## Gene panel information

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| **Gene panel** | **BeGECS** |
| **Version** | 3 |
| **Total genes** | 2818 |
| **Activation date** | Friday 03 april 2026 |
| **Publisher** | Center for Medical Genetics, Ghent |

## Genes

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **AAAS** | 99.43 % | 605378 | Achalasia-addisonianism-alacrimia syndrome, 231550 (3), Autosomal recessive |
| **AARS1** | 99.82 % | 601065 | Developmental and epileptic encephalopathy 29, 616339 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2N, 613287 (3), Autosomal dominant; ?Leukoencephalopathy, hereditary diffuse, with spheroids 2, 619661 (3), Autosomal dominant; Trichothiodystrophy 8, nonphotosensitive, 619691 (3), Autosomal recessive |
| **AARS2** | 99.71 % | 612035 | Leukoencephalopathy, progressive, with ovarian failure, 615889 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 8, 614096 (3), Autosomal recessive |
| **AASS** | 98.76 % | 605113 | Hyperlysinemia, 238700 (3), Autosomal recessive |
| **ABAT** | 99.93 % | 137150 | GABA-transaminase deficiency, 613163 (3), Autosomal recessive |
| **ABCA1** | 99.79 % | 600046 | Tangier disease, 205400 (3), Autosomal recessive; HDL deficiency, familial, 1, 604091 (3), Autosomal dominant |
| **ABCA12** | 99.81 % | 607800 | Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500 (3), Autosomal recessive; Ichthyosis, congenital, autosomal recessive 4A, 601277 (3), Autosomal recessive |
| **ABCA2** | 100 % | 600047 | Intellectual developmental disorder with poor growth and with or without seizures or ataxia, 618808 (3), Autosomal recessive |
| **ABCA3** | 99.75 % | 601615 | Surfactant metabolism dysfunction, pulmonary, 3, 610921 (3), Autosomal recessive |
| **ABCA4** | 99.11 % | 601691 | Retinal dystrophy, early-onset severe, 248200 (3), Autosomal recessive; Retinitis pigmentosa 19, 601718 (3), Autosomal recessive; {Macular degeneration, age-related, 2}, 153800 (3), Autosomal dominant; Cone-rod dystrophy 3, 604116 (3), Autosomal recessive; Fundus flavimaculatus, 248200 (3), Autosomal recessive; Stargardt disease 1, 248200 (3), Autosomal recessive |
| **ABCA5** | 99.75 % | 612503 | ?Hypertrichosis, congenital generalized, with gingival hyperplasia, 135400 (3), Autosomal recessive |
| **ABCB11** | 99.58 % | 603201 | Cholestasis, benign recurrent intrahepatic, 2, 605479 (3), Autosomal recessive; Cholestasis, progressive familial intrahepatic 2, 601847 (3), Autosomal recessive |
| **ABCB4** | 99.47 % | 171060 | Gallbladder disease 1, 600803 (3), Autosomal recessive, Autosomal dominant; Cholestasis, intrahepatic, of pregnancy, 3, 614972 (3), Autosomal recessive, Autosomal dominant; Cholestasis, progressive familial intrahepatic 3, 602347 (3), Autosomal recessive |
| **ABCB7** | 99.6 % | 300135 | Anemia, sideroblastic, with ataxia, 301310 (3), X-linked |
| **ABCC2** | 99.7 % | 601107 | Dubin-Johnson syndrome, 237500 (3), Autosomal recessive |
| **ABCC6** | 96.83 % | 603234 | Pseudoxanthoma elasticum, 264800 (3), Autosomal recessive; Arterial calcification, generalized, of infancy, 2, 614473 (3), Autosomal recessive; Pseudoxanthoma elasticum, forme fruste, 177850 (3), Autosomal dominant |

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|---|---|---|---|
| **ABCC8** | 99.78 % | 600509 | Diabetes mellitus, permanent neonatal 3, 618857 (3), Autosomal recessive, Autosomal dominant; Maturity-onset diabetes of the young, type 12, 621196 (3), Autosomal dominant; Diabetes mellitus, transient neonatal 2, 610374 (3), Autosomal dominant; Diabetes mellitus, noninsulin-dependent, 125853 (3), Autosomal dominant; Hypoglycemia of infancy, leucine-sensitive, 240800 (3), Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 1, 256450 (3), Autosomal recessive, Autosomal dominant |
| **ABCC9** | 99.79 % | 601439 | Cardiomyopathy, dilated, 1O, 608569 (3), Autosomal dominant; Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850 (3), Autosomal dominant; ?Atrial fibrillation, familial, 12, 614050 (3), Autosomal dominant; Intellectual disability and myopathy syndrome, 619719 (3), Autosomal recessive |
| **ABCD1** | 99.81 % | 300371 | Adrenoleukodystrophy, 300100 (3), X-linked recessive; Adrenomyeloneuropathy, adult, 300100 (3), X-linked recessive |
| **ABCD3** | 97.8 % | 170995 | ?Bile acid synthesis defect, congenital, 5, 616278 (3), Autosomal recessive |
| **ABCD4** | 99.8 % | 603214 | Methylmalonic aciduria and homocystinuria, cblJ type, 614857 (3), Autosomal recessive |
| **ABCG5** | 99.83 % | 605459 | Sitosterolemia 2, 618666 (3), Autosomal recessive |
| **ABCG8** | 99.84 % | 605460 | Sitosterolemia 1, 210250 (3), Autosomal recessive; {Gallbladder disease 4}, 611465 (3) |
| **ABHD12** | 99.88 % | 613599 | Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674 (3), Autosomal recessive |
| **ABHD16A** | 99.91 % | 142620 | Spastic paraplegia 86, autosomal recessive, 619735 (3), Autosomal recessive |
| **ABHD5** | 99.8 % | 604780 | Chanarin-Dorfman syndrome, 275630 (3), Autosomal recessive |
| **ACACA** | 99.98 % | 200350 | Acetyl-CoA carboxylase deficiency, 613933 (3), Autosomal recessive |
| **ACAD8** | 99.89 % | 604773 | Isobutyryl-CoA dehydrogenase deficiency, 611283 (3), Autosomal recessive |
| **ACAD9** | 100 % | 611103 | Mitochondrial complex I deficiency, nuclear type 20, 611126 (3), Autosomal recessive |
| **ACADM** | 96.58 % | 607008 | Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450 (3), Autosomal recessive |
| **ACADS** | 99.84 % | 606885 | Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470 (3), Autosomal recessive |
| **ACADSB** | 99.78 % | 600301 | 2-methylbutyrylglycinuria, 610006 (3), Autosomal recessive |
| **ACADVL** | 99.92 % | 609575 | VLCAD deficiency, 201475 (3), Autosomal recessive |
| **ACAN** | 91.04 % | 155760 | ?Spondyloepiphyseal dysplasia, Kimberley type, 608361 (3), Autosomal dominant; Short stature and advanced bone age, with or without early-onset osteoarthritis and/or osteochondritis dissecans, 165800 (3), Autosomal dominant; Spondyloepimetaphyseal dysplasia, aggrecan type, 612813 (3), Autosomal recessive |
| **ACAT1** | 99.88 % | 607809 | Alpha-methylacetoacetic aciduria, 203750 (3), Autosomal recessive |
| **ACBD5** | 99.9 % | 616618 | Retinal dystrophy with leukodystrophy, 618863 (3), Autosomal recessive |
| **ACBD6** | 97.92 % | 616352 | Neurodevelopmental disorder with progressive movement abnormalities, 620785 (3), Autosomal recessive |
| **ACD** | 99.7 % | 609377 | ?Dyskeratosis congenita, autosomal recessive 7, 616553 (3), Autosomal recessive, Autosomal dominant; ?Dyskeratosis congenita, autosomal dominant 6, 616553 (3), Autosomal recessive, Autosomal dominant |
| **ACE** | 99.54 % | 106180 | {Stroke, hemorrhagic}, 614519 (3); Renal tubular dysgenesis, 267430 (3), Autosomal recessive; {Myocardial infarction, susceptibility to} (3); {Microvascular complications of diabetes 3}, 612624 (3); [Angiotensin I-converting enzyme, benign serum increase] (3); {SARS, progression of} (3) |

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|---|---|---|---|
| **ACER3** | 99.52 % | 617036 | ?Leukodystrophy, progressive, early childhood-onset, 617762 (3), Autosomal recessive |
| **ACO2** | 99.96 % | 100850 | Optic atrophy 9, 616289 (3), Autosomal recessive, Autosomal dominant; Infantile cerebellar-retinal degeneration, 614559 (3), Autosomal recessive |
| **ACOX1** | 99.93 % | 609751 | Mitchell syndrome, 618960 (3), Autosomal dominant; Peroxisomal acyl-CoA oxidase deficiency, 264470 (3), Autosomal recessive |
| **ACOX2** | 99.54 % | 601641 | Bile acid synthesis defect, congenital, 6, 617308 (3), Autosomal recessive |
| **ACP4** | 99.71 % | 606362 | Amelogenesis imperfecta, type IJ, 617297 (3), Autosomal recessive |
| **ACP5** | 99.91 % | 171640 | Spondyloenchondrodysplasia with immune dysregulation, 607944 (3), Autosomal recessive |
| **ACSF3** | 99.49 % | 614245 | Combined malonic and methylmalonic aciduria, 614265 (3), Autosomal recessive |
| **ACSL4** | 99.62 % | 300157 | Intellectual developmental disorder, X-linked 63, 300387 (3), X-linked dominant |
| **ACSL5** | 99.77 % | 605677 | ?Diarrhea 13, 620357 (3), Autosomal recessive |
| **ACTA1** | 99.94 % | 102610 | Congenital myopathy 2B, severe infantile, autosomal recessive, 620265 (3), Autosomal recessive; ?Myopathy, scapulohumeroperoneal, 616852 (3), Autosomal dominant; Congenital myopathy 2C, severe infantile, autosomal dominant, 620278 (3), Autosomal dominant; Congenital myopathy 2A, typical, autosomal dominant, 161800 (3), Autosomal dominant |
| **ACTL6B** | 99.21 % | 612458 | Developmental and epileptic encephalopathy 76, 618468 (3), Autosomal recessive; Intellectual developmental disorder with severe speech and ambulation defects, 618470 (3), Autosomal dominant |
| **ACY1** | 100 % | 104620 | Aminoacylase 1 deficiency, 609924 (3), Autosomal recessive |
| **ADA** | 99.81 % | 608958 | Adenosine deaminase deficiency, partial, 102700 (3), Autosomal recessive, Somatic mosaicism; Severe combined immunodeficiency due to ADA deficiency, 102700 (3), Autosomal recessive, Somatic mosaicism |
| **ADA2** | 99.97 % | 607575 | Sneddon syndrome, 182410 (3), Autosomal recessive; Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688 (3), Autosomal recessive |
| **ADAM22** | 98.91 % | 603709 | Developmental and epileptic encephalopathy 61, 617933 (3), Autosomal recessive |
| **ADAM9** | 99.84 % | 602713 | Cone-rod dystrophy 9, 612775 (3), Autosomal recessive |
| **ADAMTS10** | 99.84 % | 608990 | Weill-Marchesani syndrome 1, recessive, 277600 (3), Autosomal recessive |
| **ADAMTS13** | 99.89 % | 604134 | Thrombotic thrombocytopenic purpura, hereditary, 274150 (3), Autosomal recessive |
| **ADAMTS17** | 99.86 % | 607511 | Weill-Marchesani 4 syndrome, recessive, 613195 (3), Autosomal recessive |
| **ADAMTS18** | 99.86 % | 607512 | Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458 (3), Autosomal recessive |
| **ADAMTS19** | 99.77 % | 607513 | Cardiac valvular dysplasia 2, 620067 (3), Autosomal recessive |
| **ADAMTS2** | 99.78 % | 604539 | Ehlers-Danlos syndrome, dermatosparaxis type, 225410 (3), Autosomal recessive |
| **ADAMTS3** | 99.8 % | 605011 | Hennekam lymphangiectasia-lymphedema syndrome 3, 618154 (3), Autosomal recessive |
| **ADAMTS9** | 99.64 % | 605421 | *No OMIM phenotypes* |
| **ADAMTSL2** | 99.69 % | 612277 | Geleophysic dysplasia 1, 231050 (3), Autosomal recessive |
| **ADAMTSL4** | 99.12 % | 610113 | Ectopia lentis et pupillae, 225200 (3), Autosomal recessive; Ectopia lentis, isolated, autosomal recessive, 225100 (3), Autosomal recessive |
| **ADAR** | 98.86 % | 146920 | Dyschromatosis symmetrica hereditaria, 127400 (3), Autosomal dominant; Aicardi-Goutieres syndrome 6, 615010 (3), Autosomal recessive |
| **ADARB1** | 96.97 % | 601218 | Neurodevelopmental disorder with hypotonia, microcephaly, and seizures, 618862 (3), Autosomal recessive |

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|---|---|---|---|
| **ADAT3** | 99.28 % | 615302 | Neurodevelopmental disorder with brain abnormalities, poor growth, and dysmorphic facies, 615286 (3), Autosomal recessive |
| **ADCY1** | 99.1 % | 103072 | ?Deafness, autosomal recessive 44, 610154 (3), Autosomal recessive |
| **ADCY3** | 99.6 % | 600291 | {Obesity, susceptibility to, BMIQ19}, 617885 (3), Autosomal recessive |
| **ADCY5** | 99.69 % | 600293 | Dyskinesia with orofacial involvement, autosomal dominant, 606703 (3), Autosomal dominant; Neurodevelopmental disorder with hyperkinetic movements and dyskinesia, 619651 (3), Autosomal recessive; Dyskinesia with orofacial involvement, autosomal recessive, 619647 (3), Autosomal recessive |
| **ADCY6** | 99.71 % | 600294 | Lethal congenital contracture syndrome 8, 616287 (3), Autosomal recessive |
| **ADD3** | 99.94 % | 601568 | Cerebral palsy, spastic quadriplegic, 3, 617008 (3), Autosomal recessive |
| **ADGRG1** | 99.13 % | 604110 | Cortical dysplasia, complex, with other brain malformations 14B, (bilateral perisylvian), 615752 (3); Cortical dysplasia, complex, with other brain malformations 14A, (bilateral frontoparietal), 606854 (3), Autosomal recessive |
| **ADGRG6** | 99.76 % | 612243 | Lethal congenital contracture syndrome 9, 616503 (3), Autosomal recessive |
| **ADGRV1** | 99.71 % | 602851 | Usher syndrome, type 2C, 605472 (3), Digenic dominant, Autosomal recessive; Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472 (3), Digenic dominant, Autosomal recessive; ?Febrile seizures, familial, 4, 604352 (3), Autosomal dominant |
| **ADIPOR1** | 99.65 % | 607945 | *No OMIM phenotypes* |
| **ADK** | 99.63 % | 102750 | Hypermethioninemia due to adenosine kinase deficiency, 614300 (3), Autosomal recessive |
| **ADPRS** | 99.57 % | 610624 | Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170 (3), Autosomal recessive |
| **ADSL** | 99.71 % | 608222 | Adenylosuccinase deficiency, 103050 (3), Autosomal recessive |
| **ADSS1** | 99.6 % | 612498 | Myopathy, distal, 5, 617030 (3), Autosomal recessive |
| **AEBP1** | 99.46 % | 602981 | Ehlers-Danlos syndrome, classic-like, 2, 618000 (3), Autosomal recessive |
| **AFF2** | 99.82 % | 300806 | Intellectual developmental disorder, X-linked 109, 309548 (3), X-linked recessive |
| **AFG3L2** | 99.9 % | 604581 | Spastic ataxia 5, autosomal recessive, 614487 (3), Autosomal recessive; Optic atrophy 12, 618977 (3), Autosomal dominant; Spinocerebellar ataxia 28, 610246 (3), Autosomal dominant |
| **AGA** | 99.77 % | 613228 | Aspartylglucosaminuria, 208400 (3), Autosomal recessive |
| **AGBL5** | 99.66 % | 615900 | Retinitis pigmentosa 75, 617023 (3), Autosomal recessive |
| **AGK** | 99.91 % | 610345 | Cataract 38, autosomal recessive, 614691 (3), Autosomal recessive; Sengers syndrome, 212350 (3), Autosomal recessive |
| **AGL** | 97.75 % | 610860 | Glycogen storage disease IIIa, 232400 (3), Autosomal recessive; Glycogen storage disease IIIb, 232400 (3), Autosomal recessive |
| **AGMO** | 99.72 % | 613738 | *No OMIM phenotypes* |
| **AGPAT2** | 99.75 % | 603100 | Lipodystrophy, congenital generalized, type 1, 608594 (3), Autosomal recessive |
| **AGPS** | 98.17 % | 603051 | Rhizomelic chondrodysplasia punctata, type 3, 600121 (3), Autosomal recessive |
| **AGR2** | 99.95 % | 606358 | Respiratory infections, recurrent, and failure to thrive with or without diarrhea, 620233 (3), Autosomal recessive |
| **AGRN** | 99.89 % | 103320 | Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120 (3), Autosomal recessive |
| **AGT** | 99.94 % | 106150 | Renal tubular dysgenesis, 267430 (3), Autosomal recessive |
| **AGTPBP1** | 99.58 % | 606830 | Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276 (3), Autosomal recessive |
| **AGTR1** | 99.96 % | 106165 | {Hypertension, essential}, 145500 (3), Multifactorial; Renal tubular dysgenesis, 267430 (3), Autosomal recessive |

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|---|---|---|---|
| **AGXT** | 99.95 % | 604285 | Hyperoxaluria, primary, type 1, 259900 (3), Autosomal recessive |
| **AHCY** | 99.93 % | 180960 | Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752 (3), Autosomal recessive |
| **AHI1** | 99.8 % | 608894 | Joubert syndrome 3, 608629 (3), Autosomal recessive |
| **AHR** | 99.81 % | 600253 | Foveal hypoplasia 3, 620958 (3), Autosomal recessive; ?Retinitis pigmentosa 85, 618345 (3), Autosomal recessive |
| **AICDA** | 99.3 % | 605257 | Immunodeficiency with hyper-IgM, type 2, 605258 (3), Autosomal recessive |
| **AIFM1** | 99.79 % | 300169 | Combined oxidative phosphorylation deficiency 6, 300816 (3), X-linked recessive; Cowchock syndrome, 310490 (3), X-linked recessive; Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 (3), X-linked recessive; Deafness, X-linked 5, 300614 (3), X-linked recessive |
| **AIMP1** | 99.91 % | 603605 | Leukodystrophy, hypomyelinating, 3, 260600 (3), Autosomal recessive |
| **AIMP2** | 99.9 % | 600859 | Leukodystrophy, hypomyelinating, 17, 618006 (3), Autosomal recessive |
| **AIPL1** | 99.64 % | 604392 | Leber congenital amaurosis 4, 604393 (3), Autosomal recessive, Autosomal dominant; Retinitis pigmentosa, juvenile, 604393 (3), Autosomal recessive, Autosomal dominant; Cone-rod dystrophy, 604393 (3), Autosomal recessive, Autosomal dominant |
| **AIRE** | 99.61 % | 607358 | Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300 (3), Autosomal recessive, Autosomal dominant |
| **AK1** | 99.93 % | 103000 | Anemia, congenital, nonspherocytic hemolytic, 3, adenylate kinase deficient, 612631 (3), Autosomal recessive |
| **AK2** | 99.57 % | 103020 | Reticular dysgenesis, 267500 (3), Autosomal recessive |
| **AKR1C2** | 90.29 % | 600450 | 46XY sex reversal 8, 614279 (3), Autosomal recessive |
| **AKR1D1** | 99.9 % | 604741 | Bile acid synthesis defect, congenital, 2, 235555 (3), Autosomal recessive |
| **ALAD** | 99.91 % | 125270 | Porphyria, acute hepatic, 612740 (3), Autosomal recessive; {Lead poisoning, susceptibility to}, 612740 (3), Autosomal recessive |
| **ALAS2** | 99.87 % | 301300 | Anemia, sideroblastic, 1, 300751 (3), X-linked recessive; Protoporphyria, erythropoietic, X-linked, 300752 (3), X-linked |
| **ALB** | 99.84 % | 103600 | ?[Dysalbuminemic hypertriiodothyroninemia], 615999 (3), Autosomal recessive, Autosomal dominant; Analbuminemia, 616000 (3), Autosomal recessive; [Dysalbuminemic hyperthyroxinemia], 615999 (3), Autosomal recessive, Autosomal dominant |
| **ALDH18A1** | 99.91 % | 138250 | Spastic paraplegia 9A, autosomal dominant, 601162 (3), Autosomal dominant; Cutis laxa, autosomal recessive, type IIIA, 219150 (3), Autosomal recessive; Spastic paraplegia 9B, autosomal recessive, 616586 (3), Autosomal recessive; Cutis laxa, autosomal dominant 3, 616603 (3), Autosomal dominant |
| **ALDH1A2** | 99.92 % | 603687 | Diaphragmatic hernia 4, with cardiovascular defects, 620025 (3), Autosomal recessive |
| **ALDH1A3** | 99.57 % | 600463 | Microphthalmia, isolated 8, 615113 (3), Autosomal recessive |
| **ALDH3A2** | 99.66 % | 609523 | Sjogren-Larsson syndrome, 270200 (3), Autosomal recessive |
| **ALDH4A1** | 98.39 % | 606811 | Hyperprolinemia, type II, 239510 (3), Autosomal recessive |
| **ALDH5A1** | 99.65 % | 610045 | Succinic semialdehyde dehydrogenase deficiency, 271980 (3), Autosomal recessive |
| **ALDH6A1** | 99.92 % | 603178 | Methylmalonate semialdehyde dehydrogenase deficiency, 614105 (3), Autosomal recessive |
| **ALDH7A1** | 99.31 % | 107323 | Epilepsy, early-onset, 4, vitamin B6-dependent, 266100 (3), Autosomal recessive |
| **ALDOA** | 99.92 % | 103850 | Glycogen storage disease XII, 611881 (3), Autosomal recessive |
| **ALDOB** | 99.95 % | 612724 | Fructose intolerance, hereditary, 229600 (3), Autosomal recessive |
| **ALG1** | 99.58 % | 605907 | Congenital disorder of glycosylation, type Ik, 608540 (3), Autosomal recessive |

