# Fetal anomalies

Gene panel

## Gene panel information

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| **Gene panel** | **Fetal anomalies** |
| **Version** | 7 |
| **Total genes** | 2116 |
| **Activation date** | Monday 29 june 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 |
| **ABCA12** | 99.81 % | 607800 | Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500 (3), Autosomal recessive; Ichthyosis, congenital, autosomal recessive 4A, 601277 (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 |
| **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 |
| **ABCD4** | 99.8 % | 603214 | Methylmalonic aciduria and homocystinuria, cblJ type, 614857 (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 |
| **ABI2** | 99.34 % | 606442 | *No OMIM phenotypes* |
| **ABL1** | 99.94 % | 189980 | Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 608232 (3), Somatic mutation; Congenital heart defects and skeletal malformations syndrome, 617602 (3), Autosomal dominant |
| **ACAD9** | 100 % | 611103 | Mitochondrial complex I deficiency, nuclear type 20, 611126 (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 |
| **ACBD6** | 97.92 % | 616352 | Neurodevelopmental disorder with progressive movement abnormalities, 620785 (3), Autosomal recessive |

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|---|---|---|---|
| **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) |
| **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 |
| **ACP5** | 99.91 % | 171640 | Spondyloenchondrodysplasia with immune dysregulation, 607944 (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 |
| **ACTA2** | 99.81 % | 102620 | Smooth muscle dysfunction syndrome, 613834 (3), Autosomal dominant; Aortic aneurysm, familial thoracic 6, 611788 (3), Autosomal dominant; Moyamoya disease 5, 614042 (3) |
| **ACTB** | 99.98 % | 102630 | Baraitser-Winter syndrome 1, 243310 (3), Autosomal dominant; Becker nevus, syndromic or isolated, somatic mosaic, 604919 (3); Thrombocytopenia 8, with dysmorphic features and developmental delay, 620475 (3), Autosomal dominant; Dystonia-deafness syndrome 1, 607371 (3), Autosomal dominant; Congenital smooth muscle hamartoma with or without hemihypertrophy, somatic mosaic, 620470 (3) |
| **ACTC1** | 99.97 % | 102540 | Left ventricular noncompaction 4, 613424 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 11, 612098 (3), Autosomal dominant; Atrial septal defect 5, 612794 (3), Autosomal dominant; Cardiomyopathy, dilated, 1R, 613424 (3), Autosomal dominant |
| **ACTG1** | 100 % | 102560 | Deafness, autosomal dominant 20/26, 604717 (3), Autosomal dominant; Baraitser-Winter syndrome 2, 614583 (3), Autosomal dominant |
| **ACTG2** | 99.94 % | 102545 | Megacystis-microcolon-intestinal hypoperistalsis syndrome 5, 619431 (3), Autosomal dominant; Visceral myopathy 1, 155310 (3), Autosomal dominant |
| **ACVR1** | 99.67 % | 102576 | Fibrodysplasia ossificans progressiva, 135100 (3), Autosomal dominant |
| **ACVRL1** | 99.38 % | 601284 | Telangiectasia, hereditary hemorrhagic, type 2, 600376 (3), Autosomal dominant |
| **ACY1** | 100 % | 104620 | Aminoacylase 1 deficiency, 609924 (3), Autosomal recessive |
| **ADAMTS10** | 99.84 % | 608990 | Weill-Marchesani syndrome 1, recessive, 277600 (3), Autosomal recessive |
| **ADAMTS15** | 99.88 % | 607509 | Arthrogryposis, distal, type 12, 620545 (3), Autosomal recessive |
| **ADAMTS17** | 99.86 % | 607511 | Weill-Marchesani 4 syndrome, recessive, 613195 (3), Autosomal recessive |
| **ADAMTS19** | 99.77 % | 607513 | Cardiac valvular dysplasia 2, 620067 (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 |
| **ADAR** | 98.86 % | 146920 | Dyschromatosis symmetrica hereditaria, 127400 (3), Autosomal dominant; Aicardi-Goutieres syndrome 6, 615010 (3), Autosomal recessive |
| **ADAT3** | 99.28 % | 615302 | Neurodevelopmental disorder with brain abnormalities, poor growth, and dysmorphic facies, 615286 (3), Autosomal recessive |
| **ADCY6** | 99.71 % | 600294 | Lethal congenital contracture syndrome 8, 616287 (3), Autosomal recessive |
| **ADD1** | 99.91 % | 102680 | {Hypertension, essential, salt-sensitive}, 145500 (3), Multifactorial |

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|---|---|---|---|
| **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 |
| **ADNP** | 100 % | 611386 | Helsmoortel-van der Aa syndrome, 615873 (3), Autosomal dominant |
| **ADSL** | 99.71 % | 608222 | Adenylosuccinase deficiency, 103050 (3), Autosomal recessive |
| **AFF3** | 99.09 % | 601464 | KINSSHIP syndrome, 619297 (3), Autosomal dominant |
| **AFF4** | 99.87 % | 604417 | CHOPS syndrome, 616368 (3), Autosomal dominant |
| **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 |
| **AGPS** | 98.17 % | 603051 | Rhizomelic chondrodysplasia punctata, type 3, 600121 (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 |
| **AGTR1** | 99.96 % | 106165 | {Hypertension, essential}, 145500 (3), Multifactorial; Renal tubular dysgenesis, 267430 (3), Autosomal recessive |
| **AHCY** | 99.93 % | 180960 | Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752 (3), Autosomal recessive |
| **AHDC1** | 99.99 % | 615790 | Xia-Gibbs syndrome, 615829 (3), Autosomal dominant |
| **AHI1** | 99.8 % | 608894 | Joubert syndrome 3, 608629 (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 |
| **AKT1** | 99.96 % | 164730 | Proteus syndrome, somatic mosaic, 176920 (3); Breast cancer, somatic, 114480 (3); Cowden syndrome 6, 615109 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3); Ovarian cancer, somatic, 167000 (3) |
| **AKT2** | 99.9 % | 164731 | Diabetes mellitus, type II, 125853 (3), Autosomal dominant; Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900 (3), Autosomal dominant |
| **AKT3** | 99.57 % | 611223 | Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937 (3), 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 |
| **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 |
| **ALG1** | 99.58 % | 605907 | Congenital disorder of glycosylation, type Ik, 608540 (3), Autosomal recessive |
| **ALG11** | 98.83 % | 613666 | Congenital disorder of glycosylation, type Ip, 613661 (3), Autosomal recessive |

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|---|---|---|---|
| **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 |
| **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 |
| **AMER1** | 99.95 % | 300647 | Osteopathia striata with cranial sclerosis, 300373 (3), X-linked dominant |
| **AMMECR1** | 99.72 % | 300195 | Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990 (3), X-linked recessive |
| **AMOT** | 99.8 % | 300410 | *No OMIM phenotypes* |
| **AMOTL1** | 99.58 % | 614657 | Craniofaciocardiohepatic syndrome, 621192 (3), Autosomal dominant |
| **AMPD2** | 99 % | 102771 | Pontocerebellar hypoplasia, type 9, 615809 (3), Autosomal recessive; ?Spastic paraplegia 63, autosomal recessive, 615686 (3), Autosomal recessive |
| **AMT** | 99.98 % | 238310 | Glycine encephalopathy 2, 620398 (3), Autosomal recessive |
| **ANAPC1** | 72.56 % | 608473 | Rothmund-Thomson syndrome, type 1, 618625 (3), Autosomal recessive |
| **ANGPT2** | 99.86 % | 601922 | Lymphatic malformation 10, 619369 (3), Autosomal dominant |
| **ANKH** | 99.76 % | 605145 | Chondrocalcinosis 2, 118600 (3), Autosomal dominant; Craniometaphyseal dysplasia, 123000 (3), Autosomal dominant |
| **ANKLE2** | 99.94 % | 616062 | Microcephaly 16, primary, autosomal recessive, 616681 (3), Autosomal recessive |
| **ANKRD11** | 99.54 % | 611192 | KBG syndrome, 148050 (3), Autosomal dominant |

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|---|---|---|---|
| **ANKRD17** | 99.81 % | 615929 | Chopra-Amiel-Gordon syndrome, 619504 (3), Autosomal dominant |
| **ANKRD26** | 99.79 % | 610855 | Thrombocytopenia 2, 188000 (3), Autosomal dominant |
| **ANKS6** | 99.91 % | 615370 | Nephronophthisis 16, 615382 (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 |
| **AP1S2** | 99.72 % | 300629 | Pettigrew syndrome, 304340 (3), X-linked recessive |
| **AP3B2** | 99.97 % | 602166 | Developmental and epileptic encephalopathy 48, 617276 (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 |
| **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 |
| **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 |
| **ARCN1** | 99.9 % | 600820 | Short stature-micrognathia syndrome, 617164 (3), Autosomal dominant |
| **ARF1** | 98.49 % | 103180 | Periventricular nodular heterotopia 8, 618185 (3), Autosomal dominant |
| **ARF3** | 99.33 % | 103190 | *No OMIM phenotypes* |
| **ARFGEF2** | 99.88 % | 605371 | Periventricular heterotopia with microcephaly, 608097 (3), Autosomal recessive |
| **ARHGAP29** | 96.3 % | 610496 | *No OMIM phenotypes* |
| **ARHGAP31** | 99.98 % | 610911 | Adams-Oliver syndrome 1, 100300 (3), Autosomal dominant |
| **ARHGEF40** | 99.86 % | 610018 | *No OMIM phenotypes* |
| **ARID1A** | 99.61 % | 603024 | Coffin-Siris syndrome 2, 614607 (3), Autosomal dominant |
| **ARID1B** | 99.8 % | 614556 | Coffin-Siris syndrome 1, 135900 (3), Autosomal dominant |
| **ARID2** | 99.49 % | 609539 | Coffin-Siris syndrome 6, 617808 (3), Autosomal dominant |
| **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 |
| **ARSA** | 99.83 % | 607574 | Metachromatic leukodystrophy, 250100 (3), Autosomal recessive |
| **ARSB** | 99.79 % | 611542 | Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200 (3), Autosomal recessive |
| **ARSL** | 99.68 % | 300180 | Chondrodysplasia punctata, X-linked recessive, 302950 (3), X-linked recessive |

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|---|---|---|---|
| **ARV1** | 99.82 % | 611647 | Developmental and epileptic encephalopathy 38, 617020 (3), Autosomal recessive |
| **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 |
| **ASNS** | 99.17 % | 108370 | Asparagine synthetase deficiency, 615574 (3), Autosomal recessive |
| **ASPA** | 99.88 % | 608034 | Canavan disease, 271900 (3), Autosomal recessive |
| **ASPM** | 99.52 % | 605481 | Microcephaly 5, primary, autosomal recessive, 608716 (3), Autosomal recessive |
| **ASS1** | 99.83 % | 603470 | Citrullinemia, 215700 (3), Autosomal recessive |
| **ASXL1** | 99.93 % | 612990 | Myelodysplastic syndrome, somatic, 614286 (3); Bohring-Opitz syndrome, 605039 (3), Autosomal dominant |
| **ASXL2** | 99.83 % | 612991 | Shashi-Pena syndrome, 617190 (3), Autosomal dominant |
| **ASXL3** | 99.96 % | 615115 | Bainbridge-Ropers syndrome, 615485 (3), Autosomal dominant |
| **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 |
| **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 |
| **ATN1** | 99.82 % | 607462 | Dentatorubral-pallidoluysian atrophy, 125370 (3), Autosomal dominant; Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494 (3), Autosomal dominant |
| **ATP11C** | 99.66 % | 300516 | ?Hemolytic anemia, congenital, X-linked, 301015 (3), X-linked 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 |
| **ATP1A3** | 99.8 % | 182350 | Alternating hemiplegia of childhood 2, 614820 (3), Autosomal dominant; Dystonia-12, 128235 (3), Autosomal dominant; CAPOS syndrome, 601338 (3), Autosomal dominant; Developmental and epileptic encephalopathy 99, 619606 (3), Autosomal dominant |
| **ATP5PO** | 99.76 % | 600828 | Mitochondrial complex V (ATP synthase) deficiency, nuclear type 7, 620359 (3), Autosomal 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 |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **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 |
| **ATR** | 99.75 % | 601215 | Seckel syndrome 1, 210600 (3), Autosomal recessive; ?Cutaneous telangiectasia and cancer syndrome, familial, 614564 (3), Autosomal dominant |
| **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 |
| **AXIN1** | 99.91 % | 603816 | Hepatocellular carcinoma, somatic, 114550 (3); Craniometadiaphyseal osteosclerosis with hip dysplasia, 620558 (3), Autosomal recessive; ?Caudal duplication anomaly, 607864 (3) |
| **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 |
| **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 |
| **BAIAP2** | 99.86 % | 605475 | *No OMIM phenotypes* |
| **BANF1** | 97.05 % | 603811 | Nestor-Guillermo progeria syndrome, 614008 (3), Autosomal recessive |
| **BAZ2B** | 99.37 % | 605683 | *No OMIM phenotypes* |
| **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 |
| **BCL11A** | 99.95 % | 606557 | Dias-Logan syndrome, 617101 (3), Autosomal dominant |
| **BCL9L** | 99.81 % | 609004 | *No OMIM phenotypes* |
| **BCOR** | 99.92 % | 300485 | Microphthalmia, syndromic 2, 300166 (3), X-linked dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **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 |
| **BFSP2** | 99.91 % | 603212 | Cataract 12, multiple types, 611597 (3), Autosomal dominant |
| **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 |
| **BHLHE22** | 99.98 % | 613483 | *No OMIM phenotypes* |
| **BICD2** | 99.95 % | 609797 | Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291 (3), Autosomal dominant; Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290 (3), Autosomal dominant |
| **BICRA** | 99.71 % | 605690 | Coffin-Siris syndrome 12, 619325 (3), Autosomal dominant |
| **BIN1** | 99.35 % | 601248 | Centronuclear myopathy 2, 255200 (3), Autosomal recessive |
| **BLM** | 99.87 % | 604610 | Bloom syndrome, 210900 (3), Autosomal recessive |
| **BLOC1S6** | 99.66 % | 604310 | Hermansky-Pudlak syndrome 9, 614171 (3), Autosomal recessive |
| **BMP1** | 99.61 % | 112264 | Osteogenesis imperfecta, type XIII, 614856 (3), Autosomal recessive |
| **BMP2** | 99.76 % | 112261 | Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 1, 617877 (3), Autosomal dominant; Brachydactyly, type A2, 112600 (3), Autosomal dominant; {HFE hemochromatosis, modifier of}, 235200 (3), Autosomal recessive |
| **BMP4** | 99.99 % | 112262 | Orofacial cleft 11, 600625 (3), Autosomal dominant; Microphthalmia, syndromic 6, 607932 (3), Autosomal dominant |
| **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 |
| **BNC2** | 99.85 % | 608669 | Lower urinary tract obstruction, congenital, 618612 (3), Autosomal dominant |
| **BNIP1** | 99.84 % | 603291 | Spondylopeiphyseal dysplasia, Holling type, 621345 (3), Autosomal recessive |
| **BOLA3** | 97.22 % | 613183 | Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299 (3), Autosomal recessive |
| **BORCS5** | 99.99 % | 616598 | *No OMIM phenotypes* |
| **BPNT2** | 99.96 % | 614010 | Chondrodysplasia with joint dislocations, GPAPP type, 614078 (3), Autosomal recessive |
| **BRAF** | 99.53 % | 164757 | Melanoma, malignant, somatic, 155600 (3); LEOPARD syndrome 3, 613707 (3), Autosomal dominant; Cardiofaciocutaneous syndrome, 115150 (3), Autosomal dominant; Adenocarcinoma of lung, somatic, 211980 (3); Noonan syndrome 7, 613706 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3); Nonsmall cell lung cancer, somatic, 211980 (3) |
| **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) |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **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 |
| **BRD4** | 99.81 % | 608749 | Cornelia de Lange syndrome 6, 620568 (3), Autosomal dominant |
| **BRF1** | 99.82 % | 604902 | Cerebellofaciodental syndrome, 616202 (3), Autosomal recessive |
| **BRF2** | 99.97 % | 607013 | *No 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 |
| **BRPF1** | 99.91 % | 602410 | Intellectual developmental disorder with dysmorphic facies and ptosis, 617333 (3), Autosomal dominant |
| **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 |
| **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 |
| **C12orf57** | 99.96 % | 615140 | Temtamy syndrome, 218340 (3), Autosomal recessive |
| **C1GALT1C1** | 99.93 % | 300611 | Hemolytic uremic syndrome, atypical, 8, with rhizomelic short stature, 301110 (3), X-linked recessive; Tn polyagglutination syndrome, somatic, 300622 (3) |
| **C1orf127** | 99.82 % | 619700 | Heterotaxy, visceral, 14, autosomal, 621080 (3), Autosomal recessive |
| **C1QBP** | 99.84 % | 601269 | Combined oxidative phosphorylation deficiency 33, 617713 (3), Autosomal recessive |
| **C2CD3** | 99.73 % | 615944 | Orofaciodigital syndrome XIV, 615948 (3), Autosomal recessive |
| **C2orf69** | 99.66 % | 619219 | Combined oxidative phosphorylation deficiency 53, 619423 (3), Autosomal recessive |
| **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 |
| **CACHD1** | 97.39 % | 620144 | *No OMIM phenotypes* |
| **CACNA1A** | 99.72 % | 601011 | Spinocerebellar ataxia 6, 183086 (3), Autosomal dominant; Episodic ataxia, type 2, 108500 (3), Autosomal dominant; Developmental and epileptic encephalopathy 42, 617106 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, 141500 (3), Autosomal dominant |
| **CACNA1C** | 99.92 % | 114205 | Timothy syndrome, 601005 (3), Autosomal dominant; Long QT syndrome 8, 618447 (3), Autosomal dominant; Neurodevelopmental disorder with hypotonia, language delay, and skeletal defects with or without seizures, 620029 (3), Autosomal dominant; Brugada syndrome 3, 611875 (3), Autosomal dominant |
| **CACNA1D** | 99.82 % | 114206 | Primary aldosteronism, seizures, and neurologic abnormalities, 615474 (3), Autosomal dominant; Sinoatrial node dysfunction and deafness, 614896 (3), Autosomal recessive |
| **CACNA1E** | 99.14 % | 601013 | Developmental and epileptic encephalopathy 69, 618285 (3), Autosomal dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **CACNA1G** | 99.67 % | 604065 | Spinocerebellar ataxia 42, 616795 (3), Autosomal dominant; Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087 (3), Autosomal dominant |
| **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 |
| **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 |
| **CAMK2B** | 98.89 % | 607707 | Intellectual developmental disorder, autosomal dominant 54, 617799 (3), Autosomal dominant |
| **CAMSAP1** | 99.95 % | 613774 | Cortical dysplasia, complex, with other brain malformations 12, 620316 (3), Autosomal recessive |
| **CAMTA1** | 99.88 % | 611501 | Cerebellar dysfunction with variable cognitive and behavioral abnormalities, 614756 (3), Autosomal dominant |
| **CANT1** | 99.62 % | 613165 | Desbuquois dysplasia 1, 251450 (3), Autosomal recessive; Epiphyseal dysplasia, multiple, 7, 617719 (3), Autosomal recessive |
| **CAPN15** | 99.95 % | 603267 | Oculogastrointestinal neurodevelopmental syndrome, 619318 (3), Autosomal recessive |
| **CAPRIN1** | 99.53 % | 601178 | Neurodevelopmental disorder with language impairment, autism, and attention deficit-hyperactivity disorder, 620782 (3), Autosomal dominant; Neurodegeneration, childhood-onset, with cerebellar ataxia and cognitive decline, 620636 (3), Autosomal dominant |
| **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 |
| **CASP2** | 99.77 % | 600639 | Intellectual developmental disorder, autosomal recessive 80, with variant lissencephaly, 620653 (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 |
| **CBFB** | 99.83 % | 121360 | Cleidocranial dysplasia 2, 620099 (3), Autosomal dominant |
| **CBL** | 99.82 % | 165360 | Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 (3), Autosomal dominant; ?Juvenile myelomonocytic leukemia, 607785 (3), Somatic mutation, Autosomal dominant |
| **CBY1** | 99.98 % | 607757 | *No OMIM phenotypes* |
| **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 |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **CCDC107** | 99.51 % | | *No OMIM phenotypes* |
| **CCDC22** | 99.56 % | 300859 | Ritscher-Schinzel syndrome 2, 300963 (3), X-linked recessive |
| **CCDC32** | 99.81 % | 618941 | Cardiofacioneurodevelopmental syndrome, 619123 (3), Autosomal 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 |
| **CCDC78** | 99.67 % | 614666 | Centronuclear myopathy 4, 614807 (3), Autosomal dominant |
| **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 |
| **CCN2** | 99.87 % | 121009 | ?Spondyloepimetaphyseal dysplasia, Li-Shao-Li type, 621099 (3), Autosomal dominant; Kyphomelic dysplasia, 211350 (3), Autosomal recessive |
| **CCND2** | 99.58 % | 123833 | Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938 (3), Autosomal dominant |
| **CCNQ** | 99.66 % | 300708 | STAR syndrome, 300707 (3), X-linked dominant |
| **CCP110** | 98.96 % | 609544 | *No OMIM phenotypes* |
| **CCT3** | 97.96 % | 600114 | Neurodevelopmental disorder with speech or visual impairment and brain hypomyelination, 621034 (3), Autosomal dominant |
| **CCT8** | 99.7 % | 617786 | *No OMIM phenotypes* |
| **CD40LG** | 99.81 % | 300386 | Immunodeficiency, X-linked, with hyper-IgM, 308230 (3), X-linked recessive |
| **CD96** | 99.72 % | 606037 | C syndrome, 211750 (3), Autosomal dominant |
| **CDAN1** | 99.93 % | 607465 | Dyserythropoietic anemia, congenital, type Ia, 224120 (3), Autosomal recessive |
| **CDC40** | 99.61 % | 605585 | ?Pontocerebellar hypoplasia, type 15, 619302 (3), Autosomal recessive |
| **CDC42** | 95.76 % | 116952 | Takenouchi-Kosaki syndrome, 616737 (3), Autosomal dominant |
| **CDC42BPB** | 99.9 % | 614062 | Chilton-Okur-Chung neurodevelopmental syndrome, 619841 (3), Autosomal dominant |
| **CDC45** | 98.79 % | 603465 | Meier-Gorlin syndrome 7, 617063 (3), Autosomal recessive |
| **CDC6** | 99.45 % | 602627 | Meier-Gorlin syndrome 5, 613805 (3), Autosomal recessive |
| **CDH1** | 99.69 % | 192090 | Ovarian cancer, somatic, 167000 (3); Blepharocheilodontic syndrome 1, 119580 (3), Autosomal dominant; Diffuse gastric and lobular breast cancer syndrome with or without cleft lip and/or palate, 137215 (3), Autosomal dominant; Endometrial carcinoma, somatic, 608089 (3); Breast cancer, lobular, somatic, 114480 (3) |
| **CDH11** | 99.65 % | 600023 | Teebi hypertelorism syndrome 2, 619736 (3), Autosomal dominant; Elsahy-Waters syndrome, 211380 (3), Autosomal recessive |
| **CDH2** | 99.82 % | 114020 | Arrhythmogenic right ventricular dysplasia 14, 618920 (3), Autosomal dominant; ?Attention deficit-hyperactivity disorder 8, 619957 (3), Autosomal recessive; Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929 (3), Autosomal dominant |
| **CDH3** | 99.86 % | 114021 | Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 (3), Autosomal recessive; Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 (3), Autosomal recessive |
| **CDK10** | 99.58 % | 603464 | Al Kaissi syndrome, 617694 (3), Autosomal recessive |
| **CDK13** | 99.69 % | 603309 | Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360 (3), Autosomal dominant |
| **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 |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **CDK8** | 99.82 % | 603184 | Intellectual developmental disorder with hypotonia and behavioral abnormalities, 618748 (3), Autosomal dominant |
