

Mackenzie's Mission Gene & Condition List

What conditions are being screened for in Mackenzie's Mission?

Genetic carrier screening offered through this research study has been carefully developed. It is focused on providing people with information about their chance of having children with a severe genetic condition occurring in childhood. The screening is designed to provide genetic information that is relevant and useful, and to minimise uncertain and unclear information.

How the conditions and genes are selected

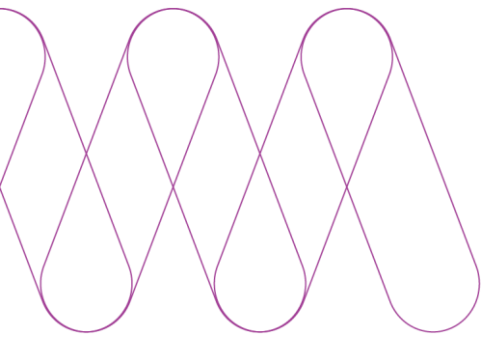
The Mackenzie's Mission reproductive genetic carrier screen currently includes approximately 1300 genes which are associated with about 750 conditions. The reason there are fewer conditions than genes is that some genetic conditions can be caused by changes in more than one gene. The gene list is reviewed regularly.

To select the conditions and genes to be screened, a committee comprised of experts in genetics and screening was established including: clinical geneticists, genetic scientists, a genetic pathologist, genetic counsellors, an ethicist and a parent of a child with a genetic condition. The following criteria were developed and are used to select the genes to be included:

- Screening the gene is **technically possible** using currently available technology
- The gene is **known to cause a genetic condition**
- The condition affects people in **childhood**
- The condition has a **serious** impact on a person's quality of life and/or is life-limiting
 - For many of the conditions there is no treatment or the treatment is very burdensome for the child and their family. For some conditions very early diagnosis and treatment can make a difference for the child.

Types of conditions included

The conditions included in the screening vary in the way that they affect people and can involve one or many different parts of the body. Some of the ways that the conditions affect children can include:



Shortened life expectancy

Some conditions screened lead to a shortened life – either causing death in childhood, or with symptoms in childhood and early death in adulthood.

Intellectual disability

Some conditions cause intellectual disability which limits a person's ability to learn and develop independence. In some conditions this is severe – the child with the condition may never learn to walk or talk, whereas in others it is less severe – the child may be able to do many things for him or herself, but may need extra help and may not be able to live independently as an adult.

Physical conditions

Some conditions may affect the person physically, such as causing congenital heart disease or differences in how the limbs develop. In some cases these symptoms may be treatable, whereas in other cases there is no treatment available.

Neurological and muscular conditions

Some conditions are due to a problem with the brain itself, problems with the way the brain sends signals through the spinal cord and nerves to the body, or because the muscles themselves are weak. Sometimes these conditions can get worse over time.

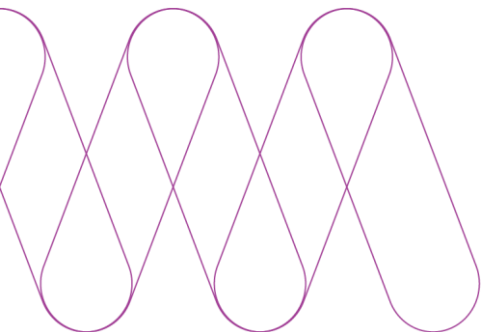
Important information about analysis and reporting of results

In addition to knowing what genes are being screened in Mackenzie's Mission, it is essential to also understand how the results are being analysed and reported. The screening is designed to be offered to a large number of people, with a focus on providing meaningful information that is useful to inform family planning.

Although a gene may be screened through Mackenzie's Mission, as outlined below, there are situations where particular genetic changes may not be analysed or reported:

A focus on severe conditions that occur in childhood

Some genetic conditions may vary in how much they affect people. This is because some genetic changes can have a more severe effect than others. Knowing about a chance of having a child with a mild form of a genetic condition often does not alter parents' reproductive plans and can cause confusion and distress. The focus of screening in this study is to provide information about the genetic chance of having a child with a severe



genetic condition. If a particular change in a gene is only associated with a mild form of the condition, this will not usually be reported to participants.

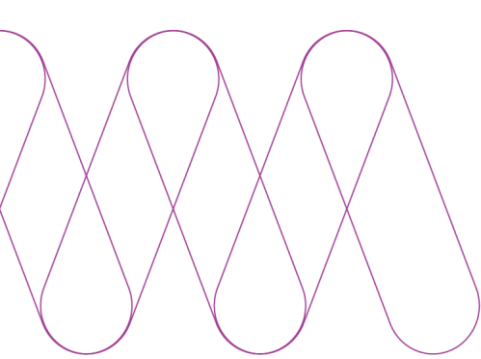
A ‘couple screen’

In this study, a couple screening approach is used, meaning both biological parents of the pregnancy or planned pregnancy are screened at the same time. We are all genetic carriers for inherited conditions, however, many of the severe genetic conditions that occur in childhood are caused by **both** the biological mother and the biological father being carriers for the same autosomal recessive condition, or the biological mother being a carrier for an X-linked condition. Because of the very large number of genes screened, screening both biological parents at the same time and issuing a combined result provides the most useful information for that couple. If only one partner is a genetic carrier for an autosomal recessive condition/s, this will not be reported. This is because together, the couple will have a low chance of having a child with the condition. It is not practical to issue individual results for every person screened, and the results are most meaningful when combined together. If in the future either person has a new partner, that new couple should consider screening, as the results for the original couple are not relevant to the new couple.

A screening approach

There are many different types of genetic changes that can cause genetic conditions. It is important to understand that even with a ‘low chance’ result, there remains a small chance of a couple having a child with a genetic condition that was screened through Mackenzie’s Mission. The genetic testing offered through this study is referred to as ‘screening’, because the technology used will detect many, but not all, genetic changes causing these conditions.

For fragile X syndrome and spinal muscular atrophy, targeted tests are used (each testing laboratory uses different methods which are described in the Mackenzie’s Mission genetic carrier screening laboratory reports). In some circumstances, fragile X screening may also include AGG interruption analysis. For all other conditions, massively parallel sequencing is used. The testing techniques will not detect all genetic changes in each gene screened. For example, larger sections of extra or missing genetic material (called copy number variants, >50bp) or rearrangements will not be detected, which in some instances may be the main cause, or a major cause of a particular condition; examples include the *DMD*, *F8* and *TANGO2* genes. Additionally, in some cases this screening may not cover all genes associated with a particular genetic condition; this may be because the gene is associated with a mild form of the condition, or there are technical challenges in screening the gene.



Screening results are based on current knowledge

Knowledge about our genes is changing every day. Results from the genetic carrier screening performed through this study are being analysed and interpreted by experienced laboratory scientists. Their interpretation of the genetic information will be based on currently available information. So far, detailed genetic studies have not been done in people from all of the ethnic backgrounds found in the Australian population. This can make it more challenging to interpret some types of genetic results. For people from backgrounds for which there is less information, there may be a higher chance that couples who have an increased chance of having an affected child will not be identified.