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|---|---|---|---|
| **ALG11** | 98.83 % | 613666 | Congenital disorder of glycosylation, type Ip, 613661 (3), Autosomal recessive |
| **ALG12** | 99.92 % | 607144 | Congenital disorder of glycosylation, type Ig, 607143 (3), Autosomal recessive |
| **ALG13** | 99.75 % | 300776 | Developmental and epileptic encephalopathy 36, 300884 (3), X-linked |
| **ALG14** | 97.7 % | 612866 | Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies, 619031 (3), Autosomal recessive; Myopathy, epilepsy, and progressive cerebral atrophy, 619036 (3), Autosomal recessive; ?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227 (3), Autosomal recessive |
| **ALG2** | 99.98 % | 607905 | Congenital disorder of glycosylation, type Ii, 607906 (3), Autosomal recessive; Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 (3), Autosomal recessive |
| **ALG3** | 99.96 % | 608750 | Congenital disorder of glycosylation, type Id, 601110 (3), Autosomal recessive |
| **ALG6** | 95.75 % | 604566 | Congenital disorder of glycosylation, type Ic, 603147 (3), Autosomal recessive |
| **ALG8** | 98.96 % | 608103 | Congenital disorder of glycosylation, type Ih, 608104 (3), Autosomal recessive; Polycystic liver disease 3 with or without kidney cysts, 617874 (3), Autosomal dominant |
| **ALG9** | 99.52 % | 606941 | Gillessen-Kaesbach-Nishimura syndrome, 263210 (3), Autosomal recessive; Congenital disorder of glycosylation, type Il, 608776 (3), Autosomal recessive |
| **ALKBH8** | 99.89 % | 613306 | Intellectual developmental disorder, autosomal recessive 71, 618504 (3), Autosomal recessive |
| **ALMS1** | 99.82 % | 606844 | Alstrom syndrome, 203800 (3), Autosomal recessive |
| **ALOX12B** | 99.65 % | 603741 | Ichthyosis, congenital, autosomal recessive 2, 242100 (3), Autosomal recessive |
| **ALOXE3** | 99.8 % | 607206 | Ichthyosis, congenital, autosomal recessive 3, 606545 (3), Autosomal recessive |
| **ALPK3** | 99.91 % | 617608 | Cardiomyopathy, familial hypertrophic 27, 618052 (3), Autosomal recessive |
| **ALPL** | 99.53 % | 171760 | Odontohypophosphatasia, 146300 (3), Autosomal recessive, Autosomal dominant; Hypophosphatasia, infantile, 241500 (3), Autosomal recessive; Hypophosphatasia, childhood, 241510 (3), Autosomal recessive; Hypophosphatasia, adult, 146300 (3), Autosomal recessive, Autosomal dominant |
| **ALS2** | 99.73 % | 606352 | Primary lateral sclerosis, juvenile, 606353 (3), Autosomal recessive; Spastic paralysis, infantile onset ascending, 607225 (3), Autosomal recessive; Amyotrophic lateral sclerosis 2, juvenile, 205100 (3), Autosomal recessive |
| **ALX1** | 98.39 % | 601527 | Frontonasal dysplasia 3, 613456 (3), Autosomal recessive |
| **ALX3** | 98.56 % | 606014 | Frontonasal dysplasia 1, 136760 (3), Autosomal recessive |
| **ALX4** | 99.89 % | 605420 | Parietal foramina 2, 609597 (3), Autosomal dominant; {Craniosynostosis 5, susceptibility to}, 615529 (3), Autosomal dominant; Frontonasal dysplasia 2, 613451 (3), Autosomal recessive |
| **AMACR** | 99.93 % | 604489 | Alpha-methylacyl-CoA racemase deficiency, 614307 (3), Autosomal recessive; Bile acid synthesis defect, congenital, 4, 214950 (3), Autosomal recessive |
| **AMBN** | 99.93 % | 601259 | Amelogenesis imperfecta, type IF, 616270 (3), Autosomal recessive |
| **AMELX** | 99.94 % | 300391 | Amelogenesis imperfecta, type 1E, 301200 (3), X-linked dominant |
| **AMER1** | 99.95 % | 300647 | Osteopathia striata with cranial sclerosis, 300373 (3), X-linked dominant |
| **AMFR** | 99.22 % | 603243 | Spastic paraplegia 89, autosomal recessive, 620379 (3), Autosomal recessive |
| **AMH** | 99.73 % | 600957 | Persistent Mullerian duct syndrome, type I, 261550 (3), Autosomal recessive |
| **AMHR2** | 99.48 % | 600956 | Persistent Mullerian duct syndrome, type II, 261550 (3), Autosomal recessive |
| **AMMECR1** | 99.72 % | 300195 | Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990 (3), X-linked recessive |
| **AMN** | 99.7 % | 605799 | Imerslund-Grasbeck syndrome 2, 618882 (3), Autosomal recessive |
| **AMPD2** | 99 % | 102771 | Pontocerebellar hypoplasia, type 9, 615809 (3), Autosomal recessive; ?Spastic paraplegia 63, autosomal recessive, 615686 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **AMT** | 99.98 % | 238310 | Glycine encephalopathy 2, 620398 (3), Autosomal recessive |
| **ANAPC1** | 72.56 % | 608473 | Rothmund-Thomson syndrome, type 1, 618625 (3), Autosomal recessive |
| **ANAPC7** | 99.94 % | 606949 | Ferguson-Bonni neurodevelopmental syndrome, 619699 (3), Autosomal recessive |
| **ANK3** | 99.75 % | 600465 | Intellectual developmental disorder, autosomal recessive 37, 615493 (3), Autosomal recessive |
| **ANKLE2** | 99.94 % | 616062 | Microcephaly 16, primary, autosomal recessive, 616681 (3), Autosomal recessive |
| **ANKS6** | 99.91 % | 615370 | Nephronophthisis 16, 615382 (3), Autosomal recessive |
| **ANO1** | 99.81 % | 610108 | Moyamoya disease 7, 620687 (3), Autosomal recessive, Autosomal dominant; ?Intestinal dysmotility syndrome, 620045 (3), Autosomal recessive |
| **ANO10** | 99.87 % | 613726 | Spinocerebellar ataxia, autosomal recessive 10, 613728 (3), Autosomal recessive |
| **ANO5** | 99.62 % | 608662 | Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307 (3), Autosomal recessive; Miyoshi muscular dystrophy 3, 613319 (3), Autosomal recessive; Gnathodiaphyseal dysplasia, 166260 (3), Autosomal dominant |
| **ANO6** | 98.65 % | 608663 | Scott syndrome, 262890 (3), Autosomal recessive |
| **ANOS1** | 99.91 % | 300836 | Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700 (3), X-linked recessive |
| **ANTXR1** | 99.77 % | 606410 | GAPO syndrome, 230740 (3), Autosomal recessive; {?Hemangioma, capillary infantile, susceptibility to}, 602089 (3), Autosomal dominant |
| **ANTXR2** | 99.56 % | 608041 | Hyaline fibromatosis syndrome, 228600 (3), Autosomal recessive |
| **AOPEP** | 99.82 % | 619600 | Dystonia 31, 619565 (3), Autosomal recessive |
| **AP1B1** | 99.9 % | 600157 | Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150 (3), Autosomal recessive |
| **AP1G1** | 99.87 % | 603533 | Usmani-Riazuddin syndrome, autosomal recessive, 619548 (3), Autosomal recessive; Usmani-Riazuddin syndrome, autosomal dominant, 619467 (3), Autosomal dominant |
| **AP1S1** | 98.44 % | 603531 | MEDNIK syndrome, 609313 (3), Autosomal recessive |
| **AP1S2** | 99.72 % | 300629 | Pettigrew syndrome, 304340 (3), X-linked recessive |
| **AP3B1** | 99.78 % | 603401 | Hermansky-Pudlak syndrome 2, 608233 (3), Autosomal recessive |
| **AP3B2** | 99.97 % | 602166 | Developmental and epileptic encephalopathy 48, 617276 (3), Autosomal recessive |
| **AP3D1** | 99.68 % | 607246 | ?Hermansky-Pudlak syndrome 10, 617050 (3), Autosomal recessive |
| **AP4B1** | 98.34 % | 607245 | Spastic paraplegia 47, autosomal recessive, 614066 (3), Autosomal recessive |
| **AP4E1** | 99.93 % | 607244 | Stuttering, familial persistent, 1, 184450 (3), Autosomal dominant; Spastic paraplegia 51, autosomal recessive, 613744 (3), Autosomal recessive |
| **AP4M1** | 98.54 % | 602296 | Spastic paraplegia 50, autosomal recessive, 612936 (3), Autosomal recessive |
| **AP4S1** | 88.62 % | 607243 | Spastic paraplegia 52, autosomal recessive, 614067 (3), Autosomal recessive |
| **AP5Z1** | 99.89 % | 613653 | Spastic paraplegia 48, autosomal recessive, 613647 (3), Autosomal recessive |
| **APC2** | 99.7 % | 612034 | Cortical dysplasia, complex, with other brain malformations 10, 618677 (3), Autosomal recessive; Intellectual developmental disorder, autosomal recessive 74, 617169 (3), Autosomal recessive |
| **APOA1** | 99.81 % | 107680 | Hypoalphalipoproteinemia, primary, 2, 618463 (3), Autosomal recessive; Amyloidosis, hereditary systemic 3, 620657 (3), Autosomal dominant; Hypoalphalipoproteinemia, primary, 2, intermediate, 619836 (3), Autosomal dominant |
| **APOB** | 99.97 % | 107730 | Hypercholesterolemia, familial, 2, 144010 (3), Autosomal dominant; Hypobetalipoproteinemia, 615558 (3), Autosomal recessive |
| **APOC2** | 99.97 % | 608083 | Hyperlipoproteinemia, type Ib, 207750 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **APOE** | 99.81 % | 107741 | Alzheimer disease 2, 104310 (3), Autosomal dominant; Sea-blue histiocyte disease, 269600 (3), Autosomal recessive; {?Alzheimer disease, protection against, due to APOE3-Christchurch}, 607822 (3), Autosomal dominant; {Coronary artery disease, severe, susceptibility to}, 617347 (3); Lipoprotein glomerulopathy, 611771 (3); {?Macular degeneration, age-related}, 603075 (3), Autosomal dominant; Hyperlipoproteinemia, type III, 617347 (3) |
| **APRT** | 99.46 % | 102600 | Adenine phosphoribosyltransferase deficiency, 614723 (3), Autosomal recessive |
| **APTX** | 99.92 % | 606350 | Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920 (3), Autosomal recessive |
| **AQP2** | 99.95 % | 107777 | Diabetes insipidus, nephrogenic, 2, 125800 (3), Autosomal recessive, Autosomal dominant |
| **AR** | 99.96 % | 313700 | Androgen insensitivity, partial, with or without breast cancer, 312300 (3), X-linked recessive; Spinal and bulbar muscular atrophy, X-linked 1, 313200 (3), X-linked recessive; {Prostate cancer, susceptibility to}, 301120 (3), X-linked; Androgen insensitivity, 300068 (3), X-linked recessive; Hypospadias 1, X-linked, 300633 (3), X-linked recessive |
| **ARFGEF2** | 99.88 % | 605371 | Periventricular heterotopia with microcephaly, 608097 (3), Autosomal recessive |
| **ARG1** | 99.88 % | 608313 | Argininemia, 207800 (3), Autosomal recessive |
| **ARHGDIA** | 99.94 % | 601925 | Nephrotic syndrome, type 8, 615244 (3), Autosomal recessive |
| **ARHGEF2** | 99.19 % | 607560 | ?Neurodevelopmental disorder with midbrain and hindbrain malformations, 617523 (3), Autosomal recessive |
| **ARHGEF9** | 99.77 % | 300429 | Developmental and epileptic encephalopathy 8, 300607 (3), X-linked |
| **ARL13B** | 99.78 % | 608922 | Joubert syndrome 8, 612291 (3), Autosomal recessive |
| **ARL2BP** | 99.68 % | 615407 | Retinitis pigmentosa 82 with or without situs inversus, 615434 (3), Autosomal recessive |
| **ARL3** | 99.94 % | 604695 | Retinitis pigmentosa 83, 618173 (3), Autosomal dominant; Joubert syndrome 35, 618161 (3), Autosomal recessive |
| **ARL6** | 99.89 % | 608845 | Retinitis pigmentosa 55, 613575 (3), Autosomal recessive; {Bardet-Biedl syndrome 1, modifier of}, 209900 (3), Autosomal recessive, Digenic recessive; Bardet-Biedl syndrome 3, 600151 (3), Autosomal recessive |
| **ARL6IP1** | 99.29 % | 607669 | Spastic paraplegia 61, autosomal recessive, 615685 (3), Autosomal recessive |
| **ARMC9** | 99.29 % | 617612 | Joubert syndrome 30, 617622 (3), Autosomal recessive |
| **ARNT2** | 99.72 % | 606036 | ?Webb-Dattani syndrome, 615926 (3), Autosomal recessive |
| **ARPC1B** | 99.72 % | 604223 | Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia, 617718 (3), Autosomal recessive |
| **ARPC5** | 98.62 % | 604227 | Immunodeficiency 113 with autoimmunity and autoinflammation, 620565 (3), Autosomal recessive |
| **ARR3** | 99.76 % | 301770 | Myopia 26, X-linked, female-limited, 301010 (3), X-linked |
| **ARSA** | 99.83 % | 607574 | Metachromatic leukodystrophy, 250100 (3), Autosomal recessive |
| **ARSB** | 99.79 % | 611542 | Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200 (3), Autosomal recessive |
| **ARSG** | 99.72 % | 610008 | Usher syndrome, type IV, 618144 (3), Autosomal recessive |
| **ARSK** | 99.74 % | 610011 | Mucopolysaccharidosis, type X, 619698 (3), Autosomal recessive |
| **ARSL** | 99.68 % | 300180 | Chondrodysplasia punctata, X-linked recessive, 302950 (3), X-linked recessive |
| **ARV1** | 99.82 % | 611647 | Developmental and epileptic encephalopathy 38, 617020 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **ARX** | 99.44 % | 300382 | Proud syndrome, 300004 (3), X-linked; Hydranencephaly with abnormal genitalia, 300215 (3), X-linked; Partington syndrome, 309510 (3), X-linked recessive; Developmental and epileptic encephalopathy 1, 308350 (3), X-linked recessive; Lissencephaly, X-linked 2, 300215 (3), X-linked; Intellectual developmental disorder, X-linked 29, 300419 (3), X-linked recessive |
| **ASAH1** | 99.6 % | 613468 | Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 (3), Autosomal recessive; Farber lipogranulomatosis, 228000 (3), Autosomal recessive |
| **ASCC1** | 98.23 % | 614215 | Spinal muscular atrophy with congenital bone fractures 2, 616867 (3), Autosomal recessive; Barrett esophagus/esophageal adenocarcinoma, 614266 (3) |
| **ASCC3** | 99.66 % | 614217 | Intellectual developmental disorder, autosomal recessive 81, 620700 (3), Autosomal recessive |
| **ASL** | 99.6 % | 608310 | Argininosuccinic aciduria, 207900 (3), Autosomal recessive |
| **ASNS** | 99.17 % | 108370 | Asparagine synthetase deficiency, 615574 (3), Autosomal recessive |
| **ASPA** | 99.88 % | 608034 | Canavan disease, 271900 (3), Autosomal recessive |
| **ASPH** | 99.83 % | 600582 | Traboulsi syndrome, 601552 (3), Autosomal recessive |
| **ASPM** | 99.52 % | 605481 | Microcephaly 5, primary, autosomal recessive, 608716 (3), Autosomal recessive |
| **ASRGL1** | 99.48 % | 609212 | *No OMIM phenotypes* |
| **ASS1** | 99.83 % | 603470 | Citrullinemia, 215700 (3), Autosomal recessive |
| **ATAD1** | 99.42 % | 614452 | Hyperekplexia 4, 618011 (3), Autosomal recessive |
| **ATAD3A** | 98.72 % | 612316 | Harel-Yoon syndrome, 617183 (3), Autosomal recessive, Autosomal dominant; Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810 (3), Autosomal recessive |
| **ATCAY** | 99.99 % | 608179 | Ataxia, cerebellar, Cayman type, 601238 (3), Autosomal recessive |
| **ATF6** | 97.86 % | 605537 | Achromatopsia 7, 616517 (3), Autosomal recessive |
| **ATG4D** | 99.55 % | 611340 | Spermatogenic failure 101, 621269 (3), Autosomal recessive |
| **ATG7** | 99.86 % | 608760 | Spinocerebellar ataxia, autosomal recessive 31, 619422 (3), Autosomal recessive |
| **ATIC** | 99.81 % | 601731 | AICA-ribosiduria due to ATIC deficiency, 608688 (3), Autosomal recessive |
| **ATM** | 99.88 % | 607585 | Lymphoma, B-cell non-Hodgkin, somatic (3); Ataxia-telangiectasia, 208900 (3), Autosomal recessive; {Breast cancer, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant; T-cell prolymphocytic leukemia, somatic (3); Lymphoma, mantle cell, somatic (3) |
| **ATOH7** | 99.55 % | 609875 | Persistent hyperplastic primary vitreous, autosomal recessive, 221900 (3), Autosomal recessive |
| **ATP11C** | 99.66 % | 300516 | ?Hemolytic anemia, congenital, X-linked, 301015 (3), X-linked recessive |
| **ATP13A2** | 99.27 % | 610513 | Spastic paraplegia 78, autosomal recessive, 617225 (3), Autosomal recessive; Kufor-Rakeb syndrome, 606693 (3), Autosomal recessive |
| **ATP13A3** | 99.85 % | 610232 | Pulmonary hypertension, primary, 5, 265400 (3), Autosomal recessive |
| **ATP1A2** | 99.36 % | 182340 | Developmental and epileptic encephalopathy 98, 619605 (3), Autosomal dominant; Fetal akinesia, respiratory insufficiency, microcephaly, polymicrogyria, and dysmorphic facies, 619602 (3), Autosomal recessive; Alternating hemiplegia of childhood 1, 104290 (3), Autosomal dominant; Migraine, familial basilar, 602481 (3), Autosomal dominant; Migraine, familial hemiplegic, 2, 602481 (3), Autosomal dominant |
| **ATP2A1** | 98.96 % | 108730 | Brody myopathy, 601003 (3), Autosomal recessive |
| **ATP2B3** | 99.81 % | 300014 | ?Spinocerebellar ataxia, X-linked 1, 302500 (3), X-linked recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **ATP5F1A** | 99.98 % | 164360 | Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4A, 620358 (3), Autosomal dominant; ?Combined oxidative phosphorylation deficiency 22, 616045 (3), Autosomal recessive; ?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4B, encephalopathic type, 615228 (3), Autosomal recessive |
| **ATP5F1D** | 99.04 % | 603150 | Mitochondrial complex V (ATP synthase) deficiency, 618120 (3), Autosomal recessive |
| **ATP5F1E** | 100 % | 606153 | Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053 (3), Autosomal recessive |
| **ATP5MK** | 100 % | 615204 | Mitochondrial complex V (ATP synthase) deficiency, nuclear type 6, 618683 (3), Autosomal recessive |
| **ATP5PO** | 99.76 % | 600828 | Mitochondrial complex V (ATP synthase) deficiency, nuclear type 7, 620359 (3), Autosomal recessive |
| **ATP6AP1** | 99.75 % | 300197 | Immunodeficiency 47, 300972 (3), X-linked recessive |
| **ATP6AP2** | 99.55 % | 300556 | Intellectual developmental disorder, X-linked syndromic, Hedera type, 300423 (3), X-linked recessive; ?Parkinsonism with spasticity, X-linked, 300911 (3), X-linked recessive; Congenital disorder of glycosylation, type IIr, 301045 (3), X-linked recessive |
| **ATP6V0A1** | 99.56 % | 192130 | Neurodevelopmental disorder with epilepsy and brain atrophy, 619971 (3), Autosomal recessive; Developmental and epileptic encephalopathy 104, 619970 (3), Autosomal dominant |
| **ATP6V0A2** | 99.77 % | 611716 | Wrinkly skin syndrome, 278250 (3), Autosomal recessive; Cutis laxa, autosomal recessive, type IIA, 219200 (3), Autosomal recessive |
| **ATP6V0A4** | 99.67 % | 605239 | Distal renal tubular acidosis 3, with or without sensorineural hearing loss, 602722 (3), Autosomal recessive |
| **ATP6V1A** | 99.41 % | 607027 | Cutis laxa, autosomal recessive, type IID, 617403 (3), Autosomal recessive; Developmental and epileptic encephalopathy 93, 618012 (3), Autosomal dominant |
| **ATP6V1B1** | 99.59 % | 192132 | Distal renal tubular acidosis 2 with progressive sensorineural hearing loss, 267300 (3), Autosomal recessive |
| **ATP6V1E1** | 99.83 % | 108746 | Cutis laxa, autosomal recessive, type IIC, 617402 (3), Autosomal recessive |
| **ATP7A** | 99.79 % | 300011 | Occipital horn syndrome, 304150 (3), X-linked recessive; Neuronopathy, distal hereditary motor, X-linked, 300489 (3), X-linked recessive; Menkes disease, 309400 (3), X-linked recessive |
| **ATP7B** | 99.95 % | 606882 | Wilson disease, 277900 (3), Autosomal recessive |
| **ATP8A2** | 99.87 % | 605870 | Cerebellar ataxia, impaired intellectual development, and dysequilibrium syndrome 4, 615268 (3), Autosomal recessive |
| **ATP8B1** | 99.9 % | 602397 | Cholestasis, progressive familial intrahepatic 1, 211600 (3), Autosomal recessive; Cholestasis, intrahepatic, of pregnancy, 1, 147480 (3), Autosomal dominant; Cholestasis, benign recurrent intrahepatic, 243300 (3), Autosomal recessive |
| **ATP9A** | 99.85 % | 609126 | Neurodevelopmental disorder with poor growth and behavioral abnormalities, 620242 (3), Autosomal recessive |
| **ATPAF2** | 99.49 % | 608918 | ?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273 (3), Autosomal recessive |
| **ATR** | 99.75 % | 601215 | Seckel syndrome 1, 210600 (3), Autosomal recessive; ?Cutaneous telangiectasia and cancer syndrome, familial, 614564 (3), Autosomal dominant |
| **ATRIP** | 99.73 % | 606605 | *No OMIM phenotypes* |
| **ATRX** | 99.57 % | 300032 | Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 (3); Intellectual disability-hypotonic facies syndrome, X-linked, 309580 (3), X-linked recessive; Alpha-thalassemia/impaired intellectual development syndrome, 301040 (3), X-linked dominant |
| **AUH** | 99.96 % | 600529 | 3-methylglutaconic aciduria, type I, 250950 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **AVIL** | 99.84 % | 613397 | Nephrotic syndrome, type 21, 618594 (3), Autosomal recessive |
| **AVPR2** | 99.9 % | 300538 | Diabetes insipidus, nephrogenic, 1, 304800 (3), X-linked recessive; Nephrogenic syndrome of inappropriate antidiuresis, 300539 (3), X-linked recessive |
| **B3GALNT2** | 97.89 % | 610194 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 11, 615181 (3), Autosomal recessive |
| **B3GALT6** | 99.98 % | 615291 | Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 (3), Autosomal recessive; Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640 (3), Autosomal recessive; Al-Gazali syndrome, 609465 (3), Autosomal recessive |
| **B3GAT3** | 99.42 % | 606374 | Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600 (3), Autosomal recessive |
| **B3GLCT** | 99.97 % | 610308 | Peters-plus syndrome, 261540 (3), Autosomal recessive |
| **B4GALNT1** | 98.13 % | 601873 | Spastic paraplegia 26, autosomal recessive, 609195 (3), Autosomal recessive |
| **B4GALT1** | 99.4 % | 137060 | Combined low LDL and fibrinogen, 620364 (3), Autosomal recessive; Congenital disorder of glycosylation, type IId, 607091 (3), Autosomal recessive |
| **B4GALT7** | 99.6 % | 604327 | Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070 (3), Autosomal recessive |
| **B4GAT1** | 99.94 % | 605517 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287 (3), Autosomal recessive |
| **B9D1** | 99.49 % | 614144 | ?Meckel syndrome 9, 614209 (3), Autosomal recessive; Joubert syndrome 27, 617120 (3), Autosomal recessive |
| **B9D2** | 99.25 % | 611951 | ?Meckel syndrome 10, 614175 (3), Autosomal recessive; Joubert syndrome 34, 614175 (3), Autosomal recessive |
| **BAAT** | 99.94 % | 602938 | Hypercholanemia, familial 3, 619232 (3), Autosomal recessive |
| **BAG5** | 100 % | 603885 | Cardiomyopathy, dilated, 2F, 619747 (3), Autosomal recessive |
| **BANF1** | 97.05 % | 603811 | Nestor-Guillermo progeria syndrome, 614008 (3), Autosomal recessive |
| **BBIP1** | 99.91 % | 613605 | Bardet-Biedl syndrome 18, 615995 (3), Autosomal recessive |
| **BBS1** | 99.96 % | 209901 | Bardet-Biedl syndrome 1, 209900 (3), Autosomal recessive, Digenic recessive |
| **BBS10** | 99.8 % | 610148 | Bardet-Biedl syndrome 10, 615987 (3), Autosomal recessive |
| **BBS12** | 99.98 % | 610683 | Bardet-Biedl syndrome 12, 615989 (3), Autosomal recessive |
| **BBS2** | 99.4 % | 606151 | Retinitis pigmentosa 74, 616562 (3), Autosomal recessive; Bardet-Biedl syndrome 2, 615981 (3), Autosomal recessive |
| **BBS4** | 99.38 % | 600374 | Bardet-Biedl syndrome 4, 615982 (3), Autosomal recessive |
| **BBS5** | 98.81 % | 603650 | Bardet-Biedl syndrome 5, 615983 (3), Autosomal recessive |
| **BBS7** | 99.54 % | 607590 | Bardet-Biedl syndrome 7, 615984 (3), Autosomal recessive |
| **BBS9** | 99.75 % | 607968 | Bardet-Biedl syndrome 9, 615986 (3), Autosomal recessive |
| **BCAP31** | 99.64 % | 300398 | Deafness, dystonia, and cerebral hypomyelination, 300475 (3), X-linked recessive |
| **BCAS3** | 98.22 % | 607470 | Hengel-Maroofian-Schols syndrome, 619641 (3), Autosomal recessive |
| **BCAT2** | 99.64 % | 113530 | Hypervalinemia and hyperleucine-isoleucinemia, 618850 (3), Autosomal recessive |
| **BCKDHA** | 99.82 % | 608348 | Maple syrup urine disease, type Ia, 248600 (3), Autosomal recessive |
| **BCKDHB** | 99.69 % | 248611 | Maple syrup urine disease, type Ib, 620698 (3), Autosomal recessive |
| **BCKDK** | 99.8 % | 614901 | Branched-chain keto acid dehydrogenase kinase deficiency, 614923 (3), Autosomal recessive |
| **BCL10** | 98.89 % | 603517 | {Lymphoma, follicular, somatic}, 605027 (3); ?Immunodeficiency 37, 616098 (3), Autosomal recessive; {Sezary syndrome, somatic} (3); {Male germ cell tumor, somatic}, 273300 (3); Lymphoma, MALT, somatic, 137245 (3); {Mesothelioma, somatic}, 156240 (3) |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **BCOR** | 99.92 % | 300485 | Microphthalmia, syndromic 2, 300166 (3), X-linked dominant |
| **BCORL1** | 99.33 % | 300688 | Shukla-Vernon syndrome, 301029 (3), X-linked recessive |
| **BCS1L** | 99.9 % | 603647 | GRACILE syndrome, 603358 (3), Autosomal recessive; Mitochondrial complex III deficiency, nuclear type 1, 124000 (3), Autosomal recessive; Bjornstad syndrome, 262000 (3), Autosomal recessive |
| **BEST1** | 99.95 % | 607854 | Macular dystrophy, vitelliform, 2, 153700 (3), Autosomal dominant; ?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 2, 193220 (3), Autosomal dominant; Retinitis pigmentosa-50, 613194 (3); Retinitis pigmentosa, concentric, 613194 (3); Vitreoretinochoroidopathy, 193220 (3), Autosomal dominant; Bestrophinopathy, autosomal recessive, 611809 (3) |
| **BET1** | 99.64 % | 605456 | Muscular dystrophy, congenital, with rapid progression, 254100 (3), Autosomal recessive |
| **BGN** | 99.87 % | 301870 | Meester-Loeys syndrome, 300989 (3), X-linked; Spondyloepimetaphyseal dysplasia, X-linked, 300106 (3), X-linked recessive |
| **BHLHA9** | 99.36 % | 615416 | ?Camptosynpolydactyly, complex, 607539 (3), Autosomal recessive; Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432 (3), Autosomal recessive |
| **BIN1** | 99.35 % | 601248 | Centronuclear myopathy 2, 255200 (3), Autosomal recessive |
| **BLM** | 99.87 % | 604610 | Bloom syndrome, 210900 (3), Autosomal recessive |
| **BLNK** | 99.97 % | 604515 | ?Agammaglobulinemia 4, 613502 (3), Autosomal recessive |
| **BLOC1S3** | 99.88 % | 609762 | Hermansky-Pudlak syndrome 8, 614077 (3), Autosomal recessive |
| **BLOC1S5** | 99.76 % | 607289 | Hermansky-Pudlak syndrome 11, 619172 (3), Autosomal recessive |
| **BLOC1S6** | 99.66 % | 604310 | Hermansky-Pudlak syndrome 9, 614171 (3), Autosomal recessive |
| **BLVRA** | 99.74 % | 109750 | Hyperbiliverdinemia, 614156 (3), Autosomal recessive, Autosomal dominant |
| **BMP1** | 99.61 % | 112264 | Osteogenesis imperfecta, type XIII, 614856 (3), Autosomal recessive |
| **BMPER** | 99.68 % | 608699 | Diaphanospondylodysostosis, 608022 (3), Autosomal recessive |
| **BMPR1B** | 99.82 % | 603248 | Acromesomelic dysplasia 3, 609441 (3), Autosomal recessive; Brachydactyly, type A2, 112600 (3), Autosomal dominant; Brachydactyly, type A1, D, 616849 (3), Autosomal dominant |
| **BOLA3** | 97.22 % | 613183 | Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299 (3), Autosomal recessive |
| **BPGM** | 100 % | 613896 | Erythrocytosis, familial, 8, 222800 (3), Autosomal recessive |
| **BPNT2** | 99.96 % | 614010 | Chondrodysplasia with joint dislocations, GPAPP type, 614078 (3), Autosomal recessive |
| **BRAT1** | 99.92 % | 614506 | Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056 (3), Autosomal recessive; Rigidity and multifocal seizure syndrome, lethal neonatal, 614498 (3), Autosomal recessive |
| **BRCA1** | 99.73 % | 113705 | Fanconi anemia, complementation group S, 617883 (3), Autosomal recessive; {Breast-ovarian cancer, familial, 1}, 604370 (3), Autosomal dominant; {Pancreatic cancer, susceptibility to, 4}, 614320 (3) |
| **BRCA2** | 99.97 % | 600185 | Fanconi anemia, complementation group D1, 605724 (3), Autosomal recessive; {Glioblastoma 3}, 613029 (3), Autosomal recessive; {Medulloblastoma}, 155255 (3), Somatic mutation, Autosomal recessive, Autosomal dominant; {Prostate cancer}, 176807 (3), Somatic mutation, Autosomal dominant; {Breast-ovarian cancer, familial, 2}, 612555 (3), Autosomal dominant; {Breast cancer, male, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant; {Pancreatic cancer 2}, 613347 (3); Wilms tumor, 194070 (3), Somatic mutation, Autosomal dominant |
| **BRF1** | 99.82 % | 604902 | Cerebellofaciodental syndrome, 616202 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **BRIP1** | 99.53 % | 605882 | Fanconi anemia, complementation group J, 609054 (3); {Breast cancer, early-onset, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant |
| **BRWD1** | 99.77 % | 617824 | Ciliary dyskinesia, primary, 51, 620438 (3), Autosomal recessive |
| **BRWD3** | 99.53 % | 300553 | Intellectual developmental disorder, X-linked 93, 300659 (3), X-linked recessive |
| **BSCL2** | 99.73 % | 606158 | Lipodystrophy, congenital generalized, type 2, 269700 (3), Autosomal recessive; Neuronopathy, distal hereditary motor, autosomal dominant 13, 619112 (3), Autosomal dominant; Silver spastic paraplegia syndrome, 270685 (3), Autosomal dominant; Encephalopathy, progressive, with or without lipodystrophy, 615924 (3), Autosomal recessive |
| **BSND** | 99.73 % | 606412 | Sensorineural deafness with mild renal dysfunction, 602522 (3), Autosomal recessive; Bartter syndrome, type 4a, 602522 (3), Autosomal recessive |
| **BTD** | 99.93 % | 609019 | Biotinidase deficiency, 253260 (3), Autosomal recessive |
| **BTK** | 99.77 % | 300300 | Agammaglobulinemia, X-linked 1, 300755 (3), X-linked recessive; Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200 (3), X-linked recessive |
| **BUB1** | 98.53 % | 602452 | Colorectal cancer with chromosomal instability, somatic, 114500 (3); Microcephaly 30, primary, autosomal recessive, 620183 (3), Autosomal recessive |
| **BUB1B** | 99.99 % | 602860 | Colorectal cancer, somatic, 114500 (3); [Premature chromatid separation trait], 176430 (3), Autosomal dominant; Mosaic variegated aneuploidy syndrome 1, 257300 (3), Autosomal recessive |
| **BVES** | 99.69 % | 604577 | Muscular dystrophy, limb-girdle, autosomal recessive 25, 616812 (3), Autosomal recessive |
| **C12orf4** | 99.8 % | 616082 | Intellectual developmental disorder, autosomal recessive 66, 618221 (3), Autosomal recessive |
| **C12orf57** | 99.96 % | 615140 | Temtamy syndrome, 218340 (3), Autosomal recessive |
| **C19orf12** | 99.83 % | 614297 | Neurodegeneration with brain iron accumulation 4, 614298 (3), Autosomal recessive, Autosomal dominant; ?Spastic paraplegia 43, autosomal recessive, 615043 (3), Autosomal recessive |
| **C1orf127** | 99.82 % | 619700 | Heterotaxy, visceral, 14, autosomal, 621080 (3), Autosomal recessive |
| **C1QA** | 99.8 % | 120550 | C1q deficiency 1, 613652 (3), Autosomal recessive |
| **C1QB** | 98.46 % | 120570 | C1q deficiency 2, 620321 (3), Autosomal recessive |
| **C1QBP** | 99.84 % | 601269 | Combined oxidative phosphorylation deficiency 33, 617713 (3), Autosomal recessive |
| **C1QC** | 99.81 % | 120575 | C1q deficiency 3, 620322 (3), Autosomal recessive |
| **C1S** | 99.95 % | 120580 | C1s deficiency, 613783 (3); Ehlers-Danlos syndrome, periodontal type, 2, 617174 (3), Autosomal dominant |
| **C2CD3** | 99.73 % | 615944 | Orofaciodigital syndrome XIV, 615948 (3), Autosomal recessive |
| **C2orf69** | 99.66 % | 619219 | Combined oxidative phosphorylation deficiency 53, 619423 (3), Autosomal recessive |
| **C3** | 99.86 % | 120700 | C3 deficiency, 613779 (3), Autosomal recessive; {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 (3), Autosomal dominant; {Macular degeneration, age-related, 9}, 611378 (3) |
| **C3orf52** | 99.63 % | 611956 | Hypotrichosis 15, 620177 (3), Autosomal recessive |
| **C5** | 99.89 % | 120900 | C5 deficiency, 609536 (3), Autosomal recessive; [Eculizumab, poor response to], 615749 (3), Autosomal dominant |
| **C6** | 99.88 % | 217050 | C6 deficiency, 612446 (3), Autosomal recessive |
| **C7** | 99.78 % | 217070 | C7 deficiency, 610102 (3) |
| **C8A** | 99.25 % | 120950 | C8 deficiency, type I, 613790 (3), Autosomal recessive |
| **C8B** | 98.96 % | 120960 | C8 deficiency, type II, 613789 (3), Autosomal recessive |
| **CA12** | 99.78 % | 603263 | Hyperchlorhidrosis, isolated, 143860 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **CA2** | 97.98 % | 611492 | Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730 (3), Autosomal recessive |
| **CA5A** | 99.86 % | 114761 | Hyperammonemia due to carbonic anhydrase VA deficiency, 615751 (3), Autosomal recessive |
| **CA8** | 99.58 % | 114815 | Spinocerebellar ataxia, autosomal recessive 34, 613227 (3), Autosomal recessive |
| **CABP2** | 99.09 % | 607314 | Deafness, autosomal recessive 93, 614899 (3), Autosomal recessive |
| **CABP4** | 99.92 % | 608965 | Cone-rod synaptic disorder, congenital nonprogressive, 610427 (3), Autosomal recessive |
| **CACNA1B** | 99.93 % | 601012 | Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497 (3), Autosomal recessive |
| **CACNA1D** | 99.82 % | 114206 | Primary aldosteronism, seizures, and neurologic abnormalities, 615474 (3), Autosomal dominant; Sinoatrial node dysfunction and deafness, 614896 (3), Autosomal recessive |
| **CACNA1F** | 99.76 % | 300110 | Cone-rod dystrophy, X-linked, 3, 300476 (3), X-linked recessive; Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071 (3), X-linked; Aland Island eye disease, 300600 (3), X-linked |
| **CACNA1S** | 99.59 % | 114208 | {Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580 (3), Autosomal dominant; Congenital myopathy 18 due to dihydropyridine receptor defect, 620246 (3), Autosomal recessive, Autosomal dominant; Hypokalemic periodic paralysis, type 1, 170400 (3), Autosomal dominant; {Malignant hyperthermia susceptibility 5}, 601887 (3), Autosomal dominant |
| **CACNA2D1** | 97.59 % | 114204 | Developmental and epileptic encephalopathy 110, 620149 (3), Autosomal recessive |
| **CACNA2D2** | 99.6 % | 607082 | Cerebellar atrophy with seizures and variable developmental delay, 618501 (3), Autosomal recessive |
| **CACNA2D4** | 99.94 % | 608171 | Retinal cone dystrophy 4, 610478 (3), Autosomal recessive |
| **CAD** | 99.7 % | 114010 | Developmental and epileptic encephalopathy 50, 616457 (3), Autosomal recessive |
| **CALCRL** | 99.51 % | 114190 | ?Lymphatic malformation 8, 618773 (3), Autosomal recessive |
| **CAMK2A** | 99.95 % | 114078 | Intellectual developmental disorder, autosomal dominant 53, 617798 (3), Autosomal dominant; ?Intellectual developmental disorder, autosomal recessive 63, 618095 (3), Autosomal recessive |
| **CAMLG** | 98.41 % | 601118 | ?Congenital disorder of glycosylation, type IIz, 620201 (3), Autosomal recessive |
| **CAMSAP1** | 99.95 % | 613774 | Cortical dysplasia, complex, with other brain malformations 12, 620316 (3), Autosomal recessive |
| **CANT1** | 99.62 % | 613165 | Desbuquois dysplasia 1, 251450 (3), Autosomal recessive; Epiphyseal dysplasia, multiple, 7, 617719 (3), Autosomal recessive |
| **CAPN1** | 99.68 % | 114220 | Spastic paraplegia 76, autosomal recessive, 616907 (3), Autosomal recessive |
| **CAPN15** | 99.95 % | 603267 | Oculogastrointestinal neurodevelopmental syndrome, 619318 (3), Autosomal recessive |
| **CAPN3** | 99.96 % | 114240 | Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129 (3), Autosomal dominant |
| **CARD11** | 99.86 % | 607210 | B-cell expansion with NFKB and T-cell anergy, 616452 (3), Autosomal dominant; Immunodeficiency 11B with atopic dermatitis, 617638 (3), Autosomal dominant; Immunodeficiency 11A, 615206 (3), Autosomal recessive |
| **CARD9** | 99.88 % | 607212 | Immunodeficiency 103, susceptibility to fungal infection, 212050 (3), Autosomal recessive |
| **CARMIL2** | 99.83 % | 610859 | Immunodeficiency 58, 618131 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **CARS1** | 99.94 % | 123859 | Microcephaly, developmental delay, and brittle hair syndrome, 618891 (3), Autosomal recessive |
| **CARS2** | 99.95 % | 612800 | Combined oxidative phosphorylation deficiency 27, 616672 (3), Autosomal recessive |
| **CASK** | 99.29 % | 300172 | Intellectual developmental disorder, with or without nystagmus, 300422 (3), X-linked recessive; Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia, 300749 (3), X-linked; FG syndrome 4, 300422 (3), X-linked recessive |
| **CASP8** | 99.81 % | 601763 | {Breast cancer, protection against}, 114480 (3), Somatic mutation, Autosomal dominant; ?Caspase 8 lymphadenopathy syndrome, 607271 (3), Autosomal recessive; Hepatocellular carcinoma, somatic, 114550 (3); {Lung cancer, protection against}, 211980 (3), Somatic mutation, Autosomal dominant |
| **CASQ2** | 96.79 % | 114251 | Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938 (3), Autosomal recessive |
| **CASR** | 99.93 % | 601199 | Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 (3), Autosomal dominant; Hyperparathyroidism, neonatal, 239200 (3), Autosomal recessive, Autosomal dominant; Hypocalcemia, autosomal dominant, 601198 (3), Autosomal dominant; Hypocalciuric hypercalcemia, type I, 145980 (3), Autosomal dominant; {?Epilepsy idiopathic generalized, susceptibility to, 8}, 612899 (3), Autosomal dominant |
| **CAST** | 99.89 % | 114090 | Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295 (3), Autosomal recessive |
| **CAV1** | 99.66 % | 601047 | Lipodystrophy, congenital generalized, type 3, 612526 (3), Autosomal recessive; Pulmonary hypertension, primary, 3, 615343 (3), Autosomal dominant; Lipodystrophy, familial partial, type 7, 606721 (3), Autosomal dominant |
| **CAV3** | 99.99 % | 601253 | Myopathy, distal, Tateyama type, 614321 (3), Autosomal dominant; Creatine phosphokinase, elevated serum, 123320 (3), Autosomal dominant; Cardiomyopathy, familial hypertrophic, 192600 (3), Digenic dominant, Autosomal dominant; Rippling muscle disease 2, 606072 (3), Autosomal dominant; Long QT syndrome 9, 611818 (3), Autosomal dominant |
| **CAVIN1** | 99.8 % | 603198 | Lipodystrophy, congenital generalized, type 4, 613327 (3), Autosomal recessive |
| **CBLB** | 99.62 % | 604491 | Autoimmune disease, multisystem, infantile-onset, 3, 620430 (3), Autosomal recessive |
| **CBS** | 16.71 % | 613381 | Thrombosis, hyperhomocysteinemic, 236200 (3), Autosomal recessive; Homocystinuria, B6-responsive and nonresponsive types, 236200 (3), Autosomal recessive |
| **CBY1** | 99.98 % | 607757 | *No OMIM phenotypes* |
| **CC2D1A** | 99.77 % | 610055 | Intellectual developmental disorder, autosomal recessive 3, 608443 (3), Autosomal recessive |
| **CC2D2A** | 99.85 % | 612013 | COACH syndrome 2, 619111 (3), Autosomal recessive; Retinitis pigmentosa 93, 619845 (3), Autosomal recessive; Meckel syndrome 6, 612284 (3), Autosomal recessive; Joubert syndrome 9, 612285 (3), Autosomal recessive |
| **CCBE1** | 98.41 % | 612753 | Hennekam lymphangiectasia-lymphedema syndrome 1, 235510 (3), Autosomal recessive |
| **CCDC103** | 99.55 % | 614677 | Ciliary dyskinesia, primary, 17, 614679 (3), Autosomal recessive |
| **CCDC115** | 98.87 % | 613734 | Congenital disorder of glycosylation, type IIo, 616828 (3), Autosomal recessive |
| **CCDC134** | 99.99 % | 618788 | Osteogenesis imperfecta, type XXII, 619795 (3), Autosomal recessive |
| **CCDC174** | 99.98 % | 616735 | Hypotonia, infantile, with psychomotor retardation, 616816 (3), Autosomal recessive |
| **CCDC186** | 99.84 % | 619249 | *No OMIM phenotypes* |
| **CCDC22** | 99.56 % | 300859 | Ritscher-Schinzel syndrome 2, 300963 (3), X-linked recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **CCDC28B** | 99.56 % | 610162 | {Bardet-Biedl syndrome 1, modifier of}, 209900 (3), Autosomal recessive, Digenic recessive |
| **CCDC39** | 99.76 % | 613798 | Ciliary dyskinesia, primary, 14, 613807 (3), Autosomal recessive |
| **CCDC40** | 99.94 % | 613799 | Ciliary dyskinesia, primary, 15, 613808 (3), Autosomal recessive |
| **CCDC47** | 99.91 % | 618260 | Trichohepatoneurodevelopmental syndrome, 618268 (3), Autosomal recessive |
| **CCDC65** | 99.63 % | 611088 | Ciliary dyskinesia, primary, 27, 615504 (3), Autosomal recessive |
| **CCDC8** | 99.89 % | 614145 | 3-M syndrome 3, 614205 (3), Autosomal recessive |
| **CCDC88A** | 99.68 % | 609736 | PEHO syndrome-like, 617507 (3), Autosomal recessive |
| **CCDC88C** | 99.88 % | 611204 | ?Spinocerebellar ataxia 40, 616053 (3), Autosomal dominant; Hydrocephalus, congenital, 1, 236600 (3), Autosomal recessive |
| **CCN6** | 99.9 % | 603400 | Progressive pseudorheumatoid dysplasia, 208230 (3), Autosomal recessive |
| **CCNO** | 99.94 % | 607752 | Ciliary dyskinesia, primary, 29, 615872 (3), Autosomal recessive |
| **CCNQ** | 99.66 % | 300708 | STAR syndrome, 300707 (3), X-linked dominant |
| **CCT2** | 98.63 % | 605139 | *No OMIM phenotypes* |
| **CCT5** | 99.89 % | 610150 | ?Neuropathy, hereditary sensory, with spastic paraplegia, 256840 (3), Autosomal recessive |
| **CD151** | 99.93 % | 602243 | [Blood group, Raph], 179620 (3); Epidermolysis bullosa simplex 7, with nephropathy and deafness, 609057 (3), Autosomal recessive |
| **CD19** | 99.5 % | 107265 | Immunodeficiency, common variable, 3, 613493 (3), Autosomal recessive |
| **CD247** | 98.25 % | 186780 | ?Immunodeficiency 25, 610163 (3), Autosomal recessive |
| **CD27** | 99.95 % | 186711 | Lymphoproliferative syndrome 2, 615122 (3), Autosomal recessive |
| **CD320** | 99.86 % | 606475 | Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646 (3), Autosomal recessive |
| **CD36** | 99.4 % | 173510 | Platelet glycoprotein IV deficiency, 608404 (3), Autosomal recessive; {Coronary heart disease, susceptibility to, 7}, 610938 (3); {Malaria, cerebral, susceptibility to}, 611162 (3); {Malaria, cerebral, reduced risk of}, 611162 (3) |
| **CD3D** | 99.98 % | 186790 | Immunodeficiency 19, severe combined, 615617 (3), Autosomal recessive |
| **CD3E** | 99.64 % | 186830 | Immunodeficiency 18, 615615 (3), Autosomal recessive; Immunodeficiency 18, SCID variant, 615615 (3), Autosomal recessive |
| **CD3G** | 99.96 % | 186740 | Immunodeficiency 17, CD3 gamma deficient, 615607 (3), Autosomal recessive |
| **CD40** | 99.97 % | 109535 | Immunodeficiency with hyper-IgM, type 3, 606843 (3), Autosomal recessive |
| **CD40LG** | 99.81 % | 300386 | Immunodeficiency, X-linked, with hyper-IgM, 308230 (3), X-linked recessive |
| **CD55** | 95.68 % | 125240 | [Blood group Cromer], 613793 (3), Autosomal recessive; Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300 (3), Autosomal recessive |
| **CD59** | 99.85 % | 107271 | Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300 (3), Autosomal recessive |
| **CD70** | 99.89 % | 602840 | Lymphoproliferative syndrome 3, 618261 (3), Autosomal recessive |
| **CD79A** | 99.91 % | 112205 | Agammaglobulinemia 3, 613501 (3), Autosomal recessive |
| **CD79B** | 99.06 % | 147245 | Agammaglobulinemia 6, 612692 (3), Autosomal recessive |
| **CD81** | 99.85 % | 186845 | Immunodeficiency, common variable, 6, 613496 (3), Autosomal recessive |
| **CDAN1** | 99.93 % | 607465 | Dyserythropoietic anemia, congenital, type Ia, 224120 (3), Autosomal recessive |
| **CDC14A** | 97.87 % | 603504 | Deafness, autosomal recessive 32, with or without immotile sperm, 608653 (3), Autosomal recessive |
| **CDC40** | 99.61 % | 605585 | ?Pontocerebellar hypoplasia, type 15, 619302 (3), Autosomal recessive |
| **CDC45** | 98.79 % | 603465 | Meier-Gorlin syndrome 7, 617063 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **CDC6** | 99.45 % | 602627 | Meier-Gorlin syndrome 5, 613805 (3), Autosomal recessive |
| **CDH11** | 99.65 % | 600023 | Teebi hypertelorism syndrome 2, 619736 (3), Autosomal dominant; Elsahy-Waters syndrome, 211380 (3), Autosomal recessive |
| **CDH23** | 99.56 % | 605516 | Usher syndrome, type 1D, 601067 (3), Autosomal recessive, Digenic recessive; {Pituitary adenoma 5, multiple types}, 617540 (3), Autosomal dominant; Usher syndrome, type 1D/F digenic, 601067 (3), Autosomal recessive, Digenic recessive; Deafness, autosomal recessive 12, 601386 (3), Autosomal recessive |
| **CDH3** | 99.86 % | 114021 | Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 (3), Autosomal recessive; Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 (3), Autosomal recessive |
| **CDHR1** | 99.33 % | 609502 | Macular dystrophy, retinal, 613660 (3), Autosomal recessive; Cone-rod dystrophy 15, 613660 (3), Autosomal recessive; Retinitis pigmentosa 65, 613660 (3), Autosomal recessive |
| **CDIN1** | 99.8 % | 615626 | Dyserythropoietic anemia, congenital, type Ib, 615631 (3), Autosomal recessive |
| **CDK10** | 99.58 % | 603464 | Al Kaissi syndrome, 617694 (3), Autosomal recessive |
| **CDK5** | 99.86 % | 123831 | ?Lissencephaly 7 with cerebellar hypoplasia, 616342 (3), Autosomal recessive |
| **CDK5RAP2** | 99.88 % | 608201 | Microcephaly 3, primary, autosomal recessive, 604804 (3), Autosomal recessive |
| **CDK6** | 98.37 % | 603368 | ?Microcephaly 12, primary, autosomal recessive, 616080 (3), Autosomal recessive |
| **CDKL5** | 99.79 % | 300203 | Developmental and epileptic encephalopathy 2, 300672 (3), X-linked dominant |
| **CDSN** | 99.91 % | 602593 | Hypotrichosis 2, 146520 (3), Autosomal dominant; Peeling skin syndrome 1, 270300 (3), Autosomal recessive |
| **CDT1** | 99.68 % | 605525 | Meier-Gorlin syndrome 4, 613804 (3), Autosomal recessive |
| **CEACAM16** | 99.91 % | 614591 | Deafness, autosomal dominant 4B, 614614 (3), Autosomal dominant; Deafness, autosomal recessive 113, 618410 (3), Autosomal recessive |
| **CEBPE** | 99.87 % | 600749 | ?Immunodeficiency 108 with autoinflammation, 260570 (3), Autosomal recessive; Specific granule deficiency, 245480 (3), Autosomal recessive, Autosomal dominant |
| **CENPF** | 99.88 % | 600236 | Stromme syndrome, 243605 (3), Autosomal recessive |
| **CENPJ** | 99.9 % | 609279 | Microcephaly 6, primary, autosomal recessive, 608393 (3), Autosomal recessive; ?Seckel syndrome 4, 613676 (3), Autosomal recessive |
| **CENPT** | 99.5 % | 611510 | ?Short stature and microcephaly with genital anomalies, 618702 (3), Autosomal recessive |
| **CEP104** | 99.96 % | 616690 | Joubert syndrome 25, 616781 (3), Autosomal recessive; Intellectual developmental disorder, autosomal recessive 77, 619988 (3), Autosomal recessive |
| **CEP120** | 99.85 % | 613446 | Short-rib thoracic dysplasia 13 with or without polydactyly, 616300 (3), Autosomal recessive; Joubert syndrome 31, 617761 (3), Autosomal recessive |
| **CEP135** | 99.82 % | 611423 | Microcephaly 8, primary, autosomal recessive, 614673 (3), Autosomal recessive |
| **CEP152** | 99.94 % | 613529 | Microcephaly 9, primary, autosomal recessive, 614852 (3), Autosomal recessive; Seckel syndrome 5, 613823 (3), Autosomal recessive |
| **CEP164** | 99.9 % | 614848 | Nephronophthisis 15, 614845 (3), Autosomal recessive |
| **CEP19** | 99.44 % | 615586 | Morbid obesity and spermatogenic failure, 615703 (3), Autosomal recessive |
| **CEP290** | 98.33 % | 610142 | Leber congenital amaurosis 10, 611755 (3); Joubert syndrome 5, 610188 (3), Autosomal recessive; Senior-Loken syndrome 6, 610189 (3), Autosomal recessive; ?Bardet-Biedl syndrome 14, 615991 (3), Autosomal recessive; Meckel syndrome 4, 611134 (3), Autosomal recessive |
| **CEP295** | 99.93 % | 617728 | Seckel syndrome 11, 620767 (3), Autosomal recessive |
| **CEP41** | 99.97 % | 610523 | Joubert syndrome 15, 614464 (3), Autosomal recessive |
| **CEP55** | 99.77 % | 610000 | Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **CEP57** | 99.81 % | 607951 | Mosaic variegated aneuploidy syndrome 2, 614114 (3), Autosomal recessive |
| **CEP63** | 99.15 % | 614724 | ?Seckel syndrome 6, 614728 (3), Autosomal recessive |
| **CEP78** | 99.88 % | 617110 | Cone-rod dystrophy and hearing loss, 617236 (3), Autosomal recessive |
| **CERKL** | 99.81 % | 608381 | Retinitis pigmentosa 26, 608380 (3), Autosomal recessive |
| **CERS1** | 99.83 % | 606919 | Epilepsy, progressive myoclonic, 8, 616230 (3), Autosomal recessive |
| **CERS3** | 99.83 % | 615276 | Ichthyosis, congenital, autosomal recessive 9, 615023 (3), Autosomal recessive |
| **CFAP20** | 99.89 % | 617906 | *No OMIM phenotypes* |
| **CFAP300** | 99.81 % | 618058 | Ciliary dyskinesia, primary, 38, 618063 (3), Autosomal recessive |
| **CFAP410** | 99.95 % | 603191 | Retinal dystrophy with macular staphyloma, 617547 (3), Autosomal recessive; Spondylometaphyseal dysplasia, axial, 602271 (3), Autosomal recessive |
| **CFAP418** | 99.92 % | 614477 | Retinitis pigmentosa 64, 614500 (3), Autosomal recessive; Cone-rod dystrophy 16, 614500 (3), Autosomal recessive; Bardet-Biedl syndrome 21, 617406 (3), Autosomal recessive |
| **CFAP45** | 99.68 % | 605152 | Heterotaxy, visceral, 11, autosomal, with male infertility, 619608 (3), Autosomal recessive |
| **CFAP53** | 99.91 % | 614759 | Heterotaxy, visceral, 6, autosomal recessive, 614779 (3), Autosomal recessive |
| **CFAP74** | 99.51 % | 620187 | Ciliary dyskinesia, primary, 49, without situs inversus, 620197 (3), Autosomal recessive |
| **CFD** | 99.94 % | 134350 | Complement factor D deficiency, 613912 (3), Autosomal recessive |
| **CFH** | 99.24 % | 134370 | {Macular degeneration, age-related, 4}, 610698 (3), Autosomal dominant; Basal laminar drusen, 126700 (3), Autosomal dominant; Complement factor H deficiency, 609814 (3), Autosomal recessive, Autosomal dominant; {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 (3), Autosomal recessive, Autosomal dominant |
| **CFI** | 99.7 % | 217030 | {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 (3), Autosomal dominant; {Macular degeneration, age-related, 13, susceptibility to}, 615439 (3), Autosomal dominant; Complement factor I deficiency, 610984 (3), Autosomal recessive |
| **CFL2** | 99.69 % | 601443 | Nemaline myopathy 7, autosomal recessive, 610687 (3), Autosomal recessive |
| **CFP** | 99.63 % | 300383 | Properdin deficiency, X-linked, 312060 (3), X-linked recessive |
| **CFTR** | 99.89 % | 602421 | Cystic fibrosis, 219700 (3), Autosomal recessive; Sweat chloride elevation without CF (3); Congenital bilateral absence of vas deferens, 277180 (3), Autosomal recessive; {Pancreatitis, hereditary}, 167800 (3), Autosomal dominant; {Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400 (3), Autosomal dominant; {Hypertrypsinemia, neonatal} (3) |
| **CHAT** | 96.79 % | 118490 | Myasthenic syndrome, congenital, 6, presynaptic, 254210 (3), Autosomal recessive |
| **CHKA** | 99.41 % | 118491 | Neurodevelopmental disorder with microcephaly, movement abnormalities, and seizures, 620023 (3), Autosomal recessive |
| **CHKB** | 99.96 % | 612395 | Muscular dystrophy, congenital, megaconial type, 602541 (3), Autosomal recessive |
| **CHM** | 97.19 % | 300390 | Choroideremia, 303100 (3), X-linked |
| **CHMP1A** | 99.98 % | 164010 | Pontocerebellar hypoplasia, type 8, 614961 (3), Autosomal recessive |
| **CHP1** | 99.56 % | 606988 | ?Spastic ataxia 9, autosomal recessive, 618438 (3), Autosomal recessive |
| **CHRDL1** | 99.67 % | 300350 | Megalocornea 1, X-linked, 309300 (3), X-linked recessive |
| **CHRM3** | 99.96 % | 118494 | Prune belly syndrome, 100100 (3), Autosomal recessive |
| **CHRNA1** | 99.46 % | 100690 | Myasthenic syndrome, congenital, 1B, fast-channel, 608930 (3), Autosomal recessive, Autosomal dominant; Myasthenic syndrome, congenital, 1A, slow-channel, 601462 (3), Autosomal dominant; Multiple pterygium syndrome, lethal type, 253290 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **CHRNA3** | 99.67 % | 118503 | {Lung cancer susceptibility 2}, 612052 (3); Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT, 191800 (3), Autosomal recessive |
| **CHRNB1** | 99.93 % | 100710 | ?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314 (3), Autosomal recessive; Myasthenic syndrome, congenital, 2A, slow-channel, 616313 (3), Autosomal dominant |
| **CHRND** | 99.96 % | 100720 | ?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323 (3), Autosomal recessive; Multiple pterygium syndrome, lethal type, 253290 (3), Autosomal recessive; Myasthenic syndrome, congenital, 3B, fast-channel, 616322 (3), Autosomal recessive; ?Myasthenic syndrome, congenital, 3A, slow-channel, 616321 (3), Autosomal dominant |
| **CHRNE** | 99.92 % | 100725 | Myasthenic syndrome, congenital, 4A, slow-channel, 605809 (3), Autosomal recessive, Autosomal dominant; Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931 (3), Autosomal recessive; Myasthenic syndrome, congenital, 4B, fast-channel, 616324 (3), Autosomal recessive |
| **CHRNG** | 99.93 % | 100730 | Multiple pterygium syndrome, lethal type, 253290 (3), Autosomal recessive; Escobar syndrome, 265000 (3), Autosomal recessive |
| **CHST14** | 99.87 % | 608429 | Ehlers-Danlos syndrome, musculocontractural type 1, 601776 (3), Autosomal recessive |
| **CHST3** | 99.92 % | 603799 | Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095 (3), Autosomal recessive |
| **CHST6** | 99.94 % | 605294 | Macular corneal dystrophy, 217800 (3), Autosomal recessive |
| **CHSY1** | 99.99 % | 608183 | Temtamy preaxial brachydactyly syndrome, 605282 (3), Autosomal recessive |
| **CHUK** | 99.58 % | 600664 | ?Popliteal pterygium syndrome, Bartsocas-Papas type 2, 619339 (3), Autosomal recessive; ?Cocoon syndrome, 613630 (3), Autosomal recessive |
| **CIB1** | 99.85 % | 602293 | {Epidermodysplasia verruciformis, susceptibility to, 3}, 618267 (3), Autosomal recessive |
| **CIB2** | 99.93 % | 605564 | Deafness, autosomal recessive 48, 609439 (3), Autosomal recessive |
| **CIITA** | 99.81 % | 600005 | {Rheumatoid arthritis, susceptibility to}, 180300 (3); MHC class II deficiency 1, 209920 (3), Autosomal recessive |
| **CILK1** | 99.45 % | 612325 | Cranioectodermal dysplasia 6, 621337 (3), Autosomal recessive; {Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924 (3), Autosomal dominant; Endocrine-cerebroosteodysplasia, 612651 (3), Autosomal recessive |
| **CISD2** | 96.99 % | 611507 | Wolfram syndrome 2, 604928 (3), Autosomal recessive |
| **CIT** | 99.88 % | 605629 | Microcephaly 17, primary, autosomal recessive, 617090 (3), Autosomal recessive |
| **CKAP2L** | 99.44 % | 616174 | Filippi syndrome, 272440 (3), Autosomal recessive |
| **CLCC1** | 97.65 % | 617539 | Retinitis pigmentosa 32, 609913 (3), Autosomal recessive |
| **CLCF1** | 99.86 % | 607672 | Cold-induced sweating syndrome 2, 610313 (3), Autosomal recessive |
| **CLCN1** | 99.93 % | 118425 | Myotonia congenita, recessive, 255700 (3), Autosomal recessive; Myotonia congenita, dominant, 160800 (3), Autosomal dominant; Myotonia levior, 160800 (3), Autosomal dominant |
| **CLCN2** | 99.85 % | 600570 | Leukoencephalopathy with ataxia, 615651 (3), Autosomal recessive; Hyperaldosteronism, familial, type II, 605635 (3), Autosomal dominant; {Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628 (3), Autosomal dominant; {Epilepsy, juvenile absence, susceptibility to, 2}, 607628 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628 (3), Autosomal dominant |
| **CLCN3** | 99.87 % | 600580 | Neurodevelopmental disorder with seizures and brain abnormalities, 619517 (3), Autosomal recessive; Neurodevelopmental disorder with hypotonia and brain abnormalities, 619512 (3), Autosomal dominant |
| **CLCN4** | 99.91 % | 302910 | Raynaud-Claes syndrome, 300114 (3), X-linked dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **CLCN5** | 99.79 % | 300008 | Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990 (3), X-linked recessive; Hypophosphatemic rickets, 300554 (3), X-linked recessive; Dent disease 1, 300009 (3), X-linked recessive; Nephrolithiasis, type I, 310468 (3), X-linked recessive |
| **CLCN7** | 99.83 % | 602727 | Hypopigmentation, organomegaly, and delayed myelination and development, 618541 (3), Autosomal dominant; Osteopetrosis, autosomal recessive 4, 611490 (3), Autosomal recessive; Osteopetrosis, autosomal dominant 2, 166600 (3), Autosomal dominant |
| **CLCNKB** | 99.59 % | 602023 | Bartter syndrome, type 3, 607364 (3), Autosomal recessive; Bartter syndrome, type 4b, digenic, 613090 (3), Digenic recessive |
| **CLDN1** | 99.87 % | 603718 | Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626 (3), Autosomal recessive |
| **CLDN10** | 99.86 % | 617579 | HELIX syndrome, 617671 (3), Autosomal recessive |
| **CLDN14** | 99.99 % | 605608 | Deafness, autosomal recessive 29, 614035 (3), Autosomal recessive |
| **CLDN16** | 99.92 % | 603959 | Hypomagnesemia 3, renal, 248250 (3), Autosomal recessive |
| **CLDN19** | 98.74 % | 610036 | Hypomagnesemia 5, renal, with ocular involvement, 248190 (3), Autosomal recessive |
| **CLDN9** | 99.99 % | 615799 | Deafness, autosomal recessive 116, 619093 (3), Autosomal recessive |
| **CLIC5** | 99.89 % | 607293 | ?Deafness, autosomal recessive 103, 616042 (3), Autosomal recessive |
| **CLMP** | 99.81 % | 611693 | Congenital short bowel syndrome, 615237 (3), Autosomal recessive |
| **CLN3** | 99.6 % | 607042 | Ceroid lipofuscinosis, neuronal, 3, 204200 (3), Autosomal recessive |
| **CLN5** | 99.94 % | 608102 | Ceroid lipofuscinosis, neuronal, 5, 256731 (3), Autosomal recessive |
| **CLN6** | 99.89 % | 606725 | Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 6A, 601780 (3), Autosomal recessive |
| **CLN8** | 100 % | 607837 | Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 8, 600143 (3), Autosomal recessive |
| **CLP1** | 99.97 % | 608757 | Pontocerebellar hypoplasia, type 10, 615803 (3), Autosomal recessive |
| **CLPB** | 99.82 % | 616254 | Neutropenia, severe congenital, 9, autosomal dominant, 619813 (3), Autosomal dominant; 3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271 (3), Autosomal recessive; 3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835 (3), Autosomal dominant |
| **CLPP** | 99.86 % | 601119 | Perrault syndrome 3, 614129 (3), Autosomal recessive |
| **CLRN1** | 99.97 % | 606397 | Usher syndrome, type 3A, 276902 (3), Autosomal recessive; Retinitis pigmentosa 61, 614180 (3) |
| **CLRN2** | 99.95 % | 618988 | Deafness, autosomal recessive 117, 619174 (3), Autosomal recessive |
| **CNGA1** | 99.56 % | 123825 | Retinitis pigmentosa 49, 613756 (3), Autosomal recessive |
| **CNGA3** | 99.46 % | 600053 | Achromatopsia 2, 216900 (3), Autosomal recessive |
| **CNGB1** | 99.76 % | 600724 | Retinitis pigmentosa 45, 613767 (3), Autosomal recessive |
| **CNGB3** | 99.92 % | 605080 | Achromatopsia 3, 262300 (3), Autosomal recessive |
| **CNKSR2** | 99.5 % | 300724 | Intellectual developmental disorder, X-linked syndromic, Houge type, 301008 (3), X-linked |
| **CNNM2** | 99.8 % | 607803 | Hypomagnesemia 6, renal, 613882 (3), Autosomal dominant; Hypomagnesemia, seizures, and impaired intellectual development 1, 616418 (3), Autosomal recessive, Autosomal dominant |
