis 2,juvenile,205100<br>Primary lateral sclerosis, juvenile, 606353<br>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<br>Parietal foramina 2, 609597<br>{Craniosynostosis 5,susceptibility to},615529 |
| AMACR | 132.1 | 100% | 99% | Alpha-methylacyl-CoA racemase deficiency, 614307<br>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<br>Long QT syndrome-4, 600919 |
| ANKH | 104.1 | 100% | 100% | Chondrocalcinosis 2, 118600<br>Craniometaphyseal dysplasia, 123000 |

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| 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<br>Miyoshi muscular dystrophy 3, 613319<br>Muscular dystrophy, limb-girdle, type 2L, 611307 |
| ANO6 | 125.7 | 97% | 94% | Scott syndrome,262890 |
| ANTXR1 | 127.4 | 97% | 96% | GAPO syndrome, 230740<br>{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<br>Adenomatous polyposis coli, 175100<br>Brain tumor-polyposis syndrome 2, 175100<br>Colorectal cancer, somatic, 114500<br>Desmoid disease, hereditary, 135290<br>Gardner syndrome,175100<br>Gastric cancer, somatic, 613659<br>Hepatoblastoma, somatic, 114550 |
| APCDD1 | 162.2 | 100% | 97% | Hypotrichosis 1,605389 |
| APOA1 | 90 | 100% | 100% | Amyloidosis,3 or more types,105200<br>ApoA-I and ApoC-III deficiency,combined<br>Corneal clouding,autosomal recessive |

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| | | | | Hypoalphalipoproteinemia,604091 |
| APOA2 | 91.3 | 81% | 81% | Apolipoprotein A-II deficiency<br>{Hypercholesterolemia,familial,modifier of},143890 |
| APOA5 | 120.8 | 100% | 100% | Hyperchylomicronemia,late-onset,144650<br>{Hypertriglyceridemia,susceptibility to},145750 |
| APOB | 181.6 | 99% | 99% | Hypercholesterolemia,due to ligand-defective apo B,144010<br>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<br>Hyperlipoproteinemia,type III<br>Lipoprotein glomerulopathy,611771<br>Sea-blue histiocyte disease,269600<br>{?Macular degeneration,age-related},603075<br>{Myocardial infarction sisceptibility} |
| APP | 149.5 | 100% | 100% | Alzheimer disease 1,familial,104300<br>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<br>Androgen insensitivity,partial,with/without breast cancer,312300<br>Hypospadias 1,X-linked,300633<br>Spinal and bulbar muscular atrophy of Kennedy,313200<br>{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 |

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| ARHGEF12 | 143.5 | 98% | 97% | No OMIM phenotype<br>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<br>Retinitis pigmentosa 55, 613575<br>{Bardet-Biedl syndrome 1, modifier of}, 209900 |
| ARMC4 | 123.8 | 90% | 89% | Ciliary dyskinesia, primary, 23, 615451 |
| ARNT | 121 | 97% | 95% | No OMIM phenotype<br>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<br>Hydranencephaly with abnormal genitalia,300215<br>Lissencephaly, X-linked 2, 300215<br>Mental retardation, X-linked 29 and others, 300419<br>Partington syndrome, 309510<br>Proud syndrome, 300004 |
| ASAH1 | 118.4 | 96% | 87% | Farber lipogranulomatosis, 228000<br>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<br>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 |

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|---|---|---|---|---|
| ASS1 | 86.8 | 95% | 88% | Citrullinemia, 215700 |
| ASXL1 | 145 | 99% | 98% | Bohring-Opitz syndrome, 605039<br>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<br>Spastic paraplegia 3A, autosomal dominant, 182600 |
| ATL3 | 117.6 | 97% | 92% | Neuropathy,hereditary sensory,type IF,615632 |
| ATM | 112 | 98% | 95% | Ataxia-telangiectasia, 208900<br>Lymphoma, B-cell non-Hodgkin, somatic<br>Lymphoma, mantle cell<br>T-cell prolymphocytic leukemia, somatic<br>{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<br>Migraine, familial basilar, 602481<br>Migraine, familial hemiplegic, 2, 602481 |
| ATP1A3 | 169.4 | 100% | 100% | Alternating hemiplegia of childhood 2,614820<br>CAPOS syndrome,601338<br>Dystonia-12,128235 |
| ATP2A1 | 150.9 | 100% | 100% | Brody myopathy, 601003 |
| ATP2A2 | 166.4 | 100% | 99% | Acrokeratosis verruciformis, 101900<br>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<br>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<br>Occipital horn syndrome, 304150<br>Spinal muscular atrophy, distal, X-linked 3, 300489 |

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|---|---|---|---|---|
| ATP7B | 147.1 | 100% | 99% | Wilson disease, 277900 |
| ATP8B1 | 149.6 | 95% | 93% | Cholestasis, benign recurrent intrahepatic, 243300<br>Cholestasis, intrahepatic, of pregnancy, 1, 147480<br>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<br>Seckel syndrome 1, 210600 |
| ATRX | 54.3 | 96% | 88% | Alpha-thalassemia myelodysplasia syndrome, somatic, 300448<br>Alpha-thalassemia/mental retardation syndrome, 301040<br>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<br>{Amyotrophic lateral sclerosis,susceptibility to,13},183090<br>{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<br>Nephrogenic syndrome of inappropriate antidiuresis, 300539 |
| AXIN1 | 128.3 | 98% | 98% | ?Caudal duplication anomaly,607864<br>Hepatocellular carcinoma,somatic,114550 |
| AXIN2 | 111.4 | 100% | 99% | Colorectal cancer somatic,114500<br>Oligodontia-colorectal cancer syndrome,608615 |
| B2M | 319.4 | 100% | 100% | ?Amyloidosis,familial visceral,105200<br>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<br>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 |

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|---|---|---|---|---|
| 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<br>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<br>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<br>Lymphoma,MALT,somatic,137245<br>{Lymphoma,follicular,somatic},605027<br>{Male germ cell tumor,somatic},273300<br>{Mesothelioma,somatic},156240<br>{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 |

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|---|---|---|---|---|
| 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<br>Leukemia,chronic myeloid,somatic,608232 |
| BCS1L | 147 | 100% | 100% | Bjornstad syndrome, 262000<br>GRACILE syndrome, 603358<br>Leigh syndrome, 256000<br>Mitochondrial complex III deficiency, nuclear type 1, 124000 |
| BDNF | 174.6 | 100% | 100% | Central hypoventilation syndrome,congenital,209880<br>{Anorexia nervosa,susceptibility to},610269<br>{Bulimia nervosa,age of onset of weight loss in},607499<br>{Memory impairment,susceptibility to}<br>{Obsessive-compulsive disorder,protection against},164230 |
| BEAN1 | 117.9 | 94% | 85% | Spinocerebellar ataxia 31,117210 |
| BEST1 | 111.7 | 99% | 96% | Bestrophinopathy, 611809<br>Macular dystrophy,vitelliform,2,153700<br>Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma, 193220<br>Retinitis pigmentosa,concentric,613194<br>Retinitis pigmentosa 50,613194<br>Vitreoretinochoroidopathy,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<br>Premature ovarian failure 4,300510 |
| BMP2 | 166.2 | 100% | 100% | Brachydactyly, type A2, 112600 |

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| | | | | {HFE hemochromatosis, modifier of}, 235200 |
| BMP4 | 120.8 | 100% | 98% | Microphthalmia, syndromic 6, 607932<br>Orofacial cleft 11, 600625 |
| BMPER | 144.9 | 100% | 100% | Diaphanospondylodysostosis,608022 |
| BMPR1A | 98.6 | 99% | 89% | Juvenile polyposis syndrome, infantile form, 174900<br>Polyposis syndrome, hereditary mixed, 2, 610069<br>Polyposis, juvenile intestinal, 174900 |
| BMPR1B | 191.5 | 100% | 99% | Acromesomelic dysplasia,Demirhan type,609441<br>Brachydactyly,type A2,112600 |
| BMPR2 | 195.7 | 100% | 100% | Pulmonary hypertension, familial primary, 1, with or without HHT, 178600<br>Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600<br>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<br>Cardiofaciocutaneous syndrome, 115150<br>Colorectal cancer, somatic<br>LEOPARD syndrome 3, 613707<br>Melanoma, malignant, somatic<br>Nonsmall cell lung cancer, somatic<br>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<br>Wilms tumor, 194070<br>{Breast cancer, male, susceptibility to}, 114480<br>{Breast-ovarian cancer, familial, 2}, 612555<br>{Glioblastoma 3}, 613029<br>{Medulloblastoma}, 155255<br>{Pancreatic cancer 2},613347<br>{Prostate cancer}, 176807 |
| BRIP1 | 120.7 | 99% | 97% | ?Breast cancer, early-onset, 114480<br>Fanconi anemia, complementation group J, 609054 |
| BRWD3 | 65.9 | 96% | 90% | Mental retardation, X-linked 93, 300659 |

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|---|---|---|---|---|
| BSCL2 | 107.4 | 100% | 98% | Encephalopathy,progressive,with or without lipodystrophy,615924<br>Lipodystrophy, congenital generalized, type 2, 269700<br>Neuropathy, distal hereditary motor, type V, 600794<br>Silver spastic paraplegia syndrome, 270685 |
| BSND | 131.6 | 100% | 100% | Bartter syndrome, type 4a, 602522<br>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<br>Mosaic variegated aneuploidy syndrome 1, 257300<br>[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<br>Perrault syndrome 5,616138<br>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<br>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<br>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 |

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| 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<br>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<br>Migraine, familial hemiplegic, 1, 141500<br>Migraine, familial hemiplegic,1,with progressive cerebellar ataxia,141500<br>Spinocerebellar ataxia 6,183086 |
| CACNA1C | 145.4 | 99% | 98% | Brugada syndrome 3, 611875<br>Timothy syndrome, 601005 |
| CACNA1D | 143.9 | 100% | 99% | Sinoatrial node dysfunction and deafness, 614896 |
| CACNA1F | 62.7 | 98% | 91% | Aland Island eye disease, 300600<br>Cone-rod dystropy, X-linked, 3, 300476<br>Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071 |
| CACNA1S | 132.8 | 100% | 99% | Hypokalemic periodic paralysis, type 1, 170400<br>{Malignant hyperthermia susceptibility 5}, 601887<br>{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 |

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| CACNB4 | 110.2 | 95% | 94% | Episodic ataxia, type 5, 613855<br>{Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682<br>{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<br>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<br>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<br>Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749<br>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<br>Hypocalcemia,autosomal dominant,601198<br>Hypocalciuric hypercalcemia,type I,145980<br>{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<br>?Partial lipodystrophy, congenital cataracts and neurodegeneration syndrome,606721<br>Pulmonary hypertension, primary, 3,615343 |

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| CAV3 | 269.3 | 100% | 100% | Cardiomyopathy,familial hypertrophic,192600<br>Creatine phosphokinase,elevated serum,123320<br>Long QT syndrome 9, 611818<br>Muscular dystrophy,limb-girdle,type IC,607801<br>Myopathy,distal,Tateyama type,614321<br>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<br>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<br>Joubert syndrome 9,612285<br>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<br>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<br>[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 |

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| CD320 | 87.4 | 100% | 97% | Methylmalonic aciduria due to transcobalamin receptor defect,613646 |
| CD36 | 124.4 | 100% | 99% | Platelet glycoprotein IV deficiency,608404<br>{Coronary heart disease,susceptibility to,7},610938<br>{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<br>Hyperparathyroidism-jaw tumor syndrome, 145001<br>Parathyroid adenoma with cystic changes, 145001<br>Parathyroid carcinoma, 608266 |
| CDH1 | 107.7 | 99% | 98% | Endometrial carcinoma, somatic, 608089<br>Gastric cancer, familial diffuse, with or without cleft lip and/or palate, 137215<br>Ovarian carcinoma, somatic, 167000<br>{Breast cancer, lobular}, 114480<br>{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<br>Usher syndrome, type 1D, 601067<br>Usher syndrome, type 1D/F digenic, 601067 |
| CDH3 | 132.2 | 100% | 97% | Ectodermal dysplasia,ectrodactyly and macular dystrophy,225280<br>Hypotrichosis, congenital, with juvenile macular dystrophy,601553 |
| CDHR1 | 141.1 | 100% | 99% | Cone-rod dystrophy 15, 613660 |

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| | | | | 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<br>IMAGE syndrome, 614732 |
| CDKN2A | 56.4 | 93% | 91% | Melanoma and neural system tumor syndrome, 155755<br>Orolaryngeal cancer, multiple<br>Pancreatic cancer/melanoma syndrome, 606719<br>{Melanoma, cutaneous malignant, 2}, 155601 |
| CDON | 132.5 | 100% | 98% | Holoprosencephaly 11, 614226 |
| CDSN | 16.5 | 59% | 32% | Hypotrichosis 2,146520<br>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<br>Polyarteritis nodosa,childhood-onset,615688 |
| CEL | 110 | 84% | 79% | Homocystinuria, B6-responsive and nonresponsive types, 236200<br>Thrombosis, hyperhomocysteinemic, 236200 |
| CENPJ | 144.5 | 99% | 96% | Microcephaly 6, primary, autosomal recessive, 608393<br>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<br>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<br>Joubert syndrome 5,610188<br>Leber congenital amaurosis 10,611775<br>Meckel syndrome 4,611134<br>Senior-Loken syndrome 6,610189 |
| CEP41 | 83.4 | 97% | 91% | Joubert syndrome 15, 614464 |

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| 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% | Ichtyosis, congenital, autosomal recessive 9, 615023 |
| CES1 | 138 | 97% | 93% | Carboxylesterase 1 deficiency |
| CETP | 132.3 | 100% | 100% | Hyperalphalipoproteinemia,143470<br>[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<br>Cystic fibrosis, 219700<br>Sweat chloride elevation without CF<br>{Bronchiestasis with or without elevated sweat chloride 1, modifier of}, 211400<br>{Hypertrypsinemia, neonatal}<br>{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<br>Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 |
| CHEK2 | 99 | 82% | 80% | Li-Fraumeni syndrome, 609265<br>Osteosarcoma, somatic, 259500<br>{Breast cancer, susceptibility to}, 114480<br>{Prostate cancer, familial, susceptibility to}, 176807<br>{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<br>Dementia, familial, nonspecific, 600795 |
| CHMP4B | 125.3 | 99% | 96% | Cataract 31, multiple types, 605387 |

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|---|---|---|---|---|
| CHN1 | 143.6 | 97% | 96% | Duane retraction syndrome 2,604356 |
| CHRDL1 | 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<br>Myasthenic syndrome, fast-channel congenital, 60893<br>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<br>{Nicotine addiction,susceptibility to},188890 |
| CHRNB1 | 130.2 | 98% | 93% | Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931<br>Myasthenic syndrome, slow-channel congenital, 601462 |
| CHRNB2 | 249.4 | 100% | 100% | Epilepsy, nocturnal frontal lobe, 3, 605375 |
| CHRND | 144.5 | 99% | 96% | Multiple pterygium syndrome, lethal type, 253290<br>Myasthenic syndrome, slow-channel congenital, 601462<br>Myasthenic syndrome, fast-channel congenital, 608930 |
| CHRNE | 123.5 | 100% | 95% | Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931<br>Myasthenic syndrome, fast-channel congenital, 608930<br>Myasthenic syndrome, slow-channel congenital, 601462 |
| CHRNG | 147 | 100% | 100% | Escobar syndrome,26500<br>Multiple pterygium syndrome,lethal type,253290 |
| CHST14 | 155.8 | 94% | 92% | Ehlers-Danlos syndrome, musculocantractural 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<br>Usher syndrome, type IJ, 614869 |
| CIITA | 104.5 | 100% | 100% | Bare lymphocyte syndrome type II, complementation group A, 209920<br>{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<br>Atrial septal defect 8, 614433 |
| CLCF1 | 84.2 | 97% | 97% | Cold-induced sweating syndrome 2,610313 |