When there is a family history of a genetic condition

While genetic carrier screening is relevant to everyone, regardless of whether there is a family history of a genetic condition, there will be some people who take part in this study who have a genetic condition themselves, or who have a relative/s with a genetic condition. It is important for people with a family history of a genetic condition who are wanting to have screening through Mackenzie's Mission to speak to a member of our study genetic counselling team, to determine whether the reproductive genetic carrier screen offered through this study is right for them. **Even if the gene causing the condition in their family is on the Mackenzie's Mission gene list, it is important to clarify whether the screening offered is able to detect the genetic change(s) present in that family.**

Please don't hesitate to contact our study team

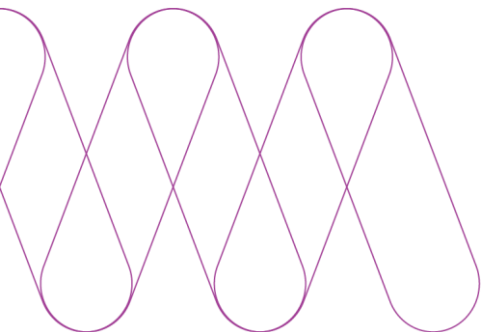
Our study team includes experienced genetic counsellors, clinical geneticists and laboratory scientists. We encourage healthcare providers and potential participants to contact us to discuss any queries they may have about the conditions screened through Mackenzie's Mission.

Mackenzie's Mission Study Team

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Phone: 1800 976 299

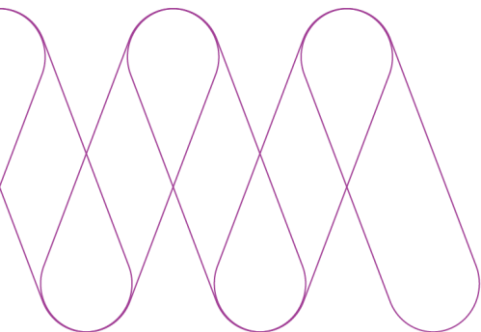
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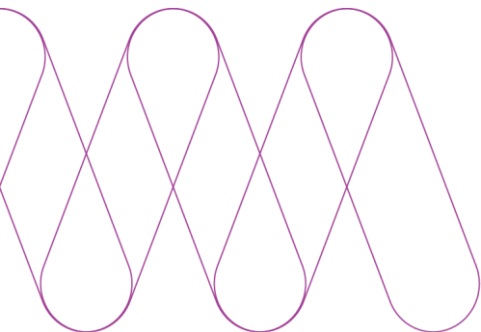
List of genes and conditions screened in Mackenzie's Mission

Please note that some genes appear on this list more than once, as changes in some genes can cause more than one different condition.

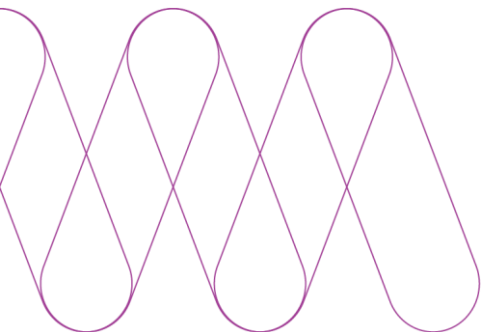
| Condition | Genes |
|---|---|
| Syndromes with intellectual disability | |
| Multiple congenital abnormalities with intellectual disability | |
| Achalasia-addisonianism-alacrimia syndrome | AAAS |
| Al Kaissi syndrome | CDK10 |
| Athabaskan brainstem dysgenesis syndrome | HOXA1 |
| Arthrogyroposis, intellectual disability, and seizure disorder | SLC35A3 |
| 3MC syndrome | COLEC11, MASP1 |
| Bardet-Biedl syndrome | ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, LZTFL1, MKKS, MKS1, SDCCAG8, TTC8 |
| Basel-Vanagait-Smirin-Yosef syndrome | MED25 |
| Behr syndrome | OPA1 |
| Boucher-Neuhauser syndrome | PNPLA6 |
| Bosley-Salih-Alorainy syndrome | HOXA1 |
| Brunner syndrome | MAOA |
| Goldberg-Shprintzen megacolon syndrome | KIFBP |
| Borjeson-Forssman-Lehmann syndrome | PHF6 |
| Bloom syndrome | BLM |
| Partington syndrome | ARX |
| Pitt-Hopkins-like syndrome | CNTNAP2 |
| Polyhydramnios, megalencephaly, and symptomatic epilepsy | STRADA |
| PERCHING syndrome | KLHL7 |
| Shaheen syndrome | COG6 |
| Growth retardation, intellectual developmental disorder, hypotonia, and hepatopathy | IARS1 |
| Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia (CAGSSS) | IARS2 |
| Carey-Fineman-Ziter syndrome | MYMK |
| Cerebellofaciodental syndrome | BRF1 |
| Craniofacial dysmorphism, skeletal anomalies, and intellectual disability syndrome | TMCO1 |



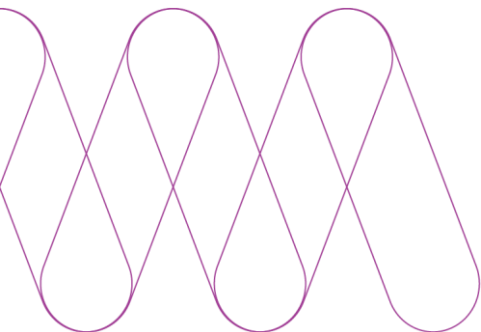
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| CHIME syndrome | PIGL |
| COACH syndrome | CC2D2A, RPGRIP1L, TMEM67 |
| Cockayne syndrome | ERCC4, ERCC5, ERCC6, ERCC8 |
| Cohen syndrome | VPS13B |
| Cerebrooculofacioskeletal syndrome (COFS) | ERCC2, ERCC6 |
| Coffin-Lowry syndrome | RPS6KA3 |
| Cowchock syndrome | AIFM1 |
| De Sanctis-Cacchione syndrome | ERCC6 |
| Developmental delay with short stature, dysmorphic features, and sparse hair | DPH1 |
| Donnai-Barrow syndrome | LRP2 |
| DOOR syndrome | TBC1D24 |
| XFE progeroid syndrome | ERCC4 |
| Desmosterolosis | DHCR24 |
| Dyggve-Melchior-Clausen disease | DYM |
| Elsahy-Waters syndrome | CDH11 |
| Fragile X syndrome | FMR1 |
| Frontometaphyseal dysplasia | FLNA |
| Galloway-Mowat syndrome | WDR73, OSGEP |
| Gillespie syndrome | ITPR1 |
| Griscelli syndrome | RAB27A |
| HSAN2D syndrome | SCN9A |
| Hypoparathyroidism-retardation-dysmorphism syndrome | TBCE |
| Hypotonia, infantile, with psychomotor retardation and characteristic facies | TBCK, UNC80, NALCN |
| Jawad syndrome | RBBP8 |
| Jensen syndrome | TIMM8A |
| Johanson-Blizzard syndrome | UBR1 |
| IFAP syndrome with or without BRESHECK syndrome | MBTPS2 |
| Immunoskeletal dysplasia with neurodevelopmental abnormalities | EXTL3 |
| Infantile liver failure syndrome | LARS1 |
| Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies | OTUD6B |
| Intellectual developmental disorder with cardiac arrhythmia | GNB5 |
| Lujan-Fryns syndrome | MED12 |
| Kohlschutter-tonz syndrome | ROGDI |
| Ohdo syndrome | MED12 |