| **CNNM4** | 99.81 % | 607805 | Jalili syndrome, 217080 (3), Autosomal recessive |
| **CNP** | 99.78 % | 123830 | ?Leukodystrophy, hypomyelinating, 20, 619071 (3), Autosomal recessive |
| **CNPY3** | 99.53 % | 610774 | Developmental and epileptic encephalopathy 60, 617929 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **CNTN1** | 98.8 % | 600016 | Congenital myopathy 12, 612540 (3), Autosomal recessive |
| **CNTN2** | 99.61 % | 190197 | Epilepsy, early-onset, 5, with or without developmental delay, 615400 (3), Autosomal recessive |
| **CNTNAP1** | 99.64 % | 602346 | Lethal congenital contracture syndrome 7, 616286 (3), Autosomal recessive; Hypomyelinating neuropathy, congenital, 3, 618186 (3), Autosomal recessive |
| **CNTNAP2** | 99.93 % | 604569 | Pitt-Hopkins like syndrome 1, 610042 (3), Autosomal recessive; {Autism susceptibility 15}, 612100 (3) |
| **COA6** | 99.84 % | 614772 | Mitochondrial complex IV deficiency, nuclear type 13, 616501 (3), Autosomal recessive |
| **COA7** | 98.41 % | 615623 | Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387 (3), Autosomal recessive |
| **COA8** | 99.95 % | 616003 | Mitochondrial complex IV deficiency, nuclear type 17, 619061 (3), Autosomal recessive |
| **COASY** | 99.73 % | 609855 | Pontocerebellar hypoplasia, type 12, 618266 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 6, 615643 (3), Autosomal recessive |
| **COCH** | 99.88 % | 603196 | Deafness, autosomal dominant 9, 601369 (3), Autosomal dominant; ?Deafness, autosomal recessive 110, 618094 (3), Autosomal recessive |
| **COG1** | 99.95 % | 606973 | Congenital disorder of glycosylation, type IIg, 611209 (3), Autosomal recessive |
| **COG4** | 99.68 % | 606976 | Congenital disorder of glycosylation, type IIj, 613489 (3), Autosomal recessive; Saul-Wilson syndrome, 618150 (3), Autosomal dominant |
| **COG5** | 99.85 % | 606821 | Congenital disorder of glycosylation, type IIi, 613612 (3), Autosomal recessive |
| **COG6** | 99.89 % | 606977 | Shaheen syndrome, 615328 (3), Autosomal recessive; Congenital disorder of glycosylation, type IIl, 614576 (3), Autosomal recessive |
| **COG7** | 98.84 % | 606978 | Congenital disorder of glycosylation, type IIe, 608779 (3), Autosomal recessive |
| **COG8** | 99.86 % | 606979 | Congenital disorder of glycosylation, type IIh, 611182 (3) |
| **COL11A1** | 97.48 % | 120280 | Fibrochondrogenesis 1, 228520 (3), Autosomal recessive; Stickler syndrome, type II, 604841 (3), Autosomal dominant; Marshall syndrome, 154780 (3), Autosomal dominant; Deafness, autosomal dominant 37, 618533 (3), Autosomal dominant; {Lumbar disc herniation, susceptibility to}, 603932 (3) |
| **COL11A2** | 99.65 % | 120290 | Deafness, autosomal dominant 13, 601868 (3), Autosomal dominant; Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150 (3), Autosomal recessive; Fibrochondrogenesis 2, 614524 (3), Autosomal recessive, Autosomal dominant; Deafness, autosomal recessive 53, 609706 (3), Autosomal recessive; Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840 (3), Autosomal dominant |
| **COL12A1** | 99.72 % | 120320 | Bethlem myopathy 2, 616471 (3), Autosomal dominant; ?Ullrich congenital muscular dystrophy 2, 616470 (3), Autosomal recessive |
| **COL17A1** | 99.88 % | 113811 | Epithelial recurrent erosion dystrophy, 122400 (3), Autosomal dominant; Epidermolysis bullosa, junctional 4, intermediate, 619787 (3), Autosomal recessive |
| **COL18A1** | 99.79 % | 120328 | Knobloch syndrome, type 1, 267750 (3), Autosomal recessive; Glaucoma, primary closed-angle, 618880 (3), Autosomal dominant |
| **COL1A2** | 98.99 % | 120160 | Osteogenesis imperfecta, type III, 259420 (3), Autosomal dominant; {Osteoporosis, postmenopausal}, 166710 (3), Autosomal dominant; Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821 (3), Autosomal dominant; Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2, 619120 (3), Autosomal dominant; Ehlers-Danlos syndrome, cardiac valvular type, 225320 (3), Autosomal recessive; Osteogenesis imperfecta, type IV, 166220 (3), Autosomal dominant; Osteogenesis imperfecta, type II, 166210 (3), Autosomal dominant |
| **COL25A1** | 99.7 % | 610004 | Fibrosis of extraocular muscles, congenital, 5, 616219 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **COL27A1** | 99.6 % | 608461 | Steel syndrome, 615155 (3), Autosomal recessive |
| **COL2A1** | 99.31 % | 120140 | ?Vitreoretinopathy with phalangeal epiphyseal dysplasia, 619248 (3), Autosomal dominant; Czech dysplasia, 609162 (3), Autosomal dominant; Achondrogenesis, type II or hypochondrogenesis, 200610 (3), Autosomal dominant; Spondyloperipheral dysplasia, 271700 (3), Autosomal dominant; SMED Strudwick type, 184250 (3), Autosomal dominant; ?Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 (3), Autosomal dominant; Spondylometaphyseal dysplasia, Algerian type, 184253 (3), Autosomal dominant; SED congenita, 183900 (3), Autosomal dominant; Kniest dysplasia, 156550 (3), Autosomal dominant; Stickler syndrome, type I, nonsyndromic ocular, 609508 (3), Autosomal dominant; Osteoarthritis with mild chondrodysplasia, 604864 (3), Autosomal dominant; Stickler syndrome, type I, 108300 (3), Autosomal dominant; Platyspondylic skeletal dysplasia, Torrance type, 151210 (3), Autosomal dominant; Spondyloepiphyseal dysplasia, Stanescu type, 616583 (3), Autosomal dominant; Avascular necrosis of the femoral head, 608805 (3), Autosomal dominant; Legg-Calve-Perthes disease, 150600 (3), Autosomal dominant |
| **COL3A1** | 99.79 % | 120180 | Ehlers-Danlos syndrome, vascular type, 130050 (3), Autosomal dominant; Polymicrogyria with or without vascular-type EDS, 618343 (3), Autosomal recessive |
| **COL4A3** | 99.92 % | 120070 | Alport syndrome 3A, autosomal dominant, 104200 (3), Autosomal dominant; Hematuria, benign familial, 2, 620320 (3), Autosomal dominant; Alport syndrome 3B, autosomal recessive, 620536 (3), Autosomal recessive |
| **COL4A4** | 99.72 % | 120131 | Hematuria, familial benign, 1, 141200 (3), Autosomal dominant; Alport syndrome 2, autosomal recessive, 203780 (3), Autosomal recessive |
| **COL4A5** | 99.66 % | 303630 | Alport syndrome 1, X-linked, 301050 (3), X-linked dominant |
| **COL4A6** | 99.83 % | 303631 | ?Deafness, X-linked 6, 300914 (3), X-linked recessive |
| **COL6A1** | 99.88 % | 120220 | Ullrich congenital muscular dystrophy 1A, 254090 (3), Autosomal recessive, Autosomal dominant; Bethlem myopathy 1A, 158810 (3), Autosomal dominant |
| **COL6A2** | 99.93 % | 120240 | ?Myosclerosis, congenital, 255600 (3), Autosomal recessive; Ullrich congenital muscular dystrophy 1B, 620727 (3), Autosomal recessive, Autosomal dominant; Bethlem myopathy 1B, 620725 (3), Autosomal recessive, Autosomal dominant |
| **COL6A3** | 99.97 % | 120250 | Bethlem myopathy 1C, 620726 (3), Autosomal recessive, Autosomal dominant; Ullrich congenital muscular dystrophy 1C, 620728 (3), Autosomal recessive, Autosomal dominant; Dystonia 27, 616411 (3), Autosomal recessive |
| **COL7A1** | 99.82 % | 120120 | Nail disorder, nonsyndromic congenital, 8, 607523 (3), Autosomal dominant; Epidermolysis bullosa dystrophica, Bart type, 132000 (3), Autosomal dominant; Epidermolysis bullosa dystrophica inversa, 226600 (3), Autosomal recessive; Epidermolysis bullosa dystrophica, autosomal recessive, 226600 (3), Autosomal recessive; Epidermolysis bullosa, pretibial, 131850 (3), Autosomal recessive, Autosomal dominant; Epidermolysis bullosa dystrophica, autosomal dominant, 131750 (3), Autosomal dominant; Transient bullous of the newborn, 131705 (3), Autosomal recessive, Autosomal dominant; Epidermolysis bullosa pruriginosa, 604129 (3), Autosomal recessive, Autosomal dominant; Epidermolysis bullosa dystrophica, localisata variant, 226600 (3), Autosomal recessive |
| **COL9A1** | 99.84 % | 120210 | Stickler syndrome, type IV, 614134 (3), Autosomal recessive; ?Epiphyseal dysplasia, multiple, 6, 614135 (3), Autosomal dominant |
| **COL9A2** | 98.61 % | 120260 | Epiphyseal dysplasia, multiple, 2, 600204 (3), Autosomal dominant; ?Stickler syndrome, type V, 614284 (3), Autosomal recessive |
| **COL9A3** | 99.7 % | 120270 | {Intervertebral disc disease, susceptibility to}, 603932 (3); Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 (3), Autosomal dominant; Stickler syndrome, type VI, 620022 (3), Autosomal recessive |
| **COLEC10** | 99.96 % | 607620 | 3MC syndrome 3, 248340 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **COLEC11** | 99.85 % | 612502 | 3MC syndrome 2, 265050 (3), Autosomal recessive |
| **COLGALT1** | 99.43 % | 617531 | Brain small vessel disease 3, 618360 (3), Autosomal recessive |
| **COLQ** | 99.9 % | 603033 | Myasthenic syndrome, congenital, 5, 603034 (3), Autosomal recessive |
| **COPB2** | 99.58 % | 606990 | Osteoporosis, childhood- or juvenile-onset, with developmental delay, 619884 (3), Autosomal dominant; ?Microcephaly 19, primary, autosomal recessive, 617800 (3), Autosomal recessive |
| **COQ2** | 99.65 % | 609825 | {Multiple system atrophy, susceptibility to}, 146500 (3), Autosomal recessive, Autosomal dominant; Coenzyme Q10 deficiency, primary, 1, 607426 (3), Autosomal recessive |
| **COQ4** | 99.77 % | 612898 | Coenzyme Q10 deficiency, primary, 7, 616276 (3), Autosomal recessive; Spastic ataxia 10, autosomal recessive, 620666 (3), Autosomal recessive |
| **COQ5** | 99.53 % | 616359 | ?Coenzyme Q10 deficiency, primary, 9, 619028 (3), Autosomal recessive |
| **COQ6** | 99.35 % | 614647 | Coenzyme Q10 deficiency, primary, 6, 614650 (3), Autosomal recessive |
| **COQ7** | 99.89 % | 601683 | Coenzyme Q10 deficiency, primary, 8, 616733 (3), Autosomal recessive; Neuronopathy, distal hereditary motor, autosomal recessive 9, 620402 (3), Autosomal recessive |
| **COQ8A** | 99.89 % | 606980 | Coenzyme Q10 deficiency, primary, 4, 612016 (3), Autosomal recessive |
| **COQ8B** | 99.09 % | 615567 | Nephrotic syndrome, type 9, 615573 (3), Autosomal recessive |
| **COQ9** | 98.19 % | 612837 | Coenzyme Q10 deficiency, primary, 5, 614654 (3), Autosomal recessive |
| **CORO1A** | 91.68 % | 605000 | Immunodeficiency 8, 615401 (3), Autosomal recessive |
| **COX10** | 99.87 % | 602125 | Mitochondrial complex IV deficiency, nuclear type 3, 619046 (3), Autosomal recessive |
| **COX11** | 99.59 % | 603648 | Mitochondrial complex IV deficiency, nuclear type 23, 620275 (3), Autosomal recessive |
| **COX14** | 98.98 % | 614478 | ?Mitochondrial complex IV deficiency, nuclear type 10, 619053 (3), Autosomal recessive |
| **COX15** | 99.92 % | 603646 | Mitochondrial complex IV deficiency, nuclear type 6, 615119 (3), Autosomal recessive |
| **COX16** | 99.96 % | 618064 | Mitochondrial complex IV deficiency, nuclear type 22, 619355 (3), Autosomal recessive |
| **COX20** | 99.83 % | 614698 | Mitochondrial complex IV deficiency, nuclear type 11, 619054 (3), Autosomal recessive |
| **COX4I1** | 99.99 % | 123864 | Mitochondrial complex IV deficiency, nuclear type 16, 619060 (3), Autosomal recessive |
| **COX5A** | 95.85 % | 603773 | Mitochondrial complex IV deficiency, nuclear type 20, 619064 (3), Autosomal recessive |
| **COX6A1** | 99.61 % | 602072 | Charcot-Marie-Tooth disease, recessive intermediate D, 616039 (3), Autosomal recessive |
| **COX6A2** | 97.72 % | 602009 | Mitochondrial complex IV deficiency, nuclear type 18, 619062 (3), Autosomal recessive |
| **COX6B1** | 99.98 % | 124089 | Mitochondrial complex IV deficiency, nuclear type 7, 619051 (3), Autosomal recessive |
| **COX7B** | 99.97 % | 300885 | Linear skin defects with multiple congenital anomalies 2, 300887 (3), X-linked dominant |
| **CP** | 99.82 % | 117700 | Aceruloplasminemia, 604290 (3), Autosomal recessive |
| **CPA6** | 99.9 % | 609562 | Febrile seizures, familial, 11, 614418 (3), Autosomal recessive; Epilepsy, familial temporal lobe, 5, 614417 (3), Autosomal recessive, Autosomal dominant |
| **CPAMD8** | 99.58 % | 608841 | Anterior segment dysgenesis 8, 617319 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **CPE** | 99.81 % | 114855 | BDV syndrome, 619326 (3), Autosomal recessive |
| **CPLANE1** | 99.74 % | 614571 | Orofaciodigital syndrome VI, 277170 (3), Autosomal recessive; Joubert syndrome 17, 614615 (3), Autosomal recessive |
| **CPLX1** | 99.99 % | 605032 | Developmental and epileptic encephalopathy 63, 617976 (3), Autosomal recessive |
| **CPN1** | 99.77 % | 603103 | Carboxypeptidase N deficiency, 212070 (3), Autosomal recessive |
| **CPOX** | 99.85 % | 612732 | Coproporphyria, 121300 (3), Autosomal recessive, Autosomal dominant; Harderoporphyria, 618892 (3), Autosomal recessive |
| **CPS1** | 99.76 % | 608307 | Carbamoylphosphate synthetase I deficiency, 237300 (3), Autosomal recessive |
| **CPSF3** | 99.62 % | 606029 | Neurodevelopmental disorder with microcephaly, hypotonia, nystagmus, and seizures, 619876 (3), Autosomal recessive |
| **CPT1A** | 99.55 % | 600528 | CPT deficiency, hepatic, type IA, 255120 (3), Autosomal recessive |
| **CPT2** | 99.22 % | 600650 | {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212 (3), Autosomal recessive, Autosomal dominant; CPT II deficiency, infantile, 600649 (3), Autosomal recessive; CPT II deficiency, lethal neonatal, 608836 (3), Autosomal recessive; CPT II deficiency, myopathic, stress-induced, 255110 (3), Autosomal recessive, Autosomal dominant |
| **CR2** | 99.76 % | 120650 | {Systemic lupus erythematosus, susceptibility to, 9}, 610927 (3), Autosomal dominant; ?Immunodeficiency, common variable, 7, 614699 (3), Autosomal recessive |
| **CRADD** | 99.62 % | 603454 | Intellectual developmental disorder, autosomal recessive 34, with variant lissencephaly, 614499 (3), Autosomal recessive |
| **CRAT** | 99.92 % | 600184 | ?Neurodegeneration with brain iron accumulation 8, 617917 (3), Autosomal recessive |
| **CRB1** | 99.78 % | 604210 | Leber congenital amaurosis 8, 613835 (3), Autosomal recessive; Retinitis pigmentosa-12, 600105 (3), Autosomal recessive; Pigmented paravenous chorioretinal atrophy, 172870 (3), Autosomal dominant |
| **CRB2** | 99.65 % | 609720 | Focal segmental glomerulosclerosis 9, 616220 (3), Autosomal recessive; Ventriculomegaly with cystic kidney disease, 219730 (3), Autosomal recessive |
| **CRBN** | 99.9 % | 609262 | Intellectual developmental disorder, autosomal recessive 2, 607417 (3), Autosomal recessive |
| **CREB3L1** | 99.3 % | 616215 | Osteogenesis imperfecta, type XVI, 616229 (3), Autosomal recessive |
| **CRIPT** | 99.97 % | 604594 | Rothmund-Thomson syndrome, type 3, 615789 (3), Autosomal recessive |
| **CRLF1** | 99.76 % | 604237 | Cold-induced sweating syndrome 1, 272430 (3), Autosomal recessive |
| **CRPPA** | 99.83 % | 614631 | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 (3), Autosomal recessive |
| **CRTAP** | 99.84 % | 605497 | Osteogenesis imperfecta, type VII, 610682 (3), Autosomal recessive |
| **CRYAB** | 99.85 % | 123590 | Myopathy, myofibrillar, 2B, infantile-onset, 613869 (3), Autosomal recessive; Myopathy, myofibrillar, 2A, adult-onset, 608810 (3), Autosomal dominant; Cataract 16, multiple types, 613763 (3), Autosomal recessive, Autosomal dominant; Cardiomyopathy, dilated, 1II, 615184 (3), Autosomal dominant |
| **CSF1R** | 99.71 % | 164770 | Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 (3), Autosomal recessive; Leukoencephalopathy, diffuse hereditary, with spheroids 1, 221820 (3), Autosomal dominant |
| **CSF2RB** | 99.96 % | 138981 | Surfactant metabolism dysfunction, pulmonary, 5, 614370 (3), Autosomal recessive |
| **CSF3R** | 99.23 % | 138971 | Neutropenia, severe congenital, 7, autosomal recessive, 617014 (3), Autosomal recessive; ?Neutrophilia, hereditary, 162830 (3), Autosomal dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **CSGALNACT1** | 99.95 % | 616615 | Skeletal dysplasia, mild, with joint laxity and advanced bone age, 618870 (3), Autosomal recessive |
| **CSPP1** | 99.85 % | 611654 | Joubert syndrome 21, 615636 (3), Autosomal recessive |
| **CSTA** | 99.8 % | 184600 | Peeling skin syndrome 4, 607936 (3), Autosomal recessive |
| **CSTB** | 99.88 % | 601145 | Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800 (3), Autosomal recessive |
| **CTC1** | 99.9 % | 613129 | Cerebroretinal microangiopathy with calcifications and cysts, 612199 (3), Autosomal recessive |
| **CTDP1** | 99.85 % | 604927 | Congenital cataracts, facial dysmorphism, and neuropathy, 604168 (3), Autosomal recessive |
| **CTNNA2** | 99.52 % | 114025 | Cortical dysplasia, complex, with other brain malformations 9, 618174 (3), Autosomal recessive |
| **CTNS** | 99.93 % | 606272 | Cystinosis, nephropathic, 219800 (3), Autosomal recessive; Cystinosis, ocular nonnephropathic, 219750 (3), Autosomal recessive; Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 (3), Autosomal recessive; Cystinosis, atypical nephropathic, 219800 (3), Autosomal recessive |
| **CTPS1** | 97.51 % | 123860 | Immunodeficiency 24, 615897 (3), Autosomal recessive |
| **CTSA** | 99.77 % | 613111 | Galactosialidosis, 256540 (3), Autosomal recessive; Brain small vessel disease 6 with leukoencephalopathy, 621394 (3), Autosomal dominant |
| **CTSC** | 99.79 % | 602365 | Periodontitis 1, juvenile, 170650 (3), Autosomal recessive; Haim-Munk syndrome, 245010 (3), Autosomal recessive; Papillon-Lefevre syndrome, 245000 (3), Autosomal recessive |
| **CTSD** | 99.84 % | 116840 | Ceroid lipofuscinosis, neuronal, 10, 610127 (3), Autosomal recessive |
| **CTSF** | 99.65 % | 603539 | Ceroid lipofuscinosis, neuronal, 13 (Kufs type), 615362 (3), Autosomal recessive |
| **CTSK** | 99.31 % | 601105 | Pycnodysostosis, 265800 (3), Autosomal recessive |
| **CTU2** | 99.82 % | 617057 | Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142 (3), Autosomal recessive |
| **CUBN** | 99.88 % | 602997 | [Proteinuria, chronic benign], 618884 (3), Autosomal recessive; Imerslund-Grasbeck syndrome 1, 261100 (3), Autosomal recessive |
| **CUL4B** | 99.71 % | 300304 | Intellectual developmental disorder, X-linked syndromic, Cabezas type, 300354 (3), X-linked recessive |
| **CUL7** | 99.89 % | 609577 | 3-M syndrome 1, 273750 (3), Autosomal recessive |
| **CWC27** | 99.62 % | 617170 | Retinitis pigmentosa with or without skeletal anomalies, 250410 (3), Autosomal recessive |
| **CWF19L1** | 99.07 % | 616120 | Spinocerebellar ataxia, autosomal recessive 17, 616127 (3), Autosomal recessive |
| **CXCR2** | 100 % | 146928 | ?WHIM syndrome 2, 619407 (3), Autosomal recessive |
| **CYB561** | 99.78 % | 600019 | Orthostatic hypotension 2, 618182 (3), Autosomal recessive |
| **CYB5A** | 99.99 % | 613218 | Methemoglobinemia and ambiguous genitalia, 250790 (3), Autosomal recessive |
| **CYB5R3** | 99.25 % | 613213 | Methemoglobinemia, type I, 250800 (3), Autosomal recessive; Methemoglobinemia, type II, 250800 (3), Autosomal recessive |
| **CYBA** | 99.5 % | 608508 | Chronic granulomatous disease 4, autosomal recessive, 233690 (3), Autosomal recessive |
| **CYBB** | 99.67 % | 300481 | Immunodeficiency 34, mycobacteriosis, X-linked, 300645 (3), X-linked recessive; Chronic granulomatous disease, X-linked, 306400 (3), X-linked recessive |
| **CYBC1** | 99.91 % | 618334 | Chronic granulomatous disease 5, autosomal recessive, 618935 (3), Autosomal recessive |
| **CYC1** | 99.61 % | 123980 | Mitochondrial complex III deficiency, nuclear type 6, 615453 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **CYP11A1** | 99.76 % | 118485 | Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743 (3) |
| **CYP11B1** | 99.96 % | 610613 | Aldosteronism, glucocorticoid-remediable, 103900 (3), Autosomal dominant; Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 (3), Autosomal recessive |
| **CYP11B2** | 99.91 % | 124080 | Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 (3), Autosomal recessive; Aldosterone to renin ratio raised (3); {Low renin hypertension, susceptibility to} (3); Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 (3), Autosomal recessive |
| **CYP17A1** | 99.94 % | 609300 | 17,20-lyase deficiency, isolated, 202110 (3), Autosomal recessive; 17-alpha-hydroxylase/17,20-lyase deficiency, 202110 (3), Autosomal recessive |
| **CYP19A1** | 99.89 % | 107910 | Aromatase deficiency, 613546 (3) |
| **CYP1B1** | 99.86 % | 601771 | Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 (3), Autosomal recessive; Anterior segment dysgenesis 6, multiple subtypes, 617315 (3), Autosomal recessive |
| **CYP21A2** | 98.92 % | 613815 | Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910 (3), Autosomal recessive; Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 (3), Autosomal recessive |
| **CYP24A1** | 99.98 % | 126065 | Hypercalcemia, infantile, 1, 143880 (3), Autosomal recessive |
| **CYP26C1** | 99.58 % | 608428 | Focal facial dermal dysplasia 4, 614974 (3), Autosomal recessive |
| **CYP27A1** | 99.91 % | 606530 | Cerebrotendinous xanthomatosis, 213700 (3), Autosomal recessive |
| **CYP27B1** | 99.52 % | 609506 | Vitamin D-dependent rickets, type I, 264700 (3), Autosomal recessive |
| **CYP2R1** | 99.94 % | 608713 | Rickets due to defect in vitamin D 25-hydroxylation deficiency, 600081 (3), Autosomal recessive |
| **CYP2U1** | 99.95 % | 610670 | Spastic paraplegia 56, autosomal recessive, 615030 (3), Autosomal recessive |
| **CYP4F22** | 99.69 % | 611495 | Ichthyosis, congenital, autosomal recessive 5, 604777 (3), Autosomal recessive |
| **CYP4V2** | 99.91 % | 608614 | Bietti crystalline corneoretinal dystrophy, 210370 (3), Autosomal recessive |
| **CYP7B1** | 99.92 % | 603711 | Spastic paraplegia 5A, autosomal recessive, 270800 (3), Autosomal recessive; Bile acid synthesis defect, congenital, 3, 613812 (3), Autosomal recessive |
| **D2HGDH** | 99.94 % | 609186 | D-2-hydroxyglutaric aciduria, 600721 (3), Autosomal recessive |
| **DAAM2** | 99.64 % | 606627 | Nephrotic syndrome, type 24, 619263 (3), Autosomal recessive |
| **DAG1** | 99.98 % | 128239 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818 (3), Autosomal recessive |
| **DALRD3** | 99.64 % | 618904 | Developmental and epileptic encephalopathy 86, 618910 (3), Autosomal recessive |
| **DARS1** | 99.01 % | 603084 | Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281 (3), Autosomal recessive |
| **DARS2** | 98.38 % | 610956 | Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105 (3), Autosomal recessive |
| **DAW1** | 99.83 % | 620279 | Ciliary dyskinesia, primary, 52, 620570 (3), Autosomal recessive |
| **DBH** | 99.81 % | 609312 | Orthostatic hypotension 1, due to DBH deficiency, 223360 (3), Autosomal recessive |
| **DBR1** | 99.92 % | 607024 | Xerosis and growth failure with immune and pulmonary dysfunction syndrome, 620510 (3), Autosomal recessive; {Encephalitis, acute, infection (viral)-induced, susceptibility to, 11}, 619441 (3), Autosomal recessive |
| **DBT** | 96.32 % | 248610 | Maple syrup urine disease, type II, 620699 (3), Autosomal recessive |
| **DCAF17** | 99.65 % | 612515 | Woodhouse-Sakati syndrome, 241080 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **DCC** | 99.9 % | 120470 | Mirror movements 1 and/or agenesis of the corpus callosum, 157600 (3), Autosomal dominant; Esophageal carcinoma, somatic, 133239 (3); Colorectal cancer, somatic, 114500 (3); Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 (3), Autosomal recessive |
| **DCDC2** | 99.89 % | 605755 | Nephronophthisis 19, 616217 (3), Autosomal recessive; ?Deafness, autosomal recessive 66, 610212 (3), Autosomal recessive; Sclerosing cholangitis, neonatal, 617394 (3), Autosomal recessive |
| **DCHS1** | 99.88 % | 603057 | Mitral valve prolapse 2, 607829 (3), Autosomal dominant; Van Maldergem syndrome 1, 601390 (3), Autosomal recessive |
| **DCLRE1C** | 99.78 % | 605988 | Severe combined immunodeficiency, Athabascan type, 602450 (3), Autosomal recessive; Omenn syndrome, 603554 (3), Autosomal recessive |
| **DCPS** | 99.81 % | 610534 | Al-Raqad syndrome, 616459 (3), Autosomal recessive |
| **DCT** | 99.97 % | 191275 | Oculocutaneous albinism, type VIII, 619165 (3), Autosomal recessive |
| **DCX** | 99.97 % | 300121 | Subcortical laminal heterotopia, X-linked, 300067 (3), X-linked; Lissencephaly, X-linked, 300067 (3), X-linked |
| **DDB2** | 99.98 % | 600811 | Xeroderma pigmentosum, group E, DDB-negative subtype, 278740 (3), Autosomal recessive |
| **DDC** | 99.23 % | 107930 | Aromatic L-amino acid decarboxylase deficiency, 608643 (3), Autosomal recessive |
| **DDHD1** | 99.8 % | 614603 | Spastic paraplegia 28, autosomal recessive, 609340 (3), Autosomal recessive |
| **DDHD2** | 99.87 % | 615003 | Spastic paraplegia 54, autosomal recessive, 615033 (3), Autosomal recessive |
| **DDR2** | 98.65 % | 191311 | Warburg-Cinotti syndrome, 618175 (3), Autosomal dominant; Spondylometaepiphyseal dysplasia, short limb-hand type, 271665 (3), Autosomal recessive |
| **DDRGK1** | 99.9 % | 616177 | Spondyloepimetaphyseal dysplasia, Shohat type, 602557 (3), Autosomal recessive |
| **DDX11** | 98.29 % | 601150 | Warsaw breakage syndrome, 613398 (3), Autosomal recessive |
| **DDX3X** | 99.22 % | 300160 | Intellectual developmental disorder, X-linked syndromic, Snijders Blok type, 300958 (3), X-linked dominant, X-linked recessive |
| **DDX59** | 99.38 % | 615464 | Orofaciodigital syndrome V, 174300 (3), Autosomal recessive |
| **DEAF1** | 98.98 % | 602635 | Vulto-van Silfout-de Vries syndrome, 615828 (3), Autosomal dominant; Neurodevelopmental disorder with hypotonia, impaired expressive language, and with or without seizures, 617171 (3), Autosomal recessive |
| **DEF6** | 99.92 % | 610094 | Immunodeficiency 87 and autoimmunity, 619573 (3), Autosomal recessive |
| **DEGS1** | 99.97 % | 615843 | Leukodystrophy, hypomyelinating, 18, 618404 (3), Autosomal recessive |
| **DENND5A** | 99.88 % | 617278 | Developmental and epileptic encephalopathy 49, 617281 (3), Autosomal recessive |
| **DES** | 99.93 % | 125660 | Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 (3), Autosomal dominant; Cardiomyopathy, dilated, 1I, 604765 (3), Autosomal dominant; Myopathy, myofibrillar, 1, 601419 (3), Autosomal recessive, Autosomal dominant |
| **DGAT1** | 99.93 % | 604900 | Diarrhea 7, protein-losing enteropathy type, 615863 (3), Autosomal recessive |
| **DGKE** | 99.15 % | 601440 | {Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008 (3), Autosomal recessive; Nephrotic syndrome, type 7, 615008 (3), Autosomal recessive |
| **DGUOK** | 99.38 % | 601465 | Portal hypertension, noncirrhotic, 1, 617068 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880 (3), Autosomal recessive |
| **DHCR24** | 99.28 % | 606418 | Desmosterolosis, 602398 (3), Autosomal recessive |
| **DHCR7** | 99.89 % | 602858 | Smith-Lemli-Opitz syndrome, 270400 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **DHDDS** | 98.73 % | 608172 | Developmental delay and seizures with or without movement abnormalities, 617836 (3), Autosomal dominant; ?Congenital disorder of glycosylation, type 1bb, 613861 (3), Autosomal recessive; Retinitis pigmentosa 59, 613861 (3), Autosomal recessive |
| **DHFR** | 98.75 % | 126060 | Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839 (3), Autosomal recessive |
| **DHH** | 99.78 % | 605423 | 46XY gonadal dysgenesis with minifascicular neuropathy, 607080 (3), Autosomal recessive; 46XY sex reversal 7, 233420 (3), Autosomal recessive |
| **DHODH** | 99.75 % | 126064 | Miller syndrome, 263750 (3), Autosomal recessive |
| **DHX38** | 99.66 % | 605584 | Retinitis pigmentosa 84, 618220 (3), Autosomal recessive |
| **DIAPH1** | 99.77 % | 602121 | Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900 (3), Autosomal dominant; Seizures, cortical blindness, microcephaly syndrome, 616632 (3), Autosomal recessive |
| **DIS3L2** | 99.75 % | 614184 | Perlman syndrome, 267000 (3), Autosomal recessive |
| **DKC1** | 99.67 % | 300126 | ?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108 (3), X-linked dominant; Dyskeratosis congenita, X-linked, 305000 (3), X-linked recessive |
| **DLAT** | 99.65 % | 608770 | Pyruvate dehydrogenase E2 deficiency, 245348 (3), Autosomal recessive |
| **DLD** | 99.78 % | 238331 | Dihydrolipoamide dehydrogenase deficiency, 246900 (3), Autosomal recessive |
| **DLG3** | 99.84 % | 300189 | Intellectual developmental disorder, X-linked 90, 300850 (3), X-linked recessive |
| **DLG5** | 99.56 % | 604090 | Yuksel-Vogel-Bauser syndrome, 620703 (3), Autosomal recessive |
| **DLL3** | 99.67 % | 602768 | Spondylocostal dysostosis 1, autosomal recessive, 277300 (3), Autosomal recessive |
| **DLX5** | 99.09 % | 600028 | Split-hand/foot malformation 1, 183600 (3), Autosomal dominant; ?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600 (3), Autosomal recessive |
| **DMD** | 99.79 % | 300377 | Becker muscular dystrophy, 300376 (3), X-linked recessive; Cardiomyopathy, dilated, 3B, 302045 (3), X-linked; Duchenne muscular dystrophy, 310200 (3), X-linked recessive |
| **DMGDH** | 99.82 % | 605849 | Dimethylglycine dehydrogenase deficiency, 605850 (3), Autosomal recessive |
| **DMP1** | 100 % | 600980 | Hypophosphatemic rickets, AR, 241520 (3), Autosomal recessive |
| **DMXL2** | 99.85 % | 612186 | Developmental and epileptic encephalopathy 81, 618663 (3), Autosomal recessive; ?Deafness, autosomal dominant 71, 617605 (3), Autosomal dominant; ?Polyendocrine-polyneuropathy syndrome, 616113 (3), Autosomal recessive |
| **DNA2** | 99.6 % | 601810 | Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156 (3), Autosomal dominant; Rothmund-Thomson syndrome, type 4, 620819 (3), Autosomal recessive; Seckel syndrome 8, 615807 (3), Autosomal recessive |
| **DNAAF1** | 99.66 % | 613190 | Ciliary dyskinesia, primary, 13, 613193 (3), Autosomal recessive |
| **DNAAF11** | 99.73 % | 614930 | Ciliary dyskinesia, primary, 19, 614935 (3), Autosomal recessive |
| **DNAAF2** | 99.89 % | 612517 | Ciliary dyskinesia, primary, 10, 612518 (3), Autosomal recessive |
| **DNAAF3** | 99.83 % | 614566 | Ciliary dyskinesia, primary, 2, 606763 (3), Autosomal recessive |
| **DNAAF4** | 99.72 % | 608706 | {Dyslexia, susceptibility to, 1}, 127700 (3), Autosomal dominant; Ciliary dyskinesia, primary, 25, 615482 (3), Autosomal recessive |
| **DNAAF5** | 99.79 % | 614864 | Ciliary dyskinesia, primary, 18, 614874 (3), Autosomal recessive |
| **DNAAF6** | 99.34 % | 300933 | Ciliary dyskinesia, primary, 36, X-linked, 300991 (3), X-linked recessive |
| **DNAH1** | 99.67 % | 603332 | Spermatogenic failure 18, 617576 (3), Autosomal recessive; Ciliary dyskinesia, primary, 37, 617577 (3), Autosomal recessive |
| **DNAH11** | 99.87 % | 603339 | Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **DNAH5** | 99.89 % | 603335 | Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644 (3), Autosomal recessive |
| **DNAH7** | 99.57 % | 610061 | Ciliary dyskinesia, primary, 50, 620356 (3), Autosomal recessive |
| **DNAH9** | 99.89 % | 603330 | Ciliary dyskinesia, primary, 40, 618300 (3), Autosomal recessive |
| **DNAI1** | 99.56 % | 604366 | Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400 (3), Autosomal recessive |
| **DNAI2** | 99.82 % | 605483 | Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444 (3), Autosomal recessive |
| **DNAJB13** | 99.46 % | 610263 | Ciliary dyskinesia, primary, 34, 617091 (3), Autosomal recessive |
| **DNAJB2** | 99.48 % | 604139 | Neuronopathy, distal hereditary motor, autosomal recessive 5, 614881 (3), Autosomal recessive |
| **DNAJB4** | 98.29 % | 611327 | Congenital myopathy 21 with early respiratory failure, 620326 (3), Autosomal recessive |
| **DNAJC12** | 99.44 % | 606060 | Hyperphenylalaninemia, mild, non-BH4-deficient, 617384 (3), Autosomal recessive |
| **DNAJC19** | 99.55 % | 608977 | 3-methylglutaconic aciduria, type V, 610198 (3), Autosomal recessive |
| **DNAJC21** | 99.71 % | 617048 | Bone marrow failure syndrome 3, 617052 (3), Autosomal recessive |
| **DNAJC3** | 99.79 % | 601184 | Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192 (3), Autosomal recessive |
| **DNAJC30** | 99.84 % | 618202 | Leber-like hereditary optic neuropathy, autosomal recessive 1, 619382 (3), Autosomal recessive |
| **DNAJC6** | 98.58 % | 608375 | Parkinson disease 19a, juvenile-onset, 615528 (3), Autosomal recessive; Parkinson disease 19b, early-onset, 615528 (3), Autosomal recessive |
| **DNAL1** | 99.88 % | 610062 | Ciliary dyskinesia, primary, 16, 614017 (3), Autosomal recessive |
| **DNASE1L3** | 99.09 % | 602244 | Systemic lupus erythematosus 16, 614420 (3), Autosomal recessive |
| **DNASE2** | 99.92 % | 126350 | Autoinflammatory-pancytopenia syndrome, 619858 (3), Autosomal recessive |
| **DNM1** | 99.96 % | 602377 | Developmental and epileptic encephalopathy 31B, autosomal recessive, 620352 (3), Autosomal recessive; Developmental and epileptic encephalopathy 31A, autosomal dominant, 616346 (3), Autosomal dominant |
| **DNM1L** | 99.26 % | 603850 | Optic atrophy 5, 610708 (3), Autosomal dominant; Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 (3), Autosomal recessive, Autosomal dominant |
| **DNM2** | 99.86 % | 602378 | Centronuclear myopathy 1, 160150 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal type 2M, 606482 (3), Autosomal dominant; Charcot-Marie-Tooth disease, dominant intermediate B, 606482 (3), Autosomal dominant; Lethal congenital contracture syndrome 5, 615368 (3), Autosomal recessive |
| **DNMBP** | 99.82 % | 611282 | Cataract 48, 618415 (3), Autosomal recessive |
| **DNMT3B** | 99.92 % | 602900 | Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 (3), Autosomal recessive; Facioscapulohumeral muscular dystrophy 4, digenic, 619478 (3), Digenic dominant |
| **DOCK11** | 99.52 % | 300681 | Autoinflammatory disease, multisystem, with immune dysregulation, X-linked, 301109 (3), X-linked recessive |
| **DOCK2** | 99.93 % | 603122 | Immunodeficiency 40, 616433 (3), Autosomal recessive |
| **DOCK3** | 99.83 % | 603123 | Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292 (3), Autosomal recessive |
| **DOCK6** | 99.4 % | 614194 | Adams-Oliver syndrome 2, 614219 (3), Autosomal recessive |
| **DOCK7** | 95.61 % | 615730 | Developmental and epileptic encephalopathy 23, 615859 (3), Autosomal recessive |
| **DOCK8** | 99.75 % | 611432 | Hyper-IgE syndrome 2, autosomal recessive, with recurrent infections, 243700 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **DOHH** | 99.36 % | 611262 | Neurodevelopmental disorder with microcephaly, cerebral atrophy, and visual impairment, 620066 (3), Autosomal recessive |
| **DOK7** | 99.22 % | 610285 | Fetal akinesia deformation sequence 3, 618389 (3), Autosomal recessive; Myasthenic syndrome, congenital, 10, 254300 (3), Autosomal recessive |
| **DOLK** | 99.93 % | 610746 | Congenital disorder of glycosylation, type Im, 610768 (3), Autosomal recessive |
| **DONSON** | 99.84 % | 611428 | Microcephaly, short stature, and limb abnormalities, 617604 (3), Autosomal recessive; Microcephaly-micromelia syndrome, 251230 (3), Autosomal recessive |
| **DPAGT1** | 99.95 % | 191350 | Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750 (3), Autosomal recessive; Congenital disorder of glycosylation, type Ij, 608093 (3), Autosomal recessive |
| **DPH1** | 99.89 % | 603527 | Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901 (3), Autosomal recessive |
| **DPH2** | 99.73 % | 603456 | Developmental delay with short stature, dysmorphic facial features, and sparse hair 2, 620062 (3), Autosomal recessive |
| **DPH5** | 96.35 % | 611075 | Neurodevelopmental disorder with short stature, prominent forehead, and feeding difficulties, 620070 (3), Autosomal recessive |
| **DPM1** | 96.61 % | 603503 | Congenital disorder of glycosylation, type Ie, 608799 (3), Autosomal recessive |
| **DPM2** | 99.92 % | 603564 | Congenital disorder of glycosylation, type Iu, 615042 (3), Autosomal recessive |
| **DPM3** | 99.47 % | 605951 | ?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937 (3), Autosomal recessive |
| **DPP9** | 99.96 % | 608258 | Hatipoglu immunodeficiency syndrome, 620331 (3), Autosomal recessive |
| **DPYS** | 99.89 % | 613326 | Dihydropyrimidinuria, 222748 (3), Autosomal recessive |
| **DRC1** | 99.73 % | 615288 | Spermatogenic failure 80, 620222 (3), Autosomal recessive; Ciliary dyskinesia, primary, 21, 615294 (3), Autosomal recessive |
| **DRP2** | 99.79 % | 300052 | *No OMIM phenotypes* |
| **DSC2** | 99.51 % | 125645 | Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 (3), Autosomal recessive, Autosomal dominant; Arrhythmogenic right ventricular dysplasia 11, 610476 (3), Autosomal recessive, Autosomal dominant |
| **DSC3** | 99.58 % | 600271 | Hypotrichosis and recurrent skin vesicles, 613102 (3), Autosomal recessive |
| **DSE** | 99.99 % | 605942 | Ehlers-Danlos syndrome, musculocontractural type 2, 615539 (3), Autosomal recessive |
| **DSG1** | 99.9 % | 125670 | Keratosis palmoplantaris striata I, AD, 148700 (3), Autosomal dominant; Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508 (3), Autosomal recessive |
| **DSG2** | 99.87 % | 125671 | Cardiomyopathy, dilated, 1BB, 612877 (3), Autosomal recessive; Arrhythmogenic right ventricular dysplasia 10, 610193 (3), Autosomal dominant |
| **DSG4** | 99.88 % | 607892 | Hypotrichosis 6, 607903 (3), Autosomal recessive |
| **DSP** | 99.97 % | 125647 | Arrhythmogenic right ventricular dysplasia 8, 607450 (3), Autosomal dominant; Epidermolysis bullosa, lethal acantholytic, 609638 (3), Autosomal recessive; Keratosis palmoplantaris striata II, 612908 (3), Autosomal dominant; Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821 (3), Autosomal dominant; Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 (3), Autosomal recessive |
| **DST** | 99.87 % | 113810 | Neuropathy, hereditary sensory and autonomic, type VI, 614653 (3), Autosomal recessive; Epidermolysis bullosa simplex 3, localized or generalized intermediate, with bp230 deficiency, 615425 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **DSTYK** | 99.39 % | 612666 | Spastic paraplegia 23, autosomal recessive, 270750 (3), Autosomal recessive; Congenital anomalies of kidney and urinary tract 1, 610805 (3), Autosomal dominant |
| **DTNBP1** | 99.86 % | 607145 | Hermansky-Pudlak syndrome 7, 614076 (3), Autosomal recessive |
| **DTYMK** | 99.94 % | 188345 | Neurodegeneration, childhood-onset, with progressive microcephaly, 619847 (3), Autosomal recessive |
| **DUOX2** | 97.61 % | 606759 | Thyroid dyshormonogenesis 6, 607200 (3), Autosomal recessive |
| **DUOXA2** | 100 % | 612772 | Thyroid dyshormonogenesis 5, 274900 (3), Autosomal recessive |
| **DUT** | 99.66 % | 601266 | Bone marrow failure and diabetes mellitus syndrome, 620044 (3), Autosomal recessive |
| **DYM** | 99.76 % | 607461 | Smith-McCort dysplasia, 607326 (3), Autosomal recessive; Dyggve-Melchior-Clausen disease, 223800 (3), Autosomal recessive |
| **DYNC2H1** | 99.74 % | 603297 | Short-rib thoracic dysplasia 3 with or without polydactyly, 613091 (3), Autosomal recessive, Digenic recessive |
| **DYNC2I1** | 99.94 % | 615462 | Short-rib thoracic dysplasia 8 with or without polydactyly, 615503 (3), Autosomal recessive |
| **DYNC2I2** | 99.68 % | 613363 | Short-rib thoracic dysplasia 11 with or without polydactyly, 615633 (3), Autosomal recessive |
| **DYNC2LI1** | 99.86 % | 617083 | Short-rib thoracic dysplasia 15 with polydactyly, 617088 (3), Autosomal recessive |
| **DYNLT2B** | 99.98 % | 617353 | Short-rib thoracic dysplasia 17 with or without polydactyly, 617405 (3), Autosomal recessive |
| **DYSF** | 99.57 % | 603009 | Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601 (3), Autosomal recessive; Miyoshi muscular dystrophy 1, 254130 (3), Autosomal recessive; Myopathy, distal, with anterior tibial onset, 606768 (3), Autosomal recessive |
| **DZIP1L** | 99.53 % | 617570 | Polycystic kidney disease 5, 617610 (3), Autosomal recessive |
| **EARS2** | 99.44 % | 612799 | Combined oxidative phosphorylation deficiency 12, 614924 (3), Autosomal recessive |
| **EBP** | 99.7 % | 300205 | MEND syndrome, 300960 (3), X-linked recessive; Chondrodysplasia punctata, X-linked dominant, 302960 (3), X-linked dominant |
| **ECEL1** | 99.85 % | 605896 | Arthrogryposis, distal, type 5D, 615065 (3), Autosomal recessive |
| **ECHS1** | 99.94 % | 602292 | Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277 (3), Autosomal recessive |
| **ECM1** | 99.76 % | 602201 | Urbach-Wiethe disease, 247100 (3), Autosomal recessive |
| **EDA** | 99.51 % | 300451 | Tooth agenesis, selective, X-linked 1, 313500 (3), X-linked dominant; Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100 (3), X-linked recessive |
| **EDAR** | 99.34 % | 604095 | [Hair morphology 1, hair thickness], 612630 (3); Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490 (3), Autosomal dominant; Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900 (3), Autosomal recessive |
| **EDARADD** | 99.95 % | 606603 | Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941 (3), Autosomal recessive; Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940 (3), Autosomal dominant |
| **EDEM3** | 98.75 % | 610214 | Congenital disorder of glycosylation, type IIv, 619493 (3), Autosomal recessive |
| **EDN1** | 99.89 % | 131240 | Question mark ears, isolated, 612798 (3), Autosomal dominant; Auriculocondylar syndrome 3, 615706 (3), Autosomal recessive |
| **EDN3** | 99.75 % | 131242 | Waardenburg syndrome, type 4B, 613265 (3), Autosomal recessive, Autosomal dominant; {Hirschsprung disease, susceptibility to, 4}, 613712 (3), Autosomal dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **EDNRB** | 99.98 % | 131244 | {Hirschsprung disease, susceptibility to, 2}, 600155 (3), Autosomal dominant; ?ABCD syndrome, 600501 (3), Autosomal recessive; Waardenburg syndrome, type 4A, 277580 (3), Autosomal recessive, Autosomal dominant |
| **EEF1B2** | 99.87 % | 600655 | *No OMIM phenotypes* |
| **EEF1D** | 99.9 % | 130592 | Neurodevelopmental disorder with thin corpus callosum, hypotonia, and absent language, 621150 (3), Autosomal recessive |
| **EFEMP2** | 99.7 % | 604633 | Cutis laxa, autosomal recessive, type IB, 614437 (3), Autosomal recessive |
| **EFL1** | 98.86 % | 617538 | Shwachman-Diamond syndrome 2, 617941 (3), Autosomal recessive |
| **EFNB1** | 99.61 % | 300035 | Craniofrontonasal dysplasia, 304110 (3), X-linked dominant |
| **EGFR** | 98.85 % | 131550 | Neonatal nephrocutaneous inflammatory syndrome, 616069 (3), Autosomal recessive; Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980 (3), Somatic mutation, Autosomal dominant; Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980 (3), Somatic mutation, Autosomal dominant; {Nonsmall cell lung cancer, susceptibility to}, 211980 (3), Somatic mutation, Autosomal dominant |
| **EGR2** | 99.98 % | 129010 | Dejerine-Sottas disease, 145900 (3), Autosomal recessive, Autosomal dominant; Charcot-Marie-Tooth disease, type 1D, 607678 (3), Autosomal dominant; Hypomyelinating neuropathy, congenital, 1, 605253 (3), Autosomal recessive, Autosomal dominant |
| **EIF2AK3** | 98.74 % | 604032 | Wolcott-Rallison syndrome, 226980 (3), Autosomal recessive |
| **EIF2AK4** | 99.77 % | 609280 | Pulmonary venoocclusive disease 2, 234810 (3), Autosomal recessive |
| **EIF2B1** | 99.75 % | 606686 | Leukoencephalopathy with vanishing white matter 1, with or without ovarian failure, 603896 (3), Autosomal recessive |
| **EIF2B2** | 99.84 % | 606454 | Leukoencephalopathy with vanishing white matter 2, with or without ovarian failure, 620312 (3), Autosomal recessive |
| **EIF2B3** | 96.56 % | 606273 | Leukoencephalopathy with vanishing white matter 3, with or without ovarian failure, 620313 (3), Autosomal recessive |
| **EIF2B4** | 99.52 % | 606687 | Leukoencephalopathy with vanishing white matter 4, with or without ovarian failure, 620314 (3), Autosomal recessive |
| **EIF2B5** | 99.54 % | 603945 | Leukoencephalopathy with vanishing white matter 5, with or without ovarian failure, 620315 (3), Autosomal recessive |
| **EIF2S3** | 99.23 % | 300161 | MEHMO syndrome, 300148 (3), X-linked recessive |
| **EIF3F** | 99.63 % | 603914 | Intellectual developmental disorder, autosomal recessive 67, 618295 (3), Autosomal recessive |
| **EIF4A3** | 99.96 % | 608546 | Robin sequence with cleft mandible and limb anomalies, 268305 (3), Autosomal recessive |
| **ELAC2** | 99.62 % | 605367 | {Prostate cancer, hereditary, 2, susceptibility to}, 614731 (3); Combined oxidative phosphorylation deficiency 17, 615440 (3), Autosomal recessive |
| **ELF4** | 99.9 % | 300775 | Autoinflammatory syndrome, familial, X-linked, Behcet-like 2, 301074 (3), X-linked recessive |
| **ELMO2** | 99.98 % | 606421 | Vascular malformation, primary intraosseous, 606893 (3), Autosomal recessive |
| **ELOVL1** | 99.89 % | 611813 | Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527 (3), Autosomal recessive, Autosomal dominant |
| **ELOVL4** | 99.62 % | 605512 | Spinocerebellar ataxia 34, 133190 (3), Autosomal dominant; Stargardt disease 3, 600110 (3), Autosomal dominant; Ichthyosis, spastic quadriplegia, and impaired intellectual development, 614457 (3), Autosomal recessive |
| **ELP1** | 99.78 % | 603722 | {Medulloblastoma}, 155255 (3), Somatic mutation, Autosomal recessive, Autosomal dominant; Dysautonomia, familial, 223900 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **ELP2** | 99.66 % | 616054 | Intellectual developmental disorder, autosomal recessive 58, 617270 (3), Autosomal recessive |
| **EMC1** | 99.35 % | 616846 | Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875 (3), Autosomal recessive |
| **EMC10** | 99.78 % | 614545 | Neurodevelopmental disorder with dysmorphic facies and variable seizures, 619264 (3), Autosomal recessive |
| **EMD** | 99.65 % | 300384 | Emery-Dreifuss muscular dystrophy 1, X-linked, 310300 (3), X-linked recessive |
| **EMG1** | 99.78 % | 611531 | Bowen-Conradi syndrome, 211180 (3), Autosomal recessive |
| **EML1** | 99.94 % | 602033 | Band heterotopia, 600348 (3), Autosomal recessive |
| **EN1** | 99.98 % | 131290 | ?ENDOVE syndrome, limb-brain type, 619218 (3), Autosomal recessive |
| **ENAM** | 100 % | 606585 | Amelogenesis imperfecta, type IC, 204650 (3), Autosomal recessive; Amelogenesis imperfecta, type IB, 104500 (3), Autosomal dominant |
| **ENO3** | 99.96 % | 131370 | Glycogen storage disease XIII, 612932 (3), Autosomal recessive |
| **ENPP1** | 99.81 % | 173335 | {Obesity, susceptibility to}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial; Hypophosphatemic rickets, autosomal recessive, 2, 613312 (3), Autosomal recessive; {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853 (3), Autosomal dominant; Arterial calcification, generalized, of infancy, 1, 208000 (3), Autosomal recessive; Cole disease, 615522 (3), Autosomal dominant |
| **ENSA** | 99.4 % | 603061 | *No OMIM phenotypes* |
| **ENTPD1** | 99.95 % | 601752 | Spastic paraplegia 64, autosomal recessive, 615683 (3), Autosomal recessive |
| **EOGT** | 98.82 % | 614789 | Adams-Oliver syndrome 4, 615297 (3), Autosomal recessive |
| **EPB41** | 98.12 % | 130500 | Elliptocytosis-1, 611804 (3), Autosomal recessive, Autosomal dominant |
| **EPB41L3** | 99.77 % | 605331 | *No OMIM phenotypes* |
| **EPB42** | 99.94 % | 177070 | Spherocytosis, type 5, 612690 (3) |
| **EPCAM** | 99.85 % | 185535 | Diarrhea 5, with tufting enteropathy, congenital, 613217 (3), Autosomal recessive; Lynch syndrome 8, 613244 (3), Autosomal dominant |
| **EPG5** | 99.86 % | 615068 | Vici syndrome, 242840 (3), Autosomal recessive |
| **EPM2A** | 99.83 % | 607566 | Myoclonic epilepsy of Lafora 1, 254780 (3), Autosomal recessive |
| **EPRS1** | 99.13 % | 138295 | Leukodystrophy, hypomyelinating, 15, 617951 (3), Autosomal recessive |
| **EPS8** | 99.09 % | 600206 | ?Deafness, autosomal recessive 102, 615974 (3), Autosomal recessive |
| **EPS8L2** | 99.78 % | 614988 | Deafness autosomal recessive 106, 617637 (3), Autosomal recessive |
| **ERBB3** | 99.23 % | 190151 | ?Lethal congenital contractural syndrome 2, 607598 (3), Autosomal recessive; {?Erythroleukemia, familial, susceptibility to}, 133180 (3), Autosomal dominant; Visceral neuropathy, familial, 1, autosomal recessive, 243180 (3), Autosomal recessive |
| **ERCC1** | 99.38 % | 126380 | Cerebrooculofacioskeletal syndrome 4, 610758 (3), Autosomal recessive |
| **ERCC2** | 99.66 % | 126340 | Xeroderma pigmentosum, group D, 278730 (3), Autosomal recessive; Trichothiodystrophy 1, photosensitive, 601675 (3), Autosomal recessive; ?Cerebrooculofacioskeletal syndrome 2, 610756 (3), Autosomal recessive |
| **ERCC3** | 99.76 % | 133510 | Trichothiodystrophy 2, photosensitive, 616390 (3), Autosomal recessive; Xeroderma pigmentosum, group B, 610651 (3), Autosomal recessive |
| **ERCC4** | 99.73 % | 133520 | Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 (3), Autosomal recessive; XFE progeroid syndrome, 610965 (3), Autosomal recessive; Xeroderma pigmentosum, group F, 278760 (3), Autosomal recessive; Fanconi anemia, complementation group Q, 615272 (3), Autosomal recessive |
| **ERCC5** | 99.95 % | 133530 | Xeroderma pigmentosum, group G, 278780 (3), Autosomal recessive; Cerebrooculofacioskeletal syndrome 3, 616570 (3), Autosomal recessive; Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **ERCC6** | 99.56 % | 609413 | UV-sensitive syndrome 1, 600630 (3), Autosomal recessive; Cerebrooculofacioskeletal syndrome 1, 214150 (3), Autosomal recessive; ?De Sanctis-Cacchione syndrome, 278800 (3), Autosomal recessive; Cockayne syndrome, type B, 133540 (3), Autosomal recessive; {Macular degeneration, age-related, susceptibility to, 5}, 613761 (3); Premature ovarian failure 11, 616946 (3), Autosomal dominant; {Lung cancer, susceptibility to}, 211980 (3), Somatic mutation, Autosomal dominant |
| **ERCC6L2** | 99.92 % | 615667 | Bone marrow failure syndrome 2, 615715 (3), Autosomal recessive |
| **ERCC8** | 99.6 % | 609412 | UV-sensitive syndrome 2, 614621 (3), Autosomal recessive; Cockayne syndrome, type A, 216400 (3), Autosomal recessive |
| **ERGIC1** | 99.87 % | 617946 | ?Arthrogryposis multiplex congenita 2, neurogenic type, 208100 (3), Autosomal recessive |
| **ERLIN1** | 99.86 % | 611604 | Spastic paraplegia 62, autosomal recessive, 615681 (3), Autosomal recessive |
| **ERLIN2** | 99.83 % | 611605 | Spastic paraplegia 18A, autosomal dominant, 620512 (3), Autosomal dominant; Spastic paraplegia 18B, autosomal recessive, 611225 (3), Autosomal recessive |
| **ESAM** | 99.81 % | 614281 | Neurodevelopmental disorder with intracranial hemorrhage, seizures, and spasticity, 620371 (3), Autosomal recessive |
| **ESCO2** | 99.95 % | 609353 | Juberg-Hayward syndrome, 216100 (3), Autosomal recessive; Roberts-SC phocomelia syndrome, 268300 (3), Autosomal recessive |
| **ESPN** | 99.83 % | 606351 | Deafness, neurosensory, without vestibular involvement, autosomal dominant, 609006 (3), Autosomal recessive; Deafness, autosomal recessive 36, 609006 (3), Autosomal recessive; ?Usher syndrome, type 1M, 618632 (3), Autosomal recessive |
| **ESRP1** | 99.76 % | 612959 | ?Deafness, autosomal recessive 109, 618013 (3), Autosomal recessive |
| **ESRRB** | 99.9 % | 602167 | Deafness, autosomal recessive 35, 608565 (3), Autosomal recessive |
| **ETFA** | 99.45 % | 608053 | Glutaric acidemia IIA, 231680 (3), Autosomal recessive |
| **ETFB** | 99.84 % | 130410 | Glutaric acidemia IIB, 231680 (3), Autosomal recessive |
| **ETFDH** | 99.85 % | 231675 | Glutaric acidemia IIC, 231680 (3), Autosomal recessive |
| **ETHE1** | 99.8 % | 608451 | Ethylmalonic encephalopathy, 602473 (3), Autosomal recessive |
| **EVC** | 99.73 % | 604831 | Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; ?Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant |
| **EVC2** | 99.79 % | 607261 | Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant |
| **EXOC2** | 99.88 % | 615329 | Neurodevelopmental disorder with dysmorphic facies and cerebellar hypoplasia, 619306 (3), Autosomal recessive |
| **EXOC3L2** | 99.81 % | 616927 | Brain malformation renal syndrome, 620943 (3), Autosomal recessive |
| **EXOC6B** | 99.39 % | 607880 | Spondyloepimetaphyseal dysplasia with joint laxity, type 3, 618395 (3), Autosomal recessive |
| **EXOC7** | 99.93 % | 608163 | Neurodevelopmental disorder with seizures and brain atrophy, 619072 (3), Autosomal recessive |
| **EXOSC1** | 99.97 % | 606493 | ?Pontocerebellar hypoplasia, type 1F, 619304 (3), Autosomal recessive |
| **EXOSC2** | 99.98 % | 602238 | Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763 (3), Autosomal recessive |
| **EXOSC3** | 99.98 % | 606489 | Pontocerebellar hypoplasia, type 1B, 614678 (3), Autosomal recessive |
| **EXOSC5** | 98.85 % | 606492 | Cerebellar ataxia, brain abnormalities, and cardiac conduction defects, 619576 (3), Autosomal recessive |
| **EXOSC8** | 99.91 % | 606019 | Pontocerebellar hypoplasia, type 1C, 616081 (3), Autosomal recessive |
| **EXOSC9** | 99.11 % | 606180 | Pontocerebellar hypoplasia, type 1D, 618065 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **EXPH5** | 99.88 % | 612878 | Epidermolysis bullosa simplex 4, localized or generalized intermediate, autosomal recessive, 615028 (3), Autosomal recessive |
| **EXT2** | 99.84 % | 608210 | Seizures, scoliosis, and macrocephaly syndrome, 616682 (3), Autosomal recessive; Exostoses, multiple, type 2, 133701 (3), Autosomal dominant |
| **EXTL3** | 99.94 % | 605744 | Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425 (3), Autosomal recessive |
| **EYS** | 99.84 % | 612424 | Retinitis pigmentosa 25, 602772 (3), Autosomal recessive |
| **F10** | 99.8 % | 613872 | Factor X deficiency, 227600 (3), Autosomal recessive |
| **F11** | 99.97 % | 264900 | Factor XI deficiency, autosomal dominant, 612416 (3); Factor XI deficiency, autosomal recessive, 612416 (3) |
| **F13A1** | 99.56 % | 134570 | Factor XIIIA deficiency, 613225 (3), Autosomal recessive; {Myocardial infarction, protection against}, 608446 (3); {Venous thrombosis, protection against}, 188050 (3), Autosomal dominant |
| **F13B** | 99.28 % | 134580 | Factor XIIIB deficiency, 613235 (3), Autosomal recessive |
| **F2** | 99.82 % | 176930 | Hypoprothrombinemia, 613679 (3), Autosomal recessive; {Pregnancy loss, recurrent, susceptibility to, 2}, 614390 (3), Autosomal dominant; Dysprothrombinemia, 613679 (3), Autosomal recessive; Thrombophilia 1 due to thrombin defect, 188050 (3), Autosomal dominant; {Stroke, ischemic, susceptibility to}, 601367 (3), Multifactorial |
| **F5** | 99.31 % | 612309 | Thrombophilia 2 due to activated protein C resistance, 188055 (3), Autosomal dominant; {Pregnancy loss, recurrent, susceptibility to, 1}, 614389 (3), Autosomal dominant; {Thrombophilia, susceptibility to, due to factor V Leiden}, 188055 (3), Autosomal dominant; {Budd-Chiari syndrome}, 600880 (3), Autosomal recessive; {Stroke, ischemic, susceptibility to}, 601367 (3), Multifactorial; Factor V deficiency, 227400 (3), Autosomal recessive |
| **F7** | 99.98 % | 613878 | {Myocardial infarction, decreased susceptibility to}, 608446 (3); Factor VII deficiency, 227500 (3), Autosomal recessive |
| **F8** | 99.88 % | 300841 | Thrombophilia 13, X-linked, due to factor VIII defect, 301071 (3), X-linked; Hemophilia A, 306700 (3), X-linked recessive |
| **F9** | 99.81 % | 300746 | {Deep venous thrombosis, protection against}, 300807 (3), X-linked recessive; Hemophilia B, 306900 (3), X-linked recessive; Thrombophilia 8, X-linked, due to factor IX defect, 300807 (3), X-linked recessive; {Warfarin sensitivity}, 301052 (3), X-linked |
| **FA2H** | 99.5 % | 611026 | Spastic paraplegia 35, autosomal recessive, 612319 (3), Autosomal recessive |
| **FADD** | 99.61 % | 602457 | Immunodeficiency 90 with encephalopathy, functional hyposplenia, and hepatic dysfunction, 613759 (3), Autosomal recessive |
| **FAH** | 97.69 % | 613871 | Tyrosinemia, type I, 276700 (3), Autosomal recessive |
| **FAM126A** | 99.87 % | 610531 | Leukodystrophy, hypomyelinating, 5, 610532 (3), Autosomal recessive |
| **FAM149B1** | 98.11 % | 618413 | Joubert syndrome 36, 618763 (3), Autosomal recessive |
| **FAM161A** | 99.86 % | 613596 | Retinitis pigmentosa 28, 606068 (3), Autosomal recessive |
| **FAM20A** | 99.95 % | 611062 | Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690 (3), Autosomal recessive |
| **FAM20C** | 99.92 % | 611061 | Raine syndrome, 259775 (3), Autosomal recessive |
| **FAN1** | 99.45 % | 613534 | Interstitial nephritis, karyomegalic, 614817 (3), Autosomal recessive |
| **FANCA** | 99.94 % | 607139 | Fanconi anemia, complementation group A, 227650 (3), Autosomal recessive |
| **FANCB** | 99.51 % | 300515 | Fanconi anemia, complementation group B, 300514 (3), X-linked recessive |
| **FANCC** | 99.91 % | 613899 | Fanconi anemia, complementation group C, 227645 (3), Autosomal recessive |
| **FANCD2** | 99.24 % | 613984 | Fanconi anemia, complementation group D2, 227646 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **FANCE** | 99.85 % | 613976 | Fanconi anemia, complementation group E, 600901 (3), Autosomal recessive |
| **FANCF** | 99.98 % | 613897 | Fanconi anemia, complementation group F, 603467 (3), Autosomal recessive |