| **CDKL5** | 99.79 % | 300203 | Developmental and epileptic encephalopathy 2, 300672 (3), X-linked dominant |
| **CDKN1C** | 99.99 % | 600856 | IMAGE syndrome, 614732 (3), Autosomal dominant; Beckwith-Wiedemann syndrome, 130650 (3), Autosomal dominant |
| **CDON** | 99.9 % | 608707 | Holoprosencephaly 11, 614226 (3), Autosomal dominant |
| **CDT1** | 99.68 % | 605525 | Meier-Gorlin syndrome 4, 613804 (3), Autosomal recessive |
| **CDX1** | 99.34 % | 600746 | *No OMIM phenotypes* |
| **CDX2** | 99.95 % | 600297 | *No OMIM phenotypes* |
| **CELSR1** | 99.9 % | 604523 | Lymphatic malformation 9, 619319 (3), Autosomal dominant; Yellow nail syndrome, 153300 (3), Autosomal recessive |
| **CELSR3** | 99.8 % | 604264 | *No OMIM phenotypes* |
| **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 |
| **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 |
| **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 |
| **CEP57** | 99.81 % | 607951 | Mosaic variegated aneuploidy syndrome 2, 614114 (3), Autosomal recessive |
| **CEP63** | 99.15 % | 614724 | ?Seckel syndrome 6, 614728 (3), Autosomal recessive |
| **CEP76** | 99.86 % | 620791 | *No OMIM phenotypes* |
| **CEP83** | 98.61 % | 615847 | Nephronophthisis 18, 615862 (3), Autosomal recessive |
| **CEP85L** | 99.98 % | 618865 | Lissencephaly 10, 618873 (3), Autosomal dominant |
| **CERS3** | 99.83 % | 615276 | Ichthyosis, congenital, autosomal recessive 9, 615023 (3), Autosomal recessive |
| **CERT1** | 99.44 % | 604677 | Neurodevelopmental disorder with hypotonia, speech delay, and dysmorphic facies, 616351 (3), Autosomal dominant |
| **CFAP298** | 99.7 % | 615494 | Ciliary dyskinesia, primary, 26, 615500 (3), Autosomal recessive |
| **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 |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **CFAP45** | 99.68 % | 605152 | Heterotaxy, visceral, 11, autosomal, with male infertility, 619608 (3), Autosomal recessive |
| **CFAP52** | 99.77 % | 609804 | Heterotaxy, visceral, 10, autosomal, with male infertility, 619607 (3), Autosomal recessive |
| **CFAP53** | 99.91 % | 614759 | Heterotaxy, visceral, 6, autosomal recessive, 614779 (3), Autosomal recessive |
| **CFC1** | 20.68 % | 605194 | Heterotaxy, visceral, 2, autosomal, 605376 (3), 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 |
| **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) |
| **CHAF1A** | 99.9 % | 601246 | *No OMIM phenotypes* |
| **CHAMP1** | 99.97 % | 616327 | Neurodevelopmental disorder with hypotonia, impaired language, and dysmorphic features, 616579 (3), Autosomal dominant |
| **CHASERR** | 0 % | 620993 | Neurodevelopmental disorder with dysmorphic facies, absent speech and ambulation, and brain abnormalities, 621012 (3), Autosomal dominant |
| **CHAT** | 96.79 % | 118490 | Myasthenic syndrome, congenital, 6, presynaptic, 254210 (3), Autosomal recessive |
| **CHD3** | 99.8 % | 602120 | Snijders Blok-Campeau syndrome, 618205 (3), Autosomal dominant |
| **CHD4** | 99.9 % | 603277 | Sifrim-Hitz-Weiss syndrome, 617159 (3), Autosomal dominant |
| **CHD7** | 99.9 % | 608892 | Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant; CHARGE syndrome, 214800 (3), Autosomal dominant |
| **CHD8** | 99.89 % | 610528 | Intellectual developmental disorder with autism and macrocephaly, 615032 (3), Autosomal dominant |
| **CHKB** | 99.96 % | 612395 | Muscular dystrophy, congenital, megaconial type, 602541 (3), Autosomal recessive |
| **CHMP1A** | 99.98 % | 164010 | Pontocerebellar hypoplasia, type 8, 614961 (3), Autosomal 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 |
| **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 |
| **CHRNB2** | 99.68 % | 118507 | Epilepsy, nocturnal frontal lobe, 3, 605375 (3) |
| **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 |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **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 |
| **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 |
| **CIBAR1** | 99.89 % | 617273 | ?Polydactyly, postaxial, type A9, 618219 (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 |
| **CIT** | 99.88 % | 605629 | Microcephaly 17, primary, autosomal recessive, 617090 (3), Autosomal recessive |
| **CITED2** | 99.97 % | 602937 | Atrial septal defect 8, 614433 (3), Autosomal dominant; Ventricular septal defect 2, 614431 (3), Autosomal dominant |
| **CKAP2L** | 99.44 % | 616174 | Filippi syndrome, 272440 (3), Autosomal recessive |
| **CLASP1** | 99.96 % | 605852 | *No OMIM phenotypes* |
| **CLCF1** | 99.86 % | 607672 | Cold-induced sweating syndrome 2, 610313 (3), Autosomal recessive |
| **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 |
| **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 |
| **CLCNKA** | 99.81 % | 602024 | Bartter syndrome, type 4b, digenic, 613090 (3), Digenic recessive |
| **CLCNKB** | 99.59 % | 602023 | Bartter syndrome, type 3, 607364 (3), Autosomal recessive; Bartter syndrome, type 4b, digenic, 613090 (3), Digenic 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 |
| **CLTC** | 99.3 % | 118955 | Intellectual developmental disorder, autosomal dominant 56, 617854 (3), Autosomal dominant |
| **CNKSR2** | 99.5 % | 300724 | Intellectual developmental disorder, X-linked syndromic, Houge type, 301008 (3), X-linked |
| **CNOT1** | 99.71 % | 604917 | Vissers-Bodmer syndrome, 619033 (3), Autosomal dominant; Holoprosencephaly 12, with or without pancreatic agenesis, 618500 (3), Autosomal dominant |
| **CNOT2** | 98.68 % | 604909 | Intellectual developmental disorder with nasal speech, dysmorphic facies, and variable skeletal anomalies, 618608 (3), Autosomal dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **CNOT3** | 99.88 % | 604910 | Intellectual developmental disorder with speech delay, autism, and dysmorphic facies, 618672 (3), Autosomal dominant |
| **CNTN1** | 98.8 % | 600016 | Congenital myopathy 12, 612540 (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 |
| **COASY** | 99.73 % | 609855 | Pontocerebellar hypoplasia, type 12, 618266 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 6, 615643 (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) |
| **COL10A1** | 99.94 % | 120110 | Metaphyseal chondrodysplasia, Schmid type, 156500 (3), Autosomal dominant |
| **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 |
| **COL13A1** | 99.59 % | 120350 | Myasthenic syndrome, congenital, 19, 616720 (3), Autosomal recessive |
| **COL18A1** | 99.79 % | 120328 | Knobloch syndrome, type 1, 267750 (3), Autosomal recessive; Glaucoma, primary closed-angle, 618880 (3), Autosomal dominant |
| **COL1A1** | 99.22 % | 120150 | Osteogenesis imperfecta, type II, 166210 (3), Autosomal dominant; Caffey disease, 114000 (3), Autosomal dominant; Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060 (3), Autosomal dominant; Osteogenesis imperfecta, type I, 166200 (3), Autosomal dominant; {Bone mineral density variation QTL, osteoporosis}, 166710 (3), Autosomal dominant; Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1, 619115 (3), Autosomal dominant; Osteogenesis imperfecta, type IV, 166220 (3), Autosomal dominant; Osteogenesis imperfecta, type III, 259420 (3), Autosomal dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **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 |
| **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 |
| **COL4A1** | 99.93 % | 120130 | ?Retinal arteries, tortuosity of, 180000 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3); Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 (3), Autosomal dominant; Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564 (3), Autosomal dominant; Brain small vessel disease with or without ocular anomalies, 175780 (3), Autosomal dominant |
| **COL4A2** | 99.69 % | 120090 | Brain small vessel disease 2, 614483 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3) |
| **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 |
| **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 |
| **COLEC10** | 99.96 % | 607620 | 3MC syndrome 3, 248340 (3), Autosomal recessive |
| **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 |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **COMP** | 99.77 % | 600310 | Pseudoachondroplasia, 177170 (3), Autosomal dominant; Carpal tunnel syndrome 2, 619161 (3), Autosomal dominant; Epiphyseal dysplasia, multiple, 1, 132400 (3), Autosomal dominant |
| **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 |
| **COQ7** | 99.89 % | 601683 | Coenzyme Q10 deficiency, primary, 8, 616733 (3), Autosomal recessive; Neuronopathy, distal hereditary motor, autosomal recessive 9, 620402 (3), Autosomal recessive |
| **COQ9** | 98.19 % | 612837 | Coenzyme Q10 deficiency, primary, 5, 614654 (3), Autosomal recessive |
| **COX14** | 98.98 % | 614478 | ?Mitochondrial complex IV deficiency, nuclear type 10, 619053 (3), Autosomal recessive |
| **COX7B** | 99.97 % | 300885 | Linear skin defects with multiple congenital anomalies 2, 300887 (3), X-linked dominant |
| **CPAMD8** | 99.58 % | 608841 | Anterior segment dysgenesis 8, 617319 (3), Autosomal recessive |
| **CPLANE1** | 99.74 % | 614571 | Orofaciodigital syndrome VI, 277170 (3), Autosomal recessive; Joubert syndrome 17, 614615 (3), Autosomal recessive |
| **CPLANE2** | 99.81 % | 620487 | *No OMIM phenotypes* |
| **CPOX** | 99.85 % | 612732 | Coproporphyria, 121300 (3), Autosomal recessive, Autosomal dominant; Harderoporphyria, 618892 (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 |
| **CRADD** | 99.62 % | 603454 | Intellectual developmental disorder, autosomal recessive 34, with variant lissencephaly, 614499 (3), Autosomal recessive |
| **CRB2** | 99.65 % | 609720 | Focal segmental glomerulosclerosis 9, 616220 (3), Autosomal recessive; Ventriculomegaly with cystic kidney disease, 219730 (3), Autosomal recessive |
| **CREB3L1** | 99.3 % | 616215 | Osteogenesis imperfecta, type XVI, 616229 (3), Autosomal recessive |
| **CREBBP** | 99.79 % | 600140 | Menke-Hennekam syndrome 1, 618332 (3), Autosomal dominant; Rubinstein-Taybi syndrome 1, 180849 (3), Autosomal dominant |
| **CRELD1** | 99.98 % | 607170 | Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217 (3), Autosomal dominant; Jeffries-Lakhani neurodevelopmental syndrome, 620771 (3), Autosomal recessive; {Atrioventricular septal defect, susceptibility to, 2}, 606217 (3), Autosomal dominant |
| **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 |
| **CRYAA** | 17.8 % | 123580 | Cataract 9, multiple types, 604219 (3), Autosomal recessive, Autosomal dominant |
| **CRYBA1** | 99.91 % | 123610 | Cataract 10, multiple types, 600881 (3), Autosomal dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **CRYBA4** | 99.98 % | 123631 | Cataract 23, 610425 (3), Autosomal dominant |
| **CRYBB1** | 99.51 % | 600929 | Cataract 17, multiple types, 611544 (3), Autosomal recessive, Autosomal dominant |
| **CRYBB2** | 98.14 % | 123620 | Cataract 3, multiple types, 601547 (3), Autosomal dominant |
| **CRYBB3** | 99.9 % | 123630 | Cataract 22, 609741 (3), Autosomal recessive, Autosomal dominant |
| **CRYGC** | 100 % | 123680 | Cataract 2, multiple types, 604307 (3), Autosomal dominant |
| **CRYGD** | 99.88 % | 123690 | Cataract 4, multiple types, 115700 (3), Autosomal dominant |
| **CSDE1** | 98.03 % | 191510 | *No OMIM phenotypes* |
| **CSF1R** | 99.71 % | 164770 | Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 (3), Autosomal recessive; Leukoencephalopathy, diffuse hereditary, with spheroids 1, 221820 (3), Autosomal dominant |
| **CSGALNACT1** | 99.95 % | 616615 | Skeletal dysplasia, mild, with joint laxity and advanced bone age, 618870 (3), Autosomal recessive |
| **CSMD1** | 99.9 % | 608397 | *No OMIM phenotypes* |
| **CSNK2A1** | 99.7 % | 115440 | Okur-Chung neurodevelopmental syndrome, 617062 (3), Autosomal dominant |
| **CSPP1** | 99.85 % | 611654 | Joubert syndrome 21, 615636 (3), Autosomal recessive |
| **CTC1** | 99.9 % | 613129 | Cerebroretinal microangiopathy with calcifications and cysts, 612199 (3), Autosomal recessive |
| **CTCF** | 99.6 % | 604167 | Intellectual developmental disorder, autosomal dominant 21, 615502 (3), Autosomal dominant |
| **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 |
| **CTNNB1** | 99.85 % | 116806 | Exudative vitreoretinopathy 7, 617572 (3), Autosomal dominant; Pilomatricoma, somatic, 132600 (3); Colorectal cancer, somatic, 114500 (3); Neurodevelopmental disorder with spastic diplegia and visual defects, 615075 (3), Autosomal dominant; Medulloblastoma, somatic, 155255 (3); Ovarian cancer, somatic, 167000 (3); Hepatocellular carcinoma, somatic, 114550 (3) |
| **CTNND1** | 99.62 % | 601045 | Blepharocheilodontic syndrome 2, 617681 (3), Autosomal dominant |
| **CTSA** | 99.77 % | 613111 | Galactosialidosis, 256540 (3), Autosomal recessive; Brain small vessel disease 6 with leukoencephalopathy, 621394 (3), Autosomal dominant |
| **CTSD** | 99.84 % | 116840 | Ceroid lipofuscinosis, neuronal, 10, 610127 (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 |
| **CUL3** | 99.71 % | 603136 | Neurodevelopmental disorder with or without autism or seizures, 619239 (3), Autosomal dominant; Pseudohypoaldosteronism, type IIE, 614496 (3), Autosomal dominant |
| **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 |
| **CUX2** | 99.92 % | 610648 | Developmental and epileptic encephalopathy 67, 618141 (3), Autosomal dominant |
| **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 |
| **CYB5R3** | 99.25 % | 613213 | Methemoglobinemia, type I, 250800 (3), Autosomal recessive; Methemoglobinemia, type II, 250800 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **CYBB** | 99.67 % | 300481 | Immunodeficiency 34, mycobacteriosis, X-linked, 300645 (3), X-linked recessive; Chronic granulomatous disease, X-linked, 306400 (3), X-linked recessive |
| **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 |
| **CYP17A1** | 99.94 % | 609300 | 17,20-lyase deficiency, isolated, 202110 (3), Autosomal recessive; 17-alpha-hydroxylase/17,20-lyase deficiency, 202110 (3), Autosomal recessive |
| **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 |
| **CYP26B1** | 99.92 % | 605207 | Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416 (3) |
| **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 |
| **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 |
| **DAND5** | 99.84 % | 609068 | Heterotaxy, visceral, 13, autosomal, 621079 (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 |
| **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 |
| **DCX** | 99.97 % | 300121 | Subcortical laminal heterotopia, X-linked, 300067 (3), X-linked; Lissencephaly, X-linked, 300067 (3), X-linked |
| **DDR1** | 99.82 % | 600408 | *No OMIM phenotypes* |
| **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 |
| **DDX23** | 99.68 % | 612172 | *No OMIM phenotypes* |
| **DDX3X** | 99.22 % | 300160 | Intellectual developmental disorder, X-linked syndromic, Snijders Blok type, 300958 (3), X-linked dominant, X-linked recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **DDX59** | 99.38 % | 615464 | Orofaciodigital syndrome V, 174300 (3), Autosomal recessive |
| **DENND5A** | 99.88 % | 617278 | Developmental and epileptic encephalopathy 49, 617281 (3), Autosomal recessive |
| **DEPDC5** | 99.62 % | 614191 | Epilepsy, familial focal, with variable foci 1, 604364 (3), Autosomal dominant; Developmental and epileptic encephalopathy 111, 620504 (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 |
| **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 |
| **DHODH** | 99.75 % | 126064 | Miller syndrome, 263750 (3), Autosomal recessive |
| **DHPS** | 98.35 % | 600944 | Neurodevelopmental disorder with seizures and speech and walking impairment, 618480 (3), Autosomal recessive |
| **DHRS3** | 99.8 % | 612830 | *No OMIM phenotypes* |
| **DHRSX** | 99.53 % | 301034 | Congenital disorder of glycosylation, type 1DD, 301133 (3), Pseudoautosomal recessive |
| **DHTKD1** | 99.87 % | 614984 | ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025 (3), Autosomal dominant; Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750 (3), Autosomal recessive |
| **DHX30** | 99.71 % | 616423 | Neurodevelopmental disorder with variable motor and speech impairment, 617804 (3), Autosomal dominant |
| **DHX37** | 98.86 % | 617362 | Neurodevelopmental disorder with brain anomalies and with or without vertebral or cardiac anomalies, 618731 (3), Autosomal recessive; 46XY sex reversal 11, 273250 (3), Autosomal dominant |
| **DHX9** | 99.42 % | 603115 | Intellectual developmental disorder, autosomal dominant 75, 620988 (3), Autosomal dominant |
| **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 |
| **DIP2C** | 99.9 % | 611380 | *No OMIM phenotypes* |
| **DIS3L2** | 99.75 % | 614184 | Perlman syndrome, 267000 (3), Autosomal recessive |
| **DISP1** | 99.82 % | 607502 | Holoprosencephaly 10, 621143 (3), Autosomal recessive, Autosomal dominant |
| **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 |
| **DLG3** | 99.84 % | 300189 | Intellectual developmental disorder, X-linked 90, 300850 (3), X-linked recessive |
| **DLG4** | 99.82 % | 602887 | Intellectual developmental disorder, autosomal dominant 62, 618793 (3), Autosomal dominant |
| **DLG5** | 99.56 % | 604090 | Yuksel-Vogel-Bauser syndrome, 620703 (3), Autosomal recessive |
| **DLL1** | 99.85 % | 606582 | Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizures, 618709 (3), Autosomal dominant |
| **DLL3** | 99.67 % | 602768 | Spondylocostal dysostosis 1, autosomal recessive, 277300 (3), Autosomal recessive |
| **DLL4** | 99.71 % | 605185 | Adams-Oliver syndrome 6, 616589 (3), Autosomal dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **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 |
| **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 |
| **DNAH11** | 99.87 % | 603339 | Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884 (3), Autosomal recessive |
| **DNAH5** | 99.89 % | 603335 | Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644 (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 |
| **DNAJB11** | 99.85 % | 611341 | Polycystic kidney disease 6 with or without polycystic liver disease, 618061 (3), Autosomal dominant |
| **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 |
| **DNAL1** | 99.88 % | 610062 | Ciliary dyskinesia, primary, 16, 614017 (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 |
| **DNMT3A** | 99.69 % | 602769 | Tatton-Brown-Rahman syndrome, 615879 (3), Autosomal dominant; Acute myeloid leukemia, somatic, 601626 (3); Heyn-Sproul-Jackson syndrome, 618724 (3), Autosomal dominant |
| **DNMT3B** | 99.92 % | 602900 | Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 (3), Autosomal recessive; Facioscapulohumeral muscular dystrophy 4, digenic, 619478 (3), Digenic dominant |
| **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 |

| 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 |
| **DOT1L** | 99.7 % | 607375 | Nil-Deshwar neurodevelopmental syndrome, 621265 (3), Autosomal dominant |
| **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 |
| **DPF2** | 99.89 % | 601671 | Coffin-Siris syndrome 7, 618027 (3), Autosomal dominant |
| **DPH1** | 99.89 % | 603527 | Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901 (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 |
| **DPYSL5** | 99.77 % | 608383 | Ritscher-Schinzel syndrome 4, 619435 (3), Autosomal dominant |
| **DRC1** | 99.73 % | 615288 | Spermatogenic failure 80, 620222 (3), Autosomal recessive; Ciliary dyskinesia, primary, 21, 615294 (3), Autosomal recessive |
| **DRG1** | 99.81 % | 603952 | Tan-Almurshedi syndrome, 620641 (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 |
| **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 |
| **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 |
| **DVL1** | 99.85 % | 601365 | Robinow syndrome, autosomal dominant 2, 616331 (3), Autosomal dominant |
| **DVL2** | 99.52 % | 602151 | *No OMIM phenotypes* |
| **DVL3** | 99.83 % | 601368 | Robinow syndrome, autosomal dominant 3, 616894 (3), Autosomal dominant |
| **DYM** | 99.76 % | 607461 | Smith-McCort dysplasia, 607326 (3), Autosomal recessive; Dyggve-Melchior-Clausen disease, 223800 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **DYNC1H1** | 99.92 % | 600112 | Charcot-Marie-Tooth disease, axonal, type 2O, 614228 (3), Autosomal dominant; Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 (3), Autosomal dominant; Cortical dysplasia, complex, with other brain malformations 13, 614563 (3), Autosomal dominant |
| **DYNC1I1** | 98.76 % | 603772 | *No OMIM phenotypes* |
| **DYNC1I2** | 99.16 % | 603331 | Neurodevelopmental disorder with microcephaly and structural brain anomalies, 618492 (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 |
| **DYRK1A** | 99.92 % | 600855 | Intellectual developmental disorder, autosomal dominant 7, 614104 (3), Autosomal dominant |
| **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 |
| **EBF3** | 99.7 % | 607407 | Hypotonia, ataxia, and delayed development syndrome, 617330 (3), Autosomal dominant |
| **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 |
| **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 |
| **EDNRA** | 99.92 % | 131243 | {Migraine, resistance to}, 157300 (3), Autosomal dominant; Mandibulofacial dysostosis with alopecia, 616367 (3), Autosomal dominant |
| **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 |
| **EED** | 99.59 % | 605984 | Cohen-Gibson syndrome, 617561 (3), Autosomal dominant |
| **EEF1A2** | 99.86 % | 602959 | Developmental and epileptic encephalopathy 33, 616409 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 38, 616393 (3), Autosomal dominant |
| **EEF2** | 99.52 % | 130610 | ?Spinocerebellar ataxia 26, 609306 (3), Autosomal dominant |
| **EEFSEC** | 99.67 % | 607695 | Neurodevelopmental disorder with progressive spasticity and brain abnormalities, 621102 (3), Autosomal recessive |
| **EFCAB1** | 99.82 % | 619564 | Ciliary dyskinesia, primary, 53, 620642 (3), Autosomal recessive |
| **EFEMP1** | 99.4 % | 601548 | Doyne honeycomb degeneration of retina, 126600 (3), Autosomal dominant; Cutis laxa, autosomal recessive, type ID, 620780 (3), Autosomal recessive; Glaucoma 1, open angle, H, 611276 (3), Autosomal dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **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 |
| **EFTUD2** | 99.55 % | 603892 | Mandibulofacial dysostosis, Guion-Almeida type, 610536 (3), Autosomal dominant |
| **EHBP1L1** | 99.91 % | 619583 | *No OMIM phenotypes* |
| **EHMT1** | 99.87 % | 607001 | Kleefstra syndrome 1, 610253 (3), Autosomal dominant |