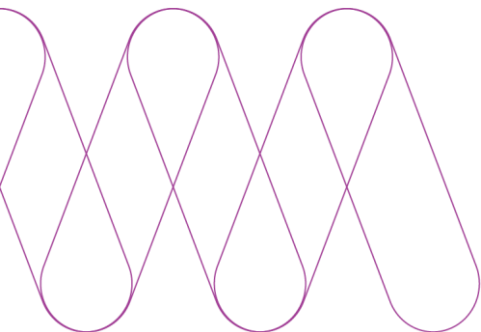
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| Opitz-Kaveggia syndrome | MED12 |
| Opitz GBBB syndrome | MID1 |
| Oliver-McFarlane syndrome | PNPLA6 |
| Mosaic variegated aneuploidy syndrome | BUB1B |
| MEHMO syndrome | EIF2S3 |
| Muscular dystrophy, congenital, with cataracts and intellectual disability | INPP5K |
| Nijmegen breakage syndrome | NBN, RAD50 |
| Nance-Horan syndrome | NHS |
| Neurodevelopmental disorder with brain anomalies and additional features | PLAA, PRUNE1, VARS1, WDR45B |
| Multiple congenital anomalies-hypotonia-seizures syndrome | PIGA, PIGN, PIGT |
| Renpenning syndrome | PQBP1 |
| Salt and pepper developmental regression syndrome | ST3GAL5 |
| Seckel syndrome | ATR, CENPJ, CEP152, RBBP8 |
| SESAME syndrome | KCNJ10 |
| Smith-Lemli-Opitz syndrome | DHCR7 |
| Spastic paraplegia and psychomotor retardation with or without seizures | HACE1 |
| LIG4 syndrome | LIG4 |
| Wieacker-Wolff syndrome | ZC4H2 |
| Alacrima, achalasia, and intellectual disability syndrome | GMPPA |
| Chudley-McCullough syndrome | GPSM2 |
| Growth retardation, developmental delay, coarse facies, and early death | FTO |
| Martsolf syndrome | RAB3GAP2 |
| Pierson syndrome | LAMB2 |
| Hemorrhagic destruction of the brain with subependymal calcification and cataracts | JAM3 |
| Hennekam lymphangiectasia-lymphedema syndrome | CCBE1, FAT4 |
| Perlman syndrome | DIS3L2 |
| Temtamy preaxial brachydactyly syndrome | CHSY1 |
| Filippi syndrome | CKAP2L |
| Fraser syndrome | FRAS1, FREM2 |
| Orofaciodigital syndrome | CPLANE1, C2CD3, DDX59, SERPINH1, TMEM107, TCTN3 |
| Roberts syndrome | ESCO2 |



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|---|---------------------------|
| SC phocomelia syndrome | ESCO2 |
| Warburg micro syndrome | RAB18, RAB3GAP1, RAB3GAP2 |
| Woodhouse-Sakati syndrome | DCAF17 |
| Van Maldergem syndrome | DCHS1, FAT4 |
| Warsaw breakage syndrome | DDX11 |
| You-Hoover-Fong syndrome | TELO2 |
| Syndromic microcephaly | |
| Microcephaly, epilepsy, and diabetes syndrome | IER3IP1 |
| Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy | QARS1 |
| Microcephaly-capillary malformation syndrome | STAMPB |
| Microcephaly, short stature, and impaired glucose metabolism | TRMT10A |
| Microcephaly, short-stature and endocrine dysfunction | XRCC4 |
| Microcephaly, short stature, and limb abnormalities | DONSON |
| Microcephaly and chorioretinopathy | TUBGCP4, TUBGCP6 |
| Microcephaly, seizures, spasticity, and brain calcification | PCDH12 |
| X-linked syndromic intellectual disability | |
| Turner type | HUWE1 |
| Claes-Jensen type | KDM5C |
| Christianson type | SLC9A6 |
| Siderius type | PHF8 |
| Type 14 | UPF3B |
| CK syndrome | NSDHL |
| Snyder-Robinson type | SMS |
| Nascimento type | UBE2A |
| Raymond type | ZDHHC9 |
| Intellectual disability, truncal obesity, retinal dystrophy, and micropenis | INPP5E |
| Intellectual disability, X-linked, with cerebellar hypoplasia and distinctive facial appearance | OPHN1 |
| Syndromic brain malformations | |
| MASA syndrome | L1CAM |
| CRASH syndrome | L1CAM |
| Agenesis of the corpus callosum with peripheral neuropathy (Andermann syndrome) | SLC12A6 |
| Acrocallosal syndrome | KIF7 |



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| Proud syndrome | ARX |
| Temtamy syndrome | C12orf57 |
| Cerebroretinal microangiopathy with calcifications and cysts | CTC1 |
| Vici syndrome | EPG5 |
| Proliferative vasculopathy and hydraencephaly-hydrocephaly syndrome | FLVCR2 |
| Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity | PTPN23 |
| Syndromic skin conditions with intellectual disability | |
| Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome | SNAP29 |
| Adams-Oliver syndrome | DOCK6, EOGT |
| Syndromic vision conditions with intellectual disability | |
| Peter's plus syndrome | B3GLCT |
| Congenital cataracts, hearing loss, and neurodegeneration | SLC33A1 |
| Knobloch syndrome | COL18A1 |
| Lowe syndrome | OCRL |
| Kaufman oculocerebrofacial syndrome | UBE3B |
| Kahrizi syndrome | SRD5A3 |
| Optic atrophy with or without ataxia, intellectual disability, and seizures | RTN4IP1 |
| Norrie disease | NDP |
| Syndromic growth conditions with intellectual disability | |
| Simpson-Golabi-Behmel syndrome | OFD1, GPC3 |
| Severe, lethal, neonatal syndromes | |
| Meckel syndrome | CC2D2A, CEP290, MKS1, NPHP3, RPGRIP1L, TMEM216, TMEM231, TMEM67 |
| Alkuraya-Kucinkas syndrome | KIAA1109 |
| Bowen-Conradi syndrome | EMG1 |
| Fetal akinesia deformation sequence | RAPSN |
| Lethal congenital contracture syndrome | CNTNAP1, GLE1, GLDN |
| Ventriculomegaly with cystic kidney disease | CRB2 |
| Hydroletharus syndrome | HYLS1, KIF7 |
| TARP syndrome | RBM10 |
| Rigidity and multifocal seizure syndrome, lethal neonatal | BRAT1 |



Syndromes without intellectual disability

Multiple pterygium syndrome

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| Lethal type | CHRNA1, RIPK4 |
| Escobar syndrome | CHRNA1 |

Multiple congenital abnormalities

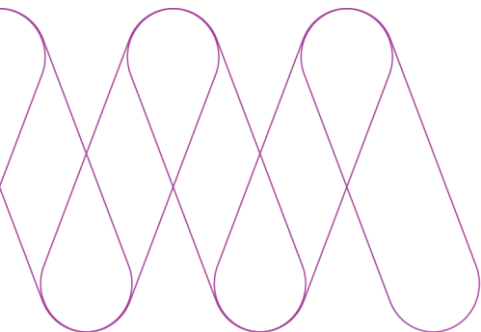
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| Burn-McKeown syndrome | TXNL4A |
| Bifid nose with or without anorectal and renal anomalies | FREM1 |
| Crisponi syndrome | CRLF1, CLCF1 |
| McKusick-Kaufman syndrome | MKKS |
| Shwachman-Diamond syndrome | SBDS |
| Split-hand foot malformation | WNT10B |
| Werner syndrome | WRN |
| VACTERL association X-linked | ZIC3 |
| Lipodystrophy, congenital generalized | BSCL2, CAVIN1 |
| Wolfram syndrome | CISD2, WFS1 |
| Urofacial syndrome | HPSE2, LRIG2 |