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| CLCN1 | 136.3 | 100% | 99% | Myotonia congenita, dominant, 160800<br>Myotonia congenita, recessive, 255700<br>Myotonia levior, recessive |
| CLCN2 | 96.1 | 98% | 98% | Leukoencephalopathy with ataxia,615651<br>{Epilepsy,idiopathic generalized,susceptibility to,11},607628 |
| CLCN5 | 89.5 | 95% | 89% | Dent disease, 300009<br>Hypophosphatemic rickets,300554<br>Nephrolithiasis,type I,310468<br>Proteinuria,low molecular weight,with hypercalciuric nephrocalcinosis,308990 |
| CLCN7 | 124.4 | 100% | 97% | Osteopetrosis,autosomal dominant 2,166600<br>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<br>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<br>Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300 |
| CLN8 | 206.6 | 100% | 100% | Ceroid lipofuscinosis, neuronal, 8, 600143<br>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<br>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 |

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| CNGB3 | 104.7 | 91% | 86% | Achromatopsia-3, 262300<br>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<br>Pitt-Hopkins like syndrome 1, 610042<br>{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<br>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<br>Marshall syndrome, 154780<br>Stickler syndrome, type II, 604841<br>{Lumbar disc herniation, susceptibility to}, 603932 |
| COL11A2 | 11 | 46% | 13% | Deafness, autosomal dominant 13, 601868<br>Deafness, autosomal recessive 53, 609706<br>Fibrochondrogenesis 2, 614524<br>Otospondylomegaepiphyseal dysplasia, 215150<br>Stickler syndrome, type III, 184840<br>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 |

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

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

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| COMP | 117.6 | 94% | 92% | Epiphyseal dysplasia,multiple,1,132400<br>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<br>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<br>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<br>Hemosiderosis, systemic, due to aceruloplasminemia, 604290<br>[Hypoceruloplasminemia, hereditary], 604290 |
| CPA6 | 110.9 | 100% | 100% | Epilepsy, familial temporal lobe, 5, 614417<br>Febrile seizures,familial,11,614418 |
| CPN1 | 107.1 | 97% | 94% | Carboxypeptidase N deficiency,212070 |
| CPOX | 96.6 | 84% | 79% | Coproporphyria, 121300<br>Harderoporphyria, 121300 |
| CPS1 | 148.8 | 100% | 99% | Carbamoylphosphate synthetase I deficiency, 237300<br>{Pulmonary hypertension, neonatal, susceptibility to}, 615371<br>{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<br>CPT II deficiency, lethal neonatal, 608836<br>Myopathy due to CPT II deficiency, 255110<br>{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 |

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|---|---|---|---|---|
| CRB1 | 199.9 | 100% | 99% | Leber congenital amaurosis 8, 613835<br>Pigmented paravenous chorioretinal atrophy, 172870<br>Retinitis pigmentosa-12, autosomal recessive, 600105 |
| CRBN | 135.5 | 100% | 97% | Mental retardation, autosomal recessive 2, 607417 |
| CREB1 | 136 | 100% | 93% | Histiocytoma,angiomatoid fibrous,somatic,612160 |
| CREBBP | 119.9 | 99% | 96% | Rubinstein-Taybi syndrome, 180849 |
| CRELD1 | 94 | 98% | 96% | Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217<br>{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<br>Leber congenital amaurosis 7, 613829 |
| CRYAA | 121.6 | 84% | 82% | Cataract 9, multiple types, 604219 |
| CRYAB | 120.2 | 97% | 95% | Cardiomyopathy, dilated, 1II, 615184<br>Cataract 16, multiple types, 613763<br>Myopathy, myofibrillar, 2, 608810<br>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 |

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| CSRP3 | 101 | 100% | 100% | Cardiomyopathy, dilated, 1M, 607482<br>Cardiomyopathy, familial hypertrophic, 12, 612124 |
| CST3 | 106.8 | 78% | 46% | Cerebral amyloid angiopathy,105150<br>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<br>Homocysteine, total plasma, elevated |
| CTHRC1 | 97.6 | 89% | 77% | Barrett esophagus/esophageal adenocarcinoma,614266 |
| CTNNA3 | 151 | 100% | 100% | Arrhytmogenic right ventricular dysplasia, familial, 13, 615616 |
| CTNNB1 | 163.2 | 100% | 100% | Colorectal cancer, somatic, 114500<br>Hepatocellular carcinoma, somatic, 114550<br>Mental retardation, autosomal dominant 19, 615075<br>Ovarian cancer, somatic, 167000<br>Pilomatricoma, somatic, 132600 |
| CTNS | 117.3 | 100% | 100% | Cystinosis, atypical nephropathic, 219800<br>Cystinosis, late-onset juvenile or adolescent nephropathic,219900<br>Cystinosis,ocular nonnephropathic,219750 |
| CTSA | 126.3 | 99% | 96% | Galactosialidosis, 256540 |
| CTSC | 128.6 | 100% | 100% | Haim-Munk syndrome, 245010<br>Papillon-Lefevre syndrome, 245000<br>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 |

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|---|---|---|---|---|
| CYB5R3 | 152.2 | 98% | 98% | Methemoglobinemia, type I, 250800<br>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<br>Cylindromatosis, familial, 132700<br>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<br>Aldosteronism, glucocorticoid-remediable, 103900 |
| CYP11B2 | 153.5 | 100% | 99% | Aldosterone to renin ratio raised<br>Hypoaldosteronism, congenital, due to CMO II deficiency, 610600<br>Hypoaldosteronism, congenital, due to CMO I deficiency, 203400<br>Low renin hypertension, susceptibility to |
| CYP17A1 | 132.3 | 100% | 99% | 17-alpha-hydroxylase/17,20-lyase deficiency, 202110<br>17,20-lyase deficiency, isolated, 202110 |
| CYP19A1 | 175.4 | 100% | 100% | Aromatase deficiency, 613546<br>Aromatase excess syndrome, 139300 |
| CYP1B1 | 106.3 | 100% | 99% | Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300<br>Peters anomaly, 604229 |
| CYP21A2 | 14.5 | 54% | 23% | Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910<br>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<br>{Lung cancer,resistance to},211980<br>{Nicotine addiction,protection from},188890 |
| CYP2B6 | 92.7 | 99% | 94% | Efavirenz,poor metabolism of,614546<br>{Efavirenz central nervous system toxicity,susceptibility to},614546 |

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|---|---|---|---|---|
| CYP2C19 | 182.4 | 99% | 96% | Clopidogrel,impaired responsiveness to,609535<br>Mephenytoin poor metabolizer,609535<br>Omeprazole poor metabolizer,609535<br>Proguanil poor metabolizer,609535 |
| CYP2C8 | 112.2 | 97% | 96% | Rhabdomyolysis,cerivastatin-induced |
| CYP2C9 | 188.1 | 100% | 98% | Tolbutamide poor metabolizer<br>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<br>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 involvment 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<br>[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<br>Esophageal carcinoma,somatic,133239<br>Mirror movements 1,157600 |
| DCHS1 | 138.6 | 99% | 98% | Mitral valve prolapse 2,607829<br>Van Maldergem syndrome 1,601390 |
| DCLRE1C | 120.5 | 95% | 92% | Severe combined immunodeficiency, Athabascan type, 602450 |
| DCN | 141.1 | 95% | 94% | Corneal dystrophy, congenital stromal, 610048 |
| DCTN1 | 119.2 | 100% | 98% | Neuropathy, distal hereditary motor, type VIIB, 607641<br>Perry syndrome,168605<br>{Amyotrophic lateral sclerosis,susceptibility to},105400 |
| DCX | 76.6 | 100% | 100% | Lissencephaly, X-linked, 300067<br>Subcortical laminal heteropia, X-linked, 300067 |
| DDB2 | 148.5 | 100% | 98% | Xeroderma pigmentosum, group E, DDB-negative subtype, 278740 |

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| 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 Ir, 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<br>Cardiomyopathy, dilated, 1I, 604765<br>Myopathy, myofibrillar, 1, 601419<br>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<br>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<br>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<br>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<br>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<br>Pleuropulmonary blastoma, 601200 |

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| | | | | 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<br>Trichodontoosseous syndrome,190320 |
| DMD | 83.9 | 99% | 94% | Becker muscular dystrophy, 300376<br>Cardiomyopathy, dilated, 3B, 302045<br>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<br>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 |

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|---|---|---|---|---|
| DNASE1L3 | 146.1 | 100% | 100% | Systemic lupus erythematosus 16,614420 |
| DNM1L | 109.2 | 100% | 94% | Encephalopahty, lethal, due to defective mitochondrial peroxisomal fission, 614388 |
| DNM2 | 116 | 96% | 93% | Charcot-Marie-Tooth disease, axonal, type 2M, 606482<br>Charcot-Marie-Tooth disease, dominant intermediate B, 606482<br>Lethal congenital contracture syndrome 5, 615368<br>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<br>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 Ij, 608093<br>Myasthenic syndrome, congenital, with tubular aggregates 2, 614750 |
| DPM1 | 112.1 | 89% | 84% | Congenital disorder of glycosylation, type Ie, 608799 |
| DPM2 | 98.3 | 100% | 100% | Congenital disorder of glycosylation, type Iu, 615042 |
| DPM3 | 136 | 100% | 100% | Congenital disorder of glycosylation, type Io, 612937 |
| DPP6 | 132.1 | 98% | 94% | Mental retardation, autosomal dominant 33, 616311<br>{Ventricular fibrillation, paroxysmal familial, 2} |
| DPY19L2 | 84.6 | 71% | 57% | Spermatogenic failure,613958 |
| DPYD | 166.8 | 97% | 93% | Dihydropyrimidine dehydrogenase deficiency, 274270<br>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<br>Myoclonus dystonia (Klein (1999) Proc Natl Acad Sci USA 96,5173<br>Schizophrenia, association with (Arinami (1994) Lancet 343,703) |
| DRD4 | 68.6 | 80% | 76% | Autonomic nervous system dysfunction<br>[Novelty seeking personality],601696<br>{Attention deficit-hyperactivity disorder},143465 |
| DRD5 | 84.5 | 100% | 100% | Dystonia,primary cervical<br>{Attention deficit-hyperactivity disorder,susceptibility to},143465<br>{Blepharospasm,primary benign},606798 |

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| 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<br>Keratosis palmoplantaris striata I,AD,148700 |
| DSG2 | 128.9 | 100% | 99% | Arrhythmogenic right ventricular dysplasia 10, 610193<br>Cardiomyopathy, dilated, 1BB, 612877 |
| DSG4 | 214.6 | 98% | 96% | Hypotrichosis 6,607903 |
| DSP | 131.6 | 99% | 97% | Arrhythmogenic right ventricular dysplasia 8,607450<br>Cardiomyopathy, dilated, with woolly hair and keratoderma,605676<br>Dilated cardiomyopathy with woolly hair, keratoderma and tooth agenesis,615821<br>Epidermolysis bullosa,lethal acantholytic,609638 |
| DSPP | 182.5 | 100% | 98% | Deafness,autosomal dominant 36,with dentinogenesis,605594<br>Dentin dysplasia,type II,125420<br>Dentinogenesis imperfecta, Shields type II,125490<br>Dentinogenesis imperfecta, Shields type III, 125500 |
| DST | 159.5 | 99% | 97% | ?Neuropathy, hereditary sensory and autonomic, type VI, 614653<br>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<br>{Schizophrenia},181500 |
| DUOX2 | 131.9 | 95% | 94% | Thyroid dyshormonogenesis 6,607200 |
| DUOXA2 | 106.9 | 100% | 98% | Thyroid dyshormonogenesis 5,274900 |
| DUSP6 | 144.6 | 100% | 100% | Hypogonadotropic hypogonadism 19 with or without anosmia, 615269 |
| DYM | 105 | 97% | 95% | Dyggve-Melchior-Clausen disease, 223800<br>Smith-McCort dysplasia, 607326 |
| DYNC1H1 | 166.1 | 99% | 98% | Charcot-Marie-Tooth disease, axonal, type 20, 614228<br>Mental retardation, autosomal dominant 13, 614563<br>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<br>Muscular dystrophy, limb-girdle, type 2B, 253601<br>Myopathy, distal, with anterior tibial onset, 606768 |

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| DYX1C1 | 78 | 96% | 84% | Ciliary dyskinesia, primary, 25, 615482<br>{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<br>{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<br>Tooth agenesis,selective,X-linked 1,313500 |
| EDAR | 118.2 | 100% | 99% | Ectodermal dysplasia 10A,hypohidrotic/hair/nail type, autosomal dominant,129490<br>Ectodermal dysplasia 10B,hypohidrotic/hair/tooth type, autosomal recessive,224900<br>[Hair morphology 1,hair thickness],612630 |
| EDARADD | 75.6 | 91% | 91% | Ectodermal dysplasia 11A,hypohidrotic/hair/tooth type, autosomal dominant,614940<br>Ectodermal dysplasia 11B,hypohidrotic/hair/tooth type, autosomal recessive,614941 |
| EDN1 | 138.6 | 100% | 100% | auriculocondylar syndrome 3,615706<br>Question mark ears,isolated,612798<br>{High density lipoprotein cholesterol level QTL 7} |
| EDN3 | 106.9 | 100% | 95% | Central hypoventilation syndrome congenital,209880<br>Waardenburg syndrome, type 4B,613265<br>{Hirshprung disease,susceptibility to,4},613712 |
| EDNRA | 206.4 | 100% | 100% | mandibulofacial dysostosis with alopecia, 616367<br>{Migraine, resistance to},157300 |
| EDNRB | 124.7 | 95% | 89% | ?{Hirschsprung disease, susceptibility to}, 600155<br>ABCD syndrome, 600501<br>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 IB,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<br>Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980<br>Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980 |

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| | | | | {Nonsmall cell lung cancer, susceptibility to}, 211980 |
| EGLN1 | 42.9 | 81% | 66% | Erythrocytosis,familial,3,609820<br>[Hemoglobin,high altitude adaptation],609070 |
| EGR2 | 110 | 100% | 100% | Charcot-Marie-Tooth disease,type 1D,607678<br>Dejerine-Sottas disease,145900<br>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 venooclusive 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<br>Ovarioleukodystrophy, 603896 |
| EIF2B3 | 150.2 | 100% | 100% | Leukoencephalopathy with vanishing white matter, 603896 |
| EIF2B4 | 140.8 | 99% | 96% | Leukoencephaly with vanishing white matter, 603896<br>Ovarioleukodystrophy, 603896 |
| EIF2B5 | 113.7 | 100% | 99% | Leukoencephalopathy with vanishing white matter, 603896<br>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<br>{Prostate cancer, hereditary, 2, susceptibility to}, 614731 |
| ELANE | 93.3 | 100% | 93% | Neutropenia, cyclic, 162800<br>Neutropenia, severe congenital 1, autosomal dominant, 202700 |
| ELN | 91.5 | 99% | 96% | Cutis laxa AD,123700<br>Supravalvar aortic stenosis,185500 |
| ELOVL4 | 77.5 | 100% | 98% | ?Spinocerebellar ataxia 34,133190<br>Ichthyosis, spastic quadriplegia, and mental retardation, 614457<br>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 |