| **EIF2AK3** | 98.74 % | 604032 | Wolcott-Rallison syndrome, 226980 (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 |
| **EIF2S3** | 99.23 % | 300161 | MEHMO syndrome, 300148 (3), X-linked recessive |
| **EIF3B** | 99.94 % | 603917 | *No OMIM phenotypes* |
| **EIF3F** | 99.63 % | 603914 | Intellectual developmental disorder, autosomal recessive 67, 618295 (3), Autosomal recessive |
| **EIF4A2** | 99.91 % | 601102 | Neurodevelopmental disorder with hypotonia and speech delay, with or without seizures, 620455 (3), Autosomal recessive, Autosomal dominant |
| **EIF4A3** | 99.96 % | 608546 | Robin sequence with cleft mandible and limb anomalies, 268305 (3), Autosomal recessive |
| **EIF5A** | 100 % | 600187 | Faundes-Banka syndrome, 619376 (3), Autosomal dominant |
| **ELAC2** | 99.62 % | 605367 | {Prostate cancer, hereditary, 2, susceptibility to}, 614731 (3); Combined oxidative phosphorylation deficiency 17, 615440 (3), Autosomal recessive |
| **ELMO2** | 99.98 % | 606421 | Vascular malformation, primary intraosseous, 606893 (3), Autosomal recessive |
| **ELN** | 98.97 % | 130160 | Cutis laxa, autosomal dominant, 123700 (3), Autosomal dominant; Supravalvar aortic stenosis, 185500 (3), 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 |
| **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 |
| **EMILIN1** | 99.81 % | 130660 | Neuronopathy, distal hereditary motor, autosomal dominant 10, 620080 (3), Autosomal dominant; Arterial tortuosity-bone fragility syndrome, 620908 (3), Autosomal recessive |
| **EML1** | 99.94 % | 602033 | Band heterotopia, 600348 (3), Autosomal recessive |
| **EMX2** | 99.86 % | 600035 | Schizencephaly, 269160 (3) |
| **EN1** | 99.98 % | 131290 | ?ENDOVE syndrome, limb-brain type, 619218 (3), Autosomal recessive |
| **ENG** | 99.82 % | 131195 | Telangiectasia, hereditary hemorrhagic, type 1, 187300 (3), Autosomal dominant |
| **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 |
| **ENPP5** | 99.98 % | 617001 | *No OMIM phenotypes* |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **EOGT** | 98.82 % | 614789 | Adams-Oliver syndrome 4, 615297 (3), Autosomal recessive |
| **EP300** | 99.9 % | 602700 | Menke-Hennekam syndrome 2, 618333 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3); Rubinstein-Taybi syndrome 2, 613684 (3), Autosomal dominant |
| **EPG5** | 99.86 % | 615068 | Vici syndrome, 242840 (3), Autosomal recessive |
| **EPHB4** | 99.37 % | 600011 | Capillary malformation-arteriovenous malformation 2, 618196 (3), Autosomal dominant; Lymphatic malformation 7, 617300 (3), Autosomal dominant |
| **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 |
| **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 |
| **ERCC8** | 99.6 % | 609412 | UV-sensitive syndrome 2, 614621 (3), Autosomal recessive; Cockayne syndrome, type A, 216400 (3), Autosomal recessive |
| **ERF** | 99.68 % | 611888 | Craniosynostosis 4, 600775 (3), Autosomal dominant; Chitayat syndrome, 617180 (3), Autosomal dominant |
| **ERG** | 99.9 % | 165080 | Lymphatic malformation 14, 620602 (3), Autosomal dominant |
| **ERGIC1** | 99.87 % | 617946 | ?Arthrogryposis multiplex congenita 2, neurogenic type, 208100 (3), Autosomal recessive |
| **ERI1** | 99.21 % | 608739 | Hoxha-Aliu syndrome, 620662 (3), Autosomal recessive; Spondyloepimetaphyseal dysplasia, Guo-Campeau type, 620663 (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 |
| **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 |
| **ETV2** | 99.59 % | 609358 | *No OMIM phenotypes* |
| **EVC** | 99.73 % | 604831 | Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; ?Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **EVC2** | 99.79 % | 607261 | Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant |
| **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 |
| **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 |
| **EXT1** | 99.88 % | 608177 | Exostoses, multiple, type 1, 133700 (3), Autosomal dominant; Chondrosarcoma, 215300 (3), Somatic mutation |
| **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 |
| **EYA1** | 99.41 % | 601653 | Branchiootic syndrome 1, 602588 (3), Autosomal dominant; Branchiootorenal syndrome 1, with or without cataracts, 113650 (3), Autosomal dominant; Anterior segment anomalies with or without cataract, 602588 (3), Autosomal dominant; ?Otofaciocervical syndrome, 166780 (3), Autosomal dominant |
| **EZH2** | 99.77 % | 601573 | Weaver syndrome, 277590 (3), Autosomal dominant |
| **FAAP100** | 99.83 % | 611301 | Fanconi anemia, complementation group X, 621258 (3), Autosomal recessive |
| **FAH** | 97.69 % | 613871 | Tyrosinemia, type I, 276700 (3), Autosomal recessive |
| **FAM111A** | 99.98 % | 615292 | Kenny-Caffey syndrome, type 2, 127000 (3), Autosomal dominant; Gracile bone dysplasia, 602361 (3), Autosomal dominant |
| **FAM126A** | 99.87 % | 610531 | Leukodystrophy, hypomyelinating, 5, 610532 (3), Autosomal recessive |
| **FAM149B1** | 98.11 % | 618413 | Joubert syndrome 36, 618763 (3), Autosomal recessive |
| **FAM177A1** | 99.96 % | 619181 | Neurodevelopmental disorder with white matter abnormalities and gait disturbance, 621152 (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 |
| **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 |
| **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 |
| **FANCM** | 99.83 % | 609644 | Premature ovarian failure 15, 618096 (3), Autosomal recessive; Spermatogenic failure 28, 618086 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **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 |
| **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 |
| **FAT1** | 99.97 % | 600976 | *No OMIM phenotypes* |
| **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 |
| **FBN1** | 99.73 % | 134797 | Geleophysic dysplasia 2, 614185 (3), Autosomal dominant; Weill-Marchesani syndrome 2, dominant, 608328 (3), Autosomal dominant; Ectopia lentis, familial, 129600 (3), Autosomal dominant; MASS syndrome, 604308 (3), Autosomal dominant; Marfan lipodystrophy syndrome, 616914 (3), Autosomal dominant; Acromicric dysplasia, 102370 (3), Autosomal dominant; Marfan syndrome, 154700 (3), Autosomal dominant; Stiff skin syndrome, 184900 (3), Autosomal dominant |
| **FBN2** | 99.55 % | 612570 | Macular degeneration, early-onset, 616118 (3), Autosomal dominant; Contractural arachnodactyly, congenital, 121050 (3), Autosomal dominant |
| **FBRSL1** | 99.84 % | 620123 | *No OMIM phenotypes* |
| **FBXL4** | 99.94 % | 605654 | Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471 (3), Autosomal recessive |
| **FBXO22** | 99.87 % | 609096 | Tayoun-Maawali syndrome, 621184 (3), Autosomal recessive |
| **FBXO28** | 99.6 % | 609100 | Developmental and epileptic encephalopathy 100, 619777 (3), Autosomal dominant |
| **FBXW11** | 99.8 % | 605651 | Neurodevelopmental, jaw, eye, and digital syndrome, 618914 (3), Autosomal dominant |
| **FBXW7** | 99.84 % | 606278 | Developmental delay, hypotonia, and impaired language, 620012 (3), Autosomal dominant |
| **FEM1B** | 99.96 % | 613539 | Neurodevelopmental disorder with behavioral, ear, and skeletal abnormalities, 621263 (3), Autosomal dominant |
| **FEZF1** | 99.97 % | 613301 | Hypogonadotropic hypogonadism 22, with or without anosmia, 616030 (3), Autosomal recessive |
| **FGD1** | 99.65 % | 300546 | Aarskog-Scott syndrome, 305400 (3), X-linked recessive |
| **FGF10** | 99.83 % | 602115 | LADD syndrome 3, 620193 (3), Autosomal dominant; Aplasia of lacrimal and salivary glands, 180920 (3), Autosomal dominant |
| **FGF16** | 99.54 % | 300827 | Metacarpal 4-5 fusion, 309630 (3), X-linked recessive |
| **FGF3** | 99.74 % | 164950 | Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706 (3), Autosomal recessive |
| **FGF4** | 99.45 % | 164980 | Short-rib thoracic dysplasia 22 without polydactyly, 621260 (3), Autosomal recessive |
| **FGF8** | 99.65 % | 600483 | Hypogonadotropic hypogonadism 6 with or without anosmia, 612702 (3), Autosomal dominant |
| **FGF9** | 99.96 % | 600921 | Multiple synostoses syndrome 3, 612961 (3), Autosomal dominant |
| **FGFR1** | 99.77 % | 136350 | Pfeiffer syndrome, 101600 (3), Autosomal dominant; Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 (3), Autosomal dominant; Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; Hartsfield syndrome, 615465 (3), Autosomal dominant; Trigonocephaly 1, 190440 (3), Autosomal dominant; Osteoglophonic dysplasia, 166250 (3), Autosomal dominant; Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001 (3) |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **FGFR2** | 99.76 % | 176943 | Bent bone dysplasia syndrome, 614592 (3), Autosomal dominant; LADD syndrome 1, 149730 (3), Autosomal dominant; Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 (3), Autosomal dominant; Scaphocephaly and Axenfeld-Rieger anomaly (3); Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; Gastric cancer, somatic, 613659 (3); Craniofacial-skeletal-dermatologic dysplasia, 101600 (3), Autosomal dominant; Apert syndrome, 101200 (3), Autosomal dominant; Pfeiffer syndrome, 101600 (3), Autosomal dominant; Craniosynostosis, nonspecific (3); ?Scaphocephaly, maxillary retrusion, and impaired intellectual development, 609579 (3); Beare-Stevenson cutis gyrata syndrome, 123790 (3), Autosomal dominant; Crouzon syndrome, 123500 (3), Autosomal dominant; Saethre-Chotzen syndrome, 101400 (3), Autosomal dominant |
| **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 |
| **FH** | 99.84 % | 136850 | Leiomyomatosis and renal cell cancer, 150800 (3), Autosomal dominant; Fumarase deficiency, 606812 (3), Autosomal recessive |
| **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 |
| **FILIP1** | 99.9 % | 607307 | Neuromuscular disorder, congenital, with dysmorphic facies, 620775 (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 |
| **FKBP8** | 99.77 % | 604840 | *No OMIM phenotypes* |
| **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 |
| **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 |
| **FLII** | 99.6 % | 600362 | Cardiomyopathy, dilated, 2J, 620635 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **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 |
| **FLNC** | 99.75 % | 102565 | Cardiomyopathy, familial hypertrophic, 26, 617047 (3), Autosomal dominant; Arrhythmogenic right ventricular dysplasia, familial, 617047 (3), Autosomal dominant; Cardiomyopathy, familial restrictive 5, 617047 (3), Autosomal dominant; Myopathy, distal, 4, 614065 (3), Autosomal dominant; Myopathy, myofibrillar, 5, 609524 (3), Autosomal dominant |
| **FLT4** | 99.64 % | 136352 | Hemangioma, capillary infantile, somatic, 602089 (3); Lymphatic malformation 1, 153100 (3), Autosomal dominant; Congenital heart defects, multiple types, 7, 618780 (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 |
| **FN1** | 99.65 % | 135600 | Spondylometaphyseal dysplasia, corner fracture type, 184255 (3), Autosomal dominant; Glomerulopathy with fibronectin deposits 2, 601894 (3), Autosomal dominant |
| **FOLR1** | 99.95 % | 136430 | Neurodegeneration due to cerebral folate transport deficiency, 613068 (3), Autosomal recessive |
| **FOSL2** | 99.72 % | 601575 | Aplasia cutis-enamel dysplasia syndrome, 620789 (3), Autosomal dominant |
| **FOXC1** | 99.97 % | 601090 | Axenfeld-Rieger syndrome, type 3, 602482 (3), Autosomal dominant; Anterior segment dysgenesis 3, multiple subtypes, 601631 (3), Autosomal dominant |
| **FOXC2** | 99.91 % | 602402 | Lymphedema-distichiasis syndrome, 153400 (3), Autosomal dominant; Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400 (3), Autosomal dominant |
| **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 |
| **FOXF1** | 99.94 % | 601089 | Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380 (3), Autosomal dominant |
| **FOXG1** | 99.97 % | 164874 | Rett syndrome, congenital variant, 613454 (3), Autosomal dominant |
| **FOXI3** | 99.8 % | 612351 | Craniofacial microsomia 2, 620444 (3), Autosomal recessive, Autosomal dominant |
| **FOXJ1** | 99.85 % | 602291 | Ciliary dyskinesia, primary, 43, 618699 (3), Autosomal dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **FOXL2** | 98.7 % | 605597 | Premature ovarian failure 3, 608996 (3), Autosomal dominant; Blepharophimosis, epicanthus inversus, and ptosis, types 1 and 2, 110100 (3), Autosomal recessive, Autosomal dominant |
| **FOXP2** | 99.84 % | 605317 | Speech-language disorder-1, 602081 (3), Autosomal dominant |
| **FOXP3** | 99.49 % | 300292 | Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790 (3), X-linked recessive |
| **FOXP4** | 99.33 % | 608924 | *No OMIM phenotypes* |
| **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 |
| **FRRS1L** | 99.69 % | 604574 | Developmental and epileptic encephalopathy 37, 616981 (3), Autosomal recessive |
| **FRYL** | 99.67 % | 620798 | Pan-Chung-Bellen syndrome, 621049 (3), Autosomal dominant |
| **FSD1L** | 99.84 % | 609829 | *No OMIM phenotypes* |
| **FTL** | 99.52 % | 134790 | Hyperferritinemia-cataract syndrome, 600886 (3), Autosomal dominant; L-ferritin deficiency, dominant and recessive, 615604 (3), Autosomal recessive, Autosomal dominant; Neurodegeneration with brain iron accumulation 3, 606159 (3), Autosomal dominant |
| **FTO** | 99.17 % | 610966 | Growth retardation, developmental delay, facial dysmorphism, 612938 (3), Autosomal recessive; {Obesity, susceptibility to, BMIQ14}, 612460 (3), Autosomal 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 |
| **FUZ** | 99.56 % | 610622 | {Neural tube defects, susceptibility to}, 182940 (3), Autosomal dominant |
| **FYCO1** | 99.95 % | 607182 | Cataract 18, autosomal recessive, 610019 (3), Autosomal recessive |
| **FZD2** | 99.89 % | 600667 | Omodysplasia 2, 164745 (3), Autosomal dominant |
| **FZD6** | 99.92 % | 603409 | Nail disorder, nonsyndromic congenital, 1, 161050 (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 |
| **GABRA1** | 99.93 % | 137160 | {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136 (3); Developmental and epileptic encephalopathy 19, 615744 (3), Autosomal dominant; {Epilepsy, childhood absence, susceptibility to, 4}, 611136 (3) |
| **GABRB2** | 99.81 % | 600232 | Developmental and epileptic encephalopathy 92, 617829 (3), Autosomal dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **GABRG2** | 98.61 % | 137164 | Developmental and epileptic encephalopathy 74, 618396 (3), Autosomal dominant; Febrile seizures, familial, 8, 607681 (3), Autosomal dominant; Generalized epilepsy with febrile seizures plus, type 3, 607681 (3), Autosomal dominant |
| **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 |
| **GALNS** | 99.66 % | 612222 | Mucopolysaccharidosis IVA, 253000 (3), Autosomal recessive |
| **GALNT2** | 98.46 % | 602274 | Congenital disorder of glycosylation, type IIt, 618885 (3), Autosomal recessive |
| **GALT** | 99.95 % | 606999 | Galactosemia, 230400 (3), Autosomal recessive |
| **GANAB** | 99.79 % | 104160 | Polycystic kidney disease 3, 600666 (3), Autosomal dominant |
| **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 |
| **GATA2** | 99.91 % | 137295 | {Leukemia, acute myeloid, susceptibility to}, 601626 (3), Somatic mutation, Autosomal dominant; Emberger syndrome, 614038 (3), Autosomal dominant; Immunodeficiency 21, 614172 (3), Autosomal dominant; {Myelodysplastic syndrome, susceptibility to}, 614286 (3) |
| **GATA3** | 99.81 % | 131320 | Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255 (3), Autosomal dominant |
| **GATA4** | 99.65 % | 600576 | Tetralogy of Fallot, 187500 (3), Autosomal dominant; Atrial septal defect 2, 607941 (3), Autosomal dominant; Ventricular septal defect 1, 614429 (3), Autosomal dominant; Atrioventricular septal defect 4, 614430 (3), Autosomal dominant; ?Testicular anomalies with or without congenital heart disease, 615542 (3), Autosomal dominant |
| **GATA5** | 99.91 % | 611496 | Congenital heart defects, multiple types, 5, 617912 (3), Autosomal recessive, Autosomal dominant |
| **GATA6** | 99.78 % | 601656 | Atrial septal defect 9, 614475 (3), Autosomal dominant; Persistent truncus arteriosus, 217095 (3); Pancreatic agenesis and congenital heart defects, 600001 (3), Autosomal dominant; Atrioventricular septal defect 5, 614474 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant |
| **GATAD2B** | 99.08 % | 614998 | GAND syndrome, 615074 (3), Autosomal dominant |
| **GATB** | 99.78 % | 603645 | ?Combined oxidative phosphorylation deficiency 41, 618838 (3), Autosomal recessive |
| **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 |
| **GDF1** | 99.15 % | 602880 | Congenital heart defects, multiple types, 6, 613854 (3), Autosomal dominant; Right atrial isomerism (Ivemark), 208530 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **GDF11** | 99.86 % | 603936 | ?Vertebral hypersegmentation and orofacial anomalies, 619122 (3), Autosomal dominant |
| **GDF2** | 99.92 % | 605120 | Telangiectasia, hereditary hemorrhagic, type 5, 615506 (3), Autosomal dominant |
| **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 |
| **GEMIN4** | 99.93 % | 606969 | Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities, 617913 (3), Autosomal recessive |
| **GFAP** | 99.92 % | 137780 | Alexander disease, 203450 (3), Autosomal dominant |
| **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 |
| **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 |
| **GINS3** | 89.38 % | 610610 | *No 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 |
| **GJA3** | 99.97 % | 121015 | Cataract 14, multiple types, 601885 (3), Autosomal dominant |
| **GJA8** | 99.95 % | 600897 | Cataract 1, multiple types, 116200 (3), Autosomal dominant |
| **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 |
| **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 |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **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 |
| **GLI2** | 99.67 % | 165230 | Culler-Jones syndrome, 615849 (3), Autosomal dominant; Holoprosencephaly 9, 610829 (3), Autosomal dominant |
| **GLI3** | 99.97 % | 165240 | Greig cephalopolysyndactyly syndrome, 175700 (3), Autosomal dominant; Polydactyly, postaxial, types A1 and B, 174200 (3), Autosomal dominant; Pallister-Hall syndrome, 146510 (3), Autosomal dominant; Polydactyly, preaxial, type IV, 174700 (3), Autosomal dominant |
| **GLIS3** | 99.86 % | 610192 | Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199 (3), Autosomal recessive |
| **GLMN** | 93.7 % | 601749 | Glomuvenous malformations, 138000 (3), Autosomal dominant |
| **GLUL** | 99.4 % | 138290 | Glutamine deficiency, congenital, 610015 (3), Autosomal recessive; Developmental and epileptic encephalopathy 116, 620806 (3), Autosomal dominant |
| **GM2A** | 99.95 % | 613109 | GM2-gangliosidosis, AB variant, 272750 (3), Autosomal recessive |
| **GMNN** | 99.29 % | 602842 | Meier-Gorlin syndrome 6, 616835 (3), Autosomal dominant |
| **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 |
| **GNA11** | 99.84 % | 139313 | Hypocalciuric hypercalcemia, type II, 145981 (3), Autosomal dominant; Hypocalcemia, autosomal dominant 2, 615361 (3), Autosomal dominant |
| **GNA14** | 99.75 % | 604397 | *No OMIM phenotypes* |
| **GNAI2** | 99.81 % | 139360 | Pituitary adenoma, ACTH-secreting, somatic (3); Ventricular tachycardia, somatic, 192605 (3) |
| **GNAI3** | 98.72 % | 139370 | Auriculocondylar syndrome 1, 602483 (3), Autosomal dominant |
| **GNAO1** | 97.95 % | 139311 | Developmental and epileptic encephalopathy 17, 615473 (3), Autosomal dominant; Neurodevelopmental disorder with involuntary movements, 617493 (3), Autosomal dominant |
| **GNAS** | 99.97 % | 139320 | Pituitary adenoma 3, multiple types, somatic, 617686 (3); Pseudohypoparathyroidism Ic, 612462 (3), Autosomal dominant; Pseudohypoparathyroidism Ia, 103580 (3), Autosomal dominant; Osseous heteroplasia, progressive, 166350 (3), Autosomal dominant; McCune-Albright syndrome, somatic mosaic, 174800 (3); Pseudohypoparathyroidism Ib, 603233 (3), Autosomal dominant; Pseudopseudohypoparathyroidism, 612463 (3), Autosomal dominant; ACTH-independent macronodular adrenal hyperplasia 1, somatic, 219080 (3) |
| **GNB1** | 99.88 % | 139380 | Myelodysplastic syndrome, somatic, 614286 (3); Leukemia, acute lymphoblastic, somatic, 613065 (3); Intellectual developmental disorder, autosomal dominant 42, 616973 (3), Autosomal dominant |
| **GNB2** | 99.65 % | 139390 | Neurodevelopmental disorder with hypotonia and dysmorphic facies, 619503 (3), Autosomal dominant; ?Sick sinus syndrome 4, 619464 (3), Autosomal dominant |
| **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 |
| **GNPAT** | 99.73 % | 602744 | Rhizomelic chondrodysplasia punctata, type 2, 222765 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **GNPNAT1** | 100 % | 616510 | ?Rhizomelic dysplasia, Ain-Naz type, 619598 (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 |
| **GNS** | 98.91 % | 607664 | Mucopolysaccharidosis type IIID, 252940 (3), Autosomal recessive |
| **GON4L** | 97.66 % | 610393 | Li-Takada-Miyake syndrome, 621212 (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 |
| **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 |
| **GPI** | 99.77 % | 172400 | Anemia, congenital, nonspherocytic hemolytic, 4, glucose phosphate isomerase deficient, 613470 (3), Autosomal recessive |
| **GPKOW** | 99.6 % | 301003 | *No OMIM phenotypes* |
| **GPSM2** | 97.34 % | 609245 | Chudley-McCullough syndrome, 604213 (3), Autosomal recessive |
| **GPX4** | 99.94 % | 138322 | Spondylometaphyseal dysplasia, Sedaghatian type, 250220 (3), Autosomal recessive |
| **GREB1L** | 99.91 % | 617782 | Deafness, autosomal dominant 80, 619274 (3), Autosomal dominant; Renal hypodysplasia/aplasia 3, 617805 (3), Autosomal dominant |
| **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 |
| **GRHL3** | 99.99 % | 608317 | van der Woude syndrome 2, 606713 (3), Autosomal dominant |
| **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 |
| **GRIN2B** | 99.93 % | 138252 | Developmental and epileptic encephalopathy 27, 616139 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 6, with or without seizures, 613970 (3), Autosomal dominant |
| **GRIN2D** | 99.71 % | 602717 | Developmental and epileptic encephalopathy 46, 617162 (3), Autosomal dominant |
| **GRIP1** | 99.39 % | 604597 | Fraser syndrome 3, 617667 (3), Autosomal recessive |
| **GRM7** | 99.98 % | 604101 | Neurodevelopmental disorder with seizures, hypotonia, and brain abnormalities, 618922 (3), Autosomal recessive |
| **GSC** | 99.86 % | 138890 | Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471 (3), Autosomal recessive |
| **GSPT2** | 99.97 % | 300418 | *No OMIM phenotypes* |