| **FANCG** | 99.83 % | 602956 | Fanconi anemia, complementation group G, 614082 (3), Autosomal recessive |
| **FANCI** | 99.87 % | 611360 | Fanconi anemia, complementation group I, 609053 (3), Autosomal recessive |
| **FANCL** | 99.3 % | 608111 | Fanconi anemia, complementation group L, 614083 (3), Autosomal recessive |
| **FAR1** | 99.8 % | 616107 | Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154 (3), Autosomal recessive; Cataracts, spastic paraparesis, and speech delay, 619338 (3), Autosomal dominant |
| **FARS2** | 99.89 % | 611592 | Combined oxidative phosphorylation deficiency 14, 614946 (3), Autosomal recessive; Spastic paraplegia 77, autosomal recessive, 617046 (3), Autosomal recessive |
| **FARSA** | 99.84 % | 602918 | ?Rajab interstitial lung disease with brain calcifications 2, 619013 (3), Autosomal recessive |
| **FARSB** | 99.24 % | 609690 | Rajab interstitial lung disease with brain calcifications 1, 613658 (3), Autosomal recessive |
| **FAS** | 99.94 % | 134637 | Squamous cell carcinoma, burn scar-related, somatic (3); Autoimmune lymphoproliferative syndrome, type IA, 601859 (3), Autosomal dominant; {Autoimmune lymphoproliferative syndrome}, 601859 (3), Autosomal dominant |
| **FASTKD2** | 99.78 % | 612322 | Combined oxidative phosphorylation deficiency 44, 618855 (3), Autosomal recessive |
| **FAT4** | 99.96 % | 612411 | Van Maldergem syndrome 2, 615546 (3), Autosomal recessive; Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 (3), Autosomal recessive |
| **FBLN5** | 99.73 % | 604580 | Cutis laxa, autosomal recessive, type IA, 219100 (3), Autosomal recessive; Charcot-Marie-Tooth disease, demyelinating, type 1H, 619764 (3), Autosomal dominant; Macular degeneration, age-related, 3, 608895 (3), Autosomal dominant; ?Cutis laxa, autosomal dominant 2, 614434 (3), Autosomal dominant |
| **FBP1** | 99.91 % | 611570 | Fructose-1,6-bisphosphatase deficiency, 229700 (3), Autosomal recessive |
| **FBXL3** | 99.76 % | 605653 | Intellectual developmental disorder with short stature, facial anomalies, and speech defects, 606220 (3), Autosomal recessive |
| **FBXL4** | 99.94 % | 605654 | Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471 (3), Autosomal recessive |
| **FBXO31** | 99.02 % | 609102 | ?Intellectual developmental disorder, autosomal recessive 45, 615979 (3), Autosomal recessive |
| **FBXO7** | 99.89 % | 605648 | Parkinson disease 15, autosomal recessive, 260300 (3), Autosomal recessive |
| **FCHO1** | 99.69 % | 613437 | Immunodeficiency 76, 619164 (3), Autosomal recessive |
| **FCSK** | 99.69 % | 608675 | Congenital disorder of glycosylation with defective fucosylation 2, 618324 (3), Autosomal recessive |
| **FDFT1** | 99.66 % | 184420 | Squalene synthase deficiency, 618156 (3), Autosomal recessive |
| **FDX2** | 99.47 % | 614585 | Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900 (3), Autosomal recessive |
| **FDXR** | 99.59 % | 103270 | Multiple mitochondrial dysfunctions syndrome 9B, 620887 (3), Autosomal recessive; Auditory neuropathy and optic atrophy, 617717 (3), Autosomal recessive |
| **FECH** | 99.96 % | 612386 | Protoporphyria, erythropoietic, 1, 177000 (3), Autosomal recessive |
| **FERMT1** | 99.85 % | 607900 | Kindler syndrome, 173650 (3), Autosomal recessive |
| **FERMT3** | 99.73 % | 607901 | Leukocyte adhesion deficiency, type III, 612840 (3), Autosomal recessive |
| **FGA** | 99.96 % | 134820 | Amyloidosis, hereditary systemic 2, 105200 (3), Autosomal dominant; Hypodysfibrinogenemia, congenital, 616004 (3), Autosomal dominant; Dysfibrinogenemia, congenital, 616004 (3), Autosomal dominant; Afibrinogenemia, congenital, 202400 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **FGB** | 99.88 % | 134830 | Hypofibrinogenemia, congenital, 202400 (3), Autosomal recessive; Dysfibrinogenemia, congenital, 616004 (3), Autosomal dominant; Afibrinogenemia, congenital, 202400 (3), Autosomal recessive |
| **FGD1** | 99.65 % | 300546 | Aarskog-Scott syndrome, 305400 (3), X-linked recessive |
| **FGD4** | 99.69 % | 611104 | Charcot-Marie-Tooth disease, type 4H, 609311 (3), Autosomal recessive |
| **FGF13** | 99.74 % | 300070 | Developmental and epileptic encephalopathy 90, 301058 (3), X-linked; Intellectual developmental disorder, X-linked 110, 301095 (3), X-linked recessive |
| **FGF3** | 99.74 % | 164950 | Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706 (3), Autosomal recessive |
| **FGFR3** | 99.87 % | 134934 | Muenke syndrome, 602849 (3), Autosomal dominant; SADDAN, 616482 (3), Autosomal dominant; Hypochondroplasia, 146000 (3), Autosomal dominant; Thanatophoric dysplasia, type II, 187601 (3), Autosomal dominant; Nevus, epidermal, somatic, 162900 (3); CATSHL syndrome, 610474 (3), Autosomal recessive, Autosomal dominant; Thanatophoric dysplasia, type I, 187600 (3), Autosomal dominant; Spermatocytic seminoma, somatic, 273300 (3); Bladder cancer, somatic, 109800 (3); LADD syndrome 2, 620192 (3), Autosomal dominant; Achondroplasia, 100800 (3), Autosomal dominant; Cervical cancer, somatic, 603956 (3); Colorectal cancer, somatic, 114500 (3); Crouzon syndrome with acanthosis nigricans, 612247 (3), Autosomal dominant |
| **FGG** | 99.89 % | 134850 | Dysfibrinogenemia, congenital, 616004 (3), Autosomal dominant; Hypodysfibrinogenemia, 616004 (3), Autosomal dominant; Hypofibrinogenemia, congenital, 202400 (3), Autosomal recessive; Afibrinogenemia, congenital, 202400 (3), Autosomal recessive |
| **FH** | 99.84 % | 136850 | Leiomyomatosis and renal cell cancer, 150800 (3), Autosomal dominant; Fumarase deficiency, 606812 (3), Autosomal recessive |
| **FHL1** | 99.9 % | 300163 | Myopathy, X-linked, with postural muscle atrophy, 300696 (3), X-linked recessive; Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 (3), X-linked recessive; ?Uruguay faciocardiomusculoskeletal syndrome, 300280 (3), X-linked recessive; Scapuloperoneal myopathy, X-linked dominant, 300695 (3), X-linked dominant; Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718 (3), X-linked; Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717 (3), X-linked dominant |
| **FIBP** | 99.97 % | 608296 | Thauvin-Robinet-Faivre syndrome, 617107 (3), Autosomal recessive |
| **FIG4** | 99.67 % | 609390 | Yunis-Varon syndrome, 216340 (3), Autosomal recessive; ?Polymicrogyria, bilateral temporooccipital, 612691 (3), Autosomal recessive; Amyotrophic lateral sclerosis 11, 612577 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 4J, 611228 (3), Autosomal recessive |
| **FITM2** | 99.68 % | 612029 | Siddiqi syndrome, 618635 (3), Autosomal recessive |
| **FKBP10** | 99.7 % | 607063 | Osteogenesis imperfecta, type XI, 610968 (3), Autosomal recessive; Bruck syndrome 1, 259450 (3), Autosomal recessive |
| **FKBP14** | 99.96 % | 614505 | Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557 (3), Autosomal recessive |
| **FKRP** | 99.64 % | 606596 | Muscular dystrophy-dystroglycanopathy (congenital with or without impaired intellectual development), type B, 5, 606612 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **FKTN** | 99.91 % | 607440 | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 (3), Autosomal recessive; Cardiomyopathy, dilated, 1X, 611615 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital without impaired intellectual development), type B, 4, 613152 (3), Autosomal recessive |
| **FLAD1** | 99.79 % | 610595 | Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100 (3), Autosomal recessive |
| **FLG2** | 99.88 % | 616284 | Peeling skin syndrome 6, 618084 (3), Autosomal recessive |
| **FLI1** | 99.96 % | 193067 | Bleeding disorder, platelet-type, 21, 617443 (3), Autosomal recessive, Autosomal dominant |
| **FLNA** | 99.9 % | 300017 | Otopalatodigital syndrome, type II, 304120 (3), X-linked dominant; Intestinal pseudoobstruction, neuronal, 300048 (3), X-linked recessive; Cardiac valvular dysplasia, X-linked, 314400 (3), X-linked; ?FG syndrome 2, 300321 (3), X-linked; Melnick-Needles syndrome, 309350 (3), X-linked dominant; Terminal osseous dysplasia, 300244 (3), X-linked dominant; Congenital short bowel syndrome, 300048 (3), X-linked recessive; Otopalatodigital syndrome, type I, 311300 (3), X-linked dominant; Heterotopia, periventricular, 1, 300049 (3), X-linked dominant; Frontometaphyseal dysplasia 1, 305620 (3), X-linked recessive |
| **FLNB** | 99.84 % | 603381 | Larsen syndrome, 150250 (3), Autosomal dominant; Atelosteogenesis, type I, 108720 (3), Autosomal dominant; Atelosteogenesis, type III, 108721 (3), Autosomal dominant; Spondylocarpotarsal synostosis syndrome, 272460 (3), Autosomal recessive; Boomerang dysplasia, 112310 (3), Autosomal dominant |
| **FLVCR1** | 99.67 % | 609144 | Retinopathy-sensory neuropathy syndrome, 609033 (3), Autosomal recessive; Neurodevelopmental disorder with microcephaly, absent speech, and hypotonia, 621060 (3), Autosomal recessive |
| **FLVCR2** | 99.92 % | 610865 | Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790 (3), Autosomal recessive |
| **FMN2** | 99.94 % | 606373 | Intellectual developmental disorder, autosomal recessive 47, 616193 (3), Autosomal recessive |
| **FMO3** | 99.68 % | 136132 | Trimethylaminuria, 602079 (3), Autosomal recessive |
| **FMR1** | 99.65 % | 309550 | Fragile X tremor/ataxia syndrome, 300623 (3), X-linked dominant; Fragile X syndrome, 300624 (3), X-linked dominant; Premature ovarian failure 1, 311360 (3), X-linked |
| **FNIP1** | 99.71 % | 610594 | Immunodeficiency 93 and hypertrophic cardiomyopathy, 619705 (3), Autosomal recessive |
| **FOCAD** | 99.56 % | 614606 | Liver disease, severe congenital, 619991 (3), Autosomal recessive |
| **FOLR1** | 99.95 % | 136430 | Neurodegeneration due to cerebral folate transport deficiency, 613068 (3), Autosomal recessive |
| **FOXE1** | 99.79 % | 602617 | Bamforth-Lazarus syndrome, 241850 (3), Autosomal recessive; {Thyroid cancer, nonmedullary, 4}, 616534 (3), Autosomal dominant |
| **FOXE3** | 99.91 % | 601094 | Anterior segment dysgenesis 2, multiple subtypes, 610256 (3), Autosomal recessive; {Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349 (3), Autosomal dominant; Cataract 34, multiple types, 612968 (3), Autosomal recessive |
| **FOXI1** | 99.94 % | 601093 | Enlarged vestibular aqueduct, 600791 (3), Autosomal recessive |
| **FOXN1** | 99.44 % | 600838 | T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806 (3), Autosomal dominant; T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705 (3), Autosomal recessive; ?T-cell immunodeficiency with thymic aplasia, 242700 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **FOXP3** | 99.49 % | 300292 | Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790 (3), X-linked recessive |
| **FOXRED1** | 99.82 % | 613622 | Mitochondrial complex I deficiency, nuclear type 19, 618241 (3), Autosomal recessive |
| **FRA10AC1** | 99.88 % | 608866 | Neurodevelopmental disorder with growth retardation, dysmorphic facies, and corpus callosum abnormalities, 620113 (3), Autosomal recessive |
| **FRAS1** | 99.75 % | 607830 | Fraser syndrome 1, 219000 (3), Autosomal recessive |
| **FREM1** | 99.89 % | 608944 | Manitoba oculotrichoanal syndrome, 248450 (3), Autosomal recessive; Bifid nose with or without anorectal and renal anomalies, 608980 (3), Autosomal recessive; Trigonocephaly 2, 614485 (3), Autosomal dominant |
| **FREM2** | 99.94 % | 608945 | Fraser syndrome 2, 617666 (3), Autosomal recessive; Cryptophthalmos, unilateral or bilateral, isolated, 123570 (3), Autosomal recessive |
| **FRMD4A** | 99.87 % | 616305 | ?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819 (3), Autosomal recessive |
| **FRMD7** | 99.9 % | 300628 | Nystagmus, infantile periodic alternating, X-linked, 310700 (3), X-linked; Nystagmus 1, congenital, X-linked, 310700 (3), X-linked |
| **FRMPD4** | 99.87 % | 300838 | Intellectual developmental disorder, X-linked 104, 300983 (3), X-linked |
| **FRRS1L** | 99.69 % | 604574 | Developmental and epileptic encephalopathy 37, 616981 (3), Autosomal recessive |
| **FSHB** | 99.98 % | 136530 | Hypogonadotropic hypogonadism 24 without anosmia, 229070 (3), Autosomal recessive |
| **FSHR** | 99.86 % | 136435 | Ovarian hyperstimulation syndrome, 608115 (3), Autosomal dominant; Ovarian dysgenesis 1, 233300 (3), Autosomal recessive |
| **FTCD** | 99.74 % | 606806 | Glutamate formiminotransferase deficiency, 229100 (3), Autosomal recessive |
| **FTO** | 99.17 % | 610966 | Growth retardation, developmental delay, facial dysmorphism, 612938 (3), Autosomal recessive; {Obesity, susceptibility to, BMIQ14}, 612460 (3), Autosomal recessive |
| **FTSJ1** | 99.67 % | 300499 | Intellectual developmental disorder, X-linked 9, 309549 (3), X-linked recessive |
| **FUCA1** | 98.82 % | 612280 | Fucosidosis, 230000 (3), Autosomal recessive |
| **FUT8** | 99.8 % | 602589 | Congenital disorder of glycosylation with defective fucosylation 1, 618005 (3), Autosomal recessive |
| **FXN** | 99.85 % | 606829 | Friedreich ataxia with retained reflexes, 229300 (3), Autosomal recessive; Friedreich ataxia, 229300 (3), Autosomal recessive |
| **FXR1** | 99.63 % | 600819 | Congenital myopathy 9B, proximal, with minicore lesions, 618823 (3), Autosomal recessive; ?Congenital myopathy 9A with respiratory insufficiency and bone fractures, 618822 (3), Autosomal recessive |
| **FYB1** | 99.9 % | 602731 | Thrombocytopenia 3, 273900 (3), Autosomal recessive |
| **FYCO1** | 99.95 % | 607182 | Cataract 18, autosomal recessive, 610019 (3), Autosomal recessive |
| **FZD6** | 99.92 % | 603409 | Nail disorder, nonsyndromic congenital, 1, 161050 (3), Autosomal recessive |
| **G6PC1** | 99.79 % | 613742 | Glycogen storage disease Ia, 232200 (3), Autosomal recessive |
| **G6PC3** | 99.69 % | 611045 | Dursun syndrome, 612541 (3), Autosomal recessive; Neutropenia, severe congenital 4, autosomal recessive, 612541 (3), Autosomal recessive |
| **G6PD** | 99.61 % | 305900 | Anemia, congenital, nonspherocytic hemolytic, 1, G6PD deficient, 300908 (3), X-linked; {Resistance to malaria due to G6PD deficiency}, 611162 (3) |
| **GAA** | 99.83 % | 606800 | Pompe disease, late-onset, 621314 (3), Autosomal recessive; Pompe disease, infantile-onset, 232300 (3), Autosomal recessive |
| **GABRA3** | 99.9 % | 305660 | Epilepsy, X-linked 2, with or without impaired intellectual development and dysmorphic features, 301091 (3), X-linked |
| **GAD1** | 99.69 % | 605363 | Developmental and epileptic encephalopathy 89, 619124 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **GALC** | 99.8 % | 606890 | Krabbe disease, 245200 (3), Autosomal recessive |
| **GALE** | 99.13 % | 606953 | Thrombocytopenia 13, syndromic, 620776 (3), Autosomal recessive; Galactose epimerase deficiency, 230350 (3), Autosomal recessive |
| **GALK1** | 99.96 % | 604313 | Galactokinase deficiency with cataracts, 230200 (3), Autosomal recessive |
| **GALM** | 99.95 % | 137030 | Galactosemia IV, 618881 (3), Autosomal recessive |
| **GALNS** | 99.66 % | 612222 | Mucopolysaccharidosis IVA, 253000 (3), Autosomal recessive |
| **GALNT2** | 98.46 % | 602274 | Congenital disorder of glycosylation, type IIt, 618885 (3), Autosomal recessive |
| **GALNT3** | 99.16 % | 601756 | Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900 (3), Autosomal recessive |
| **GALT** | 99.95 % | 606999 | Galactosemia, 230400 (3), Autosomal recessive |
| **GAMT** | 99.7 % | 601240 | Cerebral creatine deficiency syndrome 2, 612736 (3), Autosomal recessive |
| **GAN** | 99.87 % | 605379 | Giant axonal neuropathy-1, 256850 (3), Autosomal recessive |
| **GAS2L2** | 99.91 % | 611398 | ?Ciliary dyskinesia, primary, 41, 618449 (3), Autosomal recessive |
| **GAS8** | 99.95 % | 605178 | Ciliary dyskinesia, primary, 33, 616726 (3), Autosomal recessive |
| **GATA1** | 99.78 % | 305371 | Anemia, congenital, nonspherocytic hemolytic, 9, 301083 (3), X-linked recessive; Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 159595 (3); Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367 (3), X-linked recessive; Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835 (3), X-linked recessive; Thrombocytopenia with beta-thalassemia, X-linked, 314050 (3), X-linked recessive |
| **GATC** | 99.97 % | 617210 | Combined oxidative phosphorylation deficiency 42, 618839 (3), Autosomal recessive |
| **GATM** | 99.75 % | 602360 | Cerebral creatine deficiency syndrome 3, 612718 (3), Autosomal recessive; Fanconi renotubular syndrome 1, 134600 (3), Autosomal dominant |
| **GBA** | 93.64 % | 606463 | {Lewy body dementia, susceptibility to}, 127750 (3), Autosomal dominant; Gaucher disease, type II, 230900 (3), Autosomal recessive; Gaucher disease, type IIIC, 231005 (3), Autosomal recessive; Gaucher disease, type III, 231000 (3), Autosomal recessive; Gaucher disease, type I, 230800 (3), Autosomal recessive; Gaucher disease, perinatal lethal, 608013 (3), Autosomal recessive; {Parkinson disease, late-onset, susceptibility to}, 168600 (3), Autosomal dominant, Multifactorial |
| **GBA2** | 99.9 % | 609471 | Spastic paraplegia 46, autosomal recessive, 614409 (3), Autosomal recessive |
| **GBE1** | 99.57 % | 607839 | Glycogen storage disease IV, 232500 (3), Autosomal recessive; Polyglucosan body disease, adult form, 263570 (3), Autosomal recessive |
| **GCDH** | 100 % | 608801 | Glutaricaciduria, type I, 231670 (3), Autosomal recessive |
| **GCGR** | 99.86 % | 138033 | Mahvash disease, 619290 (3), Autosomal recessive |
| **GCH1** | 99.88 % | 600225 | Dystonia, DOPA-responsive, 128230 (3), Autosomal recessive, Autosomal dominant; Hyperphenylalaninemia, BH4-deficient, B, 233910 (3), Autosomal recessive |
| **GCK** | 99.78 % | 138079 | MODY, type II, 125851 (3), Autosomal dominant; Diabetes mellitus, permanent neonatal 1, 606176 (3), Autosomal recessive; Hyperinsulinemic hypoglycemia, familial, 3, 602485 (3), Autosomal dominant; Diabetes mellitus, noninsulin-dependent, late onset, 125853 (3), Autosomal dominant |
| **GCLC** | 99.76 % | 606857 | {Myocardial infarction, susceptibility to}, 608446 (3); Anemia, congenital, nonspherocytic hemolytic, 7, 230450 (3), Autosomal recessive |
| **GCM2** | 99.96 % | 603716 | Hypoparathyroidism, familial isolated 2, 618883 (3), Autosomal recessive, Autosomal dominant; Hyperparathyroidism 4, 617343 (3), Autosomal dominant |
| **GCSH** | 97.94 % | 238330 | Multiple mitochondrial dysfunctions syndrome 7, 620423 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **GDAP1** | 99.9 % | 606598 | Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706 (3), Autosomal recessive; Charcot-Marie-Tooth disease, recessive intermediate, A, 608340 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2K, 607831 (3), Autosomal recessive, Autosomal dominant; Charcot-Marie-Tooth disease, type 4A, 214400 (3), Autosomal recessive |
| **GDF1** | 99.15 % | 602880 | Congenital heart defects, multiple types, 6, 613854 (3), Autosomal dominant; Right atrial isomerism (Ivemark), 208530 (3), Autosomal recessive |
| **GDF5** | 99.97 % | 601146 | Acromesomelic dysplasia 2A, 200700 (3), Autosomal recessive; Acromesomelic dysplasia 2B, 228900 (3), Autosomal recessive; Multiple synostoses syndrome 2, 610017 (3), Autosomal dominant; Symphalangism, proximal, 1B, 615298 (3), Autosomal dominant; Brachydactyly, type A2, 112600 (3), Autosomal dominant; ?Acromesomelic dysplasia 2C, Hunter-Thompson type, 201250 (3), Autosomal recessive; Brachydactyly, type C, 113100 (3), Autosomal dominant; {Osteoarthritis-5}, 612400 (3); Brachydactyly, type A1, C, 615072 (3), Autosomal recessive, Autosomal dominant |
| **GDF6** | 99.99 % | 601147 | Microphthalmia with coloboma 6, digenic, 613703 (3), Autosomal dominant; Microphthalmia, isolated 4, 613094 (3); Leber congenital amaurosis 17, 615360 (3), Autosomal recessive; Multiple synostoses syndrome 4, 617898 (3), Autosomal dominant; Klippel-Feil syndrome 1, autosomal dominant, 118100 (3), Autosomal dominant |
| **GDI1** | 99.85 % | 300104 | Intellectual developmental disorder, X-linked 41, 300849 (3), X-linked dominant |
| **GEMIN5** | 99.8 % | 607005 | Neurodevelopmental disorder with cerebellar atrophy and motor dysfunction, 619333 (3), Autosomal recessive |
| **GET3** | 99.64 % | 601913 | ?Cardiomyopathy, dilated, 2H, 620203 (3), Autosomal recessive |
| **GFER** | 99.98 % | 600924 | Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076 (3), Autosomal recessive |
| **GFM1** | 99.9 % | 606639 | Combined oxidative phosphorylation deficiency 1, 609060 (3), Autosomal recessive |
| **GFM2** | 99.53 % | 606544 | Combined oxidative phosphorylation deficiency 39, 618397 (3), Autosomal recessive |
| **GFPT1** | 99.58 % | 138292 | Myasthenia, congenital, 12, with tubular aggregates, 610542 (3), Autosomal recessive |
| **GFRA1** | 99.71 % | 601496 | Renal hypodysplasia/aplasia 4, 619887 (3), Autosomal recessive |
| **GGCX** | 99.83 % | 137167 | Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450 (3), Autosomal recessive; Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842 (3) |
| **GGPS1** | 99.91 % | 606982 | Muscular dystrophy, congenital hearing loss, and ovarian insufficiency syndrome, 619518 (3), Autosomal recessive |
| **GH1** | 100 % | 139250 | Kowarski syndrome, 262650 (3), Autosomal recessive; Growth hormone deficiency, isolated, type II, 173100 (3), Autosomal dominant; Growth hormone deficiency, isolated, type IB, 612781 (3); Growth hormone deficiency, isolated, type IA, 262400 (3), Autosomal recessive |
| **GHR** | 99.52 % | 600946 | Laron dwarfism, 262500 (3), Autosomal recessive; Increased responsiveness to growth hormone, 604271 (3), Autosomal dominant; Growth hormone insensitivity, partial, 604271 (3), Autosomal dominant; {Hypercholesterolemia, familial, modifier of}, 143890 (3), Autosomal recessive, Autosomal dominant |
| **GHRHR** | 99.96 % | 139191 | Growth hormone deficiency, isolated, type IV, 618157 (3), Autosomal recessive |
| **GIMAP5** | 100 % | 608086 | Portal hypertension, noncirrhotic, 2, 619463 (3), Autosomal recessive |
| **GIMAP6** | 100 % | 616960 | *No OMIM phenotypes* |
| **GINS1** | 99.96 % | 610608 | Immunodeficiency 55, 617827 (3), Autosomal recessive |
| **GIPC3** | 99.63 % | 608792 | Deafness, autosomal recessive 15, 601869 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **GJA1** | 100 % | 121014 | Erythrokeratodermia variabilis et progressiva 3, 617525 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Oculodentodigital dysplasia, 164200 (3), Autosomal dominant; Palmoplantar keratoderma with congenital alopecia, 104100 (3), Autosomal dominant; Syndactyly, type III, 186100 (3), Autosomal dominant; Oculodentodigital dysplasia, autosomal recessive, 257850 (3), Autosomal recessive |
| **GJB1** | 99.98 % | 304040 | Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800 (3), X-linked dominant |
| **GJB2** | 100 % | 121011 | Keratoderma, palmoplantar, with deafness, 148350 (3), Autosomal dominant; Deafness, autosomal recessive 1A, 220290 (3), Digenic dominant, Autosomal recessive; Deafness, autosomal dominant 3A, 601544 (3), Autosomal dominant; Hystrix-like ichthyosis with deafness, 602540 (3), Autosomal dominant; Bart-Pumphrey syndrome, 149200 (3), Autosomal dominant; Keratitis-ichthyosis-deafness syndrome, 148210 (3), Autosomal dominant; Vohwinkel syndrome, 124500 (3), Autosomal dominant |
| **GJB3** | 100 % | 603324 | Deafness, digenic, GJB2/GJB3, 220290 (3), Digenic dominant, Autosomal recessive; Erythrokeratodermia variabilis et progressiva 1, 133200 (3), Autosomal recessive, Autosomal dominant; Deafness, autosomal dominant 2B, with or without peripheral neuropathy, 612644 (3), Autosomal dominant |
| **GJB6** | 100 % | 604418 | Ectodermal dysplasia 2, Clouston type, 129500 (3), Autosomal dominant; Deafness, autosomal dominant 3B, 612643 (3), Autosomal dominant; Deafness, autosomal recessive 1B, 612645 (3), Autosomal recessive; Deafness, digenic GJB2/GJB6, 220290 (3), Digenic dominant, Autosomal recessive |
| **GJC2** | 99.98 % | 608803 | Lymphatic malformation 3, 613480 (3), Autosomal dominant; ?Spastic paraplegia 44, autosomal recessive, 613206 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 2, 608804 (3), Autosomal recessive |
| **GK** | 99.15 % | 300474 | Glycerol kinase deficiency, 307030 (3), X-linked recessive |
| **GLA** | 99.85 % | 300644 | Fabry disease, cardiac variant, 301500 (3), X-linked; Fabry disease, 301500 (3), X-linked |
| **GLB1** | 99.98 % | 611458 | GM1-gangliosidosis, type I, 230500 (3), Autosomal recessive; GM1-gangliosidosis, type III, 230650 (3), Autosomal recessive; Mucopolysaccharidosis type IVB (Morquio), 253010 (3), Autosomal recessive; GM1-gangliosidosis, type II, 230600 (3), Autosomal recessive |
| **GLDC** | 99.78 % | 238300 | Glycine encephalopathy1, 605899 (3), Autosomal recessive |
| **GLDN** | 99.82 % | 608603 | Lethal congenital contracture syndrome 11, 617194 (3), Autosomal recessive |
| **GLE1** | 99.92 % | 603371 | Lethal congenital contracture syndrome 1, 253310 (3), Autosomal recessive; Congenital arthrogryposis with anterior horn cell disease, 611890 (3), Autosomal recessive |
| **GLI1** | 99.29 % | 165220 | Polydactyly, preaxial I, 174400 (3), Autosomal recessive; Polydactyly, postaxial, type A8, 618123 (3), Autosomal recessive |
| **GLIS3** | 99.86 % | 610192 | Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199 (3), Autosomal recessive |
| **GLRA1** | 99.79 % | 138491 | Hyperekplexia 1, 149400 (3), Autosomal recessive, Autosomal dominant |
| **GLRA2** | 99.77 % | 305990 | Intellectual developmental disorder, X-linked syndromic, Pilorge type, 301076 (3), X-linked |
| **GLRB** | 99.37 % | 138492 | Hyperekplexia 2, 614619 (3), Autosomal recessive |
| **GLRX5** | 99.99 % | 609588 | Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 (3), Autosomal recessive; Spasticity, childhood-onset, with hyperglycinemia, 616859 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **GLS** | 99.51 % | 138280 | CASGID syndrome, 618339 (3), Autosomal dominant; Global developmental delay, progressive ataxia, and elevated glutamine, 618412 (3), Autosomal recessive; Developmental and epileptic encephalopathy 71, 618328 (3), Autosomal recessive |
| **GLUL** | 99.4 % | 138290 | Glutamine deficiency, congenital, 610015 (3), Autosomal recessive; Developmental and epileptic encephalopathy 116, 620806 (3), Autosomal dominant |
| **GLYCTK** | 99.9 % | 610516 | D-glyceric aciduria, 220120 (3), Autosomal recessive |
| **GM2A** | 99.95 % | 613109 | GM2-gangliosidosis, AB variant, 272750 (3), Autosomal recessive |
| **GMPPA** | 99.86 % | 615495 | Alacrima, achalasia, and impaired intellectual development syndrome, 615510 (3), Autosomal recessive |
| **GMPPB** | 99.92 % | 615320 | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 14, 615351 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 (3), Autosomal recessive |
| **GNAT1** | 99.81 % | 139330 | Night blindness, congenital stationary, autosomal dominant 3, 610444 (3), Autosomal dominant; Night blindness, congenital stationary, type 1G, 616389 (3), Autosomal recessive |
| **GNAT2** | 99.54 % | 139340 | Achromatopsia 4, 613856 (3) |
| **GNB3** | 99.95 % | 139130 | Night blindness, congenital stationary, type 1H, 617024 (3), Autosomal recessive; {Hypertension, essential, susceptibility to}, 145500 (3), Multifactorial |
| **GNB5** | 99.73 % | 604447 | Lodder-Merla syndrome, type 2, with developmental delay and with or without cardiac arrhythmia, 617182 (3), Autosomal recessive; Lodder-Merla syndrome, type 1, with impaired intellectual development and cardiac arrhythmia, 617173 (3), Autosomal recessive |
| **GNE** | 99.91 % | 603824 | Sialuria, 269921 (3), Autosomal dominant; Thrombocytopenia 12 with or without myopathy, 620757 (3), Autosomal recessive; Nonaka myopathy, 605820 (3), Autosomal recessive |
| **GNMT** | 99.47 % | 606628 | Glycine N-methyltransferase deficiency, 606664 (3), Autosomal recessive |
| **GNPAT** | 99.73 % | 602744 | Rhizomelic chondrodysplasia punctata, type 2, 222765 (3), Autosomal recessive |
| **GNPTAB** | 99.54 % | 607840 | Mucolipidosis III alpha/beta, 252600 (3), Autosomal recessive; Mucolipidosis II alpha/beta, 252500 (3), Autosomal recessive |
| **GNPTG** | 99.93 % | 607838 | Mucolipidosis III gamma, 252605 (3), Autosomal recessive |
| **GNRH1** | 91.65 % | 152760 | ?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841 (3), Autosomal recessive |
| **GNRHR** | 99.75 % | 138850 | Hypogonadotropic hypogonadism 7 without anosmia, 146110 (3), Autosomal recessive |
| **GNS** | 98.91 % | 607664 | Mucopolysaccharidosis type IIID, 252940 (3), Autosomal recessive |
| **GOLGA2** | 100 % | 602580 | Developmental delay with hypotonia, myopathy, and brain abnormalities, 620240 (3), Autosomal recessive |
| **GON7** | 99.48 % | 617436 | Galloway-Mowat syndrome 9, 619603 (3), Autosomal recessive |
| **GORAB** | 99.51 % | 607983 | Geroderma osteodysplasticum, 231070 (3), Autosomal recessive |
| **GOSR2** | 99.7 % | 604027 | Epilepsy, progressive myoclonic 6, 614018 (3), Autosomal recessive; Muscular dystrophy, congenital, with or without seizures, 620166 (3), Autosomal recessive |
| **GOT2** | 99.33 % | 138150 | Developmental and epileptic encephalopathy 82, 618721 (3), Autosomal recessive |
| **GP1BA** | 99.93 % | 606672 | Bernard-Soulier syndrome, type A1 (recessive), 231200 (3), Autosomal recessive; Bernard-Soulier syndrome, type A2 (dominant), 153670 (3), Autosomal dominant; von Willebrand disease, platelet-type, 177820 (3), Autosomal dominant; {Nonarteritic anterior ischemic optic neuropathy, susceptibility to}, 258660 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **GP1BB** | 98.93 % | 138720 | Giant platelet disorder, isolated, 231200 (3), Autosomal recessive; Bernard-Soulier syndrome, type B, 231200 (3), Autosomal recessive |
| **GP6** | 99.97 % | 605546 | Bleeding disorder, platelet-type, 11, 614201 (3), Autosomal recessive |
| **GP9** | 99.95 % | 173515 | Bernard-Soulier syndrome, type C, 231200 (3), Autosomal recessive |
| **GPAA1** | 99.71 % | 603048 | Glycosylphosphatidylinositol biosynthesis defect 15, 617810 (3), Autosomal recessive |
| **GPC3** | 99.69 % | 300037 | Wilms tumor, somatic, 194070 (3); Simpson-Golabi-Behmel syndrome, type 1, 312870 (3), X-linked recessive |
| **GPC4** | 97.57 % | 300168 | Keipert syndrome, 301026 (3), X-linked recessive |
| **GPC6** | 99.73 % | 604404 | Omodysplasia 1, 258315 (3), Autosomal recessive |
| **GPD1** | 99.37 % | 138420 | Hypertriglyceridemia, transient infantile, 614480 (3), Autosomal recessive |
| **GPHN** | 99.56 % | 603930 | Molybdenum cofactor deficiency C, 615501 (3), Autosomal recessive |
| **GPI** | 99.77 % | 172400 | Anemia, congenital, nonspherocytic hemolytic, 4, glucose phosphate isomerase deficient, 613470 (3), Autosomal recessive |
| **GPIHBP1** | 99.83 % | 612757 | Hyperlipoproteinemia, type 1D, 615947 (3), Autosomal recessive |
| **GPNMB** | 99.81 % | 604368 | Amyloidosis, primary localized cutaneous, 3, 617920 (3), Autosomal recessive |
| **GPR101** | 100 % | 300393 | Pituitary adenoma 2, GH-secreting, 300943 (3), X-linked |
| **GPR143** | 99.66 % | 300808 | Ocular albinism, type I, Nettleship-Falls type, 300500 (3), X-linked; Nystagmus 6, congenital, X-linked, 300814 (3), X-linked recessive |
| **GPR179** | 99.93 % | 614515 | Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565 (3), Autosomal recessive |
| **GPR68** | 99.98 % | 601404 | Amelogenesis imperfecta, hypomaturation type, IIA6, 617217 (3), Autosomal recessive |
| **GPSM2** | 97.34 % | 609245 | Chudley-McCullough syndrome, 604213 (3), Autosomal recessive |
| **GPT2** | 99.47 % | 138210 | Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281 (3), Autosomal recessive |
| **GPX4** | 99.94 % | 138322 | Spondylometaphyseal dysplasia, Sedaghatian type, 250220 (3), Autosomal recessive |
| **GRHL2** | 99.68 % | 608576 | Deafness, autosomal dominant 28, 608641 (3), Autosomal dominant; Ectodermal dysplasia/short stature syndrome, 616029 (3), Autosomal recessive; Corneal dystrophy, posterior polymorphous, 4, 618031 (3), Autosomal dominant |
| **GRHPR** | 99.46 % | 604296 | Hyperoxaluria, primary, type II, 260000 (3), Autosomal recessive |
| **GRIA1** | 99.77 % | 138248 | ?Intellectual developmental disorder, autosomal recessive 76, 619931 (3), Autosomal recessive; Intellectual developmental disorder, autosomal dominant 67, 619927 (3), Autosomal dominant |
| **GRIA3** | 99.83 % | 305915 | Intellectual developmental disorder, X-linked syndromic, Wu type, 300699 (3), X-linked recessive |
| **GRID2** | 99.91 % | 602368 | Spinocerebellar ataxia, autosomal recessive 18, 616204 (3), Autosomal recessive |
| **GRIK2** | 99.77 % | 138244 | Neurodevelopmental disorder with impaired language and ataxia and with or without seizures, 619580 (3), Autosomal dominant; Intellectual developmental disorder, autosomal recessive 6, 611092 (3), Autosomal recessive |
| **GRIN1** | 99.96 % | 138249 | Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820 (3), Autosomal recessive; Developmental and epileptic encephalopathy 101, 619814 (3), Autosomal recessive; Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254 (3), Autosomal dominant |
| **GRIP1** | 99.39 % | 604597 | Fraser syndrome 3, 617667 (3), Autosomal recessive |
| **GRM1** | 99.95 % | 604473 | Spinocerebellar ataxia, autosomal recessive 13, 614831 (3), Autosomal recessive; Spinocerebellar ataxia 44, 617691 (3), Autosomal dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **GRM6** | 99.94 % | 604096 | Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270 (3), Autosomal recessive |
| **GRM7** | 99.98 % | 604101 | Neurodevelopmental disorder with seizures, hypotonia, and brain abnormalities, 618922 (3), Autosomal recessive |
| **GRN** | 99.78 % | 138945 | Frontotemporal dementia 2, 607485 (3), Autosomal recessive, Autosomal dominant; Aphasia, primary progressive, 607485 (3), Autosomal recessive, Autosomal dominant; Ceroid lipofuscinosis, neuronal, 11, 614706 (3), Autosomal recessive |
| **GRXCR1** | 99.9 % | 613283 | Deafness, autosomal recessive 25, 613285 (3), Autosomal recessive |
| **GRXCR2** | 100 % | 615762 | ?Deafness, autosomal recessive 101, 615837 (3), Autosomal recessive |
| **GSR** | 98.35 % | 138300 | Anemia, congenital, nonspherocytic hemolytic, 10, glutathione reductase deficient, 618660 (3), Autosomal recessive |
| **GSS** | 99.62 % | 601002 | Anemia, congenital, nonspherocytic hemolytic, 6, glutatione synthetase deficient, 231900 (3), Autosomal recessive; Glutathione synthetase deficiency, 266130 (3), Autosomal recessive |
| **GSX2** | 99.97 % | 616253 | Diencephalic-mesencephalic junction dysplasia syndrome 2, 618646 (3), Autosomal recessive |
| **GTF2E2** | 99.98 % | 189964 | Trichothiodystrophy 6, nonphotosensitive, 616943 (3), Autosomal recessive |
| **GTF2H5** | 99.97 % | 608780 | Trichothiodystrophy 3, photosensitive, 616395 (3), Autosomal recessive |
| **GTF3A** | 99.87 % | 600860 | *No OMIM phenotypes* |
| **GTPBP2** | 99.76 % | 607434 | Jaberi-Elahi syndrome, 617988 (3), Autosomal recessive |
| **GTPBP3** | 99.4 % | 608536 | Combined oxidative phosphorylation deficiency 23, 616198 (3), Autosomal recessive |
| **GUCY1A1** | 99.94 % | 139396 | Moyamoya 6 with achalasia, 615750 (3), Autosomal recessive |
| **GUCY2C** | 99.69 % | 601330 | Diarrhea 6, 614616 (3), Autosomal dominant; Meconium ileus, 614665 (3), Autosomal recessive |
| **GUCY2D** | 99.86 % | 600179 | Cone-rod dystrophy 6, 601777 (3), Autosomal recessive, Autosomal dominant; ?Choroidal dystrophy, central areolar 1, 215500 (3), Autosomal dominant; Leber congenital amaurosis 1, 204000 (3), Autosomal recessive; Night blindness, congenital stationary, type 1I, 618555 (3), Autosomal recessive |
| **GUSB** | 99.73 % | 611499 | Mucopolysaccharidosis VII, 253220 (3), Autosomal recessive |
| **GYG1** | 99.86 % | 603942 | ?Glycogen storage disease XV, 613507 (3), Autosomal recessive; Polyglucosan body myopathy 2, 616199 (3), Autosomal recessive |
| **GYS1** | 99.81 % | 138570 | Glycogen storage disease 0, muscle, 611556 (3), Autosomal recessive |
| **GYS2** | 99.88 % | 138571 | Glycogen storage disease 0, liver, 240600 (3), Autosomal recessive |
| **GZF1** | 99.98 % | 613842 | Joint laxity, short stature, and myopia, 617662 (3), Autosomal recessive |
| **H6PD** | 99.92 % | 138090 | Cortisone reductase deficiency 1, 604931 (3), Autosomal recessive |
| **HAAO** | 99.28 % | 604521 | Vertebral, cardiac, renal, and limb defects syndrome 1, 617660 (3), Autosomal recessive |
| **HACD1** | 99.76 % | 610467 | Congenital myopathy 11, 619967 (3), Autosomal recessive |
| **HACE1** | 99.65 % | 610876 | Spastic paraplegia and psychomotor retardation with or without seizures, 616756 (3), Autosomal recessive |
| **HADH** | 99.35 % | 601609 | Hyperinsulinemic hypoglycemia, familial, 4, 609975 (3), Autosomal recessive; 3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 (3), Autosomal recessive |
| **HADHA** | 99.79 % | 600890 | HELLP syndrome, maternal, of pregnancy, 609016 (3), Autosomal recessive; LCHAD deficiency, 609016 (3), Autosomal recessive; Mitochondrial trifunctional protein deficiency 1, 609015 (3), Autosomal recessive; Fatty liver, acute, of pregnancy, 609016 (3), Autosomal recessive |
| **HADHB** | 99.79 % | 143450 | Mitochondrial trifunctional protein deficiency 2, 620300 (3), Autosomal recessive |
| **HAL** | 99.5 % | 609457 | [Histidinemia], 235800 (3), Autosomal recessive, Autosomal dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **HAMP** | 99.94 % | 606464 | Hemochromatosis, type 2B, 613313 (3), Autosomal recessive |
| **HARS1** | 99.74 % | 142810 | Charcot-Marie-Tooth disease, axonal, type 2W, 616625 (3), Autosomal dominant; Usher syndrome type 3B, 614504 (3), Autosomal recessive |
| **HARS2** | 99.74 % | 600783 | Perrault syndrome 2, 614926 (3), Autosomal recessive |
| **HAX1** | 99.76 % | 605998 | Neutropenia, severe congenital 3, autosomal recessive, 610738 (3), Autosomal recessive |
| **HBA1** | 97.41 % | 141800 | Hemoglobin H disease, nondeletional, 613978 (3); Thalassemias, alpha-, 604131 (3); Heinz body anemias, alpha-, 140700 (3), Autosomal dominant; Methemoglobinemia, alpha type, 617973 (3), Autosomal dominant; Erythrocytosis, familial, 7, 617981 (3), Autosomal dominant |
| **HBA2** | 66.76 % | 141850 | Heinz body anemia, 140700 (3), Autosomal dominant; Thalassemia, alpha-, 604131 (3); Erythrocytosis, familial, 7, 617981 (3), Autosomal dominant; Hemoglobin H disease, deletional and nondeletional, 613978 (3) |
| **HBB** | 100 % | 141900 | Methemoglobinemia, beta type, 617971 (3), Autosomal dominant; Thalassemia-beta, dominant inclusion-body, 603902 (3), Autosomal dominant; Sickle cell disease, 603903 (3), Autosomal recessive; Thalassemia, beta, 613985 (3); Delta-beta thalassemia, 141749 (3), Autosomal dominant; {Malaria, resistance to}, 611162 (3); Hereditary persistence of fetal hemoglobin, 141749 (3), Autosomal dominant; Erythrocytosis, familial, 6, 617980 (3), Autosomal dominant; Heinz body anemia, 140700 (3), Autosomal dominant |
| **HCCS** | 99.85 % | 300056 | Linear skin defects with multiple congenital anomalies 1, 309801 (3), X-linked dominant |
| **HCFC1** | 99.82 % | 300019 | Methylmalonic aciduria and homocysteinemia, cblX type, 309541 (3), X-linked recessive |
| **HDAC8** | 99.57 % | 300269 | Cornelia de Lange syndrome 5, 300882 (3), X-linked dominant |
| **HEATR3** | 99.52 % | 614951 | Diamond-Blackfan anemia 21, 620072 (3), Autosomal recessive |
| **HECTD4** | 99.87 % | 620209 | Neurodevelopmental disorder with seizures, spasticity, and complete or partial agenesis of the corpus callosum, 620250 (3), Autosomal recessive |
| **HEPACAM** | 99.96 % | 611642 | Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 (3), Autosomal recessive; Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without impaired intellectual development, 613926 (3), Autosomal dominant |
| **HERC1** | 99.79 % | 605109 | Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011 (3), Autosomal recessive |
| **HERC2** | 94.55 % | 605837 | Intellectual developmental disorder, autosomal recessive 38, 615516 (3), Autosomal recessive; [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 (3), Autosomal recessive; [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 (3), Autosomal recessive |
| **HES7** | 99.74 % | 608059 | Spondylocostal dysostosis 4, autosomal recessive, 613686 (3), Autosomal recessive |
| **HESX1** | 99.81 % | 601802 | Pituitary hormone deficiency, combined, 5, 182230 (3), Autosomal recessive, Autosomal dominant; Septooptic dysplasia, 182230 (3), Autosomal recessive, Autosomal dominant; Growth hormone deficiency with pituitary anomalies, 182230 (3), Autosomal recessive, Autosomal dominant |
| **HEXA** | 99.88 % | 606869 | [Hex A pseudodeficiency], 272800 (3), Autosomal recessive; GM2-gangliosidosis, several forms, 272800 (3), Autosomal recessive; Tay-Sachs disease, 272800 (3), Autosomal recessive |
| **HEXB** | 99.83 % | 606873 | Sandhoff disease, infantile, juvenile, and adult forms, 268800 (3), Autosomal recessive |
| **HGD** | 98.76 % | 607474 | Alkaptonuria, 203500 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **HGF** | 99.05 % | 142409 | Deafness, autosomal recessive 39, 608265 (3), Autosomal recessive |
| **HGSNAT** | 99.82 % | 610453 | Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 (3), Autosomal recessive; Retinitis pigmentosa 73, 616544 (3), Autosomal recessive |
| **HHAT** | 99.88 % | 605743 | Nivelon-Nivelon-Mabille syndrome, 600092 (3), Autosomal recessive |
| **HIBCH** | 99.59 % | 610690 | 3-hydroxyisobutryl-CoA hydrolase deficiency, 250620 (3), Autosomal recessive |
| **HID1** | 99.88 % | 605752 | Developmental and epileptic encephalopathy 105 with hypopituitarism, 619983 (3), Autosomal recessive |
| **HIKESHI** | 99.85 % | 614908 | Leukodystrophy, hypomyelinating, 13, 616881 (3), Autosomal recessive |
| **HINT1** | 99.88 % | 601314 | Neuromyotonia and axonal neuropathy, autosomal recessive, 137200 (3), Autosomal recessive |
| **HJV** | 99.86 % | 608374 | Hemochromatosis, type 2A, 602390 (3), Autosomal recessive |
| **HK1** | 98.93 % | 142600 | Anemia, congenital, nonspherocytic hemolytic, 5, hexokinase deficient, 235700 (3), Autosomal recessive; Retinitis pigmentosa 79, 617460 (3), Autosomal dominant; Neuropathy, hereditary motor and sensory, Russe type, 605285 (3), Autosomal recessive; Neurodevelopmental disorder with visual defects and brain anomalies, 618547 (3), Autosomal dominant |
| **HLCS** | 99.81 % | 609018 | Holocarboxylase synthetase deficiency, 253270 (3), Autosomal recessive |
| **HMGCL** | 97.26 % | 613898 | HMG-CoA lyase deficiency, 246450 (3), Autosomal recessive |
| **HMGCR** | 99.71 % | 142910 | Muscular dystrophy, limb-girdle, autosomal recessive 28, 620375 (3), Autosomal recessive; [Statins, response to], 620410 (3); [Low density lipoprotein cholesterol level QTL 3], 620410 (3) |
| **HMGCS2** | 98.43 % | 600234 | HMG-CoA synthase-2 deficiency, 605911 (3), Autosomal recessive |
| **HMOX1** | 99.82 % | 141250 | Heme oxygenase-1 deficiency, 614034 (3), Autosomal recessive; {Pulmonary disease, chronic obstructive, susceptibility to}, 606963 (3) |
| **HMX1** | 99.9 % | 142992 | Oculoauricular syndrome, 612109 (3), Autosomal recessive |
| **HNMT** | 99.28 % | 605238 | Intellectual developmental disorder, autosomal recessive 51, 616739 (3), Autosomal recessive; {Asthma, susceptibility to}, 600807 (3), Autosomal dominant |
| **HNRNPH2** | 99.98 % | 300610 | Intellectual developmental disorder, X-linked syndromic, Bain type, 300986 (3), X-linked dominant |
| **HOGA1** | 99.93 % | 613597 | Hyperoxaluria, primary, type III, 613616 (3), Autosomal recessive |
| **HOXA1** | 99.98 % | 142955 | Bosley-Salih-Alorainy syndrome, 601536 (3), Autosomal recessive; Athabaskan brainstem dysgenesis syndrome, 601536 (3), Autosomal recessive |
| **HOXA2** | 100 % | 604685 | Microtia with or without hearing impairment (AD), 612290 (3), Autosomal recessive, Autosomal dominant; ?Microtia, hearing impairment, and cleft palate (AR), 612290 (3), Autosomal recessive, Autosomal dominant |
| **HOXB1** | 99.76 % | 142968 | Facial paresis, hereditary congenital, 3, 614744 (3), Autosomal recessive |
| **HOXC13** | 99.63 % | 142976 | Ectodermal dysplasia 9, hair/nail type, 614931 (3), Autosomal recessive |
| **HPCA** | 99.11 % | 142622 | Dystonia 2, torsion, autosomal recessive, 224500 (3), Autosomal recessive |
| **HPD** | 99.87 % | 609695 | Hawkinsinuria, 140350 (3), Autosomal dominant; Tyrosinemia, type III, 276710 (3), Autosomal recessive |
| **HPDL** | 99.87 % | 618994 | Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026 (3), Autosomal recessive; Spastic paraplegia 83, autosomal recessive, 619027 (3), Autosomal recessive |
| **HPGD** | 99.85 % | 601688 | ?Digital clubbing, isolated congenital, 119900 (3), Autosomal recessive; Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100 (3), Autosomal recessive; Cranioosteoarthropathy, 259100 (3), Autosomal recessive |
| **HPRT1** | 97.88 % | 308000 | Hyperuricemia, HRPT-related, 300323 (3), X-linked recessive; Lesch-Nyhan syndrome, 300322 (3), X-linked recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **HPS1** | 99.94 % | 604982 | Hermansky-Pudlak syndrome 1, 203300 (3), Autosomal recessive |
| **HPS3** | 99.85 % | 606118 | Hermansky-Pudlak syndrome 3, 614072 (3), Autosomal recessive |
| **HPS4** | 99.84 % | 606682 | Hermansky-Pudlak syndrome 4, 614073 (3), Autosomal recessive |
| **HPS5** | 99.85 % | 607521 | Hermansky-Pudlak syndrome 5, 614074 (3), Autosomal recessive |
| **HPS6** | 99.81 % | 607522 | Hermansky-Pudlak syndrome 6, 614075 (3), Autosomal recessive |
| **HPSE2** | 99.96 % | 613469 | Urofacial syndrome 1, 236730 (3), Autosomal recessive |
| **HR** | 99.88 % | 602302 | Atrichia with papular lesions, 209500 (3), Autosomal recessive; Alopecia universalis, 203655 (3), Autosomal recessive |
| **HS2ST1** | 93.17 % | 604844 | Neurofacioskeletal syndrome with or without renal agenesis, 619194 (3), Autosomal recessive |
| **HS6ST2** | 97.33 % | 300545 | ?Paganini-Miozzo syndrome, 301025 (3), X-linked recessive |
| **HSD11B2** | 99.86 % | 614232 | Apparent mineralocorticoid excess, 218030 (3), Autosomal recessive |
| **HSD17B10** | 99.84 % | 300256 | HSD10 mitochondrial disease, 300438 (3), X-linked dominant |
| **HSD17B3** | 99.49 % | 605573 | Pseudohermaphroditism, male, with gynecomastia, 264300 (3), Autosomal recessive |
| **HSD17B4** | 99.55 % | 601860 | D-bifunctional protein deficiency, 261515 (3), Autosomal recessive; Perrault syndrome 1, 233400 (3), Autosomal recessive |
| **HSD3B2** | 99.99 % | 613890 | Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810 (3), Autosomal recessive |
| **HSD3B7** | 99.89 % | 607764 | Bile acid synthesis defect, congenital, 1, 607765 (3), Autosomal recessive |
| **HSPA9** | 99.8 % | 600548 | Even-plus syndrome, 616854 (3), Autosomal recessive; Anemia, sideroblastic, 4, 182170 (3), Autosomal dominant |
| **HSPD1** | 99.77 % | 118190 | Spastic paraplegia 13, autosomal dominant, 605280 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 4, 612233 (3), Autosomal recessive |
| **HSPG2** | 99.18 % | 142461 | Dyssegmental dysplasia, Silverman-Handmaker type, 224410 (3), Autosomal recessive; Schwartz-Jampel syndrome, type 1, 255800 (3), Autosomal recessive |
| **HTRA1** | 99.75 % | 602194 | Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy 2, 616779 (3), Autosomal dominant; Cerebral arteriopathy, autosomal recessive, with subcortical infarcts and leukoencephalopathy 2, 600142 (3), Autosomal recessive |
| **HTRA2** | 99.73 % | 606441 | {Parkinson disease 13}, 610297 (3); 3-methylglutaconic aciduria, type VIII, 617248 (3), Autosomal recessive |
| **HUWE1** | 99.73 % | 300697 | Intellectual developmental disorder, X-linked syndromic, Turner type, 309590 (3), X-linked |
| **HYAL1** | 99.88 % | 607071 | Mucopolysaccharidosis type IX, 601492 (3), Autosomal recessive |
| **HYDIN** | 76.66 % | 610812 | Ciliary dyskinesia, primary, 5, 608647 (3), Autosomal recessive |
| **HYLS1** | 100 % | 610693 | Hydrolethalus syndrome, 236680 (3), Autosomal recessive |
| **IARS1** | 99.6 % | 600709 | Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093 (3), Autosomal recessive |
| **IARS2** | 99.59 % | 612801 | Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007 (3), Autosomal recessive |
| **IBA57** | 99.5 % | 615316 | Multiple mitochondrial dysfunctions syndrome 3, 615330 (3), Autosomal recessive; ?Spastic paraplegia 74, autosomal recessive, 616451 (3), Autosomal recessive |
| **ICOS** | 99.82 % | 604558 | Immunodeficiency, common variable, 1, 607594 (3), Autosomal recessive |
| **IDH3A** | 99.85 % | 601149 | Retinitis pigmentosa 90, 619007 (3), Autosomal recessive |
| **IDH3B** | 99.98 % | 604526 | Retinitis pigmentosa 46, 612572 (3), Autosomal recessive |
| **IDS** | 99.17 % | 300823 | Mucopolysaccharidosis II, 309900 (3), X-linked recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **IDUA** | 99.91 % | 252800 | Mucopolysaccharidosis Is, 607016 (3), Autosomal recessive; Mucopolysaccharidosis Ih/s, 607015 (3), Autosomal recessive; Mucopolysaccharidosis Ih, 607014 (3), Autosomal recessive |
| **IER3IP1** | 99.97 % | 609382 | Microcephaly, epilepsy, and diabetes syndrome, 614231 (3), Autosomal recessive |
| **IFIH1** | 99.64 % | 606951 | Immunodeficiency 95, 619773 (3), Autosomal recessive; Aicardi-Goutieres syndrome 7, 615846 (3), Autosomal dominant; Singleton-Merten syndrome 1, 182250 (3), Autosomal dominant |
| **IFNAR1** | 99.77 % | 107450 | Immunodeficiency 106, susceptibility to viral infections, 619935 (3), Autosomal recessive |
| **IFNAR2** | 99.65 % | 602376 | {Hepatitis B virus, susceptibility to}, 610424 (3); Immunodeficiency 45, 616669 (3), Autosomal recessive |
| **IFNGR1** | 99.85 % | 107470 | {H. pylori infection, susceptibility to}, 600263 (3); Immunodeficiency 27A, mycobacteriosis, AR, 209950 (3), Autosomal recessive; Immunodeficiency 27B, mycobacteriosis, AD, 615978 (3), Autosomal dominant; {Tuberculosis infection, protection against}, 607948 (3); {Tuberculosis, susceptibility to}, 607948 (3); {Hepatitis B virus infection, susceptibility to}, 610424 (3) |
| **IFNGR2** | 99.81 % | 147569 | Immunodeficiency 28, mycobacteriosis, 614889 (3), Autosomal recessive |
| **IFT122** | 99.49 % | 606045 | Cranioectodermal dysplasia 1, 218330 (3), Autosomal recessive |
| **IFT140** | 99.77 % | 614620 | {Polycystic kidney disease 9, susceptibility to}, 621164 (3), Autosomal dominant; Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 (3), Autosomal recessive; Retinitis pigmentosa 80, 617781 (3), Autosomal recessive; Cranioectodermal dysplasia 5, 621180 (3), Autosomal recessive |
| **IFT172** | 99.83 % | 607386 | Retinitis pigmentosa 71, 616394 (3), Autosomal recessive; Bardet-Biedl syndrome 20, 619471 (3), Autosomal recessive; Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 (3), Autosomal recessive |
| **IFT27** | 99.94 % | 615870 | Bardet-Biedl syndrome 19, 615996 (3), Autosomal recessive |
| **IFT43** | 99.83 % | 614068 | ?Cranioectodermal dysplasia 3, 614099 (3), Autosomal recessive; ?Retinitis pigmentosa 81, 617871 (3), Autosomal recessive; Short-rib thoracic dysplasia 18 with polydactyly, 617866 (3), Autosomal recessive |
| **IFT52** | 99.81 % | 617094 | Short-rib thoracic dysplasia 16 with or without polydactyly, 617102 (3), Autosomal recessive |
| **IFT57** | 99.29 % | 606621 | ?Orofaciodigital syndrome XVIII, 617927 (3), Autosomal recessive |
| **IFT74** | 99.71 % | 608040 | Bardet-Biedl syndrome 22, 617119 (3), Autosomal recessive; Spermatogenic failure 58, 619585 (3), Autosomal recessive; Joubert syndrome 40, 619582 (3), Autosomal recessive |
| **IFT80** | 99.71 % | 611177 | Short-rib thoracic dysplasia 2 with or without polydactyly, 611263 (3), Autosomal recessive |
| **IFT81** | 94.88 % | 605489 | Short-rib thoracic dysplasia 19 with or without polydactyly, 617895 (3), Autosomal recessive |
| **IGBP1** | 99.89 % | 300139 | ?Corpus callosum, agenesis of, with impaired intellectual development, ocular coloboma and micrognathia, 300472 (3), X-linked recessive |
| **IGF1** | 99.94 % | 147440 | Insulin-like growth factor I deficiency, 608747 (3), Autosomal recessive |
| **IGF1R** | 99.82 % | 147370 | Insulin-like growth factor I, resistance to, 270450 (3), Autosomal recessive, Autosomal dominant |
| **IGFALS** | 99.95 % | 601489 | Acid-labile subunit, deficiency of, 615961 (3), Autosomal recessive |
| **IGFBP7** | 99.77 % | 602867 | Retinal arterial macroaneurysm with supravalvular pulmonic stenosis, 614224 (3), Autosomal recessive |
| **IGHM** | 99.96 % | 147020 | Agammaglobulinemia 1, 601495 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **IGHMBP2** | 99.71 % | 600502 | Charcot-Marie-Tooth disease, axonal, type 2S, 616155 (3), Autosomal recessive; Neuronopathy, distal hereditary motor, autosomal recessive 1, 604320 (3), Autosomal recessive |
| **IGLL1** | 99.89 % | 146770 | Agammaglobulinemia 2, 613500 (3), Autosomal recessive |
| **IGSF1** | 99.91 % | 300137 | Hypothyroidism, central, and testicular enlargement, 300888 (3), X-linked recessive |
| **IHH** | 99.91 % | 600726 | Acrocapitofemoral dysplasia, 607778 (3), Autosomal recessive; Brachydactyly, type A1, 112500 (3), Autosomal dominant |
| **IKBKB** | 99.03 % | 603258 | Immunodeficiency 15B, 615592 (3), Autosomal recessive; Immunodeficiency 15A, 618204 (3), Autosomal dominant |
| **IKBKG** | 54.61 % | 300248 | Incontinentia pigmenti, 308300 (3), X-linked dominant; Ectodermal dysplasia and immunodeficiency 1, 300291 (3), X-linked recessive; Immunodeficiency 33, 300636 (3), X-linked recessive; Autoinflammatory disease, systemic, X-linked, 301081 (3), X-linked |
| **IL10RA** | 99.78 % | 146933 | Inflammatory bowel disease 28, early onset, autosomal recessive, 613148 (3), Autosomal recessive |
| **IL10RB** | 99.58 % | 123889 | {Hepatitis B virus, susceptibility to}, 610424 (3); Inflammatory bowel disease 25, early onset, autosomal recessive, 612567 (3), Autosomal recessive |
| **IL11RA** | 99.83 % | 600939 | Craniosynostosis and dental anomalies, 614188 (3), Autosomal recessive |
| **IL12B** | 99.83 % | 161561 | Immunodeficiency 29, mycobacteriosis, 614890 (3), Autosomal recessive |
| **IL12RB1** | 98.54 % | 601604 | Immunodeficiency 30, 614891 (3), Autosomal recessive |
| **IL17RA** | 99.91 % | 605461 | Immunodeficiency 51, 613953 (3), Autosomal recessive |
| **IL17RC** | 99.92 % | 610925 | Candidiasis, familial, 9, 616445 (3), Autosomal recessive |
| **IL1RAPL1** | 99.71 % | 300206 | Intellectual developmental disorder, X-linked 21, 300143 (3), X-linked recessive |
| **IL1RN** | 99.11 % | 147679 | Chronic recurrent multifocal osteomyelitis 2, with periostitis and pustulosis, 612852 (3), Autosomal recessive; {Gastric cancer risk after H. pylori infection}, 613659 (3); {Microvascular complications of diabetes 4}, 612628 (3); Interleukin 1 receptor antagonist deficiency, 612852 (3), Autosomal recessive |
| **IL21R** | 98.46 % | 605383 | Immunodeficiency 56, 615207 (3), Autosomal recessive |
| **IL2RA** | 99.89 % | 147730 | Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367 (3), Autosomal recessive; {Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942 (3) |
| **IL2RB** | 99.87 % | 146710 | Immunodeficiency 63 with lymphoproliferation and autoimmunity, 618495 (3), Autosomal recessive |
| **IL2RG** | 99.85 % | 308380 | Combined immunodeficiency, X-linked, moderate, 312863 (3), X-linked recessive; Severe combined immunodeficiency, X-linked, 300400 (3), X-linked recessive |
| **IL36RN** | 99.77 % | 605507 | Psoriasis 14, pustular, 614204 (3), Autosomal recessive |
| **IL6R** | 98.93 % | 147880 | [Interleukin 6, serum level of, QTL], 614752 (3); Hyper-IgE syndrome 5, autosomal recessive, with recurrent infections, 618944 (3), Autosomal recessive; [Interleukin-6 receptor, soluble, serum level of, QTL], 614689 (3) |
| **IL6ST** | 99.8 % | 600694 | Hyper-IgE syndrome 4A, autosomal dominant, with recurrent infections, 619752 (3), Autosomal dominant; Stuve-Wiedemann syndrome 2, 619751 (3), Autosomal recessive; Hyper-IgE syndrome 4B, autosomal recessive, with recurrent infections, 618523 (3), Autosomal recessive; ?Immunodeficiency 94 with autoinflammation and dysmorphic facies, 619750 (3), Autosomal dominant |
| **IL7** | 99.68 % | 146660 | Immunodeficiency 130 with HPV-related verrucosis, 618309 (3), Autosomal recessive |
| **IL7R** | 99.79 % | 146661 | Immunodeficiency 104, severe combined, 608971 (3), Autosomal recessive |
| **ILDR1** | 99.88 % | 609739 | Deafness, autosomal recessive 42, 609646 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **IMPG2** | 99.91 % | 607056 | Retinitis pigmentosa 56, 613581 (3), Autosomal recessive; Macular dystrophy, vitelliform, 5, 616152 (3), Autosomal dominant |
| **INPP5E** | 99.35 % | 613037 | Impaired intellectual development, truncal obesity, retinal dystrophy, and micropenis syndrome, 610156 (3), Autosomal recessive; Joubert syndrome 1, 213300 (3), Autosomal recessive |
| **INPP5K** | 99.78 % | 607875 | Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404 (3), Autosomal recessive |