Syndromic skin and skeletal conditions

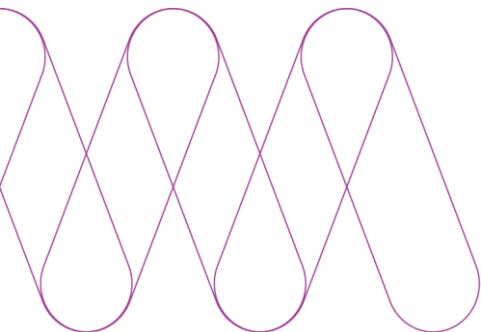
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| Rothmund-Thomson syndrome | RECQL4 |
| Alstrom syndrome | ALMS1 |
| GAPO syndrome | ANTXR1 |
| HELIX syndrome | CLDN10 |
| Haim-Munk syndrome | CTSC |
| Laryngoonychocutaneous syndrome | LAMA3 |
| Miller syndrome | DHODH |
| Macrocephaly, alopecia, cutis laxa, and scoliosis | RIN2 |
| Mandibuloacral dysplasia with type B lipodystrophy | ZMPSTE24 |
| Dyskeratosis congenita | DKC1, RTEL1, WRAP53 |
| Papillon-Lefevre syndrome | CTSC |
| Spondyloocular syndrome | XYLT2 |
| Treacher-Collins syndrome | POLR1C |
| Schimke immunosseous dysplasia | SMARCAL1 |

Syndromic vision and hearing conditions

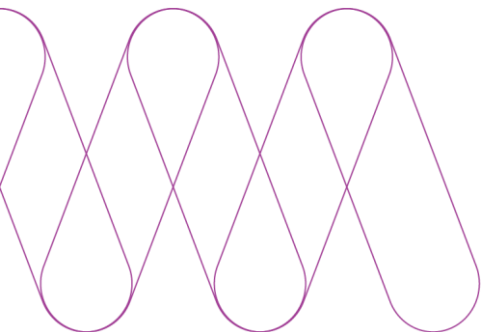
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| Usher syndrome | ADGRV1, CDH23, CLRN1, MYO7A, PCDH15, USH1C, USH1G, USH2A, WHRN |
| Retinitis pigmentosa with skeletal anomalies | CWC27 |



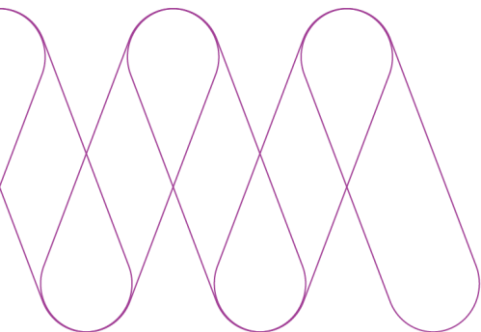
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| Jalili syndrome | CNNM4 |
| Syndromic vision and renal conditions | |
| Senior-Loken syndrome | CEP290, NPHP1, NPHP4, SDCCAG8, IQCB1, WDR19 |
| Mitochondrial conditions | |
| Conditions affecting multiple body systems | |
| Combined oxidative phosphorylation deficiency | AARS2, C12orf65, CARS2, FARS2, ELAC2, GFM1, GTPBP3, MTFMT, MTO1, NARS2, RMND1, TSFM, TUFM, VARS2, TRIT1, EARS2 |
| Leigh and Leigh-like syndrome | |
| Mitochondrial complex I deficiency | ACAD9, FOXRED1, NUBPL, NDUFA1, NDUFAF2, NDUFAF5, NDUFAF6, NDUFA10, NDUFA11, NDUFS6, NDUFS4, NDUFS2, NDUFS7, NDUFS8, NDUFS1, NDUFV1, NDUFV2 |
| Leigh syndrome due to cytochrome c oxidase deficiency | COX15 |
| Leigh syndrome, French Canadian type | LRPPRC |
| Other mitochondrial conditions | |
| Mitochondrial complex II deficiency | SDHAF1 |
| Mitochondrial complex III deficiency | BCS1L, LYRM7, TTC19, UQCRQ |
| Mitochondrial complex IV deficiency | COX10, COA8, COX20, SURF1, PET100 |
| Mitochondrial complex V deficiency | TMEM70 |
| Mitochondrial DNA depletion syndrome | DGUOK, FBXL4, MGME1, MPV17, RRM2B, SUCLA2, SUCLG1, TK2, TWNK, TYMP |
| Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE) | TWNK |
| Multiple mitochondrial dysfunctions syndrome | BOLA3, IBA57, ISCA2, NFU1 |
| Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2 | COX15, SCO2 |
| Sideroblastic anaemia with B-cell immunodeficiency, periodic fevers, and developmental delay | TRNT1 |
| Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation | DARS2 |
| Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis (HUPRA syndrome) | SARS2 |
| HSD10 disease | HSD17B10 |
| Mohr-Tranebjaerg syndrome | TIMM8A |



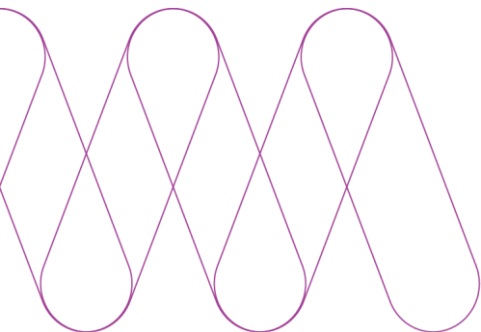
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| Mitochondrial neurodevelopmental disorder, with abnormal movements and lactic acidosis | WARS2 |
| Myopathy, lactic acidosis, and sideroblastic anaemia | PUS1, LARS2, YARS2 |
| Myopathy, mitochondrial, and ataxia | MSTO1 |
| Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency | ECHS1 |
| Lysosomal storage disorders | |
| Mannosidosis | |
| Alpha | MAN2B1 |
| Beta | MANBA |
| Mucopolysaccharidosis | |
| Mucopolysaccharidosis | GALNS, GNS, GUSB, IDS, IDUA |
| Type VI (Maroteaux-Lamy) | ARSB |
| Type IVB (Morquio) | GLB1 |
| Type IIIA (Sanfilippo A) | SGSH |
| Type IIIB (Sanfilippo B) | NAGLU |
| Type IIIC (Sanfilippo C) | HGSNAT |
| Cystinosis | |
| Atypical nephropathic | CTNS |
| Nephropathic | CTNS |
| Late-onset juvenile or adolescent nephropathic | CTNS |
| Ocular non-nephropathic | CTNS |
| Other lysosomal storage disorders | |
| Galactosialidosis | CTSA |
| Yunis-Varon syndrome | FIG4 |
| Fucosidosis | FUCA1 |
| Farber lipogranulomatosis | ASAH1 |
| Glycogen storage disease (Pompe) | GAA |
| Geleophysic dysplasia | ADAMTSL2 |
| Krabbe disease | GALC, PSAP |
| Fabry disease | GLA |
| GM1-gangliosidosis | GLB1 |
| GM2-gangliosidosis | HEXA, GM2A |
| Metachromatic leukodystrophy | ARSA, PSAP |
| Mucopolysaccharidosis | GNPTAB, GNPTG, MCOLN1 |
| Polyglucosan body myopathy 1 with or without immunodeficiency | RBCK1 |