| **GTF2E2** | 99.98 % | 189964 | Trichothiodystrophy 6, nonphotosensitive, 616943 (3), Autosomal recessive |
| **GTF2H5** | 99.97 % | 608780 | Trichothiodystrophy 3, photosensitive, 616395 (3), Autosomal recessive |
| **GTF3C3** | 99.72 % | 604888 | Neurodevelopmental disorder with dysmorphic facies, brain anomalies, and seizures, 621201 (3), Autosomal recessive |
| **GTPBP1** | 99.93 % | 602245 | Neurodevelopmental disorder with characteristic facial and ectodermal features and tetraparesis 1, 620888 (3), Autosomal recessive |
| **GTPBP2** | 99.76 % | 607434 | Jaberi-Elahi syndrome, 617988 (3), Autosomal recessive |
| **GTPBP3** | 99.4 % | 608536 | Combined oxidative phosphorylation deficiency 23, 616198 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **GUCY2C** | 99.69 % | 601330 | Diarrhea 6, 614616 (3), Autosomal dominant; Meconium ileus, 614665 (3), Autosomal recessive |
| **GUSB** | 99.73 % | 611499 | Mucopolysaccharidosis VII, 253220 (3), Autosomal recessive |
| **GZF1** | 99.98 % | 613842 | Joint laxity, short stature, and myopia, 617662 (3), Autosomal recessive |
| **H1-4** | 99.98 % | 142220 | Rahman syndrome, 617537 (3), Autosomal dominant |
| **H3-3A** | 93.93 % | 601128 | Bryant-Li-Bhoj neurodevelopmental syndrome 1, 619720 (3), Autosomal dominant |
| **H3-3B** | 100 % | 601058 | Bryant-Li-Bhoj neurodevelopmental syndrome 2, 619721 (3), Autosomal dominant |
| **H4C3** | 99.96 % | 602827 | Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 1, 619758 (3), Autosomal dominant |
| **HAAO** | 99.28 % | 604521 | Vertebral, cardiac, renal, and limb defects syndrome 1, 617660 (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 |
| **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) |
| **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 |
| **HDAC2** | 99.62 % | 605164 | *No OMIM phenotypes* |
| **HDAC3** | 99.78 % | 605166 | *No OMIM phenotypes* |
| **HDAC8** | 99.57 % | 300269 | Cornelia de Lange syndrome 5, 300882 (3), X-linked dominant |
| **HECTD1** | 99.85 % | 618649 | *No OMIM phenotypes* |
| **HECTD4** | 99.87 % | 620209 | Neurodevelopmental disorder with seizures, spasticity, and complete or partial agenesis of the corpus callosum, 620250 (3), Autosomal recessive |
| **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 |
| **HEY2** | 99.78 % | 604674 | *No OMIM phenotypes* |
| **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 |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **HIBCH** | 99.59 % | 610690 | 3-hydroxyisobutryl-CoA hydrolase deficiency, 250620 (3), Autosomal recessive |
| **HIRA** | 99.48 % | 600237 | *No OMIM phenotypes* |
| **HIVEP2** | 99.98 % | 143054 | Intellectual developmental disorder, autosomal dominant 43, 616977 (3), Autosomal dominant |
| **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 |
| **HMGA2** | 90.06 % | 600698 | Silver-Russell syndrome 5, 618908 (3), Autosomal dominant |
| **HMGB1** | 99.95 % | 163905 | *No OMIM phenotypes* |
| **HMX1** | 99.9 % | 142992 | Oculoauricular syndrome, 612109 (3), Autosomal recessive |
| **HNF1B** | 99.77 % | 189907 | Type 2 diabetes mellitus, 125853 (3), Autosomal dominant; Renal cysts and diabetes syndrome, 137920 (3), Autosomal dominant; {Renal cell carcinoma}, 144700 (3) |
| **HNF4A** | 99.93 % | 600281 | Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026 (3), Autosomal dominant; {Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant; MODY, type I, 125850 (3), Autosomal dominant |
| **HNRNPH1** | 99.88 % | 601035 | Neurodevelopmental disorder with craniofacial dysmorphism and skeletal defects, 620083 (3), Autosomal dominant |
| **HNRNPH2** | 99.98 % | 300610 | Intellectual developmental disorder, X-linked syndromic, Bain type, 300986 (3), X-linked dominant |
| **HNRNPK** | 99.59 % | 600712 | Au-Kline syndrome, 616580 (3), Autosomal dominant |
| **HNRNPR** | 98.74 % | 607201 | Neurodevelopmental disorder with dysmorphic facies and skeletal and brain abnormalities, 620073 (3), Autosomal dominant |
| **HNRNPU** | 99.92 % | 602869 | Developmental and epileptic encephalopathy 54, 617391 (3), Autosomal dominant |
| **HOXA1** | 99.98 % | 142955 | Bosley-Salih-Alorainy syndrome, 601536 (3), Autosomal recessive; Athabaskan brainstem dysgenesis syndrome, 601536 (3), Autosomal recessive |
| **HOXA13** | 99.85 % | 142959 | Hand-foot-genital syndrome, 140000 (3), Autosomal dominant; ?Guttmacher syndrome, 176305 (3), Autosomal dominant |
| **HOXB1** | 99.76 % | 142968 | Facial paresis, hereditary congenital, 3, 614744 (3), Autosomal recessive |
| **HOXD13** | 99.85 % | 142989 | Syndactyly, type V, 186300 (3), Autosomal dominant; Synpolydactyly 1, 186000 (3), Autosomal dominant; Brachydactyly, type E, 113300 (3), Autosomal dominant; Brachydactyly, type D, 113200 (3), Autosomal dominant; ?Brachydactyly-syndactyly syndrome, 610713 (3) |
| **HPD** | 99.87 % | 609695 | Hawkinsinuria, 140350 (3), Autosomal dominant; Tyrosinemia, type III, 276710 (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 |
| **HRAS** | 99.97 % | 190020 | Bladder cancer, somatic, 109800 (3); Thyroid carcinoma, follicular, somatic, 188470 (3); Congenital myopathy with excess of muscle spindles, 218040 (3), Autosomal dominant; Nevus sebaceous or woolly hair nevus, somatic, 162900 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Spitz nevus or nevus spilus, somatic, 137550 (3); Costello syndrome, 218040 (3), Autosomal dominant |
| **HS2ST1** | 93.17 % | 604844 | Neurofacioskeletal syndrome with or without renal agenesis, 619194 (3), Autosomal recessive |
| **HSD17B3** | 99.49 % | 605573 | Pseudohermaphroditism, male, with gynecomastia, 264300 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **HSD17B4** | 99.55 % | 601860 | D-bifunctional protein deficiency, 261515 (3), Autosomal recessive; Perrault syndrome 1, 233400 (3), Autosomal recessive |
| **HSF4** | 99.83 % | 602438 | Cataract 5, multiple types, 116800 (3), Autosomal dominant |
| **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 |
| **HUWE1** | 99.73 % | 300697 | Intellectual developmental disorder, X-linked syndromic, Turner type, 309590 (3), X-linked |
| **HYAL2** | 99.86 % | 603551 | Muggenthaler-Chowdhury-Chioza syndrome, 621063 (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 |
| **IBA57** | 99.5 % | 615316 | Multiple mitochondrial dysfunctions syndrome 3, 615330 (3), Autosomal recessive; ?Spastic paraplegia 74, autosomal recessive, 616451 (3), Autosomal recessive |
| **IDH1** | 99.85 % | 147700 | {Glioma, susceptibility to, somatic}, 137800 (3) |
| **IDS** | 99.17 % | 300823 | Mucopolysaccharidosis II, 309900 (3), X-linked recessive |
| **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 |
| **IFITM5** | 99.96 % | 614757 | Osteogenesis imperfecta, type V, 610967 (3), Autosomal dominant |
| **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 |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **IFT81** | 94.88 % | 605489 | Short-rib thoracic dysplasia 19 with or without polydactyly, 617895 (3), Autosomal 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 |
| **IGF2** | 99.9 % | 147470 | Silver-Russell syndrome 3, 616489 (3), Autosomal dominant |
| **IGFBP7** | 99.77 % | 602867 | Retinal arterial macroaneurysm with supravalvular pulmonic stenosis, 614224 (3), Autosomal recessive |
| **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 |
| **IHH** | 99.91 % | 600726 | Acrocapitofemoral dysplasia, 607778 (3), Autosomal recessive; Brachydactyly, type A1, 112500 (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 |
| **IKZF1** | 99.58 % | 603023 | Immunodeficiency, common variable, 13, 616873 (3), Autosomal dominant |
| **IKZF2** | 99.88 % | 606234 | Immunodysregulation, craniofacial anomalies, hearing impairment, athelia, and developmental delay, 621234 (3), Autosomal dominant; Immunodysregulation with variable immunodeficiency and autoimmunity, 621233 (3), Autosomal recessive, Autosomal dominant |
| **IL11RA** | 99.83 % | 600939 | Craniosynostosis and dental anomalies, 614188 (3), Autosomal recessive |
| **IL1RAPL1** | 99.71 % | 300206 | Intellectual developmental disorder, X-linked 21, 300143 (3), X-linked recessive |
| **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 |
| **INPP4A** | 99.29 % | 600916 | Neurodevelopmental disorder with growth impairment, quadriparesis, and poor or absent speech, 621354 (3), Autosomal recessive |
| **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 |
| **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 |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **IQCB1** | 99.56 % | 609237 | Senior-Loken syndrome 5, 609254 (3), Autosomal recessive |
| **IQCE** | 99.87 % | 617631 | Polydactyly, postaxial, type A7, 617642 (3), Autosomal recessive |
| **IRF6** | 99.64 % | 607199 | {Orofacial cleft 6}, 608864 (3), Autosomal dominant; Popliteal pterygium syndrome 1, 119500 (3), Autosomal dominant; van der Woude syndrome 1, 119300 (3), Autosomal dominant |
| **IRX5** | 99.7 % | 606195 | Hamamy syndrome, 611174 (3), Autosomal recessive |
| **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 |
| **ITGA8** | 99.75 % | 604063 | Renal hypodysplasia/aplasia 1, 191830 (3), Autosomal recessive |
| **ITGAV** | 99.26 % | 193210 | Immune dysregulation, neurodevelopmental defects, and colitis, 621375 (3), Autosomal recessive |
| **ITGB4** | 99.89 % | 147557 | Epidermolysis bullosa, junctional 5B, with pyloric atresia, 226730 (3), Autosomal recessive; Epidermolysis bullosa, junctional 5A, intermediate, 619816 (3), Autosomal recessive |
| **JAG1** | 99.94 % | 601920 | ?Deafness, congenital heart defects, and posterior embryotoxon, 617992 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2HH, 619574 (3), Autosomal dominant; Alagille syndrome 1, 118450 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant |
| **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 |
| **KANSL1** | 98.99 % | 612452 | Koolen-De Vries syndrome, 610443 (3), Autosomal dominant |
| **KAT5** | 99.81 % | 601409 | Neurodevelopmental disorder with dysmorphic facies, sleep disturbance, and brain abnormalities, 619103 (3), Autosomal dominant |
| **KAT6A** | 99.88 % | 601408 | Arboleda-Tham syndrome, 616268 (3), Autosomal dominant |
| **KAT6B** | 99.82 % | 605880 | SBBYSS syndrome, 603736 (3), Autosomal dominant; Genitopatellar syndrome, 606170 (3), Autosomal dominant |
| **KATNB1** | 99.46 % | 602703 | Lissencephaly 6, with microcephaly, 616212 (3), Autosomal recessive |
| **KATNIP** | 99.1 % | 616650 | Joubert syndrome 26, 616784 (3), Autosomal recessive |
| **KBTBD2** | 99.85 % | 619393 | *No OMIM phenotypes* |
| **KCNB1** | 99.93 % | 600397 | Developmental and epileptic encephalopathy 26, 616056 (3), Autosomal dominant |
| **KCNC3** | 99.59 % | 176264 | Spinocerebellar ataxia 13, 605259 (3), Autosomal dominant |
| **KCNH1** | 99.86 % | 603305 | Zimmermann-Laband syndrome 1, 135500 (3), Autosomal dominant; Temple-Baraitser syndrome, 611816 (3), Autosomal dominant |
| **KCNH2** | 99.79 % | 152427 | Short QT syndrome 1, 609620 (3); Long QT syndrome 2, 613688 (3), Autosomal dominant |
| **KCNJ1** | 100 % | 600359 | Bartter syndrome, type 2, 241200 (3), Autosomal recessive |
| **KCNJ2** | 100 % | 600681 | Atrial fibrillation, familial, 9, 613980 (3), Autosomal dominant; Andersen syndrome, 170390 (3), Autosomal dominant; Short QT syndrome 3, 609622 (3), Autosomal dominant |
| **KCNJ8** | 99.97 % | 600935 | *No OMIM phenotypes* |
| **KCNK3** | 99.99 % | 603220 | Developmental delay with sleep apnea, 621402 (3), Autosomal dominant; Pulmonary hypertension, primary, 4, 615344 (3), Autosomal dominant |
| **KCNK9** | 100 % | 605874 | Birk-Barel syndrome, 612292 (3), Autosomal dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **KCNN3** | 99.78 % | 602983 | Zimmermann-Laband syndrome 3, 618658 (3), Autosomal dominant |
| **KCNN4** | 99.86 % | 602754 | Dehydrated hereditary stomatocytosis 2, 616689 (3), Autosomal dominant |
| **KCNT1** | 99.52 % | 608167 | Developmental and epileptic encephalopathy 14, 614959 (3), Autosomal dominant; Epilepsy nocturnal frontal lobe, 5, 615005 (3), Autosomal dominant |
| **KCTD1** | 99.84 % | 613420 | Scalp-ear-nipple syndrome, 181270 (3), Autosomal dominant |
| **KDELR2** | 99.83 % | 609024 | Osteogenesis imperfecta, type XXI, 619131 (3), Autosomal recessive |
| **KDM1A** | 96.19 % | 609132 | Cleft palate, psychomotor retardation, and distinctive facial features, 616728 (3), Autosomal dominant; {ACTH-independent macronodular adrenal hyperplasia 3}, 620990 (3), Autosomal dominant |
| **KDM2B** | 99.91 % | 609078 | *No OMIM phenotypes* |
| **KDM4B** | 99.73 % | 609765 | Intellectual developmental disorder, autosomal dominant 65, 619320 (3), Autosomal dominant |
| **KDM5A** | 99.81 % | 180202 | El Hayek-Chahrour neurodevelopmental syndrome, 620820 (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 |
| **KDM6B** | 99.9 % | 611577 | Stolerman neurodevelopmental syndrome, 618505 (3), Autosomal dominant |
| **KDR** | 99.66 % | 191306 | {Hemangioma, capillary infantile, susceptibility to}, 602089 (3), Autosomal dominant; Hemangioma, capillary infantile, somatic, 602089 (3) |
| **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 |
| **KICS2** | 99.71 % | 617420 | Intellectual developmental disorder, autosomal recessive 83, 621100 (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 |
| **KIF11** | 99.79 % | 148760 | Microcephaly with or without chorioretinopathy, lymphedema, or impaired intellectual development, 152950 (3), Autosomal dominant |
| **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 |
| **KIF21A** | 98.45 % | 608283 | Fibrosis of extraocular muscles, congenital, 3B, 135700 (3), Autosomal dominant; Fibrosis of extraocular muscles, congenital, 1, 135700 (3), Autosomal dominant |
| **KIF21B** | 99.07 % | 608322 | *No OMIM phenotypes* |
| **KIF22** | 99.68 % | 603213 | Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546 (3), Autosomal dominant |
| **KIF24** | 99.87 % | 613747 | *No OMIM phenotypes* |
| **KIF26A** | 99.91 % | 613231 | Cortical dysplasia, complex, with other brain malformations 11, 620156 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **KIF2A** | 99.75 % | 602591 | Cortical dysplasia, complex, with other brain malformations 3, 615411 (3), Autosomal dominant |
| **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 |
| **KIF5B** | 99.82 % | 602809 | *No OMIM phenotypes* |
| **KIF5C** | 99.77 % | 604593 | Cortical dysplasia, complex, with other brain malformations 2, 615282 (3), Autosomal dominant |
| **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 |
| **KLF1** | 99.56 % | 600599 | Blood group--Lutheran inhibitor, 111150 (3), Autosomal dominant; [Hereditary persistence of fetal hemoglobin], 613566 (3), Autosomal dominant; Anemia, dyserythropoietic congenital, type IVa, 613673 (3), Autosomal dominant; Anemia, congenital dyserythropoietic, type IVb, 620969 (3), Autosomal recessive |
| **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 |
| **KMT2A** | 99.91 % | 159555 | Wiedemann-Steiner syndrome, 605130 (3), Autosomal dominant |
| **KMT2B** | 99.65 % | 606834 | Intellectual developmental disorder, autosomal dominant 68, 619934 (3), Autosomal dominant; Dystonia 28, childhood-onset, 617284 (3), Autosomal dominant |
| **KMT2C** | 99.28 % | 606833 | Kleefstra syndrome 2, 617768 (3), Autosomal dominant |
| **KMT2D** | 99.72 % | 602113 | Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186 (3), Autosomal dominant; Kabuki syndrome 1, 147920 (3), Autosomal dominant |
| **KMT2E** | 99.82 % | 608444 | O'Donnell-Luria-Rodan syndrome, 618512 (3), Autosomal dominant |
| **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 |
| **KRAS** | 97.26 % | 190070 | Gastric cancer, somatic, 613659 (3); Oculoectodermal syndrome, somatic, 600268 (3); Breast cancer, somatic, 114480 (3); Noonan syndrome 3, 609942 (3), Autosomal dominant; RAS-associated autoimmune leukoproliferative disorder, 614470 (3), Autosomal dominant; Arteriovenous malformation of the brain, somatic, 108010 (3); Lung cancer, somatic, 211980 (3); Pancreatic carcinoma, somatic, 260350 (3); Leukemia, acute myeloid, somatic, 601626 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Cardiofaciocutaneous syndrome 2, 615278 (3), Autosomal dominant; Bladder cancer, somatic, 109800 (3) |
| **KRIT1** | 99.03 % | 604214 | Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations, 116860 (3), Autosomal dominant; Cerebral cavernous malformations-1, 116860 (3), Autosomal dominant; Cavernous malformations of CNS and retina, 116860 (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 |
| **KYNU** | 99.13 % | 605197 | ?Hydroxykynureninuria, 236800 (3), Autosomal recessive; Vertebral, cardiac, renal, and limb defects syndrome 2, 617661 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **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 |
| **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 |
| **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 |
| **LAMC3** | 99.83 % | 604349 | Cortical malformations, occipital, 614115 (3), Autosomal recessive |
| **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 |
| **LARS2** | 99.68 % | 604544 | Perrault syndrome 4, 615300 (3), Autosomal recessive; Hydrops, lactic acidosis, and sideroblastic anemia, 617021 (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 |
| **LDB1** | 99.63 % | 603451 | *No OMIM phenotypes* |
| **LDB3** | 98.75 % | 605906 | Left ventricular noncompaction 3, 601493 (3), Autosomal dominant; Cardiomyopathy, dilated, 2L, 621237 (3), Autosomal recessive; Cardiomyopathy, hypertrophic, 24, 601493 (3), Autosomal dominant; Myopathy, myofibrillar, 4, 609452 (3), Autosomal dominant; Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 (3), Autosomal dominant |
| **LEF1** | 99.81 % | 153245 | Ectodermal dysplasia 17 with or without limb malformations, 621224 (3), Autosomal dominant |
| **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 |
| **LHX3** | 99.79 % | 600577 | Pituitary hormone deficiency, combined, 3, 221750 (3), Autosomal recessive |
| **LHX4** | 99.92 % | 602146 | Pituitary hormone deficiency, combined, 4, 262700 (3), Autosomal dominant |
| **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 |
| **LIG4** | 99.99 % | 601837 | LIG4 syndrome, 606593 (3), Autosomal recessive; {Multiple myeloma, resistance to}, 254500 (3), Somatic mutation |
| **LINS1** | 99.88 % | 610350 | Intellectual developmental disorder, autosomal recessive 27, 614340 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **LIPA** | 99.94 % | 613497 | Wolman disease, 620151 (3), Autosomal recessive; Cholesteryl ester storage disease, 278000 (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 |
| **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 |
| **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 |
| **LMNB1** | 99.59 % | 150340 | Microcephaly 26, primary, autosomal dominant, 619179 (3), Autosomal dominant |
| **LMNB2** | 99.74 % | 150341 | Microcephaly 27, primary, autosomal dominant, 619180 (3), Autosomal dominant; ?Epilepsy, progressive myoclonic, 9, 616540 (3), Autosomal recessive; {Lipodystrophy, partial, acquired, susceptibility to}, 608709 (3), Autosomal dominant |
| **LMOD2** | 99.98 % | 608006 | Cardiomyopathy, dilated, 2G, 619897 (3), Autosomal recessive |
| **LMOD3** | 99.97 % | 616112 | Nemaline myopathy 10, 616165 (3), Autosomal recessive |
| **LMX1B** | 99.92 % | 602575 | Focal segmental glomerulosclerosis 10, 256020 (3), Autosomal dominant; Nail-patella syndrome, 161200 (3), Autosomal dominant |
| **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 |
| **LOX** | 99.8 % | 153455 | Aortic aneurysm, familial thoracic 10, 617168 (3), Autosomal dominant |
| **LRIG2** | 98.22 % | 608869 | Urofacial syndrome 2, 615112 (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 |
| **LRRC32** | 99.96 % | 137207 | Cleft palate, proliferative retinopathy, and developmental delay, 619074 (3), Autosomal recessive |
| **LRRC56** | 99.77 % | 618227 | Ciliary dyskinesia, primary, 39, 618254 (3), Autosomal recessive |
| **LRRC8C** | 99.38 % | 612889 | TIMES syndrome, 621056 (3), Autosomal dominant |
| **LRRK1** | 99.93 % | 610986 | Osteosclerotic metaphyseal dysplasia, 615198 (3), Autosomal recessive |
| **LSM1** | 99.73 % | 607281 | FICUS syndrome, 621193 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **LSM11** | 99.93 % | 617910 | ?Aicardi-Goutieres syndrome 8, 619486 (3), Autosomal recessive |
| **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 |
| **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 |
| **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 |
| **MAB21L1** | 100 % | 601280 | Cerebellar, ocular, craniofacial, and genital syndrome, 618479 (3), Autosomal recessive |
| **MAB21L2** | 99.99 % | 604357 | Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877 (3), Autosomal recessive, Autosomal dominant |
| **MACF1** | 98.83 % | 608271 | Lissencephaly 9 with complex brainstem malformation, 618325 (3), Autosomal dominant |
| **MAF** | 99.89 % | 177075 | Cataract 21, multiple types, 610202 (3), Autosomal dominant; Ayme-Gripp syndrome, 601088 (3), Autosomal dominant |
| **MAFB** | 99.57 % | 608968 | Duane retraction syndrome 3, 617041 (3), Autosomal dominant; Multicentric carpotarsal osteolysis syndrome, 166300 (3), Autosomal dominant |
| **MAGED2** | 99.94 % | 300470 | Bartter syndrome, type 5, antenatal, transient, 300971 (3), X-linked recessive |
| **MAGEL2** | 99.99 % | 605283 | Schaaf-Yang syndrome, 615547 (3), Autosomal dominant |
| **MAL** | 99.57 % | 188860 | ?Leukodystrophy, hypomyelinating, 28, 620978 (3), Autosomal recessive |
| **MAMLD1** | 99.9 % | 300120 | Hypospadias 2, X-linked, 300758 (3), X-linked recessive |
| **MAN1B1** | 99.8 % | 604346 | Rafiq syndrome, 614202 (3), Autosomal recessive |
| **MAN2B2** | 99.82 % | 618899 | Congenital disorder of glycosylation type 1EE with or without immunodeficiency, 621140 (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 |
| **MAP1B** | 99.99 % | 157129 | ?Deafness, autosomal dominant 83, 619808 (3), Autosomal dominant; Periventricular nodular heterotopia 9, 618918 (3), Autosomal dominant |
| **MAP2K1** | 99.43 % | 176872 | Cardiofaciocutaneous syndrome 3, 615279 (3), Autosomal dominant; Melorheostosis, isolated, somatic mosaic, 155950 (3) |
| **MAP2K2** | 99.93 % | 601263 | Cardiofaciocutaneous syndrome 4, 615280 (3), Autosomal dominant |
| **MAP3K1** | 99.76 % | 600982 | 46XY sex reversal 6, 613762 (3), Autosomal dominant |
| **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 |
| **MAP3K7** | 98.99 % | 602614 | Frontometaphyseal dysplasia 2, 617137 (3), Autosomal dominant; Cardiospondylocarpofacial syndrome, 157800 (3), Autosomal dominant |