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|---|---|---|---|---|
| ENAM | 124.6 | 100% | 100% | Amelogenesis imperfecta type IB,104500<br>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<br>Cole disease,615522<br>Hypophosphatemic rickets,autosomal recessive,2,613312<br>{Diabetes mellitus,non-insulin-dependent,susceptibility to},125853<br>{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<br>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<br>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<br>Diphenylhydantoin toxicity<br>Hypercholanemia, familial, 607748<br>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<br>Gastric cancer,somatic,613659<br>Glioblastoma,somatic,137800<br>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 |

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|---|---|---|---|---|
| ERCC2 | 120 | 100% | 99% | Cerebrooculofacioskeletal syndrome 2, 610756<br>Trichothiodystrophy, 601675<br>Xeroderma pigmentosum, group D, 278730 |
| ERCC3 | 94.3 | 100% | 99% | Trichothiodystrophy, 601675<br>Xeroderma pigmentosum, group B, 610651 |
| ERCC4 | 144.3 | 100% | 99% | ?XFE progeroid syndrome, 610965<br>Fanconi anemia, complementation group Q, 615272<br>Xeroderma pigmentosum, group F, 278760<br>Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 |
| ERCC5 | 133.9 | 100% | 96% | Xeroderma pigmentosum, group G, 278780<br>Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 |
| ERCC6 | 165.9 | 100% | 100% | Cerebrooculofacioskeletal syndrome 1, 214150<br>Cockayne syndrome, type B, 133540<br>De Sanctis-Cacchione syndrome, 278800<br>Premature ovarian failure 11,616946<br>UV-sensitive syndrome 1, 600630<br>{Lung cancer, susceptibility to}, 211980<br>{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<br>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<br>SC phocomelia syndrome, 269000 |
| ESPN | 39.8 | 70% | 56% | Deafness, autosomal recessive 36, 609006<br>Deafness, neurosensory, without vestibular involvement, autosomal dominant |
| ESR1 | 109.7 | 99% | 98% | Estrogen resistance,615363<br>{Breast cancer},114480<br>{Migraine,susceptibility to},157300<br>{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 |

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|---|---|---|---|---|
| ETHE1 | 64.8 | 99% | 88% | Ethylmalonic encephalopathy, 602473 |
| ETV6 | 114.9 | 100% | 100% | Leukemia,acute myeloid,somatic,601626<br>Thrombocytopenia 5,616216 |
| EVC | 95.9 | 94% | 89% | Ellis-van Creveld syndrome,225500<br>Weyers acrodental dysostosis,193530 |
| EVC2 | 105.5 | 95% | 91% | Ellis-van Creveld syndrome,225500<br>Weyers acrodental dysostosis,193530 |
| EWSR1 | 65.8 | 93% | 79% | Ewing sarcoma,612219<br>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<br>Exostoses, multiple, type 1, 133700 |
| EXT2 | 170.1 | 99% | 97% | ?Seizures,scoliosis and macrocephaly syndrome,616682<br>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<br>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<br>{Myocardial infarction,protection against},608446<br>{Venous thrombosis,protection against},188050 |
| F13B | 141.4 | 95% | 85% | Factor XIIIB deficiency,613235 |
| F2 | 106.3 | 100% | 97% | Dysprothrombinemia,613679<br>Hypoprothrombinemia,613679<br>Thrombophilia due to thrombin defect,188050<br>{Pregnancy loss,recurrent,susceptibility to,2},614390<br>{Stroke,ischemic,susceptibility to},601367 |

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|---|---|---|---|---|
| F5 | 176.1 | 99% | 97% | Factor V deficiency,227400<br>Thrombophilia due to activated prtein C resistance,188055<br>{Budd-Chiari syndrome},600880<br>{Pregnancy loss,recurrent,susceptibility to,1},614389<br>{Stroke,ischemic,susceptibility to},601367 |
| F7 | 138.6 | 100% | 96% | Factor VII deficiency,227500<br>{Myocardial infarction,decreased susceptibility to},608446 |
| F8 | 79.5 | 98% | 96% | Hemophilia A,306700 |
| F9 | 102.4 | 95% | 86% | Hemophilia B,306900<br>Thrombophilia,X-linked,due to factor IX defect},300807<br>{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<br>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 |

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|---|---|---|---|---|
| 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<br>{Lung cancer, susceptibility to}, 211980 |
| FAT4 | 220.5 | 100% | 100% | Hennekam lymphangiectasia-lymphedema syndrome 2,616006<br>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<br>Cutis laxa,autosomal recessive,type IA,219100<br>Macular degeneration,age-related,3,608895 |
| FBN1 | 161.3 | 99% | 98% | Acromicric dysplasia, 102370<br>Aortic aneurysm, ascending, and dissection<br>Ectopia lentis, familial, 129600<br>Geleophysic dysplasia 2,614185<br>Marfan lipodystrophy syndrome,616914<br>Marfan syndrome, 154700<br>MASS syndrome, 604308<br>Stiff skin syndrome, 184900<br>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% | Protoporphyria, erythropoietic, autosomal recessive, 177000 |
| FERMT1 | 97.3 | 98% | 94% | Kindler syndrome,173650 |
| FERMT3 | 103.7 | 100% | 97% | Leukocyte adhesion deficiency, type III, 612840 |

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|---|---|---|---|---|
| FGA | 149.6 | 97% | 95% | Afibrinogenemia,congenital,202400<br>Amyloidosis,familial visceral,105200<br>Dysfibrinogenemia,congenital,616004<br>Hypodysfibrinogenemia,congenital,616004 |
| FGB | 168 | 100% | 98% | Afibrinogenemia,congenital,202400<br>Dysfibrinogenemia,congenital,616004<br>Hypofibrinogenemia,congenital,202400 |
| FGD1 | 53.8 | 89% | 77% | Aarskog-Scott syndrome, 305400<br>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<br>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<br>Osteomalacia,tumor-induced<br>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<br>Hartsfield syndrome, 615465<br>Hypogonadotropic hypogonadism 2 with or without anosmia, 147950<br>Jackson-Weiss syndrome, 123150<br>Osteoglophonic dysplasia, 166250<br>Pfeiffer syndrome, 101600<br>Trigonocephaly 1, 190440 |

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

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| FHL1 | 58.5 | 96% | 75% | Emery-Dreifuss muscular dystrophy 6,X-linked,300696<br>Myopathy,X-linked,with postural muscle atrophy,300696<br>Reducing body myopathy,X-linked 1a,severe,infantile or early childhood onset,300717<br>Reducing body myopathy,X-linked 1b,with late childhood or adult onset,300718<br>Scapuloperoneal myopathy,X-linked dominant,300695 |
| FIG4 | 155.7 | 100% | 99% | ?Polymicrogyria,bilateral temporooccipital,612691<br>Amyotrophic lateral sclerosis 11,612577<br>Charcot-Marie-Tooth disease, type 4J, 611228<br>Yunis-Varon syndrome,216340 |
| FIGLA | 84.3 | 88% | 84% | Premature ovarian failure,612310 |
| FKBP10 | 141.6 | 97% | 88% | Bruck syndrome 1,259450<br>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<br>Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 |
| FKTN | 137.2 | 97% | 89% | Cardiomyopathy,dilated,1X,611615<br>Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies),type A,4,253800<br>Muscular dystrophy-dystroglycanopathy (congenital without mental retardation),type B,4,613152<br>Muscular dystrophy-dystroglycanopathy (limb-girdle),type C,4,611588 |
| FLCN | 142.8 | 100% | 98% | Birt-Hogg-Dube syndrome, 135150<br>Colorectal cancer, somatic, 114500<br>Pneumothorax, primary spontaneous, 173600<br>Renal carcinoma, chromophobe, somatic, 144700 |
| FLG | 212.1 | 100% | 99% | Ichthyosis vulgaris,146700<br>{Dermatitis,atopic,susceptibility to,2},605803 |

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|---|---|---|---|---|
| FLNA | 85.1 | 99% | 97% | Cardiac valvular dysplasia,X-linked,314400<br>Congenital short bowel syndrome,300048<br>FG syndrome 2,300321<br>Frontometaphyseal dysplasia,305620<br>Heterotopia,periventricular,300049<br>Heterotopia,periventricular,ED variant,300537<br>Intestinal pseudoobstruction,neuronal,300048<br>Melnick-Needles syndrome,309350<br>Otopalatodigital syndrome,type I,311300<br>Otopalatodigital syndrome,type II,304120<br>Terminal osseous dysplasia,300244 |
| FLNB | 139.4 | 100% | 99% | Atelosteogenesis,type I,108720<br>Atelosteogenesis,type III,108721<br>Boomerang dysplasia,112310<br>Larsen syndrome,150250<br>Spondylocarpotarsal synostosis syndrome,272460 |
| FLNC | 149.8 | 100% | 99% | Myopathy, distal, 4, 614065<br>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<br>Leukemia,acute myeloid,reduced survival in,somatic,601626 |
| FLT4 | 148 | 99% | 95% | Hemangioma,capillary infantile,somatic,602089<br>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<br>Fragile X tremor/ataxia syndrome, 300623<br>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<br>Iridogoniodysgenesis, type 1, 601631<br>Iris hypoplasia and glaucoma, 601631<br>Rieger or Axenfeld anomalies, 602482 |

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| 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<br>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<br>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<br>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<br>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<br>Ovarian hyperstimulation syndrome,608115<br>Ovarian response to FSH stimulation,276400 |
| FTCD | 79.6 | 88% | 80% | Glutamate formiminotransferase deficiency, 229100 |
| FTL | 104.5 | 99% | 82% | Hyperferritinemia-cataract syndrome, 600886<br>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<br>Tremor, hereditary essential, 4, 614782 |

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| 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<br>Friedreich ataxia with retained reflexes, 229300 |
| FXYD2 | 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<br>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<br>Neutropenia,severe congenital 4,autosomal recessive,612541 |
| G6PD | 69.8 | 98% | 96% | Favism, 134700<br>Hemolytic anemia due to G6PD deficiency,300908<br>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<br>{Epilepsy, childhood absence, susceptibility to, 4}<br>{Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136 |
| GABRB3 | 143.2 | 95% | 95% | {Epilepsy,childhood absence, susceptibility to, 5},612269<br>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<br>Febrile seizures,familial,8,611277<br>{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 |

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| GARS | 127.7 | 100% | 99% | Charcot-Marie-Tooth disease, type 2D, 601472<br>Neuropathy,distal hereditary motor,type VA,600794 |
| GATA1 | 49.6 | 97% | 90% | Anemia,X-linked,with/without neutropenia and/or platelet abnormalities,300835<br>Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685<br>Thrombocytopenia with beta-thalassemia, X-linked, 314050<br>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<br>Atrial septal defect 2,607941<br>Atrioventricular septal defect 4,614430<br>Tetralogy of Fallot,187500<br>Ventricular septal defect 1,614429 |
| GATA6 | 49.7 | 83% | 66% | Atrioventricular septal defect 5, 614474<br>Atrial septal defect 9, 614475<br>Pancreatic agenesis and congenital heart defects, 600001<br>Persistent truncus arteriosus, 217095<br>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<br>Gaucher disease, type II, 230900<br>Gaucher disease, type III, 231000<br>Gaucher disease, type IIIC, 231005<br>Gaucher disease, perinatal lethal, 608013<br>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<br>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<br>Hyperphenylalaninemia, BH4-deficient, B, 233910 |

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

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| GDNF | 138.3 | 98% | 94% | Central hypoventilation syndrome, 209880<br>{Pheochromocytoma, modifier of}, 171300<br>{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<br>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% | Psuedoxanthoma elasticum-like disorder with multiple coagulation factor deficiency,610842<br>Vitamin K-dependent clotting factors,combined deficiency of,1,277450 |
| GH1 | 180.2 | 100% | 100% | Growth hormone deficiency,isolated,type IA,262400<br>Growth hormone deficiency,isolated,type IB,612781<br>Growth hormone deficiency,isolated,type II,173100<br>Kowarski syndrome,262650 |
| GHR | 208.7 | 100% | 100% | Growth hormone insensitivity,partial,604271<br>Laron dwarfism,262500<br>{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<br>Craniometaphyseal dysplasia, autosomal recessive,218400<br>Erythrokeratodermia variabilis et progressiva,133200<br>Hypoplastic left heart syndrome 1,241550<br>Oculodentodigital dysplasia,164200<br>Oculodentodigital dysplasia,autosomal recessive,257850<br>Palmoplantar keratoderma with congenital alopecia,104100<br>Syndactyly,type III,186100 |
| GJA3 | 152.3 | 100% | 100% | Cataract 14, multiple types, 601885 |

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| GJA5 | 225.4 | 100% | 100% | Atrial fibrillation, familial, 11, 614049<br>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<br>Deafness,autosomal recessive 1A,220290<br>Hystrix-like ichthyosis with deafness,602540<br>Keratitis,ichthyosis-deafness syndrome,148210<br>Keratoderma,palmoplantar,with deafness,148350<br>Vohwinkel syndrome,124500 |
| GJB3 | 300.8 | 100% | 100% | Deafness autosomal dominant 2B,612644<br>Deafness,digenic,GJB2/GJB3,220290<br>Erythrokeratodermia variabilis et progressiva,133200 |
| GJB4 | 363.8 | 100% | 100% | Erythrokeratodermia variabilis with erythema gyratum repens,133200 |
| GJB6 | 191.1 | 100% | 100% | Deafness,autosomal dominant 3B,612643<br>Deafness,autosomal recessive 1B,612645<br>Deafness,digenic GJB2/GJB6,220290<br>Ectodermal dysplasia 2,Clouston type,129500 |
| GJC2 | 34.3 | 81% | 62% | Leukodystrophy, hypomyelinating, 2, 608804<br>Lymphedema, hereditary, IC, 613480<br>Spastic paraplegia 44, autosomal recessive, 613206 |
| GK | 29.1 | 66% | 50% | Glycerol kinase deficiency, 307030 |
| GLA | 47.8 | 99% | 90% | Fabry disease, 301500<br>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<br>Lethal congenital contracture syndrome 1,253310 |
| GLI2 | 110.3 | 97% | 93% | Holoprosencephaly-9, 610829 |
| GLI3 | 138.5 | 100% | 100% | Greig cephalopolysyndactyly syndrome, 175700<br>Pallister-Hall syndrome, 146510<br>Polydactyly, preaxial, type IV, 174700<br>Polydactyly, postaxial, types A1 and B, 174200<br>{Hypothalamic hamartomas, somatic}, 241800 |
| GLIS2 | 75.5 | 99% | 90% | Nephronophthisis 7, 611498 |

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| 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<br>Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351<br>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<br>Hypocalciuric hypercalcemia, type II, 145981 |
| GNAI2 | 144.5 | 100% | 100% | Pituitary ACTH-secreting adenoma<br>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<br>Sturge-Weber syndrome, somatic, mosaic,185300 |
| GNAS | 110.5 | 95% | 94% | Acromegaly,somatic,102200<br>ACTH-independent macronodular adrenal hyperplasia,219080<br>McCune-Albright syndrome,somatic,mosaic 174800<br>Osseous heteroplasia, progressive, 166350<br>Pseudohypoparathyroidism Ia, 103580<br>Pseudohypoparathyroidism Ib, 603233<br>Pseudohypoparathyroidism Ic, 612462<br>Pseudopseudohypoparathyroidysm,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 |

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| GNE | 150.9 | 100% | 100% | Nonaka myopathy, 605820<br>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% | Mucolipidosis III alpha/beta, 252600<br>Mucolipidosis II alpha/beta, 252500 |
| GNPTG | 125.2 | 90% | 87% | Mucolipidosis 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<br>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<br>Bernard-Soulier syndrome, type A2 (dominant),153670<br>von Willebrand disease,platelet-type,177820<br>{Nonarteric anterior ischemic optic neuropathy,susceptibility to},258660 |
| GP1BB | 37 | 86% | 65% | Bernard-Soulier syndrome,type B,231200<br>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<br>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<br>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 |