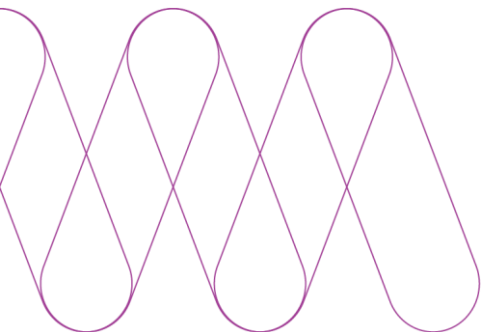
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| Tay-Sachs disease | HEXA |
| Sandhoff disease | HEXB |
| Chediak-Higashi syndrome | LYST |
| Aspartylglucosaminuria | AGA |
| Schindler disease | NAGA |
| Sialidosis | NEU1 |
| Combined SAP deficiency | PSAP |
| Marinesco-Sjogren syndrome | SIL1 |
| Sialic acid storage disorder | SLC17A5 |
| Niemann-Pick disease | NPC1, NPC2, SMPD1 |
| Metabolic conditions | |
| Peroxisome biogenesis disorders | |
| Including Zellweger syndrome, neonatal adrenoleukodystrophy and infantile Refsum disease | PEX1, PEX10, PEX11B, PEX12, PEX13, PEX16, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7 |
| Organic acidemias | |
| Argininosuccinic aciduria | ASL |
| 3-methylglutaconic aciduria | AUH, CLPB, DNAJC19, HTRA2, OPA3, SERAC1 |
| D-2-hydroxyglutaric aciduria | D2HGDH |
| Glutaric aciduria | GCDH |
| D-glyceric aciduria | GLYCTK |
| L-2-hydroxyglutaric aciduria | L2HGDH |
| Methylmalonic aciduria | MMADHC, MMUT |
| Methylmalonic aciduria and homocystinuria | LMBRD1, MMACHC, MMADHC |
| Alpha-methylacetoacetic aciduria | ACAT1 |
| Methylmalonic aciduria, vitamin B12-responsive | MMAA, MMAB |
| Mevalonic aciduria | MVK |
| Combined D-2- and L-2-hydroxyglutaric aciduria | SLC25A1 |
| Isovaleric acidemia | IVD |
| Glutaric acidemia | ETFA, ETFB, ETFDH |
| Other metabolic conditions | |
| Adenylosuccinase deficiency | ADSL |
| Arts syndrome | PRPS1 |
| Chanarin-Dorfman syndrome | ABHD5 |
| Galactosemia | GALT |



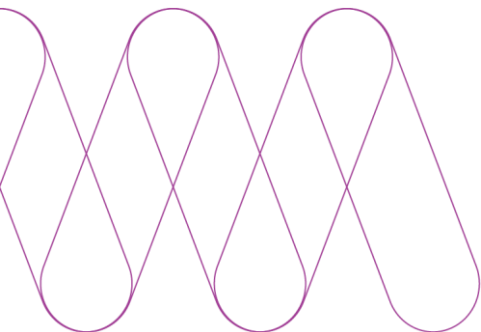
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| Glycogen storage disease | AGL, G6PC, GYS2, GBE1, LDHA, PFKM, SLC37A4 |
| GABA-transaminase deficiency | ABAT |
| Fanconi-Bickel syndrome | SLC2A2 |
| Hyperinsulinemic hypoglycemia | ABCC8, HADH, KCNJ11 |
| Hyperoxaluria | AGXT |
| Hypermanganesemia with dystonia | SLC39A14 |
| Succinic semialdehyde dehydrogenase deficiency | ALDH5A1 |
| Fructose intolerance | ALDOB |
| Congenital disorders of glycosylation | ALG1, ALG11, ALG12, ALG3, ALG6, ALG8, ALG9, CCDC115, COG6, COG7, DOLK, DPAGT1, MGAT2, MPI, PGM1, PMM2, RFT1, SLC39A8, SSR4, SRD5A3, TMEM165 |
| Congenital disorder of deglycosylation | NGLY1 |
| Glycine encephalopathy | AMT, GLDC |
| Glycosylphosphatidylinositol biosynthesis defect | GPAA1 |
| Argininemia | ARG1 |
| Asparagine synthetase deficiency | ASNS |
| Canavan disease | ASPA |
| Citrullinemia | ASS1, SLC25A13 |
| Chylomicron retention disease | SAR1B |
| Menkes disease and occipital horn syndrome | ATP7A |
| Maple syrup urine disease | BCKDHA, BCKDHB, DBT |
| Branched-chain ketoacid dehydrogenase kinase deficiency | BCKDK |
| GRACILE syndrome | BCS1L |
| Homocystinuria | MMADHC, MTHFR, MTR, MTRR |
| Lysinuric protein intolerance | SLC7A7 |
| Proteinuria | CLCN5 |
| Prolidase deficiency | PEPD |
| Hypomagnesemia | CLDN19, SLC30A10, TRPM6 |
| Coenzyme Q10 deficiency | COQ2, COQ4, COQ6, COQ8A |
| Carbamoylphosphate synthetase I deficiency | CPS1 |
| CPT 2 deficiency | CPT1A, CPT2 |
| Methemoglobinemia | CYB5R3 |
| Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration | TANGO2 |



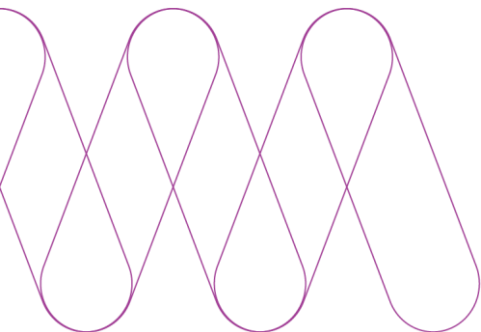
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| Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency | FLAD1 |
| Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency | ACADM |
| Peroxisomal acyl-CoA oxidase deficiency | ACOX1 |
| 17-alpha-hydroxylase deficiency | CYP17A1 |
| 17,20-lyase deficiency | CYP17A1 |
| Cerebrotendinous xanthomatosis | CYP27A1 |
| Aromatic L-amino acid decarboxylase deficiency | DDC |
| Dihydrolipoamide dehydrogenase deficiency | DLD |
| Wolcott-Rallison syndrome | EIF2AK3 |
| Hypophosphatemic rickets | ENPP1 |
| Hyperphosphatasia with intellectual disability syndrome | PIGV, PIGO, PGAP2, PGAP3 |
| Ethylmalonic encephalopathy | ETHE1 |
| Tyrosinemia | FAH, HPD |
| Fructose-1,6-bisphosphatase deficiency | FBP1 |
| Fumarase deficiency | FH |
| Glutamate formiminotransferase deficiency | FTCD |
| Cerebral creatine deficiency syndrome | GAMT, GATM, SLC6A8 |
| Gaucher disease | GBA, PSAP |
| Glycerol kinase deficiency | GK |
| Molybdenum cofactor deficiency | GPHN, MOCS1, MOCS2 |
| Glutathione synthetase deficiency | GSS |
| 3-hydroxyacyl-CoA dehydrogenase deficiency | HADH |
| LCHAD deficiency | HADHA |
| Trifunctional protein deficiency | HADHA, HADHB |
| Hemochromatosis | HAMP, HJV |
| 3-hydroxyisobutryl-CoA hydrolase deficiency | HIBCH |
| Holocarboxylase synthetase deficiency | HLCS |
| HMG-CoA lyase deficiency | HMGCL |
| HMG-CoA synthase-2 deficiency | HMGCS2 |
| Lesch-Nyhan syndrome | HPRT1 |
| D-bifunctional protein deficiency | HSD17B4 |
| Leprechaunism | INSR |
| Norum disease | LCAT |
| Lactate dehydrogenase-B deficiency | LDHB |
| Familial hypercholesterolemia | LDLR, LDLRAP1 |