| **INPPL1** | 99.35 % | 600829 | Opsismodysplasia, 258480 (3), Autosomal recessive |
| **INS** | 99.82 % | 176730 | Diabetes mellitus, insulin-dependent, 2, 125852 (3), Autosomal dominant; Maturity-onset diabetes of the young, type 10, 613370 (3), Autosomal dominant; Hyperproinsulinemia, 616214 (3), Autosomal dominant; Diabetes mellitus, permanent neonatal 4, 618858 (3), Autosomal recessive, Autosomal dominant |
| **INSR** | 99.87 % | 147670 | Rabson-Mendenhall syndrome, 262190 (3), Autosomal recessive; Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 (3); Donohue syndrome, 246200 (3), Autosomal recessive; Hyperinsulinemic hypoglycemia, familial, 5, 609968 (3), Autosomal dominant |
| **INTS1** | 99.84 % | 611345 | Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies, 618571 (3), Autosomal recessive |
| **INTS11** | 99.71 % | 611354 | Neurodevelopmental disorder with motor and language delay, ocular defects, and brain abnormalities, 620428 (3), Autosomal recessive |
| **INTS13** | 98.85 % | 615079 | *No OMIM phenotypes* |
| **INTU** | 99.87 % | 610621 | ?Orofaciodigital syndrome XVII, 617926 (3), Autosomal recessive; ?Short-rib thoracic dysplasia 20 with polydactyly, 617925 (3), Autosomal recessive |
| **INVS** | 99.93 % | 243305 | Nephronophthisis 2, infantile, 602088 (3), Autosomal recessive |
| **IPO8** | 98.33 % | 605600 | VISS syndrome, 619472 (3), Autosomal recessive |
| **IQCB1** | 99.56 % | 609237 | Senior-Loken syndrome 5, 609254 (3), Autosomal recessive |
| **IQSEC1** | 99.87 % | 610166 | Intellectual developmental disorder with short stature and behavioral abnormalities, 618687 (3), Autosomal recessive |
| **IQSEC2** | 99.75 % | 300522 | Intellectual developmental disorder, X-linked 1, 309530 (3), X-linked dominant |
| **IRAK4** | 98.8 % | 606883 | Immunodeficiency 67, 607676 (3), Autosomal recessive |
| **IREB2** | 99.83 % | 147582 | Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451 (3), Autosomal recessive |
| **IRF7** | 99.61 % | 605047 | Immunodeficiency 39, 616345 (3), Autosomal recessive |
| **IRF8** | 99.74 % | 601565 | Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893 (3), Autosomal dominant; Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990 (3), Autosomal recessive |
| **IRF9** | 99.93 % | 147574 | Immunodeficiency 65, susceptibility to viral infections, 618648 (3), Autosomal recessive |
| **IRS4** | 99.97 % | 300904 | Hypothyroidism, congenital, nongoitrous, 9, 301035 (3), X-linked recessive |
| **IRX5** | 99.7 % | 606195 | Hamamy syndrome, 611174 (3), Autosomal recessive |
| **ISCA1** | 99.72 % | 611006 | Multiple mitochondrial dysfunctions syndrome 5, 617613 (3), Autosomal recessive |
| **ISCA2** | 99.68 % | 615317 | Multiple mitochondrial dysfunctions syndrome 4, 616370 (3), Autosomal recessive |
| **ISCU** | 99.35 % | 611911 | Myopathy with lactic acidosis, hereditary, 255125 (3), Autosomal recessive |
| **ISG15** | 99.99 % | 147571 | Immunodeficiency 38, 616126 (3), Autosomal recessive |
| **ITCH** | 98.63 % | 606409 | Autoimmune disease, multisystem, with facial dysmorphism, 613385 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **ITGA2B** | 99.37 % | 607759 | Fetomaternal alloimmune thrombocytopenia 2, 621266 (3), Autosomal dominant; Glanzmann thrombasthenia 1, 273800 (3), Autosomal recessive; Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 (3), Autosomal dominant |
| **ITGA3** | 99.11 % | 605025 | Epidermolysis bullosa, junctional 7, with interstitial lung disease and nephrotic syndrome, 614748 (3), Autosomal recessive |
| **ITGA6** | 99.34 % | 147556 | Epidermolysis bullosa, junctional 6, with pyloric atresia, 619817 (3), Autosomal recessive |
| **ITGA7** | 99.4 % | 600536 | Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204 (3), Autosomal recessive |
| **ITGA8** | 99.75 % | 604063 | Renal hypodysplasia/aplasia 1, 191830 (3), Autosomal recessive |
| **ITGB2** | 99.91 % | 600065 | Leukocyte adhesion deficiency, 116920 (3), Autosomal recessive |
| **ITGB3** | 99.29 % | 173470 | Bleeding disorder, platelet-type, 24, autosomal dominant, 619271 (3), Autosomal dominant; Glanzmann thrombasthenia 2, 619267 (3), Autosomal recessive; Fetomaternal alloimmune thrombocytopenia 1, 621264 (3), Autosomal dominant |
| **ITGB4** | 99.89 % | 147557 | Epidermolysis bullosa, junctional 5B, with pyloric atresia, 226730 (3), Autosomal recessive; Epidermolysis bullosa, junctional 5A, intermediate, 619816 (3), Autosomal recessive |
| **ITGB6** | 99.3 % | 147558 | Amelogenesis imperfecta, type IH, 616221 (3), Autosomal recessive |
| **ITK** | 99.59 % | 186973 | Lymphoproliferative syndrome 1, 613011 (3), Autosomal recessive |
| **ITPA** | 99.92 % | 147520 | [Inosine triphosphatase deficiency], 613850 (3); Developmental and epileptic encephalopathy 35, 616647 (3), Autosomal recessive |
| **ITPR1** | 99.82 % | 147265 | Gillespie syndrome, 206700 (3), Autosomal recessive, Autosomal dominant; Spinocerebellar ataxia 29, congenital nonprogressive, 117360 (3), Autosomal dominant; Spinocerebellar ataxia 15, 606658 (3), Autosomal dominant |
| **IVD** | 99.86 % | 607036 | Isovaleric acidemia, 243500 (3), Autosomal recessive |
| **IYD** | 99.86 % | 612025 | Thyroid dyshormonogenesis 4, 274800 (3), Autosomal recessive |
| **JAG2** | 99.89 % | 602570 | Muscular dystrophy, limb-girdle, autosomal recessive 27, 619566 (3), Autosomal recessive |
| **JAGN1** | 100 % | 616012 | Neutropenia, severe congenital, 6, autosomal recessive, 616022 (3), Autosomal recessive |
| **JAK3** | 99.74 % | 600173 | Severe combined immunodeficiency, autosomal recessive, T-negative/B-positive type, 600802 (3), Autosomal recessive |
| **JAM2** | 99.41 % | 606870 | Basal ganglia calcification, idiopathic, 8, autosomal recessive, 618824 (3), Autosomal recessive |
| **JAM3** | 99.94 % | 606871 | Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730 (3), Autosomal recessive |
| **JPH1** | 99.95 % | 605266 | Congenital myopathy 25, 620964 (3), Autosomal recessive; {?Charcot-Marie-Tooth disease, axonal, autosomal dominant, type 2K, modifier of}, 607831 (3), Autosomal recessive, Autosomal dominant |
| **JPH2** | 99.77 % | 605267 | Cardiomyopathy, dilated, 2E, 619492 (3), Autosomal recessive; Cardiomyopathy, hypertrophic, 17, 613873 (3), Autosomal dominant |
| **JUP** | 99.59 % | 173325 | Naxos disease, 601214 (3), Autosomal recessive; ?Arrhythmogenic right ventricular dysplasia 12, 611528 (3), Autosomal dominant |
| **KARS1** | 99.8 % | 601421 | Deafness, autosomal recessive 89, 613916 (3), Autosomal recessive; Leukoencephalopathy, progressive, infantile-onset, with or without deafness, 619147 (3), Autosomal recessive; ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 (3), Autosomal recessive; Deafness, congenital, and adult-onset progressive leukoencephalopathy, 619196 (3), Autosomal recessive |
| **KATNB1** | 99.46 % | 602703 | Lissencephaly 6, with microcephaly, 616212 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **KATNIP** | 99.1 % | 616650 | Joubert syndrome 26, 616784 (3), Autosomal recessive |
| **KCNE1** | 86.07 % | 176261 | Jervell and Lange-Nielsen syndrome 2, 612347 (3), Autosomal recessive; Long QT syndrome 5, 613695 (3), Autosomal dominant |
| **KCNJ1** | 100 % | 600359 | Bartter syndrome, type 2, 241200 (3), Autosomal recessive |
| **KCNJ10** | 99.83 % | 602208 | Enlarged vestibular aqueduct, digenic, 600791 (3), Autosomal recessive; SESAME syndrome, 612780 (3), Autosomal recessive |
| **KCNJ11** | 100 % | 600937 | Diabetes, permanent neonatal 2, with or without neurologic features, 618856 (3), Autosomal dominant; Maturity-onset diabetes of the young, type 13, 616329 (3), Autosomal dominant; Diabetes mellitus, transient neonatal 3, 610582 (3), Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 2, 601820 (3), Autosomal recessive, Autosomal dominant |
| **KCNJ13** | 99.97 % | 603208 | Snowflake vitreoretinal degeneration, 193230 (3), Autosomal dominant; Leber congenital amaurosis 16, 614186 (3), Autosomal recessive |
| **KCNJ16** | 100 % | 605722 | Hypokalemic tubulopathy and deafness, 619406 (3), Autosomal recessive |
| **KCNMA1** | 99.72 % | 600150 | {Epilepsy, idiopathic generalized, susceptibility to, 16}, 618596 (3), Autosomal dominant; Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446 (3), Autosomal dominant; Cerebellar atrophy, developmental delay, and seizures, 617643 (3), Autosomal recessive; Liang-Wang syndrome, 618729 (3), Autosomal dominant |
| **KCNQ1** | 99.9 % | 607542 | Short QT syndrome 2, 609621 (3), Autosomal dominant; Atrial fibrillation, familial, 3, 607554 (3), Autosomal dominant; Long QT syndrome 1, 192500 (3), Autosomal dominant; {Long QT syndrome 1, acquired, susceptibility to}, 192500 (3), Autosomal dominant; Jervell and Lange-Nielsen syndrome, 220400 (3), Autosomal recessive |
| **KCNV2** | 99.95 % | 607604 | Cone dystrophy with supernormal rod responses, 610356 (3), Autosomal recessive |
| **KCTD3** | 99.38 % | 613272 | *No OMIM phenotypes* |
| **KCTD7** | 99.92 % | 611725 | Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726 (3), Autosomal recessive |
| **KDELR2** | 99.83 % | 609024 | Osteogenesis imperfecta, type XXI, 619131 (3), Autosomal recessive |
| **KDM5B** | 98.77 % | 605393 | Intellectual developmental disorder, autosomal recessive 65, 618109 (3), Autosomal recessive |
| **KDM5C** | 99.83 % | 314690 | Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type, 300534 (3), X-linked recessive |
| **KDM6A** | 99.7 % | 300128 | Kabuki syndrome 2, 300867 (3), X-linked dominant |
| **KDSR** | 99.92 % | 136440 | Erythrokeratodermia variabilis et progressiva 4, 617526 (3), Autosomal recessive |
| **KERA** | 99.43 % | 603288 | Cornea plana 2, autosomal recessive, 217300 (3), Autosomal recessive |
| **KHDC3L** | 99.99 % | 611687 | Hydatidiform mole, recurrent, 2, 614293 (3), Autosomal recessive |
| **KIAA0586** | 99.33 % | 610178 | Short-rib thoracic dysplasia 14 with polydactyly, 616546 (3), Autosomal recessive; Joubert syndrome 23, 616490 (3), Autosomal recessive |
| **KIAA0753** | 99.95 % | 617112 | ?Orofaciodigital syndrome XV, 617127 (3), Autosomal recessive; ?Joubert syndrome 38, 619476 (3), Autosomal recessive; Short-rib thoracic dysplasia 21 without polydactyly, 619479 (3), Autosomal recessive |
| **KIAA0825** | 99.85 % | 617266 | Polydactyly, postaxial, type A10, 618498 (3), Autosomal recessive |
| **KIAA1109** | 99.76 % | 611565 | Alkuraya-Kucinskas syndrome, 617822 (3), Autosomal recessive |
| **KIAA1549** | 99.87 % | 613344 | Retinitis pigmentosa 86, 618613 (3), Autosomal recessive |
| **KIDINS220** | 99.86 % | 615759 | Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296 (3), Autosomal dominant; Ventriculomegaly and arthrogryposis, 619501 (3), Autosomal recessive |
| **KIF12** | 99.81 % | 611278 | Cholestasis, progressive familial intrahepatic, 8, 619662 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **KIF14** | 98.75 % | 611279 | Microcephaly 20, primary, autosomal recessive, 617914 (3), Autosomal recessive; ?Meckel syndrome 12, 616258 (3), Autosomal recessive |
| **KIF1A** | 99.77 % | 601255 | NESCAV syndrome, 614255 (3), Autosomal dominant; Neuropathy, hereditary sensory, type IIC, 614213 (3), Autosomal recessive; Spastic paraplegia 30, autosomal dominant, 610357 (3), Autosomal dominant; Spastic paraplegia 30, autosomal recessive, 620607 (3), Autosomal recessive |
| **KIF1C** | 99.87 % | 603060 | Spastic ataxia 2, autosomal recessive, 611302 (3), Autosomal recessive |
| **KIF21A** | 98.45 % | 608283 | Fibrosis of extraocular muscles, congenital, 3B, 135700 (3), Autosomal dominant; Fibrosis of extraocular muscles, congenital, 1, 135700 (3), Autosomal dominant |
| **KIF26A** | 99.91 % | 613231 | Cortical dysplasia, complex, with other brain malformations 11, 620156 (3), Autosomal recessive |
| **KIF4A** | 99.61 % | 300521 | Taurodontism, microdontia, and dens invaginatus, 313490 (3), X-linked recessive; Intellectual developmental disorder, X-linked 100, 300923 (3), X-linked recessive |
| **KIF7** | 99.84 % | 611254 | Joubert syndrome 12, 200990 (3), Autosomal recessive; Acrocallosal syndrome, 200990 (3), Autosomal recessive; ?Hydrolethalus syndrome 2, 614120 (3), Autosomal recessive; ?Al-Gazali-Bakalinova syndrome, 607131 (3), Autosomal recessive |
| **KIFBP** | 99.5 % | 609367 | Goldberg-Shprintzen megacolon syndrome, 609460 (3), Autosomal recessive |
| **KIRREL1** | 98.9 % | 607428 | Nephrotic syndrome, type 23, 619201 (3), Autosomal recessive |
| **KISS1R** | 99.93 % | 604161 | Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 (3), Autosomal recessive; ?Precocious puberty, central, 1, 176400 (3), Autosomal dominant |
| **KIZ** | 99.95 % | 615757 | Retinitis pigmentosa 69, 615780 (3), Autosomal recessive |
| **KLC2** | 99.76 % | 611729 | Spastic paraplegia, optic atrophy, and neuropathy, 609541 (3), Autosomal recessive |
| **KLHL15** | 99.83 % | 300980 | Intellectual developmental disorder, X-linked 103, 300982 (3), X-linked recessive |
| **KLHL24** | 99.87 % | 611295 | Cardiomyopathy, familial hypertrophic, 29, with polyglucosan bodies, 620236 (3), Autosomal recessive; Epidermolysis bullosa simplex 6, generalized intermediate, with or without cardiomyopathy, 617294 (3), Autosomal dominant |
| **KLHL3** | 99.68 % | 605775 | Pseudohypoaldosteronism, type IID, 614495 (3), Autosomal recessive, Autosomal dominant |
| **KLHL40** | 99.87 % | 615340 | Nemaline myopathy 8, autosomal recessive, 615348 (3), Autosomal recessive |
| **KLHL41** | 99.69 % | 607701 | Nemaline myopathy 9, 615731 (3), Autosomal recessive |
| **KLHL7** | 99.8 % | 611119 | Retinitis pigmentosa 42, 612943 (3), Autosomal dominant; PERCHING syndrome, 617055 (3), Autosomal recessive |
| **KLK4** | 99.86 % | 603767 | Amelogenesis imperfecta, type IIA1, 204700 (3), Autosomal recessive |
| **KNL1** | 99.65 % | 609173 | Microcephaly 4, primary, autosomal recessive, 604321 (3), Autosomal recessive |
| **KPTN** | 99.71 % | 615620 | Intellectual developmental disorder, autosomal recessive 41, 615637 (3), Autosomal recessive |
| **KREMEN1** | 99.75 % | 609898 | Ectodermal dysplasia 13, hair/tooth type, 617392 (3), Autosomal recessive |
| **KRT1** | 99.7 % | 139350 | Ichthyosis, annular epidermolytic 2, 620148 (3), Autosomal dominant; Palmoplantar keratoderma, nonepidermolytic, 600962 (3), Autosomal dominant; Epidermolytic hyperkeratosis 1, 113800 (3), Autosomal dominant; Palmoplantar keratoderma, epidermolytic, 2, 620411 (3), Autosomal dominant; Keratosis palmoplantaris striata III, 607654 (3); Ichthyosis histrix, Curth-Macklin type, 146590 (3), Autosomal dominant |
| **KRT10** | 99.88 % | 148080 | Ichthyosis, annular epidermolytic 1, 607602 (3), Autosomal dominant; Epidermolytic hyperkeratosis 2B, autosomal recessive, 620707 (3), Autosomal recessive; Epidermolytic hyperkeratosis 2A, autosomal dominant, 620150 (3), Autosomal dominant; ?Ichthyosis histrix, Lambert type, 146600 (3), Autosomal dominant; Ichthyosis with confetti, 609165 (3), Autosomal dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **KRT14** | 99.88 % | 148066 | Epidermolysis bullosa simplex 1D, generalized, intermediate or severe, autosomal recessive, 601001 (3), Autosomal recessive; Epidermolysis bullosa simplex 1C, localized, 131800 (3), Autosomal dominant; Dermatopathia pigmentosa reticularis, 125595 (3), Autosomal dominant; Epidermolysis bullosa simplex 1A, generalized severe, 131760 (3), Autosomal dominant; Naegeli-Franceschetti-Jadassohn syndrome, 161000 (3), Autosomal dominant; Epidermolysis bullosa simplex 1B, generalized intermediate, 131900 (3), Autosomal dominant |
| **KRT18** | 99.75 % | 148070 | Cirrhosis, cryptogenic, 215600 (3), Autosomal recessive; {Cirrhosis, noncryptogenic, susceptibility to}, 215600 (3), Autosomal recessive |
| **KRT25** | 99.2 % | 616646 | Woolly hair, autosomal recessive 3, 616760 (3), Autosomal recessive |
| **KRT5** | 99.91 % | 148040 | Epidermolysis bullosa simplex 2A, generalized severe, 619555 (3), Autosomal dominant; Dowling-Degos disease 1, 179850 (3), Autosomal dominant; Epidermolysis bullosa simplex 2F, with mottled pigmentation, 131960 (3), Autosomal dominant; Epidermolysis bullosa simplex 2D, generalized, intermediate or severe, autosomal recessive, 619599 (3), Autosomal recessive; Epidermolysis bullosa simplex 2B, generalized intermediate, 619588 (3), Autosomal dominant; Epidermolysis bullosa simplex 2C, localized, 619594 (3), Autosomal dominant; Epidermolysis bullosa simplex 2E, with migratory circinate erythema, 609352 (3), Autosomal dominant |
| **KRT74** | 98.52 % | 608248 | Woolly hair, autosomal dominant, 194300 (3), Autosomal dominant; ?Hypotrichosis 3, 613981 (3), Autosomal dominant; ?Ectodermal dysplasia 7, hair/nail type, 614929 (3), Autosomal recessive |
| **KRT85** | 99.74 % | 602767 | Ectodermal dysplasia 4, hair/nail type, 602032 (3), Autosomal recessive |
| **KY** | 99.81 % | 605739 | Myopathy, myofibrillar, 7, 617114 (3), Autosomal recessive |
| **KYNU** | 99.13 % | 605197 | ?Hydroxykynureninuria, 236800 (3), Autosomal recessive; Vertebral, cardiac, renal, and limb defects syndrome 2, 617661 (3), Autosomal recessive |
| **L1CAM** | 99.88 % | 308840 | MASA syndrome, 303350 (3), X-linked recessive; Hydrocephalus, congenital, X-linked, 307000 (3), X-linked recessive; ?Corpus callosum, partial agenesis of, 304100 (3), X-linked recessive |
| **L2HGDH** | 99.76 % | 609584 | L-2-hydroxyglutaric aciduria, 236792 (3), Autosomal recessive |
| **LACC1** | 99.96 % | 613409 | Juvenile arthritis, 618795 (3), Autosomal recessive |
| **LAGE3** | 99.82 % | 300060 | Galloway-Mowat syndrome 2, X-linked, 301006 (3), X-linked recessive |
| **LAMA1** | 99.81 % | 150320 | Poretti-Boltshauser syndrome, 615960 (3), Autosomal recessive |
| **LAMA2** | 99.69 % | 156225 | Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 (3), Autosomal recessive; Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855 (3), Autosomal recessive |
| **LAMA3** | 99.76 % | 600805 | Epidermolysis bullosa, junctional 2A, intermediate, 619783 (3), Autosomal recessive; Epidermolysis bullosa, junctional 2C, laryngoonychocutaneous, 245660 (3), Autosomal recessive; Epidermolysis bullosa, junctional 2B, severe, 619784 (3), Autosomal recessive |
| **LAMA5** | 99.73 % | 601033 | Nephrotic syndrome, type 26, 620049 (3), Autosomal recessive; ?Bent bone dysplasia syndrome 2, 620076 (3), Autosomal recessive |
| **LAMB1** | 99.56 % | 150240 | Lissencephaly 5, 615191 (3), Autosomal recessive |
| **LAMB2** | 99.88 % | 150325 | Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 (3), Autosomal recessive; Pierson syndrome, 609049 (3), Autosomal recessive |
| **LAMB3** | 99.71 % | 150310 | Epidermolysis bullosa, junctional 1B, severe, 226700 (3), Autosomal recessive; Epidermolysis bullosa, junctional 1A, intermediate, 226650 (3), Autosomal recessive; Amelogenesis imperfecta, type IA, 104530 (3), Autosomal dominant |
| **LAMC2** | 99.5 % | 150292 | Epidermolysis bullosa, junctional 3B, severe, 619786 (3), Autosomal recessive; Epidermolysis bullosa, junctional 3A, intermediate, 619785 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **LAMC3** | 99.83 % | 604349 | Cortical malformations, occipital, 614115 (3), Autosomal recessive |
| **LAMP2** | 99.41 % | 309060 | Danon disease, 300257 (3), X-linked dominant |
| **LARGE1** | 99.86 % | 603590 | Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 6, 608840 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 (3), Autosomal recessive |
| **LARP7** | 99.8 % | 612026 | Alazami syndrome, 615071 (3), Autosomal recessive |
| **LARS1** | 99.83 % | 151350 | ?Infantile liver failure syndrome 1, 615438 (3), Autosomal recessive |
| **LARS2** | 99.68 % | 604544 | Perrault syndrome 4, 615300 (3), Autosomal recessive; Hydrops, lactic acidosis, and sideroblastic anemia, 617021 (3), Autosomal recessive |
| **LAS1L** | 99.88 % | 300964 | Wilson-Turner syndrome, 309585 (3), X-linked recessive |
| **LAT** | 99.26 % | 602354 | Immunodeficiency 52, 617514 (3), Autosomal recessive |
| **LBR** | 99.84 % | 600024 | Pelger-Huet anomaly, 169400 (3), Autosomal dominant; ?Reynolds syndrome, 613471 (3), Autosomal dominant; Rhizomelic skeletal dysplasia with or without Pelger-Huet anomaly, 618019 (3), Autosomal recessive; Greenberg skeletal dysplasia, 215140 (3), Autosomal recessive |
| **LCA5** | 99.89 % | 611408 | Leber congenital amaurosis 5, 604537 (3), Autosomal recessive |
| **LCAT** | 99.91 % | 606967 | Fish-eye disease, 136120 (3), Autosomal recessive; Norum disease, 245900 (3), Autosomal recessive |
| **LCK** | 99.72 % | 153390 | Immunodeficiency 22, 615758 (3), Autosomal recessive |
| **LCP2** | 99.55 % | 601603 | Immunodeficiency 81, 619374 (3), Autosomal recessive |
| **LCT** | 99.66 % | 603202 | Lactase deficiency, congenital, 223000 (3), Autosomal recessive |
| **LDHA** | 99.77 % | 150000 | Glycogen storage disease XI, 612933 (3), Autosomal recessive |
| **LDHB** | 99.2 % | 150100 | [Lactate dehydrogenase-B deficiency], 614128 (3) |
| **LDLR** | 99.93 % | 606945 | LDL cholesterol level QTL2, 143890 (3), Autosomal recessive, Autosomal dominant; Hypercholesterolemia, familial, 1, 143890 (3), Autosomal recessive, Autosomal dominant |
| **LDLRAP1** | 96.07 % | 605747 | Hypercholesterolemia, familial, 4, 603813 (3), Autosomal recessive |
| **LEMD2** | 99.88 % | 616312 | Marbach-Rustad progeroid syndrome, 619322 (3), Autosomal dominant; Cataract 46, juvenile-onset, 212500 (3), Autosomal recessive |
| **LEP** | 99.98 % | 164160 | Obesity, morbid, due to leptin deficiency, 614962 (3), Autosomal recessive |
| **LEPR** | 97.13 % | 601007 | Obesity, morbid, due to leptin receptor deficiency, 614963 (3), Autosomal recessive |
| **LETM1** | 99.7 % | 604407 | Neurodegeneration, childhood-onset, with multisystem involvement due to mitochondrial dysfunction, 620089 (3), Autosomal recessive |
| **LFNG** | 99.97 % | 602576 | Spondylocostal dysostosis 3, autosomal recessive, 609813 (3), Autosomal recessive |
| **LGI3** | 99.98 % | 608302 | Intellectual developmental disorder with muscle tone abnormalities and distal skeletal defects, 620007 (3), Autosomal recessive |
| **LGI4** | 99.84 % | 608303 | Arthrogryposis multiplex congenita 1, neurogenic, with myelin defect, 617468 (3), Autosomal recessive |
| **LHB** | 99.93 % | 152780 | Hypogonadotropic hypogonadism 23 with or without anosmia, 228300 (3), Autosomal recessive |
| **LHCGR** | 99.74 % | 152790 | Leydig cell adenoma, somatic, with precocious puberty, 176410 (3); Leydig cell hypoplasia with pseudohermaphroditism, 238320 (3), Autosomal recessive; Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320 (3), Autosomal recessive; Luteinizing hormone resistance, female, 238320 (3), Autosomal recessive; Precocious puberty, male, 176410 (3), Autosomal dominant |
| **LHFPL5** | 99.98 % | 609427 | Deafness, autosomal recessive 67, 610265 (3), Autosomal recessive |
| **LHX3** | 99.79 % | 600577 | Pituitary hormone deficiency, combined, 3, 221750 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **LIAS** | 99.91 % | 607031 | Hyperglycinemia, lactic acidosis, and seizures, 614462 (3), Autosomal recessive |
| **LIFR** | 99.8 % | 151443 | Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559 (3), Autosomal recessive |
| **LIG1** | 99.2 % | 126391 | Immunodeficiency 96, 619774 (3), Autosomal recessive |
| **LIG3** | 99.93 % | 600940 | Mitochondrial DNA depletion syndrome 20 (MNGIE type), 619780 (3), Autosomal recessive |
| **LIG4** | 99.99 % | 601837 | LIG4 syndrome, 606593 (3), Autosomal recessive; {Multiple myeloma, resistance to}, 254500 (3), Somatic mutation |
| **LINGO1** | 99.98 % | 609791 | Intellectual developmental disorder, autosomal recessive 64, 618103 (3), Autosomal recessive |
| **LINS1** | 99.88 % | 610350 | Intellectual developmental disorder, autosomal recessive 27, 614340 (3), Autosomal recessive |
| **LIPA** | 99.94 % | 613497 | Wolman disease, 620151 (3), Autosomal recessive; Cholesteryl ester storage disease, 278000 (3), Autosomal recessive |
| **LIPC** | 99.32 % | 151670 | {Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant; Hepatic lipase deficiency, 614025 (3), Autosomal recessive; [High density lipoprotein cholesterol level QTL 12], 612797 (3) |
| **LIPE** | 99.9 % | 151750 | Lipodystrophy, familial partial, type 6, 615980 (3), Autosomal recessive |
| **LIPN** | 99.82 % | 613924 | Ichthyosis, congenital, autosomal recessive 8, 613943 (3), Autosomal recessive |
| **LIPT1** | 99.89 % | 610284 | Lipoyltransferase 1 deficiency, 616299 (3), Autosomal recessive |
| **LIPT2** | 99.92 % | 617659 | Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668 (3), Autosomal recessive |
| **LMAN1** | 99.91 % | 601567 | Combined factor V and VIII deficiency, 227300 (3), Autosomal recessive |
| **LMBR1** | 99.88 % | 605522 | Syndactyly, type IV, 186200 (3), Autosomal dominant; Laurin-Sandrow syndrome, 135750 (3), Autosomal dominant; Acheiropody, 200500 (3), Autosomal recessive; Triphalangeal thumb-polysyndactyly syndrome, 190605 (3), Autosomal dominant |
| **LMBRD1** | 99.1 % | 612625 | Methylmalonic aciduria and homocystinuria, cblF type, 277380 (3), Autosomal recessive |
| **LMF1** | 99.94 % | 611761 | Lipase deficiency, combined, 246650 (3), Autosomal recessive |
| **LMNA** | 99.84 % | 150330 | Mandibuloacral dysplasia, 248370 (3), Autosomal recessive; Heart-hand syndrome, Slovenian type, 610140 (3), Autosomal dominant; Cardiomyopathy, dilated, 1A, 115200 (3), Autosomal dominant; Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 (3), Autosomal recessive; Restrictive dermopathy 2, 619793 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2B1, 605588 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 (3), Autosomal dominant; Hutchinson-Gilford progeria, 176670 (3), Autosomal dominant; Lipodystrophy, familial partial, type 2, 151660 (3), Autosomal dominant; Muscular dystrophy, congenital, 613205 (3), Autosomal dominant; Malouf syndrome, 212112 (3), Autosomal dominant |
| **LMOD1** | 99.98 % | 602715 | ?Megacystis-microcolon-intestinal hypoperistalsis syndrome 3, 619362 (3), Autosomal recessive |
| **LMOD2** | 99.98 % | 608006 | Cardiomyopathy, dilated, 2G, 619897 (3), Autosomal recessive |
| **LMOD3** | 99.97 % | 616112 | Nemaline myopathy 10, 616165 (3), Autosomal recessive |
| **LNPK** | 97.53 % | 610236 | Neurodevelopmental disorder with epilepsy and hypoplasia of the corpus callosum, 618090 (3), Autosomal recessive |
| **LONP1** | 99.79 % | 605490 | CODAS syndrome, 600373 (3), Autosomal recessive |
| **LOXHD1** | 99.89 % | 613072 | Deafness, autosomal recessive 77, 613079 (3), Autosomal recessive |
| **LOXL3** | 98.95 % | 607163 | Myopia 28, autosomal recessive, 619781 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **LPAR6** | 99.99 % | 609239 | Hypotrichosis 8, 278150 (3), Autosomal recessive; Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150 (3), Autosomal recessive |
| **LPIN1** | 99.78 % | 605518 | Myoglobinuria, acute recurrent, autosomal recessive, 268200 (3), Autosomal recessive |
| **LPIN2** | 99.9 % | 605519 | Majeed syndrome, 609628 (3) |
| **LPL** | 99.92 % | 609708 | Lipoprotein lipase deficiency, 238600 (3), Autosomal recessive; [High density lipoprotein cholesterol level QTL 11], 238600 (3), Autosomal recessive; Combined hyperlipidemia, familial, 144250 (3), Autosomal dominant |
| **LRAT** | 99.99 % | 604863 | Leber congenital amaurosis 14, 613341 (3), Autosomal recessive; Retinal dystrophy, early-onset severe, 613341 (3), Autosomal recessive; Retinitis pigmentosa, juvenile, 613341 (3), Autosomal recessive |
| **LRBA** | 99.8 % | 606453 | Immunodeficiency, common variable, 8, with autoimmunity, 614700 (3), Autosomal recessive |
| **LRIG2** | 98.22 % | 608869 | Urofacial syndrome 2, 615112 (3), Autosomal recessive |
| **LRIT3** | 99.86 % | 615004 | Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058 (3), Autosomal recessive |
| **LRMDA** | 99 % | 614537 | Albinism, oculocutaneous, type VII, 615179 (3), Autosomal recessive |
| **LRP2** | 99.52 % | 600073 | Donnai-Barrow syndrome, 222448 (3), Autosomal recessive |
| **LRP4** | 99.58 % | 604270 | ?Myasthenic syndrome, congenital, 17, 616304 (3), Autosomal recessive; Sclerosteosis 2, 614305 (3), Autosomal recessive, Autosomal dominant; Cenani-Lenz syndactyly syndrome, 212780 (3), Autosomal recessive |
| **LRP5** | 99.56 % | 603506 | Osteopetrosis, autosomal dominant 1, 607634 (3), Autosomal dominant; Polycystic liver disease 4 with or without kidney cysts, 617875 (3), Autosomal dominant; Endosteal hyperostosis, 144750 (3), Autosomal dominant; Osteoporosis-pseudoglioma syndrome, 259770 (3), Autosomal recessive; [Bone mineral density variability 1, high bone mass], 601884 (3), Autosomal dominant; Exudative vitreoretinopathy 4, 601813 (3), Autosomal recessive, Autosomal dominant |
| **LRPAP1** | 99.81 % | 104225 | Myopia 23, autosomal recessive, 615431 (3), Autosomal recessive |
| **LRPPRC** | 99.32 % | 607544 | Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111 (3), Autosomal recessive |
| **LRRC32** | 99.96 % | 137207 | Cleft palate, proliferative retinopathy, and developmental delay, 619074 (3), Autosomal recessive |
| **LRRC45** | 99.77 % | 621312 | *No OMIM phenotypes* |
| **LRRC56** | 99.77 % | 618227 | Ciliary dyskinesia, primary, 39, 618254 (3), Autosomal recessive |
| **LRRK1** | 99.93 % | 610986 | Osteosclerotic metaphyseal dysplasia, 615198 (3), Autosomal recessive |
| **LRSAM1** | 99.95 % | 610933 | Charcot-Marie-Tooth disease, axonal, type 2P, 614436 (3), Autosomal recessive, Autosomal dominant |
| **LRTOMT** | 99.6 % | 612414 | Deafness, autosomal recessive 63, 611451 (3), Autosomal recessive |
| **LSR** | 99.71 % | 616582 | *No OMIM phenotypes* |
| **LSS** | 99.85 % | 600909 | Hypotrichosis 14, 618275 (3), Autosomal recessive; Cataract 44, 616509 (3), Autosomal recessive; Alopecia-intellectual disability syndrome 4, 618840 (3), Autosomal recessive |
| **LTBP1** | 99.63 % | 150390 | Cutis laxa, autosomal recessive, type IIE, 619451 (3), Autosomal recessive |
| **LTBP2** | 99.69 % | 602091 | Glaucoma 3, primary congenital, D, 613086 (3); Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750 (3), Autosomal recessive; ?Weill-Marchesani syndrome 3, recessive, 614819 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **LTBP3** | 99.41 % | 602090 | Dental anomalies and short stature, 601216 (3), Autosomal recessive; Geleophysic dysplasia 3, 617809 (3), Autosomal dominant |
| **LTBP4** | 99.61 % | 604710 | Cutis laxa, autosomal recessive, type IC, 613177 (3), Autosomal recessive |
| **LTV1** | 99.89 % | 620074 | Inflammatory poikiloderma with hair abnormalities and acral keratoses, 620199 (3), Autosomal recessive |
| **LYRM4** | 95.94 % | 613311 | ?Combined oxidative phosphorylation deficiency 19, 615595 (3), Autosomal recessive |
| **LYRM7** | 99.99 % | 615831 | Mitochondrial complex III deficiency, nuclear type 8, 615838 (3), Autosomal recessive |
| **LYST** | 99.87 % | 606897 | Chediak-Higashi syndrome, 214500 (3), Autosomal recessive |
| **LZTFL1** | 99.98 % | 606568 | Bardet-Biedl syndrome 17, 615994 (3), Autosomal recessive |
| **LZTR1** | 99.3 % | 600574 | Noonan syndrome 2, 605275 (3), Autosomal recessive; Noonan syndrome 10, 616564 (3), Autosomal dominant; {Schwannomatosis-2, susceptibility to}, 615670 (3), Autosomal dominant |
| **MADD** | 99.72 % | 603584 | Neurodevelopmental disorder with dysmorphic facies, impaired speech and hypotonia, 619005 (3), Autosomal recessive; DEEAH syndrome, 619004 (3), Autosomal recessive |
| **MAG** | 99.67 % | 159460 | Spastic paraplegia 75, autosomal recessive, 616680 (3), Autosomal recessive |
| **MAGED2** | 99.94 % | 300470 | Bartter syndrome, type 5, antenatal, transient, 300971 (3), X-linked recessive |
| **MAGI2** | 99.61 % | 606382 | Nephrotic syndrome, type 15, 617609 (3), Autosomal recessive |
| **MAGT1** | 99.56 % | 300715 | Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853 (3), X-linked recessive; Congenital disorder of glycosylation, type Icc, 301031 (3), X-linked recessive |
| **MAK** | 99.6 % | 154235 | Retinitis pigmentosa 62, 614181 (3), Autosomal recessive |
| **MALT1** | 99.59 % | 604860 | Immunodeficiency 12, 615468 (3), Autosomal recessive |
| **MAN1B1** | 99.8 % | 604346 | Rafiq syndrome, 614202 (3), Autosomal recessive |
| **MAN2B1** | 99.51 % | 609458 | Mannosidosis, alpha-, types I and II, 248500 (3), Autosomal recessive |
| **MAN2C1** | 99.64 % | 154580 | Congenital disorder of deglycosylation 2, 619775 (3), Autosomal recessive |
| **MANBA** | 99.66 % | 609489 | Mannosidosis, beta, 248510 (3), Autosomal recessive |
| **MAOA** | 99.12 % | 309850 | Brunner syndrome, 300615 (3), X-linked recessive |
| **MAP3K14** | 98.93 % | 604655 | Immunodeficiency 112, 620449 (3), Autosomal recessive |
| **MAP3K20** | 99.66 % | 609479 | Centronuclear myopathy 6 with fiber-type disproportion, 617760 (3), Autosomal recessive; Split-foot malformation with mesoaxial polydactyly, 616890 (3), Autosomal recessive |
| **MAPKAPK5** | 99.78 % | 606723 | Neurocardiofaciodigital syndrome, 619869 (3), Autosomal recessive |
| **MAPKBP1** | 99.77 % | 616786 | Nephronophthisis 20, 617271 (3), Autosomal recessive |
| **MAPT** | 99.6 % | 157140 | Supranuclear palsy, progressive, 601104 (3), Autosomal dominant; Frontotemporal dementia 1, with or without parkinsonism, 600274 (3), Autosomal dominant; Supranuclear palsy, progressive atypical, 260540 (3), Autosomal recessive; {Parkinson disease, susceptibility to}, 168600 (3), Autosomal dominant, Multifactorial; Pick disease, 172700 (3), Autosomal dominant |
| **MARS1** | 99.43 % | 156560 | Spastic paraplegia 70, autosomal recessive, 620323 (3), Autosomal recessive; Interstitial lung and liver disease, 615486 (3), Autosomal recessive; ?Trichothiodystrophy 9, nonphotosensitive, 619692 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2U, 616280 (3), Autosomal dominant |
| **MARS2** | 99.98 % | 609728 | ?Combined oxidative phosphorylation deficiency 25, 616430 (3), Autosomal recessive; Spastic ataxia 3, autosomal recessive, 611390 (3), Autosomal recessive |
| **MARVELD2** | 99.79 % | 610572 | Deafness, autosomal recessive 49, 610153 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **MASP1** | 99.57 % | 600521 | 3MC syndrome 1, 257920 (3), Autosomal recessive |
| **MAT1A** | 98.69 % | 610550 | Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 (3), Autosomal recessive, Autosomal dominant; Methionine adenosyltransferase deficiency, autosomal recessive, 250850 (3), Autosomal recessive, Autosomal dominant |
| **MATN3** | 99.65 % | 602109 | {Osteoarthritis susceptibility 2}, 140600 (3), Autosomal dominant; Spondyloepimetaphyseal dysplasia, Borochowitz-Cormier-Daire type, 608728 (3), Autosomal recessive; Epiphyseal dysplasia, multiple, 5, 607078 (3), Autosomal dominant |
| **MBD4** | 99.87 % | 603574 | {Uveal melanoma, susceptibility to, 1}, 606660 (3), Autosomal dominant; Tumor predisposition syndrome 2, 619975 (3), Autosomal recessive |
| **MBOAT7** | 99.81 % | 606048 | Intellectual developmental disorder, autosomal recessive 57, 617188 (3), Autosomal recessive |
| **MBTPS1** | 99.4 % | 603355 | Spondyloepiphyseal dysplasia, Kondo-Fu type, 618392 (3), Autosomal recessive; CAOP syndrome, 621252 (3), Autosomal recessive |
| **MBTPS2** | 99.85 % | 300294 | Keratosis follicularis spinulosa decalvans, X-linked, 308800 (3), X-linked recessive; Osteogenesis imperfecta, type XIX, 301014 (3), X-linked recessive; IFAP syndrome with or without BRESHECK syndrome, 308205 (3), X-linked recessive; ?Olmsted syndrome, X-linked, 300918 (3), X-linked recessive |
| **MC2R** | 99.99 % | 607397 | Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200 (3), Autosomal recessive |
| **MC4R** | 99.96 % | 155541 | Obesity (BMIQ20), 618406 (3), Autosomal recessive, Autosomal dominant; {Obesity, resistance to (BMIQ20)}, 618406 (3), Autosomal recessive, Autosomal dominant |
| **MCAT** | 99.86 % | 614479 | Optic atrophy 15, 620583 (3), Autosomal recessive |
| **MCCC1** | 99.44 % | 609010 | 3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200 (3), Autosomal recessive |
| **MCCC2** | 99.86 % | 609014 | 3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210 (3), Autosomal recessive |
| **MCEE** | 99.74 % | 608419 | Methylmalonyl-CoA epimerase deficiency, 251120 (3), Autosomal recessive |
| **MCFD2** | 99.94 % | 607788 | Factor V and factor VIII, combined deficiency of, 613625 (3) |
| **MCIDAS** | 99.93 % | 614086 | Ciliary dyskinesia, primary, 42, 618695 (3), Autosomal recessive |
| **MCM10** | 99.8 % | 609357 | Immunodeficiency 80 with or without cardiomyopathy, 619313 (3), Autosomal recessive |
| **MCM3AP** | 99.89 % | 603294 | Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124 (3), Autosomal recessive |
| **MCM4** | 99.77 % | 602638 | Immunodeficiency 54, 609981 (3), Autosomal recessive |
| **MCOLN1** | 99.64 % | 605248 | Lisch epithelial corneal dystrophy, 620763 (3), Autosomal dominant; Mucolipidosis IV, 252650 (3), Autosomal recessive |
| **MCPH1** | 99.56 % | 607117 | Microcephaly 1, primary, autosomal recessive, 251200 (3), Autosomal recessive |
| **MCTS1** | 99.69 % | 300587 | Immunodeficiency 118, mycobacteriosis, 301115 (3), X-linked recessive |
| **MDFIC** | 99.59 % | 614511 | Lymphatic malformation 12, 620014 (3), Autosomal recessive |
| **MDH2** | 98.1 % | 154100 | Developmental and epileptic encephalopathy 51, 617339 (3), Autosomal recessive |
| **MDM2** | 98.57 % | 164785 | {Accelerated tumor formation, susceptibility to}, 614401 (3), Autosomal dominant; ?Lessel-Kubisch syndrome, 618681 (3), Autosomal recessive |
| **MECP2** | 99.91 % | 300005 | Rett syndrome, atypical, 312750 (3), X-linked dominant; Encephalopathy, neonatal severe, 300673 (3), X-linked recessive; Intellectual developmental disorder, X-linked syndromic, Lubs type, 300260 (3), X-linked recessive; {Autism susceptibility, X-linked 3}, 300496 (3), X-linked; Intellectual developmental disorder, X-linked syndromic 13, 300055 (3), X-linked recessive; Rett syndrome, 312750 (3), X-linked dominant; Rett syndrome, preserved speech variant, 312750 (3), X-linked dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **MECR** | 98.95 % | 608205 | Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282 (3), Autosomal recessive; Optic atrophy 16, 620629 (3), Autosomal recessive |
| **MED11** | 99.79 % | 612383 | Neurodegeneration with developmental delay, early respiratory failure, myoclonic seizures, and brain abnormalities, 620327 (3), Autosomal recessive |
| **MED12** | 99.77 % | 300188 | Lujan-Fryns syndrome, 309520 (3), X-linked recessive; Ohdo syndrome, X-linked, 300895 (3), X-linked recessive; Hardikar syndrome, 301068 (3), X-linked dominant; Opitz-Kaveggia syndrome, 305450 (3), X-linked recessive |
| **MED17** | 99.66 % | 603810 | Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668 (3), Autosomal recessive |
| **MED23** | 99.79 % | 605042 | Intellectual developmental disorder, autosomal recessive 18, with or without epilepsy, 614249 (3), Autosomal recessive |
| **MED25** | 98.48 % | 610197 | Basel-Vanagaite-Smirin-Yosef syndrome, 616449 (3), Autosomal recessive |
| **MED27** | 99.82 % | 605044 | Neurodevelopmental disorder with spasticity, cataracts, and cerebellar hypoplasia, 619286 (3), Autosomal recessive |
| **MEFV** | 99.97 % | 608107 | Neutrophilic dermatosis, acute febrile, 608068 (3), Autosomal dominant; Familial Mediterranean fever, AR, 249100 (3), Autosomal recessive; Familial Mediterranean fever, AD, 134610 (3), Autosomal dominant |
| **MEGF10** | 99.65 % | 612453 | Congenital myopathy 10A, severe variant, 614399 (3), Autosomal recessive; Congenital myopathy 10B, mild variant, 620249 (3), Autosomal recessive |
| **MEGF8** | 99.36 % | 604267 | Carpenter syndrome 2, 614976 (3), Autosomal recessive |
| **MEOX1** | 99.87 % | 600147 | Klippel-Feil syndrome 2, 214300 (3), Autosomal recessive |
| **MERTK** | 98.41 % | 604705 | Retinitis pigmentosa 38, 613862 (3), Autosomal recessive |
| **MESD** | 99.71 % | 607783 | Osteogenesis imperfecta, type XX, 618644 (3), Autosomal recessive |
| **MESP2** | 99.91 % | 605195 | Spondylocostal dysostosis 2, autosomal recessive, 608681 (3), Autosomal recessive |
| **METTL23** | 99.99 % | 615262 | Intellectual developmental disorder, autosomal recessive 44, 615942 (3), Autosomal recessive |
| **METTL5** | 99.65 % | 618628 | Intellectual developmental disorder, autosomal recessive 72, 618665 (3), Autosomal recessive |
| **MFF** | 99.89 % | 614785 | Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086 (3), Autosomal recessive |
| **MFN2** | 99.82 % | 608507 | Lipomatosis, multiple symmetric, with or without peripheral neuropathy, 151800 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 (3), Autosomal recessive; Hereditary motor and sensory neuropathy VIA, 601152 (3), Autosomal dominant |
| **MFRP** | 99.6 % | 606227 | Microphthalmia, isolated 5, 611040 (3), Autosomal recessive; Nanophthalmos 2, 609549 (3), Autosomal recessive |
| **MFSD2A** | 99.18 % | 614397 | Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities, 616486 (3), Autosomal recessive |
| **MFSD8** | 99.75 % | 611124 | Macular dystrophy with central cone involvement, 616170 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 7, 610951 (3), Autosomal recessive |
| **MGAT2** | 99.99 % | 602616 | Congenital disorder of glycosylation, type IIa, 212066 (3), Autosomal recessive |
| **MGME1** | 99.96 % | 615076 | Mitochondrial DNA depletion syndrome 11, 615084 (3), Autosomal recessive |
| **MGP** | 99.98 % | 154870 | Keutel syndrome, 245150 (3), Autosomal recessive |
| **MIA3** | 99.84 % | 613455 | ?Ondontochondrodysplasia 2 with hearing loss and diabetes, 619269 (3), Autosomal recessive |
| **MICOS13** | 99.75 % | 616658 | Combined oxidative phosphorylation deficiency 37, 618329 (3), Autosomal recessive |
| **MICU1** | 99.69 % | 605084 | Myopathy with extrapyramidal signs, 615673 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **MID1** | 99.87 % | 300552 | Opitz GBBB syndrome, 300000 (3), X-linked recessive |
| **MINAR2** | 99.76 % | 620215 | Deafness, autosomal recessive 120, 620238 (3), Autosomal recessive |
| **MINPP1** | 99.84 % | 605391 | {Thyroid carcinoma, follicular}, 188470 (3), Somatic mutation, Autosomal dominant; Pontocerebellar hypoplasia, type 16, 619527 (3), Autosomal recessive |
| **MIPEP** | 99.93 % | 602241 | Combined oxidative phosphorylation deficiency 31, 617228 (3), Autosomal recessive |
| **MKKS** | 99.99 % | 604896 | McKusick-Kaufman syndrome, 236700 (3), Autosomal recessive; Bardet-Biedl syndrome 6, 605231 (3), Autosomal recessive |
| **MKS1** | 99.62 % | 609883 | Bardet-Biedl syndrome 13, 615990 (3), Autosomal recessive; Meckel syndrome 1, 249000 (3), Autosomal recessive; Joubert syndrome 28, 617121 (3), Autosomal recessive |
| **MLC1** | 99.91 % | 605908 | Megalencephalic leukoencephalopathy with subcortical cysts 1, 604004 (3), Autosomal recessive |
| **MLH1** | 99.69 % | 120436 | Lynch syndrome 2, 609310 (3); Muir-Torre syndrome, 158320 (3), Autosomal dominant; Mismatch repair cancer syndrome 1, 276300 (3), Autosomal recessive |
| **MLIP** | 99.81 % | 614106 | Myopathy with myalgia, increased serum creatine kinase, and with or without episodic rhabdomyolysis, 620138 (3), Autosomal recessive |
| **MLPH** | 99.88 % | 606526 | Griscelli syndrome, type 3, 609227 (3), Autosomal recessive |
| **MLYCD** | 99.82 % | 606761 | Malonyl-CoA decarboxylase deficiency, 248360 (3), Autosomal recessive |
| **MMAA** | 99.8 % | 607481 | Methylmalonic aciduria, vitamin B12-responsive, cblA type, 251100 (3), Autosomal recessive |
| **MMAB** | 99.94 % | 607568 | Methylmalonic aciduria, vitamin B12-responsive, cblB type, 251110 (3), Autosomal recessive |
| **MMACHC** | 99.9 % | 609831 | Methylmalonic aciduria and homocystinuria, cblC type, 277400 (3), Autosomal recessive |
| **MMADHC** | 99.76 % | 611935 | Methylmalonic aciduria and homocystinuria, cblD type, 277410 (3), Autosomal recessive; Methylmalonic aciduria, cblD type, 620953 (3), Autosomal recessive; Homocystinuria-megaloblastic anemia, cblD type, 620952 (3), Autosomal recessive |
| **MME** | 99.79 % | 120520 | ?Spinocerebellar ataxia 43, 617018 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2T, 617017 (3), Autosomal recessive, Autosomal dominant |
| **MMP13** | 99.5 % | 600108 | ?Spondyloepimetaphyseal dysplasia, Missouri type, 602111 (3), Autosomal dominant; Metaphyseal anadysplasia 1, 602111 (3), Autosomal dominant; Metaphyseal dysplasia, Spahr type, 250400 (3), Autosomal recessive |
| **MMP2** | 99.16 % | 120360 | Multicentric osteolysis, nodulosis, and arthropathy, 259600 (3), Autosomal recessive |
| **MMP20** | 99.97 % | 604629 | Amelogenesis imperfecta, type IIA2, 612529 (3), Autosomal recessive |
| **MMP21** | 99.82 % | 608416 | Heterotaxy, visceral, 7, autosomal, 616749 (3), Autosomal recessive |
| **MMUT** | 99.71 % | 609058 | Methylmalonic aciduria, mut(0) type, 251000 (3), Autosomal recessive |
| **MNS1** | 99.9 % | 610766 | Heterotaxy, visceral, 9, autosomal, with male infertility, 618948 (3), Autosomal recessive |
| **MOCOS** | 99.87 % | 613274 | Xanthinuria, type II, 603592 (3), Autosomal recessive |
| **MOCS1** | 99.68 % | 603707 | Molybdenum cofactor deficiency A, 252150 (3), Autosomal recessive |
| **MOCS2** | 99.79 % | 603708 | Molybdenum cofactor deficiency B1, 252160 (3), Autosomal recessive |
| **MOGS** | 99.81 % | 601336 | Congenital disorder of glycosylation, type IIb, 606056 (3), Autosomal recessive |
| **MPC1** | 99.85 % | 614738 | Mitochondrial pyruvate carrier deficiency, 614741 (3), Autosomal recessive |
| **MPDU1** | 99.91 % | 604041 | Congenital disorder of glycosylation, type If, 609180 (3), Autosomal recessive |
| **MPDZ** | 99.73 % | 603785 | Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219 (3), Autosomal recessive |
| **MPI** | 99.73 % | 154550 | Congenital disorder of glycosylation, type Ib, 602579 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **MPIG6B** | 99.96 % | 606520 | ?Thrombocytopenia, anemia, and myelofibrosis, 617441 (3), Autosomal recessive |
| **MPL** | 99.37 % | 159530 | Myelofibrosis with myeloid metaplasia, somatic, 254450 (3); Amegakaryocytic thrombocytopenia, congenital, 1, 604498 (3), Autosomal recessive; Thrombocythemia 2, 601977 (3), Somatic mutation, Autosomal dominant |
| **MPLKIP** | 98.89 % | 609188 | Trichothiodystrophy 4, nonphotosensitive, 234050 (3), Autosomal recessive |
| **MPV17** | 99.85 % | 137960 | Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810 (3), Autosomal recessive |
| **MPZ** | 99.36 % | 159440 | Charcot-Marie-Tooth disease, type 2I, 607677 (3), Autosomal dominant; Dejerine-Sottas disease, 145900 (3), Autosomal recessive, Autosomal dominant; Charcot-Marie-Tooth disease, type 1B, 118200 (3), Autosomal dominant; Roussy-Levy syndrome, 180800 (3), Autosomal dominant; Charcot-Marie-Tooth disease, dominant intermediate D, 607791 (3), Autosomal dominant; Hypomyelinating neuropathy, congenital, 2, 618184 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2J, 607736 (3), Autosomal dominant |
| **MPZL2** | 99.89 % | 604873 | Deafness, autosomal recessive 111, 618145 (3), Autosomal recessive |
| **MRAP** | 99.97 % | 609196 | Glucocorticoid deficiency 2, 607398 (3), Autosomal recessive |
| **MRE11** | 99.93 % | 600814 | Ataxia-telangiectasia-like disorder 1, 604391 (3), Autosomal recessive |
| **MRM2** | 99.99 % | 606906 | Mitochondrial DNA depletion syndrome 17, 618567 (3), Autosomal recessive |
| **MRPL3** | 99.87 % | 607118 | Combined oxidative phosphorylation deficiency 9, 614582 (3), Autosomal recessive |
| **MRPL44** | 99.54 % | 611849 | Combined oxidative phosphorylation deficiency 16, 615395 (3), Autosomal recessive |
| **MRPS16** | 99.43 % | 609204 | Combined oxidative phosphorylation deficiency 2, 610498 (3), Autosomal recessive |
| **MRPS2** | 99.94 % | 611971 | Combined oxidative phosphorylation deficiency 36, 617950 (3), Autosomal recessive |
| **MRPS22** | 99.83 % | 605810 | Ovarian dysgenesis 7, 618117 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 5, 611719 (3), Autosomal recessive |
| **MRPS23** | 99.81 % | 611985 | ?Combined oxidative phosphorylation deficiency 46, 618952 (3), Autosomal recessive |
| **MRPS34** | 99.94 % | 611994 | Combined oxidative phosphorylation deficiency 32, 617664 (3), Autosomal recessive |
| **MSH2** | 99.77 % | 609309 | Lynch syndrome 1, 120435 (3), Autosomal dominant; Muir-Torre syndrome, 158320 (3), Autosomal dominant; Mismatch repair cancer syndrome 2, 619096 (3), Autosomal recessive |
| **MSH3** | 99.43 % | 600887 | Familial adenomatous polyposis 4, 617100 (3), Autosomal recessive; Endometrial carcinoma, somatic, 608089 (3) |
| **MSH6** | 99.86 % | 600678 | Lynch syndrome 5, 614350 (3), Autosomal dominant; Mismatch repair cancer syndrome 3, 619097 (3), Autosomal recessive; {Endometrial cancer, familial}, 608089 (3), Somatic mutation, Autosomal dominant |
| **MSMO1** | 99.89 % | 607545 | Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834 (3), Autosomal recessive |
| **MSN** | 99.62 % | 309845 | Immunodeficiency 50, 300988 (3), X-linked recessive |
| **MSRB3** | 98.94 % | 613719 | Deafness, autosomal recessive 74, 613718 (3), Autosomal recessive |
| **MSTO1** | 68.42 % | 617619 | Myopathy, mitochondrial, and ataxia, 617675 (3), Autosomal recessive, Autosomal dominant |
| **MTFMT** | 99.93 % | 611766 | Combined oxidative phosphorylation deficiency 15, 614947 (3), Autosomal recessive; Mitochondrial complex I deficiency, nuclear type 27, 618248 (3), Autosomal recessive |
| **MTHFD1** | 99.87 % | 172460 | {Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive; Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **MTHFR** | 99.8 % | 607093 | {Vascular disease, susceptibility to} (3); Homocystinuria due to MTHFR deficiency, 236250 (3), Autosomal recessive; {Thromboembolism, susceptibility to}, 188050 (3), Autosomal dominant; {Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant; {Neural tube defects, susceptibility to}, 601634 (3), Autosomal recessive |
| **MTHFS** | 99.4 % | 604197 | Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367 (3), Autosomal recessive |
| **MTM1** | 99.71 % | 300415 | Myopathy, centronuclear, X-linked, 310400 (3), X-linked recessive |
| **MTMR2** | 99.63 % | 603557 | Charcot-Marie-Tooth disease, type 4B1, 601382 (3), Autosomal recessive |
| **MTO1** | 98.56 % | 614667 | Combined oxidative phosphorylation deficiency 10, 614702 (3), Autosomal recessive |
| **MTPAP** | 99.9 % | 613669 | ?Spastic ataxia 4, autosomal recessive, 613672 (3), Autosomal recessive |
| **MTR** | 99.92 % | 156570 | {Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive; Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 (3), Autosomal recessive |
| **MTRFR** | 99.89 % | 613541 | Spastic paraplegia 55, autosomal recessive, 615035 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 7, 613559 (3), Autosomal recessive |
| **MTRR** | 99.94 % | 602568 | Homocystinuria-megaloblastic anemia, cbl E type, 236270 (3), Autosomal recessive; {Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive |
| **MTTP** | 99.82 % | 157147 | Abetalipoproteinemia, 200100 (3), Autosomal recessive |
| **MTX2** | 99.38 % | 608555 | Mandibuloacral dysplasia progeroid syndrome, 619127 (3), Autosomal recessive |
| **MUSK** | 99.79 % | 601296 | Fetal akinesia deformation sequence 1, 208150 (3), Autosomal recessive; Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325 (3), Autosomal recessive |
| **MUTYH** | 99.89 % | 604933 | Adenomas, multiple colorectal, 608456 (3), Autosomal recessive; Gastric cancer, somatic, 613659 (3) |
| **MVK** | 99.8 % | 251170 | Hyper-IgD syndrome, 260920 (3), Autosomal recessive; Porokeratosis 3, multiple types, 175900 (3), Autosomal dominant; Mevalonic aciduria, 610377 (3), Autosomal recessive |
| **MYBPC1** | 99.46 % | 160794 | Congenital myopathy 16, 618524 (3), Autosomal dominant; Lethal congenital contracture syndrome 4, 614915 (3), Autosomal recessive; Arthrogryposis, distal, type 1B, 614335 (3), Autosomal dominant |
| **MYBPC3** | 99.84 % | 600958 | Cardiomyopathy, hypertrophic, 4, 115197 (3), Autosomal recessive, Autosomal dominant; Cardiomyopathy, dilated, 1MM, 615396 (3), Autosomal dominant; Left ventricular noncompaction 10, 615396 (3), Autosomal dominant |
| **MYD88** | 99.89 % | 602170 | Macroglobulinemia, Waldenstrom, somatic, 153600 (3); Immunodeficiency 68, 612260 (3), Autosomal recessive |
| **MYF5** | 99.64 % | 159990 | Ophthalmoplegia, external, with rib and vertebral anomalies, 618155 (3), Autosomal recessive |
| **MYH11** | 99.23 % | 160745 | Megacystis-microcolon-intestinal hypoperistalsis syndrome 2, 619351 (3), Autosomal recessive; Aortic aneurysm, familial thoracic 4, 132900 (3), Autosomal dominant; Visceral myopathy 2, 619350 (3), Autosomal dominant |
| **MYH2** | 99.98 % | 160740 | Congenital myopathy 6 with ophthalmoplegia, 605637 (3), Autosomal recessive, Autosomal dominant |
| **MYH3** | 99.96 % | 160720 | Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A, 178110 (3), Autosomal dominant; Contractures, pterygia, and spondylocarpotarsal fusion syndrome 1B, 618469 (3), Autosomal recessive; Arthrogryposis, distal, type 2B3 (Sheldon-Hall), 618436 (3), Autosomal dominant; Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700 (3), Autosomal dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **MYH7** | 99.83 % | 160760 | Laing distal myopathy, 160500 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 1, 192600 (3), Digenic dominant, Autosomal dominant; Left ventricular noncompaction 5, 613426 (3), Autosomal dominant; Cardiomyopathy, dilated, 1S, 613426 (3), Autosomal dominant; Congenital myopathy 7B, myosin storage, autosomal recessive, 255160 (3), Autosomal recessive; Congenital myopathy 7A, myosin storage, autosomal dominant, 608358 (3), Autosomal dominant |
| **MYL1** | 99.84 % | 160780 | Congenital myopathy 14, 618414 (3), Autosomal recessive |
| **MYL2** | 99.95 % | 160781 | Cardiomyopathy, hypertrophic, 10, 608758 (3), Autosomal dominant; Myopathy, myofibrillar, 12, infantile-onset, with cardiomyopathy, 619424 (3), Autosomal recessive |
| **MYL3** | 99.91 % | 160790 | Cardiomyopathy, hypertrophic, 8, 608751 (3), Autosomal recessive, Autosomal dominant |
| **MYL9** | 99.97 % | 609905 | ?Megacystis-microcolon-intestinal hypoperistalsis syndrome 4, 619365 (3), Autosomal recessive |
| **MYLK** | 99.83 % | 600922 | Megacystis-microcolon-intestinal hypoperistalsis syndrome 1, 249210 (3), Autosomal recessive; Aortic aneurysm, familial thoracic 7, 613780 (3), Autosomal dominant |
| **MYLPF** | 98.49 % | 617378 | Arthrogryposis, distal, type 1C, 619110 (3), Autosomal recessive, Autosomal dominant |
| **MYMK** | 99.37 % | 615345 | Carey-Fineman-Ziter syndrome, 254940 (3), Autosomal recessive |
| **MYMX** | 99.86 % | 619912 | Carey-Fineman-Ziter syndrome 2, 619941 (3), Autosomal recessive |
| **MYO15A** | 99.43 % | 602666 | Deafness, autosomal recessive 3, 600316 (3), Autosomal recessive |
| **MYO18B** | 99.89 % | 607295 | Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549 (3), Autosomal recessive |
| **MYO1E** | 99.95 % | 601479 | Glomerulosclerosis, focal segmental, 6, 614131 (3), Autosomal recessive |
| **MYO3A** | 99.85 % | 606808 | Deafness, autosomal recessive 30, 607101 (3), Autosomal recessive; Deafness, autosomal dominant 90, 620722 (3), Autosomal dominant |
| **MYO5A** | 99.86 % | 160777 | Griscelli syndrome, type 1, 214450 (3), Autosomal recessive |
| **MYO5B** | 99.9 % | 606540 | Diarrhea 2, with microvillus atrophy, with or without cholestasis, 251850 (3), Autosomal recessive; Cholestasis, progressive familial intrahepatic, 10, 619868 (3), Autosomal recessive |
| **MYO6** | 99.63 % | 600970 | Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346 (3), Autosomal dominant; Deafness, autosomal dominant 22, 606346 (3), Autosomal dominant; Deafness, autosomal recessive 37, 607821 (3), Autosomal recessive |
| **MYO7A** | 99.66 % | 276903 | Deafness, autosomal recessive 2, 600060 (3), Autosomal recessive; Usher syndrome, type 1B, 276900 (3), Autosomal recessive; Deafness, autosomal dominant 11, 601317 (3), Autosomal dominant |
| **MYO9A** | 99.85 % | 604875 | Myasthenic syndrome, congenital, 24, presynaptic, 618198 (3), Autosomal recessive |
| **MYOD1** | 99.76 % | 159970 | Congenital myopathy 17, 618975 (3), Autosomal recessive |
| **MYPN** | 99.68 % | 608517 | Cardiomyopathy, hypertrophic, 22, 615248 (3), Autosomal dominant; Congenital myopathy 24, 617336 (3), Autosomal recessive; Cardiomyopathy, familial restrictive, 4, 615248 (3), Autosomal dominant; Cardiomyopathy, dilated, 1KK, 615248 (3), Autosomal dominant |
| **MYSM1** | 95.13 % | 612176 | Bone marrow failure syndrome 4, 618116 (3), Autosomal recessive |
| **MYZAP** | 100 % | 614071 | Cardiomyopathy, dilated, 2K, 620894 (3), Autosomal recessive |