| **MAP4K4** | 98.85 % | 604666 | *No OMIM phenotypes* |
| **MAPK1** | 99.73 % | 176948 | Noonan syndrome 13, 619087 (3), Autosomal dominant |
| **MAPK8IP3** | 99.91 % | 605431 | Neurodevelopmental disorder with or without variable brain abnormalities, 618443 (3), Autosomal dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **MAPKAPK5** | 99.78 % | 606723 | Neurocardiofaciodigital syndrome, 619869 (3), Autosomal recessive |
| **MAPRE2** | 99.74 % | 605789 | Symmetric circumferential skin creases, congenital, 2, 616734 (3), Autosomal dominant |
| **MASP1** | 99.57 % | 600521 | 3MC syndrome 1, 257920 (3), Autosomal recessive |
| **MAST1** | 99.84 % | 612256 | Mega-corpus-callosum syndrome with cerebellar hypoplasia and cortical malformations, 618273 (3), Autosomal dominant |
| **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 |
| **MAX** | 99.8 % | 154950 | Polydactyly-macrocephaly syndrome, 620712 (3), Autosomal dominant; {Pheochromocytoma, susceptibility to}, 171300 (3), Autosomal dominant |
| **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 |
| **MCIDAS** | 99.93 % | 614086 | Ciliary dyskinesia, primary, 42, 618695 (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 |
| **MDFIC** | 99.59 % | 614511 | Lymphatic malformation 12, 620014 (3), Autosomal recessive |
| **MDH2** | 98.1 % | 154100 | Developmental and epileptic encephalopathy 51, 617339 (3), Autosomal recessive |
| **MECOM** | 99.85 % | 165215 | Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738 (3), Autosomal dominant |
| **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 |
| **MED13L** | 99.9 % | 608771 | Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789 (3), Autosomal dominant |
| **MED17** | 99.66 % | 603810 | Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668 (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 |
| **MEF2C** | 98.93 % | 600662 | Chromosome 5q14.3 deletion syndrome, 613443 (4), Autosomal dominant; Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language, 613443 (3), Autosomal dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **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 |
| **MEIS2** | 99.82 % | 601740 | Cleft palate, cardiac defects, and impaired intellectual development, 600987 (3), Autosomal dominant |
| **MEOX1** | 99.87 % | 600147 | Klippel-Feil syndrome 2, 214300 (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 |
| **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 |
| **MGAT2** | 99.99 % | 602616 | Congenital disorder of glycosylation, type IIa, 212066 (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 |
| **MID1** | 99.87 % | 300552 | Opitz GBBB syndrome, 300000 (3), X-linked recessive |
| **MINPP1** | 99.84 % | 605391 | {Thyroid carcinoma, follicular}, 188470 (3), Somatic mutation, Autosomal dominant; Pontocerebellar hypoplasia, type 16, 619527 (3), Autosomal recessive |
| **MIR17HG** | 0 % | 609415 | *No OMIM phenotypes* |
| **MITF** | 99.9 % | 156845 | Waardenburg syndrome, type 2A, 193510 (3), Autosomal dominant; {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456 (3); Tietz albinism-deafness syndrome, 103500 (3), Autosomal dominant; COMMAD syndrome, 617306 (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 |
| **MLYCD** | 99.82 % | 606761 | Malonyl-CoA decarboxylase deficiency, 248360 (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 |
| **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 |
| **MMP15** | 99.53 % | 602261 | *No OMIM phenotypes* |
| **MMP2** | 99.16 % | 120360 | Multicentric osteolysis, nodulosis, and arthropathy, 259600 (3), Autosomal recessive |
| **MMP21** | 99.82 % | 608416 | Heterotaxy, visceral, 7, autosomal, 616749 (3), Autosomal recessive |
| **MMP9** | 99.8 % | 120361 | Metaphyseal anadysplasia 2, 613073 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **MN1** | 99.99 % | 156100 | CEBALID syndrome, 618774 (3), Autosomal dominant; Meningioma, 607174 (3), Autosomal dominant |
| **MNS1** | 99.9 % | 610766 | Heterotaxy, visceral, 9, autosomal, with male infertility, 618948 (3), Autosomal recessive |
| **MNX1** | 99.01 % | 142994 | Currarino syndrome, 176450 (3), Autosomal dominant |
| **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 |
| **MPC2** | 99.27 % | 614737 | *No OMIM phenotypes* |
| **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 |
| **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 |
| **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 |
| **MRAS** | 99.91 % | 608435 | Noonan syndrome 11, 618499 (3), Autosomal dominant |
| **MRPS14** | 99.84 % | 611978 | ?Combined oxidative phosphorylation deficiency 38, 618378 (3), Autosomal recessive |
| **MRPS16** | 99.43 % | 609204 | Combined oxidative phosphorylation deficiency 2, 610498 (3), Autosomal recessive |
| **MRPS22** | 99.83 % | 605810 | Ovarian dysgenesis 7, 618117 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 5, 611719 (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 |
| **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 |
| **MSL2** | 99.99 % | 614802 | Karayol-Borroto-Haghshenas neurodevelopmental syndrome, 620985 (3), Autosomal dominant |
| **MSL3** | 99.83 % | 300609 | Basilicata-Akhtar syndrome, 301032 (3), X-linked dominant |
| **MSMO1** | 99.89 % | 607545 | Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834 (3), Autosomal recessive |
| **MSTO1** | 68.42 % | 617619 | Myopathy, mitochondrial, and ataxia, 617675 (3), Autosomal recessive, Autosomal dominant |
| **MSX1** | 99.87 % | 142983 | Tooth agenesis, selective, 1, with or without orofacial cleft, 106600 (3), Autosomal dominant; Ectodermal dysplasia 3, Witkop type, 189500 (3), Autosomal dominant; Orofacial cleft 5, 608874 (3), Autosomal dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **MSX2** | 99.96 % | 123101 | Parietal foramina with cleidocranial dysplasia, 168550 (3), Autosomal dominant; Craniosynostosis 2, 604757 (3), Autosomal dominant; Parietal foramina 1, 168500 (3), Autosomal dominant |
| **MT-TE** | 2.79 % | | *No OMIM phenotypes* |
| **MT-TL1** | 2.67 % | | *No OMIM phenotypes* |
| **MTFMT** | 99.93 % | 611766 | Combined oxidative phosphorylation deficiency 15, 614947 (3), Autosomal recessive; Mitochondrial complex I deficiency, nuclear type 27, 618248 (3), Autosomal recessive |
| **MTM1** | 99.71 % | 300415 | Myopathy, centronuclear, X-linked, 310400 (3), X-linked recessive |
| **MTO1** | 98.56 % | 614667 | Combined oxidative phosphorylation deficiency 10, 614702 (3), Autosomal recessive |
| **MTOR** | 99.78 % | 601231 | Focal cortical dysplasia, type II, somatic, 607341 (3); Smith-Kingsmore syndrome, 616638 (3), Autosomal dominant |
| **MTPAP** | 99.9 % | 613669 | ?Spastic ataxia 4, autosomal recessive, 613672 (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 |
| **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 |
| **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 |
| **MYBBP1A** | 99.89 % | 604885 | *No OMIM phenotypes* |
| **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 |
| **MYCN** | 99.94 % | 164840 | Feingold syndrome 1, 164280 (3), Autosomal dominant; Megalencephaly-polydactyly syndrome, 620748 (3), Autosomal dominant |
| **MYH10** | 99.89 % | 160776 | *No OMIM phenotypes* |
| **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 |
| **MYH6** | 99.87 % | 160710 | ?Atrial septal defect 3, 614089 (3), Autosomal dominant; {Sick sinus syndrome 3}, 614090 (3); Cardiomyopathy, dilated, 1EE, 613252 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 14, 613251 (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 |
| **MYH8** | 99.99 % | 160741 | Carney complex variant, 608837 (3); Trismus-pseudocamptodactyly syndrome, 158300 (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 |
| **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 |
| **MYO18B** | 99.89 % | 607295 | Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549 (3), Autosomal recessive |
| **MYO9A** | 99.85 % | 604875 | Myasthenic syndrome, congenital, 24, presynaptic, 618198 (3), Autosomal recessive |
| **MYOCD** | 99.95 % | 606127 | Megabladder, congenital, 618719 (3), Autosomal dominant |
| **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 |
| **MYRF** | 99.59 % | 608329 | Nanophthalmos 1, 600165 (3), Autosomal dominant; Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113 (3), Autosomal dominant; Cardiac-urogenital syndrome, 618280 (3), Autosomal dominant |
| **MYSM1** | 95.13 % | 612176 | Bone marrow failure syndrome 4, 618116 (3), Autosomal recessive |
| **MYT1** | 99.94 % | 600379 | *No OMIM phenotypes* |
| **NAA10** | 99.94 % | 300013 | Microphthalmia, syndromic 1, 309800 (3), X-linked; Ogden syndrome, 300855 (3), X-linked dominant, X-linked recessive |
| **NAA15** | 99.72 % | 608000 | Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787 (3), Autosomal dominant |
| **NACC1** | 99.98 % | 610672 | Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393 (3), Autosomal dominant |
| **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 |
| **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 |
| **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 |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **NANS** | 99.82 % | 605202 | Spondyloepimetaphyseal dysplasia, Genevieve type, 610442 (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 |
| **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 |
| **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 |
| **NDUFA10** | 99.92 % | 603835 | Mitochondrial complex I deficiency, nuclear type 22, 618243 (3), Autosomal recessive |
| **NDUFA6** | 99.93 % | 602138 | Mitochondrial complex I deficiency, nuclear type 33, 618253 (3), Autosomal recessive |
| **NDUFAF2** | 99.96 % | 609653 | Mitochondrial complex I deficiency, nuclear type 10, 618233 (3), Autosomal recessive |
| **NDUFAF5** | 99.87 % | 612360 | Mitochondrial complex I deficiency, nuclear type 16, 618238 (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 |
| **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 |
| **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 |
| **NEDD4L** | 99.58 % | 606384 | Periventricular nodular heterotopia 7, 617201 (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 |
| **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) |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **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 |
| **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 |
| **NEUROD1** | 99.99 % | 601724 | {Type 2 diabetes mellitus, susceptibility to}, 125853 (3), Autosomal dominant; Maturity-onset diabetes of the young 6, 606394 (3) |
| **NEXMIF** | 99.97 % | 300524 | Intellectual developmental disorder, X-linked 98, 300912 (3), X-linked dominant |
| **NEXN** | 98.12 % | 613121 | Cardiomyopathy, dilated, 2M, autosomal recessive, 621261 (3), Autosomal recessive; Cardiomyopathy, dilated, 1CC, 613122 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 20, 613876 (3), Autosomal dominant |
| **NF1** | 99.83 % | 613113 | Watson syndrome, 193520 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, 607785 (3), Somatic mutation, Autosomal dominant; Neurofibromatosis, familial spinal, 162210 (3), Autosomal dominant; Neurofibromatosis, type 1, 162200 (3), Autosomal dominant; Neurofibromatosis-Noonan syndrome, 601321 (3), Autosomal dominant |
| **NFIA** | 97.78 % | 600727 | Brain malformations with or without urinary tract defects, 613735 (3), Autosomal dominant |
| **NFIB** | 99.84 % | 600728 | Macrocephaly, acquired, with impaired intellectual development, 618286 (3), Autosomal dominant |
| **NFIX** | 99.82 % | 164005 | Marshall-Smith syndrome, 602535 (3), Autosomal dominant; Malan syndrome, 614753 (3), Autosomal dominant |
| **NHEJ1** | 99.61 % | 611290 | Microphthalmia/coloboma 13, 620968 (3), Autosomal recessive; Immunodeficiency 124, severe combined, 611291 (3), Autosomal recessive |
| **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 |
| **NID1** | 99.81 % | 131390 | *No OMIM phenotypes* |
| **NIPAL4** | 99.97 % | 609383 | Ichthyosis, congenital, autosomal recessive 6, 612281 (3), Autosomal recessive |
| **NIPBL** | 99.72 % | 608667 | Cornelia de Lange syndrome 1, 122470 (3), Autosomal dominant |
| **NKX2-5** | 99.89 % | 600584 | Hypoplastic left heart syndrome 2, 614435 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant; Hypothyroidism, congenital nongoitrous, 5, 225250 (3), Autosomal dominant; Conotruncal heart malformations, variable, 217095 (3); Ventricular septal defect 3, 614432 (3), Autosomal dominant; Atrial septal defect 7, with or without AV conduction defects, 108900 (3), Autosomal dominant |
| **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 |
| **NLRP3** | 99.95 % | 606416 | CINCA syndrome, 607115 (3), Autosomal dominant; Familial cold inflammatory syndrome 1, 120100 (3), Autosomal dominant; Keratoendothelitis fugax hereditaria, 148200 (3), Autosomal dominant; Deafness, autosomal dominant 34, with or without inflammation, 617772 (3), Autosomal dominant; Muckle-Wells syndrome, 191900 (3), Autosomal dominant |
| **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* |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **NOG** | 99.02 % | 602991 | Symphalangism, proximal, 1A, 185800 (3), Autosomal dominant; Brachydactyly, type B2, 611377 (3), Autosomal dominant; Stapes ankylosis with broad thumbs and toes, 184460 (3), Autosomal dominant; Tarsal-carpal coalition syndrome, 186570 (3), Autosomal dominant; Multiple synostoses syndrome 1, 186500 (3), Autosomal dominant |
| **NONO** | 99.89 % | 300084 | Intellectual developmental disorder, X-linked syndromic 34, 300967 (3), X-linked |
| **NOTCH1** | 99.8 % | 190198 | Adams-Oliver syndrome 5, 616028 (3), Autosomal dominant; Aortic valve disease 1, 109730 (3), Autosomal dominant |
| **NOTCH2** | 98.36 % | 600275 | Alagille syndrome 2, 610205 (3), Autosomal dominant; Hajdu-Cheney syndrome, 102500 (3), Autosomal dominant |
| **NOTCH3** | 99.56 % | 600276 | Lateral meningocele syndrome, 130720 (3), Autosomal dominant; Lipodystrophy, familial partial, type 1, 608600 (3), Autosomal dominant; Cerebral arteriopathy, autosomal recessive, with subcortical infarcts and leukoencephalopathy 1, 621295 (3), Autosomal recessive; ?Myofibromatosis, infantile 2, 615293 (3), Autosomal dominant; Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy 1, 125310 (3), Autosomal recessive, Autosomal dominant |
| **NOVA2** | 99.75 % | 601991 | Neurodevelopmental disorder with or without autistic features and/or structural brain abnormalities, 618859 (3), Autosomal dominant |
| **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 |
| **NPNT** | 99.66 % | 610306 | *No OMIM phenotypes* |
| **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 |
| **NR0B1** | 99.94 % | 300473 | Adrenal hypoplasia, congenital, 300200 (3), X-linked recessive; 46XY sex reversal 2, dosage-sensitive, 300018 (3), X-linked |
| **NR2F1** | 99.88 % | 132890 | Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722 (3), Autosomal dominant |
| **NR2F2** | 99.91 % | 107773 | 46XX sex reversal 5, 618901 (3), Autosomal dominant; Congenital heart defects, multiple types, 4, 615779 (3), Autosomal dominant |
| **NR5A1** | 99.09 % | 184757 | 46XX sex reversal 4, 617480 (3), Autosomal dominant; Premature ovarian failure 7, 612964 (3), Autosomal dominant; 46XY sex reversal 3, 612965 (3), Autosomal dominant; Adrenocortical insufficiency, 612964 (3), Autosomal dominant; Spermatogenic failure 8, 613957 (3), Autosomal dominant |
| **NR6A1** | 99.92 % | 602778 | Oculovertebral syndrome, 621277 (3), Autosomal dominant |
| **NRAS** | 99.69 % | 164790 | Noonan syndrome 6, 613224 (3), Autosomal dominant; ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 (3); Melanocytic nevus syndrome, congenital, somatic, 137550 (3); Epidermal nevus, somatic, 162900 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Thyroid carcinoma, follicular, somatic, 188470 (3); Neurocutaneous melanosis, somatic, 249400 (3); Colorectal cancer, somatic, 114500 (3) |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **NSD1** | 99.93 % | 606681 | Sotos syndrome, 117550 (3), Autosomal dominant |
| **NSD2** | 99.84 % | 602952 | Rauch-Steindl syndrome, 619695 (3), Autosomal dominant |
| **NSDHL** | 99.66 % | 300275 | CK syndrome, 300831 (3), X-linked recessive; CHILD syndrome, 308050 (3), X-linked dominant |
| **NSRP1** | 99.89 % | 616173 | Neurodevelopmental disorder with spasticity, seizures, and brain abnormalities, 620001 (3), Autosomal recessive |
| **NSUN6** | 99.82 % | 617199 | Intellectual developmental disorder, autosomal recessive 82, 620779 (3), Autosomal recessive |
| **NTRK2** | 99.57 % | 600456 | Developmental and epileptic encephalopathy 58, 617830 (3), Autosomal dominant; Obesity, hyperphagia, and developmental delay, 613886 (3), Autosomal dominant |
| **NUBPL** | 99.77 % | 613621 | Mitochondrial complex I deficiency, nuclear type 21, 618242 (3), Autosomal recessive |
| **NUDCD2** | 99.95 % | 620136 | *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 |
| **NUP88** | 99.83 % | 602552 | Fetal akinesia deformation sequence 4, 618393 (3), Autosomal recessive |
| **NUS1** | 99.55 % | 610463 | Intellectual developmental disorder, autosomal dominant 55, with seizures, 617831 (3), Autosomal dominant; ?Congenital disorder of glycosylation, type 1aa, 617082 (3), Autosomal recessive |
| **NXN** | 99.94 % | 612895 | Robinow syndrome, autosomal recessive 2, 618529 (3), Autosomal recessive |
| **OBSL1** | 99.88 % | 610991 | 3-M syndrome 2, 612921 (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 |
| **ODC1** | 99.65 % | 165640 | Bachmann-Bupp syndrome, 619075 (3), Autosomal dominant |
| **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 |
| **OPHN1** | 99.73 % | 300127 | Intellectual developmental disorder, X-linked syndromic, Billuart type, 300486 (3), X-linked recessive |
| **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 |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **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 |
| **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 |
| **OTX2** | 99.98 % | 600037 | Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125 (3), Autosomal dominant; Pituitary hormone deficiency, combined, 6, 613986 (3), Autosomal dominant; Microphthalmia, syndromic 5, 610125 (3), Autosomal dominant |
| **P3H1** | 99.36 % | 610339 | Osteogenesis imperfecta, type VIII, 610915 (3), Autosomal recessive |
| **P4HB** | 99.98 % | 176790 | Cole-Carpenter syndrome 1, 112240 (3), Autosomal dominant |
| **PACS1** | 99.57 % | 607492 | Schuurs-Hoeijmakers syndrome, 615009 (3), Autosomal dominant |
| **PACS2** | 99.88 % | 610423 | Developmental and epileptic encephalopathy 66, 618067 (3), Autosomal dominant |
| **PAFAH1B1** | 99.88 % | 601545 | Subcortical laminar heterotopia, 607432 (3), Autosomal dominant; Lissencephaly 1, 607432 (3), Autosomal dominant |
| **PAICS** | 99.98 % | 172439 | ?Phosphoribosylaminoimidazole carboxylase deficiency, 619859 (3), Autosomal recessive |
| **PAK2** | 99.71 % | 605022 | ?Knobloch syndrome 2, 618458 (3), Autosomal dominant |
| **PAK3** | 99.76 % | 300142 | Intellectual developmental disorder, X-linked 30, 300558 (3), X-linked recessive |
| **PALB2** | 99.33 % | 610355 | {Breast-ovarian cancer, familial, susceptibility to, 5}, 620442 (3), Autosomal dominant; {Pancreatic cancer, susceptibility to, 3}, 613348 (3), Autosomal dominant; Fanconi anemia, complementation group N, 610832 (3), Autosomal 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 |
| **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 |
| **PARP6** | 99.9 % | 619439 | *No OMIM phenotypes* |
| **PATJ** | 96.31 % | 603199 | *No OMIM phenotypes* |
| **PAX1** | 99.8 % | 167411 | Otofaciocervical syndrome 2 with T-cell deficiency, 615560 (3), Autosomal recessive |
| **PAX2** | 99.8 % | 167409 | Glomerulosclerosis, focal segmental, 7, 616002 (3), Autosomal dominant; Papillorenal syndrome, 120330 (3), Autosomal dominant |
| **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 |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **PAX6** | 99.91 % | 607108 | Optic nerve hypoplasia, 165550 (3), Autosomal dominant; Cataract with late-onset corneal dystrophy, 106210 (3), Autosomal dominant; Microphthalmia/coloboma 12, 120200 (3), Autosomal dominant; ?Coloboma of optic nerve, 120430 (3), Autosomal dominant; Aniridia, 106210 (3), Autosomal dominant; Anterior segment dysgenesis 5, multiple subtypes, 604229 (3), Autosomal dominant; ?Morning glory disc anomaly, 120430 (3), Autosomal dominant; Foveal hypoplasia 1, 136520 (3), Autosomal dominant; Keratitis, 148190 (3), Autosomal dominant |
| **PAX7** | 99.3 % | 167410 | Congenital myopathy 19, 618578 (3), Autosomal recessive; Rhabdomyosarcoma 2, alveolar, 268220 (3), Somatic mutation |
| **PAX8** | 99.38 % | 167415 | Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700 (3), Autosomal dominant |
| **PBX1** | 99.56 % | 176310 | Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641 (3), Autosomal dominant |
| **PC** | 99.85 % | 608786 | Pyruvate carboxylase deficiency, 266150 (3), Autosomal recessive |
| **PCDH12** | 99.97 % | 605622 | Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280 (3), Autosomal recessive |
| **PCGF2** | 99.25 % | 600346 | Turnpenny-Fry syndrome, 618371 (3), Autosomal dominant |
| **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 |
| **PCYT1A** | 99.93 % | 123695 | Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940 (3), Autosomal recessive; Lipodystrophy, congenital generalized, type 5, 620680 (3), Autosomal recessive |
| **PDCD10** | 99.52 % | 609118 | Cerebral cavernous malformations-3, 603285 (3), Autosomal dominant |
| **PDCD2** | 99.98 % | 600866 | *No OMIM phenotypes* |
| **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 |
| **PDE12** | 99.9 % | 616519 | *No OMIM phenotypes* |
| **PDE3A** | 99.9 % | 123805 | Hypertension and brachydactyly syndrome, 112410 (3), Autosomal dominant |
| **PDE4D** | 99.76 % | 600129 | Acrodysostosis 2, with or without hormone resistance, 614613 (3), Autosomal dominant |
| **PDE6D** | 99.44 % | 602676 | Joubert syndrome 22, 615665 (3), Autosomal recessive |
| **PDE6H** | 99.97 % | 601190 | Achromatopsia 6, 610024 (3), Autosomal recessive |
| **PDGFRB** | 99.81 % | 173410 | Premature aging syndrome, Penttinen type, 601812 (3), Autosomal dominant; ?Ocular pterygium-digital keloid dysplasia syndrome, 621091 (3), Autosomal dominant; Kosaki overgrowth syndrome, 616592 (3), Autosomal dominant; Myofibromatosis, infantile, 1, 228550 (3), Autosomal dominant; Basal ganglia calcification, idiopathic, 4, 615007 (3), Autosomal dominant |