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| Pyruvate dehydrogenase lipoic acid synthetase deficiency | LIAS |
| Cholesteryl ester storage disease | LIPA |
| Wolman disease | LIPA |
| Lipoyltransferase 1 deficiency | LIPT1 |
| Lipoprotein lipase deficiency | LPL |
| Malonyl-CoA decarboxylase deficiency | MLYCD |
| Abetalipoproteinemia | MTTP |
| N-acetylglutamate synthase deficiency | NAGS |
| N-terminal acetyltransferase deficiency | NAA10 |
| Ornithine transcarbamylase deficiency | OTC |
| Phenylketonuria (PKU) | PAH |
| Pyruvate carboxylase deficiency | PC |
| Hyperphenylalaninemia | PTS, QDPR, DNAJC12 |
| Propionicacidemia | PCCA, PCCB |
| Proprotein convertase 1 deficiency | PCSK1 |
| Pyruvate dehydrogenase deficiency | PDHA1, PDHB, PDP1 |
| Phosphoglycerate kinase 1 deficiency | PGK1 |
| Phosphoglycerate dehydrogenase deficiency | PHGDH |
| Refsum disease | PHYH |
| Pyruvate kinase deficiency | PKLR |
| Plasminogen deficiency | PLG |
| Dysplasminogenemia | PLG |
| Pyridoxamine 5'-phosphate oxidase deficiency | PNPO |
| Phosphoribosylpyrophosphate synthetase superactivity | PRPS1 |
| Phosphoserine phosphatase deficiency | PSPH |
| Neu-Laxova syndrome | PHGDH, PSAT1 |
| Riboflavin transport deficiency syndrome | SLC52A2, SLC52A3 |
| Tumoral calcinosis, normophosphatemic | SAMD9 |
| Lathosterolosis | SC5D |
| Emphysema-cirrhosis, due to AAT deficiency | SERPINA1 |
| Hemorrhagic diathesis due to antithrombin Pittsburgh | SERPINA1 |
| Monocarboxylate transporter 1 deficiency | SLC16A1 |
| Thiamine metabolism dysfunction syndrome | SLC19A2, SLC19A3, SLC25A19, TPK1 |
| Carnitine deficiency | SLC22A5 |
| Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome | SLC25A15 |
| Acrodermatitis enteropathica | SLC39A4 |



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| Multiple sulfatase deficiency | SUMF1 |
| Salla disease | SLC17A5 |
| Sjogren-Larsson syndrome | ALDH3A2 |
| Sulfite oxidase deficiency | SUOX |
| Transaldolase deficiency | TALDO1 |
| Barth syndrome | TAZ |
| Adrenocorticotrophic hormone deficiency | TBX19 |
| Transcobalamin II deficiency | TCN2 |
| Hemolytic anaemia due to triosephosphate isomerase deficiency | TPI1 |
| Crigler-Najjar syndrome | UGT1A1 |
| Orotic aciduria | UMPS |
| VLCAD deficiency | ACADVL |
| Wilson disease | ATP7B |
| Endocrine conditions | |
| Congenital adrenal hyperplasia* | |
| Severe salt wasting type | CYP11A1, CYP11B2, NR0B1, POU1F1, PROP1, HSD3B2 |
| Lipoid type | STAR |
| <i>*Excludes 21-hydroxylase deficiency, as the CYP21A2 gene is not screened for technical reasons</i> | |
| Diabetes mellitus | |
| Neonatal, with congenital hypothyroidism | GLIS3 |
| Insulin-resistant, with acanthosis nigricans | INSR |
| Other endocrine conditions | |
| Disordered steroidogenesis due to cytochrome P450 oxidoreductase | POR |
| Glucocorticoid deficiency | MC2R, MRAP, NNT |
| Growth hormone deficiency with pituitary anomalies | HESX1 |
| Hyperparathyroidism, neonatal severe | CASR |
| Hypothyroidism, congenital | TSHB |
| Insulin-like growth factor resistance | IGF1R |
| Laron syndrome | GHR |
| Obesity, morbid, due to leptin deficiency | LEP |
| Pituitary hormone deficiency | HESX1, LHX3 |
| Proopiomelanocortin (POMC) deficiency | POMC |
| Rabson-Mendenhall syndrome | INSR |



Neurological conditions

White matter disorders

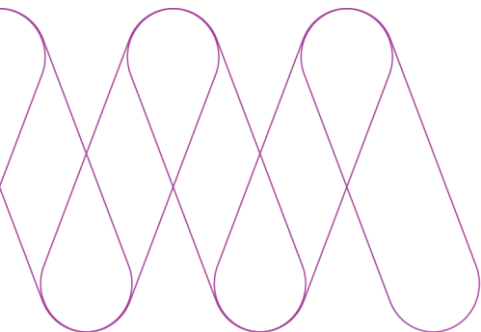
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| Adrenoleukodystrophy | ABCD1 |
| Aicardi-Goutieres syndrome | ADAR, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1 |
| Leukodystrophy, hypomyelinating | AIMP1, FAM126A, GJC2, HSPD1, POLR3A, POLR3B, PYCR2, RARS1, UFM1, VPS11 |
| Leukoencephalopathy with ataxia | CLCN2 |
| Leukoencephalopathy with vanishing white matter | EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5 |
| Leukoencephalopathy, cystic, without megalencephaly | RNASET2 |
| Megalencephalic leukoencephalopathy with subcortical cysts | HEPACAM, MLC1 |
| Hypomyelination with brainstem and spinal cord involvement and leg spasticity (HBSL) | DARS1 |
| Pelizaeus-Merzbacher disease | PLP1 |

Congenital brain malformations

| | |
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| Pontocerebellar hypoplasia | AMPD2, CLP1, EXOSC3, EXOSC8, RARS2, SEPSECS, TBC1D23, TOE1, TSEN2, TSEN54, VPS53, VRK1 |
| Lissencephaly | ARX, KATNB1, LAMB1, NDE1, DCX, TMTC3 |
| Joubert syndrome | AHI1, ARL13B, CC2D2A, CEP290, CEP41, CPLANE1, CSPP1, INPP5E, KIF7, NPHP1, OFD1, RPGRIP1L, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67 |
| Polymicrogyria | ADGRG1, RTTN |
| Septo-optic dysplasia | HESX1 |
| Band heterotopia | DCX, EML1 |
| Band-like calcification with simplified gyration and polymicrogyria | OCLN |
| Cerebellar hypoplasia and intellectual disability with or without quadrupedal locomotion | VLDLR |
| Periventricular heterotopia with microcephaly | ARFGF2 |
| Poretti-Boltshauser syndrome | LAMA1 |
| Cortical malformations, occipital | LAMC3 |