| **NAA10** | 99.94 % | 300013 | Microphthalmia, syndromic 1, 309800 (3), X-linked; Ogden syndrome, 300855 (3), X-linked dominant, X-linked recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **NAA20** | 99.63 % | 610833 | Intellectual developmental disorder, autosomal recessive 73, 619717 (3), Autosomal recessive |
| **NADK2** | 99.85 % | 615787 | 2,4-dienoyl-CoA reductase deficiency, 616034 (3), Autosomal recessive |
| **NADSYN1** | 99.53 % | 608285 | Vertebral, cardiac, renal, and limb defects syndrome 3, 618845 (3), Autosomal recessive |
| **NAE1** | 99.65 % | 603385 | Neurodevelopmental disorder with dysmorphic facies and ischiopubic hypoplasia, 620210 (3), Autosomal recessive |
| **NAGA** | 99.95 % | 104170 | Schindler disease, type I, 609241 (3), Autosomal recessive; Kanzaki disease, 609242 (3), Autosomal recessive; Schindler disease, type III, 609241 (3), Autosomal recessive |
| **NAGLU** | 99.86 % | 609701 | ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 (3), Autosomal dominant; Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 (3), Autosomal recessive |
| **NAGS** | 99.32 % | 608300 | N-acetylglutamate synthase deficiency, 237310 (3), Autosomal recessive |
| **NALCN** | 99.93 % | 611549 | Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 (3), Autosomal dominant; Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419 (3), Autosomal recessive |
| **NANS** | 99.82 % | 605202 | Spondyloepimetaphyseal dysplasia, Genevieve type, 610442 (3), Autosomal recessive |
| **NAPB** | 99.97 % | 611270 | Developmental and epileptic encephalopathy 107, 620033 (3), Autosomal recessive |
| **NARS1** | 99.25 % | 108410 | Neurodevelopmental disorder with microcephaly, impaired language, epilepsy, and gait abnormalities, autosomal dominant, 619092 (3), Autosomal dominant; Neurodevelopmental disorder with microcephaly, impaired language, and gait abnormalities, autosomal recessive, 619091 (3), Autosomal recessive |
| **NARS2** | 99.46 % | 612803 | Combined oxidative phosphorylation deficiency 24, 616239 (3), Autosomal recessive; ?Deafness, autosomal recessive 94, 618434 (3), Autosomal recessive |
| **NAXD** | 99.48 % | 615910 | Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321 (3), Autosomal recessive |
| **NAXE** | 99.25 % | 608862 | Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186 (3), Autosomal recessive |
| **NBAS** | 99.73 % | 608025 | Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 (3), Autosomal recessive; Infantile liver failure syndrome 2, 616483 (3), Autosomal recessive |
| **NBEAL2** | 99.76 % | 614169 | Gray platelet syndrome, 139090 (3), Autosomal recessive |
| **NBN** | 99.79 % | 602667 | Leukemia, acute lymphoblastic, 613065 (3); Aplastic anemia, 609135 (3); Nijmegen breakage syndrome, 251260 (3), Autosomal recessive |
| **NCAPD2** | 99.93 % | 615638 | Microcephaly 21, primary, autosomal recessive, 617983 (3), Autosomal recessive |
| **NCAPD3** | 99.86 % | 609276 | Microcephaly 22, primary, autosomal recessive, 617984 (3), Autosomal recessive |
| **NCF1** | 52.65 % | 608512 | Chronic granulomatous disease 1, autosomal recessive, 233700 (3), Autosomal recessive |
| **NCF2** | 98.87 % | 608515 | Chronic granulomatous disease 2, autosomal recessive, 233710 (3), Autosomal recessive |
| **NCF4** | 99.78 % | 601488 | Chronic granulomatous disease 3, autosomal recessive, 613960 (3), Autosomal recessive |
| **NCKAP1L** | 99.08 % | 141180 | Immunodeficiency 72 with autoinflammation, 618982 (3), Autosomal recessive |
| **NDE1** | 100 % | 609449 | Microhydranencephaly, 605013 (3), Autosomal recessive; Lissencephaly 4 (with microcephaly), 614019 (3), Autosomal recessive |
| **NDP** | 99.93 % | 300658 | Exudative vitreoretinopathy 2, X-linked, 305390 (3), X-linked dominant, X-linked recessive; Norrie disease, 310600 (3), X-linked recessive |
| **NDRG1** | 99.95 % | 605262 | Charcot-Marie-Tooth disease, type 4D, 601455 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **NDST1** | 99.96 % | 600853 | Intellectual developmental disorder, autosomal recessive 46, 616116 (3), Autosomal recessive |
| **NDUFA1** | 99.78 % | 300078 | Mitochondrial complex I deficiency, nuclear type 12, 301020 (3), X-linked recessive |
| **NDUFA10** | 99.92 % | 603835 | Mitochondrial complex I deficiency, nuclear type 22, 618243 (3), Autosomal recessive |
| **NDUFA11** | 99.84 % | 612638 | Mitochondrial complex I deficiency, nuclear type 14, 618236 (3), Autosomal recessive |
| **NDUFA12** | 99.02 % | 614530 | Mitochondrial complex I deficiency, nuclear type 23, 618244 (3), Autosomal recessive |
| **NDUFA13** | 99.91 % | 609435 | {Thyroid carcinoma, Hurthle cell}, 607464 (3); Mitochondrial complex I deficiency, nuclear type 28, 618249 (3), Autosomal recessive |
| **NDUFA2** | 99.85 % | 602137 | Mitochondrial complex I deficiency, nuclear type 13, 618235 (3), Autosomal recessive |
| **NDUFA4** | 100 % | 603833 | ?Mitochondrial complex IV deficiency, nuclear type 21, 619065 (3), Autosomal recessive |
| **NDUFA6** | 99.93 % | 602138 | Mitochondrial complex I deficiency, nuclear type 33, 618253 (3), Autosomal recessive |
| **NDUFA8** | 99.94 % | 603359 | Mitochondrial complex I deficiency, nuclear type 37, 619272 (3), Autosomal recessive |
| **NDUFA9** | 99.97 % | 603834 | Mitochondrial complex I deficiency, nuclear type 26, 618247 (3), Autosomal recessive |
| **NDUFAF1** | 99.99 % | 606934 | Mitochondrial complex I deficiency, nuclear type 11, 618234 (3), Autosomal recessive |
| **NDUFAF2** | 99.96 % | 609653 | Mitochondrial complex I deficiency, nuclear type 10, 618233 (3), Autosomal recessive |
| **NDUFAF3** | 99.85 % | 612911 | Mitochondrial complex I deficiency, nuclear type 18, 618240 (3), Autosomal recessive |
| **NDUFAF4** | 99.85 % | 611776 | Mitochondrial complex I deficiency, nuclear type 15, 618237 (3), Autosomal recessive |
| **NDUFAF5** | 99.87 % | 612360 | Mitochondrial complex I deficiency, nuclear type 16, 618238 (3), Autosomal recessive |
| **NDUFAF6** | 99.67 % | 612392 | Mitochondrial complex I deficiency, nuclear type 17, 618239 (3), Autosomal recessive; Fanconi renotubular syndrome 5, 618913 (3), Autosomal recessive |
| **NDUFAF8** | 98.38 % | 618461 | Mitochondrial complex I deficiency, nuclear type 34, 618776 (3), Autosomal recessive |
| **NDUFB10** | 99.52 % | 603843 | ?Mitochondrial complex I deficiency, nuclear type 35, 619003 (3), Autosomal recessive |
| **NDUFB11** | 98.75 % | 300403 | Linear skin defects with multiple congenital anomalies 3, 300952 (3), X-linked dominant; ?Mitochondrial complex I deficiency, nuclear type 30, 301021 (3), X-linked |
| **NDUFB3** | 99.89 % | 603839 | Mitochondrial complex I deficiency, nuclear type 25, 618246 (3), Autosomal recessive |
| **NDUFB7** | 98.66 % | 603842 | ?Mitochondrial complex I deficiency, nuclear type 39, 620135 (3), Autosomal recessive |
| **NDUFB8** | 99.93 % | 602140 | Mitochondrial complex I deficiency, nuclear type 32, 618252 (3), Autosomal recessive |
| **NDUFC2** | 99.88 % | 603845 | Mitochondrial complex I deficiency, nuclear type 36, 619170 (3), Autosomal recessive |
| **NDUFS1** | 99.56 % | 157655 | Mitochondrial complex I deficiency, nuclear type 5, 618226 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **NDUFS2** | 98.85 % | 602985 | ?Leber-like hereditary optic neuropathy, autosomal recessive 2, 620569 (3), Autosomal recessive; Mitochondrial complex I deficiency, nuclear type 6, 618228 (3), Autosomal recessive |
| **NDUFS3** | 99.73 % | 603846 | Mitochondrial complex I deficiency, nuclear type 8, 618230 (3), Autosomal recessive |
| **NDUFS4** | 99.96 % | 602694 | Mitochondrial complex I deficiency, nuclear type 1, 252010 (3), Autosomal recessive |
| **NDUFS6** | 99.83 % | 603848 | Mitochondrial complex I deficiency, nuclear type 9, 618232 (3), Autosomal recessive |
| **NDUFS7** | 98.5 % | 601825 | Mitochondrial complex I deficiency, nuclear type 3, 618224 (3), Autosomal recessive |
| **NDUFS8** | 99.65 % | 602141 | Mitochondrial complex I deficiency, nuclear type 2, 618222 (3), Autosomal recessive |
| **NDUFV1** | 99.74 % | 161015 | Mitochondrial complex I deficiency, nuclear type 4, 618225 (3), Autosomal recessive |
| **NDUFV2** | 99.87 % | 600532 | Mitochondrial complex I deficiency, nuclear type 7, 618229 (3), Autosomal recessive |
| **NEB** | 87.05 % | 161650 | Nemaline myopathy 2, autosomal recessive, 256030 (3), Autosomal recessive; Arthrogryposis multiplex congenita 6, 619334 (3), Autosomal recessive |
| **NECTIN1** | 99.95 % | 600644 | Cleft lip/palate-ectodermal dysplasia syndrome, 225060 (3), Autosomal recessive; Orofacial cleft 7, 225060 (3), Autosomal recessive |
| **NECTIN4** | 99.17 % | 609607 | Ectodermal dysplasia-syndactyly syndrome 1, 613573 (3), Autosomal recessive |
| **NEFL** | 99.99 % | 162280 | Charcot-Marie-Tooth disease, type 1F, 607734 (3), Autosomal recessive, Autosomal dominant; Charcot-Marie-Tooth disease, dominant intermediate G, 617882 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2E, 607684 (3), Autosomal dominant |
| **NEK1** | 99.87 % | 604588 | Short-rib thoracic dysplasia 6 with or without polydactyly, 263520 (3), Autosomal recessive, Digenic recessive; ?Orofaciodigital syndrome II, 252100 (3), Autosomal recessive; {Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892 (3), Autosomal dominant |
| **NEK10** | 99.75 % | 618726 | Ciliary dyskinesia, primary, 44, 618781 (3), Autosomal recessive |
| **NEK8** | 99.8 % | 609799 | Renal-hepatic-pancreatic dysplasia 2, 615415 (3), Autosomal recessive; Polycystic kidney disease 8, 620903 (3), Autosomal dominant; ?Nephronophthisis 9, 613824 (3) |
| **NEK9** | 99.84 % | 609798 | ?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262 (3), Autosomal recessive; Nevus comedonicus, somatic, 617025 (3); Lethal congenital contracture syndrome 10, 617022 (3), Autosomal recessive |
| **NEMF** | 99.91 % | 608378 | Intellectual developmental disorder with speech delay and axonal peripheral neuropathy, 619099 (3), Autosomal recessive |
| **NEPRO** | 99.47 % | 617089 | Anauxetic dysplasia 3, 618853 (3), Autosomal recessive |
| **NEU1** | 99.94 % | 608272 | Sialidosis, type II, 256550 (3), Autosomal recessive; Sialidosis, type I, 256550 (3), Autosomal recessive |
| **NEUROG3** | 99.96 % | 604882 | Diarrhea 4, malabsorptive, congenital, 610370 (3), Autosomal recessive |
| **NEXMIF** | 99.97 % | 300524 | Intellectual developmental disorder, X-linked 98, 300912 (3), X-linked dominant |
| **NFASC** | 99.55 % | 609145 | Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356 (3), Autosomal recessive |
| **NFS1** | 99.9 % | 603485 | Combined oxidative phosphorylation deficiency 52, 619386 (3), Autosomal recessive |
| **NFU1** | 99.01 % | 608100 | Spastic paraplegia 93, autosomal recessive, 620938 (3), Autosomal recessive; Multiple mitochondrial dysfunctions syndrome 1, 605711 (3), Autosomal recessive |
| **NGF** | 99.95 % | 162030 | Neuropathy, hereditary sensory and autonomic, type V, 608654 (3), Autosomal recessive |
| **NGLY1** | 99.83 % | 610661 | Congenital disorder of deglycosylation 1, 615273 (3), Autosomal recessive |
| **NHEJ1** | 99.61 % | 611290 | Microphthalmia/coloboma 13, 620968 (3), Autosomal recessive; Immunodeficiency 124, severe combined, 611291 (3), Autosomal recessive |
| **NHLRC1** | 99.81 % | 608072 | Myoclonic epilepsy of Lafora 2, 620681 (3), Autosomal recessive |
| **NHLRC2** | 99.65 % | 618277 | FINCA syndrome, 618278 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **NHP2** | 100 % | 606470 | Dyskeratosis congenita, autosomal recessive 2, 613987 (3), Autosomal recessive |
| **NHS** | 99.91 % | 300457 | Cataract 40, X-linked, 302200 (3), X-linked; Nance-Horan syndrome, 302350 (3), X-linked dominant |
| **NIN** | 99.7 % | 608684 | ?Seckel syndrome 7, 614851 (3), Autosomal recessive |
| **NIPAL4** | 99.97 % | 609383 | Ichthyosis, congenital, autosomal recessive 6, 612281 (3), Autosomal recessive |
| **NKAP** | 99.54 % | 300766 | Intellectual developmental disorder, X-linked syndromic, Hackman-Di Donato type, 301039 (3), X-linked recessive |
| **NKX3-2** | 99.62 % | 602183 | Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330 (3), Autosomal recessive |
| **NKX6-2** | 99.96 % | 605955 | Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560 (3), Autosomal recessive |
| **NLGN3** | 99.89 % | 300336 | {Autism susceptibility, X-linked 1}, 300425 (3), X-linked |
| **NLGN4X** | 99.85 % | 300427 | Intellectual developmental disorder, X-linked, 300495 (3), X-linked; {Autism susceptibility, X-linked 2}, 300495 (3), X-linked |
| **NLRP7** | 99.9 % | 609661 | Hydatidiform mole, recurrent, 1, 231090 (3), Autosomal recessive |
| **NME5** | 99.69 % | 603575 | Ciliary dyskinesia, primary, 48, without situs inversus, 620032 (3), Autosomal recessive |
| **NMNAT1** | 99.58 % | 608700 | Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis, 619260 (3), Autosomal recessive; Leber congenital amaurosis 9, 608553 (3), Autosomal recessive |
| **NMNAT2** | 98.92 % | 608701 | *No OMIM phenotypes* |
| **NNT** | 99.79 % | 607878 | Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736 (3), Autosomal recessive |
| **NONO** | 99.89 % | 300084 | Intellectual developmental disorder, X-linked syndromic 34, 300967 (3), X-linked |
| **NOS1AP** | 99.32 % | 605551 | Nephrotic syndrome, type 22, 619155 (3), Autosomal recessive |
| **NPC1** | 99.9 % | 607623 | Niemann-Pick disease, type C1, 257220 (3), Autosomal recessive; Niemann-Pick disease, type D, 257220 (3), Autosomal recessive |
| **NPC2** | 99.76 % | 601015 | Niemann-pick disease, type C2, 607625 (3), Autosomal recessive |
| **NPHP1** | 97.52 % | 607100 | Joubert syndrome 4, 609583 (3), Autosomal recessive; Nephronophthisis 1, juvenile, 256100 (3), Autosomal recessive; Senior-Loken syndrome-1, 266900 (3), Autosomal recessive |
| **NPHP3** | 99.49 % | 608002 | Nephronophthisis 3, 604387 (3), Autosomal recessive; Renal-hepatic-pancreatic dysplasia 1, 208540 (3), Autosomal recessive; Meckel syndrome 7, 267010 (3), Autosomal recessive |
| **NPHP4** | 99.89 % | 607215 | Senior-Loken syndrome 4, 606996 (3), Autosomal recessive; Nephronophthisis 4, 606966 (3), Autosomal recessive |
| **NPHS1** | 99.69 % | 602716 | Nephrotic syndrome, type 1, 256300 (3), Autosomal recessive |
| **NPHS2** | 99.52 % | 604766 | Nephrotic syndrome, type 2, 600995 (3), Autosomal recessive |
| **NPR2** | 99.89 % | 108961 | Epiphyseal chondrodysplasia, Miura type, 615923 (3), Autosomal dominant; Short stature with nonspecific skeletal abnormalities, 616255 (3), Autosomal dominant; Acromesomelic dysplasia 1, Maroteaux type, 602875 (3), Autosomal recessive |
| **NPR3** | 99.96 % | 108962 | Boudin-Mortier syndrome, 619543 (3), Autosomal recessive |
| **NR0B1** | 99.94 % | 300473 | Adrenal hypoplasia, congenital, 300200 (3), X-linked recessive; 46XY sex reversal 2, dosage-sensitive, 300018 (3), X-linked |
| **NR1H4** | 99.36 % | 603826 | Cholestasis, progressive familial intrahepatic, 5, 617049 (3), Autosomal recessive |
| **NR2E3** | 99.88 % | 604485 | Retinitis pigmentosa 37, 611131 (3), Autosomal recessive, Autosomal dominant; Enhanced S-cone syndrome 1, 268100 (3), Autosomal recessive |
| **NRCAM** | 99.71 % | 601581 | Neurodevelopmental disorder with neuromuscular and skeletal abnormalities, 619833 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **NRL** | 99.77 % | 162080 | Enhanced S-cone syndrome 2, 621371 (3), Autosomal recessive; Retinitis pigmentosa 27, 613750 (3), Autosomal dominant |
| **NRROS** | 99.99 % | 615322 | Seizures, early-onset, with neurodegeneration and brain calcification, 618875 (3), Autosomal recessive |
| **NRXN1** | 99.81 % | 600565 | Pitt-Hopkins-like syndrome 2, 614325 (3), Autosomal recessive; {Schizophrenia, susceptibility to, 17}, 621407 (3) |
| **NSDHL** | 99.66 % | 300275 | CK syndrome, 300831 (3), X-linked recessive; CHILD syndrome, 308050 (3), X-linked dominant |
| **NSMCE3** | 99.45 % | 608243 | Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241 (3), Autosomal recessive |
| **NSRP1** | 99.89 % | 616173 | Neurodevelopmental disorder with spasticity, seizures, and brain abnormalities, 620001 (3), Autosomal recessive |
| **NSUN2** | 99.88 % | 610916 | Intellectual developmental disorder, autosomal recessive 5, 611091 (3), Autosomal recessive |
| **NSUN3** | 99.88 % | 617491 | Combined oxidative phosphorylation deficiency 48, 619012 (3), Autosomal recessive |
| **NSUN6** | 99.82 % | 617199 | Intellectual developmental disorder, autosomal recessive 82, 620779 (3), Autosomal recessive |
| **NT5C2** | 99.86 % | 600417 | Spastic paraplegia 45, autosomal recessive, 613162 (3), Autosomal recessive |
| **NT5C3A** | 99.64 % | 606224 | Anemia, congenital, nonspherocytic hemolytic, 8, 266120 (3), Autosomal recessive |
| **NT5E** | 99.61 % | 129190 | Calcification of joints and arteries, 211800 (3), Autosomal recessive |
| **NTHL1** | 99.71 % | 602656 | Familial adenomatous polyposis 3, 616415 (3), Autosomal recessive |
| **NTNG2** | 99.69 % | 618689 | Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718 (3), Autosomal recessive |
| **NTRK1** | 99.31 % | 191315 | Insensitivity to pain, congenital, with anhidrosis, 256800 (3), Autosomal recessive |
| **NUBPL** | 99.77 % | 613621 | Mitochondrial complex I deficiency, nuclear type 21, 618242 (3), Autosomal recessive |
| **NUDCD3** | 99.33 % | 610296 | *No OMIM phenotypes* |
| **NUDT2** | 99.92 % | 602852 | Intellectual developmental disorder with or without peripheral neuropathy, 619844 (3), Autosomal recessive |
| **NUP107** | 98.89 % | 607617 | ?Ovarian dysgenesis 6, 618078 (3), Autosomal recessive; Galloway-Mowat syndrome 7, 618348 (3), Autosomal recessive; Nephrotic syndrome, type 11, 616730 (3), Autosomal recessive |
| **NUP133** | 99.73 % | 607613 | ?Galloway-Mowat syndrome 8, 618349 (3), Autosomal recessive; Nephrotic syndrome, type 18, 618177 (3), Autosomal recessive |
| **NUP188** | 99.6 % | 615587 | Sandestig-Stefanova syndrome, 618804 (3), Autosomal recessive |
| **NUP214** | 99.93 % | 114350 | Leukemia, T-cell acute lymphoblastic, somatic, 613065 (3); Leukemia, acute myeloid, somatic, 601626 (3); {Encephalopathy, acute, infection-induced, susceptibility to, 9}, 618426 (3), Autosomal recessive |
| **NUP62** | 100 % | 605815 | Striatonigral degeneration, infantile, 271930 (3), Autosomal recessive |
| **NUP85** | 99.7 % | 170285 | Nephrotic syndrome, type 17, 618176 (3), Autosomal recessive |
| **NUP88** | 99.83 % | 602552 | Fetal akinesia deformation sequence 4, 618393 (3), Autosomal recessive |
| **NUP93** | 99.21 % | 614351 | Nephrotic syndrome, type 12, 616892 (3), Autosomal recessive |
| **NXN** | 99.94 % | 612895 | Robinow syndrome, autosomal recessive 2, 618529 (3), Autosomal recessive |
| **NYX** | 99.78 % | 300278 | Night blindness, congenital stationary (complete), 1A, X-linked, 310500 (3), X-linked recessive |
| **OAT** | 99.79 % | 613349 | Gyrate atrophy of choroid and retina with or without ornithinemia, 258870 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **OBSCN** | 99.84 % | 608616 | {Rhabdomyolysis, susceptibility to, 1}, 620235 (3), Autosomal recessive |
| **OBSL1** | 99.88 % | 610991 | 3-M syndrome 2, 612921 (3), Autosomal recessive |
| **OCA2** | 97.93 % | 611409 | [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 (3), Autosomal recessive; [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 (3), Autosomal recessive; Albinism, brown oculocutaneous, 203200 (3), Autosomal recessive; Albinism, oculocutaneous, type II, 203200 (3), Autosomal recessive |
| **OCLN** | 81.1 % | 602876 | Pseudo-TORCH syndrome 1, 251290 (3), Autosomal recessive |
| **OCRL** | 99.78 % | 300535 | Dent disease 2, 300555 (3), X-linked recessive; Lowe syndrome, 309000 (3), X-linked recessive |
| **ODAD1** | 99.76 % | 615038 | Ciliary dyskinesia, primary, 20, 615067 (3), Autosomal recessive |
| **ODAD2** | 98.01 % | 615408 | Ciliary dyskinesia, primary, 23, 615451 (3), Autosomal recessive |
| **ODAD3** | 99.65 % | 615956 | Ciliary dyskinesia, primary, 30, 616037 (3), Autosomal recessive |
| **ODAD4** | 99.5 % | 617095 | Ciliary dyskinesia, primary, 35, 617092 (3), Autosomal recessive |
| **ODAPH** | 99.87 % | 614829 | Amelogenesis imperfecta, type IIA4, 614832 (3), Autosomal recessive |
| **OFD1** | 99.71 % | 300170 | Simpson-Golabi-Behmel syndrome, type 2, 300209 (3), X-linked recessive; ?Retinitis pigmentosa 23, 300424 (3), X-linked recessive; Orofaciodigital syndrome I, 311200 (3), X-linked dominant; Joubert syndrome 10, 300804 (3), X-linked recessive |
| **OGDHL** | 99.47 % | 617513 | *No OMIM phenotypes* |
| **OGT** | 99.77 % | 300255 | Intellectual developmental disorder, X-linked 106, 300997 (3), X-linked recessive |
| **OPA1** | 99.94 % | 605290 | Optic atrophy plus syndrome, 125250 (3), Autosomal dominant; {Glaucoma, normal tension, susceptibility to}, 606657 (3); Optic atrophy 1, 165500 (3), Autosomal dominant; Behr syndrome, 210000 (3), Autosomal recessive; ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896 (3), Autosomal recessive |
| **OPA3** | 99.96 % | 606580 | 3-methylglutaconic aciduria, type III, 258501 (3), Autosomal recessive; Optic atrophy 3 with cataract, 165300 (3), Autosomal dominant |
| **OPHN1** | 99.73 % | 300127 | Intellectual developmental disorder, X-linked syndromic, Billuart type, 300486 (3), X-linked recessive |
| **OPN1LW** | 71.07 % | 300822 | Blue cone monochromacy, 303700 (3), X-linked recessive; Colorblindness, protan, 303900 (3), X-linked |
| **OPN1MW** | 28.98 % | 300821 | Colorblindness, deutan, 303800 (3), X-linked; Blue cone monochromacy, 303700 (3), X-linked recessive |
| **OPTN** | 99.93 % | 602432 | Glaucoma 1, open angle, E, 137760 (3), Autosomal dominant; Amyotrophic lateral sclerosis 12 with or without frontotemporal dementia, 613435 (3), Autosomal recessive, Autosomal dominant; {Glaucoma, normal tension, susceptibility to}, 606657 (3) |
| **ORAI1** | 99.4 % | 610277 | Immunodeficiency 9, 612782 (3), Autosomal recessive; Myopathy, tubular aggregate, 2, 615883 (3), Autosomal dominant |
| **ORC1** | 98.12 % | 601902 | Meier-Gorlin syndrome 1, 224690 (3), Autosomal recessive |
| **ORC4** | 99.59 % | 603056 | Meier-Gorlin syndrome 2, 613800 (3), Autosomal recessive |
| **ORC6** | 99.1 % | 607213 | Meier-Gorlin syndrome 3, 613803 (3), Autosomal recessive |
| **OSGEP** | 99.94 % | 610107 | Galloway-Mowat syndrome 3, 617729 (3), Autosomal recessive |
| **OSTM1** | 99.58 % | 607649 | Osteopetrosis, autosomal recessive 5, 259720 (3), Autosomal recessive |
| **OTC** | 99.67 % | 300461 | Ornithine transcarbamylase deficiency, 311250 (3), X-linked |
| **OTOA** | 75.1 % | 607038 | Deafness, autosomal recessive 22, 607039 (3), Autosomal recessive |
| **OTOF** | 99.8 % | 603681 | Auditory neuropathy, autosomal recessive, 1, 601071 (3), Autosomal recessive; Deafness, autosomal recessive 9, 601071 (3), Autosomal recessive |
| **OTOG** | 99.74 % | 604487 | Deafness, autosomal recessive 18B, 614945 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **OTOGL** | 98.71 % | 614925 | Deafness, autosomal recessive 84B, 614944 (3), Autosomal recessive |
| **OTUD5** | 99.59 % | 300713 | Multiple congenital anomalies-neurodevelopmental syndrome, X-linked, 301056 (3), X-linked recessive |
| **OTUD6B** | 99.85 % | 612021 | Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452 (3), Autosomal recessive |
| **OTUD7A** | 98.44 % | 612024 | Neurodevelopmental disorder with hypotonia and seizures, 620790 (3), Autosomal recessive |
| **OTULIN** | 99.66 % | 615712 | Autoinflammation, panniculitis, and dermatosis syndrome, autosomal recessive, 617099 (3), Autosomal recessive; {Immunodeficiency 107, susceptibility to invasive staphylococcus aureus infection}, 619986 (3), Autosomal dominant; Autoinflammation, panniculitis, and dermatosis syndrome, autosomal dominant, 621030 (3), Autosomal dominant |
| **OXCT1** | 99.17 % | 601424 | Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050 (3), Autosomal recessive |
| **OXR1** | 99.87 % | 605609 | Cerebellar hypoplasia/atrophy, epilepsy, and global developmental delay, 213000 (3), Autosomal recessive |
| **P2RY12** | 100 % | 600515 | Bleeding disorder, platelet-type, 8, 609821 (3), Autosomal recessive |
| **P3H1** | 99.36 % | 610339 | Osteogenesis imperfecta, type VIII, 610915 (3), Autosomal recessive |
| **P3H2** | 99.84 % | 610341 | Myopia, high, with cataract and vitreoretinal degeneration, 614292 (3), Autosomal recessive |
| **P4HTM** | 99.94 % | 614584 | Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493 (3), Autosomal recessive |
| **PAH** | 99.79 % | 612349 | [Hyperphenylalaninemia, non-PKU mild], 261600 (3), Autosomal recessive; Phenylketonuria, 261600 (3), Autosomal recessive |
| **PAK3** | 99.76 % | 300142 | Intellectual developmental disorder, X-linked 30, 300558 (3), X-linked recessive |
| **PAM16** | 99.81 % | 614336 | Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320 (3), Autosomal recessive |
| **PAN2** | 99.43 % | 617447 | Developmental delay with variable cardiac and renal congenital anomalies and dysmorphic facies, 621384 (3), Autosomal recessive |
| **PANK2** | 99.86 % | 606157 | Neurodegeneration with brain iron accumulation 1, 234200 (3), Autosomal recessive |
| **PAPPA2** | 99.54 % | 619485 | Short stature, Dauber-Argente type, 619489 (3), Autosomal recessive |
| **PAPSS2** | 99.86 % | 603005 | Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847 (3), Autosomal recessive |
| **PARN** | 99.09 % | 604212 | Dyskeratosis congenita, autosomal recessive 6, 616353 (3), Autosomal recessive; Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 4, 616371 (3), Autosomal dominant |
| **PARS2** | 99.97 % | 612036 | Developmental and epileptic encephalopathy 75, 618437 (3), Autosomal recessive |
| **PAX1** | 99.8 % | 167411 | Otofaciocervical syndrome 2 with T-cell deficiency, 615560 (3), Autosomal recessive |
| **PAX3** | 99.62 % | 606597 | Craniofacial-deafness-hand syndrome, 122880 (3), Autosomal dominant; Waardenburg syndrome, type 3, 148820 (3), Autosomal recessive, Autosomal dominant; Waardenburg syndrome, type 1, 193500 (3), Autosomal dominant; Rhabdomyosarcoma 2, alveolar, 268220 (3), Somatic mutation |
| **PAX7** | 99.3 % | 167410 | Congenital myopathy 19, 618578 (3), Autosomal recessive; Rhabdomyosarcoma 2, alveolar, 268220 (3), Somatic mutation |
| **PC** | 99.85 % | 608786 | Pyruvate carboxylase deficiency, 266150 (3), Autosomal recessive |
| **PCARE** | 99.99 % | 613425 | Retinitis pigmentosa 54, 613428 (3), Autosomal recessive |
| **PCBD1** | 98.89 % | 126090 | Hyperphenylalaninemia, BH4-deficient, D, 264070 (3), Autosomal recessive |
| **PCCA** | 99.91 % | 232000 | Propionicacidemia, 606054 (3), Autosomal recessive |
| **PCCB** | 99.86 % | 232050 | Propionicacidemia, 606054 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **PCDH12** | 99.97 % | 605622 | Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280 (3), Autosomal recessive |
| **PCDH15** | 99.32 % | 605514 | Usher syndrome, type 1D/F digenic, 601067 (3), Autosomal recessive, Digenic recessive; Deafness, autosomal recessive 23, 609533 (3), Autosomal recessive; Usher syndrome, type 1F, 602083 (3), Autosomal recessive |
| **PCDH19** | 99.91 % | 300460 | Developmental and epileptic encephalopathy 9, 300088 (3), X-linked |
| **PCDHGC4** | 99.53 % | 606305 | Neurodevelopmental disorder with poor growth and skeletal anomalies, 619880 (3), Autosomal recessive |
| **PCK1** | 99.96 % | 614168 | Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680 (3), Autosomal recessive |
| **PCLO** | 99.55 % | 604918 | Pontocerebellar hypoplasia, type 3, 608027 (3), Autosomal recessive |
| **PCNT** | 99.91 % | 605925 | Microcephalic osteodysplastic primordial dwarfism, type II, 210720 (3), Autosomal recessive |
| **PCSK1** | 99.92 % | 162150 | {Obesity, susceptibility to, BMIQ12}, 612362 (3); Endocrinopathy due to proprotein convertase 1/3 deficiency, 600955 (3), Autosomal recessive |
| **PCYT1A** | 99.93 % | 123695 | Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940 (3), Autosomal recessive; Lipodystrophy, congenital generalized, type 5, 620680 (3), Autosomal recessive |
| **PCYT2** | 99.78 % | 602679 | Spastic paraplegia 82, autosomal recessive, 618770 (3), Autosomal recessive |
| **PDCD6IP** | 99.83 % | 608074 | ?Microcephaly 29, primary, autosomal recessive, 620047 (3), Autosomal recessive |
| **PDE10A** | 99.74 % | 610652 | Striatal degeneration, autosomal dominant, 616922 (3), Autosomal dominant; Dyskinesia, limb and orofacial, infantile-onset, 616921 (3), Autosomal recessive |
| **PDE2A** | 99.7 % | 602658 | Intellectual developmental disorder with paroxysmal dyskinesia or seizures, 619150 (3), Autosomal recessive |
| **PDE6A** | 99.9 % | 180071 | Retinitis pigmentosa 43, 613810 (3), Autosomal recessive |
| **PDE6B** | 99.97 % | 180072 | Retinitis pigmentosa-40, 613801 (3), Autosomal recessive; Night blindness, congenital stationary, autosomal dominant 2, 163500 (3), Autosomal dominant |
| **PDE6C** | 99.89 % | 600827 | Cone dystrophy 4, 613093 (3), Autosomal recessive |
| **PDE6D** | 99.44 % | 602676 | Joubert syndrome 22, 615665 (3), Autosomal recessive |
| **PDE6G** | 100 % | 180073 | Retinitis pigmentosa 57, 613582 (3), Autosomal recessive |
| **PDE6H** | 99.97 % | 601190 | Achromatopsia 6, 610024 (3), Autosomal recessive |
| **PDHA1** | 99.86 % | 300502 | Pyruvate dehydrogenase E1-alpha deficiency, 312170 (3), X-linked dominant |
| **PDHB** | 99.66 % | 179060 | Pyruvate dehydrogenase E1-beta deficiency, 614111 (3), Autosomal recessive |
| **PDHX** | 99.54 % | 608769 | Lacticacidemia due to PDX1 deficiency, 245349 (3), Autosomal recessive |
| **PDK3** | 99.62 % | 300906 | ?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905 (3), X-linked dominant |
| **PDP1** | 99.99 % | 605993 | Pyruvate dehydrogenase phosphatase deficiency, 608782 (3), Autosomal recessive |
| **PDSS1** | 98.29 % | 607429 | Coenzyme Q10 deficiency, primary, 2, 614651 (3), Autosomal recessive |
| **PDSS2** | 99.8 % | 610564 | Coenzyme Q10 deficiency, primary, 3, 614652 (3), Autosomal recessive |
| **PDX1** | 99.95 % | 600733 | {Diabetes mellitus, type II, susceptibility to}, 125853 (3), Autosomal dominant; Pancreatic agenesis 1, 260370 (3), Autosomal recessive; MODY, type IV, 606392 (3) |
| **PDXK** | 99.35 % | 179020 | Neuropathy, hereditary motor and sensory, type VIC, with optic atrophy, 618511 (3), Autosomal recessive |
| **PDZD7** | 99.58 % | 612971 | Deafness, autosomal recessive 57, 618003 (3), Autosomal recessive; {Retinal disease in Usher syndrome type IIA, modifier of}, 276901 (3), Autosomal recessive; Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472 (3), Digenic dominant, Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **PDZD8** | 99.92 % | 614235 | Intellectual developmental disorder with autism and dysmorphic facies, 620021 (3), Autosomal recessive |
| **PEPD** | 99.42 % | 613230 | Prolidase deficiency, 170100 (3), Autosomal recessive |
| **PERP** | 99.9 % | 609301 | Erythrokeratodermia variabilis et progressiva 7, 619209 (3), Autosomal recessive; Olmsted syndrome 2, 619208 (3), Autosomal dominant |
| **PET100** | 100 % | 614770 | Mitochondrial complex IV deficiency, nuclear type 12, 619055 (3), Autosomal recessive |
| **PEX1** | 98.36 % | 602136 | Heimler syndrome 1, 234580 (3), Autosomal recessive; Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 (3), Autosomal recessive; Peroxisome biogenesis disorder 1A (Zellweger), 214100 (3), Autosomal recessive |
| **PEX10** | 99.94 % | 602859 | Peroxisome biogenesis disorder 6A (Zellweger), 614870 (3), Autosomal recessive; Peroxisome biogenesis disorder 6B, 614871 (3), Autosomal recessive |
| **PEX11B** | 98.66 % | 603867 | Peroxisome biogenesis disorder 14B, 614920 (3), Autosomal recessive |
| **PEX12** | 99.94 % | 601758 | Peroxisome biogenesis disorder 3B, 266510 (3), Autosomal recessive; Peroxisome biogenesis disorder 3A (Zellweger), 614859 (3), Autosomal recessive |
| **PEX13** | 99.73 % | 601789 | Peroxisome biogenesis disorder 11A (Zellweger), 614883 (3), Autosomal recessive; Peroxisome biogenesis disorder 11B, 614885 (3), Autosomal recessive |
| **PEX14** | 99.89 % | 601791 | Peroxisome biogenesis disorder 13A (Zellweger), 614887 (3), Autosomal recessive |
| **PEX16** | 99.55 % | 603360 | Peroxisome biogenesis disorder 8B, 614877 (3), Autosomal recessive; Peroxisome biogenesis disorder 8A (Zellweger), 614876 (3), Autosomal recessive |
| **PEX19** | 98.91 % | 600279 | Peroxisome biogenesis disorder 12A (Zellweger), 614886 (3), Autosomal recessive |
| **PEX2** | 100 % | 170993 | Peroxisome biogenesis disorder 5A (Zellweger), 614866 (3), Autosomal recessive; Peroxisome biogenesis disorder 5B, 614867 (3), Autosomal recessive |
| **PEX26** | 99.99 % | 608666 | Peroxisome biogenesis disorder 7B, 614873 (3), Autosomal recessive; Peroxisome biogenesis disorder 7A (Zellweger), 614872 (3), Autosomal recessive |
| **PEX3** | 99.56 % | 603164 | Peroxisome biogenesis disorder 10A (Zellweger), 614882 (3), Autosomal recessive; ?Peroxisome biogenesis disorder 10B, 617370 (3), Autosomal recessive |
| **PEX5** | 99.93 % | 600414 | Peroxisome biogenesis disorder 2B, 202370 (3), Autosomal recessive; Peroxisome biogenesis disorder 2A (Zellweger), 214110 (3), Autosomal recessive; Rhizomelic chondrodysplasia punctata, type 5, 616716 (3), Autosomal recessive |
| **PEX6** | 99.89 % | 601498 | Peroxisome biogenesis disorder 4B, 614863 (3), Autosomal recessive, Autosomal dominant; Peroxisome biogenesis disorder 4A (Zellweger), 614862 (3), Autosomal recessive; Heimler syndrome 2, 616617 (3), Autosomal recessive |
| **PEX7** | 99.68 % | 601757 | Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive; Peroxisome biogenesis disorder 9B, 614879 (3), Autosomal recessive |
| **PFKM** | 99.29 % | 610681 | Glycogen storage disease VII, 232800 (3), Autosomal recessive |
| **PGAM2** | 99.99 % | 612931 | Glycogen storage disease X, 261670 (3), Autosomal recessive |
| **PGAP1** | 99.48 % | 611655 | Neurodevelopmental disorder with dysmorphic features, spasticity, and brain abnormalities, 615802 (3), Autosomal recessive |
| **PGAP2** | 99.91 % | 615187 | Hyperphosphatasia with impaired intellectual development syndrome 3, 614207 (3), Autosomal recessive |
| **PGAP3** | 99.25 % | 611801 | Hyperphosphatasia with impaired intellectual development syndrome 4, 615716 (3), Autosomal recessive |
| **PGK1** | 99.85 % | 311800 | Phosphoglycerate kinase 1 deficiency, 300653 (3), X-linked recessive |
| **PGM1** | 97.39 % | 171900 | Congenital disorder of glycosylation, type It, 614921 (3), Autosomal recessive |
| **PGM2L1** | 99.71 % | 611610 | Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skin abnormalities, 620191 (3), Autosomal recessive |
| **PGM3** | 99.78 % | 172100 | Immunodeficiency 23, 615816 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **PHEX** | 99.73 % | 300550 | Hypophosphatemic rickets, X-linked dominant, 307800 (3), X-linked dominant |
| **PHF6** | 99.38 % | 300414 | Borjeson-Forssman-Lehmann syndrome, 301900 (3), X-linked recessive |
| **PHF8** | 99.68 % | 300560 | Intellectual developmental disorder, X-linked syndromic, Siderius type, 300263 (3), X-linked recessive |
| **PHGDH** | 99.34 % | 606879 | Neu-Laxova syndrome 1, 256520 (3), Autosomal recessive; Phosphoglycerate dehydrogenase deficiency, 601815 (3), Autosomal recessive |
| **PHKA1** | 99.71 % | 311870 | Muscle glycogenosis, 300559 (3), X-linked recessive |
| **PHKA2** | 99.73 % | 300798 | Glycogen storage disease, type IXa2, 306000 (3), X-linked recessive; Glycogen storage disease, type IXa1, 306000 (3), X-linked recessive |
| **PHKB** | 99.47 % | 172490 | Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750 (3), Autosomal recessive |
| **PHKG2** | 99.93 % | 172471 | Glycogen storage disease IXc, 613027 (3), Autosomal recessive |
| **PHOX2A** | 99.11 % | 602753 | Fibrosis of extraocular muscles, congenital, 2, 602078 (3), Autosomal recessive |
| **PHYH** | 99.98 % | 602026 | Refsum disease, 266500 (3), Autosomal recessive |
| **PI4K2A** | 99.33 % | 609763 | Neurodevelopmental disorder with hyperkinetic movements, seizures and structural brain abnormalities, 620732 (3), Autosomal recessive |
| **PI4KA** | 99.17 % | 600286 | Spastic paraplegia 84, autosomal recessive, 619621 (3), Autosomal recessive; Gastrointestinal defects and immunodeficiency syndrome 2, 619708 (3), Autosomal recessive; Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531 (3), Autosomal recessive |
| **PIBF1** | 99.91 % | 607532 | Joubert syndrome 33, 617767 (3), Autosomal recessive |
| **PIDD1** | 99.83 % | 605247 | Intellectual developmental disorder, autosomal recessive 75, with neuropsychiatric features and variant lissencephaly, 619827 (3), Autosomal recessive |
| **PIEZO1** | 99.83 % | 611184 | [ER blood group system], 620207 (3), Autosomal recessive; Lymphatic malformation 6, 616843 (3), Autosomal recessive; Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380 (3), Autosomal dominant |
| **PIEZO2** | 99.67 % | 613629 | Arthrogryposis, distal, type 5, 108145 (3), Autosomal dominant; Arthrogryposis, distal, with impaired proprioception and touch, 617146 (3), Autosomal recessive; Arthrogryposis, distal, type 3, 114300 (3), Autosomal dominant; ?Marden-Walker syndrome, 248700 (3), Autosomal dominant |
| **PIGA** | 99.57 % | 311770 | Paroxysmal nocturnal hemoglobinuria, somatic, 300818 (3); Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 (3), X-linked recessive; Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072 (3), X-linked recessive |
| **PIGB** | 99.91 % | 604122 | Developmental and epileptic encephalopathy 80, 618580 (3), Autosomal recessive |
| **PIGC** | 99.94 % | 601730 | Glycosylphosphatidylinositol biosynthesis defect 16, 617816 (3), Autosomal recessive |
| **PIGG** | 99.92 % | 616918 | [Blood group, EMM system], 619812 (3), Autosomal recessive; Neurodevelopmental disorder with or without hypotonia, seizures, and cerebellar atrophy, 616917 (3), Autosomal recessive |
| **PIGH** | 99.93 % | 600154 | Glycosylphosphatidylinositol biosynthesis defect 17, 618010 (3), Autosomal recessive |
| **PIGK** | 92.32 % | 605087 | Neurodevelopmental disorder with hypotonia and cerebellar atrophy, with or without seizures, 618879 (3), Autosomal recessive |
| **PIGL** | 99.79 % | 605947 | CHIME syndrome, 280000 (3), Autosomal recessive |
| **PIGM** | 99.57 % | 610273 | Glycosylphosphatidylinositol deficiency, 610293 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **PIGN** | 99.86 % | 606097 | Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080 (3), Autosomal recessive |
| **PIGO** | 99.96 % | 614730 | Hyperphosphatasia with impaired intellectual development syndrome 2, 614749 (3), Autosomal recessive |
| **PIGP** | 99.7 % | 605938 | Developmental and epileptic encephalopathy 55, 617599 (3), Autosomal recessive |
| **PIGQ** | 99.88 % | 605754 | Multiple congenital anomalies-hypotonia-seizures syndrome 4, 618548 (3), Autosomal recessive |
| **PIGS** | 99.83 % | 610271 | Developmental and epileptic encephalopathy 95, 618143 (3), Autosomal recessive |
| **PIGT** | 99.83 % | 610272 | ?Paroxysmal nocturnal hemoglobinuria 2, 615399 (3), Somatic mutation, Autosomal dominant; Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 (3), Autosomal recessive |
| **PIGU** | 99.96 % | 608528 | Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis, 618590 (3), Autosomal recessive |
| **PIGV** | 99.98 % | 610274 | Hyperphosphatasia with impaired intellectual development syndrome 1, 239300 (3), Autosomal recessive |
| **PIGW** | 99.94 % | 610275 | Glycosylphosphatidylinositol biosynthesis defect 11, 616025 (3), Autosomal recessive |
| **PIK3C2A** | 99.81 % | 603601 | Oculoskeletodental syndrome, 618440 (3), Autosomal recessive |
| **PIK3CD** | 99.86 % | 602839 | Immunodeficiency 14A, autosomal dominant, 615513 (3), Autosomal dominant; Immunodeficiency 14B, autosomal recessive, 619281 (3), Autosomal recessive; ?Roifman-Chitayat syndrome, digenic, 613328 (3), Digenic recessive |
| **PIK3CG** | 99.43 % | 601232 | Immunodeficiency 97 with autoinflammation, 619802 (3), Autosomal recessive |
| **PINK1** | 98.62 % | 608309 | Parkinson disease 6, early onset, 605909 (3), Autosomal recessive |
| **PIP5K1C** | 99.81 % | 606102 | Lethal congenital contractural syndrome 3, 611369 (3), Autosomal recessive |
| **PISD** | 99.94 % | 612770 | Liberfarb syndrome, 618889 (3), Autosomal recessive |
| **PITRM1** | 99.81 % | 618211 | Spinocerebellar ataxia, autosomal recessive 30, 619405 (3), Autosomal recessive |
| **PITX3** | 99.89 % | 602669 | Cataract 11, multiple types, 610623 (3), Autosomal recessive, Autosomal dominant; Anterior segment dysgenesis 1, multiple subtypes, 107250 (3), Autosomal dominant; Cataract 11, syndromic, autosomal recessive, 610623 (3), Autosomal recessive, Autosomal dominant |
| **PJVK** | 99.66 % | 610219 | Deafness, autosomal recessive 59, 610220 (3), Autosomal recessive |
| **PKD1L1** | 99.29 % | 609721 | Heterotaxy, visceral, 8, autosomal, 617205 (3), Autosomal recessive |
| **PKDCC** | 99.25 % | 614150 | Rhizomelic limb shortening with dysmorphic features, 618821 (3), Autosomal recessive |
| **PKHD1** | 99.85 % | 606702 | Polycystic kidney disease 4, with or without hepatic disease, 263200 (3), Autosomal recessive |
| **PKLR** | 99.68 % | 609712 | Anemia, congenital, nonspherocytic hemolytic, 2, pyruvate kinase deficient, 266200 (3), Autosomal recessive; [Adenosine triphosphate, elevated, of erythrocytes], 102900 (3), Autosomal dominant |
| **PKP1** | 99.71 % | 601975 | Ectodermal dysplasia/skin fragility syndrome, 604536 (3), Autosomal recessive |
| **PLA2G4A** | 98.81 % | 600522 | Gastrointestinal ulceration, recurrent, with dysfunctional platelets, 618372 (3), Autosomal recessive |
| **PLA2G6** | 99.79 % | 603604 | Parkinson disease 14, autosomal recessive, 612953 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 2B, 610217 (3), Autosomal recessive; Infantile neuroaxonal dystrophy 1, 256600 (3), Autosomal recessive |
| **PLAA** | 99.59 % | 603873 | Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527 (3), Autosomal recessive |
| **PLCB1** | 99.78 % | 607120 | Developmental and epileptic encephalopathy 12, 613722 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **PLCB4** | 99.95 % | 600810 | Auriculocondylar syndrome 2B, 620458 (3), Autosomal recessive; Auriculocondylar syndrome 2A, 614669 (3), Autosomal dominant |
| **PLCE1** | 99.87 % | 608414 | Nephrotic syndrome, type 3, 610725 (3), Autosomal recessive |
| **PLD1** | 99.72 % | 602382 | Cardiac valvular dysplasia 1, 212093 (3), Autosomal recessive |
| **PLEC** | 99.95 % | 601282 | ?Epidermolysis bullosa simplex 5D, generalized intermediate, autosomal recessive, 616487 (3), Autosomal recessive; Epidermolysis bullosa simplex 5B, with muscular dystrophy, 226670 (3), Autosomal recessive; Epidermolysis bullosa simplex 5C, with pyloric atresia, 612138 (3), Autosomal recessive; Epidermolysis bullosa simplex 5A, Ogna type, 131950 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723 (3), Autosomal recessive |
| **PLEKHG2** | 99.87 % | 611893 | Leukodystrophy and acquired microcephaly with or without dystonia, 616763 (3), Autosomal recessive |
| **PLEKHG5** | 99.89 % | 611101 | Neuronopathy, distal hereditary motor, autosomal recessive 4, 611067 (3), Autosomal recessive; Charcot-Marie-Tooth disease, recessive intermediate C, 615376 (3), Autosomal recessive |
| **PLEKHM1** | 99.02 % | 611466 | ?Osteopetrosis, autosomal recessive 6, 611497 (3), Autosomal recessive; Osteopetrosis, autosomal dominant 3, 618107 (3), Autosomal dominant |
| **PLG** | 99.61 % | 173350 | Dysplasminogenemia, 217090 (3), Autosomal recessive; Angioedema, hereditary, 4, 619360 (3), Autosomal dominant; Plasminogen deficiency, type I, 217090 (3), Autosomal recessive |
| **PLK4** | 99.71 % | 605031 | Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171 (3), Autosomal recessive |
| **PLOD1** | 98.95 % | 153454 | Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400 (3), Autosomal recessive |
| **PLOD2** | 99.59 % | 601865 | Bruck syndrome 2, 609220 (3), Autosomal recessive |
| **PLOD3** | 98.22 % | 603066 | BCARD syndrome (lysyl hydroxylase 3 deficiency), 612394 (3), Autosomal recessive |
| **PLP1** | 99.93 % | 300401 | Pelizaeus-Merzbacher disease, 312080 (3), X-linked recessive; Spastic paraplegia 2, X-linked, 312920 (3), X-linked recessive |
| **PLPBP** | 99.56 % | 604436 | Epilepsy, early-onset, 1, vitamin B6-dependent, 617290 (3), Autosomal recessive |
| **PLS3** | 99.74 % | 300131 | Bone mineral density QTL18, osteoporosis, 300910 (3), X-linked dominant; Diaphragmatic hernia 5, X-linked, 306950 (3), X-linked |
| **PLVAP** | 99.96 % | 607647 | Diarrhea 10, protein-losing enteropathy type, 618183 (3), Autosomal recessive |
| **PLXNA1** | 99.89 % | 601055 | Dworschak-Punetha neurodevelopmental syndrome, 619955 (3), Autosomal recessive |
| **PLXND1** | 99.74 % | 604282 | Congenital heart defects, multiple types, 9, 620294 (3), Autosomal recessive |
| **PMM2** | 99.59 % | 601785 | Congenital disorder of glycosylation, type Ia, 212065 (3), Autosomal recessive |
| **PMP22** | 99.88 % | 601097 | Charcot-Marie-Tooth disease, type 1A, 118220 (3), Autosomal dominant; Roussy-Levy syndrome, 180800 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 1E, 118300 (3), Autosomal dominant; ?Neuropathy, inflammatory demyelinating, 139393 (3), ?Autosomal dominant; Neuropathy, recurrent, with pressure palsies, 162500 (3), Autosomal dominant; Dejerine-Sottas disease, 145900 (3), Autosomal recessive, Autosomal dominant |
| **PMPCA** | 99.9 % | 613036 | Spinocerebellar ataxia, autosomal recessive 2, 213200 (3), Autosomal recessive |
| **PMPCB** | 99.76 % | 603131 | Multiple mitochondrial dysfunctions syndrome 6, 617954 (3), Autosomal recessive |
| **PMS2** | 92.98 % | 600259 | Lynch syndrome 4, 614337 (3); Mismatch repair cancer syndrome 4, 619101 (3), Autosomal recessive |
| **PNKP** | 99.82 % | 605610 | ?Charcot-Marie-Tooth disease, type 2B2, 605589 (3), Autosomal recessive; Ataxia-oculomotor apraxia 4, 616267 (3), Autosomal recessive; Microcephaly, seizures, and developmental delay, 613402 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **PNLIP** | 99.65 % | 246600 | ?Pancreatic lipase deficiency, 614338 (3), Autosomal recessive |
| **PNP** | 99.97 % | 164050 | Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179 (3), Autosomal recessive |
| **PNPLA1** | 99.92 % | 612121 | Ichthyosis, congenital, autosomal recessive 10, 615024 (3), Autosomal recessive |
| **PNPLA2** | 99.9 % | 609059 | Neutral lipid storage disease with myopathy, 610717 (3), Autosomal recessive |
| **PNPLA6** | 99.93 % | 603197 | Spastic paraplegia 39, autosomal recessive, 612020 (3), Autosomal recessive; Oliver-McFarlane syndrome, 275400 (3), Autosomal recessive; ?Laurence-Moon syndrome, 245800 (3), Autosomal recessive; Boucher-Neuhauser syndrome, 215470 (3), Autosomal recessive |
| **PNPLA8** | 99.88 % | 612123 | Mitochondrial myopathy with lactic acidosis, 251950 (3), Autosomal recessive |
| **PNPO** | 98.99 % | 603287 | Pyridoxamine 5'-phosphate oxidase deficiency, 610090 (3), Autosomal recessive |
| **PNPT1** | 99.38 % | 610316 | Spinocerebellar ataxia 25, 608703 (3), Autosomal dominant; Deafness, autosomal recessive 70, with or without adult-onset neurodegeneration, 614934 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 13, 614932 (3), Autosomal recessive |
| **POC1A** | 99.49 % | 614783 | Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813 (3), Autosomal recessive |
| **POC1B** | 98.54 % | 614784 | Cone-rod dystrophy 20, 615973 (3), Autosomal recessive |
| **POC5** | 99.74 % | 617880 | *No OMIM phenotypes* |
| **POGLUT1** | 99.82 % | 615618 | Dowling-Degos disease 4, 615696 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232 (3), Autosomal recessive |
| **POLA1** | 99.61 % | 312040 | Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 (3), X-linked recessive; Van Esch-O'Driscoll syndrome, 301030 (3), X-linked recessive |
| **POLD3** | 99.74 % | 611415 | Immunodeficiency 122, 620869 (3), Autosomal recessive |
| **POLE** | 99.91 % | 174762 | {Colorectal cancer, susceptibility to, 12}, 615083 (3), Autosomal dominant; FILS syndrome, 615139 (3), Autosomal recessive; IMAGE-I syndrome, 618336 (3), Autosomal recessive |
| **POLG** | 99.94 % | 174763 | Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 (3), Autosomal recessive; Progressive external ophthalmoplegia, autosomal dominant 1, 157640 (3), Autosomal dominant; Progressive external ophthalmoplegia, autosomal recessive 1, 258450 (3), Autosomal recessive |
| **POLG2** | 99.51 % | 604983 | Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131 (3), Autosomal dominant; ?Mitochondrial DNA depletion syndrome 16 (hepatic type), 618528 (3), Autosomal recessive; ?Mitochondrial DNA depletion syndrome 16B (neuroophthalmic type), 619425 (3), Autosomal recessive |
| **POLH** | 99.77 % | 603968 | Xeroderma pigmentosum, variant type, 278750 (3), Autosomal recessive |
| **POLR1C** | 100 % | 610060 | Leukodystrophy, hypomyelinating, 11, 616494 (3), Autosomal recessive; Treacher Collins syndrome 3, 248390 (3), Autosomal recessive |
| **POLR1D** | 99.99 % | 613715 | Treacher Collins syndrome 2, 613717 (3), Autosomal recessive, Autosomal dominant |
| **POLR3A** | 99.79 % | 614258 | Wiedemann-Rautenstrauch syndrome, 264090 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3), Autosomal recessive |
| **POLR3B** | 99.7 % | 614366 | Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381 (3), Autosomal recessive; Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742 (3), Autosomal dominant |
| **POLR3K** | 99.99 % | 606007 | Leukodystrophy, hypomyelinating, 21, 619310 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **POLRMT** | 99.8 % | 601778 | Combined oxidative phosphorylation deficiency 55, 619743 (3), Autosomal recessive, Autosomal dominant |
| **POMC** | 99.85 % | 176830 | {Obesity, early-onset, susceptibility to}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial; Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734 (3), Autosomal recessive |
| **POMGNT1** | 99.2 % | 606822 | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 3, 613151 (3), Autosomal recessive; Retinitis pigmentosa 76, 617123 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 (3), Autosomal recessive |
| **POMGNT2** | 99.95 % | 614828 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 8, 614830 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135 (3), Autosomal recessive |
| **POMK** | 99.98 % | 615247 | ?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249 (3), Autosomal recessive |
| **POMP** | 99.97 % | 613386 | Proteasome-associated autoinflammatory syndrome 2, 618048 (3), Autosomal dominant; Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952 (3), Autosomal recessive |
| **POMT1** | 99.7 % | 607423 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 1, 613155 (3), Autosomal recessive |
| **POMT2** | 99.84 % | 607439 | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 2, 613156 (3), Autosomal recessive |
| **POP1** | 99.89 % | 602486 | Anauxetic dysplasia 2, 617396 (3), Autosomal recessive |
| **POPDC3** | 99.93 % | 605824 | Muscular dystrophy, limb-girdle, autosomal recessive 26, 618848 (3), Autosomal recessive |
| **POR** | 99.46 % | 124015 | Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 (3), Autosomal recessive; Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571 (3) |
| **PORCN** | 99.81 % | 300651 | Focal dermal hypoplasia, 305600 (3), X-linked dominant |
| **POU1F1** | 99.91 % | 173110 | Pituitary hormone deficiency, combined or isolated, 1, 613038 (3), Autosomal recessive, Autosomal dominant |
| **POU3F4** | 99.97 % | 300039 | Deafness, X-linked 2, 304400 (3), X-linked recessive |
| **PPA2** | 99.76 % | 609988 | ?Sudden cardiac failure, alcohol-induced, 617223 (3), Autosomal recessive; Sudden cardiac failure, infantile, 617222 (3), Autosomal recessive |
| **PPCS** | 98.58 % | 609853 | Cardiomyopathy, dilated, 2C, 618189 (3), Autosomal recessive |
| **PPFIBP1** | 98.01 % | 603141 | Neurodevelopmental disorder with seizures, microcephaly, and brain abnormalities, 620024 (3), Autosomal recessive |
| **PPIB** | 99.91 % | 123841 | Osteogenesis imperfecta, type IX, 259440 (3), Autosomal recessive |
| **PPIL1** | 99.97 % | 601301 | Pontocerebellar hypoplasia, type 14, 619301 (3), Autosomal recessive |
| **PPIP5K2** | 99.69 % | 611648 | Deafness, autosomal recessive 100, 618422 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **PPP1R15B** | 99.89 % | 613257 | Microcephaly, short stature, and impaired glucose metabolism 2, 616817 (3), Autosomal recessive |
| **PPP2R3C** | 99.62 % | 615902 | Spermatogenic failure 36, 618420 (3), Autosomal dominant; Myoectodermal gonadal dysgenesis syndrome, 618419 (3), Autosomal recessive |
| **PPT1** | 97.93 % | 600722 | Ceroid lipofuscinosis, neuronal, 1, 256730 (3), Autosomal recessive |
| **PQBP1** | 99.91 % | 300463 | Renpenning syndrome, 309500 (3), X-linked recessive |
| **PRDM12** | 99.99 % | 616458 | Neuropathy, hereditary sensory and autonomic, type VIII, 616488 (3), Autosomal recessive |
| **PRDM13** | 99.84 % | 616741 | Pontocerebellar hypoplasia, type 17, 619909 (3), Autosomal recessive; Cerebellar dysfunction, impaired intellectual development, and hypogonadotropic hypogonadism, 619761 (3), Autosomal recessive |
| **PRDM5** | 99.75 % | 614161 | Brittle cornea syndrome 2, 614170 (3), Autosomal recessive |
| **PRDX3** | 99.97 % | 604769 | Spinocerebellar ataxia, autosomal recessive 32, 619862 (3), Autosomal recessive; Corneal dystrophy, punctiform and polychromatic pre-Descemet, 619871 (3), Autosomal dominant |
| **PREPL** | 99.57 % | 609557 | Myasthenic syndrome, congenital, 22, 616224 (3), Autosomal recessive |
| **PRF1** | 99.89 % | 170280 | Hemophagocytic lymphohistiocytosis, familial, 2, 603553 (3), Autosomal recessive; Aplastic anemia, 609135 (3); Lymphoma, non-Hodgkin, 605027 (3) |
| **PRG4** | 99.44 % | 604283 | Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250 (3), Autosomal recessive |
| **PRICKLE1** | 99.7 % | 608500 | Epilepsy, progressive myoclonic 1B, 612437 (3), Autosomal recessive |
| **PRIM1** | 98.37 % | 176635 | Primordial dwarfism-immunodeficiency-lipodystrophy syndrome, 620005 (3), Autosomal recessive |
| **PRKCD** | 99.51 % | 176977 | Autoimmune lymphoproliferative syndrome, type III, 615559 (3), Autosomal recessive |
| **PRKDC** | 99.89 % | 600899 | Immunodeficiency 26, with or without neurologic abnormalities, 615966 (3), Autosomal recessive |
| **PRKG2** | 99.81 % | 601591 | Spondylometaphyseal dysplasia, Pagnamenta type, 619638 (3), Autosomal recessive; Acromesomelic dysplasia 4, 619636 (3), Autosomal recessive |
| **PRKN** | 99.74 % | 602544 | Adenocarcinoma of lung, somatic, 211980 (3); Parkinson disease, juvenile, type 2, 600116 (3), Autosomal recessive; Ovarian cancer, somatic, 167000 (3) |
| **PRKRA** | 99.56 % | 603424 | Dystonia 16, 612067 (3), Autosomal recessive |
| **PRMT7** | 99.65 % | 610087 | Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157 (3), Autosomal recessive |
| **PROC** | 99.58 % | 612283 | Thrombophilia 3 due to protein C deficiency, autosomal dominant, 176860 (3), Autosomal dominant; Thrombophilia 3 due to protein C deficiency, autosomal recessive, 612304 (3), Autosomal recessive |
| **PRODH** | 4.01 % | 606810 | {Schizophrenia, susceptibility to, 4}, 600850 (3), Autosomal dominant; Hyperprolinemia, type I, 239500 (3), Autosomal recessive |
| **PROM1** | 99.9 % | 604365 | Macular dystrophy, retinal, 2, 608051 (3), Autosomal dominant; Retinitis pigmentosa 41, 612095 (3), Autosomal recessive; Stargardt disease 4, 603786 (3), Autosomal dominant; Cone-rod dystrophy 12, 612657 (3), Autosomal recessive, Autosomal dominant |
| **PROP1** | 99.75 % | 601538 | Pituitary hormone deficiency, combined, 2, 262600 (3), Autosomal recessive |
| **PRORP** | 99.83 % | 609947 | Combined oxidative phosphorylation deficiency 54, 619737 (3), Autosomal recessive |
| **PROS1** | 99.58 % | 176880 | Thrombophilia 5 due to protein S deficiency, autosomal recessive, 614514 (3), Autosomal recessive; Thrombophilia 5 due to protein S deficiency, autosomal dominant, 612336 (3), Autosomal dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **PRPH2** | 99.9 % | 179605 | Macular dystrophy, patterned, 1, 169150 (3), Autosomal dominant; Choroidal dystrophy, central areolar 2, 613105 (3), Autosomal dominant; Retinitis punctata albescens, 136880 (3), Autosomal recessive, Autosomal dominant; Leber congenital amaurosis 18, 608133 (3), Digenic dominant, Autosomal recessive, Autosomal dominant; Macular dystrophy, vitelliform, 3, 608161 (3), Autosomal dominant; Retinitis pigmentosa 7 and digenic form, 608133 (3), Digenic dominant, Autosomal recessive, Autosomal dominant |
| **PRPS1** | 99.76 % | 311850 | Arts syndrome, 301835 (3), X-linked recessive; Phosphoribosylpyrophosphate synthetase superactivity, 300661 (3), X-linked recessive; Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 (3), X-linked recessive; Deafness, X-linked 1, 304500 (3), X-linked; Gout, PRPS-related, 300661 (3), X-linked recessive |