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| GPR98 | 145.8 | 99% | 94% | Febrile seizures, familial, 4, 604352<br>Usher syndrome, type 2C, 605472<br>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<br>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<br>Ceroid lipofuscinosis,neuronal,11,614706<br>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<br>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<br>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<br>Meconium ileus,614665 |
| GUCY2D | 79.9 | 96% | 84% | Cone-rod dystrophy 6, 601777<br>Leber congenital amaurosis 1, 204000 |

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| 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<br>Silver-Russell syndrome,180860<br>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<br>Hyperinsulinemic hypoglycemia, familial, 4, 609975 |
| HADHA | 70.5 | 93% | 86% | Fatty liver, acute, of pregnancy, 609016<br>HELLP syndrome, maternal, of pregnancy, 609016<br>LCHAD deficiency, 609016<br>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-<br>Heinz body anemias,alpha-,140700<br>Hemoglobin H disease,nondeletional,613978<br>Methemoglobinemias,alpha-<br>Thalassemias,alpha-,604131 |
| HBA2 | 65.1 | 94% | 84% | Erythrocytosis<br>Heinz body anemia,140700<br>Hemoglobin H disease,nondeletional,613978<br>Hypochromic microcytic anemia<br>Thalassemia,alpha-,604131 |

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| HBB | 133.5 | 100% | 100% | Delta-beta thalassemia,141749<br>Erythremias,beta-<br>Heinz body anemias,beta-,140700<br>Hereditary persistence of fetal hemoglobin,141749<br>Thalassemia-beta,dominant inclusion body,603902<br>Sickle cell anemia,603903<br>Thalassemias,beta-,613985 |
| HBD | 201.9 | 100% | 100% | Thalassemia due to Hb Lepore<br>Thalassemia,delta- |
| HBG1 | 123.4 | 98% | 93% | Fetal hemoglobin quantitative trait locus 1,141749 |
| HBG2 | 235.1 | 100% | 100% | Cyanosis,transient neonatal,613977<br>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<br>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<br>Wilson-Turner syndrome, 309585 |
| HEATR2 | 107.9 | 84% | 77% | Ciliary dyskinesia, primary, 18, 614874 |
| HEPACAM | 124.5 | 76% | 75% | Megalencephalic leukeoencephalopathy with subcortical cysts 2A,613925<br>Megalencephalic leukoncephalopathy with subcortical cysts 2B,remitting,with or without mental retardation,613926 |
| HERC2 | 98.8 | 77% | 72% | Mental retardation, autosomal recessive 38, 615516<br>[Skin/hair/eye pigmentation 1, blond/brown hair], 227220<br>[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<br>Pituitary hormone deficiency, combined, 5, 182230<br>Septooptic dysplasia, 182230 |

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| HEXA | 125.4 | 100% | 99% | GM2-gangliosidosis, several forms, 272800<br>Tay-Sachs disease, 272800<br>[Hex A pseudodeficiency], 272800 |
| HEXB | 134.4 | 97% | 91% | Sandhoff disease, infantile, juvenile, and adult forms, 268800 |
| HFE | 126.5 | 100% | 97% | Hemochromatosis, 235200<br>{Microvascular complications of diabetes 7}, 612635<br>{Porphyria variegata, susceptibility to}, 176200<br>{Porphyria cutanea tarda, susceptibility to}, 176100<br>{Alzheimer disease, susceptibility to}, 104300<br>[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<br>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<br>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<br>Hepatic adenoma, somatic, 142330<br>MODY, type III, 600496<br>Renal cell carcinoma, 144700<br>{Diabetes mellitus, noninsulin-dependent, 2}, 125853<br>{Diabetes mellitus, insulin-dependent}, 222100 |
| HNF1B | 103 | 100% | 97% | Diabetes mellitus,noninsulin-dependent,125853<br>Renal cysts and diabetes syndrome, 137920 |

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| | | | | {Renal cell carcinoma},144700 |
| HNF4A | 134 | 100% | 98% | Fanconi renotubular syndrome 4,with maturity-onset diabetes of the young,616026<br>MODY,type I,125850<br>{Diabetes mellitus,noninsulin-dependent},125853 |
| HNRNPA1 | 81.4 | 86% | 78% | ?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3,615424<br>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<br>Bosley-Salih-Alorainy syndrome, 601536 |
| HOXA11 | 85.6 | 82% | 79% | Radioulnar synostosis with amegakaryocytic thrombocytopenia,605432 |
| HOXA13 | 38 | 65% | 60% | Guttmacher syndrome,176305<br>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<br>Vertical talus,congenital,192950 |
| HOXD13 | 95.9 | 93% | 88% | ?Brachydactyly-syndactyly syndrome,610713<br>Brachydactyly type D,113200<br>Brachydactyly,type E,113300<br>Syndactyly,type V,186300<br>Synpolydactyly with foot anomalies,286000 |
| HPD | 131.5 | 100% | 100% | Hawkinsinuria, 140350<br>Tyrosinemia, type III, 276710 |
| HPGD | 94.2 | 97% | 89% | Cranioosteoarthropathy,259100<br>Digital clubbing,isolated congenital,119900<br>Hypertrophic osteoarthropathy,primary,autosomal recessive 1,259100 |
| HPRT1 | 45.8 | 95% | 84% | HPRT-related gout,300323<br>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 |

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|---|---|---|---|---|
| HPSE2 | 108.3 | 97% | 92% | Urofacial syndrome 1,236730 |
| HR | 76.4 | 96% | 91% | Alopecia universalis,203655<br>Atrichia with papular lesions,209500<br>Hypotrichosis 4,146550 |
| HRAS | 165.7 | 100% | 98% | Congenital myopathy with excess of muscle spindles, 218040<br>Costello syndrome, 218040<br>Schimmelpenning-Feuerstein-Mims syndrome,somatic mosaic,163200<br>{Bladder cancer, somatic}, 109800<br>{Nevus sebaceous, somatic}, 162900<br>{Spitz nevus or nevus spilus,somatic},137550<br>{Thyroid carcinoma, follicular, somatic}, 188470 |
| HRG | 153.1 | 97% | 94% | Thrombophilia due to elevated HRG,613116<br>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<br>Mental retardation, X-linked syndromic 10, 300220<br>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<br>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<br>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<br>Neuropathy, distal hereditary motor, type IIA, 158590 |
| HSPD1 | 84.8 | 97% | 89% | Leukodystrophy, hypomyelinating, 4, 612233<br>Spastic paraplegia 13, autosomal dominant, 605280 |
| HSPG2 | 108 | 99% | 97% | Dyssegmental dysplasia, Silverman-Handmaker type, 224410<br>Schwartz-Jampel syndrome, type 1, 255800 |
| HTR1A | 200.6 | 100% | 100% | Periodic fever,menstrual cycle dependent,614674 |

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|---|---|---|---|---|
| HTRA1 | 98.5 | 78% | 75% | CARASIL syndrome,600142<br>{Macular degeneration,age-related,7},610149<br>{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-hydrosyglutaric 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<br>Mucopolysaccharidosis Is, 607016<br>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 supravalvular 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 |

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|---|---|---|---|---|
| IHH | 112.6 | 100% | 100% | Acrocaptiofemoral dysplasia,607778<br>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<br>Ectodermal,dysplasia,anhidrotic,lymphedema and immunodeficiency,300301<br>Immunodeficiency 33,300636<br>Immunodeficiency,isolated,300584<br>Uncontinentia pigmenti,308300<br>Invasive pneumococcal disease,recurrent isolated,2,300640 |
| IKZF1 | 162.4 | 100% | 99% | Leukemia,acute lymphoblastic<br>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<br>{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<br>[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 |
| ILDR1 | 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<br>Retinitis pigmentosa 10, 180105 |
| IMPG2 | 166.8 | 100% | 97% | Macular dystrophy,vitelliform,5,616152<br>Retinitis pigmentosa 56, 613581 |

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| INF2 | 76.5 | 92% | 86% | Charcot-Marie-Tooth disease,dominant intermediate E,614455<br>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<br>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<br>Diabetes mellitus,permanent neonatal,606176<br>Hyperproinsulinemia,616214<br>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<br>Myelodysplastic syndrome,preleukemic<br>Myelogenous leukemia,acute<br>Nonsmall cell lung cancer,somatic,211980 |
| IRF4 | 153.5 | 99% | 97% | Multiple myeloma,254500<br>[Skin/hair/eye pigmentation, variation in,8],611724 |
| IRF6 | 121.7 | 99% | 93% | Orofacial cleft 6,608864<br>Popliteal pterygium syndrome 1,119500<br>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<br>{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<br>Glanzmann thrombasthenia,273800 |

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|---|---|---|---|---|
| | | | | 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<br>Glanzmann thrombasthenia,273800<br>Purpura,posttransfusion<br>Thrombocytopenia,neonatal alloimmune<br>{Myocardial infarction,susceptibility to},608446 |
| ITGB4 | 132.4 | 96% | 93% | Epidermolysis bullosa of hands and feet,131800<br>Epidermolysis bullosa,junctional,non-Herlitz type,226650<br>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<br>Dementia,familial British,176500<br>Dementia,familial Danish,117300 |
| ITPR1 | 157.5 | 100% | 99% | Spinocerebellar ataxia 15, 606658<br>Spinocerebellar ataxia 29, congenital nonprogressive, 117360 |
| IVD | 106.8 | 100% | 100% | Isovaleric acidemia, 243500 |
| IYD | 115.4 | 100% | 97% | Thyroid dyshormonogenesis 4,274800 |
| JAG1 | 139.8 | 100% | 97% | Alagille syndrome, 118450 |
| JAK2 | 89.8 | 95% | 93% | Erythrocytosis,somatic,133100<br>Leukemia,acute myelogenous,601626<br>Myelofibrosis,somatic,254450<br>Polycythemia vera,263300<br>Thrombocythemia 3,614521<br>{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 |

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| JUP | 134.9 | 100% | 99% | Arrhythmogenic right ventricular dysplasia 12,611528<br>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<br>Deafness,autosomal recessive 89,613916 |
| KAT6B | 167.3 | 98% | 97% | Genitopatellar syndrome, 606170<br>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<br>Long QT syndrome-5, 613695 |
| KCNE2 | 113.9 | 100% | 100% | Atrial fibrillation, familial, 4, 611493<br>Long QT syndrome-6, 613693 |
| KCNE3 | 171.2 | 100% | 100% | Brugada syndrome 6, 613119 |
| KCNH2 | 89.2 | 93% | 83% | Long QT syndrome-2, 613688<br>{Long QT syndrome-2, acquired, susceptibility to}, 613688<br>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<br>Diabetes mellitus,transient neonatal,3,610582<br>Hyperinsulinemic hypoglycemia,familial,2,601820<br>Maturity-onset diabetes of the young,type 13,616329<br>{Diabetes mellitus,type 2,susceptibility to},125853 |
| KCNJ13 | 194 | 100% | 98% | Leber congenital amaurosis 16, 614186<br>Snowflake vitreoretinal degeneration, 193230 |
| KCNJ2 | 212.3 | 100% | 100% | Andersen syndrome, 170390<br>Atrial fibrillation, familial, 9, 613980<br>Short QT syndrome-3, 609622 |

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| KCNJ5 | 179.7 | 100% | 99% | Hyperaldosteronism, familial, type III, 613677<br>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<br>Jervell and Lange-Nielsen syndrome, 220400<br>Long QT syndrome-1, 192500<br>Short QT syndrome-2, 609621<br>{Long QT syndrome 1, acquired, susceptibility to}, 192500 |
| KCNQ1OT1 | | | | Beckwith-Wiedemann syndrome,130650 |
| KCNQ2 | 80.5 | 98% | 92% | Epileptic encephalopathy, early infantile, 7, 613720<br>Myokymia, 121200<br>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<br>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<br>Neuropathy, hereditary sensory, type IIC, 614213<br>Spastic paraplegia 30, autosomal recessive, 610357 |

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| KIF1B | 149.8 | 99% | 98% | Charcot-Marie-Tooth disease, type 2A1, 118210<br>Pheochromocytoma, 171300<br>{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<br>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<br>?Hydrolethalus syndrome 2,614120<br>Acrocallosal syndrome,200990<br>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<br>?Precocious puberty,central,1,176400 |
| KIT | 153.8 | 99% | 98% | Gastrointestinal stromal tumor, familial, 606764<br>Germ cell tumors, 273300<br>Leukemia, acute myeloid, 601626<br>Mast cell disease, 154800<br>Piebaldism, 172800 |
| KITLG | 76.3 | 91% | 88% | Hyperpigmentation familial progressive 2,145250<br>[Skin/hair/eye pigmentation 7],611664 |
| KL | 162 | 98% | 95% | Tumoral calcinosis, hyperphosphatemic,211900 |
| KLF1 | 50.7 | 90% | 83% | Blood group--Lutheran inhibitor, 111150<br>Dyserthropoietic anemia,congenital,type IV,613673<br>[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<br>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 |

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| 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<br>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<br>Breast cancer,somatic,114480<br>Cardiofaciocutaneous syndrome 2,615278<br>Gastric cancer,somatic,137215<br>Leukemia,acute myeloid,601626<br>Lung cancer,somatic,211980<br>Noonan syndrome 3,609942<br>Pancreatic carcinoma, somatic,260350<br>RAS-associated autoimmune leukoproliferative disorder,614470<br>Schimmelpenning-Feuerstein-Mins syndrome,somatic mosaic,163200 |
| KRIT1 | 85.9 | 100% | 99% | Cavernous malformations of CNS and retina,116860<br>Cerebral cavernous malformations-1,116860<br>Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations,116860 |
| KRT1 | 134.3 | 99% | 91% | Epidermolytic hyperkeratosis,113800<br>Ichthyosis histrix,Curth-Macklin type,146590<br>Ichthyosis, cyclic, with epidermolytic hyperkeratosis,607602<br>Keratosis palmoplantaris striata III,607654<br>Palmoplantar keratoderma,epidermolytic,144200 |
| KRT10 | 90.8 | 94% | 91% | Epidermolytic hyperkeratosis,113800<br>Ichthyosis with confetti,609165<br>Ichthyosis,cyclic,with epidermolytic hyperkeratosis,607602 |
| KRT12 | 88 | 95% | 92% | Meesmann corneal dystrophy, 122100 |
| KRT13 | 111.4 | 100% | 94% | White sponge nevus 2,615785 |

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| KRT14 | 53.6 | 86% | 78% | Dermatopathia pigmentosa reticularis,125595<br>Epidermolysis bullosa simplex,Dowling-Meara type,131760<br>Epidermolysis bullosa simplex,Koebner type,131900<br>Epidermolysis bullosa simplex,recessive 1,601001<br>Epidermolysis bullosa simplex,Weber-Cockayne type,13 |
| KRT16 | 28.8 | 71% | 47% | Pachyonychia congenita 1,167200<br>Palmoplantar keratoderma,nonepidermolytic,focal,613000 |
| KRT17 | 19.3 | 44% | 34% | Pachyonychia congenita 2,167210<br>Steatocystoma multiplex,184500 |
| KRT18 | 30.7 | 92% | 53% | Cirrhosis,cryptogenic,215600<br>{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<br>Epidermolysis bullosa simplex,Dowling-Meara type,131760<br>Epidermolysis bullosa simplex,Koebner type,131900<br>Epidermolysis bullosa simplex,recessive 1,601001<br>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<br>?Hypotrichosis 3,613981<br>Woolly hair, autosomal dominant,194300 |
| KRT8 | 37.3 | 85% | 67% | Cirrhosis,cryptogenic,215600<br>{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 |