Microcephaly

| | |
|----------|--|
| Isolated | ASPM, CDK5RAP2, CENPJ, CEP152, CIT, KIF14, KNL1, MCPH1, MFSD2A, MED17, PNKP, SLC25A19, STIL, WDR62, ZNF335 |
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Hydrocephalus

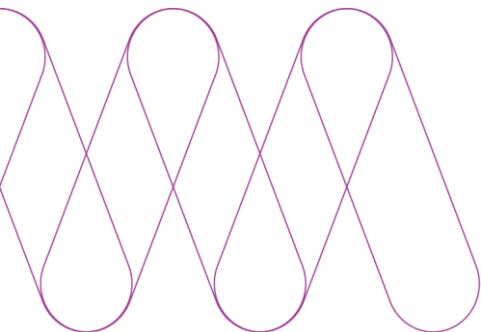
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| Non-syndromic hydrocephalus | L1CAM, CCDC88C, MPDZ |
| Hydrocephalus with congenital idiopathic intestinal pseudoobstruction | L1CAM |
| Hydrocephalus due to aqueductal stenosis | L1CAM |
| Hydrocephalus with Hirschsprung disease | L1CAM |

Neurodegenerative conditions

| | |
|--|---|
| Neuronal ceroid lipofuscinoses | CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, MFSD8, PPT1, TPP1 |
| Parkinson disease, juvenile-onset | DNAJC6, FBXO7, PLA2G6, ATP13A2 |
| Encephalopathy, progressive | BSCL2, TBCD, NAXE |
| Moyamoya disease | GUCY1A1 |
| Neurodegeneration with brain iron accumulation | C19orf12, PANK2, PLA2G6 |
| Neurodegeneration due to cerebral folate transport deficiency | FOLR1 |
| Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset | SQSTM1 |
| Neurodegeneration, stress-induced, with variable ataxia and seizures | ADPRS |
| Infantile or childhood-onset striatonigral degeneration | NUP62, VAC14 |
| PEHO syndrome | ZNHIT3 |
| Infantile cerebellar-retinal degeneration | ACO2 |
| Infantile neuroaxonal dystrophy 1 | PLA2G6 |
| Spastic tetraplegia, thin corpus callosum, and progressive microcephaly | SLC1A4 |
| Troyer syndrome | SPART |

Ataxias

| | |
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| Ataxia-telangiectasia | ATM, MRE11 |
| Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia | APTX |
| Ataxia, cerebellar, Cayman type | ATCAY |
| Ataxia, posterior column, with retinitis pigmentosa | FLVCR1 |
| Ataxia-oculomotor apraxia 4 | PNKP |
| Ataxia with isolated vitamin E deficiency | TTPA |
| Cerebellar ataxia, cognitive disability, and disequilibrium (CAMRQ) | WDR81, ATP8A2 |
| Spastic ataxia | KIF1C, MARS2, NKX6-2, SACS |
| Spinocerebellar ataxia | GRM1, PMPCA, SETX, SNX14, STUB1, SCYL1, TPP1, WWOX |



Movement disorders

| | |
|--|---------------|
| Choreoacanthocytosis | VPS13A |
| Dystonia | COL6A3, PRKRA |
| Dystonia, dopa-responsive, due to sepiapterin reductase deficiency | SPR |
| Dystonia, DOPA-responsive, with or without hyperphenylalaninemia | GCH1 |
| Parkinsonism-dystonia, infantile | SLC6A3 |
| Segawa syndrome | TH |

Epilepsy

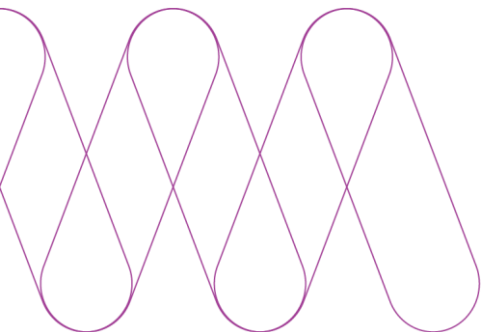
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| Epilepsy, pyridoxine-dependent | ALDH7A1 |
| Epileptic encephalopathy, early infantile | AP3B2, ARV1, ARX, ARHGEF9, DENND5A, FRRS1L, MECP2, SLC13A5, SLC12A5, SLC25A22, TBC1D24, UBA5, WWOX |
| Epilepsy, progressive myoclonic | CSTB, EPM2A, GOSR2, KCTD7, NHLRC1, PRICKLE1, SCARB2, TBC1D24 |
| Hyperekplexia | ATAD1, SLC6A5 |
| Epilepsy, early-onset, vitamin B6-dependent | PLPBP |
| Epilepsy, X-linked, with variable learning disabilities and behaviour disorders | SYN1 |
| Epilepsy, hearing loss, and intellectual disability syndrome | SPATA5 |
| Cortical dysplasia-focal epilepsy syndrome | CNTNAP2 |
| Amish infantile epilepsy syndrome | ST3GAL5 |

Intellectual disability

| | |
|---|--|
| Non-syndromic intellectual disability, X-linked | AP1S2, ARX, ATRX, BRWD3, CASK, CLCN4, CUL4B, DLG3, FTSJ1, GDI1, HCFC1, IL1RAPL1, IQSEC2, MECP2, NEXMIF, NLGN4X, PAK3, RAB39B, RLIM, SLC16A2, SYP, THOC2, TSPAN7, USP9X, ZNF711 |
| Non-syndromic intellectual disability, autosomal recessive | ADAT3, CC2D1A, ELP2, GPT2, HERC2, KPTN, LINS1, MAN1B1, MBOAT7, MED23, METTL23, NSUN2, PGAP1, PIGG, TRAPPC9, TTI2, TUSC3 |
| Intellectual developmental disorder with microcephaly and short stature | PUS7 |

Other neurological conditions

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|---|------|
| Sensorineural hearing loss, premature ovarian failure (females), variable intellectual disability, spasticity, ataxia | CLPP |
|---|------|



Cutaneous conditions

Ichthyosis

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| Ichthyosis, congenital, autosomal recessive | ABCA12, ALOX12B, ALOXE3, CERS3, CYP4F22, NIPAL4, TGM1 |
| Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis | CLDN1 |
| Epidermolytic hyperkeratosis | KRT10 |

Cutis laxa

| | |
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| Cutis laxa, autosomal recessive | ALDH18A1, ATP6V0A2, EFEMP2, FBLN5, LTBP4, PYCR1 |
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Ectodermal dysplasia

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| Ectodermal dysplasia, ectrodactyly and macular dystrophy | CDH3 |
| Ectodermal dysplasia | EDA, EDAR, IKBKG, KRT85 |