| **PRRX1** | 99.21 % | 167420 | Agnathia-otocephaly complex, 202650 (3), Autosomal recessive, Autosomal dominant |
| **PRSS56** | 99.72 % | 613858 | Microphthalmia, isolated 6, 613517 (3), Autosomal recessive |
| **PRUNE1** | 99.54 % | 617413 | Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481 (3), Autosomal recessive |
| **PRX** | 99.97 % | 605725 | Charcot-Marie-Tooth disease, type 4F, 614895 (3), Autosomal recessive; Dejerine-Sottas disease, 145900 (3), Autosomal recessive, Autosomal dominant |
| **PSAP** | 99.77 % | 176801 | Combined SAP deficiency, 611721 (3), Autosomal recessive; Krabbe disease, atypical, 611722 (3), Autosomal recessive; Metachromatic leukodystrophy due to SAP-b deficiency, 249900 (3), Autosomal recessive; Gaucher disease, atypical, 610539 (3); {Parkinson disease 24, autosomal dominant, susceptibility to}, 619491 (3), Autosomal dominant |
| **PSAT1** | 99.75 % | 610936 | Neu-Laxova syndrome 2, 616038 (3), Autosomal recessive; Phosphoserine aminotransferase deficiency, 610992 (3), Autosomal recessive |
| **PSMB1** | 99.93 % | 602017 | ?Neurodevelopmental disorder with microcephaly, hypotonia, and absent language, 620038 (3), Autosomal recessive |
| **PSMB10** | 99.58 % | 176847 | Immunodeficiency 121 with autoinflammation, 620807 (3), Autosomal dominant; Proteasome-associated autoinflammatory syndrome 5, 619175 (3), Autosomal recessive |
| **PSMB8** | 99.74 % | 177046 | Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040 (3), Autosomal recessive |
| **PSPH** | 98.61 % | 172480 | Phosphoserine phosphatase deficiency, 614023 (3), Autosomal recessive |
| **PTCD3** | 99.76 % | 614918 | Combined oxidative phosphorylation deficiency 51, 619057 (3), Autosomal recessive |
| **PTCHD1** | 99.97 % | 300828 | {Autism, susceptibility to, X-linked 4}, 300830 (3), X-linked recessive |
| **PTCRA** | 99.74 % | 606817 | Immunodeficiency 126, 620931 (3), Autosomal recessive |
| **PTF1A** | 100 % | 607194 | Pancreatic and cerebellar agenesis, 609069 (3), Autosomal recessive; Pancreatic agenesis 2, 615935 (3), Autosomal recessive |
| **PTH** | 99.91 % | 168450 | Hypoparathyroidism, familial isolated 1, 146200 (3), Autosomal recessive, Autosomal dominant |
| **PTH1R** | 99.34 % | 168468 | Metaphyseal chondrodysplasia, Murk Jansen type, 156400 (3), Autosomal dominant; Eiken syndrome, 600002 (3), Autosomal recessive; Failure of tooth eruption, primary, 125350 (3), Autosomal dominant; Chondrodysplasia, Blomstrand type, 215045 (3), Autosomal recessive |
| **PTPN14** | 99.75 % | 603155 | Choanal atresia and lymphedema, 613611 (3), Autosomal recessive |
| **PTPN23** | 99.88 % | 606584 | Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity, 618890 (3), Autosomal recessive |
| **PTPRC** | 97.95 % | 151460 | Immunodeficiency 105, severe combined, 619924 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **PTPRQ** | 98.85 % | 603317 | Deafness, autosomal dominant 73, 617663 (3), Autosomal dominant; Deafness, autosomal recessive 84A, 613391 (3), Autosomal recessive |
| **PTRH2** | 99.79 % | 608625 | Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263 (3), Autosomal recessive |
| **PTRHD1** | 99.88 % | 617342 | Neurodevelopmental disorder with early-onset parkinsonism and behavioral abnormalities, 620747 (3), Autosomal recessive |
| **PTS** | 99.9 % | 612719 | Hyperphenylalaninemia, BH4-deficient, A, 261640 (3), Autosomal recessive |
| **PUS1** | 99.96 % | 608109 | Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462 (3), Autosomal recessive |
| **PUS3** | 99.96 % | 616283 | Neurodevelopmental disorder with microcephaly and gray sclerae, 617051 (3), Autosomal recessive |
| **PUS7** | 99.84 % | 616261 | Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342 (3), Autosomal recessive |
| **PXDN** | 99.9 % | 605158 | Anterior segment dysgenesis 7, with sclerocornea, 269400 (3), Autosomal recessive |
| **PYCR1** | 99.75 % | 179035 | Cutis laxa, autosomal recessive, type IIIB, 614438 (3), Autosomal recessive; Cutis laxa, autosomal recessive, type IIB, 612940 (3), Autosomal recessive |
| **PYCR2** | 99.67 % | 616406 | Leukodystrophy, hypomyelinating, 10, 616420 (3), Autosomal recessive |
| **PYGL** | 99.88 % | 613741 | Glycogen storage disease VI, 232700 (3), Autosomal recessive |
| **PYGM** | 99.72 % | 608455 | McArdle disease, 232600 (3), Autosomal recessive |
| **PYROXD1** | 99.49 % | 617220 | Myopathy, myofibrillar, 8, 617258 (3), Autosomal recessive |
| **QARS1** | 99.82 % | 603727 | Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760 (3), Autosomal recessive |
| **QDPR** | 99.37 % | 612676 | Hyperphenylalaninemia, BH4-deficient, C, 261630 (3), Autosomal recessive |
| **QRSL1** | 99.87 % | 617209 | Combined oxidative phosphorylation deficiency 40, 618835 (3), Autosomal recessive |
| **RAB18** | 99.77 % | 602207 | Warburg micro syndrome 3, 614222 (3), Autosomal recessive |
| **RAB23** | 99.9 % | 606144 | Carpenter syndrome, 201000 (3), Autosomal recessive |
| **RAB27A** | 99.97 % | 603868 | Griscelli syndrome, type 2, 607624 (3), Autosomal recessive |
| **RAB28** | 99.88 % | 612994 | Cone-rod dystrophy 18, 615374 (3), Autosomal recessive |
| **RAB33B** | 99.98 % | 605950 | Smith-McCort dysplasia 2, 615222 (3), Autosomal recessive |
| **RAB39B** | 92.19 % | 300774 | Intellectual developmental disorder, X-linked 72, 300271 (3), X-linked recessive; Waisman syndrome, 311510 (3), X-linked recessive |
| **RAB3GAP1** | 99.26 % | 602536 | Martsolf syndrome 2, 619420 (3), Autosomal recessive; Warburg micro syndrome 1, 600118 (3), Autosomal recessive |
| **RAB3GAP2** | 99.66 % | 609275 | Martsolf syndrome 1, 212720 (3), Autosomal recessive; Warburg micro syndrome 2, 614225 (3), Autosomal recessive |
| **RABGAP1** | 99.91 % | 615882 | *No OMIM phenotypes* |
| **RAC2** | 99.81 % | 602049 | Immunodeficiency 73A with defective neutrophil chemotaxix and leukocytosis, 608203 (3), Autosomal dominant; ?Immunodeficiency 73C with defective neutrophil chemotaxis and hypogammaglobulinemia, 618987 (3), Autosomal recessive; Immunodeficiency 73B with defective neutrophil chemotaxis and lymphopenia, 618986 (3), Autosomal dominant |
| **RACGAP1** | 98.29 % | 604980 | Anemia, congenital dyserythropoietic, type IIIb, autosomal recessive, 619789 (3), Autosomal recessive |
| **RAD50** | 99.81 % | 604040 | Nijmegen breakage syndrome-like disorder, 613078 (3), Autosomal recessive |
| **RAD51C** | 98.2 % | 602774 | {Breast-ovarian cancer, familial, susceptibility to, 3}, 613399 (3); Fanconi anemia, complementation group O, 613390 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **RAG1** | 100 % | 179615 | Omenn syndrome, 603554 (3), Autosomal recessive; Severe combined immunodeficiency, B cell-negative, 601457 (3), Autosomal recessive; Combined cellular and humoral immune defects with granulomas, 233650 (3), Autosomal recessive; Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889 (3) |
| **RAG2** | 99.99 % | 179616 | Severe combined immunodeficiency, B cell-negative, 601457 (3), Autosomal recessive; Combined cellular and humoral immune defects with granulomas, 233650 (3), Autosomal recessive; Omenn syndrome, 603554 (3), Autosomal recessive |
| **RALGAPA1** | 99.75 % | 608884 | Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation, 618797 (3), Autosomal recessive |
| **RAPSN** | 99.78 % | 601592 | Fetal akinesia deformation sequence 2, 618388 (3), Autosomal recessive; Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326 (3), Autosomal recessive |
| **RARB** | 99.84 % | 180220 | Microphthalmia, syndromic 12, 615524 (3), Autosomal recessive, Autosomal dominant |
| **RARS1** | 99.85 % | 107820 | Leukodystrophy, hypomyelinating, 9, 616140 (3), Autosomal recessive |
| **RARS2** | 99.8 % | 611524 | Pontocerebellar hypoplasia, type 6, 611523 (3), Autosomal recessive |
| **RASGRP1** | 99.8 % | 603962 | Immunodeficiency 64, 618534 (3), Autosomal recessive |
| **RAX** | 99.27 % | 601881 | Microphthalmia, syndromic 16, 611038 (3), Autosomal recessive |
| **RAX2** | 99.72 % | 610362 | Retinitis pigmentosa 95, 620102 (3), Autosomal recessive; Cone-rod dystrophy 11, 610381 (3), Autosomal dominant; ?Macular degeneration, age-related, 6, 613757 (3) |
| **RBBP8** | 99.93 % | 604124 | Seckel syndrome 2, 606744 (3), Autosomal recessive; Jawad syndrome, 251255 (3), Autosomal recessive; Pancreatic carcinoma, somatic (3) |
| **RBCK1** | 99.93 % | 610924 | Polyglucosan body myopathy 1 with or without immunodeficiency, 615895 (3), Autosomal recessive |
| **RBL2** | 99.39 % | 180203 | Brunet-Wagner neurodevelopmental syndrome, 619690 (3), Autosomal recessive |
| **RBM10** | 99.72 % | 300080 | TARP syndrome, 311900 (3), X-linked recessive |
| **RBM28** | 99.92 % | 612074 | ?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079 (3), Autosomal recessive |
| **RBM8A** | 99.16 % | 605313 | Thrombocytopenia-absent radius syndrome, 274000 (3), Autosomal recessive |
| **RBMX** | 99.69 % | 300199 | ?Intellectual developmental disorder, X-linked syndromic, Gustavson type, 309555 (3), X-linked recessive; ?Intellectual developmental disorder, X-linked syndromic, Shashi type, 300238 (3), X-linked recessive |
| **RBP3** | 99.85 % | 180290 | ?Retinitis pigmentosa 66, 615233 (3), Autosomal recessive |
| **RBP4** | 99.69 % | 180250 | Microphthalmia/coloboma 10, 616428 (3), Autosomal dominant; Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147 (3), Autosomal recessive |
| **RCBTB1** | 99.89 % | 607867 | Retinal dystrophy with or without extraocular anomalies, 617175 (3), Autosomal recessive |
| **RD3** | 99.83 % | 180040 | Leber congenital amaurosis 12, 610612 (3), Autosomal recessive |
| **RDH12** | 99.87 % | 608830 | Leber congenital amaurosis 13, 612712 (3), Autosomal recessive, Autosomal dominant |
| **RDX** | 99.9 % | 179410 | Deafness, autosomal recessive 24, 611022 (3), Autosomal recessive |
| **RECQL** | 99.86 % | 600537 | RECON progeroid syndrome, 620370 (3), Autosomal recessive |
| **RECQL4** | 99.91 % | 603780 | Baller-Gerold syndrome, 218600 (3), Autosomal recessive; Rothmund-Thomson syndrome, type 2, 268400 (3), Autosomal recessive; RAPADILINO syndrome, 266280 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **REEP1** | 99.49 % | 609139 | Neuronopathy, distal hereditary motor, autosomal recessive 6, 620011 (3), Autosomal recessive; Spastic paraplegia 31, autosomal dominant, 610250 (3), Autosomal dominant; ?Neuronopathy, distal hereditary motor, autosomal dominant 12, 614751 (3), Autosomal dominant |
| **REEP6** | 99.93 % | 609346 | Retinitis pigmentosa 77, 617304 (3), Autosomal recessive |
| **REL** | 98.69 % | 164910 | Immunodeficiency 92, 619652 (3), Autosomal recessive |
| **RELB** | 99.26 % | 604758 | Immunodeficiency 53, 617585 (3), Autosomal recessive |
| **RELN** | 99.85 % | 600514 | {Epilepsy, familial temporal lobe, 7}, 616436 (3), Autosomal dominant; Lissencephaly 2 (Norman-Roberts type), 257320 (3), Autosomal recessive |
| **RELT** | 99.88 % | 611211 | Amelogenesis imperfecta, type IIIC, 618386 (3), Autosomal recessive |
| **REN** | 98.91 % | 179820 | Renal tubular dysgenesis, 267430 (3), Autosomal recessive; [Hyperproreninemia] (3); Tubulointerstitial kidney disease, autosomal dominant, 4, 613092 (3), Autosomal dominant |
| **RETREG1** | 99.79 % | 613114 | Neuropathy, hereditary sensory and autonomic, type IIB, 613115 (3), Autosomal recessive |
| **RFC1** | 99.64 % | 102579 | Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome, 614575 (3), Autosomal recessive |
| **RFT1** | 98.55 % | 611908 | Congenital disorder of glycosylation, type In, 612015 (3), Autosomal recessive |
| **RFX5** | 99.29 % | 601863 | ?MHC class II deficiency 5, 620818 (3), Autosomal recessive; MHC class II deficiency 3, 620816 (3), Autosomal recessive |
| **RFX6** | 99.63 % | 612659 | Mitchell-Riley syndrome, 615710 (3), Autosomal recessive |
| **RFXANK** | 100 % | 603200 | MHC class II deficiency 2, 620815 (3), Autosomal recessive |
| **RFXAP** | 99.87 % | 601861 | MHC class II deficiency 4, 620817 (3), Autosomal recessive |
| **RHAG** | 99.73 % | 180297 | Overhydrated hereditary stomatocytosis, 185000 (3), Autosomal dominant; Anemia, hemolytic, Rh-null, regulator type, 268150 (3), Autosomal recessive |
| **RHCE** | 95.9 % | 111700 | Rh-null disease, amorph type, 617970 (3), Autosomal recessive |
| **RHO** | 99.95 % | 180380 | Night blindness, congenital stationary, autosomal dominant 1, 610445 (3), Autosomal dominant; Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 (3), Autosomal recessive, Autosomal dominant; Retinitis punctata albescens, 136880 (3), Autosomal recessive, Autosomal dominant |
| **RHOH** | 99.87 % | 602037 | Immunodeficiency 129, 618307 (3), Autosomal recessive |
| **RIMS2** | 99.9 % | 606630 | Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970 (3), Autosomal recessive |
| **RIN2** | 99.88 % | 610222 | Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075 (3), Autosomal recessive |
| **RINT1** | 99.97 % | 610089 | Infantile liver failure syndrome 3, 618641 (3), Autosomal recessive |
| **RIPK1** | 99.91 % | 603453 | Immunodeficiency 57 with autoinflammation, 618108 (3), Autosomal recessive; Autoinflammation with episodic fever and lymphadenopathy, 618852 (3), Autosomal dominant |
| **RIPK4** | 99.68 % | 605706 | CHAND syndrome, 214350 (3), Autosomal recessive; Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650 (3), Autosomal recessive |
| **RIPOR2** | 99.89 % | 611410 | Deafness, autosomal dominant 21, 607017 (3), Autosomal dominant; ?Deafness, autosomal recessive 104, 616515 (3), Autosomal recessive |
| **RIPPLY2** | 99.74 % | 609891 | ?Spondylocostal dysostosis 6, 616566 (3), Autosomal recessive |
| **RLBP1** | 99.78 % | 180090 | Bothnia retinal dystrophy, 607475 (3), Autosomal recessive; Newfoundland rod-cone dystrophy, 607476 (3); Retinitis punctata albescens, 136880 (3), Autosomal recessive, Autosomal dominant; Fundus albipunctatus, 136880 (3), Autosomal recessive, Autosomal dominant |
| **RLIM** | 99.77 % | 300379 | Tonne-Kalscheuer syndrome, 300978 (3), X-linked |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **RMND1** | 99.79 % | 614917 | Combined oxidative phosphorylation deficiency 11, 614922 (3), Autosomal recessive |
| **RMRP** | 98.59 % | 157660 | Anauxetic dysplasia 1, 607095 (3), Autosomal recessive; Metaphyseal dysplasia without hypotrichosis, 250460 (3), Autosomal recessive; Cartilage-hair hypoplasia, 250250 (3), Autosomal recessive |
| **RNASEH1** | 99.97 % | 604123 | Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479 (3), Autosomal recessive |
| **RNASEH2A** | 99.4 % | 606034 | Aicardi-Goutieres syndrome 4, 610333 (3), Autosomal recessive |
| **RNASEH2B** | 99.89 % | 610326 | Aicardi-Goutieres syndrome 2, 610181 (3), Autosomal recessive |
| **RNASEH2C** | 99.34 % | 610330 | Aicardi-Goutieres syndrome 3, 610329 (3), Autosomal recessive |
| **RNASET2** | 99.98 % | 612944 | Leukoencephalopathy, cystic, without megalencephaly, 612951 (3), Autosomal recessive |
| **RNF113A** | 99.98 % | 300951 | Trichothiodystrophy 5, nonphotosensitive, 300953 (3), X-linked |
| **RNF168** | 99.86 % | 612688 | RIDDLE syndrome, 611943 (3), Autosomal recessive |
| **RNF170** | 99.85 % | 614649 | Ataxia, sensory, 1, autosomal dominant, 608984 (3), Autosomal dominant; Spastic paraplegia 85, autosomal recessive, 619686 (3), Autosomal recessive |
| **RNF213** | 99.89 % | 613768 | {Moyamoya disease 2, susceptibility to}, 607151 (3), Autosomal recessive, Autosomal dominant |
| **RNF216** | 99.87 % | 609948 | Cerebellar ataxia and hypogonadotropic hypogonadism, 212840 (3), Autosomal recessive |
| **RNF220** | 98.8 % | 616136 | Leukodystrophy, hypomyelinating, 23, with ataxia, deafness, liver dysfunction, and dilated cardiomyopathy, 619688 (3), Autosomal recessive |
| **RNF31** | 99.82 % | 612487 | Immunodeficiency 115 with autoinflammation, 620632 (3), Autosomal recessive |
| **RNPC3** | 95.2 % | 618016 | Pituitary hormone deficiency, combined or isolated, 7, 618160 (3), Autosomal recessive |
| **RNU4ATAC** | 99.32 % | 601428 | Roifman syndrome, 616651 (3), Autosomal recessive; Lowry-Wood syndrome, 226960 (3), Autosomal recessive; Microcephalic osteodysplastic primordial dwarfism, type I, 210710 (3), Autosomal recessive |
| **RNU7-1** | 99.98 % | 617876 | Aicardi-Goutieres syndrome 9, 619487 (3), Autosomal recessive |
| **ROBO1** | 99.79 % | 602430 | Pituitary hormone deficiency, combined or isolated, 8, 620303 (3), Autosomal dominant; Neurooculorenal syndrome, 620305 (3), Autosomal recessive; ?Nystagmus 8, congenital, autosomal recessive, 257400 (3), Autosomal recessive |
| **ROBO3** | 99.88 % | 608630 | Gaze palsy, familial horizontal, with progressive scoliosis, 1, 607313 (3), Autosomal recessive |
| **ROGDI** | 98.83 % | 614574 | Kohlschutter-Tonz syndrome, 226750 (3), Autosomal recessive |
| **ROR1** | 99.23 % | 602336 | ?Deafness, autosomal recessive 108, 617654 (3), Autosomal recessive |
| **ROR2** | 99.79 % | 602337 | Brachydactyly, type B1, 113000 (3), Autosomal dominant; Robinow syndrome, autosomal recessive, 268310 (3), Autosomal recessive |
| **RORC** | 98.53 % | 602943 | Immunodeficiency 42, 616622 (3), Autosomal recessive |
| **RP1** | 99.87 % | 603937 | Retinitis pigmentosa 1, 180100 (3), Autosomal recessive, Autosomal dominant |
| **RP1L1** | 99.94 % | 608581 | Occult macular dystrophy, 613587 (3), Autosomal dominant; Retinitis pigmentosa 88, 618826 (3), Autosomal recessive |
| **RP2** | 99.64 % | 300757 | Retinitis pigmentosa 2, 312600 (3), X-linked |
| **RPE65** | 98.26 % | 180069 | Retinitis pigmentosa 20, 613794 (3), Autosomal recessive; Retinitis pigmentosa 87 with choroidal involvement, 618697 (3), Autosomal dominant; Leber congenital amaurosis 2, 204100 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **RPGR** | 99.36 % | 312610 | Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 (3), X-linked; Cone-rod dystrophy, X-linked, 1, 304020 (3), X-linked recessive; Retinitis pigmentosa 3, 300029 (3), X-linked; Macular degeneration, X-linked atrophic, 300834 (3), X-linked recessive |
| **RPGRIP1** | 99.75 % | 605446 | Cone-rod dystrophy 13, 608194 (3), Autosomal recessive; Leber congenital amaurosis 6, 613826 (3), Autosomal recessive |
| **RPGRIP1L** | 96.51 % | 610937 | Joubert syndrome 7, 611560 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive; ?COACH syndrome 3, 619113 (3), Autosomal recessive |
| **RPIA** | 99.1 % | 180430 | Ribose 5-phosphate isomerase deficiency, 608611 (3), Autosomal recessive |
| **RPL10** | 99.72 % | 312173 | {Autism, susceptibility to, X-linked 5}, 300847 (3); Intellectual developmental disorder, X-linked syndromic 35, 300998 (3), X-linked recessive |
| **RPL3L** | 99.73 % | 617416 | Cardiomyopathy, dilated, 2D, 619371 (3), Autosomal recessive |
| **RPS6KA3** | 99.42 % | 300075 | Intellectual developmental disorder, X-linked 19, 300844 (3), X-linked dominant; Coffin-Lowry syndrome, 303600 (3), X-linked dominant |
| **RRM2B** | 99.86 % | 604712 | Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 (3), Autosomal recessive; Rod-cone dystrophy, sensorineural deafness, and Fanconi-type renal dysfunction, 268315 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 (3), Autosomal dominant |
| **RRP7A** | 96.08 % | 619449 | ?Microcephaly 28, primary, autosomal recessive, 619453 (3), Autosomal recessive |
| **RS1** | 99.87 % | 300839 | Retinoschisis, 312700 (3), X-linked recessive |
| **RSPH1** | 98.95 % | 609314 | Ciliary dyskinesia, primary, 24, 615481 (3), Autosomal recessive |
| **RSPH3** | 99.85 % | 615876 | Ciliary dyskinesia, primary, 32, 616481 (3), Autosomal recessive |
| **RSPH4A** | 99.84 % | 612647 | Ciliary dyskinesia, primary, 11, 612649 (3), Autosomal recessive |
| **RSPH9** | 99.64 % | 612648 | Ciliary dyskinesia, primary, 12, 612650 (3), Autosomal recessive |
| **RSPO1** | 99.45 % | 609595 | Palmoplantar hyperkeratosis and true hermaphroditism, 610644 (3), Autosomal recessive; Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644 (3), Autosomal recessive |
| **RSPO2** | 99.8 % | 610575 | ?Humerofemoral hypoplasia with radiotibial ray deficiency, 618022 (3), Autosomal recessive; Tetraamelia syndrome 2, 618021 (3), Autosomal recessive |
| **RSPO4** | 96.41 % | 610573 | Anonychia congenita, 206800 (3), Autosomal recessive |
| **RSPRY1** | 99.51 % | 616585 | Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723 (3), Autosomal recessive |
| **RTEL1** | 99.92 % | 608833 | Dyskeratosis congenita, autosomal dominant 4, 615190 (3), Autosomal recessive, Autosomal dominant; Dyskeratosis congenita, autosomal recessive 5, 615190 (3), Autosomal recessive, Autosomal dominant; Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 3, 616373 (3), Autosomal dominant |
| **RTN4IP1** | 99.85 % | 610502 | Optic atrophy 10 with or without ataxia, impaired intellectual development and seizures, 616732 (3), Autosomal recessive |
| **RTTN** | 99.84 % | 610436 | Microcephaly, short stature, and polymicrogyria with seizures, 614833 (3), Autosomal recessive |
| **RXYLT1** | 99.19 % | 605862 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041 (3), Autosomal recessive |
| **RYR1** | 99.68 % | 180901 | Congenital myopathy 1B, autosomal recessive, 255320 (3), Autosomal recessive; Congenital myopathy 1A, autosomal dominant, with susceptibility to malignant hyperthermia, 117000 (3), Autosomal dominant; King-Denborough syndrome, 619542 (3), Autosomal dominant; {Malignant hyperthermia susceptibility 1}, 145600 (3), Autosomal dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **S1PR2** | 99.94 % | 605111 | Deafness, autosomal recessive 68, 610419 (3), Autosomal recessive |
| **SACS** | 99.95 % | 604490 | Spastic ataxia, Charlevoix-Saguenay type, 270550 (3), Autosomal recessive |
| **SAG** | 99.9 % | 181031 | Retinitis pigmentosa 47, autosomal recessive, 613758 (3), Autosomal recessive; Retinitis pigmentosa 96, autosomal dominant, 620228 (3), Autosomal dominant; Oguchi disease-1, 258100 (3), Autosomal recessive |
| **SAMD7** | 99.94 % | 620493 | Macular dystrophy with or without cone dysfunction, 620762 (3), Autosomal recessive |
| **SAMD9** | 99.94 % | 610456 | Tumoral calcinosis, familial, normophosphatemic, 610455 (3), Autosomal recessive; Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041 (3), Autosomal dominant; MIRAGE syndrome, 617053 (3), Autosomal dominant |
| **SAMHD1** | 99.94 % | 606754 | ?Chilblain lupus 2, 614415 (3), Autosomal dominant; Aicardi-Goutieres syndrome 5, 612952 (3), Autosomal recessive |
| **SAR1B** | 99.49 % | 607690 | Chylomicron retention disease, 246700 (3), Autosomal recessive |
| **SARS1** | 97.74 % | 607529 | Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709 (3), Autosomal recessive |
| **SARS2** | 99.91 % | 612804 | Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845 (3), Autosomal recessive |
| **SASH3** | 99.88 % | 300441 | Immunodeficiency 102, 301082 (3), X-linked recessive |
| **SASS6** | 96 % | 609321 | Microcephaly 14, primary, autosomal recessive, 616402 (3), Autosomal recessive |
| **SBDS** | 99.32 % | 607444 | {Aplastic anemia, susceptibility to}, 609135 (3); Shwachman-Diamond syndrome 1, 260400 (3), Autosomal recessive |
| **SBF1** | 99.97 % | 603560 | Charcot-Marie-Tooth disease, type 4B3, 615284 (3), Autosomal recessive |
| **SBF2** | 99.53 % | 607697 | Charcot-Marie-Tooth disease, type 4B2, 604563 (3), Autosomal recessive |
| **SC5D** | 99.82 % | 602286 | Lathosterolosis, 607330 (3), Autosomal recessive |
| **SCAPER** | 99.71 % | 611611 | Intellectual developmental disorder and retinitis pigmentosa, 618195 (3), Autosomal recessive |
| **SCARB2** | 99.66 % | 602257 | Epilepsy, progressive myoclonic 4, with or without renal failure, 254900 (3), Autosomal recessive |
| **SCARF2** | 98.63 % | 613619 | Van den Ende-Gupta syndrome, 600920 (3), Autosomal recessive |
| **SCN1B** | 99.86 % | 600235 | Generalized epilepsy with febrile seizures plus, type 1, 604233 (3), Autosomal dominant; Developmental and epileptic encephalopathy 52, 617350 (3), Autosomal recessive; Cardiac conduction defect, nonspecific, 612838 (3); Atrial fibrillation, familial, 13, 615377 (3), Autosomal dominant; Brugada syndrome 5, 612838 (3) |
| **SCN4A** | 99.95 % | 603967 | Paramyotonia congenita, 168300 (3), Autosomal dominant; Hyperkalemic periodic paralysis, 170500 (3), Autosomal dominant; Congenital myopathy 22B, severe fetal, 620369 (3), Autosomal recessive; Hypokalemic periodic paralysis, type 2, 613345 (3), Autosomal dominant; Myotonia congenita, atypical, acetazolamide-responsive, 608390 (3), Autosomal dominant; Myasthenic syndrome, congenital, 16, 614198 (3), Autosomal recessive; Congenital myopathy 22A, classic, 620351 (3), Autosomal recessive |
| **SCN5A** | 99.86 % | 600163 | Ventricular fibrillation, familial, 1, 603829 (3); Heart block, progressive, type IA, 113900 (3), Autosomal dominant; Cardiomyopathy, dilated, 1E, 601154 (3), Autosomal dominant; Heart block, nonprogressive, 113900 (3), Autosomal dominant; Long QT syndrome 3, 603830 (3), Autosomal dominant; Sick sinus syndrome 1, 608567 (3), Autosomal recessive; Brugada syndrome 1, 601144 (3), Autosomal dominant; Atrial fibrillation, familial, 10, 614022 (3), Autosomal dominant; {Sudden infant death syndrome, susceptibility to}, 272120 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **SCN9A** | 99.74 % | 603415 | Erythermalgia, primary, 133020 (3), Autosomal dominant; Insensitivity to pain, congenital, 243000 (3), Autosomal recessive; Small fiber neuropathy, 133020 (3), Autosomal dominant; Paroxysmal extreme pain disorder, 167400 (3), Autosomal dominant; Neuropathy, hereditary sensory and autonomic, type IID, 243000 (3), Autosomal recessive |
| **SCNN1A** | 100 % | 600228 | Pseudohypoaldosteronism, type IB1, autosomal recessive, 264350 (3), Autosomal recessive; ?Liddle syndrome 3, 618126 (3), Autosomal dominant; Bronchiectasis with or without elevated sweat chloride 2, 613021 (3), Autosomal dominant |
| **SCNN1B** | 98.62 % | 600760 | Bronchiectasis with or without elevated sweat chloride 1, 211400 (3), Autosomal dominant; Pseudohypoaldosteronism, type IB2, autosomal recessive, 620125 (3), Autosomal recessive; Liddle syndrome 1, 177200 (3), Autosomal dominant |
| **SCNN1G** | 99.86 % | 600761 | Bronchiectasis with or without elevated sweat chloride 3, 613071 (3), Autosomal dominant; Pseudohypoaldosteronism, type IB3, autosomal recessive, 620126 (3), Autosomal recessive; Liddle syndrome 2, 618114 (3), Autosomal dominant |
| **SCO1** | 99.77 % | 603644 | Mitochondrial complex IV deficiency, nuclear type 4, 619048 (3), Autosomal recessive |
| **SCO2** | 100 % | 604272 | Myopia 6, 608908 (3), Autosomal dominant; Mitochondrial complex IV deficiency, nuclear type 2, 604377 (3), Autosomal recessive |
| **SCUBE3** | 99.44 % | 614708 | Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 2, 619184 (3), Autosomal recessive |
| **SCYL1** | 99.79 % | 607982 | Spinocerebellar ataxia, autosomal recessive 21, 616719 (3), Autosomal recessive |
| **SCYL2** | 98.99 % | 616365 | Arthrogryposis multiplex congenita 4, neurogenic, with agenesis of the corpus callosum, 618766 (3), Autosomal recessive |
| **SDCCAG8** | 100 % | 613524 | Senior-Loken syndrome 7, 613615 (3), Autosomal recessive; Bardet-Biedl syndrome 16, 615993 (3), Autosomal recessive |
| **SDHA** | 97.97 % | 600857 | Cardiomyopathy, dilated, 1GG, 613642 (3), Autosomal recessive; Mitochondrial complex II deficiency, nuclear type 1, 252011 (3), Autosomal recessive; Neurodegeneration with ataxia and late-onset optic atrophy, 619259 (3), Autosomal dominant; Pheochromocytoma/paraganglioma syndrome 5, 614165 (3), Autosomal dominant |
| **SDHAF1** | 99.95 % | 612848 | Mitochondrial complex II deficiency, nuclear type 2, 619166 (3), Autosomal recessive |
| **SDHB** | 93.61 % | 185470 | Pheochromocytoma/paraganglioma syndrome 4, 115310 (3), Autosomal dominant; Mitochondrial complex II deficiency, nuclear type 4, 619224 (3), Autosomal recessive; Gastrointestinal stromal tumor, 606764 (3), Autosomal dominant, Isolated cases; Paraganglioma and gastric stromal sarcoma, 606864 (3) |
| **SDHD** | 91.13 % | 602690 | Pheochromocytoma/paraganglioma syndrome 1, 168000 (3), Autosomal dominant; Paraganglioma and gastric stromal sarcoma, 606864 (3); Mitochondrial complex II deficiency, nuclear type 3, 619167 (3), Autosomal recessive |
| **SDR9C7** | 99.91 % | 609769 | Ichthyosis, congenital, autosomal recessive 13, 617574 (3), Autosomal recessive |
| **SEC23A** | 99.88 % | 610511 | Craniolenticulosutural dysplasia, 607812 (3), Autosomal recessive, Autosomal dominant |
| **SEC23B** | 99.84 % | 610512 | ?Cowden syndrome 7, 616858 (3), Autosomal dominant; Dyserythropoietic anemia, congenital, type II, 224100 (3), Autosomal recessive |
| **SEC24D** | 99.62 % | 607186 | Cole-Carpenter syndrome 2, 616294 (3), Autosomal recessive |
| **SEC31A** | 99.72 % | 610257 | ?Halperin-Birk syndrome, 618651 (3), Autosomal recessive |
| **SECISBP2** | 99.91 % | 607693 | Thyroid hormone metabolism, abnormal, 1, 609698 (3), Autosomal recessive |
| **SELENOI** | 99.72 % | 607915 | Spastic paraplegia 81, autosomal recessive, 618768 (3), Autosomal recessive |
| **SELENON** | 99.12 % | 606210 | Congenital myopathy 3 with rigid spine, 602771 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **SEMA4A** | 98.4 % | 607292 | Retinitis pigmentosa 35, 610282 (3), Autosomal recessive; Cone-rod dystrophy 10, 610283 (3), Autosomal recessive |
| **SEPSECS** | 99.43 % | 613009 | Pontocerebellar hypoplasia type 2D, 613811 (3), Autosomal recessive |
| **SERAC1** | 99.84 % | 614725 | 3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739 (3), Autosomal recessive |
| **SERPINA6** | 99.96 % | 122500 | Corticosteroid-binding globulin deficiency, 611489 (3), Autosomal recessive, Autosomal dominant |
| **SERPINB6** | 99.98 % | 173321 | ?Deafness, autosomal recessive 91, 613453 (3), Autosomal recessive |
| **SERPINB7** | 99.95 % | 603357 | Palmoplantar keratoderma, Nagashima type, 615598 (3), Autosomal recessive |
| **SERPINB8** | 99.76 % | 601697 | Peeling skin syndrome 5, 617115 (3), Autosomal recessive |
| **SERPINC1** | 99.6 % | 107300 | Thrombophilia 7 due to antithrombin III deficiency, 613118 (3), Autosomal recessive, Autosomal dominant |
| **SERPINE1** | 99.39 % | 173360 | Plasminogen activator inhibitor-1 deficiency, 613329 (3), Autosomal recessive, Autosomal dominant; {Transcription of plasminogen activator inhibitor, modulator of} (3) |
| **SERPINF1** | 99.91 % | 172860 | Osteogenesis imperfecta, type VI, 613982 (3), Autosomal recessive |
| **SERPINF2** | 99.61 % | 613168 | Alpha-2-plasmin inhibitor deficiency, 262850 (3), Autosomal recessive |
| **SERPING1** | 99.9 % | 606860 | Angioedema, hereditary, 1 and 2, 106100 (3), Autosomal recessive, Autosomal dominant; Complement component 4, partial deficiency of, 120790 (3), Autosomal dominant |
| **SERPINH1** | 99.67 % | 600943 | {Preterm premature rupture of the membranes, susceptibility to}, 610504 (3), Multifactorial; Osteogenesis imperfecta, type X, 613848 (3), Autosomal recessive |
| **SETX** | 99.86 % | 608465 | Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 (3), Autosomal recessive; Amyotrophic lateral sclerosis 4, juvenile, 602433 (3), Autosomal dominant |
| **SFRP4** | 99.94 % | 606570 | Pyle disease, 265900 (3), Autosomal recessive |
| **SFTPB** | 99.83 % | 178640 | Surfactant metabolism dysfunction, pulmonary, 1, 265120 (3), Autosomal recessive |
| **SFXN4** | 99.94 % | 615564 | Combined oxidative phosphorylation deficiency 18, 615578 (3), Autosomal recessive |
| **SGCA** | 99.76 % | 600119 | Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099 (3), Autosomal recessive |
| **SGCB** | 99.71 % | 600900 | Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286 (3), Autosomal recessive |
| **SGCD** | 99.98 % | 601411 | Cardiomyopathy, dilated, 1L, 606685 (3); Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287 (3), Autosomal recessive |
| **SGCG** | 99.97 % | 608896 | Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700 (3), Autosomal recessive |
| **SGO1** | 99.91 % | 609168 | Chronic atrial and intestinal dysrhythmia, 616201 (3), Autosomal recessive |
| **SGPL1** | 99.71 % | 603729 | RENI syndrome, 617575 (3), Autosomal recessive |
| **SGSH** | 99.79 % | 605270 | Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900 (3), Autosomal recessive |
| **SH2D1A** | 99.61 % | 300490 | Lymphoproliferative syndrome, X-linked, 1, 308240 (3), X-linked recessive |
| **SH3PXD2B** | 99.96 % | 613293 | Frank-ter Haar syndrome, 249420 (3), Autosomal recessive |
| **SH3TC2** | 99.93 % | 608206 | Charcot-Marie-Tooth disease, type 4C, 601596 (3), Autosomal recessive; Mononeuropathy of the median nerve, mild, 613353 (3), Autosomal dominant |
| **SHMT2** | 99.41 % | 138450 | Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121 (3), Autosomal recessive |
| **SHOX** | 95.24 % | 312865 | Short stature, idiopathic familial, 300582 (3); Leri-Weill dyschondrosteosis, 127300 (3), Pseudoautosomal dominant; Langer mesomelic dysplasia, 249700 (3), Pseudoautosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **SHQ1** | 99.68 % | 613663 | Neurodevelopmental disorder with dystonia and seizures, 619922 (3), Autosomal recessive; ?Dystonia 35, childhood-onset, 619921 (3), Autosomal recessive |
| **SI** | 99.55 % | 609845 | Sucrase-isomaltase deficiency, congenital, 222900 (3), Autosomal recessive |
| **SIGMAR1** | 99.9 % | 601978 | ?Neuronopathy, distal hereditary motor, autosomal recessive 2, 605726 (3), Autosomal recessive; ?Amyotrophic lateral sclerosis 16, juvenile, 614373 (3), Autosomal recessive |
| **SIL1** | 99.79 % | 608005 | Marinesco-Sjogren syndrome, 248800 (3), Autosomal recessive |
| **SIX6** | 99.99 % | 606326 | Optic disc anomalies with retinal and/or macular dystrophy, 212550 (3), Autosomal recessive |
| **SKIV2L** | 99.43 % | 600478 | Trichohepatoenteric syndrome 2, 614602 (3), Autosomal recessive |
| **SLC10A7** | 99.96 % | 611459 | Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363 (3), Autosomal recessive |
| **SLC11A2** | 99.2 % | 600523 | Anemia, hypochromic microcytic, with iron overload 1, 206100 (3), Autosomal recessive |
| **SLC12A1** | 99.8 % | 600839 | Bartter syndrome, type 1, 601678 (3), Autosomal recessive |
| **SLC12A3** | 99.42 % | 600968 | Gitelman syndrome, 263800 (3), Autosomal recessive |
| **SLC12A5** | 99.79 % | 606726 | {Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685 (3), Autosomal dominant; Developmental and epileptic encephalopathy 34, 616645 (3), Autosomal recessive |
| **SLC12A6** | 99.78 % | 604878 | Agenesis of the corpus callosum with peripheral neuropathy, 218000 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2II, 620068 (3), Autosomal dominant |
| **SLC13A3** | 99.86 % | 606411 | Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate, 618384 (3), Autosomal recessive |
| **SLC13A5** | 99.92 % | 608305 | Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905 (3), Autosomal recessive |
| **SLC16A1** | 99.17 % | 600682 | Hyperinsulinemic hypoglycemia, familial, 7, 610021 (3), Autosomal dominant; Erythrocyte lactate transporter defect, 245340 (3), Autosomal dominant; Monocarboxylate transporter 1 deficiency, 616095 (3), Autosomal recessive, Autosomal dominant |
| **SLC16A2** | 99.68 % | 300095 | Allan-Herndon-Dudley syndrome, 300523 (3), X-linked |
| **SLC17A5** | 99.7 % | 604322 | Salla disease, 604369 (3), Autosomal recessive; Sialic acid storage disorder, infantile, 269920 (3), Autosomal recessive |
| **SLC18A2** | 99.84 % | 193001 | Parkinsonism-dystonia, infantile, 2, 618049 (3), Autosomal recessive |
| **SLC18A3** | 99.5 % | 600336 | Myasthenic syndrome, congenital, 21, presynaptic, 617239 (3), Autosomal recessive |
| **SLC19A2** | 98.59 % | 603941 | Thiamine-responsive megaloblastic anemia syndrome, 249270 (3), Autosomal recessive |
| **SLC19A3** | 99.9 % | 606152 | Thiamine metabolism dysfunction syndrome 2 (biotin/thiamine-responsive basal ganglia disease type), 607483 (3), Autosomal recessive |
| **SLC1A4** | 99.65 % | 600229 | Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657 (3), Autosomal recessive |
| **SLC22A12** | 99.84 % | 607096 | Hypouricemia, renal, 220150 (3), Autosomal recessive |
| **SLC22A5** | 99.98 % | 603377 | Carnitine deficiency, systemic primary, 212140 (3), Autosomal recessive |
| **SLC24A4** | 99.77 % | 609840 | [Skin/hair/eye pigmentation 6, blond/brown hair], 210750 (3), Autosomal recessive; Amelogenesis imperfecta, type IIA5, 615887 (3), Autosomal recessive; [Skin/hair/eye pigmentation 6, blue/green eyes], 210750 (3), Autosomal recessive |
| **SLC24A5** | 99.96 % | 609802 | [Skin/hair/eye pigmentation 4, fair/dark skin], 113750 (3), Autosomal recessive; Albinism, oculocutaneous, type VI, 113750 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **SLC25A1** | 99.49 % | 190315 | Combined D-2- and L-2-hydroxyglutaric aciduria, 615182 (3), Autosomal recessive; Myasthenic syndrome, congenital, 23, presynaptic, 618197 (3), Autosomal recessive |
| **SLC25A12** | 99.54 % | 603667 | Developmental and epileptic encephalopathy 39, 612949 (3), Autosomal recessive |
| **SLC25A13** | 99.29 % | 603859 | Citrullinemia, type II, neonatal-onset, 605814 (3), Autosomal recessive; Citrullinemia, adult-onset type II, 603471 (3), Autosomal recessive |
| **SLC25A15** | 99.89 % | 603861 | Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 (3), Autosomal recessive |
| **SLC25A19** | 99.69 % | 606521 | Microcephaly, Amish type, 607196 (3), Autosomal recessive; Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710 (3), Autosomal recessive |
| **SLC25A20** | 99.36 % | 613698 | Carnitine-acylcarnitine translocase deficiency, 212138 (3), Autosomal recessive |
| **SLC25A22** | 99.89 % | 609302 | Developmental and epileptic encephalopathy 3, 609304 (3), Autosomal recessive |
| **SLC25A26** | 99.82 % | 611037 | Combined oxidative phosphorylation deficiency 28, 616794 (3), Autosomal recessive |
| **SLC25A3** | 99.68 % | 600370 | Mitochondrial phosphate carrier deficiency, 610773 (3), Autosomal recessive |
| **SLC25A32** | 99.94 % | 138480 | ?Exercise intolerance, riboflavin-responsive, 616839 (3), Autosomal recessive |
| **SLC25A36** | 99.68 % | 616149 | Hyperinsulinemic hypoglycemia, familial, 8, 620211 (3), Autosomal recessive |
| **SLC25A38** | 99.76 % | 610819 | Anemia, sideroblastic, 2, pyridoxine-refractory, 205950 (3), Autosomal recessive |
| **SLC25A4** | 99.95 % | 103220 | Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283 (3), Autosomal dominant; Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184 (3), Autosomal dominant |
| **SLC25A42** | 99.74 % | 610823 | Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416 (3), Autosomal recessive |
| **SLC25A46** | 99.88 % | 610826 | Neuropathy, hereditary motor and sensory, type VIB, 616505 (3), Autosomal recessive; Pontocerebellar hypoplasia, type 1E, 619303 (3), Autosomal recessive |
| **SLC26A2** | 99.91 % | 606718 | Epiphyseal dysplasia, multiple, 4, 226900 (3), Autosomal recessive; De la Chapelle dysplasia, 256050 (3), Autosomal recessive; Diastrophic dysplasia, 222600 (3), Autosomal recessive; Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 (3), Autosomal recessive; Achondrogenesis Ib, 600972 (3), Autosomal recessive; Atelosteogenesis, type II, 256050 (3), Autosomal recessive |
| **SLC26A3** | 99.88 % | 126650 | Diarrhea 1, secretory chloride, congenital, 214700 (3), Autosomal recessive |
| **SLC26A4** | 99.86 % | 605646 | Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791 (3), Autosomal recessive; Pendred syndrome, 274600 (3), Autosomal recessive |
| **SLC27A4** | 99.85 % | 604194 | Ichthyosis prematurity syndrome, 608649 (3), Autosomal recessive |
| **SLC29A3** | 99.61 % | 612373 | Histiocytosis-lymphadenopathy plus syndrome, 602782 (3), Autosomal recessive |
| **SLC2A1** | 99.5 % | 138140 | Dystonia 9, 601042 (3), Autosomal dominant; GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 (3), Autosomal recessive, Autosomal dominant; Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 (3), Autosomal dominant; GLUT1 deficiency syndrome 2, childhood onset, 612126 (3), Autosomal dominant |
| **SLC2A10** | 99.96 % | 606145 | Arterial tortuosity syndrome, 208050 (3), Autosomal recessive |
| **SLC2A2** | 99.84 % | 138160 | Fanconi-Bickel syndrome, 227810 (3), Autosomal recessive; {Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant |
| **SLC2A9** | 99.58 % | 606142 | {Uric acid concentration, serum, QTL 2}, 612076 (3), Autosomal recessive, Autosomal dominant; Hypouricemia, renal, 2, 612076 (3), Autosomal recessive, Autosomal dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **SLC30A10** | 99.72 % | 611146 | Hypermanganesemia with dystonia 1, 613280 (3), Autosomal recessive |
| **SLC30A9** | 99.71 % | 604604 | Birk-Landau-Perez syndrome, 617595 (3), Autosomal recessive |
| **SLC33A1** | 99.68 % | 603690 | Spastic paraplegia 42, autosomal dominant, 612539 (3), Autosomal dominant; Huppke-Brendel syndrome, 614482 (3), Autosomal recessive |
| **SLC34A1** | 99.89 % | 182309 | ?Fanconi renotubular syndrome 2, 613388 (3), Autosomal recessive; Hypercalcemia, infantile, 2, 616963 (3), Autosomal recessive; Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286 (3), Autosomal dominant |
| **SLC34A2** | 99.93 % | 604217 | Pulmonary alveolar microlithiasis, 265100 (3), Autosomal recessive |
| **SLC34A3** | 99.85 % | 609826 | Hypophosphatemic rickets with hypercalciuria, 241530 (3), Autosomal recessive |
| **SLC35A1** | 99.44 % | 605634 | Congenital disorder of glycosylation, type IIf, 603585 (3), Autosomal recessive |
| **SLC35A2** | 99.84 % | 314375 | Congenital disorder of glycosylation, type IIm, 300896 (3), X-linked dominant, Somatic mosaicism |
| **SLC35A3** | 94.4 % | 605632 | Arthrogryposis, impaired intellectual development, and seizures, 615553 (3), Autosomal recessive |
| **SLC35B2** | 99.96 % | 610788 | Leukodystrophy, hypomyelinating, 26, with chondrodysplasia, 620269 (3), Autosomal recessive |
| **SLC35C1** | 99.95 % | 605881 | Congenital disorder of glycosylation, type IIc, 266265 (3), Autosomal recessive |
| **SLC35D1** | 97.02 % | 610804 | Schneckenbecken dysplasia, 269250 (3), Autosomal recessive |
| **SLC37A4** | 99.86 % | 602671 | Glycogen storage disease Ib, 232220 (3), Autosomal recessive; Congenital disorder of glycosylation, type IIw, 619525 (3), Autosomal dominant; Glycogen storage disease Ic, 232240 (3), Autosomal recessive |
| **SLC38A3** | 99.8 % | 604437 | Developmental and epileptic encephalopathy 102, 619881 (3), Autosomal recessive |
| **SLC38A8** | 99.83 % | 615585 | Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218 (3), Autosomal recessive |
| **SLC39A13** | 99.3 % | 608735 | Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350 (3), Autosomal recessive |
| **SLC39A14** | 99.21 % | 608736 | ?Hyperostosis cranalis interna, 144755 (3), Autosomal dominant; Hypermanganesemia with dystonia 2, 617013 (3), Autosomal recessive |
| **SLC39A4** | 99.85 % | 607059 | Acrodermatitis enteropathica, 201100 (3), Autosomal recessive |
| **SLC39A7** | 99.9 % | 601416 | Agammaglobulinemia 9, autosomal recessive, 619693 (3), Autosomal recessive |
| **SLC39A8** | 99.75 % | 608732 | Congenital disorder of glycosylation, type IIn, 616721 (3), Autosomal recessive |
| **SLC3A1** | 99.88 % | 104614 | Cystinuria, 220100 (3), Autosomal recessive, Autosomal dominant |
| **SLC44A1** | 99.84 % | 606105 | Neurodegeneration, childhood-onset, with ataxia, tremor, optic atrophy, and cognitive decline, 618868 (3), Autosomal recessive |
| **SLC45A1** | 99.78 % | 605763 | Intellectual developmental disorder with neuropsychiatric features, 617532 (3), Autosomal recessive |
| **SLC45A2** | 99.82 % | 606202 | [Skin/hair/eye pigmentation 5, dark/light eyes], 227240 (3), Autosomal recessive; [Skin/hair/eye pigmentation 5, black/nonblack hair], 227240 (3), Autosomal recessive; Albinism, oculocutaneous, type IV, 606574 (3), Autosomal recessive; [Skin/hair/eye pigmentation 5, dark/fair skin], 227240 (3), Autosomal recessive |
| **SLC46A1** | 99.89 % | 611672 | Folate malabsorption, hereditary, 229050 (3), Autosomal recessive |
| **SLC4A1** | 99.73 % | 109270 | [Blood group, Swann], 601550 (3); [Blood group, Wright], 112050 (3); Distal renal tubular acidosis 1, 179800 (3), Autosomal dominant; [Blood group, Waldner], 112010 (3); Spherocytosis, type 4, 612653 (3), Autosomal dominant; [Blood group, Froese], 601551 (3); Distal renal tubular acidosis 4 with hemolytic anemia, 611590 (3), Autosomal recessive; {Malaria, resistance to}, 611162 (3); Cryohydrocytosis, 185020 (3), Autosomal dominant; Ovalocytosis, SA type, 166900 (3), Autosomal dominant; [Blood group, Diego], 110500 (3) |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **SLC4A11** | 99.86 % | 610206 | Corneal endothelial dystrophy, autosomal recessive, 217700 (3), Autosomal recessive; Corneal dystrophy, Fuchs endothelial, 4, 613268 (3); Corneal endothelial dystrophy and perceptive deafness, 217400 (3), Autosomal recessive |
| **SLC4A2** | 99.93 % | 109280 | ?Osteopetrosis, autosomal recessive 9, 620366 (3), Autosomal recessive |
| **SLC4A4** | 99.83 % | 603345 | Proximal renal tubular acidosis-ocular anomaly syndrome, 604278 (3), Autosomal recessive |
| **SLC52A2** | 100 % | 607882 | Brown-Vialetto-Van Laere syndrome 2, 614707 (3), Autosomal recessive |
| **SLC52A3** | 99.93 % | 613350 | ?Fazio-Londe disease, 211500 (3), Autosomal recessive; Brown-Vialetto-Van Laere syndrome 1, 211530 (3), Autosomal recessive |
| **SLC5A1** | 99.89 % | 182380 | Glucose/galactose malabsorption, 606824 (3), Autosomal recessive |
| **SLC5A2** | 99.66 % | 182381 | Renal glucosuria, 233100 (3), Autosomal recessive, Autosomal dominant |
| **SLC5A5** | 99.32 % | 601843 | Thyroid dyshormonogenesis 1, 274400 (3), Autosomal recessive |
| **SLC5A6** | 99.9 % | 604024 | Sodium-dependent multivitamin transporter deficiency, 618973 (3), Autosomal recessive; Peripheral motor neuropathy, childhood-onset, biotin-responsive, 619903 (3), Autosomal recessive |
| **SLC5A7** | 98.76 % | 608761 | Neuronopathy, distal hereditary motor, autosomal dominant 7, 158580 (3), Autosomal dominant; Myasthenic syndrome, congenital, 20, presynaptic, 617143 (3), Autosomal recessive |
| **SLC6A17** | 98.83 % | 610299 | Intellectual developmental disorder, autosomal recessive 48, 616269 (3), Autosomal recessive |
| **SLC6A19** | 99.87 % | 608893 | Hartnup disorder, 234500 (3), Autosomal recessive |
| **SLC6A3** | 99.69 % | 126455 | Parkinsonism-dystonia, infantile, 1, 613135 (3), Autosomal recessive; {Nicotine dependence, protection against}, 188890 (3) |
| **SLC6A5** | 99.71 % | 604159 | Hyperekplexia 3, 614618 (3), Autosomal recessive, Autosomal dominant |
| **SLC6A6** | 99.95 % | 186854 | Hypotaurinemic retinal degeneration and cardiomyopathy, 145350 (3), Autosomal recessive |
| **SLC6A8** | 99.67 % | 300036 | Cerebral creatine deficiency syndrome 1, 300352 (3), X-linked recessive |
| **SLC6A9** | 99.57 % | 601019 | Glycine encephalopathy with normal serum glycine, 617301 (3), Autosomal recessive |
| **SLC7A14** | 99.87 % | 615720 | Retinitis pigmentosa 68, 615725 (3), Autosomal recessive |
| **SLC7A7** | 99.91 % | 603593 | Lysinuric protein intolerance, 222700 (3), Autosomal recessive |
| **SLC7A9** | 99.86 % | 604144 | Cystinuria, 220100 (3), Autosomal recessive, Autosomal dominant |
| **SLC9A1** | 99.72 % | 107310 | Lichtenstein-Knorr syndrome, 616291 (3), Autosomal recessive |
| **SLC9A3** | 99.84 % | 182307 | Diarrhea 8, secretory sodium, congenital, 616868 (3), Autosomal recessive |
| **SLC9A6** | 99.54 % | 300231 | Neurodegenerative disorder, X-linked, female-restricted, with parkinsonism and cognitive impairment, 301142 (3), X-linked dominant; Intellectual developmental disorder, X-linked syndromic, Christianson type, 300243 (3), X-linked |
| **SLCO2A1** | 99.75 % | 601460 | Hypertrophic osteoarthropathy, primary, autosomal dominant, 167100 (3), Autosomal dominant; PHOAR2-enteropathy syndrome, 614441 (3), Autosomal recessive |
| **SLF2** | 99.78 % | 610348 | Atelis syndrome 1, 620184 (3), Autosomal recessive |
| **SLITRK6** | 99.99 % | 609681 | Deafness and myopia, 221200 (3), Autosomal recessive |
| **SLURP1** | 99.55 % | 606119 | Meleda disease, 248300 (3), Autosomal recessive |
| **SLX4** | 99.88 % | 613278 | Fanconi anemia, complementation group P, 613951 (3), Autosomal recessive |
| **SMARCAL1** | 99.78 % | 606622 | Schimke immunoosseous dysplasia, 242900 (3), Autosomal recessive |
| **SMARCD2** | 99.67 % | 601736 | Specific granule deficiency 2, 617475 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **SMC1A** | 99.93 % | 300040 | Cornelia de Lange syndrome 2, 300590 (3), X-linked dominant; Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044 (3), X-linked dominant |
| **SMC5** | 99.38 % | 609386 | Atelis syndrome 2, 620185 (3), Autosomal recessive |
| **SMG9** | 99.44 % | 613176 | Heart and brain malformation syndrome, 616920 (3), Autosomal recessive; Neurodevelopmental disorder with intention tremor, pyramidal signs, dyspraxia, and ocular anomalies, 619995 (3), Autosomal recessive |
| **SMN1** | 7.85 % | 600354 | Spinal muscular atrophy-2, 253550 (3), Autosomal recessive; Spinal muscular atrophy-4, 271150 (3), Autosomal recessive; Spinal muscular atrophy-3, 253400 (3), Autosomal recessive; Spinal muscular atrophy-1, 253300 (3), Autosomal recessive |
| **SMN2** | 6.84 % | 601627 | {Spinal muscular atrophy, type III, modifier of}, 253400 (3), Autosomal recessive |
| **SMO** | 99.78 % | 601500 | Pallister-Hall-like syndrome, 241800 (3), Autosomal recessive; Basal cell carcinoma, somatic, 605462 (3); Curry-Jones syndrome, somatic mosaic, 601707 (3) |
| **SMOC1** | 99.87 % | 608488 | Microphthalmia with limb anomalies, 206920 (3), Autosomal recessive |
| **SMOC2** | 98.78 % | 607223 | Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400 (3), Autosomal recessive |
| **SMPD1** | 99.92 % | 607608 | Niemann-Pick disease, type B, 607616 (3), Autosomal recessive; Niemann-Pick disease, type A, 257200 (3), Autosomal recessive |
| **SMPD4** | 98.85 % | 610457 | Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622 (3), Autosomal recessive |
| **SMS** | 99.74 % | 300105 | Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type, 309583 (3), X-linked recessive |
| **SNAP29** | 99.51 % | 604202 | Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528 (3), Autosomal recessive |
| **SNAPC4** | 99.92 % | 602777 | Neurodevelopmental disorder with motor regression, progressive spastic paraplegia, and oromotor dysfunction, 620515 (3), Autosomal recessive |
| **SNIP1** | 99.51 % | 608241 | Neurodevelopmental disorder with hypotonia, craniofacial abnormalities, and seizures, 614501 (3), Autosomal recessive |
| **SNORD118** | 99.84 % | 616663 | Leukoencephalopathy, brain calcifications, and cysts, 614561 (3), Autosomal recessive |
| **SNX10** | 99.97 % | 614780 | Osteopetrosis, autosomal recessive 8, 615085 (3), Autosomal recessive |
| **SNX14** | 99.73 % | 616105 | Spinocerebellar ataxia, autosomal recessive 20, 616354 (3), Autosomal recessive |
| **SNX27** | 98.63 % | 611541 | *No OMIM phenotypes* |
| **SOD1** | 99.04 % | 147450 | Spastic tetraplegia and axial hypotonia, progressive, 618598 (3), Autosomal recessive; Amyotrophic lateral sclerosis 1, 105400 (3), Autosomal recessive, Autosomal dominant |
| **SORD** | 78.67 % | 182500 | Neuronopathy, distal hereditary motor, autosomal recessive 8, 618912 (3), Autosomal recessive |
| **SOST** | 98.76 % | 605740 | Sclerosteosis 1, 269500 (3), Autosomal recessive; Craniodiaphyseal dysplasia, autosomal dominant, 122860 (3), Autosomal dominant |
| **SOX18** | 99.84 % | 601618 | Hypotrichosis-lymphedema-telangiectasia syndrome, 607823 (3), Autosomal recessive; Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940 (3), Autosomal dominant |
| **SOX3** | 99.6 % | 313430 | Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency, 300123 (3); Panhypopituitarism, X-linked, 312000 (3), X-linked |
| **SP110** | 99.91 % | 604457 | {Mycobacterium tuberculosis, susceptibility to}, 607948 (3); Hepatic venoocclusive disease with immunodeficiency, 235550 (3), Autosomal recessive |
| **SP7** | 100 % | 606633 | Osteogenesis imperfecta, type XII, 613849 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **SPAG1** | 99.63 % | 603395 | Ciliary dyskinesia, primary, 28, 615505 (3), Autosomal recessive |
| **SPARC** | 98.85 % | 182120 | Osteogenesis imperfecta, type XVII, 616507 (3), Autosomal recessive |
| **SPART** | 99.98 % | 607111 | Troyer syndrome, 275900 (3), Autosomal recessive |
| **SPATA5** | 99.32 % | 613940 | Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577 (3), Autosomal recessive |
| **SPATA5L1** | 99.79 % | 619578 | Deafness, autosomal recessive 119, 619615 (3), Autosomal recessive; Neurodevelopmental disorder with hearing loss and spasticity, 619616 (3), Autosomal recessive |
| **SPATA7** | 99.87 % | 609868 | Leber congenital amaurosis 3, 604232 (3), Autosomal recessive; Retinitis pigmentosa 94, variable age at onset, autosomal recessive, 604232 (3), Autosomal recessive |
| **SPEG** | 99.75 % | 615950 | Centronuclear myopathy 5, 615959 (3), Autosomal recessive |
| **SPG11** | 99.76 % | 610844 | Amyotrophic lateral sclerosis 5, juvenile, 602099 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2X, 616668 (3), Autosomal recessive; Spastic paraplegia 11, autosomal recessive, 604360 (3), Autosomal recessive |
| **SPG21** | 99.93 % | 608181 | Mast syndrome, 248900 (3), Autosomal recessive |
| **SPG7** | 99.89 % | 602783 | Spastic paraplegia 7, autosomal recessive, 607259 (3), Autosomal recessive, Autosomal dominant |
| **SPINK1** | 99.93 % | 167790 | Tropical calcific pancreatitis, 608189 (3), Autosomal recessive, Autosomal dominant; Pancreatitis, hereditary, 167800 (3), Autosomal dominant; {Fibrocalculous pancreatic diabetes, susceptibility to}, 608189 (3), Autosomal recessive, Autosomal dominant |
| **SPINK5** | 99.75 % | 605010 | Netherton syndrome, 256500 (3), Autosomal recessive |
| **SPINT2** | 99.84 % | 605124 | Diarrhea 3, secretory sodium, congenital, syndromic, 270420 (3), Autosomal recessive |
| **SPNS2** | 99.75 % | 612584 | ?Deafness, autosomal recessive 115, 618457 (3), Autosomal recessive |
| **SPPL2A** | 99.83 % | 608238 | Immunodeficiency 86, mycobacteriosis, 619549 (3), Autosomal recessive |
| **SPR** | 99.84 % | 182125 | Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716 (3), ?Autosomal dominant, Autosomal recessive |
| **SPRED2** | 99.86 % | 609292 | Noonan syndrome 14, 619745 (3), Autosomal recessive |
| **SPRTN** | 99.91 % | 616086 | Ruijs-Aalfs syndrome, 616200 (3), Autosomal recessive |
| **SPTA1** | 98.7 % | 182860 | Spherocytosis, type 3, 270970 (3), Autosomal recessive; Elliptocytosis-2, 130600 (3), Autosomal dominant; Pyropoikilocytosis, 266140 (3), Autosomal recessive |
| **SPTBN2** | 99.7 % | 604985 | Spinocerebellar ataxia 5, 600224 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 14, 615386 (3), Autosomal recessive |
| **SPTBN4** | 99.78 % | 606214 | Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519 (3), Autosomal recessive |
| **SQOR** | 99.84 % | 617658 | Sulfide:quinone oxidoreductase deficiency, 619221 (3), Autosomal recessive |
| **SQSTM1** | 99.97 % | 601530 | Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 (3), Autosomal recessive; Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 (3), Autosomal dominant; Myopathy, distal, with rimmed vacuoles, 617158 (3), Autosomal dominant; Paget disease of bone 3, 167250 (3), Autosomal dominant |
| **SRD5A2** | 99.79 % | 607306 | Pseudovaginal perineoscrotal hypospadias, 264600 (3), Autosomal recessive |
| **SRD5A3** | 99.98 % | 611715 | Kahrizi syndrome, 612713 (3), Autosomal recessive; Congenital disorder of glycosylation, type Iq, 612379 (3), Autosomal recessive |
| **SRY** | 50.56 % | 480000 | 46XY sex reversal 1, 400044 (3), Y-linked; 46XX sex reversal 1, 400045 (4), X-linked dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **SSR4** | 99.87 % | 300090 | Congenital disorder of glycosylation, type Iy, 300934 (3), X-linked recessive |
| **ST14** | 99.8 % | 606797 | Ichthyosis, congenital, autosomal recessive 11, 602400 (3), Autosomal recessive |
| **ST3GAL3** | 99.88 % | 606494 | Developmental and epileptic encephalopathy 15, 615006 (3), Autosomal recessive; Intellectual developmental disorder, autosomal recessive 12, 611090 (3), Autosomal recessive |
| **ST3GAL5** | 99.49 % | 604402 | Salt and pepper developmental regression syndrome, 609056 (3), Autosomal recessive |
| **STAC3** | 99.22 % | 615521 | Congenital myopathy 13, 255995 (3), Autosomal recessive |
| **STAG2** | 99.33 % | 300826 | Holoprosencephaly 13, X-linked, 301043 (3), X-linked dominant, X-linked recessive; Mullegama-Klein-Martinez syndrome, 301022 (3), X-linked |
| **STAMBP** | 99.81 % | 606247 | Microcephaly-capillary malformation syndrome, 614261 (3), Autosomal recessive |