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|---|---|---|---|---|
| L1CAM | 87.3 | 98% | 95% | Corpus callosum,partial agenesis of,304100<br>CRASH syndrome, 303350<br>Hydrocephalus due to aqueductal stenosis, 307000<br>Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000<br>Hydrocephalus with Hirschsprung disease, 307000<br>MASA syndrome, 303350 |
| L2HGDH | 120.8 | 97% | 95% | L-2-hydroxyglutaric aciduria, 236792 |
| LAMA2 | 144.2 | 99% | 98% | Muscular dystrophy, congenital merosin-deficient, 607855<br>Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855 |
| LAMA3 | 149.1 | 99% | 99% | Epidermolysis bullosa,generalized atrophic benign,226650<br>Epidermolysis bullosa,junctional,Herlitz type,226700<br>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<br>Pierson syndrome,609049 |
| LAMB3 | 107.8 | 100% | 99% | Amelogenesis imperfecta,type IA,104530<br>Epidermolysis bullosa,junctional,Herlitz type,226700<br>Epidermolysis bullosa,junctional,non-Herlitz type,226650 |
| LAMC2 | 108.7 | 100% | 97% | Epidermolysis bullosa,junctional,Herlitz type,226700<br>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<br>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<br>Greenberg skeletal dysplasia,215140<br>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 |

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| LDB3 | 108.4 | 94% | 92% | Cardiomyopathy, dilated 1C, 601493<br>Left ventricular noncompaction 3, with or without dilated cardiomyopathy, 601493<br>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<br>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<br>Melorheostosis with osteopoikilosis,155950<br>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<br>Leydig cell hypoplasia with hypergonadotropic hypogonadism,238320<br>Leydig cell hypoplasia with pseudohermaphroditism,238320<br>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<br>Severe combined immunodeficiency with sensitivity to ionizing radiation, 602450<br>{Multiple myeloma, resistance to}, 254500 |
| LIM2 | 88.4 | 100% | 99% | Cataract 19, 615277 |

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| LINS | 131.6 | 99% | 96% | Mental retardation, autosomal recessive 27, 614340 |
| LIPA | 109.7 | 95% | 95% | Cholesteryl ester storage disease, 278000<br>Wolman disease, 278000 |
| LIPC | 116.6 | 99% | 98% | Diabetes mellitus, noninsulin-dependent, 125853<br>Hepatic lipase deficiency, 614025<br>[High density lipoprotein cholesterol level QTL 12], 612797 |
| LIPH | 116.4 | 100% | 99% | Hypotrichosis 7,604379<br>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<br>Hypoplastic or aplastic tibia with polydactyly,188740<br>Laurin-Sandrow syndrome,135750<br>Polydactyly,preaxial type II,174500<br>Syndactyly,type IV,186200<br>Triphalangeal thumb type I,174500 |
| LMBRD1 | 73.2 | 87% | 82% | Methylmalonic aciduria and homocystinuria, cblF type, 277380 |
| LMF1 | 125.9 | 99% | 97% | Lipase deficiency,combined,246650 |
| LMNA | 72 | 95% | 87% | Cardiomyopathy,dilated,1A,115200<br>Charcot-Marie-Tooth disease,type 2B1,605588<br>Emery-Dreifuss muscular dystrophy 2,AD,181350<br>Emery-Dreifuss muscular dystrophy 3,AR,616516<br>Heart-hand syndrome,Slovenian type,610140<br>Hutchinson-Gilford progeria,176670<br>Lipodystrophy,familial partial,2,151660<br>Malouf syndrome,212112<br>Mandibuloacral dysplasia,248370<br>Muscular dystrophy,congenital,613205<br>Muscular dystrophy,limb-girdle,type 1B,159001<br>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 |

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| LPAR6 | 108.2 | 100% | 96% | Hypotrichosis 8,278150<br>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<br>Lipoprotein lipase deficiency, 238600<br>[High density lipoprotein cholesterol level QTL 11] |
| LPP | 118.5 | 100% | 99% | Leukemia,acute myeloid,601626<br>Lipoma |
| LRAT | 285.3 | 100% | 100% | Leber congenital amaurosis 14, 613341<br>Retinal dystrophy, early-onset severe, 613341<br>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<br>Sclerosteosis 2, 614305 |
| LRP5 | 165.5 | 98% | 96% | Exudative vitreoretinopathy 4, 601810<br>Hyperostosis, endosteal, 144750<br>Osteoporosis-pseudoglioma syndrome, 259770<br>Osteosclerosis, 144750<br>van Buchem disease, type 2, 607636<br>[Bone mineral density variability 1], 601884<br>{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<br>Microspherophakia and/or megalocornea,with ectopia lentis and with or without secondary |

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| | | | | glaucoma,251750<br>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<br>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<br>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<br>Pick disease,172700<br>Supranuclear palsy,progressive,601104<br>Supranuclear palsy,progressive atypical,260540<br>{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 |

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| 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<br>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<br>Keratosis 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% | Mucolipidosis IV, 252650 |
| MCPH1 | 133.9 | 99% | 94% | Microcephaly 1, primary, autosomal recessive, 251200 |
| MECP2 | 48.4 | 97% | 75% | Encephalopathy, neonatal severe, 300673<br>Mental retardation,X-linked,syndromic,Lubs type,300260<br>Mental retardation, X-linked, syndromic 13, 300055<br>Rett syndrome, 312750<br>Rett syndrome,atypical,312750<br>Rett syndrome,preserved speech variant,312750<br>{Autism susceptibility, X-linked 3}, 300496 |
| MED12 | 64.3 | 95% | 89% | Lujan-Fryns syndrome, 309520<br>Ohdo syndrome, X-linked, 300895<br>Opitz-Kaveggia syndrome, 305450 |
| MED13L | 131.9 | 100% | 98% | Mental retardation and distinctive facial features with or without cardiac defects,616789<br>Transposition of the great arteries, dextro-looped 1, 608808 |
| MED17 | 129.9 | 99% | 97% | Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668 |

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| MED23 | 131 | 99% | 97% | Mental retardation, autosomal recessive 18, 614249 |
| MED25 | 82.6 | 97% | 85% | ?Charcot-Marie-Tooth disease, type 2B2, 605589<br>Basel-Vanagait-Smirin-Yosef syndrome,616449 |
| MEF2C | 112.8 | 99% | 88% | Chromosome 5q14.3 deletion syndrome, 613443<br>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<br>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<br>Angiofibroma, somatic<br>Carcinoid tumor of lung<br>Lipoma, somatic<br>Multiple endocrine neoplasia 1, 131100<br>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% | Spondylococostal 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<br>Hereditary motor and sensory neuropathy VIA,601152 |
| MFRP | 117.5 | 100% | 100% | Microphthalmia, isolated 5, 611040<br>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 |

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| MIR96 | | | | Deafness,autosomal dominant 50,613074 |
| MITF | 134.3 | 100% | 100% | Tietz albinism-deafness syndrome,103500<br>Waardenburg syndrome, type 2A,193510<br>Waardenburg syndrome/ocular albinism, digenic,103470<br>{Melanoma,cutaneous malignant,susceptibility to 8},614456 |
| MKKS | 222.1 | 89% | 89% | Bardet-Biedl syndrome 6,605231<br>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<br>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<br>Mismatch repair cancer syndrome, 276300<br>Muir-Torre syndrome, 158320 |
| MLH3 | 157 | 100% | 100% | Colorectal cancer,hereditary nonpolyposis,type 7,614385<br>Colorectal cancer,somatic,114500<br>{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, cblB complementation type, 251110 |
| MMACHC | 173.1 | 100% | 100% | Methylmalonic aciduria and homocystinuria, cblC 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<br>{Epidermolysis bullosa dystrophica,autosomal recessive,modifier of},226600 |
| MMP13 | 136.6 | 92% | 91% | Metaphyseal anadysplasia 1,602111<br>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 |

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| 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 gycosylation, 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<br>Thrombocythemia 2, 601977<br>Thrombocytopenia, congenital amegakaryocytic, 604498 |
| MPLKIP | 76.3 | 88% | 52% | Trichothiodystrophy, nonphotosensitive 1, 234050 |
| MPO | 142.8 | 100% | 99% | Cardiomyopathy, dilated, 1T, 613740<br>Myeloperoxidase deficiency, 254600<br>{Alzheimer disease, susceptibility to}, 104300<br>{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<br>Charcot-Marie-Tooth disease, type 1B, 118200<br>Charcot-Marie-Tooth disease, type 2I, 607677<br>Charcot-Marie-Tooth disease, type 2J, 607736<br>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<br>Muir-Torre syndrome, 158320<br>Mismatch repair cancer syndrome, 276300 |

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|---|---|---|---|---|
| MSH3 | 109 | 96% | 92% | Endometrial carcinoma,somatic,608089 |
| MSH6 | 159.6 | 100% | 99% | Colorectal cancer, hereditary nonpolyposis, type 5, 614350<br>Endometrial cancer, familial, 608089<br>Mismatch repair cancer syndrome, 276300 |
| MSR1 | 181.2 | 100% | 100% | Barett esophagus/esophageal adenocarcinoma,614266<br>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<br>Orofacial cleft 5,608874<br>Tooth agenesis,selective,1,with or without orofacial cleft,106600 |
| MSX2 | 82.3 | 100% | 92% | Craniosynostosis, type 2, 604757<br>Parietal foramina 1, 168500<br>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<br>{Neural tube defects, susceptibility to}, 601634<br>{Schizophrenia, susceptibility to}, 181500<br>{Thromboembolism, susceptibility to}, 188050<br>{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<br>{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<br>Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600 |

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| | | | | Gastric cancer, somatic, 613659 |
| MVK | 126.3 | 100% | 100% | Hyper-IgD syndrome, 260920<br>Mevalonic aciduria, 610377<br>Porokeratosis 3, disseminated superficial actinic, 175900 |
| MXI1 | 102.6 | 89% | 85% | Neurofibrosarcoma<br>{Prostate cancer,susceptibility to},176807 |
| MYBPC1 | 159.7 | 99% | 98% | Arthrogryposis,distal,type 1B,614335<br>Lethal congenital contracture syndrome 4,614915 |
| MYBPC3 | 129.6 | 97% | 93% | Cardiomyopathy, dilated, 1MM, 615396<br>Cardiomyopathy, familial hypertrophic, 4, 115197<br>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<br>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<br>Arthrogryposis, distal, type 2B, 601680 |
| MYH6 | 107.5 | 98% | 93% | Atrial septal defect 3, 614089<br>Cardiomyopathy, dilated, 1EE, 613252<br>Cardiomyopathy, familial hypertrophic, 14, 613251<br>{Sick sinus syndrome 3}, 614090 |
| MYH7 | 106.3 | 99% | 95% | Cardiomyopathy,dilated, 1S,613426<br>Cardiomyopathy,hypertrophic,1, 192600<br>Left ventricular noncompaction 5, 613426<br>Liang distal myopathy,160500<br>Myopathy,myosin storage,autosomal dominant,608358<br>Myopathy,myosin storage,autosomal recessive,255160<br>Scapuloperoneal syndrome,myopathic type,181430 |

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| MYH8 | 128.8 | 100% | 99% | Carney complex variant,608837<br>Trismus-pseudocamptodactyly syndrome,158300 |
| MYH9 | 123.8 | 99% | 95% | Deafness,autosomal dominant 17,603622<br>Epstein syndrome,153650<br>Fechtner syndrome,153640<br>Macrothrombocytopenia and progressive sensorineural deafness,600208<br>May-Hegglin anomaly, 155100<br>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<br>Deafness,autosomal dominant 22,with hypertrophic cardiomyopathy,606346<br>Deafness,autosomal recessive 37,607821 |
| MYO7A | 127 | 98% | 95% | Deafness, autosomal dominant 11, 601317<br>Deafness, autosomal recessive 2, 600060<br>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<br>Myopathy, myofibrillar, 3, 609200<br>Myopathy, spheroid body, 182920 |
| MYOZ2 | 145.5 | 100% | 100% | Cardiomyopathy, familial hypertrophic, 16, 613838 |
| MYPN | 147 | 99% | 97% | Cardiomyopathy, dilated, 1KK, 615248<br>Cardiomypathy, familial hypertrophic, 22, 615248<br>Cardiomyopaty, familial restrictive 4, 615248 |
| NAA10 | 58 | 100% | 91% | N-terminal acetyltransferase deficiency, 300855 |

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| NAGA | 147.9 | 100% | 100% | Kanzaki disease, 609242<br>Schindler disease, type I, 609241<br>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 dysmophism, 615419 |
| NANOS1 | 36.2 | 86% | 70% | Spermatogenic failure 12,615413 |
| NBAS | 134.1 | 99% | 97% | Infantile liver failure syndrome 2,616483<br>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<br>Leukemia,acute lymphoblastic,613065<br>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<br>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<br>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 |

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| 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<br>Mitochondrial complex I deficiency, 252010 |
| NDUFS4 | 155.3 | 100% | 99% | Leigh syndrome, 256000<br>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<br>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<br>Sialidosis, type II, 256550 |
| NEUROD1 | 153.6 | 100% | 100% | Maturity-onset diabetes of the young 6, 606394<br>{Diabetes mellitus, noninsulin-dependent}, 125853 |
| NEUROG3 | 105.7 | 100% | 99% | Diarrhea 4,malabsorptive,congenital,610370 |
| NEXN | 66.2 | 87% | 71% | Cardiomyopathy, dilated, 1CC, 613122<br>Cardiomyopathy, familial hypertrophic, 20, 613876 |
| NF1 | 122.7 | 91% | 86% | Neurofibromatosis,type 1,162200 |
| NF2 | 96.7 | 100% | 100% | Neurofibromatosis, type 2, 101000<br>Meningioma, NF2-related, somatic, 607174<br>Schwannomatosis, 162091 |
| NFIX | 139.2 | 98% | 94% | Marshall-Smith syndrome, 602535<br>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 |

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| 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<br>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<br>Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978<br>{Thyroid cancer,nonmedullary,1},188550 |
| NKX2-5 | 70.2 | 100% | 94% | Atrial septal defect 7, with or without AV conduction defects, 108900<br>Conotruncal heart malformations, variable, 217095<br>Hypoplastic left heart syndrome 2, 614435<br>Hypothyroidism, congenital nongoitrous, 5, 225250<br>Tetrology of Fallot, 187500<br>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<br>{Asperger syndrome susceptibility, X-linked 2}, 300497<br>{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<br>Cold-induced autoinflammatory syndrome, familial, 120100<br>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 |

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| 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<br>Multiple synostosis syndrome 1, 186500<br>Stapes ankylosis with broad thumb and toes, 184460<br>Symphalangism, proximal, 185800<br>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<br>Aortic valve disease,109730 |
| NOTCH2 | 166.5 | 100% | 98% | Alagille syndrome 2, 610205<br>Hajdu-Cheney syndrome,102500 |
| NOTCH3 | 99.6 | 92% | 85% | ?Myofibromatosis,infantile 2,615293<br>Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy,125310 |
| NPC1 | 139.1 | 100% | 97% | Niemann-Pick disease, type C1, 257220<br>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<br>Nephronophthisis 1, juvenile, 256100<br>Senior-Loken syndrome-1,266900 |
| NPHP3 | 111.4 | 98% | 91% | Meckel syndrome 7,267010<br>Nephronophthisis 3, 604387<br>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<br>Epiphyseal chondrodysplasia,Miura type,615923<br>Short stature with nonspecific skeletal abnormalities,616255 |