| **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 |
| **PDIA6** | 99.49 % | 611099 | *No OMIM phenotypes* |
| **PDSS1** | 98.29 % | 607429 | Coenzyme Q10 deficiency, primary, 2, 614651 (3), Autosomal recessive |
| **PEPD** | 99.42 % | 613230 | Prolidase deficiency, 170100 (3), Autosomal recessive |
| **PET100** | 100 % | 614770 | Mitochondrial complex IV deficiency, nuclear type 12, 619055 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **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 |
| **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 |
| **PGM1** | 97.39 % | 171900 | Congenital disorder of glycosylation, type It, 614921 (3), Autosomal recessive |
| **PGM3** | 99.78 % | 172100 | Immunodeficiency 23, 615816 (3), Autosomal recessive |
| **PHEX** | 99.73 % | 300550 | Hypophosphatemic rickets, X-linked dominant, 307800 (3), X-linked dominant |
| **PHF21A** | 99.64 % | 608325 | Intellectual developmental disorder with behavioral abnormalities and craniofacial dysmorphism with or without seizures, 618725 (3), Autosomal dominant |
| **PHF5A** | 99.98 % | 617846 | *No OMIM phenotypes* |
| **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 |
| **PHIP** | 99.61 % | 612870 | Chung-Jansen syndrome, 617991 (3), Autosomal dominant |
| **PHLDB1** | 99.88 % | 612834 | Osteogenesis imperfecta, type XXIII, 620639 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **PHOX2B** | 99.93 % | 603851 | {Neuroblastoma, susceptibility to, 2}, 613013 (3); Neuroblastoma with Hirschsprung disease, 613013 (3); Central hypoventilation syndrome, congenital, 1, with or without Hirschsprung disease, 209880 (3), Autosomal dominant |
| **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 |
| **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 |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **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 |
| **PIGY** | 99.96 % | 610662 | Hyperphosphatasia with impaired intellectual development syndrome 6, 616809 (3), Autosomal recessive |
| **PIK3C2A** | 99.81 % | 603601 | Oculoskeletodental syndrome, 618440 (3), Autosomal recessive |
| **PIK3CA** | 99.63 % | 171834 | Hemifacial myohyperplasia, somatic, 606773 (3); CLOVE syndrome, somatic, 612918 (3); Hepatocellular carcinoma, somatic, 114550 (3); Breast cancer, somatic, 114480 (3); Cerebral cavernous malformations 4, somatic, 619538 (3); Ovarian cancer, somatic, 167000 (3); Colorectal cancer, somatic, 114500 (3); Macrodactyly, somatic, 155500 (3); CLAPO syndrome, somatic, 613089 (3); Keratosis, seborrheic, somatic, 182000 (3); Gastric cancer, somatic, 613659 (3); Nonsmall cell lung cancer, somatic, 211980 (3); Nevus, epidermal, somatic mosaic, 162900 (3); Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 (3); Cowden syndrome 5, 615108 (3) |
| **PIK3R1** | 99.65 % | 171833 | Immunodeficiency 36, 616005 (3), Autosomal dominant; SHORT syndrome, 269880 (3), Autosomal dominant; Agammaglobulinemia 7, autosomal recessive, 615214 (3), Autosomal recessive |
| **PIK3R2** | 99.28 % | 603157 | Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387 (3), Autosomal dominant |
| **PIP5K1C** | 99.81 % | 606102 | Lethal congenital contractural syndrome 3, 611369 (3), Autosomal recessive |
| **PISD** | 99.94 % | 612770 | Liberfarb syndrome, 618889 (3), Autosomal recessive |
| **PITX1** | 99.83 % | 602149 | Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800 (3), Autosomal dominant |
| **PITX2** | 99.85 % | 601542 | Ring dermoid of cornea, 180550 (3), Autosomal dominant; Axenfeld-Rieger syndrome, type 1, 180500 (3), Autosomal dominant; Anterior segment dysgenesis 4, 137600 (3), Autosomal dominant |
| **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 |
| **PKD1** | 99.48 % | 601313 | Polycystic kidney disease 1, 173900 (3), Autosomal dominant |
| **PKD1L1** | 99.29 % | 609721 | Heterotaxy, visceral, 8, autosomal, 617205 (3), Autosomal recessive |
| **PKD2** | 99.79 % | 173910 | Polycystic kidney disease 2, 613095 (3), Autosomal dominant |
| **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 |
| **PKP2** | 98.7 % | 602861 | Arrhythmogenic right ventricular dysplasia 9, 609040 (3), Autosomal dominant |
| **PLAA** | 99.59 % | 603873 | Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527 (3), Autosomal recessive |
| **PLAG1** | 99.94 % | 603026 | Adenomas, salivary gland pleomorphic, somatic, 181030 (3); Silver-Russell syndrome 4, 618907 (3), Autosomal dominant |
| **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 |
| **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 |
| **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 |
| **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 |
| **PLXNB2** | 99.82 % | 604293 | *No OMIM phenotypes* |
| **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 |
| **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 |
| **PNPLA1** | 99.92 % | 612121 | Ichthyosis, congenital, autosomal recessive 10, 615024 (3), Autosomal recessive |
| **PNPLA8** | 99.88 % | 612123 | Mitochondrial myopathy with lactic acidosis, 251950 (3), Autosomal recessive |
| **POC1A** | 99.49 % | 614783 | Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813 (3), Autosomal recessive |
| **POGZ** | 99.15 % | 614787 | White-Sutton syndrome, 616364 (3), Autosomal dominant |
| **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 |
| **POLD1** | 99.66 % | 174761 | Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381 (3), Autosomal dominant; Immunodeficiency 120, 620836 (3), Autosomal recessive; {Colorectal cancer, susceptibility to, 10}, 612591 (3), Autosomal dominant |
| **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 |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **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 |
| **POLR1A** | 99.5 % | 616404 | Leukodystrophy, hypomyelinating, 27, 620675 (3), Autosomal recessive; Acrofacial dysostosis, Cincinnati type, 616462 (3), Autosomal dominant |
| **POLR1B** | 99.59 % | 602000 | Treacher-Collins syndrome 4, 618939 (3), Autosomal dominant |
| **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 |
| **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 |
| **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 |
| **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 |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **POU1F1** | 99.91 % | 173110 | Pituitary hormone deficiency, combined or isolated, 1, 613038 (3), Autosomal recessive, Autosomal dominant |
| **POU3F3** | 99.96 % | 602480 | Snijders Blok-Fisher syndrome, 618604 (3), Autosomal dominant |
| **PPFIA3** | 99.65 % | 603144 | Paul-Chao neurodevelopmental syndrome, 621122 (3), Autosomal dominant |
| **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 |
| **PPP1CB** | 99.51 % | 600590 | Noonan syndrome-like disorder with loose anagen hair 2, 617506 (3), Autosomal dominant |
| **PPP1R12A** | 98.24 % | 602021 | Genitourinary and/or/brain malformation syndrome, 618820 (3), Autosomal dominant |
| **PPP1R13L** | 99.57 % | 607463 | Arrhythmogenic cardiomyopathy with variable ectodermal abnormalities, 620519 (3), Autosomal recessive |
| **PPP1R21** | 99.69 % | 618159 | Neurodevelopmental disorder with hypotonia, facial dysmorphism, and brain abnormalities, 619383 (3), Autosomal recessive |
| **PPP2CA** | 99.93 % | 176915 | Houge-Janssens syndrome 3, 618354 (3), Autosomal dominant |
| **PPP2R1A** | 99.87 % | 605983 | Houge-Janssens syndrome 2, 616362 (3), Autosomal dominant |
| **PPP2R3C** | 99.62 % | 615902 | Spermatogenic failure 36, 618420 (3), Autosomal dominant; Myoectodermal gonadal dysgenesis syndrome, 618419 (3), Autosomal recessive |
| **PPP2R5D** | 99.71 % | 601646 | Houge-Janssens syndrome 1, 616355 (3), Autosomal dominant |
| **PPP3CA** | 99.66 % | 114105 | Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265 (3), Autosomal dominant; Developmental and epileptic encephalopathy 91, 617711 (3), Autosomal dominant |
| **PQBP1** | 99.91 % | 300463 | Renpenning syndrome, 309500 (3), X-linked 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 |
| **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 |
| **PRIM1** | 98.37 % | 176635 | Primordial dwarfism-immunodeficiency-lipodystrophy syndrome, 620005 (3), Autosomal recessive |
| **PRKACA** | 99.74 % | 601639 | Cushing syndrome, ACTH-independent adrenal, somatic, 615830 (3); Cardioacrofacial dysplasia 1, 619142 (3), Autosomal dominant |
| **PRKACB** | 96.04 % | 176892 | Cardioacrofacial dysplasia 2, 619143 (3), Autosomal dominant, Somatic mosaicism |
| **PRKAG2** | 96.89 % | 602743 | Glycogen storage disease of heart, lethal congenital, 261740 (3), Autosomal dominant; Wolff-Parkinson-White syndrome, 194200 (3), Autosomal dominant; Cardiomyopathy, hypertrophic 6, 600858 (3), Autosomal dominant |
| **PRKAR1A** | 96.1 % | 188830 | Pigmented nodular adrenocortical disease, primary, 1, 610489 (3), Autosomal dominant; Acrodysostosis 1, with or without hormone resistance, 101800 (3), Autosomal dominant; Adrenocortical tumor, somatic (3); Carney complex, type 1, 160980 (3), Autosomal dominant; Myxoma, intracardiac, 255960 (3), Autosomal dominant |
| **PRKCI** | 99.39 % | 600539 | *No OMIM phenotypes* |
| **PRKD1** | 99.92 % | 605435 | Congenital heart defects and ectodermal dysplasia, 617364 (3), Autosomal dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **PRKG2** | 99.81 % | 601591 | Spondylometaphyseal dysplasia, Pagnamenta type, 619638 (3), Autosomal recessive; Acromesomelic dysplasia 4, 619636 (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 |
| **PRR12** | 99.55 % | 616633 | Neuroocular syndrome, 619539 (3), Autosomal dominant |
| **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 |
| **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 |
| **PSKH1** | 99.89 % | 177015 | Cholestasis, progressive familial intrahepatic, 13, 620962 (3), Autosomal recessive |
| **PSMC3** | 99.63 % | 186852 | ?Deafness, cataract, impaired intellectual development, and polyneuropathy, 619354 (3), Autosomal recessive |
| **PSMF1** | 99.69 % | 617858 | *No OMIM phenotypes* |
| **PSPH** | 98.61 % | 172480 | Phosphoserine phosphatase deficiency, 614023 (3), Autosomal recessive |
| **PTBP1** | 99.97 % | 600693 | *No OMIM phenotypes* |
| **PTCH1** | 99.87 % | 601309 | Basal cell nevus syndrome 1, 109400 (3), Autosomal dominant; Basal cell carcinoma, somatic, 605462 (3); Holoprosencephaly 7, 610828 (3), Autosomal dominant |
| **PTDSS1** | 99.94 % | 612792 | Lenz-Majewski hyperostotic dwarfism, 151050 (3), Autosomal dominant |
| **PTEN** | 99.87 % | 601728 | {Glioma susceptibility 2}, 613028 (3), Autosomal dominant; {Meningioma}, 607174 (3), Autosomal dominant; Cowden syndrome 1, 158350 (3), Autosomal dominant; Lhermitte-Duclos disease, 158350 (3), Autosomal dominant; Prostate cancer, somatic, 176807 (3); Macrocephaly/autism syndrome, 605309 (3), Autosomal dominant |
| **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 |
| **PTHLH** | 99.47 % | 168470 | Brachydactyly, type E2, 613382 (3), Autosomal dominant |
| **PTPN11** | 99.84 % | 176876 | Noonan syndrome 1, 163950 (3), Autosomal dominant; LEOPARD syndrome 1, 151100 (3), Autosomal dominant; Metachondromatosis, 156250 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, somatic, 607785 (3) |
| **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 |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **PTS** | 99.9 % | 612719 | Hyperphenylalaninemia, BH4-deficient, A, 261640 (3), Autosomal recessive |
| **PUF60** | 99.86 % | 604819 | Verheij syndrome, 615583 (3), Autosomal dominant |
| **PUM1** | 99.17 % | 607204 | Neurodevelopmental disorder with motor abnormalities, and facial dysmorphism, 620719 (3), Autosomal dominant |
| **PURA** | 99.99 % | 600473 | Neurodevelopmental disorder with neonatal respiratory insufficiency, hypotonia, and feeding difficulties, 616158 (3), Autosomal dominant |
| **PUS3** | 99.96 % | 616283 | Neurodevelopmental disorder with microcephaly and gray sclerae, 617051 (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 |
| **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 |
| **QRICH1** | 99.96 % | 617387 | Ververi-Brady syndrome, 617982 (3), Autosomal dominant |
| **QRSL1** | 99.87 % | 617209 | Combined oxidative phosphorylation deficiency 40, 618835 (3), Autosomal recessive |
| **RAB11A** | 99.95 % | 605570 | *No OMIM phenotypes* |
| **RAB11B** | 99.81 % | 604198 | Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807 (3), Autosomal dominant |
| **RAB18** | 99.77 % | 602207 | Warburg micro syndrome 3, 614222 (3), Autosomal recessive |
| **RAB23** | 99.9 % | 606144 | Carpenter syndrome, 201000 (3), Autosomal recessive |
| **RAB33B** | 99.98 % | 605950 | Smith-McCort dysplasia 2, 615222 (3), Autosomal recessive |
| **RAB34** | 99.89 % | 610917 | Orofaciodigital syndrome XX, 620718 (3), Autosomal 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 |
| **RAC1** | 99.78 % | 602048 | Intellectual developmental disorder, autosomal dominant 48, 617751 (3), Autosomal dominant |
| **RAC3** | 99.98 % | 602050 | Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577 (3), Autosomal dominant |
| **RAD21** | 99.87 % | 606462 | Cornelia de Lange syndrome 4, 614701 (3), Autosomal dominant; ?Mungan syndrome, 611376 (3), Autosomal recessive |
| **RAD50** | 99.81 % | 604040 | Nijmegen breakage syndrome-like disorder, 613078 (3), Autosomal recessive |
| **RAD51** | 97.94 % | 179617 | Mirror movements 2, 614508 (3), Autosomal dominant; {Breast cancer, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant; Fanconi anemia, complementation group R, 617244 (3), Autosomal dominant |
| **RAD51C** | 98.2 % | 602774 | {Breast-ovarian cancer, familial, susceptibility to, 3}, 613399 (3); Fanconi anemia, complementation group O, 613390 (3), Autosomal recessive |
| **RAF1** | 99.91 % | 164760 | Cardiomyopathy, dilated, 1NN, 615916 (3), Autosomal dominant; Noonan syndrome 5, 611553 (3), Autosomal dominant; LEOPARD syndrome 2, 611554 (3), Autosomal dominant |
| **RAI1** | 99.92 % | 607642 | Smith-Magenis syndrome, 182290 (3), Autosomal dominant, Isolated cases |
| **RALA** | 99.18 % | 179550 | Hiatt-Neu-Cooper neurodevelopmental syndrome, 619311 (3), Autosomal dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **RALGAPA1** | 99.75 % | 608884 | Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation, 618797 (3), Autosomal recessive |
| **RAP1B** | 98.82 % | 179530 | Thrombocytopenia 11 with multiple congenital anomalies and dysmorphic facies, 620654 (3), Autosomal dominant |
| **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 |
| **RARS2** | 99.8 % | 611524 | Pontocerebellar hypoplasia, type 6, 611523 (3), Autosomal recessive |
| **RASA1** | 99.23 % | 139150 | Capillary malformation-arteriovenous malformation 1, 608354 (3), Autosomal dominant; Basal cell carcinoma, somatic, 605462 (3) |
| **RASA2** | 99.71 % | 601589 | *No OMIM phenotypes* |
| **RAX** | 99.27 % | 601881 | Microphthalmia, syndromic 16, 611038 (3), Autosomal recessive |
| **RBBP5** | 99.51 % | 600697 | *No OMIM phenotypes* |
| **RBBP8** | 99.93 % | 604124 | Seckel syndrome 2, 606744 (3), Autosomal recessive; Jawad syndrome, 251255 (3), Autosomal recessive; Pancreatic carcinoma, somatic (3) |
| **RBFOX2** | 99.94 % | 612149 | *No OMIM phenotypes* |
| **RBM10** | 99.72 % | 300080 | TARP syndrome, 311900 (3), X-linked recessive |
| **RBM8A** | 99.16 % | 605313 | Thrombocytopenia-absent radius syndrome, 274000 (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 |
| **RBPJ** | 99.83 % | 147183 | Adams-Oliver syndrome 3, 614814 (3), Autosomal dominant |
| **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 |
| **RELN** | 99.85 % | 600514 | {Epilepsy, familial temporal lobe, 7}, 616436 (3), Autosomal dominant; Lissencephaly 2 (Norman-Roberts type), 257320 (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 |
| **RERE** | 99.62 % | 605226 | Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975 (3), Autosomal dominant |
| **RET** | 99.85 % | 164761 | {Hirschsprung disease, susceptibility to, 1}, 142623 (3), Autosomal dominant; Multiple endocrine neoplasia IIA, 171400 (3), Autosomal dominant; {Hirschsprung disease, protection against}, 142623 (3), Autosomal dominant; Medullary thyroid carcinoma, 155240 (3), Autosomal dominant; Pheochromocytoma, 171300 (3), Autosomal dominant; Multiple endocrine neoplasia IIB, 162300 (3), Autosomal dominant |
| **RFT1** | 98.55 % | 611908 | Congenital disorder of glycosylation, type In, 612015 (3), Autosomal recessive |
| **RFWD3** | 99.75 % | 614151 | ?Fanconi anemia, complementation group W, 617784 (3), Autosomal recessive |
| **RFX6** | 99.63 % | 612659 | Mitchell-Riley syndrome, 615710 (3), Autosomal recessive |
| **RHOA** | 97.54 % | 165390 | Ectodermal dysplasia with facial dysmorphism and acral, ocular, and brain anomalies, somatic mosaic, 618727 (3) |
| **RHOBTB2** | 99.95 % | 607352 | Developmental and epileptic encephalopathy 64, 618004 (3), Autosomal dominant |
| **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 |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **RIPK4** | 99.68 % | 605706 | CHAND syndrome, 214350 (3), Autosomal recessive; Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650 (3), Autosomal recessive |
| **RIPPLY2** | 99.74 % | 609891 | ?Spondylocostal dysostosis 6, 616566 (3), Autosomal recessive |
| **RIT1** | 99.11 % | 609591 | Noonan syndrome 8, 615355 (3), Autosomal dominant |
| **RLIM** | 99.77 % | 300379 | Tonne-Kalscheuer syndrome, 300978 (3), X-linked |
| **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 |
| **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 |
| **RNF125** | 99.91 % | 610432 | Tenorio syndrome, 616260 (3), Autosomal dominant |
| **RNF13** | 99.73 % | 609247 | Developmental and epileptic encephalopathy 73, 618379 (3), Autosomal dominant |
| **RNF2** | 97.16 % | 608985 | Luo-Schoch-Yamamoto syndrome, 619460 (3), Autosomal dominant |
| **RNPC3** | 95.2 % | 618016 | Pituitary hormone deficiency, combined or isolated, 7, 618160 (3), Autosomal recessive |
| **RNU12** | 0 % | 620204 | CDAGS syndrome, 603116 (3), Autosomal recessive; ?Spinocerebellar ataxia, autosomal recessive 33, 620208 (3), Autosomal recessive |
| **RNU2-2P** | 1.36 % | 621238 | Developmental and epileptic encephalopathy 119, 621304 (3), Autosomal dominant |
| **RNU4-2** | 0 % | 620823 | ReNU syndrome, 620851 (3), Autosomal dominant |
| **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 |
| **RNU5A-1** | 0 % | 180691 | *No OMIM phenotypes* |
| **RNU5B-1** | 0 % | 621090 | Neurodevelopmental disorder with seizures and joint laxity, 621302 (3), Autosomal dominant |
| **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 |
| **ROR2** | 99.79 % | 602337 | Brachydactyly, type B1, 113000 (3), Autosomal dominant; Robinow syndrome, autosomal recessive, 268310 (3), Autosomal recessive |
| **RORA** | 99.85 % | 600825 | Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060 (3), Autosomal dominant |
| **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 |
| **RPL10** | 99.72 % | 312173 | {Autism, susceptibility to, X-linked 5}, 300847 (3); Intellectual developmental disorder, X-linked syndromic 35, 300998 (3), X-linked recessive |
| **RPL11** | 98.91 % | 604175 | Diamond-Blackfan anemia 7, 612562 (3), Autosomal dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **RPL13** | 99.89 % | 113703 | Spondyloepimetaphyseal dysplasia, Isidor-Toutain type, 618728 (3), Autosomal dominant |
| **RPL15** | 99.92 % | 604174 | Diamond-Blackfan anemia 12, 615550 (3), Autosomal dominant |
| **RPL26** | 99.94 % | 603704 | ?Diamond-Blackfan anemia 11, 614900 (3), Autosomal dominant |
| **RPL35A** | 99.98 % | 180468 | Diamond-Blackfan anemia 5, 612528 (3), Autosomal dominant |
| **RPL5** | 99.93 % | 603634 | Diamond-Blackfan anemia 6, 612561 (3), Autosomal dominant |
| **RPS10** | 99.88 % | 603632 | Diamond-Blackfan anemia 9, 613308 (3), Autosomal dominant |
| **RPS17** | 99.96 % | 180472 | Diamond-Blackfan anemia 4, 612527 (3), Autosomal dominant |
| **RPS19** | 99.99 % | 603474 | Diamond-Blackfan anemia 1, 105650 (3), Autosomal dominant |
| **RPS23** | 99.99 % | 603683 | Brachycephaly, trichomegaly, and developmental delay, 617412 (3), Autosomal dominant |
| **RPS24** | 99.95 % | 602412 | Diamond-blackfan anemia 3, 610629 (3), Autosomal dominant |
| **RPS26** | 100 % | 603701 | Diamond-Blackfan anemia 10, 613309 (3), Autosomal dominant |
| **RPS6KA3** | 99.42 % | 300075 | Intellectual developmental disorder, X-linked 19, 300844 (3), X-linked dominant; Coffin-Lowry syndrome, 303600 (3), X-linked dominant |
| **RPS7** | 99.94 % | 603658 | Diamond-Blackfan anemia 8, 612563 (3), Autosomal dominant |
| **RRAGC** | 97.5 % | 608267 | Long-Olsen-Distelmaier syndrome, 620609 (3), Autosomal dominant |
| **RRAS** | 99.69 % | 165090 | *No OMIM phenotypes* |
| **RRAS2** | 99.71 % | 600098 | Ovarian carcinoma (3); Noonan syndrome 12, 618624 (3), Autosomal dominant |
| **RREB1** | 99.89 % | 602209 | *No OMIM phenotypes* |
| **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 |
| **RSPH4A** | 99.84 % | 612647 | Ciliary dyskinesia, primary, 11, 612649 (3), Autosomal recessive |
| **RSPH9** | 99.64 % | 612648 | Ciliary dyskinesia, primary, 12, 612650 (3), Autosomal recessive |
| **RSPO2** | 99.8 % | 610575 | ?Humerofemoral hypoplasia with radiotibial ray deficiency, 618022 (3), Autosomal recessive; Tetraamelia syndrome 2, 618021 (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 |
| **RTTN** | 99.84 % | 610436 | Microcephaly, short stature, and polymicrogyria with seizures, 614833 (3), Autosomal recessive |
| **RUNX2** | 99.9 % | 600211 | Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510 (3), Autosomal dominant; Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 (3), Autosomal dominant; Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600 (3), Autosomal dominant; Cleidocranial dysplasia, 119600 (3), Autosomal dominant |
| **RXYLT1** | 99.19 % | 605862 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **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 |
| **SACS** | 99.95 % | 604490 | Spastic ataxia, Charlevoix-Saguenay type, 270550 (3), Autosomal recessive |
| **SALL1** | 99.96 % | 602218 | Townes-Brocks syndrome 1, 107480 (3), Autosomal dominant; Townes-Brocks branchiootorenal-like syndrome, 107480 (3), Autosomal dominant |
| **SALL4** | 99.99 % | 607343 | ?IVIC syndrome, 147750 (3), Autosomal dominant; Duane-radial ray syndrome, 607323 (3), Autosomal dominant |
| **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 |
| **SART3** | 99.87 % | 611684 | *No OMIM phenotypes* |
| **SASS6** | 96 % | 609321 | Microcephaly 14, primary, autosomal recessive, 616402 (3), Autosomal recessive |