| **STAR** | 99.71 % | 600617 | Lipoid adrenal hyperplasia, 201710 (3), Autosomal recessive |
| **STAT1** | 99.66 % | 600555 | Immunodeficiency 31C, chronic mucocutaneous candidiasis, autosomal dominant, 614162 (3), Autosomal dominant; Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 (3), Autosomal dominant; Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796 (3), Autosomal recessive |
| **STAT2** | 99.28 % | 600556 | Pseudo-TORCH syndrome 3, 618886 (3), Autosomal recessive; Immunodeficiency 44, 616636 (3), Autosomal recessive |
| **STAT5B** | 97.68 % | 604260 | Growth hormone insensitivity with immune dysregulation 1, autosomal recessive, 245590 (3), Autosomal recessive; Growth hormone insensitivity with immune dysregulation 2, autosomal dominant, 618985 (3), Autosomal dominant |
| **STIL** | 98.88 % | 181590 | Microcephaly 7, primary, autosomal recessive, 612703 (3), Autosomal recessive |
| **STIM1** | 99.89 % | 605921 | Myopathy, tubular aggregate, 1, 160565 (3), Autosomal dominant; Stormorken syndrome, 185070 (3), Autosomal dominant; Immunodeficiency 10, 612783 (3), Autosomal recessive |
| **STK4** | 99.24 % | 604965 | T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868 (3), Autosomal recessive |
| **STN1** | 99.59 % | 613128 | Cerebroretinal microangiopathy with calcifications and cysts 2, 617341 (3), Autosomal recessive |
| **STRA6** | 99.82 % | 610745 | Microphthalmia, syndromic 9, 601186 (3), Autosomal recessive; Microphthalmia, isolated, with coloboma 8, 601186 (3), Autosomal recessive |
| **STRADA** | 99.77 % | 608626 | Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087 (3), Autosomal recessive |
| **STRC** | 42.57 % | 606440 | Deafness, autosomal recessive 16, 603720 (3), Autosomal recessive |
| **STS** | 99.8 % | 300747 | Ichthyosis, X-linked, 308100 (3), X-linked recessive |
| **STT3A** | 99.92 % | 601134 | Congenital disorder of glycosylation, type Iw, autosomal dominant, 619714 (3), Autosomal dominant; Congenital disorder of glycosylation, type Iw, autosomal recessive, 615596 (3), Autosomal recessive |
| **STUB1** | 99.91 % | 607207 | Spinocerebellar ataxia 48, 618093 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 16, 615768 (3), Autosomal recessive |
| **STX11** | 99.93 % | 605014 | Hemophagocytic lymphohistiocytosis, familial, 4, 603552 (3), Autosomal recessive |
| **STX1A** | 99.15 % | 186590 | *No OMIM phenotypes* |
| **STX3** | 99.85 % | 600876 | Retinal dystrophy and microvillus inclusion disease, 619446 (3), Autosomal recessive; Diarrhea 12, with microvillus atrophy, 619445 (3), Autosomal recessive |
| **STXBP2** | 99.73 % | 601717 | Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease, 613101 (3), Autosomal recessive |
| **STXBP3** | 96.34 % | 608339 | *No OMIM phenotypes* |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **SUCLA2** | 99.92 % | 603921 | Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 (3), Autosomal recessive |
| **SUCLG1** | 99.79 % | 611224 | Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400 (3), Autosomal recessive |
| **SULT2B1** | 99.62 % | 604125 | Ichthyosis, congenital, autosomal recessive 14, 617571 (3), Autosomal recessive |
| **SUMF1** | 99.88 % | 607939 | Multiple sulfatase deficiency, 272200 (3), Autosomal recessive |
| **SUOX** | 99.89 % | 606887 | Sulfite oxidase deficiency, 272300 (3), Autosomal recessive |
| **SURF1** | 99.88 % | 185620 | Charcot-Marie-Tooth disease, type 4K, 616684 (3), Autosomal recessive; Mitochondrial complex IV deficiency, nuclear type 1, 220110 (3), Autosomal recessive |
| **SVBP** | 97.73 % | 617853 | Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569 (3), Autosomal recessive |
| **SYN1** | 99.87 % | 313440 | Epilepsy, X-linked 1, with variable learning disabilities and behavior disorders, 300491 (3), X-linked; Intellectual developmental disorder, X-linked 50, 300115 (3), X-linked |
| **SYNE1** | 99.89 % | 608441 | Arthrogryposis multiplex congenita 3, myogenic type, 618484 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 8, 610743 (3), Autosomal recessive |
| **SYNE4** | 99.79 % | 615535 | Deafness, autosomal recessive 76, 615540 (3), Autosomal recessive |
| **SYNJ1** | 99.58 % | 604297 | Parkinson disease 20, early-onset, 615530 (3), Autosomal recessive; Developmental and epileptic encephalopathy 53, 617389 (3), Autosomal recessive |
| **SYP** | 99.81 % | 313475 | Intellectual developmental disorder, X-linked 96, 300802 (3), X-linked recessive |
| **SYT2** | 99.63 % | 600104 | Myasthenic syndrome, congenital, 7A, presynaptic, and distal motor neuropathy, autosomal dominant, 616040 (3), Autosomal dominant; Myasthenic syndrome, congenital, 7B, presynaptic, autosomal recessive, 619461 (3), Autosomal recessive |
| **SZT2** | 99.27 % | 615463 | Developmental and epileptic encephalopathy 18, 615476 (3), Autosomal recessive |
| **TACO1** | 99.34 % | 612958 | Mitochondrial complex IV deficiency, nuclear type 8, 619052 (3), Autosomal recessive |
| **TACR3** | 99.84 % | 162332 | Hypogonadotropic hypogonadism 11 with or without anosmia, 614840 (3), Autosomal recessive |
| **TACSTD2** | 99.94 % | 137290 | Corneal dystrophy, gelatinous drop-like, 204870 (3), Autosomal recessive |
| **TAF1** | 99.65 % | 313650 | Intellectual developmental disorder, X-linked syndromic 33, 300966 (3), X-linked recessive; Dystonia-Parkinsonism, X-linked, 314250 (3), X-linked recessive |
| **TAF2** | 99.59 % | 604912 | Intellectual developmental disorder, autosomal recessive 40, 615599 (3), Autosomal recessive |
| **TAF6** | 99.5 % | 602955 | Alazami-Yuan syndrome, 617126 (3), Autosomal recessive |
| **TAF8** | 99.86 % | 609514 | Neurodevelopmental disorder with severe motor impairment, absent language, cerebral hypomyelination, and brain atrophy, 619972 (3), Autosomal recessive |
| **TAFAZZIN** | 99.93 % | 300394 | Barth syndrome, 302060 (3), X-linked recessive |
| **TALDO1** | 99.87 % | 602063 | Transaldolase deficiency, 606003 (3), Autosomal recessive |
| **TANGO2** | 99.2 % | 616830 | Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878 (3), Autosomal recessive |
| **TAP1** | 99.7 % | 170260 | MHC class I deficiency 1, 604571 (3), Autosomal recessive |
| **TAP2** | 99.7 % | 170261 | MHC class I deficiency 2, 620813 (3), Autosomal recessive |
| **TAPT1** | 99.68 % | 612758 | Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897 (3), Autosomal recessive |
| **TARS1** | 99.82 % | 187790 | Trichothiodystrophy 7, nonphotosensitive, 618546 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **TARS2** | 99.24 % | 612805 | Combined oxidative phosphorylation deficiency 21, 615918 (3), Autosomal recessive |
| **TASP1** | 99.82 % | 608270 | Suleiman-El-Hattab syndrome, 618950 (3), Autosomal recessive |
| **TAT** | 99.94 % | 613018 | Tyrosinemia, type II, 276600 (3), Autosomal recessive |
| **TBC1D20** | 99.94 % | 611663 | Warburg micro syndrome 4, 615663 (3), Autosomal recessive |
| **TBC1D23** | 99.15 % | 617687 | Pontocerebellar hypoplasia, type 11, 617695 (3), Autosomal recessive |
| **TBC1D24** | 99.87 % | 613577 | Deafness, autosomal recessive 86, 614617 (3), Autosomal recessive; Epilepsy, rolandic, with paroxysmal exercise-induce dystonia and writer's cramp, 608105 (3), Autosomal recessive; Myoclonic epilepsy, infantile, familial, 605021 (3), Autosomal recessive; Deafness, autosomal dominant 65, 616044 (3), Autosomal dominant; Developmental and epileptic encephalopathy 16, 615338 (3), Autosomal recessive; DOORS syndrome, 220500 (3), Autosomal recessive |
| **TBC1D2B** | 99.76 % | 619152 | Neurodevelopmental disorder with seizures and gingival overgrowth, 619323 (3), Autosomal recessive |
| **TBC1D32** | 99.75 % | 615867 | Orofaciodigital syndrome IX, 258865 (3), Autosomal recessive; Alsahan-Harris syndrome, 621307 (3), Autosomal recessive; Retinitis pigmentosa 100, 621280 (3), Autosomal recessive |
| **TBC1D8B** | 99.43 % | 301027 | Nephrotic syndrome, type 20, 301028 (3), X-linked |
| **TBCD** | 99.88 % | 604649 | Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193 (3), Autosomal recessive |
| **TBCE** | 99.8 % | 604934 | Kenny-Caffey syndrome, type 1, 244460 (3), Autosomal recessive; Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 (3), Autosomal recessive; Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207 (3), Autosomal recessive |
| **TBCK** | 99.74 % | 616899 | Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900 (3), Autosomal recessive |
| **TBL1X** | 99.85 % | 300196 | Hypothyroidism, congenital, nongoitrous, 8, 301033 (3), X-linked |
| **TBX15** | 98.52 % | 604127 | Cousin syndrome, 260660 (3), Autosomal recessive |
| **TBX19** | 99.73 % | 604614 | Adrenocorticotropic hormone deficiency, 201400 (3), Autosomal recessive |
| **TBX22** | 99.83 % | 300307 | Cleft palate with ankyloglossia, 303400 (3), X-linked; ?Abruzzo-Erickson syndrome, 302905 (3), X-linked |
| **TBX4** | 99.72 % | 601719 | Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension, 147891 (3), Autosomal dominant; Amelia, posterior, with pelvic and pulmonary hypoplasia syndrome, 601360 (3), Autosomal recessive |
| **TBX6** | 99.57 % | 602427 | Spondylocostal dysostosis 5, 122600 (3), Autosomal recessive, Autosomal dominant |
| **TBXAS1** | 99.89 % | 274180 | Ghosal hematodiaphyseal syndrome, 231095 (3), Autosomal recessive |
| **TBXT** | 99.89 % | 601397 | Sacral agenesis with vertebral anomalies, 615709 (3), Autosomal recessive; {Neural tube defects, susceptibility to}, 182940 (3), Autosomal dominant |
| **TCAP** | 99.32 % | 604488 | Cardiomyopathy, hypertrophic, 25, 607487 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954 (3), Autosomal recessive |
| **TCEAL1** | 99.87 % | 300237 | Hijazi-Reis syndrome, 301094 (3), X-linked dominant |
| **TCF3** | 99.88 % | 147141 | Agammaglobulinemia 8B, autosomal recessive, 619824 (3), Autosomal recessive; Agammaglobulinemia 8A, autosomal dominant, 616941 (3), Autosomal dominant |
| **TCIRG1** | 99.78 % | 604592 | Osteopetrosis, autosomal recessive 1, 259700 (3), Autosomal recessive |
| **TCN2** | 99.9 % | 613441 | Transcobalamin II deficiency, 275350 (3), Autosomal recessive |
| **TCTN1** | 99.84 % | 609863 | Joubert syndrome 13, 614173 (3), Autosomal recessive |
| **TCTN2** | 99.95 % | 613846 | Joubert syndrome 24, 616654 (3), Autosomal recessive; ?Meckel syndrome 8, 613885 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **TCTN3** | 99.84 % | 613847 | Joubert syndrome 18, 614815 (3), Autosomal recessive; Orofaciodigital syndrome IV, 258860 (3), Autosomal recessive |
| **TDP1** | 99.87 % | 607198 | ?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250 (3), Autosomal recessive |
| **TDP2** | 99.91 % | 605764 | Spinocerebellar ataxia, autosomal recessive 23, 616949 (3), Autosomal recessive |
| **TDRD7** | 99.74 % | 611258 | Cataract 36, 613887 (3), Autosomal recessive |
| **TECPR2** | 99.84 % | 615000 | Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031 (3), Autosomal recessive |
| **TECRL** | 99.8 % | 617242 | Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021 (3), Autosomal recessive |
| **TECTA** | 99.84 % | 602574 | Deafness, autosomal dominant 8/12, 601543 (3), Autosomal dominant; Deafness, autosomal recessive 21, 603629 (3), Autosomal recessive |
| **TELO2** | 99.92 % | 611140 | You-Hoover-Fong syndrome, 616954 (3), Autosomal recessive |
| **TENM3** | 99.93 % | 610083 | Microphthalmia, syndromic 15, 615145 (3), Autosomal recessive; ?Microphthalmia/coloboma 9, 615145 (3), Autosomal recessive |
| **TENT5A** | 99.95 % | 611357 | Osteogenesis imperfecta, type XVIII, 617952 (3), Autosomal recessive |
| **TERT** | 99.86 % | 187270 | Dyskeratosis congenita, autosomal dominant 2, 613989 (3), Autosomal recessive, Autosomal dominant; Dyskeratosis congenita, autosomal recessive 4, 613989 (3), Autosomal recessive, Autosomal dominant; Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 1, 614742 (3), Autosomal dominant; {Melanoma, cutaneous malignant, 9}, 615134 (3), Autosomal dominant; {Leukemia, acute myeloid}, 601626 (3), Somatic mutation, Autosomal dominant |
| **TET2** | 99.98 % | 612839 | Myelodysplastic syndrome, somatic, 614286 (3); Immunodeficiency 75, 619126 (3), Autosomal recessive |
| **TET3** | 99.67 % | 613555 | Beck-Fahrner syndrome, 618798 (3), Autosomal recessive, Autosomal dominant |
| **TF** | 99.85 % | 190000 | Atransferrinemia, 209300 (3), Autosomal recessive |
| **TFAM** | 99.53 % | 600438 | ?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156 (3), Autosomal recessive |
| **TFE3** | 99.59 % | 314310 | Intellectual developmental disorder, X-linked syndromic, with pigmentary mosaicism and coarse facies, 301066 (3), X-linked; Renal cell carcinoma, papillary, 1, 300854 (3) |
| **TFG** | 98.77 % | 602498 | ?Spastic paraplegia 57, autosomal recessive, 615658 (3), Autosomal recessive; Hereditary motor and sensory neuropathy, Okinawa type, 604484 (3), Autosomal dominant |
| **TFR2** | 99.6 % | 604720 | Hemochromatosis, type 3, 604250 (3), Autosomal recessive |
| **TFRC** | 99.44 % | 190010 | Immunodeficiency 46, 616740 (3), Autosomal recessive |
| **TG** | 99.88 % | 188450 | {Autoimmune thyroid disease, susceptibility to, 3}, 608175 (3); Thyroid dyshormonogenesis 3, 274700 (3), Autosomal recessive |
| **TGDS** | 99.96 % | 616146 | Catel-Manzke syndrome, 616145 (3), Autosomal recessive |
| **TGM1** | 99.11 % | 190195 | Ichthyosis, congenital, autosomal recessive 1, 242300 (3), Autosomal recessive |
| **TGM5** | 99.86 % | 603805 | Peeling skin syndrome 2, 609796 (3), Autosomal recessive |
| **TH** | 99.72 % | 191290 | Segawa syndrome, recessive, 605407 (3), Autosomal recessive |
| **THG1L** | 99.93 % | 618802 | Spinocerebellar ataxia, autosomal recessive 28, 618800 (3), Autosomal recessive |
| **THOC2** | 99.44 % | 300395 | Arthrogryposis multiplex congenita 7, X-linked, 301127 (3), X-linked; Intellectual developmental disorder, X-linked syndromic, Kumar type, 300957 (3), X-linked |
| **THOC6** | 99.53 % | 615403 | Beaulieu-Boycott-Innes syndrome, 613680 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **THRB** | 99.68 % | 190160 | Thyroid hormone resistance, autosomal recessive, 274300 (3), Autosomal recessive; Thyroid hormone resistance, 188570 (3), Autosomal dominant; Thyroid hormone resistance, selective pituitary, 145650 (3), Autosomal dominant |
| **THSD1** | 99.97 % | 616821 | ?Aneurysm, intracranial berry, 12, 618734 (3), Autosomal dominant; Lymphatic malformation 13, 620244 (3), Autosomal recessive |
| **THUMPD1** | 99.78 % | 616662 | Neurodevelopmental disorder with speech delay and variable ocular anomalies, 619989 (3), Autosomal recessive |
| **TIAM1** | 99.9 % | 600687 | Neurodevelopmental disorder with language delay and seizures, 619908 (3), Autosomal recessive |
| **TIMM50** | 98.63 % | 607381 | 3-methylglutaconic aciduria, type IX, 617698 (3), Autosomal recessive |
| **TIMM8A** | 99.84 % | 300356 | Mohr-Tranebjaerg syndrome, 304700 (3), X-linked recessive |
| **TIMMDC1** | 99.85 % | 615534 | Mitochondrial complex I deficiency, nuclear type 31, 618251 (3), Autosomal recessive |
| **TJP2** | 99.84 % | 607709 | Hypercholanemia, familial 1, 607748 (3), Autosomal recessive; Cholestasis, progressive familial intrahepatic 4, 615878 (3), Autosomal recessive |
| **TK2** | 99.19 % | 188250 | Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560 (3), Autosomal recessive; ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069 (3), Autosomal recessive |
| **TKFC** | 98.93 % | 615844 | Triokinase and FMN cyclase deficiency syndrome, 618805 (3), Autosomal recessive |
| **TKT** | 97.06 % | 606781 | Short stature, developmental delay, and congenital heart defects, 617044 (3), Autosomal recessive |
| **TLR7** | 99.97 % | 300365 | Immunodeficiency 74, COVID19-related, X-linked, 301051 (3), X-linked recessive; Systemic lupus erythematosus 17, 301080 (3), X-linked dominant |
| **TLR8** | 99.98 % | 300366 | Immunodeficiency 98 with autoinflammation, X-linked, 301078 (3), X-linked, Somatic mosaicism |
| **TMC1** | 99.83 % | 606706 | Deafness, autosomal dominant 36, 606705 (3), Autosomal dominant; Deafness, autosomal recessive 7, 600974 (3), Autosomal recessive |
| **TMC6** | 99.86 % | 605828 | {Epidermodysplasia verruciformis, susceptibility to, 1}, 226400 (3), Autosomal recessive |
| **TMC8** | 99.72 % | 605829 | {Epidermodysplasia verruciformis, susceptibility to, 2}, 618231 (3), Autosomal recessive |
| **TMCO1** | 98.23 % | 614123 | Craniofacial dysmorphism, skeletal anomalies, and impaired intellectual development 1, 213980 (3), Autosomal recessive |
| **TMEM107** | 99.96 % | 616183 | Orofaciodigital syndrome XVI, 617563 (3), Autosomal recessive; Meckel syndrome 13, 617562 (3), Autosomal recessive; ?Joubert syndrome 29, 617562 (3), Autosomal recessive |
| **TMEM126A** | 99.87 % | 612988 | Optic atrophy 7, 612989 (3), Autosomal recessive |
| **TMEM126B** | 99.34 % | 615533 | Mitochondrial complex I deficiency, nuclear type 29, 618250 (3), Autosomal recessive |
| **TMEM132E** | 99.9 % | 616178 | Deafness, autosomal recessive 99, 618481 (3), Autosomal recessive |
| **TMEM138** | 99.94 % | 614459 | Joubert syndrome 16, 614465 (3), Autosomal recessive |
| **TMEM147** | 99.64 % | 613585 | Neurodevelopmental disorder with facial dysmorphism, absent language, and pseudo-Pelger-Huet anomaly, 620075 (3), Autosomal recessive |
| **TMEM165** | 99.89 % | 614726 | Congenital disorder of glycosylation, type IIk, 614727 (3), Autosomal recessive |
| **TMEM199** | 99.93 % | 616815 | Congenital disorder of glycosylation, type IIp, 616829 (3), Autosomal recessive |
| **TMEM216** | 99.5 % | 613277 | Joubert syndrome 2, 608091 (3), Autosomal recessive; Retinitis pigmentosa 98, 620996 (3), Autosomal recessive; Meckel syndrome 2, 603194 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **TMEM218** | 99.92 % | 619285 | Joubert syndrome 39, 619562 (3), Autosomal recessive |
| **TMEM222** | 98.57 % | 619469 | Neurodevelopmental disorder with motor and speech delay and behavioral abnormalities, 619470 (3), Autosomal recessive |
| **TMEM231** | 93.22 % | 614949 | Joubert syndrome 20, 614970 (3), Autosomal recessive; Meckel syndrome 11, 615397 (3), Autosomal recessive |
| **TMEM237** | 99.12 % | 614423 | Joubert syndrome 14, 614424 (3), Autosomal recessive |
| **TMEM251** | 99.97 % | 619332 | Dysostosis multiplex, Ain-Naz type, 619345 (3), Autosomal recessive |
| **TMEM260** | 99.88 % | 617449 | Structural heart defects and renal anomalies syndrome, 617478 (3), Autosomal recessive |
| **TMEM38B** | 99.82 % | 611236 | Osteogenesis imperfecta, type XIV, 615066 (3), Autosomal recessive |
| **TMEM53** | 99.66 % | 619722 | Craniotubular dysplasia, Ikegawa type, 619727 (3), Autosomal recessive |
| **TMEM63C** | 99.93 % | 619953 | Spastic paraplegia 87, autosomal recessive, 619966 (3), Autosomal recessive |
| **TMEM65** | 93.99 % | 616609 | *No OMIM phenotypes* |
| **TMEM67** | 99.79 % | 609884 | Nephronophthisis 11, 613550 (3), Autosomal recessive; {Bardet-Biedl syndrome 14, modifier of}, 615991 (3), Autosomal recessive; Joubert syndrome 6, 610688 (3), Autosomal recessive; Meckel syndrome 3, 607361 (3), Autosomal recessive; ?RHYNS syndrome, 602152 (3), Autosomal recessive; COACH syndrome 1, 216360 (3), Autosomal recessive |
| **TMEM70** | 99.93 % | 612418 | Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052 (3), Autosomal recessive |
| **TMEM94** | 99.74 % | 618163 | Intellectual developmental disorder with cardiac defects and dysmorphic facies, 618316 (3), Autosomal recessive |
| **TMIE** | 98.91 % | 607237 | Deafness, autosomal recessive 6, 600971 (3), Autosomal recessive |
| **TMPRSS15** | 99.72 % | 606635 | Enterokinase deficiency, 226200 (3), Autosomal recessive |
| **TMPRSS3** | 99.9 % | 605511 | Deafness, autosomal recessive 8/10, 601072 (3), Autosomal recessive |
| **TMPRSS6** | 99.88 % | 609862 | Iron-refractory iron deficiency anemia, 206200 (3), Autosomal recessive |
| **TMTC3** | 98.1 % | 617218 | Lissencephaly 8, 617255 (3), Autosomal recessive |
| **TMX2** | 99.87 % | 616715 | Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730 (3), Autosomal recessive |
| **TNFRSF11A** | 99.9 % | 603499 | Osteopetrosis, autosomal recessive 7, 612301 (3), Autosomal recessive; {Paget disease of bone 2, early-onset}, 602080 (3), Autosomal dominant; Osteolysis, familial expansile, 174810 (3), Autosomal dominant |
| **TNFRSF11B** | 99.75 % | 602643 | Paget disease of bone 5, juvenile-onset, 239000 (3), Autosomal recessive |
| **TNFRSF13B** | 99.29 % | 604907 | Immunodeficiency, common variable, 2, 240500 (3), Autosomal recessive, Autosomal dominant; Immunoglobulin A deficiency 2, 609529 (3) |
| **TNFRSF9** | 99.88 % | 602250 | Immunodeficiency 109 with lymphoproliferation, 620282 (3), Autosomal recessive |
| **TNFSF11** | 99.68 % | 602642 | Osteopetrosis, autosomal recessive 2, 259710 (3), Autosomal recessive |
| **TNIK** | 99.78 % | 610005 | Intellectual developmental disorder, autosomal recessive 54, 617028 (3), Autosomal recessive |
| **TNNI3** | 99.84 % | 191044 | ?Cardiomyopathy, dilated, 2A, 611880 (3), Autosomal recessive; Cardiomyopathy, hypertrophic, 7, 613690 (3), Autosomal dominant; Cardiomyopathy, familial restrictive, 1, 115210 (3), Autosomal dominant; Cardiomyopathy, dilated, 1FF, 613286 (3) |
| **TNNT1** | 99.63 % | 191041 | Nemaline myopathy 5C, autosomal dominant, 620389 (3), Autosomal dominant; Nemaline myopathy 5A, autosomal recessive, severe infantile, 605355 (3), Autosomal recessive; Nemaline myopathy 5B, autosomal recessive, childhood-onset, 620386 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **TNR** | 99.23 % | 601995 | Neurodevelopmental disorder, nonprogressive, with spasticity and transient opisthotonus, 619653 (3), Autosomal recessive |
| **TNXB** | 89.61 % | 600985 | Ehlers-Danlos syndrome, classic-like, 1, 606408 (3), Autosomal recessive; Vesicoureteral reflux 8, 615963 (3), Autosomal dominant |
| **TOE1** | 99.73 % | 613931 | Pontocerebellar hypoplasia, type 7, 614969 (3), Autosomal recessive |
| **TOGARAM1** | 99.92 % | 617618 | Joubert syndrome 37, 619185 (3), Autosomal recessive |
| **TONSL** | 99.87 % | 604546 | Spondyloepimetaphyseal dysplasia, sponastrime type, 271510 (3), Autosomal recessive |
| **TOP3A** | 99.77 % | 601243 | Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098 (3), Autosomal recessive |
| **TOR1A** | 99.89 % | 605204 | {Dystonia-1, modifier of} (3); Arthrogryposis multiplex congenita 5, 618947 (3), Autosomal recessive; Dystonia-1, torsion, 128100 (3), Autosomal dominant |
| **TOR1AIP1** | 99.59 % | 614512 | ?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072 (3), Autosomal recessive |
| **TP53RK** | 99.89 % | 608679 | Galloway-Mowat syndrome 4, 617730 (3), Autosomal recessive |
| **TP73** | 99.86 % | 601990 | Ciliary dyskinesia, primary, 47, and lissencephaly, 619466 (3), Autosomal recessive |
| **TPI1** | 99.97 % | 190450 | Hemolytic anemia due to triosephosphate isomerase deficiency, 615512 (3), Autosomal recessive |
| **TPK1** | 99.98 % | 606370 | Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458 (3), Autosomal recessive |
| **TPM3** | 99.32 % | 191030 | Congenital myopathy 4A, autosomal dominant, 255310 (3), Autosomal dominant; Congenital myopathy 4B, autosomal recessive, 609284 (3), Autosomal recessive |
| **TPO** | 99.93 % | 606765 | Thyroid dyshormonogenesis 2A, 274500 (3), Autosomal recessive |
| **TPP1** | 99.95 % | 607998 | Ceroid lipofuscinosis, neuronal, 2, 204500 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 7, 609270 (3), Autosomal recessive |
| **TPP2** | 99.83 % | 190470 | Immunodeficiency 78 with autoimmunity and developmental delay, 619220 (3), Autosomal recessive |
| **TPR** | 98.91 % | 189940 | ?Intellectual developmental disorder, autosomal recessive 79, 620393 (3), Autosomal recessive |
| **TPRKB** | 95.96 % | 608680 | Galloway-Mowat syndrome 5, 617731 (3), Autosomal recessive |
| **TPRN** | 99.34 % | 613354 | Deafness, autosomal recessive 79, 613307 (3), Autosomal recessive |
| **TRAC** | 100 % | 186880 | Immunodeficiency 7, TCR-alpha/beta deficient, 615387 (3), Autosomal recessive |
| **TRAF3IP1** | 99.94 % | 607380 | Senior-Loken syndrome 9, 616629 (3), Autosomal recessive |
| **TRAF3IP2** | 99.96 % | 607043 | ?Candidiasis, familial, 8, 615527 (3), Autosomal recessive; {Psoriasis susceptibility 13}, 614070 (3) |
| **TRAIP** | 99.82 % | 605958 | Seckel syndrome 9, 616777 (3), Autosomal recessive |
| **TRAK1** | 99.91 % | 608112 | Developmental and epileptic encephalopathy 68, 618201 (3), Autosomal recessive |
| **TRAPPC10** | 84.32 % | 602103 | Neurodevelopmental disorder with microcephaly, short stature, and speech delay, 620027 (3), Autosomal recessive |
| **TRAPPC11** | 99.92 % | 614138 | Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356 (3), Autosomal recessive |
| **TRAPPC12** | 99.69 % | 614139 | Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669 (3), Autosomal recessive |
| **TRAPPC14** | 99.25 % | 618350 | ?Microcephaly 25, primary, autosomal recessive, 618351 (3), Autosomal recessive |
| **TRAPPC2** | 99.47 % | 300202 | Spondyloepiphyseal dysplasia tarda, 313400 (3), X-linked recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **TRAPPC2L** | 100 % | 610970 | Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331 (3), Autosomal recessive |
| **TRAPPC4** | 99.88 % | 610971 | Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy, 618741 (3), Autosomal recessive |
| **TRAPPC6B** | 99.36 % | 610397 | Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862 (3), Autosomal recessive |
| **TRAPPC9** | 99.85 % | 611966 | Intellectual developmental disorder, autosomal recessive 13, 613192 (3), Autosomal recessive |
| **TRDN** | 99.9 % | 603283 | Cardiac arrhythmia syndrome, with or without skeletal muscle weakness, 615441 (3), Autosomal recessive |
| **TREM2** | 99.99 % | 605086 | {Alzheimer disease 17, susceptibility to}, 615080 (3), Autosomal recessive; Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193 (3), Autosomal recessive |
| **TREX1** | 100 % | 606609 | Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 (3), Autosomal dominant; Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 (3), Autosomal recessive, Autosomal dominant; {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant; Chilblain lupus, 610448 (3), Autosomal dominant |
| **TRHR** | 99.99 % | 188545 | Hypothyroidism, congenital, nongoitrous, 7, 618573 (3), Autosomal recessive |
| **TRIM2** | 99.87 % | 614141 | Charcot-Marie-Tooth disease, type 2R, 615490 (3), Autosomal recessive |
| **TRIM32** | 99.99 % | 602290 | ?Bardet-Biedl syndrome 11, 615988 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110 (3), Autosomal recessive |
| **TRIM36** | 99.78 % | 609317 | ?Anencephaly 1, 206500 (3), Autosomal recessive |
| **TRIM37** | 97.98 % | 605073 | Mulibrey nanism, 253250 (3), Autosomal recessive |
| **TRIOBP** | 99.87 % | 609761 | Deafness, autosomal recessive 28, 609823 (3), Autosomal recessive |
| **TRIP11** | 99.89 % | 604505 | Odontochondrodysplasia 1, 184260 (3), Autosomal recessive; Achondrogenesis, type IA, 200600 (3), Autosomal recessive |
| **TRIP13** | 99.98 % | 604507 | Oocyte/zygote/embryo maturation arrest 9, 619011 (3), Autosomal recessive; Mosaic variegated aneuploidy syndrome 3, 617598 (3), Autosomal recessive |
| **TRIP4** | 99.64 % | 604501 | ?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066 (3), Autosomal recessive; Spinal muscular atrophy with congenital bone fractures 1, 616866 (3), Autosomal recessive |
| **TRIT1** | 98.3 % | 617840 | Combined oxidative phosphorylation deficiency 35, 617873 (3), Autosomal recessive |
| **TRMT1** | 99.93 % | 611669 | Intellectual developmental disorder, autosomal recessive 68, 618302 (3), Autosomal recessive |
| **TRMT10A** | 99.86 % | 616013 | Microcephaly, short stature, and impaired glucose metabolism 1, 616033 (3), Autosomal recessive |
| **TRMT10C** | 99.84 % | 615423 | Combined oxidative phosphorylation deficiency 30, 616974 (3), Autosomal recessive |
| **TRMT5** | 99.99 % | 611023 | Peripheral neuropathy with variable spasticity, exercise intolerance, and developmental delay, 616539 (3), Autosomal recessive |
| **TRMU** | 99.84 % | 610230 | {Deafness, mitochondrial, modifier of}, 580000 (3), Mitochondrial; Liver failure, transient infantile, 613070 (3), Autosomal recessive |
| **TRNT1** | 99.91 % | 612907 | Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 (3), Autosomal recessive; Retinitis pigmentosa and erythrocytic microcytosis, 616959 (3), Autosomal recessive |
| **TRPM1** | 99.09 % | 603576 | Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216 (3), Autosomal recessive |
| **TRPM6** | 99.74 % | 607009 | Hypomagnesemia 1, intestinal, 602014 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **TRPV6** | 99.85 % | 606680 | Hyperparathyroidism, transient neonatal, 618188 (3), Autosomal recessive |
| **TSEN15** | 98.98 % | 608756 | Pontocerebellar hypoplasia, type 2F, 617026 (3), Autosomal recessive |
| **TSEN2** | 98.82 % | 608753 | Pontocerebellar hypoplasia type 2B, 612389 (3), Autosomal recessive |
| **TSEN34** | 99.94 % | 608754 | ?Pontocerebellar hypoplasia type 2C, 612390 (3), Autosomal recessive |
| **TSEN54** | 99.85 % | 608755 | Pontocerebellar hypoplasia type 2A, 277470 (3), Autosomal recessive; Pontocerebellar hypoplasia type 4, 225753 (3), Autosomal recessive; ?Pontocerebellar hypoplasia type 5, 610204 (3), Autosomal recessive |
| **TSFM** | 99.97 % | 604723 | Combined oxidative phosphorylation deficiency 3, 610505 (3), Autosomal recessive |
| **TSHB** | 99.49 % | 188540 | Hypothyroidism, congenital, nongoitrous 4, 275100 (3), Autosomal recessive |
| **TSHR** | 99.9 % | 603372 | Hyperthyroidism, familial gestational, 603373 (3), Autosomal dominant; Hyperthyroidism, nonautoimmune, 609152 (3), Autosomal dominant; Thyroid adenoma, hyperfunctioning, somatic, 609152 (3); Hypothyroidism, congenital, nongoitrous, 1, 275200 (3), Autosomal recessive; Thyroid carcinoma with thyrotoxicosis, somatic, 609152 (3) |
| **TSPAN7** | 99.94 % | 300096 | Intellectual developmental disorder, X-linked 58, 300210 (3), X-linked recessive |
| **TSPEAR** | 99.99 % | 612920 | Tooth agenesis, selective, 10, 620173 (3), Autosomal recessive; ?Deafness, autosomal recessive 98, 614861 (3), Autosomal recessive; Ectodermal dysplasia 14, hypohidrotic/hair/tooth/nail type, 618180 (3), Autosomal recessive |
| **TSPOAP1** | 99.17 % | 610764 | Dystonia 22, juvenile-onset, 620453 (3), Autosomal recessive; ?Dystonia 22, adult-onset, 620456 (3), Autosomal recessive |
| **TSPYL1** | 100 % | 604714 | Sudden infant death with dysgenesis of the testes syndrome, 608800 (3), Autosomal recessive |
| **TTC12** | 99.9 % | 610732 | Ciliary dyskinesia, primary, 45, 618801 (3), Autosomal recessive |
| **TTC19** | 99.92 % | 613814 | Mitochondrial complex III deficiency, nuclear type 2, 615157 (3), Autosomal recessive |
| **TTC21B** | 99.13 % | 612014 | Short-rib thoracic dysplasia 4 with or without polydactyly, 613819 (3), Autosomal recessive; Nephronophthisis 12, 613820 (3), Autosomal recessive, Autosomal dominant |
| **TTC26** | 99.82 % | 617453 | Biliary, renal, neurologic, and skeletal syndrome, 619534 (3), Autosomal recessive |
| **TTC37** | 99.79 % | 614589 | Trichohepatoenteric syndrome 1, 222470 (3), Autosomal recessive |
| **TTC5** | 99.82 % | 619014 | Neurodevelopmental disorder with cerebral atrophy and variable facial dysmorphism, 619244 (3), Autosomal recessive |
| **TTC7A** | 99.02 % | 609332 | Gastrointestinal defects and immunodeficiency syndrome, 243150 (3), Autosomal recessive |
| **TTC8** | 99.76 % | 608132 | Bardet-Biedl syndrome 8, 615985 (3), Autosomal recessive; ?Retinitis pigmentosa 51, 613464 (3), Autosomal recessive |
| **TTI1** | 99.98 % | 614425 | Neurodevelopmental disorder with microcephaly and movement abnormalities, 620445 (3), Autosomal recessive |
| **TTI2** | 99.92 % | 614426 | Intellectual developmental disorder, autosomal recessive 39, 615541 (3), Autosomal recessive |
| **TTLL5** | 99.89 % | 612268 | Cone-rod dystrophy 19, 615860 (3), Autosomal recessive |
| **TTN** | 99.03 % | 188840 | Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807 (3), Autosomal recessive; Congenital myopathy 5 with cardiomyopathy, 611705 (3), Autosomal recessive; Tibial muscular dystrophy, tardive, 600334 (3), Autosomal dominant; Cardiomyopathy, dilated, 1G, 604145 (3), Autosomal dominant; ?Cardiomyopathy, familial hypertrophic, 9, 613765 (3), Autosomal dominant; Myopathy myofibrillar, 9, with early respiratory failure, 603689 (3), Autosomal dominant |
| **TTPA** | 99.55 % | 600415 | Ataxia with isolated vitamin E deficiency, 277460 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **TUBGCP2** | 99.71 % | 617817 | Pachygyria, microcephaly, developmental delay, and dysmorphic facies, with or without seizures, 618737 (3), Autosomal recessive |
| **TUBGCP4** | 99.8 % | 609610 | Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335 (3), Autosomal recessive |
| **TUBGCP6** | 99.93 % | 610053 | Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270 (3), Autosomal recessive |
| **TUFM** | 99.97 % | 602389 | Combined oxidative phosphorylation deficiency 4, 610678 (3), Autosomal recessive |
| **TUFT1** | 98.13 % | 600087 | Woolly hair-skin fragility syndrome, 620415 (3), Autosomal recessive |
| **TULP1** | 99.84 % | 602280 | Leber congenital amaurosis 15, 613843 (3), Autosomal recessive; Retinitis pigmentosa 14, 600132 (3), Autosomal recessive |
| **TULP3** | 99.37 % | 604730 | Hepatorenocardiac degenerative fibrosis, 619902 (3), Autosomal recessive |
| **TUSC3** | 99.95 % | 601385 | Intellectual developmental disorder, autosomal recessive 7, 611093 (3), Autosomal recessive |
| **TWIST2** | 99.99 % | 607556 | Ablepharon-macrostomia syndrome, 200110 (3), Autosomal dominant; Barber-Say syndrome, 209885 (3), Autosomal dominant; Focal facial dermal dysplasia 3, Setleis type, 227260 (3), Autosomal recessive |
| **TWNK** | 99.91 % | 606075 | Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 (3), Autosomal dominant; Perrault syndrome 5, 616138 (3), Autosomal recessive |
| **TXNDC15** | 99.86 % | 617778 | Meckel syndrome 14, 619879 (3), Autosomal recessive |
| **TXNL4A** | 99.98 % | 611595 | Burn-McKeown syndrome, 608572 (3), Autosomal recessive |
| **TYK2** | 99.7 % | 176941 | Immunodeficiency 35, 611521 (3), Autosomal recessive |
| **TYMP** | 99.81 % | 131222 | Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041 (3), Autosomal recessive |
| **TYR** | 99.73 % | 606933 | [Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 (3), Autosomal dominant; [Skin/hair/eye pigmentation 3, blue/green eyes], 601800 (3), Autosomal dominant; {Melanoma, cutaneous malignant, susceptibility to, 8}, 601800 (3), Autosomal dominant; Albinism, oculocutaneous, type IB, 606952 (3), Autosomal recessive; Albinism, oculocutaneous, type IA, 203100 (3), Autosomal recessive |
| **TYROBP** | 99.88 % | 604142 | Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770 (3), Autosomal recessive |
| **TYRP1** | 99.57 % | 115501 | [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271 (3); Albinism, oculocutaneous, type III, 203290 (3), Autosomal recessive |
| **UBA1** | 99.76 % | 314370 | Spinal muscular atrophy, X-linked 2, infantile, 301830 (3), X-linked recessive; VEXAS syndrome, somatic, 301054 (3) |
| **UBA5** | 99.85 % | 610552 | ?Spinocerebellar ataxia, autosomal recessive 24, 617133 (3), Autosomal recessive; Developmental and epileptic encephalopathy 44, 617132 (3), Autosomal recessive |
| **UBE2A** | 99.61 % | 312180 | Intellectual developmental disorder, X-linked syndromic, Nascimento type, 300860 (3), X-linked recessive |
| **UBE2T** | 99.23 % | 610538 | Fanconi anemia, complementation group T, 616435 (3), Autosomal recessive |
| **UBE3B** | 99.89 % | 608047 | Kaufman oculocerebrofacial syndrome, 244450 (3), Autosomal recessive |
| **UBE4A** | 99.93 % | 603753 | Neurodevelopmental disorder with hypotonia and gross motor and speech delay, 619639 (3), Autosomal recessive |
| **UBR1** | 99.84 % | 605981 | Johanson-Blizzard syndrome, 243800 (3), Autosomal recessive |
| **UBR7** | 99.83 % | 613816 | Li-Campeau syndrome, 619189 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **UCHL1** | 99.94 % | 191342 | {?Parkinson disease 5, susceptibility to}, 613643 (3), Autosomal dominant; Spastic paraplegia 79A, autosomal dominant, 620221 (3), Autosomal dominant; Spastic paraplegia 79B, autosomal recessive, 615491 (3), Autosomal recessive |
| **UFC1** | 98.12 % | 610554 | Neurodevelopmental disorder with spasticity and poor growth, 618076 (3), Autosomal recessive |
| **UFM1** | 99.7 % | 610553 | Leukodystrophy, hypomyelinating, 14, 617899 (3), Autosomal recessive |
| **UFSP2** | 99.63 % | 611482 | ?Hip dysplasia, Beukes type, 142669 (3), Autosomal dominant; Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974 (3), Autosomal dominant; Developmental and epileptic encephalopathy 106, 620028 (3), Autosomal recessive |
| **UGDH** | 99.74 % | 603370 | Developmental and epileptic encephalopathy 84, 618792 (3), Autosomal recessive |
| **UGP2** | 99.39 % | 191760 | Developmental and epileptic encephalopathy 83, 618744 (3), Autosomal recessive |
| **UGT1A1** | 99.86 % | 191740 | Crigler-Najjar syndrome, type I, 218800 (3), Autosomal recessive; [Bilirubin, serum level of, QTL1], 601816 (3); Hyperbilirubinemia, familial transient neonatal, 237900 (3), Autosomal recessive, Autosomal dominant; Crigler-Najjar syndrome, type II, 606785 (3), Autosomal recessive; [Gilbert syndrome], 143500 (3), Autosomal recessive |
| **UMPS** | 99.76 % | 613891 | Orotic aciduria, 258900 (3), Autosomal recessive |
| **UNC13D** | 99.84 % | 608897 | Hemophagocytic lymphohistiocytosis, familial, 3, 608898 (3), Autosomal recessive |
| **UNC45A** | 99.78 % | 611219 | Osteootohepatoenteric syndrome, 619377 (3), Autosomal recessive |
| **UNC45B** | 99.91 % | 611220 | ?Cataract 43, 616279 (3), Autosomal dominant; Myofibrillar myopathy 11, 619178 (3), Autosomal recessive |
| **UNC80** | 99.76 % | 612636 | Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801 (3), Autosomal recessive |
| **UNC93B1** | 97.3 % | 608204 | {Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 1}, 610551 (3), Autosomal recessive |
| **UNG** | 99.77 % | 191525 | Immunodeficiency with hyper IgM, type 5, 608106 (3), Autosomal recessive |
| **UPB1** | 99.88 % | 606673 | Beta-ureidopropionase deficiency, 613161 (3), Autosomal recessive |
| **UPF3B** | 99.43 % | 300298 | Intellectual developmental disorder, X-linked syndromic 14, 300676 (3), X-linked recessive |
| **UQCRB** | 99.09 % | 191330 | Mitochondrial complex III deficiency, nuclear type 3, 615158 (3), Autosomal recessive |
| **UQCRC2** | 97.46 % | 191329 | Mitochondrial complex III deficiency, nuclear type 5, 615160 (3), Autosomal recessive |
| **UQCRFS1** | 99.2 % | 191327 | Mitochondrial complex III deficiency, nuclear type 10, 618775 (3), Autosomal recessive |
| **UQCRQ** | 99.87 % | 612080 | Mitochondrial complex III deficiency, nuclear type 4, 615159 (3), Autosomal recessive |
| **UROD** | 98.6 % | 613521 | Porphyria, hepatoerythropoietic, 176100 (3), Autosomal recessive, Autosomal dominant; Porphyria cutanea tarda, 176100 (3), Autosomal recessive, Autosomal dominant |
| **UROS** | 99.98 % | 606938 | Porphyria, congenital erythropoietic, 263700 (3), Autosomal recessive |
| **USB1** | 99.94 % | 613276 | Poikiloderma with neutropenia, 604173 (3), Autosomal recessive |
| **USH1C** | 99.96 % | 605242 | Usher syndrome, type 1C, 276904 (3), Autosomal recessive; Deafness, autosomal recessive 18A, 602092 (3), Autosomal recessive |
| **USH1G** | 99.97 % | 607696 | Usher syndrome, type 1G, 606943 (3), Autosomal recessive |
| **USH2A** | 99.74 % | 608400 | Usher syndrome, type 2A, 276901 (3), Autosomal recessive; Retinitis pigmentosa 39, 613809 (3), Autosomal recessive |
| **USP18** | 90.87 % | 607057 | Pseudo-TORCH syndrome 2, 617397 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **USP53** | 99.86 % | 617431 | Cholestasis, progressive familial intrahepatic, 7, with or without hearing loss, 619658 (3), Autosomal recessive |
| **USP9X** | 99.76 % | 300072 | Intellectual developmental disorder, X-linked 99, 300919 (3), X-linked recessive; Intellectual developmental disorder, X-linked 99, syndromic, female-restricted, 300968 (3), X-linked dominant |
| **UVSSA** | 99.92 % | 614632 | UV-sensitive syndrome 3, 614640 (3), Autosomal recessive |
| **VAC14** | 99.53 % | 604632 | Striatonigral degeneration, childhood-onset, 617054 (3), Autosomal recessive |
| **VAMP1** | 99.99 % | 185880 | Myasthenic syndrome, congenital, 25, 618323 (3), Autosomal recessive; Spastic ataxia 1, autosomal dominant, 108600 (3), Autosomal dominant |
| **VARS1** | 99.76 % | 192150 | Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802 (3), Autosomal recessive |
| **VARS2** | 99.67 % | 612802 | Combined oxidative phosphorylation deficiency 20, 615917 (3), Autosomal recessive |
| **VAX1** | 99.96 % | 604294 | ?Microphthalmia, syndromic 11, 614402 (3), Autosomal recessive |
| **VDR** | 99.61 % | 601769 | Rickets, vitamin D-resistant, type IIA, 277440 (3), Autosomal recessive |
| **VHL** | 99.98 % | 608537 | Hemangioblastoma, cerebellar, somatic (3); Erythrocytosis, familial, 2, 263400 (3), Autosomal recessive; von Hippel-Lindau syndrome, 193300 (3), Autosomal dominant; Renal cell carcinoma, somatic, 144700 (3); Pheochromocytoma, 171300 (3), Autosomal dominant |
| **VIPAS39** | 99.76 % | 613401 | Arthrogryposis, renal dysfunction, and cholestasis 2, 613404 (3), Autosomal recessive |
| **VKORC1** | 99.76 % | 608547 | Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473 (3), Autosomal recessive; Warfarin resistance, 122700 (3), Autosomal dominant |
| **VLDLR** | 99.94 % | 192977 | Cerebellar hypoplasia, impaired intellectual development, and dysequilibrium syndrome 1, 224050 (3), Autosomal recessive |
| **VMA21** | 99.74 % | 300913 | Myopathy, X-linked, with excessive autophagy, 310440 (3), X-linked recessive |
| **VPS11** | 99.92 % | 608549 | ?Dystonia 32, 619637 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 12, 616683 (3), Autosomal recessive |
| **VPS13A** | 99.72 % | 605978 | Choreoacanthocytosis, 200150 (3), Autosomal recessive |
| **VPS13B** | 99.81 % | 607817 | Cohen syndrome, 216550 (3), Autosomal recessive |
| **VPS13D** | 99.87 % | 608877 | Spinocerebellar ataxia, autosomal recessive 4, 607317 (3), Autosomal recessive |
| **VPS33A** | 99.08 % | 610034 | Mucopolysaccharidosis-plus syndrome, 617303 (3), Autosomal recessive |
| **VPS33B** | 99.9 % | 608552 | Keratoderma-ichthyosis-deafness syndrome, autosomal recessive, 620009 (3), Autosomal recessive; Cholestasis, progressive familial intrahepatic, 12, 620010 (3), Autosomal recessive; Arthrogryposis, renal dysfunction, and cholestasis 1, 208085 (3), Autosomal recessive |
| **VPS35L** | 98.94 % | 618981 | Ritscher-Schinzel syndrome 3, 619135 (3), Autosomal recessive |
| **VPS37A** | 99.78 % | 609927 | Spastic paraplegia 53, autosomal recessive, 614898 (3), Autosomal recessive |
| **VPS41** | 99.84 % | 605485 | Spinocerebellar ataxia, autosomal recessive 29, 619389 (3), Autosomal recessive |
| **VPS45** | 97.56 % | 610035 | Neutropenia, severe congenital, 5, autosomal recessive, 615285 (3), Autosomal recessive |
| **VPS50** | 98.46 % | 616465 | Neurodevelopmental disorder with microcephaly, seizures, and neonatal cholestasis, 619685 (3), Autosomal recessive |
| **VPS53** | 99.94 % | 615850 | Pontocerebellar hypoplasia, type 2E, 615851 (3), Autosomal recessive |
| **VRK1** | 99.91 % | 602168 | Pontocerebellar hypoplasia type 1A, 607596 (3), Autosomal recessive; Neuronopathy, distal hereditary motor, autosomal recessive 10, 620542 (3), Autosomal recessive |
| **VSX2** | 99.99 % | 142993 | Microphthalmia, isolated 2, 610093 (3), Autosomal recessive; Microphthalmia/coloboma 3, 610092 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **VWA1** | 99.7 % | 611901 | Neuronopathy, distal hereditary motor, autosomal recessive 7, 619216 (3), Autosomal recessive |
| **VWF** | 97.92 % | 613160 | von Willebrand disease, type 1, 193400 (3), Autosomal dominant; von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 (3), Autosomal recessive, Autosomal dominant; von Willebrand disease, type 3, 277480 (3), Autosomal recessive |
| **WARS1** | 99.77 % | 191050 | Neuronopathy, distal hereditary motor, autosomal dominant 9, 617721 (3), Autosomal dominant; Neurodevelopmental disorder with microcephaly and speech delay, with or without brain abnormalities, 620317 (3), Autosomal recessive |
| **WARS2** | 97.45 % | 604733 | Parkinsonism-dystonia 3, childhood-onset, 619738 (3), Autosomal recessive; Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710 (3), Autosomal recessive |
| **WAS** | 96.84 % | 300392 | Wiskott-Aldrich syndrome, 301000 (3), X-linked recessive; Neutropenia, severe congenital, X-linked, 300299 (3), X-linked recessive; Thrombocytopenia, X-linked, intermittent, 313900 (3), X-linked recessive; Thrombocytopenia, X-linked, 313900 (3), X-linked recessive |
| **WASHC4** | 99.85 % | 615748 | Intellectual developmental disorder, autosomal recessive 43, 615817 (3), Autosomal recessive |
| **WASHC5** | 99.89 % | 610657 | Ritscher-Schinzel syndrome 1, 220210 (3), Autosomal recessive; Spastic paraplegia 8, autosomal dominant, 603563 (3), Autosomal dominant |
| **WBP2** | 99.31 % | 606962 | Deafness, autosomal recessive 107, 617639 (3), Autosomal recessive |
| **WDPCP** | 99.79 % | 613580 | Bardet-Biedl syndrome 15, 615992 (3), Autosomal recessive; Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085 (3), Autosomal recessive |
| **WDR1** | 99.86 % | 604734 | Periodic fever, immunodeficiency, and thrombocytopenia syndrome, 150550 (3), Autosomal recessive |
| **WDR11** | 99.88 % | 606417 | Intellectual developmental disorder, autosomal recessive 78, 620237 (3), Autosomal recessive; Hypogonadotropic hypogonadism 14 with or without anosmia, 614858 (3), Autosomal dominant |
| **WDR19** | 99.62 % | 608151 | Nephronophthisis 13, 614377 (3), Autosomal recessive; Cranioectodermal dysplasia 4, 614378 (3), Autosomal recessive; Senior-Loken syndrome 8, 616307 (3), Autosomal recessive; Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 (3), Autosomal recessive; ?Spermatogenic failure 72, 619867 (3), Autosomal recessive |
| **WDR35** | 99.87 % | 613602 | Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 (3), Autosomal recessive; Cranioectodermal dysplasia 2, 613610 (3), Autosomal recessive |
| **WDR4** | 99.76 % | 605924 | Galloway-Mowat syndrome 6, 618347 (3), Autosomal recessive; Microcephaly, growth deficiency, seizures, and brain malformations, 618346 (3), Autosomal recessive |
| **WDR45B** | 99.91 % | 609226 | Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977 (3), Autosomal recessive |
| **WDR62** | 99.72 % | 613583 | Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317 (3), Autosomal recessive |
| **WDR72** | 96.5 % | 613214 | Amelogenesis imperfecta, type IIA3, 613211 (3), Autosomal recessive |
| **WDR73** | 99.9 % | 616144 | Galloway-Mowat syndrome 1, 251300 (3), Autosomal recessive |
| **WDR81** | 99.91 % | 614218 | Cerebellar ataxia, impaired intellectual development, and dysquilibrium syndrome 2, 610185 (3), Autosomal recessive; Hydrocephalus, congenital, 3, with brain anomalies, 617967 (3), Autosomal recessive |
| **WDR83OS** | 99.78 % | 618474 | Neurodevelopmental disorder with variable familial hypercholanemia, 621016 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **WFS1** | 99.9 % | 606201 | Deafness, autosomal dominant 6/14/38, 600965 (3), Autosomal dominant; ?Cataract 41, 116400 (3), Autosomal dominant; Wolfram-like syndrome, autosomal dominant, 614296 (3), Autosomal dominant; {Diabetes mellitus, noninsulin-dependent, association with}, 125853 (3), Autosomal dominant; Wolfram syndrome 1, 222300 (3), Autosomal recessive |
| **WHRN** | 99.62 % | 607928 | Deafness, autosomal recessive 31, 607084 (3), Autosomal recessive; Usher syndrome, type 2D, 611383 (3), Autosomal recessive |
| **WIPF1** | 99.69 % | 602357 | Wiskott-Aldrich syndrome 2, 614493 (3), Autosomal recessive |
| **WIPI2** | 99.9 % | 609225 | ?Intellectual developmental disorder with short stature and variable skeletal anomalies, 618453 (3), Autosomal recessive |
| **WLS** | 98.41 % | 611514 | Zaki syndrome, 619648 (3), Autosomal recessive |
| **WNK1** | 99.92 % | 605232 | Neuropathy, hereditary sensory and autonomic, type II, 201300 (3), Autosomal recessive; Pseudohypoaldosteronism, type IIC, 614492 (3), Autosomal dominant |
| **WNK3** | 99.8 % | 300358 | Prieto syndrome, 309610 (3), X-linked recessive |
| **WNT1** | 98.74 % | 164820 | {Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221 (3), Autosomal dominant; Osteogenesis imperfecta, type XV, 615220 (3), Autosomal recessive |
| **WNT10A** | 99.77 % | 606268 | Schopf-Schulz-Passarge syndrome, 224750 (3), Autosomal recessive; Tooth agenesis, selective, 4, 150400 (3), Autosomal recessive, Autosomal dominant; Ectodermal dysplasia 16 (odontoonychodermal dysplasia), 257980 (3), Autosomal recessive |
| **WNT10B** | 99.68 % | 601906 | Tooth agenesis, selective, 8, 617073 (3), Autosomal dominant; Split-hand/foot malformation 6, 225300 (3), Autosomal recessive |
| **WNT2B** | 98.84 % | 601968 | Diarrhea 9, 618168 (3), Autosomal recessive |
| **WNT3** | 99.83 % | 165330 | ?Tetra-amelia syndrome 1, 273395 (3), Autosomal recessive |
| **WNT4** | 99.61 % | 603490 | Mullerian aplasia and hyperandrogenism, 158330 (3), Autosomal dominant; SERKAL syndrome, 611812 (3), Autosomal recessive |
| **WNT7A** | 99.91 % | 601570 | Fuhrmann syndrome, 228930 (3), Autosomal recessive; Ulna and fibula, absence of, with severe limb deficiency, 276820 (3), Autosomal recessive; ?Santos syndrome, 613005 (3), Autosomal recessive |
| **WRAP53** | 99.62 % | 612661 | Dyskeratosis congenita, autosomal recessive 3, 613988 (3), Autosomal recessive |
| **WRN** | 99.83 % | 604611 | Werner syndrome, 277700 (3), Autosomal recessive |
| **WWOX** | 99.9 % | 605131 | Esophageal squamous cell carcinoma, somatic, 133239 (3); Developmental and epileptic encephalopathy 28, 616211 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 12, 614322 (3), Autosomal recessive |
| **XDH** | 99.67 % | 607633 | Xanthinuria, type I, 278300 (3), Autosomal recessive |
| **XIAP** | 99.62 % | 300079 | Lymphoproliferative syndrome, X-linked, 2, 300635 (3), X-linked recessive |
| **XPA** | 99.78 % | 611153 | Xeroderma pigmentosum, group A, 278700 (3), Autosomal recessive |
| **XPC** | 99.93 % | 613208 | Xeroderma pigmentosum, group C, 278720 (3), Autosomal recessive |
| **XPNPEP3** | 99.89 % | 613553 | Nephronophthisis-like nephropathy 1, 613159 (3), Autosomal recessive |
| **XRCC1** | 99.75 % | 194360 | ?Spinocerebellar ataxia, autosomal recessive 26, 617633 (3), Autosomal recessive |
| **XRCC2** | 100 % | 600375 | Spermatogenic failure 50, 619145 (3), Autosomal recessive; ?Premature ovarian failure 17, 619146 (3), Autosomal recessive; ?Fanconi anemia, complementation group U, 617247 (3), Autosomal recessive |
| **XRCC4** | 99.87 % | 194363 | Short stature, microcephaly, and endocrine dysfunction, 616541 (3), Autosomal recessive |
| **XYLT1** | 99.79 % | 608124 | Desbuquois dysplasia 2, 615777 (3), Autosomal recessive; {Pseudoxanthoma elasticum, modifier of severity of}, 264800 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **XYLT2** | 99.69 % | 608125 | {Pseudoxanthoma elasticum, modifier of severity of}, 264800 (3), Autosomal recessive; Spondyloocular syndrome, 605822 (3), Autosomal recessive |
| **YARS1** | 98.32 % | 603623 | Infantile-onset multisystem neurologic, endocrine, and pancreatic disease 2, 619418 (3), Autosomal recessive; Charcot-Marie-Tooth disease, dominant intermediate C, 608323 (3), Autosomal dominant |
| **YARS2** | 99.45 % | 610957 | Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561 (3), Autosomal recessive |
| **YIF1B** | 99.35 % | 619109 | Kaya-Barakat-Masson syndrome, 619125 (3), Autosomal recessive |
| **YIPF5** | 99.82 % | 611483 | Microcephaly, epilepsy, and diabetes syndrome 2, 619278 (3), Autosomal recessive |
| **YRDC** | 98.59 % | 612276 | Galloway-Mowat syndrome 10, 619609 (3), Autosomal recessive |
| **YY1AP1** | 99.56 % | 607860 | Grange syndrome, 602531 (3), Autosomal recessive |
| **ZAP70** | 99.08 % | 176947 | Immunodeficiency 48, 269840 (3), Autosomal recessive; Autoimmune disease, multisystem, infantile-onset, 2, 617006 (3), Autosomal recessive |
| **ZBTB24** | 99.92 % | 614064 | Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069 (3), Autosomal recessive |
| **ZC4H2** | 99.83 % | 300897 | Wieacker-Wolff syndrome, 314580 (3), X-linked recessive; Wieacker-Wolff syndrome, female-restricted, 301041 (3), X-linked dominant |
| **ZDHHC9** | 99.61 % | 300646 | Intellectual developmental disorder, X-linked syndromic, Raymond type, 300799 (3), X-linked |
| **ZFP57** | 99.97 % | 612192 | Diabetes mellitus, transient neonatal 1, 601410 (3), Autosomal recessive, Autosomal dominant |
| **ZFYVE19** | 99.85 % | 619635 | Cholestasis, progressive familial intrahepatic, 9, 619849 (3), Autosomal recessive |
| **ZFYVE26** | 99.91 % | 612012 | Spastic paraplegia 15, autosomal recessive, 270700 (3), Autosomal recessive |
| **ZIC3** | 99.87 % | 300265 | Congenital heart defects, nonsyndromic, multiple types, 1, X-linked, 306955 (3), X-linked recessive; Heterotaxy, visceral, 1, X-linked, 306955 (3), X-linked recessive; VACTERL association, X-linked, 314390 (3), X-linked recessive |
| **ZMPSTE24** | 97.94 % | 606480 | Mandibuloacral dysplasia with type B lipodystrophy, 608612 (3), Autosomal recessive; Restrictive dermopathy 1, 275210 (3), Autosomal recessive |
| **ZMYM3** | 99.84 % | 300061 | Intellectual developmental disorder, X-linked 112, 301111 (3), X-linked recessive |
| **ZMYND10** | 99.88 % | 607070 | Ciliary dyskinesia, primary, 22, 615444 (3), Autosomal recessive |
| **ZNF142** | 99.92 % | 604083 | Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425 (3), Autosomal recessive |
| **ZNF335** | 99.92 % | 610827 | Microcephaly 10, primary, autosomal recessive, 615095 (3), Autosomal recessive |
| **ZNF341** | 99.78 % | 618269 | Hyper-IgE syndrome 3, autosomal recessive, with recurrent infections, 618282 (3), Autosomal recessive |
| **ZNF407** | 99.97 % | 615894 | SIMHA syndrome, 619557 (3), Autosomal recessive |
| **ZNF408** | 99.91 % | 616454 | Retinitis pigmentosa 72, 616469 (3), Autosomal recessive; ?Exudative vitreoretinopathy 6, 616468 (3), Autosomal dominant |
| **ZNF423** | 99.92 % | 604557 | Nephronophthisis 14, 614844 (3), Autosomal recessive, Autosomal dominant; Joubert syndrome 19, 614844 (3), Autosomal recessive, Autosomal dominant |
| **ZNF469** | 99.95 % | 612078 | Brittle cornea syndrome 1, 229200 (3), Autosomal recessive |
| **ZNF513** | 99.29 % | 613598 | ?Retinitis pigmentosa 58, 613617 (3), Autosomal recessive |
| **ZNF526** | 99.97 % | 614387 | Dentici-Novelli neurodevelopmental syndrome, 619877 (3), Autosomal recessive |
| **ZNF699** | 99.98 % | 609571 | DEGCAGS syndrome, 619488 (3), Autosomal recessive |
| **ZNF711** | 99.59 % | 314990 | Intellectual developmental disorder, X-linked 97, 300803 (3), X-linked |
| **ZNFX1** | 99.95 % | 618931 | Immunodeficiency 91 and hyperinflammation, 619644 (3), Autosomal recessive |
| **ZNHIT3** | 64.64 % | 604500 | PEHO syndrome, 260565 (3), Autosomal recessive |

## Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2025-11-12**

Gene symbols used are according to the HGNC guidelines (corresponding to Ensembl database release 105).

Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.

Possible phenotype mapping keys

(1) the disorder is placed on the map based on its association with a gene, but the underlying defect is not known

(2) the disorder has been placed on the map by linkage; no mutation has been found

(3) the molecular basis for the disorder is known; a mutation has been found in the gene

(4) a contiguous gene deletion or duplication syndrome, multiple genes are deleted or duplicated causing the phenotype

Brackets, "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values (e.g., dysalbuminemic euthyroidal hyperthyroxinemia).

Braces, "{ }", indicate mutations that contribute to susceptibility to multifactorial disorders (e.g., diabetes, asthma) or to susceptibility to infection (e.g., malaria).

A question mark, "?", before the phenotype name indicates that the relationship between the phenotype and gene is provisional. More details about this relationship are provided in the comment field of the map and in the gene and phenotype OMIM entries.

* The column '% at least 20 x covered' shows the percentage of the coding sequence (+/-20 nucleotides of the flanking introns) of that gene that is on average at least 20 x covered. This according to the experience with exome sequencing in our laboratory and based on the current method.