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| NR0B1 | 63.6 | 96% | 91% | 46XY sex reversal 2,dosage-sensitive,300018<br>Adrenal hypoplasia, congenital, with hypogonadotropic hypogonadism,300200 |
| NR0B2 | 82.5 | 100% | 100% | Obesity,mild,early-onset,601665 |
| NR2E3 | 95.9 | 100% | 100% | Enhanced S-cone syndrome,268100<br>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<br>Aderenocortical insufficiency<br>Premature ovarian failure 7,612964<br>Spermatogenic failure 8,613957 |
| NRAS | 179.8 | 100% | 100% | ?RAS-associated autoimmune lymphoproliferative syndrome type IV,somatic,611470<br>Colorectal cancer,somatic,114500<br>Epidermal nevus,somatic,162900<br>Melanocytic nevus syndrome,congenital,somatic,137550<br>Neurocutaneous melanosis,somatic,249400<br>Noonan syndrome 6,613224<br>Schimmelpenning-Feuerstein-Mims syndrome,somatic mosaic,163200<br>Thyroid carcinoma,follicular,somatic,188470 |
| NRL | 61.1 | 98% | 86% | Retinal degeneration, autosomal recessive, clumped pigment type<br>Retinitis pigmentosa 27, 613750 |
| NRXN1 | 160 | 99% | 96% | Pitt-Hopkins-like syndrome 2, 614325<br>{Schizophrenia, susceptibility to, 17}, 614332 |
| NSD1 | 142.2 | 100% | 100% | Beckwith-Wiedemann syndrome, 130650<br>Leukemia, acute myeloid, 601626<br>Sotos syndrome 1, 117550 |
| NSDHL | 119.2 | 99% | 93% | CHILD syndrome, 308050<br>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 |

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| NT5E | 174.7 | 100% | 99% | Calcification of joints and arteries, 211800 |
| NTF4 | 42.2 | 90% | 77% | Glaucoma 1,open angle, 1O,613100 |
| NTRK1 | 115.4 | 97% | 93% | Insensitivity to pain, congenital, with anhidrosis, 256800<br>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<br>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<br>[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<br>Lowe syndrome, 309000 |
| OFD1 | 29.6 | 74% | 53% | ?Retinitis pigmentosa 23,300424<br>Joubert syndrome 10,300804<br>Oral-facial-digital syndrome 1, 311200<br>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<br>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<br>Colorblindness, protan, 303900 |
| OPN1MW | 64.3 | 65% | 50% | Blue cone monochromacy, 303700<br>Colorblindness, deutan, 303800 |
| OPN1SW | 120.3 | 100% | 100% | Colorblindness,tritan,190900 |

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| OPTN | 107.8 | 100% | 99% | Amyotrophic lateral sclerosis 12, 613435<br>Glaucoma 1, open angle, E, 137760<br>{Glaucoma, normal tension, susceptibility to}, 606657 |
| ORAI1 | 177.8 | 90% | 88% | Immunodeficiency 9,612782<br>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 transcarbamylase 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<br>Subcortical laminar heterotopia, 607432 |
| PAH | 170.6 | 100% | 100% | Phenylketonuria, 261600<br>[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<br>{Breast cancer, susceptibility to}, 114480<br>{Pancreatic cancer, susceptibility to, 3}, 613348 |
| PANK2 | 142.9 | 95% | 86% | HARP syndrome, 607236<br>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 |

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|---|---|---|---|---|
| PARK7 | 80.1 | 100% | 95% | Parkinson disease 7,autosomal recessive early-onset,606324 |
| PAX2 | 148.6 | 100% | 99% | Glomerulosclerosis, focal segmental, 7, 616002<br>Papillorenal syndrome, 120330<br>Renal hypoplasia, isolated, 191830 |
| PAX3 | 108.8 | 100% | 99% | Craniofacial-deafness-hand syndrome,122880<br>Rhabdomyosarcoma 2,alveolar,268220<br>Waardenburg syndrome,type 1,193500<br>Waardenburg syndrome,type 3,148820 |
| PAX4 | 84.5 | 100% | 97% | Diabetes mellitus,type 2,125853<br>Maturity-onset diabetes of the young,type IX,612225<br>{Diabetes mellitus,ketosis-prone,susceptibility to},612227 |
| PAX6 | 129.3 | 100% | 100% | ?Morning glory disc anomaly,120430<br>Aniridia,106210<br>Cataract with late-onset corneal dystrophy,106210<br>Coloboma of optic nerve,120430<br>Coloboma, ocular,120200<br>Foveal hypoplasia 1,136520<br>Keratitis,148190<br>Optic nerve hypoplasia,165550<br>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<br>Usher syndrome, type 1D/F digenic, 601067<br>Usher syndrome, type 1F, 602083 |
| PCDH19 | 133.3 | 100% | 95% | Epileptic encephalopathy, early infantile, 9, 300088 |
| PCM1 | 126 | 98% | 94% | No OMIM phenotype<br>Schizophrenia (Kamiya (2008) Arch Gen Psychiatry 65,996)<br>Breast cancer (Guan (2015) Fam Cancer 14,9) |
| PCNT | 108.1 | 97% | 93% | Microcephalic osteodysplastic primordial dwarfism, type II, 210720 |

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|---|---|---|---|---|
| PCSK1 | 148.2 | 100% | 99% | Obesity with impaired prohormone processing,60955<br>{Obesity,susceptibility to,BMIQ12},612362 |
| PCSK9 | 85.5 | 97% | 88% | Hypercholesterolemia,familial,3,603776<br>{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<br>{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<br>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<br>Retinal cone dystrophy 3, 610024 |
| PDE8B | 104.1 | 100% | 91% | Pigmented nodular adrenocortical disease, primary, 3, 614190<br>Striatal degeneration, autosomal dominant, 609161 |
| PDGFB | 87 | 100% | 100% | Basal ganglia calcification,idiopathic,5,615483<br>Dermatofibrosarcoma protuberans,607907<br>Meningioma, SIS-related,607174 |
| PDGFRA | 145.2 | 100% | 99% | Gastrointestinal stromal tumor,somatic,606764<br>Hypereosinophilic syndrome,idiopathic,resistant to imatinib,607685 |
| PDGFRB | 135.4 | 98% | 96% | Basal ganglia calcification idiopathic 4,615007<br>Myofibromatosis, infantile, 1, 228550<br>Myeloproliferative disorder with eosinophilia, 131440 |
| PDGFRL | 130 | 100% | 99% | Colorectal cancer,somatic,114500<br>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 |

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| PDX1 | 29.1 | 82% | 68% | MODY,type IV,606392<br>Pancreatic agenesis 1,260370<br>{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<br>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<br>Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 |
| PEX10 | 100.3 | 97% | 87% | Peroxisome biogenesis disorder 6A (Zellweger), 614870<br>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<br>Peroxisome biogenesis disorder 3B, 266510 |
| PEX13 | 194.6 | 100% | 100% | Peroxisome biogenesis disorder 11A (Zellweger), 614883<br>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<br>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<br>Peroxisome biogenesis disorder 5B, 614867 |
| PEX26 | 73.3 | 100% | 98% | Peroxisome biogenesis disorder 7A (Zellweger), 614872<br>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<br>Peroxisome biogenesis disorder 2B, 202370 |
| PEX6 | 75.8 | 86% | 82% | Peroxisome biogenesis disorder 4A (Zellweger), 614862<br>Peroxisome biogenesis disorder 4B, 614863 |
| PEX7 | 121.4 | 91% | 82% | Rhizomelic chondrodysplasia punctata, type 1, 215100<br>Peroxisome biogenesis disorder 9B, 614879 |
| PFKM | 149.2 | 100% | 100% | Glycogen storage disease VII, 232800 |

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|---|---|---|---|---|
| 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<br>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<br>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<br>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<br>{Neuroblastoma, susceptibility to, 2}, 613013<br>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<br>Arthrogryposis,distal,type 3,114300<br>Arthrogryposis,distal,type 5,108145 |
| PIGA | 56.5 | 76% | 73% | Multiple congenital anomalies-hypotonia-seizures syndrome 2,300868<br>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 |

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| PIGV | 149.2 | 100% | 100% | Hyperphosphatasia with mental retardation syndrome 1, 239300 |
| PIK3CA | 125.3 | 99% | 98% | Ovarian cancer, somatic, 167000<br>Breast cancer, somatic, 114480<br>Colorectal cancer, somatic, 114500<br>Gastric cancer, somatic, 613659<br>Hepatocellular carcinoma, somatic, 114550<br>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<br>Liebenberg syndrome,186550 |
| PITX2 | 128.1 | 96% | 93% | Axenfeld-Rieger syndrome type 1,180500<br>Iridogoniodysgenesis,type 2,137600<br>Peters anomaly,604229<br>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<br>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<br>Neurodegeneration with brain iron accumulation 2B, 610217<br>Parkinson disease 14, 612953 |

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| PLA2G7 | 123.9 | 100% | 98% | Asthma, susceptibility to, 600807<br>Atopy, susceptibility to, 147050<br>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<br>{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<br>Familial cold autoinflammatory syndrome 3, 614468 |
| PLEC | 87.5 | 98% | 95% | ?Epidermolysis bullosa simplex with nail dystrophy,616487<br>Epidermolysis bullosa simplex, Ogna type,131950<br>Epidermolysis bullosa simplex with muscular dystrophy,226670<br>Epidermolysis bullosa simplex with pyloric atresia,612138<br>Muscular dystrophy,limb-girdle,type 2Q,613723 |
| PLEKHG5 | 79.2 | 92% | 85% | Charcot-Marie-Tooth disease,recessive intermediate C,615376<br>Spinal muscular atrophy, distal, autosomal recessive, 4, 611067 |
| PLEKHM1 | 51.9 | 84% | 75% | Osteopetrosis,autosomal recessive 6,611497 |
| PLG | 114 | 87% | 87% | Dysplasminogenemia, 217090<br>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<br>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<br>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 |

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| PMP22 | 93.2 | 87% | 84% | Charcot-Marie-Tooth disease, type 1A, 118220<br>Charcot-Marie-Tooth disease,type 1E,118300<br>Dejerine-Sottas disease,145900<br>Neuropathy,inflammatory demyelinating,139393<br>Neuropathy,recurrent,with pressure palsies,162500<br>Roussy-Levy syndrome,180800 |
| PMS2 | 79.5 | 82% | 79% | Colorectal cancer, hereditary nonpolyposis, type 4, 614337<br>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<br>Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381 |
| POLE | 130.6 | 99% | 98% | {Colorectal cancer, susceptibility to, 12}, 615083<br>FILS syndrome, 615139 |
| POLG | 107.7 | 100% | 98% | Mitochondrial DNA depletion syndrome 4A (Alpers type),203700<br>Mitochondrial DNA depletion syndrome 4B (MNGIE type),613662<br>Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE),607459<br>Progressive external ophthalmoplegia, autosomal dominant,1,157640<br>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<br>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 |

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| | | | | 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<br>{Obesity,early-onset,susceptibility to},601665 |
| POMGNT1 | 107 | 100% | 95% | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280<br>Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151<br>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<br>Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155<br>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<br>Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156<br>Muscular dystrophy-dystroglycanopathy (limb-girdle),type C,2,613158 |
| POR | 139.2 | 100% | 97% | Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis,201750<br>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<br>Insulin resistance,severe,digenic,604367<br>Lipodystrophy,familial partial,type 3,604367<br>Obesity,severe,601665<br>[Obesity,resistance to]<br>{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 |

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|---|---|---|---|---|
| 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<br>Cardiomyopathy, dilated, 1LL, 615373 |
| PRDM5 | 134.3 | 100% | 96% | Brittle cornea syndrome 2,614170 |
| PRF1 | 113.4 | 100% | 99% | Hemophagocytic lymphohistiocytosis, familial, 2, 603553<br>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<br>Glycogen storage disease of heart, lethal congenital, 261740<br>Wolff-Parkinson-White syndrome, 194200 |
| PRKAR1A | 90.8 | 98% | 90% | Acrodysostosis 1, with or without hormone resistance,101800<br>Adrenocortical tumor, somatic<br>Carney complex, type 1, 160980<br>Myxoma, intracardiac, 255960<br>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<br>Multiple fibroadenomas of the breast,615554 |
| PRNP | 170.2 | 100% | 100% | Cerebral amyloid angiopathy,PRNP related,137440<br>Creutzfeldt-Jakob disease,123400<br>Gerstmann-Straussler disease,137440<br>Huntington disease-like 1,603218<br>Insomnia,fatal familial,600072 |

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| | | | | Prion disease with protracted course,606688 |
| PROC | 116.9 | 99% | 95% | Thrombophilia due to protein C deficiency, autosomal dominant, 176860<br>Thrombophilia due to protein C deficiency, autosomal recessive, 612304 |
| PRODH | 68.5 | 84% | 77% | Hyperprolinemia, type I, 239500<br>{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<br>Macular dystrophy, retinal, 2, 608051<br>Retinitis pigmentosa 41, 612095<br>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<br>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<br>Retinitis pigmentosa 7, 608133<br>Retinitis punctata albescens, 136880<br>Macular dystrophy, patterned, 169150<br>Macular dystrophy, vitelliform, 608161 |
| PRPS1 | 113.7 | 100% | 100% | Arts syndrome, 301835<br>Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070<br>Deafness, X-linked 1, 304500<br>Gout, PRPS-related, 300661<br>Phosphoribosylpyrophosphate synthetase superactivity, 300661 |
| PRRT2 | 62.2 | 100% | 99% | Convulsions,familial infantile,with paroxysmal choreoathetosis,602066<br>Episodic kinesigenic dyskinesia 1, 128200 |

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| | | | | Seizures,benign familial infantile, 2,605751 |
| PRRX1 | 94.8 | 100% | 100% | Agnathia-otocephaly complex,202650 |
| PRSS1 | 191 | 100% | 100% | Pancreatitis,hereditary,167800<br>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<br>Dejerine-Sottas disease, autosomal recessive, 145900 |
| PSAP | 111.2 | 100% | 97% | Combined SAP deficiency, 611721<br>Gaucher disease, atypical, 610539<br>Krabbe disease, atypical, 611722<br>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<br>Alzheimer disease, type 3, 607822<br>Cardiomyopathy, dilated, 1U, 613694<br>Dementia,frontotemporal,600274<br>Pick disease,172700 |
| PSEN2 | 123.4 | 100% | 100% | Alzheimer disease-4,606889<br>Cardiomyopathy,dilated,1V,613697 |
| PSENEN | 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<br>Basal cell nevus syndrome, 109400<br>Holoprosencephaly-7, 610828 |
| PTCH2 | 108.1 | 99% | 97% | Basal cell carcinoma somatic,605462<br>Basal cell nevus syndrome,109400<br>Medulloblastoma,155255 |
| PTDSS1 | 130.4 | 100% | 99% | Lenz-Majewski hyperostotic dwarfism, 151050 |

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| PTEN | 142.2 | 100% | 100% | Bannayan-Riley-Ruvalcaba syndrome,153480<br>Cowden syndrome 1,158350<br>Endometrial carcinoma, somatic,608089<br>Lhermitte-Duclos syndrome,158350<br>Macrocephaly/Autism syndrome,605309<br>Malignant melanoma,somatic,155600<br>PTEN hamartoma tumor syndrome<br>Squamous cell carcinoma,head and neck,somatic,275355<br>VATER association with macrocephaly and ventriculomegaly,276950<br>{Glioma susceptibility 2},613028<br>{Meningioma},607174<br>{Prostate cancer,somatic},176807 |
| PTF1A | 61.3 | 79% | 72% | Pancreatic agenesis 2,615935<br>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<br>Eiken syndrome, 600002<br>Failure of tooth eruption, primary, 125350<br>Metaphyseal chondrodysplasia, Murk-Jansen type, 156400 |
| PTHLH | 126.8 | 99% | 83% | Brachydactyly type E2,613382<br>Humoral hypercalcemia of malignancy |
| PTPN11 | 93 | 96% | 89% | LEOPARD syndrome 1, 151100<br>Leukemia, juvenile myelomonocytic, 607785<br>Metachondromatosis, 156250<br>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% | {Hepatitic C virus, susceptibility to}, 609532<br>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 |