| **SATB1** | 99.93 % | 602075 | den Hoed-de Boer-Voisin syndrome, 619229 (3), Autosomal dominant; Developmental delay with dysmorphic facies and dental anomalies, 619228 (3), Autosomal dominant |
| **SATB2** | 99.66 % | 608148 | Glass syndrome, 612313 (3), Autosomal dominant |
| **SBDS** | 99.32 % | 607444 | {Aplastic anemia, susceptibility to}, 609135 (3); Shwachman-Diamond syndrome 1, 260400 (3), Autosomal recessive |
| **SC5D** | 99.82 % | 602286 | Lathosterolosis, 607330 (3), Autosomal recessive |
| **SCAF4** | 99.82 % | 616023 | Fliedner-Zweier syndrome, 620511 (3), Autosomal dominant |
| **SCARF2** | 98.63 % | 613619 | Van den Ende-Gupta syndrome, 600920 (3), Autosomal recessive |
| **SCLT1** | 98.43 % | 611399 | *No OMIM phenotypes* |
| **SCN1A** | 99.86 % | 182389 | Developmental and epileptic encephalopathy 6B, non-Dravet, 619317 (3), Autosomal dominant; Migraine, familial hemiplegic, 3, 609634 (3), Autosomal dominant; Dravet syndrome, 607208 (3), Autosomal dominant; Febrile seizures, familial, 3A, 604403 (3), Autosomal dominant; Generalized epilepsy with febrile seizures plus, type 2, 604403 (3), Autosomal dominant |
| **SCN2A** | 99.74 % | 182390 | Seizures, benign familial infantile, 3, 607745 (3), Autosomal dominant; Developmental and epileptic encephalopathy 11, 613721 (3), Autosomal dominant; Episodic ataxia, type 9, 618924 (3), Autosomal dominant |
| **SCN3A** | 99.67 % | 182391 | Epilepsy, familial focal, with variable foci 4, 617935 (3), Autosomal dominant; Developmental and epileptic encephalopathy 62, 617938 (3), Autosomal dominant |
| **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 |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **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 |
| **SCNM1** | 98.8 % | 608095 | Orofaciodigital syndrome XIX, 620107 (3), Autosomal recessive |
| **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 |
| **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 |
| **SDR9C7** | 99.91 % | 609769 | Ichthyosis, congenital, autosomal recessive 13, 617574 (3), Autosomal recessive |
| **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 |
| **SECISBP2** | 99.91 % | 607693 | Thyroid hormone metabolism, abnormal, 1, 609698 (3), Autosomal recessive |
| **SEL1L** | 99.94 % | 602329 | Neurodevelopmental disorder with hypotonia, poor growth, dysmorphic facies, and agammaglobulinemia, 621068 (3), Autosomal recessive; ?Neurodevelopmental disorder with poor growth, absent speech, progressive ataxia, and dysmorphic facies, 621067 (3), Autosomal recessive |
| **SELENON** | 99.12 % | 606210 | Congenital myopathy 3 with rigid spine, 602771 (3), Autosomal recessive |
| **SEMA3A** | 99 % | 603961 | {Hypogonadotropic hypogonadism 16 with or without anosmia}, 614897 (3), Autosomal dominant |
| **SENP7** | 99.36 % | 612846 | *No OMIM phenotypes* |
| **SEPHS1** | 99.84 % | 600902 | Ververi-Brady syndrome 2, 621325 (3), Autosomal dominant |
| **SEPSECS** | 99.43 % | 613009 | Pontocerebellar hypoplasia type 2D, 613811 (3), Autosomal recessive |
| **SERPINF1** | 99.91 % | 172860 | Osteogenesis imperfecta, type VI, 613982 (3), Autosomal recessive |
| **SERPINH1** | 99.67 % | 600943 | {Preterm premature rupture of the membranes, susceptibility to}, 610504 (3), Multifactorial; Osteogenesis imperfecta, type X, 613848 (3), Autosomal recessive |
| **SET** | 99.92 % | 600960 | Intellectual developmental disorder, autosomal dominant 58, 618106 (3), Autosomal dominant |
| **SETBP1** | 99.98 % | 611060 | Schinzel-Giedion midface retraction syndrome, 269150 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 29, 616078 (3), Autosomal dominant |
| **SETD1A** | 99.78 % | 611052 | Epilepsy, early-onset, 2, with or without developmental delay, 618832 (3), Autosomal dominant; Neurodevelopmental disorder with speech impairment and dysmorphic facies, 619056 (3), Autosomal dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **SETD2** | 99.71 % | 612778 | Luscan-Lumish syndrome, 616831 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 70, 620157 (3), Autosomal dominant; Rabin-Pappas syndrome, 620155 (3), Autosomal dominant |
| **SETD5** | 99.92 % | 615743 | Intellectual developmental disorder, autosomal dominant 23, 615761 (3), Autosomal dominant |
| **SF3B2** | 99.75 % | 605591 | Craniofacial microsomia, 164210 (3), Autosomal dominant |
| **SF3B4** | 99.47 % | 605593 | Acrofacial dysostosis 1, Nager type, 154400 (3), Autosomal dominant |
| **SFXN4** | 99.94 % | 615564 | Combined oxidative phosphorylation deficiency 18, 615578 (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 |
| **SH3BP2** | 99.54 % | 602104 | Cherubism, 118400 (3), Autosomal dominant |
| **SH3PXD2B** | 99.96 % | 613293 | Frank-ter Haar syndrome, 249420 (3), Autosomal recessive |
| **SHANK1** | 99.83 % | 604999 | *No OMIM phenotypes* |
| **SHANK2** | 99.67 % | 603290 | {Autism susceptibility 17}, 613436 (3) |
| **SHANK3** | 99.48 % | 606230 | Phelan-McDermid syndrome, 606232 (3), Autosomal dominant; {Schizophrenia 15}, 613950 (3), Autosomal dominant |
| **SHH** | 99.93 % | 600725 | Single median maxillary central incisor, 147250 (3), Autosomal dominant; Holoprosencephaly 3, 142945 (3), Autosomal dominant; Microphthalmia/coloboma 5, 611638 (3), Autosomal dominant |
| **SHMT2** | 99.41 % | 138450 | Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121 (3), Autosomal recessive |
| **SHOC2** | 99.83 % | 602775 | Noonan syndrome-like with loose anagen hair 1, 607721 (3), Autosomal dominant |
| **SHOX** | 95.24 % | 312865 | Short stature, idiopathic familial, 300582 (3); Leri-Weill dyschondrosteosis, 127300 (3), Pseudoautosomal dominant; Langer mesomelic dysplasia, 249700 (3), Pseudoautosomal recessive |
| **SHROOM3** | 99.89 % | 604570 | *No OMIM phenotypes* |
| **SHROOM4** | 99.83 % | 300579 | *No OMIM phenotypes* |
| **SIAH1** | 99.97 % | 602212 | Buratti-Harel syndrome, 619314 (3), Autosomal dominant |
| **SIK3** | 99.88 % | 614776 | ?Spondyloepimetaphyseal dysplasia, Krakow type, 618162 (3), Autosomal recessive |
| **SIL1** | 99.79 % | 608005 | Marinesco-Sjogren syndrome, 248800 (3), Autosomal recessive |
| **SIN3A** | 99.82 % | 607776 | Witteveen-Kolk syndrome, 613406 (3), Autosomal dominant |
| **SIRT6** | 99.4 % | 606211 | *No OMIM phenotypes* |
| **SIX2** | 99.96 % | 604994 | *No OMIM phenotypes* |
| **SIX3** | 99.84 % | 603714 | Schizencephaly, 269160 (3); Holoprosencephaly 2, 157170 (3), Autosomal dominant |
| **SIX6** | 99.99 % | 606326 | Optic disc anomalies with retinal and/or macular dystrophy, 212550 (3), Autosomal recessive |
| **SKI** | 98.92 % | 164780 | Shprintzen-Goldberg syndrome, 182212 (3), Autosomal dominant |
| **SKIV2L** | 99.43 % | 600478 | Trichohepatoenteric syndrome 2, 614602 (3), Autosomal recessive |
| **SKOR2** | 99.97 % | 617138 | Valence-Farazi cerebellar ataxia syndrome, 621386 (3), Autosomal recessive |
| **SLC10A7** | 99.96 % | 611459 | Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363 (3), Autosomal recessive |
| **SLC12A1** | 99.8 % | 600839 | Bartter syndrome, type 1, 601678 (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 |
| **SLC12A9** | 99.48 % | 616861 | *No OMIM phenotypes* |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **SLC13A1** | 99.89 % | 606193 | *No OMIM phenotypes* |
| **SLC13A5** | 99.92 % | 608305 | Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905 (3), Autosomal recessive |
| **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 |
| **SLC18A3** | 99.5 % | 600336 | Myasthenic syndrome, congenital, 21, presynaptic, 617239 (3), Autosomal recessive |
| **SLC19A1** | 99.76 % | 600424 | Immunodeficiency 114, folate-responsive, 620603 (3), Autosomal recessive; ?Megaloblastic anemia, folate-responsive, 601775 (3), Autosomal recessive |
| **SLC1A2** | 99.38 % | 600300 | Developmental and epileptic encephalopathy 41, 617105 (3), Autosomal dominant |
| **SLC20A1** | 99.73 % | 137570 | *No OMIM phenotypes* |
| **SLC22A5** | 99.98 % | 603377 | Carnitine deficiency, systemic primary, 212140 (3), Autosomal recessive |
| **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 |
| **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 |
| **SLC25A24** | 96.63 % | 608744 | Fontaine progeroid syndrome, 612289 (3), Autosomal dominant |
| **SLC25A26** | 99.82 % | 611037 | Combined oxidative phosphorylation deficiency 28, 616794 (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 |
| **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 |
| **SLC27A4** | 99.85 % | 604194 | Ichthyosis prematurity syndrome, 608649 (3), Autosomal recessive |
| **SLC29A3** | 99.61 % | 612373 | Histiocytosis-lymphadenopathy plus syndrome, 602782 (3), Autosomal recessive |
| **SLC2A10** | 99.96 % | 606145 | Arterial tortuosity syndrome, 208050 (3), Autosomal recessive |
| **SLC30A5** | 99.75 % | 607819 | *No OMIM phenotypes* |
| **SLC30A7** | 96.67 % | 611149 | Ziegler-Huang syndrome, 620501 (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 |
| **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 |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **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 |
| **SLC35C1** | 99.95 % | 605881 | Congenital disorder of glycosylation, type IIc, 266265 (3), Autosomal recessive |
| **SLC35D1** | 97.02 % | 610804 | Schneckenbecken dysplasia, 269250 (3), Autosomal recessive |
| **SLC39A8** | 99.75 % | 608732 | Congenital disorder of glycosylation, type IIn, 616721 (3), Autosomal recessive |
| **SLC45A1** | 99.78 % | 605763 | Intellectual developmental disorder with neuropsychiatric features, 617532 (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) |
| **SLC4A10** | 99.21 % | 605556 | Neurodevelopmental disorder with hypotonia and characteristic brain abnormalities, 620746 (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 |
| **SLC6A9** | 99.57 % | 601019 | Glycine encephalopathy with normal serum glycine, 617301 (3), Autosomal recessive |
| **SLF2** | 99.78 % | 610348 | Atelis syndrome 1, 620184 (3), Autosomal recessive |
| **SLX4** | 99.88 % | 613278 | Fanconi anemia, complementation group P, 613951 (3), Autosomal recessive |
| **SMAD2** | 99.89 % | 601366 | Loeys-Dietz syndrome 6, 619656 (3), Autosomal dominant; Congenital heart defects, multiple types, 8, with or without heterotaxy, 619657 (3), Autosomal dominant |
| **SMAD3** | 99.95 % | 603109 | Loeys-Dietz syndrome 3, 613795 (3), Autosomal dominant |
| **SMAD4** | 99.95 % | 600993 | Pancreatic cancer, somatic, 260350 (3); Myhre syndrome, 139210 (3), Autosomal dominant; Polyposis, juvenile intestinal, 174900 (3), Autosomal dominant; Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 (3), Autosomal dominant |
| **SMAD5** | 99.74 % | 603110 | *No OMIM phenotypes* |
| **SMARCA2** | 99.82 % | 600014 | Nicolaides-Baraitser syndrome, 601358 (3), Autosomal dominant; Blepharophimosis-impaired intellectual development syndrome, 619293 (3), Autosomal dominant |
| **SMARCA4** | 99.81 % | 603254 | Coffin-Siris syndrome 4, 614609 (3), Autosomal dominant; {Rhabdoid tumor predisposition syndrome 2}, 613325 (3), Autosomal dominant; ?Otosclerosis 12, 620792 (3), Autosomal dominant |
| **SMARCB1** | 99.8 % | 601607 | Rhabdoid tumors, somatic, 609322 (3); {Schwannomatosis-1, susceptibility to}, 162091 (3), Autosomal dominant; Coffin-Siris syndrome 3, 614608 (3), Autosomal dominant; {Rhabdoid tumor predisposition syndrome 1}, 609322 (3), Autosomal dominant |
| **SMARCC1** | 99.53 % | 601732 | {Hydrocephalus, congenital, 5, susceptibility to}, 620241 (3), Autosomal dominant |
| **SMARCC2** | 99.06 % | 601734 | Coffin-Siris syndrome 8, 618362 (3), Autosomal dominant |
| **SMARCD1** | 99.36 % | 601735 | Coffin-Siris syndrome 11, 618779 (3), Autosomal dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **SMARCE1** | 99.26 % | 603111 | {Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant; Coffin-Siris syndrome 5, 616938 (3), Autosomal dominant |
| **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 |
| **SMC3** | 99.92 % | 606062 | Cornelia de Lange syndrome 3, 610759 (3), Autosomal dominant |
| **SMC5** | 99.38 % | 609386 | Atelis syndrome 2, 620185 (3), Autosomal recessive |
| **SMCHD1** | 99.84 % | 614982 | Facioscapulohumeral muscular dystrophy 2, digenic, 158901 (3), Digenic dominant; Bosma arhinia microphthalmia syndrome, 603457 (3), Autosomal dominant |
| **SMG8** | 99.89 % | 613175 | Alzahrani-Kuwahara syndrome, 619268 (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 |
| **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 |
| **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 |
| **SNAP25** | 99.12 % | 600322 | Developmental and epileptic encephalopathy 117, 616330 (3), Autosomal dominant |
| **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 |
| **SNAPIN** | 97.08 % | 607007 | Neurodevelopmental disorder with structural brain abnormalities and craniofacial abnormalities, 621393 (3), Autosomal recessive |
| **SNF8** | 98.63 % | 610904 | Developmental and epileptic encephalopathy 115, 620783 (3), Autosomal recessive; Neurodevelopmental disorder plus optic atrophy, 620784 (3), Autosomal recessive |
| **SNORD118** | 99.84 % | 616663 | Leukoencephalopathy, brain calcifications, and cysts, 614561 (3), Autosomal recessive |
| **SNRPB** | 99 % | 182282 | Cerebrocostomandibular syndrome, 117650 (3), Autosomal dominant |
| **SNUPN** | 99.9 % | 607902 | Muscular dystrophy, limb-girdle, autosomal recessive 29, 620793 (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 |
| **SON** | 99.97 % | 182465 | ZTTK syndrome, 617140 (3), Autosomal dominant |
| **SOS1** | 99.57 % | 182530 | Noonan syndrome 4, 610733 (3), Autosomal dominant; Fibromatosis, gingival, 1, 135300 (3), Autosomal dominant |
| **SOS2** | 99.81 % | 601247 | Noonan syndrome 9, 616559 (3), Autosomal dominant |
| **SOST** | 98.76 % | 605740 | Sclerosteosis 1, 269500 (3), Autosomal recessive; Craniodiaphyseal dysplasia, autosomal dominant, 122860 (3), Autosomal dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **SOX10** | 99.99 % | 602229 | Waardenburg syndrome, type 4C, 613266 (3), Autosomal dominant; PCWH syndrome, 609136 (3), Autosomal dominant; Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 (3), Autosomal dominant |
| **SOX11** | 99.95 % | 600898 | Intellectual developmental disorder with microcephaly and with or without ocular malformations or hypogonadotropic hypogonadism, 615866 (3), Autosomal dominant |
| **SOX17** | 99.97 % | 610928 | Vesicoureteral reflux 3, 613674 (3), Autosomal dominant; Pulmonary hypertension, primary, 7, 621248 (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 |
| **SOX2** | 99.98 % | 184429 | Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 (3), Autosomal dominant; Microphthalmia, syndromic 3, 206900 (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 |
| **SOX4** | 99.14 % | 184430 | Intellectual developmental disorder with speech delay and dysmorphic facies, 618506 (3), Autosomal dominant |
| **SOX5** | 99.56 % | 604975 | Lamb-Shaffer syndrome, 616803 (3), Autosomal dominant |
| **SOX6** | 99.84 % | 607257 | Tolchin-Le Caignec syndrome, 618971 (3), Autosomal dominant |
| **SOX9** | 99.94 % | 608160 | Campomelic dysplasia with autosomal sex reversal, 114290 (3), Autosomal dominant; 46XY sex reversal 10, 616425 (3), Autosomal dominant; Acampomelic campomelic dysplasia, 114290 (3), Autosomal dominant; Campomelic dysplasia, 114290 (3), Autosomal dominant; 46XX sex reversal 2, 278850 (3), Autosomal dominant |
| **SP7** | 100 % | 606633 | Osteogenesis imperfecta, type XII, 613849 (3), Autosomal recessive |
| **SPAG1** | 99.63 % | 603395 | Ciliary dyskinesia, primary, 28, 615505 (3), Autosomal recessive |
| **SPARC** | 98.85 % | 182120 | Osteogenesis imperfecta, type XVII, 616507 (3), Autosomal recessive |
| **SPATA5** | 99.32 % | 613940 | Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577 (3), Autosomal recessive |
| **SPECC1L** | 99.96 % | 614140 | Teebi hypertelorism syndrome 1, 145420 (3), Autosomal dominant; ?Facial clefting, oblique, 1, 600251 (3), Autosomal dominant |
| **SPEG** | 99.75 % | 615950 | Centronuclear myopathy 5, 615959 (3), Autosomal recessive |
| **SPEN** | 99.95 % | 613484 | Radio-Tartaglia syndrome, 619312 (3), Autosomal dominant |
| **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 |
| **SPIN4** | 99.93 % | 301113 | ?Lui-Jee-Baron syndrome, 301114 (3), X-linked |
| **SPINT2** | 99.84 % | 605124 | Diarrhea 3, secretory sodium, congenital, syndromic, 270420 (3), Autosomal recessive |
| **SPOP** | 99.61 % | 602650 | Nabais Sa-de Vries syndrome, type 1, 618828 (3), Autosomal dominant; Nabais Sa-de Vries syndrome, type 2, 618829 (3), Autosomal dominant |
| **SPOUT1** | 99.91 % | 617614 | Neurodevelopmental disorder with poor growth, seizures, and brain abnormalities, 621154 (3), Autosomal recessive |
| **SPRED1** | 99.97 % | 609291 | Legius syndrome, 611431 (3), Autosomal dominant |
| **SPRED2** | 99.86 % | 609292 | Noonan syndrome 14, 619745 (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 |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **SPTAN1** | 99.76 % | 182810 | Developmental delay with or without epilepsy, 620540 (3), Autosomal dominant; Developmental and epileptic encephalopathy 5, 613477 (3), Autosomal dominant; Spastic paraplegia 91, autosomal dominant, with or without cerebellar ataxia, 620538 (3), Autosomal dominant; Neuronopathy, distal hereditary motor, autosomal dominant 11, 620528 (3), Autosomal dominant |
| **SPTB** | 99.94 % | 182870 | Anemia, neonatal hemolytic, fatal or near-fatal, 617948 (3), Autosomal recessive, Autosomal dominant; Elliptocytosis-3, 617948 (3), Autosomal recessive, Autosomal dominant; Spherocytosis, type 2, 616649 (3), Autosomal dominant |
| **SPTBN1** | 99.8 % | 182790 | Developmental delay, impaired speech, and behavioral abnormalities, 619475 (3), Autosomal dominant |
| **SRCAP** | 99.82 % | 611421 | Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities, 619595 (3), Autosomal dominant; Floating-Harbor syndrome, 136140 (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 |
| **SRP54** | 99.84 % | 604857 | Neutropenia, severe congenital, 8, autosomal dominant, 618752 (3), Autosomal dominant |
| **SRPK3** | 99.83 % | 301002 | Intellectual developmental disorder, X-linked 114, 301134 (3), X-linked |
| **SRY** | 50.56 % | 480000 | 46XY sex reversal 1, 400044 (3), Y-linked; 46XX sex reversal 1, 400045 (4), X-linked dominant |
| **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 |
| **STAT3** | 99.7 % | 102582 | Hyper-IgE syndrome 1, autosomal dominant, with recurrent infections, 147060 (3), Autosomal dominant; Autoimmune disease, multisystem, infantile-onset, 1, 615952 (3), Autosomal dominant |
| **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 |
| **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 |
| **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 |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **STX11** | 99.93 % | 605014 | Hemophagocytic lymphohistiocytosis, familial, 4, 603552 (3), Autosomal recessive |
| **STX1B** | 99.81 % | 601485 | Generalized epilepsy with febrile seizures plus, type 9, 616172 (3), Autosomal dominant |
| **STX5** | 99.72 % | 603189 | ?Congenital disorder of glycosylation, type IIaa, 620454 (3), Autosomal recessive |
| **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 |
| **SUFU** | 99.9 % | 607035 | {Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant; Joubert syndrome 32, 617757 (3), Autosomal recessive; Basal cell nevus syndrome 2, 620343 (3); {Medulloblastoma}, 155255 (3), Somatic mutation, Autosomal recessive, Autosomal dominant |
| **SULT2B1** | 99.62 % | 604125 | Ichthyosis, congenital, autosomal recessive 14, 617571 (3), Autosomal recessive |
| **SUMF1** | 99.88 % | 607939 | Multiple sulfatase deficiency, 272200 (3), Autosomal recessive |
| **SUZ12** | 98.21 % | 606245 | Imagawa-Matsumoto syndrome, 618786 (3), Autosomal dominant |
| **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 |
| **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 |
| **TAB2** | 99.63 % | 605101 | Congenital heart defects, nonsyndromic, 2, 614980 (3), Autosomal dominant |
| **TACO1** | 99.34 % | 612958 | Mitochondrial complex IV deficiency, nuclear type 8, 619052 (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 |
| **TAF13** | 95.05 % | 600774 | Intellectual developmental disorder, autosomal recessive 60, 617432 (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 |
| **TAPT1** | 99.68 % | 612758 | Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897 (3), Autosomal recessive |
| **TASP1** | 99.82 % | 608270 | Suleiman-El-Hattab syndrome, 618950 (3), Autosomal recessive |
| **TBC1D1** | 99.86 % | 609850 | *No OMIM phenotypes* |
| **TBC1D20** | 99.94 % | 611663 | Warburg micro syndrome 4, 615663 (3), Autosomal recessive |
| **TBC1D23** | 99.15 % | 617687 | Pontocerebellar hypoplasia, type 11, 617695 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **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 |
| **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 |
| **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 |
| **TBL1XR1** | 99.93 % | 608628 | Intellectual developmental disorder, autosomal dominant 41, 616944 (3), Autosomal dominant; Pierpont syndrome, 602342 (3), Autosomal dominant |
| **TBR1** | 99.68 % | 604616 | Intellectual developmental disorder with autism and speech delay, 606053 (3), Autosomal dominant |
| **TBX1** | 98.71 % | 602054 | Tetralogy of Fallot, 187500 (3), Autosomal dominant; DiGeorge syndrome, 188400 (3), Autosomal dominant; Conotruncal anomaly face syndrome, 217095 (3); Velocardiofacial syndrome, 192430 (3), Autosomal dominant |
| **TBX15** | 98.52 % | 604127 | Cousin syndrome, 260660 (3), Autosomal recessive |
| **TBX18** | 99.76 % | 604613 | Congenital anomalies of kidney and urinary tract 2, 143400 (3), Autosomal dominant |
| **TBX20** | 99.59 % | 606061 | Atrial septal defect 4, 611363 (3) |
| **TBX22** | 99.83 % | 300307 | Cleft palate with ankyloglossia, 303400 (3), X-linked; ?Abruzzo-Erickson syndrome, 302905 (3), X-linked |
| **TBX3** | 99.88 % | 601621 | Ulnar-mammary syndrome, 181450 (3), Autosomal dominant |
| **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 |
| **TBX5** | 99.92 % | 601620 | Holt-Oram syndrome, 142900 (3), Autosomal dominant |
| **TBX6** | 99.57 % | 602427 | Spondylocostal dysostosis 5, 122600 (3), Autosomal recessive, Autosomal dominant |
| **TCF12** | 99.93 % | 600480 | Craniosynostosis 3, 615314 (3), Autosomal dominant; Hypogonadotropic hypogonadism 26 with or without anosmia, 619718 (3), Autosomal recessive, Autosomal dominant |