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| 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<br>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<br>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<br>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<br>{Breast cancer,susceptibility to},114480 |
| RAD51C | 148.2 | 100% | 100% | Fanconi anemia, complementation group O, 613390<br>{Breast-ovarian cancer, familial, susceptibility to, 3}, 613399 |
| RAD54B | 97.1 | 96% | 88% | Colon cancer,somatic,114500<br>Lymphoma,non-Hodgkin,somatic,605027 |
| RAD54L | 112.6 | 100% | 99% | Adenocarcinoma,colonic,somatic<br>Lymphoma,non-Hodgkin,somatic,605027<br>{Breast cancer,invasive ductal},114480 |

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| RAF1 | 118.6 | 100% | 99% | Cardiomyopathy,dilated,1NN,615916<br>LEOPARD syndrome 2, 611554<br>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<br>Smith-Magenis syndrome, 182290 |
| RAP1GDS1 | 92.2 | 98% | 92% | Lymphocytic leukemia,acute T-cell |
| RAPSN | 116.4 | 94% | 93% | Fetal akinesia deformation sequence, 208150<br>Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931<br>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<br>Capillary malformation-arteriovenous malformation,608354<br>Parkes Weber syndrome,608355 |
| RAX | 64 | 94% | 72% | Microphthalmia,isolated 3,611038 |
| RAX2 | 45.5 | 90% | 69% | Cone-rod dystrophy 11, 610381<br>Macular degeneration, age-related, 6,613757 |
| RB1 | 77.5 | 91% | 76% | Bladder cancer, somatic, 109800<br>Osteosarcoma, somatic, 259500<br>Retinoblastoma, 180200<br>Retinoblastoma, trilateral, 180200<br>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<br>Pancreatic carcinoma,somatic<br>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 |

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| | | | | 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<br>RAPADILINO syndrome, 266280<br>Rothmund-Thomson syndrome, 268400 |
| REEP1 | 96.5 | 95% | 95% | Neuronopathy, distal hereditary motor, type VB, 614751<br>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<br>Renal tubular dysgenesis,267430<br>[Hyperproreninemia] |
| RET | 137.7 | 99% | 97% | Central hypoventilation syndrome, congenital, 209880<br>Medullary thyroid carcinoma, 155240<br>Multiple endocrine neoplasia IIA, 171400<br>Multiple endocrine neoplasia IIB, 162300<br>Pheochromocytoma, 171300<br>{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<br>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<br>Overhydrated hereditary stomatocytosis,185000<br>Rh-mod syndrome |
| RHBDF2 | 95.5 | 98% | 95% | Tylosis with esophageal cancer,148500 |

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| RHCE | 169.9 | 99% | 99% | Rh-null disease,amorph type<br>[Blood group,Rhesus],111690 |
| RHO | 224.6 | 100% | 100% | Night blindness, congenital stationary, autosomal dominant 1, 610445<br>Retinitis pigmentosa 4, autosomal dominant or recessive, 613731<br>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<br>Fundus albipunctatus, 136880<br>Newfoundland rod-cone dystrophy, 607476<br>Retinitis punctata albescens, 136880 |
| RMND1 | 131.5 | 99% | 90% | Combined oxidative phosphorylation deficiency 11, 614922 |
| RMRP | | | | Anauxetic dysplasia,607095<br>Cartilage-hair hypoplasia,250250<br>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<br>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 |

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| ROM1 | 97.7 | 100% | 98% | Retinitis pigmentosa 7, digenic, 608133 |
| ROR2 | 155 | 99% | 97% | Brachydactyly,type B1,113000<br>Robinow syndrome, autosomal recessive,268310 |
| RP1 | 123.7 | 99% | 96% | Retinitis pigmentosa 1, 180100<br>{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<br>Retinitis pigmentosa 20, 613794 |
| RPGR | 59.2 | 81% | 66% | Cone-rod dystrophy, X-linked, 1, 304020<br>Macular degeneration, X-linked atrophic, 300834<br>Retinitis pigmentosa 3, 300029<br>Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 |
| RPGRIP1 | 145.2 | 100% | 99% | Cone-rod dystrophy 13, 608194<br>Leber congenital amaurosis 6, 613826 |
| RPGRIP1L | 133.2 | 95% | 93% | COACH syndrome,216360<br>Joubert syndrome 7, 611560<br>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<br>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 |

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| RRM2B | 133 | 98% | 96% | Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy),612075<br>Mitochondrial DNA depletion syndrome 8B (MNGIE type),612075<br>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<br>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<br>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<br>Platelet disorder, familial, with associated myeloid malignancy, 601399 |
| RUNX2 | 94.4 | 74% | 74% | Cleidocranial dysplasia, 119600<br>Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600<br>Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600<br>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<br>Central core disease, 117000<br>Minicore myopathy with external ophthalmoplegia, 255320<br>Neuromuscular disease, congenital, with uniform type 1 fiber, 117000 King-Denborough syndrome, 145600 |
| RYR2 | 134.1 | 99% | 97% | Arrhythmogenic right ventricular dysplasia 2, 600996<br>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<br>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 |

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| 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<br>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<br>Neuropathy,hereditary sensory and autonomic,type VIII,615548 |
| SCN1A | 151.4 | 99% | 98% | Dravet syndrome, 607208<br>Epilepsy, generalized, with febrile seizures plus, type 2, 604403<br>Febrile seizures, familial, 3A, 604403<br>Migraine, familial hemiplegic, 3, 609634 |
| SCN1B | 151.7 | 100% | 96% | Atrial fibrillation, familial, 13, 615377<br>Brugada syndrome 5, 612838<br>Cardiac conduction defect, nonspecific, 612838<br>Epilepsy, generalized, with febrile seizures plus, type 1, 604233 |
| SCN2A | 153.2 | 99% | 97% | Epileptic encephalopathy, early infantile, 11, 613721<br>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<br>Hypokalemic periodic paralysis, type 2, 613345<br>Myasthenic syndrome, acetazolamide-responsive, 614198<br>Myotonia congenita, atypical, acetazolamide-responsive, 608390<br>Paramyotonia congenita, 168300 |
| SCN4B | 67.8 | 100% | 99% | Long QT syndrome-10, 611819 |

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| SCN5A | 169.5 | 100% | 99% | Atrial fibrillation, familial, 10, 614022<br>Brugada syndrome 1, 601144<br>Cardiomyopathy,dilated, 1E,601154<br>Heart block,nonprogressive,113900<br>Heart block,progressive,type 1A,113900<br>Long QT syndrome-3,603830<br>Sick sinus syndrome 1, 608567<br>Ventricular fibrillation,familial,1,603829<br>{Sudden infant death syndrome, susceptibility},272120 |
| SCN8A | 192.7 | 99% | 97% | Cognitive impairment with or without cerebellar ataxia, 614306<br>Epileptic encephalopathy, early infantile, 13, 614558 |
| SCN9A | 146.4 | 96% | 94% | Epilepsy,generalized,with febrile seizures plus,type 7,613863<br>Erythermalgia, primary, 133020<br>Febrile seizures,familial,3B,613863<br>HSAN2D,autosomal recessive,243000<br>Insensitivity to pain,congenital,243000<br>Paroxysmal extreme pain disorder,167400 |
| SCNN1A | 117.7 | 94% | 92% | Bronchiectasis with or without elevated sweat chloride 2,613021<br>Pseudohypoaldosteronism, type I, 264350 |
| SCNN1B | 133.8 | 100% | 100% | Bronchiectasis with or without elevated sweat chloride 1,211400<br>Liddle syndrome, 177200<br>Pseudohypoaldosteronism,type I,264350 |
| SCNN1G | 140.6 | 98% | 96% | Bronchiectasis with or without elevated sweat chloride 3,613071<br>Liddle syndrome, 177200<br>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<br>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<br>Leigh syndrome, 256000<br>Mitochondrial respiratory chain complex II deficiency, 252011<br>Paragangliomas 5, 614165 |
| SDHAF1 | 36.9 | 100% | 94% | Mitochondrial complex II deficiency, 252011 |

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| SDHAF2 | 142.1 | 94% | 94% | Paragangliomas 2, 601650 |
| SDHB | 124.7 | 100% | 100% | Cowden syndrome 2, 612359<br>Gastrointestinal stromal tumor, 606764<br>Paraganglioma and gastric stromal sarcoma, 606864<br>Paragangliomas 4, 115310<br>Pheochromocytoma, 171300 |
| SDHC | 94 | 99% | 92% | Gastrointestinal stromal tumor, 606764<br>Paragangliomas 3, 605373<br>Paraganglioma and gastric stromal sarcoma, 606864 |
| SDHD | 51.2 | 59% | 59% | Carcinoid tumors, intestinal, 114900<br>Cowden syndrome 3, 615106<br>Merkel cell carcinoma, somatic<br>Mitochondrial complex II deficiency,252011<br>Paraganglioma and gastric stromal sarcoma, 606864<br>Paragangliomas 1, with or without deafness, 168000<br>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<br>Retinitis pigmentosa 35, 610282 |
| SEPN1 | 95.8 | 83% | 79% | Muscular dystrophy, rigid spine, 1, 602771<br>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<br>Leukemia,acute myeloid,therapy-related<br>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<br>{Pulmonary disease,chronic obstructive,susceptibility to},606963 |
| SERPINA3 | 129 | 100% | 100% | Alpha-1-antichymotrypsin deficiency |

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| | | | | 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<br>{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<br>{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<br>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<br>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% | Mucopolysaccharidisis type 3A (Sanfilippo A), 252900 |

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| SH2B3 | 79.5 | 84% | 70% | Erythrocytosis,somatic,133100<br>Myelofibrosis,somatic,254450<br>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<br>Mononeuropathy of the median nerve,mild,613353 |
| SHANK3 | 65.5 | 79% | 70% | Phelan-McDermid syndrome, 606232<br>{Schizophrenia 15}, 613950 |
| SHH | 84.8 | 95% | 91% | Holoprosencephaly-3, 142945<br>Microphthalmia with coloboma 5, 611638<br>Schizencephaly, 269160<br>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<br>Leri-Weill dyschondrosteosis,127300<br>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<br>Deafness,autosomal dominant 23,605192 |
| SIX3 | 122.8 | 97% | 89% | Holoprosencephaly-2, 157170<br>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 |

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| 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<br>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<br>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<br>Lung cancer,somatic,211980<br>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<br>[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<br>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<br>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 |

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| SLC25A4 | 125.7 | 100% | 100% | Progressive external ophthalmoplegia with mitochondrial DNA deletions 3, 609283<br>Mitochondrial DNA depletion syndrome 12 (cardiomyopathic type), 615418 |
| SLC26A2 | 223 | 100% | 100% | Achondrogenesis Ib,600972<br>Atelosteogenesis II,256050<br>De la Chapelle dysplasia,256050<br>Diastrophic dysplasia,222600<br>Diastrophic dysplasia,broad bone-platyspondylic variant,222600<br>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<br>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<br>GLUT1 deficiency syndrome 1, 606777<br>GLUT1 deficiency syndrome 2, 612126<br>Stomatin-deficient cryohydrocytosis with neurologic defects,608885<br>{Epilepsy, idiopathic generalized,susceptibility,12},614847 |
| SLC2A10 | 138.4 | 100% | 99% | Arterial tortuosity syndrome,208050 |
| SLC2A2 | 154.8 | 100% | 100% | Fanconi-Bickel syndrome,227810<br>{Diabetes mellitus, noninsulin-dependent},125853 |
| SLC2A9 | 127.5 | 100% | 93% | Hypouricemia,renal,2,612076<br>{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<br>Spastic paraplegia 42, autosomal dominant, 612539 |
| SLC34A1 | 131.8 | 100% | 99% | Fanconi renotubular syndrome 2,613388<br>Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286 |
| SLC34A2 | 162.5 | 100% | 99% | ?Testicular microlithiasis,610441<br>Pulmonary alveolar microlithiasis,265100 |
| SLC34A3 | 104.9 | 95% | 94% | Hypophosphatemic rickets with hypercalciuria, 241530 |

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| 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<br>Iminoglycinuria,digenic,242600 |
| SLC37A4 | 119.5 | 100% | 100% | Glycogen storage disease Ib, 232220<br>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<br>[skin/hair/eye pigmentation 5],227240 |
| SLC46A1 | 90.7 | 97% | 88% | Folate malabsorption, hereditary, 229050 |
| SLC4A1 | 115.7 | 100% | 99% | Ovalocytosis<br>Renal tubular acidosis,distal,AD,179800<br>Renal tubular acidosis,distal,AR,611590<br>Spherocytosis,type 4,612653 |
| SLC4A11 | 133.3 | 100% | 99% | Corneal dystrophy, Fuchs endothelial, 4, 613268<br>Corneal endothelial dystrophy 2, autosomal recessive, 217700<br>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<br>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<br>Hyperglycinuria,138500 |

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| | | | | 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<br>{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% | ?Thrichotillomania,613229<br>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<br>Myhre syndrome, 139210<br>Pancreatic cancer,somatic,260350<br>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<br>Rhabdoid tumor predisposition syndrome 2, 613325 |
| SMARCAD1 | 83 | 99% | 94% | Adermatoglyphia,136000 |
| SMARCAL1 | 121.1 | 100% | 98% | Schimke immunoosseous dysplasia, 242900 |

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| SMARCB1 | 202.6 | 100% | 100% | Coffin-Siris syndrome 3,614608<br>Rhabdoid tumors,somatic,609322<br>{Rhabdoid predisposition syndrome 1},609322<br>{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<br>Spinal muscular atrophy-2, 253550<br>Spinal muscular atrophy-3, 253400<br>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<br>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<br>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<br>Parkinson disease 1, 168601<br>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 |

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| SOS1 | 87.5 | 93% | 88% | Fibromatosis, gingival, 135300<br>Noonan syndrome 4, 610733 |
| SOST | 107.6 | 100% | 97% | Craniodiaphyseal dysplasia,autosomal dominant,122860<br>Sclerosteosis 1,269500<br>Van Buchem disease,239100 |
| SOX10 | 65.7 | 92% | 88% | PCWH syndrome, 609136<br>Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584<br>Waardenburg syndrome, type 4C, 613266 |
| SOX17 | 59.6 | 99% | 83% | Vesicoureteral reflux 3, 613674 |
| SOX18 | 13.9 | 56% | 44% | Hypotrichosis-lymphedema-telangiectasia syndrome,607823<br>Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome,137940 |
| SOX2 | 80.8 | 99% | 96% | Microphthalmia, syndromic 3, 206900<br>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<br>Panhypopituitarism, X-linked, 312000 |
| SOX9 | 96.4 | 94% | 87% | Acampomelic campomelic dysplasia,114290<br>Campomelic dysplasia,114290<br>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<br>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<br>Tropical calcific pancreatitis,608189 |
| SPINK5 | 154.4 | 100% | 95% | Netherton syndrome, 256500 |
| SPINT2 | 54.1 | 94% | 77% | Diarrhea 3 secretory sodium congenital syndromic,270420 |

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|---|---|---|---|---|
| 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<br>Pyropoikilocytosis,266140<br>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<br>Elliptocytosis-3<br>Spherocytosis,type 2,616649 |
| SPTBN2 | 103 | 99% | 98% | Spinocerebellar ataxia 5, 600224<br>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<br>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<br>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<br>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 |

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| STAT5B | 116.6 | 100% | 95% | Growth hormone insensitivity with immunodeficiency, 245590<br>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<br>Myopathy, tubular aggregate, 1, 160565<br>Stormorken syndrome, 185070 |
| STK11 | 112.3 | 100% | 97% | Melanoma, malignant, somatic<br>Peutz-Jeghers syndrome, 175200<br>Pancreatic cancer, 260350<br>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<br>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 IB, 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<br>{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<br>{Pregnancy loss,susceptibility to} |
| SYN1 | 44.3 | 70% | 56% | Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491 |

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|---|---|---|---|---|
| SYNE1 | 137.1 | 99% | 98% | Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998<br>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<br>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<br>Deafness,autosomal dominant 65,616044<br>DOOR syndrome,220500<br>Epileptic encephalopathy, early infantile, 16, 615338<br>Myoclonic epilepsy, infantile, familial, 605021 |
| TBCE | 147.1 | 99% | 98% | Hypoparathyroidism-retardation-dysmorphism syndrome, 241410<br>Kenny-Caffey syndrome-1, 244460 |

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|---|---|---|---|---|
| TBP | 118.9 | 100% | 94% | Spinocerebellar ataxia 17,607136<br>{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<br>?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<br>?Thromboxane synthase deficiency, 614158 |
| TCAP | 59.8 | 100% | 100% | Cardiomyopathy, dilated, 1N, 607487<br>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<br>Orofaciodigital syndrome IV, 258860 |
| TDGF1 | 137.6 | 99% | 85% | Forebrain defects<br>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<br>Deafness,autosomal recessive 21,603629 |

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|---|---|---|---|---|
| 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<br>{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<br>Hereditary motor and sensory neuropathy,Okinawa type,604484 |
| TFR2 | 86.8 | 98% | 95% | Hemochromatosis, type 3, 604250 |
| TG | 119 | 100% | 98% | Thyroid dyshormonogenesis 3,274700<br>{Autoimmune thyroid disease,susceptibility to},608175 |
| TGFB1 | 63.6 | 100% | 99% | Camurati-Engelmann disease, 131300<br>{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<br>Corneal dystrophy, epithelial basement membrane, 121820<br>Corneal dystrophy, Groenouw type I, 121900<br>Corneal dystrophy, lattice type I, 122200<br>Corneal dystrophy, lattice type IIIA, 608471 |
| TGFBR1 | 196.2 | 93% | 93% | Loeys-Dietz syndrome, type 1, 609192<br>{Multiple self-healing squamous epithelioma, susceptiblity to}, 132800 |
| TGFBR2 | 184.5 | 100% | 100% | Colorectal cancer, hereditary nonpolyposis, type 6, 614331<br>Esophageal cancer, somatic, 133239<br>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 |

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| THAP1 | 139.3 | 100% | 100% | Dystonia 6, torsion, 602629 |
| THBD | 85.5 | 100% | 91% | Thrombophilia due to thrombomodulin defect, 614486<br>{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<br>Thyroid hormone resistance, autosomal recessive, 274300<br>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<br>Revesz syndrome, 268130 |
| TJP2 | 108.5 | 99% | 99% | Cholestasis, progressive familial intrahepatic 4, 615878<br>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<br>{Colorectal cancer,susceptibility to},114500<br>{Macular degeneration,age-related,10},611488 |
| TMC1 | 136.6 | 95% | 94% | Deafness,autosomal dominant 36,606705<br>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<br>Meckel syndrome 2,603194 |
| TMEM231 | 86.1 | 99% | 95% | Joubert syndrome 20, 614970<br>Meckel syndrome 11,615397 |
| TMEM237 | 96.1 | 99% | 97% | Joubert syndrome 14, 614424 |

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| TMEM38B | 120.6 | 100% | 100% | Osteogenesis imperfecta,type XIV,615066 |
| TMEM43 | 116.8 | 100% | 98% | Arrhythmogenic right ventricular dysplasia 5, 604400<br>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<br>Joubert syndrome 6,610688<br>Meckel syndrome 3,607361<br>Nephronophthisis 11,613550<br>{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 hydrolylase 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<br>Paget disease of bone, 602080<br>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<br>Cardiomyopathy, familial hypertrophic, 13, 613243 |
| TNNI2 | 99.5 | 100% | 99% | Arthrogryposis multiplex congenita, distal, type 2B, 601680 |
| TNNI3 | 89.6 | 100% | 84% | Cardiomyopathy, dilated, 1FF, 613286<br>Cardiomyopathy, dilated, 2A, 611880<br>Cardiomyopathy, familial hypertrophic, 7, 613690<br>Cardiomyopathy, familial restrictive, 115210 |
| TNNT1 | 86 | 97% | 93% | Nemaline myopathy 5, Amish type, 605355 |

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| TNNT2 | 100.7 | 100% | 100% | Cardiomyopathy, dilated, 1D, 601494<br>Cardiomyopathy, familial hypertrophic, 2, 115195<br>Cardiomyopathy, familial restrictive, 3, 612422<br>Left ventricular noncompaction 6, 601494 |
| TNNT3 | 95.2 | 99% | 92% | Arthrogryposis,distal,type 2B,601680 |
| TNXB | 13.3 | 48% | 20% | Ehlers-Danlos syndrome due to tenascin X deficiency,606408<br>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<br>Dystonia, early-onset atypical, with myoclonic features<br>{Dystonia-1, modifier of} |
| TP53 | 86.8 | 98% | 91% | Adrenal cortical carcinoma, 202300<br>Breast cancer,114480<br>Choroid plexus papilloma, 260500<br>Colorectal cancer, 114500<br>Hepatocellular carcinoma, 114550<br>Li-Fraumeni syndrome, 151623<br>Nasopharyngeal carcinoma,607107<br>Osteosarcoma, 259500<br>Pancreatic cancer,260350<br>{Basal cell carcinoma 7},614740<br>{Glioma susceptibility 1},137800<br><br>Nasopharyngeal carcinoma, 607107<br>Pancreatic cancer, 260350 |
| TP63 | 186.5 | 100% | 100% | ADULT syndrome,103285<br>Ectrodactyly,ectodermal dysplasia,cleft lip/palate syndrome 3,604292<br>Hay-Wells syndrome,106260<br>Limb-mammary syndrome,603543<br>Orofacial cleft 8,129400<br>Rapp-Hodgkin syndrome,129400<br>Split-hand/foot malformation 4,605289 |
| TPI1 | 85.5 | 98% | 95% | Hemolytic anemia due to triosephosphate isomerase deficiency |

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| TPK1 | 117 | 100% | 95% | Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458 |
| TPM1 | 135.4 | 100% | 97% | Cardiomyopathy, dilated, 1Y, 611878<br>Cardiomyopathy, familial hypertrophic, 3, 115196<br>Left ventricular noncompaction 9, 611878 |
| TPM2 | 104 | 100% | 97% | Arthrogryposis, distal, type 2B, 601680<br>Arthrogryposis multiplex congenita, distal, type 1, 108120<br>CAP myopathy 2, 609285<br>Nemaline myopathy 4, autosomal dominant, 609285 |
| TPM3 | 91.3 | 89% | 88% | CAP myopathy 1, 609284<br>Myopathy congenital, with fiber-type disproportion, 255310<br>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<br>Chilblain lupus, 610448<br>Vasculopathy, retinal, with cerebral leukodystrophy, 192315<br>{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<br>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<br>Liver failure, transient infantile, 613070 |

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|---|---|---|---|---|
| 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<br>Trichorhinophalangeal syndrome,type III,190351 |
| TRPV3 | 157.8 | 100% | 99% | ?Palmoplantar keratoderma,nonepidermolytic,focal 2,616400<br>Olmsted syndrome,614594 |
| TRPV4 | 173.3 | 99% | 98% | Brachyolmia type 3, 113500<br>Hereditary motor and sensory neuropathy, type IIc, 606071<br>Metatropic dysplasia, 156530<br>Scapuloperoneal spinal muscular atrophy, 181405<br>Spondylometaphyseal dysplasia, Kozlowski type, 184252<br>[Sodium serum level QTL 1], 613508 |
| TSC1 | 127.2 | 99% | 97% | Focal cortical dysplasia, Taylor balloon cell type, 607341<br>Lymphangioleiomyomatosis, 606690<br>Tuberous sclerosis-1, 191100 |
| TSC2 | 111.2 | 99% | 98% | Lymphangioleiomyomatosis, somatic, 606690<br>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<br>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<br>Hyperthyroidism,familial gestational,603373<br>Hyperthyroidism,nonautoimmune,609152<br>Thyroid adenoma,hyperfunctioning,somatic<br>Thyroid carcinoma with thyrotoxicosis |
| TSHZ1 | 150.3 | 98% | 98% | Aural atresia,congenital,607842 |
| TSPAN12 | 135.9 | 100% | 98% | Exudative vitreoretinopathy 5, 613310 |

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|---|---|---|---|---|
| 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<br>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<br>Bardet-Biedl syndrome 8, 615985 |
| TTI2 | 92.8 | 99% | 97% | Mental retardation, autosomal recessive 39, 615541 |
| TTN | 196 | 98% | 97% | Cardiomyopathy,dilated,1G,604145<br>Cardiomyopathy,familial hypertrophic,9,613765<br>Muscular dystrophy,limb-girdle,type 2J,608807<br>Myopathy,early-onset,with fatal cardiomyopathy,611705<br>Myopathy,proximal,with early respiratory muscular involvement,603689<br>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<br>Carpal tunnel syndrome,familial,115430<br>[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<br>Fibrosis of extraocular muscles,congenital,3A,600638 |
| TUBB4A | 99.9 | 96% | 92% | Dystonia 4, torsion, autosomal dominant, 128101<br>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 |

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|---|---|---|---|---|
| TULP1 | 88.6 | 98% | 93% | Leber congenital amaurosis 15, 613843<br>Retinitis pigmentosa 14, 600132 |
| TUSC3 | 130 | 100% | 98% | Mental retardation, autosomal recessive 7, 611093 |
| TWIST1 | 104.6 | 97% | 87% | Craniosynostosis, type 1, 123100<br>Robinow-Sorauf syndrome, 180750<br>Saethre-Chotzen syndrome, 101400<br>Saethre-Chotzen syndrome with eyelid anomalies, 101400 |
| TWIST2 | 122.6 | 99% | 91% | Ablepharon-macrostomia syndrome,200110<br>Barber-Say syndrome,209885<br>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<br>Albinism, oculocutaneous, type IB, 606952<br>Waardenburg syndrome/albinism, digenic, 103470<br>[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<br>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<br>Crigler-Najjar syndrome, type II, 606785<br>Hyperbilirubinemia, familial transcient neonatal, 237900<br>[Bilirubin, serum level of, QTL1], 601816<br>[Gilbert syndrome], 143500 |
| UMOD | 109.9 | 97% | 95% | Glomerulocystic kidney disease with hyperuricemia and isothenuria,609886<br>Hyperuricemic nephropathy, familial juvenile 1, 162000<br>Medullary cystic kidney disease 2,603860 |

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|---|---|---|---|---|
| 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<br>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<br>Deafness, autosomal recessive 18A, 602092 |
| USH1G | 153.6 | 95% | 93% | Usher syndrome, type 1G, 606943 |
| USH2A | 156.8 | 99% | 98% | Retinitis pigmentosa 39, 613809<br>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<br>{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<br>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<br>Cardiomyopathy, familial hypertrophic, 15, 613255 |
| VCP | 147 | 100% | 99% | Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954<br>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<br>?Osteoporosis,involutional,166710 |

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|---|---|---|---|---|
| VHL | 91.1 | 98% | 75% | Erythrocytosis, familial, 2, 263400<br>Hemangioblastoma, cerebellar, somatic<br>Pheochromocytoma, 171300<br>Renal cell carcinoma, somatic, 144700<br>von Hippel-Lindau syndrome, 193300 |
| VIM | 117 | 100% | 93% | Cataract 30, pulverulent, 116300 |
| VIPAS39 | 135.4 | 100% | 100% | Arthrogryposis, renal dysfunction, and cholestasis 2, 613404 |
| VKORC1 | 132.8 | 100% | 100% | Vitamin K-dependent clotting factors,combined deficiency of,2,607473<br>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% | Arthrogryposis, 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<br>Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195<br>Keratoconus 1, 148300 |
| VSX2 | 67.6 | 98% | 88% | Microphthalmia, isolated 2, 610093<br>Microphthalmia with coloboma 3, 610092 |
| VWF | 107.4 | 99% | 98% | von Willebrand disease, type 1,193400<br>von Willebrand disease,type 2A,2B,2M and 2N,613554<br>von Willebrand disease,type 3,277480 |
| WAS | 35.9 | 81% | 62% | Neutropenia, severe congenital, X-linked, 300299<br>Thrombocytopenia, X-linked, 313900<br>Thrombocytopenia, X-linked, intermittent, 313900<br>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<br>?Short-rib thoracic dysplasia 5 with or without polydactyly,614376<br>Nephronophthisis 13,614377 |

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| | | | | 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<br>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<br>Deafness,autosomal dominant 6/14/38,600965<br>Wolfram syndrome,222300<br>Wolfram-like syndrome,autosomal dominant,614296<br>{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<br>Spondyloepiphyseal dysplasia tarda with progressive arthropathy,208230 |
| WNK1 | 159.4 | 98% | 97% | Neuropathy,hereditary sensory and autonomic type II,201300<br>Pseudohypoaldosteronism, type IIC, 614492 |
| WNK4 | 107.1 | 97% | 92% | Pseudohypoaldosteronism, type IIB, 614491 |
| WNT1 | 159.8 | 100% | 100% | Osteogenesis imperfecta,type XV,615220<br>{Osteoporosis,early-onset,susceptibility to,autosomal dominant,615221 |
| WNT10A | 90.5 | 100% | 96% | Odontoonychodermal dysplasia,257980<br>Schopf-Schulz-Passarge syndrome,224750<br>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<br>SERKAL syndrome, 611812 |
| WNT5A | 138.5 | 100% | 100% | Robinow syndrome autosomal dominant,180700 |

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| WNT7A | 198.8 | 100% | 100% | Fuhrmann syndrome,228930<br>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<br>Frasier syndrome, 136680<br>Meacham syndrome, 608978<br>Mesothelioma, somatic, 156240<br>Nephrotic syndrome, type 4, 256370<br>Wilms tumor, type 1, 194070 |
| WWOX | 122.5 | 100% | 99% | Epileptic encephalopathy, early infantile, 28, 616211<br>Esophageal squamous cell carcinoma, somatic, 133239<br>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<br>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 |
| ZDHHC9 | 32.9 | 93% | 74% | Mental retardation, X-linked syndromic, Raymond type, 300799 |
| ZEB1 | 188 | 100% | 99% | Corneal dystrophy, Fuchs endothelial, 6, 613270<br>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 |

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|---|---|---|---|---|
| ZFPM2 | 188.7 | 100% | 100% | Diaphragmatic hernia 3, 610187<br>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<br>Heterotaxy, visceral, 1, X-linked 306955<br>VACTERL association, X-linked, 314390 |
| ZMPSTE24 | 125.2 | 100% | 97% | Mandibuloacral dysplasia with type B lipodystrophy,608612<br>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<br>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 HGCN 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. From DG 2.5 to DG 2.6 no changes were made to the content of the gene panels.*

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