| **TCF20** | 99.99 % | 603107 | Developmental delay with variable intellectual impairment and behavioral abnormalities, 618430 (3), Autosomal dominant |
| **TCF4** | 98.39 % | 602272 | Pitt-Hopkins syndrome, 610954 (3), Autosomal dominant; Corneal dystrophy, Fuchs endothelial, 3, 613267 (3), Autosomal dominant |
| **TCIRG1** | 99.78 % | 604592 | Osteopetrosis, autosomal recessive 1, 259700 (3), Autosomal recessive |
| **TCOF1** | 99.76 % | 606847 | Treacher Collins syndrome 1, 154500 (3), Autosomal dominant |
| **TCP1** | 99.9 % | 186980 | Intellectual developmental disorder with polymicrogyria and seizures, 621021 (3), Autosomal dominant |
| **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 |
| **TECPR2** | 99.84 % | 615000 | Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031 (3), Autosomal recessive |
| **TEDC1** | 99.9 % | | *No OMIM phenotypes* |
| **TEK** | 99.7 % | 600221 | Venous malformations, multiple cutaneous and mucosal, 600195 (3), Autosomal dominant; Glaucoma 3, primary congenital, E, 617272 (3), Autosomal dominant |
| **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 |
| **TFAP2A** | 99.92 % | 107580 | Branchiooculofacial syndrome, 113620 (3), Autosomal dominant |
| **TFAP2B** | 99.8 % | 601601 | Patent ductus arteriosus 2, 617035 (3), Autosomal dominant; Char syndrome, 169100 (3), Autosomal dominant |
| **TGDS** | 99.96 % | 616146 | Catel-Manzke syndrome, 616145 (3), Autosomal recessive |
| **TGFB2** | 99.85 % | 190220 | Loeys-Dietz syndrome 4, 614816 (3), Autosomal dominant; Camurati-Engelmann disease 2, 606631 (3), Autosomal dominant |
| **TGFB3** | 99.97 % | 190230 | Arrhythmogenic right ventricular dysplasia 1, 107970 (3), Autosomal dominant; Loeys-Dietz syndrome 5, 615582 (3), Autosomal dominant |
| **TGFBR1** | 99.84 % | 190181 | {Multiple self-healing squamous epithelioma, susceptibility to}, 132800 (3), Autosomal dominant; Loeys-Dietz syndrome 1, 609192 (3), Autosomal dominant |
| **TGFBR2** | 99.91 % | 190182 | Loeys-Dietz syndrome 2, 610168 (3), Autosomal dominant; Colorectal cancer, hereditary nonpolyposis, type 6, 614331 (3); Esophageal cancer, somatic, 133239 (3) |
| **TGIF1** | 99.99 % | 602630 | Holoprosencephaly 4, 142946 (3), Autosomal dominant |
| **TGM1** | 99.11 % | 190195 | Ichthyosis, congenital, autosomal recessive 1, 242300 (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 |
| **THRA** | 99.84 % | 190120 | Hypothyroidism, congenital, nongoitrous, 6, 614450 (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 |
| **TINF2** | 99.94 % | 604319 | Dyskeratosis congenita, autosomal dominant 3, 613990 (3), Autosomal dominant; Revesz syndrome, 268130 (3), Autosomal dominant |
| **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 |
| **TKT** | 97.06 % | 606781 | Short stature, developmental delay, and congenital heart defects, 617044 (3), Autosomal recessive |
| **TLK2** | 97.61 % | 608439 | Intellectual developmental disorder, autosomal dominant 57, 618050 (3), Autosomal dominant |
| **TLL1** | 99.84 % | 606742 | Atrial septal defect 6, 613087 (3), Autosomal dominant |
| **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 |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **TMEM138** | 99.94 % | 614459 | Joubert syndrome 16, 614465 (3), Autosomal recessive |
| **TMEM165** | 99.89 % | 614726 | Congenital disorder of glycosylation, type IIk, 614727 (3), Autosomal recessive |
| **TMEM167A** | 99.91 % | 620000 | *No OMIM phenotypes* |
| **TMEM17** | 99.16 % | 614950 | *No OMIM phenotypes* |
| **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 |
| **TMEM218** | 99.92 % | 619285 | Joubert syndrome 39, 619562 (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 |
| **TMEM263** | 99.9 % | | *No OMIM phenotypes* |
| **TMEM38B** | 99.82 % | 611236 | Osteogenesis imperfecta, type XIV, 615066 (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 |
| **TMEM98** | 99.92 % | 615949 | Nanophthalmos 4, 615972 (3), Autosomal dominant |
| **TMPRSS7** | 99.78 % | | *No OMIM phenotypes* |
| **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 |
| **TNNI2** | 99.8 % | 191043 | Arthrogryposis, distal, type 2B1, 601680 (3), Autosomal dominant |
| **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 |
| **TNNT3** | 99.92 % | 600692 | Arthrogryposis, distal, type 2B2, 618435 (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 |
| **TOMM7** | 100 % | 607980 | Garg-Mishra progeroid syndrome, 620601 (3), Autosomal recessive |
| **TONSL** | 99.87 % | 604546 | Spondyloepimetaphyseal dysplasia, sponastrime type, 271510 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **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 |
| **TP63** | 99.8 % | 603273 | Premature ovarian failure 21, 620311 (3), Autosomal dominant; Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 (3), Autosomal dominant; Hay-Wells syndrome, 106260 (3), Autosomal dominant; Split-hand/foot malformation 4, 605289 (3), Autosomal dominant; Orofacial cleft 8, 618149 (3); Rapp-Hodgkin syndrome, 129400 (3), Autosomal dominant; ADULT syndrome, 103285 (3), Autosomal dominant; Limb-mammary syndrome, 603543 (3), Autosomal dominant |
| **TP73** | 99.86 % | 601990 | Ciliary dyskinesia, primary, 47, and lissencephaly, 619466 (3), Autosomal recessive |
| **TPM1** | 99.65 % | 191010 | Left ventricular noncompaction 9, 611878 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 3, 115196 (3), Autosomal dominant; Cardiomyopathy, dilated, 1Y, 611878 (3), Autosomal dominant |
| **TPM2** | 99.93 % | 190990 | Arthrogryposis, distal, type 2B4, 108120 (3), Autosomal dominant; Arthrogryposis, distal, type 1A, 108120 (3), Autosomal dominant; Congenital myopathy 23, 609285 (3), Autosomal dominant |
| **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 |
| **TPRKB** | 95.96 % | 608680 | Galloway-Mowat syndrome 5, 617731 (3), Autosomal recessive |
| **TRAF3IP1** | 99.94 % | 607380 | Senior-Loken syndrome 9, 616629 (3), Autosomal recessive |
| **TRAF7** | 99.27 % | 606692 | Cardiac, facial, and digital anomalies with developmental delay, 618164 (3), Autosomal dominant |
| **TRAIP** | 99.82 % | 605958 | Seckel syndrome 9, 616777 (3), Autosomal recessive |
| **TRAP1** | 99.81 % | 606219 | *No OMIM phenotypes* |
| **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 |
| **TRAPPC9** | 99.85 % | 611966 | Intellectual developmental disorder, autosomal recessive 13, 613192 (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 |
| **TRIM32** | 99.99 % | 602290 | ?Bardet-Biedl syndrome 11, 615988 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110 (3), Autosomal recessive |
| **TRIM37** | 97.98 % | 605073 | Mulibrey nanism, 253250 (3), Autosomal recessive |
| **TRIM71** | 99.96 % | 618570 | Hydrocephalus, congenital, 4, 618667 (3), Autosomal dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **TRIO** | 99.92 % | 601893 | Intellectual developmental disorder, autosomal dominant 44, with microcephaly, 617061 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 63, with macrocephaly, 618825 (3), Autosomal dominant |
| **TRIP11** | 99.89 % | 604505 | Odontochondrodysplasia 1, 184260 (3), Autosomal recessive; Achondrogenesis, type IA, 200600 (3), Autosomal recessive |
| **TRIP12** | 99.54 % | 604506 | Intellectual developmental disorder, autosomal dominant 49, 617752 (3), Autosomal dominant |
| **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 |
| **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 |
| **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 |
| **TRPS1** | 99.98 % | 604386 | Trichorhinophalangeal syndrome, type III, 190351 (3), Autosomal dominant; Trichorhinophalangeal syndrome, type I, 190350 (3), Autosomal dominant |
| **TRPV3** | 99.9 % | 607066 | ?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400 (3), Autosomal dominant; Olmsted syndrome 1, 614594 (3), Autosomal dominant |
| **TRPV4** | 99.9 % | 605427 | Neuronopathy, distal hereditary motor, autosomal dominant 8, 600175 (3), Autosomal dominant; Spondylometaphyseal dysplasia, Kozlowski type, 184252 (3), Autosomal dominant; Digital arthropathy-brachydactyly, familial, 606835 (3), Autosomal dominant; [Sodium serum level QTL 1], 613508 (3); SED, Maroteaux type, 184095 (3), Autosomal dominant; Metatropic dysplasia, 156530 (3), Autosomal dominant; Scapuloperoneal spinal muscular atrophy, 181405 (3), Autosomal dominant; Hereditary motor and sensory neuropathy, type IIc, 606071 (3), Autosomal dominant; ?Avascular necrosis of femoral head, primary, 2, 617383 (3), Autosomal dominant; Parastremmatic dwarfism, 168400 (3), Autosomal dominant; Brachyolmia type 3, 113500 (3), Autosomal dominant |
| **TRPV6** | 99.85 % | 606680 | Hyperparathyroidism, transient neonatal, 618188 (3), Autosomal recessive |
| **TRRAP** | 99.19 % | 603015 | ?Deafness, autosomal dominant 75, 618778 (3), Autosomal dominant; Developmental delay with or without dysmorphic facies and autism, 618454 (3), Autosomal dominant |
| **TSC1** | 99.91 % | 605284 | Focal cortical dysplasia, type II, somatic, 607341 (3); Tuberous sclerosis-1, 191100 (3), Autosomal dominant; Lymphangioleiomyomatosis, 606690 (3) |
| **TSC2** | 99.88 % | 191092 | Lymphangioleiomyomatosis, somatic, 606690 (3); ?Focal cortical dysplasia, type II, somatic, 607341 (3); Tuberous sclerosis-2, 613254 (3), Autosomal dominant |
| **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 |
| **TSHZ3** | 100 % | 614119 | *No OMIM phenotypes* |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **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 |
| **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 |
| **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 |
| **TUBA1A** | 99.89 % | 602529 | Lissencephaly 3, 611603 (3), Autosomal dominant |
| **TUBA8** | 99.95 % | 605742 | Macrothrombocytopenia, isolated, 2, autosomal dominant, 619840 (3), Autosomal dominant |
| **TUBB** | 99.77 % | 191130 | Symmetric circumferential skin creases, congenital, 1, 156610 (3), Autosomal dominant; Cortical dysplasia, complex, with other brain malformations 6, 615771 (3), Autosomal dominant |
| **TUBB2A** | 81.57 % | 615101 | Cortical dysplasia, complex, with other brain malformations 5, 615763 (3), Autosomal dominant |
| **TUBB2B** | 82.06 % | 612850 | Cortical dysplasia, complex, with other brain malformations 7, 610031 (3), Autosomal dominant |
| **TUBB3** | 99.99 % | 602661 | Fibrosis of extraocular muscles, congenital, 3A, 600638 (3), Autosomal dominant; Cortical dysplasia, complex, with other brain malformations 1, 614039 (3), Autosomal dominant |
| **TUBB4A** | 99.94 % | 602662 | Dystonia 4, torsion, autosomal dominant, 128101 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 6, 612438 (3), Autosomal dominant |
| **TUBG1** | 99.38 % | 191135 | Cortical dysplasia, complex, with other brain malformations 4, 615412 (3), Autosomal dominant |
| **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 |
| **TWIST1** | 100 % | 601622 | Craniosynostosis 1, 123100 (3), Autosomal dominant; Robinow-Sorauf syndrome, 180750 (3), Autosomal dominant; Sweeney-Cox syndrome, 617746 (3), Autosomal dominant; Saethre-Chotzen syndrome with or without eyelid anomalies, 101400 (3), Autosomal dominant |
| **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 |
| **TXN2** | 99.1 % | 609063 | ?Combined oxidative phosphorylation deficiency 29, 616811 (3), Autosomal recessive |
| **TXNDC15** | 99.86 % | 617778 | Meckel syndrome 14, 619879 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **TXNL4A** | 99.98 % | 611595 | Burn-McKeown syndrome, 608572 (3), Autosomal recessive |
| **U2AF2** | 99.96 % | 191318 | Developmental delay, dysmorphic facies, and brain anomalies, 620535 (3), Autosomal dominant |
| **UBA1** | 99.76 % | 314370 | Spinal muscular atrophy, X-linked 2, infantile, 301830 (3), X-linked recessive; VEXAS syndrome, somatic, 301054 (3) |
| **UBA2** | 99.78 % | 613295 | ACCES syndrome, 619959 (3), Autosomal dominant |
| **UBE2T** | 99.23 % | 610538 | Fanconi anemia, complementation group T, 616435 (3), Autosomal recessive |
| **UBE3B** | 99.89 % | 608047 | Kaufman oculocerebrofacial syndrome, 244450 (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 |
| **UBTF** | 99.81 % | 600673 | Neurodegeneration, childhood-onset, with brain atrophy, 617672 (3), Autosomal dominant |
| **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 |
| **UGGT1** | 98.94 % | 605897 | Congenital disorder of glycosylation, type IICC, 621381 (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 |
| **UNC50** | 99.57 % | 617826 | *No OMIM phenotypes* |
| **UQCC2** | 99.83 % | 614461 | Mitochondrial complex III deficiency, nuclear type 7, 615824 (3), Autosomal recessive |
| **UQCRB** | 99.09 % | 191330 | Mitochondrial complex III deficiency, nuclear type 3, 615158 (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 |
| **UROS** | 99.98 % | 606938 | Porphyria, congenital erythropoietic, 263700 (3), Autosomal recessive |
| **USP14** | 99.97 % | 607274 | *No OMIM phenotypes* |
| **USP18** | 90.87 % | 607057 | Pseudo-TORCH syndrome 2, 617397 (3), Autosomal recessive |
| **USP27X** | 100 % | 300975 | Intellectual developmental disorder, X-linked 105, 300984 (3), X-linked 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 |
| **VAMP1** | 99.99 % | 185880 | Myasthenic syndrome, congenital, 25, 618323 (3), Autosomal recessive; Spastic ataxia 1, autosomal dominant, 108600 (3), Autosomal dominant |
| **VARS2** | 99.67 % | 612802 | Combined oxidative phosphorylation deficiency 20, 615917 (3), Autosomal recessive |
| **VDR** | 99.61 % | 601769 | Rickets, vitamin D-resistant, type IIA, 277440 (3), Autosomal recessive |
| **VEGFC** | 99.69 % | 601528 | Lymphatic malformation 4, 615907 (3), Autosomal dominant |
| **VIPAS39** | 99.76 % | 613401 | Arthrogryposis, renal dysfunction, and cholestasis 2, 613404 (3), Autosomal recessive |
| **VLDLR** | 99.94 % | 192977 | Cerebellar hypoplasia, impaired intellectual development, and dysequilibrium syndrome 1, 224050 (3), Autosomal recessive |
| **VPS13B** | 99.81 % | 607817 | Cohen syndrome, 216550 (3), Autosomal recessive |
| **VPS33A** | 99.08 % | 610034 | Mucopolysaccharidosis-plus syndrome, 617303 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **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 |
| **VPS4A** | 99.6 % | 609982 | CIMDAG syndrome, 619273 (3), Autosomal dominant |
| **VPS50** | 98.46 % | 616465 | Neurodevelopmental disorder with microcephaly, seizures, and neonatal cholestasis, 619685 (3), Autosomal recessive |
| **VPS51** | 99.81 % | 615738 | Pontocerebellar hypoplasia, type 13, 618606 (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 |
| **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 |
| **WASHC5** | 99.89 % | 610657 | Ritscher-Schinzel syndrome 1, 220210 (3), Autosomal recessive; Spastic paraplegia 8, autosomal dominant, 603563 (3), Autosomal dominant |
| **WBP11** | 99.71 % | 618083 | Vertebral, cardiac, tracheoesophageal, renal, and limb defects, 619227 (3), Autosomal dominant |
| **WBP4** | 99.94 % | 604981 | Neurodevelopemental disorder with hypotonia, feeding difficulties, facial dysmorphism, and brain abnormalities, 620852 (3), Autosomal recessive |
| **WDHD1** | 99.64 % | 608126 | *No OMIM phenotypes* |
| **WDPCP** | 99.79 % | 613580 | Bardet-Biedl syndrome 15, 615992 (3), Autosomal recessive; Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085 (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 |
| **WDR26** | 99.72 % | 617424 | Skraban-Deardorff syndrome, 617616 (3), Autosomal dominant |
| **WDR35** | 99.87 % | 613602 | Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 (3), Autosomal recessive; Cranioectodermal dysplasia 2, 613610 (3), Autosomal recessive |
| **WDR37** | 99.56 % | 618586 | Neurooculocardiogenitourinary syndrome, 618652 (3), Autosomal dominant |
| **WDR4** | 99.76 % | 605924 | Galloway-Mowat syndrome 6, 618347 (3), Autosomal recessive; Microcephaly, growth deficiency, seizures, and brain malformations, 618346 (3), Autosomal recessive |
| **WDR44** | 99.52 % | 301070 | *No OMIM phenotypes* |
| **WDR47** | 93.11 % | 615734 | *No OMIM phenotypes* |
| **WDR62** | 99.72 % | 613583 | Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317 (3), Autosomal recessive |
| **WDR73** | 99.9 % | 616144 | Galloway-Mowat syndrome 1, 251300 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **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 |
| **WDR91** | 99.9 % | 616303 | *No OMIM phenotypes* |
| **WLS** | 98.41 % | 611514 | Zaki syndrome, 619648 (3), Autosomal recessive |
| **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 |
| **WNT10B** | 99.68 % | 601906 | Tooth agenesis, selective, 8, 617073 (3), Autosomal dominant; Split-hand/foot malformation 6, 225300 (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 |
| **WNT5A** | 99.71 % | 164975 | Robinow syndrome, autosomal dominant 1, 180700 (3), Autosomal dominant |
| **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 |
| **WNT7B** | 100 % | 601967 | *No OMIM phenotypes* |
| **WNT9B** | 99.28 % | 602864 | *No OMIM phenotypes* |
| **WRAP53** | 99.62 % | 612661 | Dyskeratosis congenita, autosomal recessive 3, 613988 (3), Autosomal recessive |
| **WSB2** | 99.87 % | | *No OMIM phenotypes* |
| **WT1** | 97.92 % | 607102 | Mesothelioma, somatic, 156240 (3); Meacham syndrome, 608978 (3), Autosomal dominant; Frasier syndrome, 136680 (3), Somatic mutation, Autosomal dominant; Nephrotic syndrome, type 4, 256370 (3), Autosomal dominant; Denys-Drash syndrome, 194080 (3), Somatic mutation, Autosomal dominant; Wilms tumor, type 1, 194070 (3), Somatic mutation, Autosomal dominant |
| **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 |
| **XPNPEP3** | 99.89 % | 613553 | Nephronophthisis-like nephropathy 1, 613159 (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 |
| **XYLT2** | 99.69 % | 608125 | {Pseudoxanthoma elasticum, modifier of severity of}, 264800 (3), Autosomal recessive; Spondyloocular syndrome, 605822 (3), Autosomal recessive |
| **YAP1** | 99.85 % | 606608 | Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or impaired intellectual development, 120433 (3), Autosomal dominant |
| **YIF1B** | 99.35 % | 619109 | Kaya-Barakat-Masson syndrome, 619125 (3), Autosomal recessive |
| **YRDC** | 98.59 % | 612276 | Galloway-Mowat syndrome 10, 619609 (3), Autosomal recessive |
| **YWHAE** | 99.51 % | 605066 | *No OMIM phenotypes* |
| **YWHAG** | 99.94 % | 605356 | Developmental and epileptic encephalopathy 56, 617665 (3), Autosomal dominant |
| **YY1** | 99.97 % | 600013 | Gabriele-de Vries syndrome, 617557 (3), Autosomal dominant |
| **YY1AP1** | 99.56 % | 607860 | Grange syndrome, 602531 (3), Autosomal recessive |
| **ZBTB18** | 99.69 % | 608433 | Intellectual developmental disorder, autosomal dominant 22, 612337 (3), Autosomal dominant |
| **ZBTB20** | 99.96 % | 606025 | Primrose syndrome, 259050 (3), Autosomal dominant |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **ZBTB24** | 99.92 % | 614064 | Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069 (3), Autosomal recessive |
| **ZBTB7A** | 99.78 % | 605878 | Macrocephaly, neurodevelopmental delay, lymphoid hyperplasia, and persistent fetal hemoglobin, 619769 (3), Autosomal dominant |
| **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 |
| **ZEB1** | 99.95 % | 189909 | Corneal dystrophy, posterior polymorphous, 3, 609141 (3), Autosomal dominant; Corneal dystrophy, Fuchs endothelial, 6, 613270 (3), Autosomal dominant |
| **ZEB2** | 99.86 % | 605802 | Mowat-Wilson syndrome, 235730 (3), Autosomal dominant |
| **ZFP57** | 99.97 % | 612192 | Diabetes mellitus, transient neonatal 1, 601410 (3), Autosomal recessive, Autosomal dominant |
| **ZFPM2** | 99.99 % | 603693 | Diaphragmatic hernia 3, 610187 (3), Autosomal dominant; 46XY sex reversal 9, 616067 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant |
| **ZFX** | 99.7 % | 314980 | Intellectual developmental disorder, X-linked syndromic 37, 301118 (3), X-linked |
| **ZIC1** | 99.95 % | 600470 | ?Craniosynostosis 6, 616602 (3), Autosomal dominant; Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736 (3), Autosomal dominant |
| **ZIC2** | 99.97 % | 603073 | Holoprosencephaly 5, 609637 (3), Autosomal dominant |
| **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 |
| **ZMIZ1** | 99.23 % | 607159 | Neurodevelopmental disorder with dysmorphic facies and distal skeletal anomalies, 618659 (3), Autosomal dominant |
| **ZMPSTE24** | 97.94 % | 606480 | Mandibuloacral dysplasia with type B lipodystrophy, 608612 (3), Autosomal recessive; Restrictive dermopathy 1, 275210 (3), Autosomal recessive |
| **ZMYM2** | 99.85 % | 602221 | Neurodevelopmental-craniofacial syndrome with variable renal and cardiac abnormalities, 619522 (3), Autosomal dominant |
| **ZMYND10** | 99.88 % | 607070 | Ciliary dyskinesia, primary, 22, 615444 (3), Autosomal recessive |
| **ZMYND11** | 99.93 % | 608668 | Intellectual developmental disorder, autosomal dominant 30, 616083 (3), Autosomal dominant |
| **ZMYND8** | 99.95 % | 615713 | *No OMIM phenotypes* |
| **ZNF335** | 99.92 % | 610827 | Microcephaly 10, primary, autosomal recessive, 615095 (3), Autosomal recessive |
| **ZNF423** | 99.92 % | 604557 | Nephronophthisis 14, 614844 (3), Autosomal recessive, Autosomal dominant; Joubert syndrome 19, 614844 (3), Autosomal recessive, Autosomal dominant |
| **ZNF462** | 99.95 % | 617371 | Weiss-Kruszka syndrome, 618619 (3), Autosomal dominant |
| **ZNF526** | 99.97 % | 614387 | Dentici-Novelli neurodevelopmental syndrome, 619877 (3), Autosomal recessive |
| **ZNF668** | 99.82 % | 617103 | Neurodevelopmental disorder with poor growth, large ears, and dysmorphic facies, 620194 (3), Autosomal recessive |
| **ZNF699** | 99.98 % | 609571 | DEGCAGS syndrome, 619488 (3), Autosomal recessive |
| **ZNF865** | 99.99 % | | *No OMIM phenotypes* |
| **ZNHIT3** | 64.64 % | 604500 | PEHO syndrome, 260565 (3), Autosomal recessive |
| **ZNRF3** | 99.52 % | 612062 | *No OMIM phenotypes* |
| **ZPR1** | 99.66 % | 603901 | ?Growth restriction, hypoplastic kidneys, alopecia, and distinctive facies, 619321 (3), Autosomal recessive |
| **ZRSR2** | 99.74 % | 300028 | Orofaciodigital syndrome XXI, 301132 (3), X-linked recessive |

# Fetal anomalies

Gene panel

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **ZSCAN10** | 99.83 % | 618365 | Otofacial neurodevelopmental syndrome, 620910 (3), Autosomal recessive |
| **ZSWIM6** | 99.51 % | 615951 | Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865 (3), Autosomal dominant; Acromelic frontonasal dysostosis, 603671 (3), Autosomal dominant |

## 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.