Cutaneous conditions affecting the nervous system

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| Xeroderma pigmentosum | ERCC2, ERCC4, ERCC5, XPA, XPC |
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Other cutaneous conditions

| | |
|--|--|
| Kindler syndrome | FERMT1 |
| Epidermolysis bullosa | COL7A1, COL17A1, DSP, ITGA6, ITGB4, KRT14, KRT5, LAMA3, LAMB3, LAMC2, PLEC |
| Hyaline fibromatosis syndrome | ANTXR2 |
| Porokeratosis 3, disseminated superficial actinic | MVK |
| Keratosis linearis with ichthyosis congenital and sclerosing keratoderma | POMP |
| Netherton syndrome | SPINK5 |
| Poikilderma with neutropenia | USB1 |
| Restrictive dermopathy, lethal | LMNA, ZMPSTE24 |
| Trichothiodystrophy | ERCC2, GTF2H5, MPLKIP |
| Transient bullous of the newborn | COL7A1 |

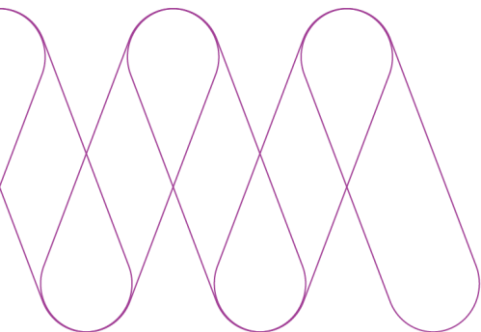
Respiratory conditions

Surfactant conditions

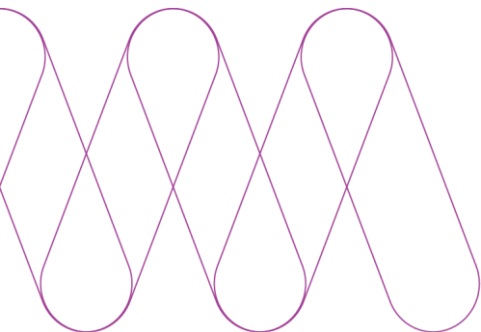
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| Surfactant metabolism dysfunction, pulmonary | ABCA3, SFTPB |
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Ciliary dyskinesia

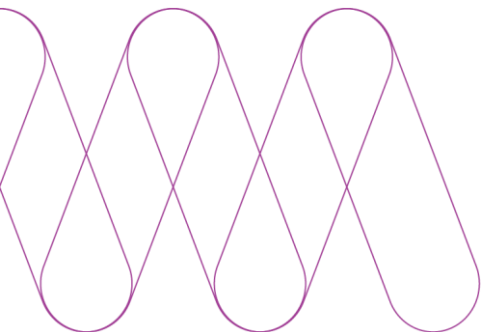
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| Ciliary dyskinesia, primary | OCAD2*, CCDC103, CCDC114, CCDC39, CCDC40, CCNO, DNAAF1, DNAAF3, DNAAF4, DNAAF5, DNAAF6 [^] , GAS8, |
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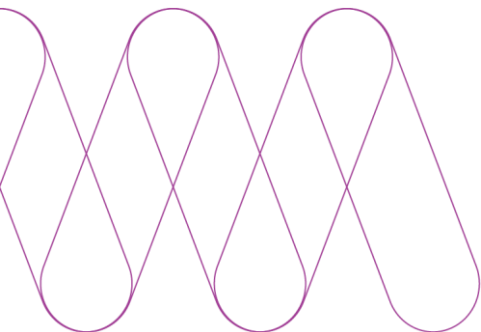
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| | HYDIN, LRRC6, RSPH1, RSPH4A, RSPH9, SPAG1, ZMYND10 <i>*Formerly known as ARMC4</i> <i>^Formerly known as PIH1D3</i> |
| Ciliary dyskinesia, primary, with or without situs inversus | DNAH11, DNAH5, DNAI1, DNAI2 |
| Other respiratory conditions | |
| Cystic fibrosis | CFTR |
| Pulmonary veno-occlusive disease | EIF2AK4 |
| Interstitial lung and liver disease | MARS1 |
| Immunological conditions | |
| Chronic granulomatous disease | |
| Deficiency of NCF-1 | NCF1 |
| Deficiency of NCF-2 | NCF2 |
| Deficiency of CYBA | CYBA |
| X-linked | CYBB |
| Combined cellular and humoral immune defects with granulomas | RAG1, RAG2 |
| Complement deficiencies | |
| C1q | C1QA, C1QB, C1QC |
| C3 | C3 |
| C5 | C5 |
| C6 | C6 |
| C7 | C7 |
| C8 | C8B |
| Factor D | CFD |
| Factor H | CFH |
| Factor I | CFI |
| Immunodeficiencies | |
| Immunodeficiency | ATP6AP1, CARD11, CD3D, CTPS1, DOCK2, ICOS, IKBKB, IL12RB1, IL17RA, LAT, LRBA, MALT1, ORAI1, PGM3, RORC, STIM1, TYK2 |
| Mycobacteriosis | CYBB, IFNGR1, IFNGR2, STAT1 |
| Purine nucleoside phosphorylase deficiency | PNP |
| Hyper-IgM | CD40, CD40LG |
| Hyper-IgD syndrome | MVK |
| Hyper-IgE recurrent infection syndrome | DOCK8 |



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| Centromeric instability-facial anomalies syndrome | DNMT3B, ZBTB24 |
| Combined immunodeficiency, moderate | IL2RG |
| Combined immunodeficiency and megaloblastic anaemia with or without hyperhomocysteinemia | MTHFD1 |
| Neutropenia | |
| Severe, congenital | G6PC3, HAX1, JAGN1, VPS45, WAS |
| Severe combined immunodeficiencies | |
| Severe combined immunodeficiency | IL2RG |
| Adenosine deaminase deficiency | ADA |
| With microcephaly, growth retardation, and sensitivity to ionizing radiation | NHEJ1 |
| Athabaskan type | DCLRE1C |
| B cell-negative | RAG1, RAG2 |
| T-cell negative, B-cell/natural killer cell-positive type | IL7R, JAK3 |
| Reticular dysgenesis | AK2 |
| Other immunological conditions | |
| Agammaglobulinemia | BTK, IGHM |
| Autoimmune disease, multisystem, with facial dysmorphism | ITCH |
| Autoinflammation, lipodystrophy, and dermatosis syndrome | PSMB8 |
| Bone marrow failure syndrome | ERCC6L2, DNAJC21 |
| Bare lymphocyte syndrome | CIITA, RFXAP, TAP1 |
| Candidiasis, familial | CARD9 |
| Histiocytosis-lymphadenopathy plus syndrome | SLC29A3 |
| Hemophagocytic lymphohistiocytosis | PRF1, STX11, STXBP2, UNC13D |
| Hepatic veno-occlusive disease with immunodeficiency | SP110 |
| Interleukin 1 receptor antagonist deficiency | IL1RN |
| Immunodysregulation, polyendocrinopathy, and enteropathy | FOXP3 |
| Leukocyte adhesion deficiency | FERMT3, ITGB2 |
| Lymphoproliferative syndrome | CD27, ITK, SH2D1A, XIAP |
| MHC class II deficiency, complementation group B | RFXANK |
| Natural killer cell and glucocorticoid deficiency with DNA repair defect | MCM4 |
| Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease | ARPC1B |
| Properdin deficiency | CFP |
| Pyogenic bacterial infections, recurrent, due to MYD88 deficiency | MYD88 |



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| Selective T-cell defect | ZAP70 |
| T-cell immunodeficiency, congenital alopecia, and nail dystrophy | FOXN1 |
| Darsun syndrome | G6PC3 |
| Majeed syndrome | LPIN2 |
| Omenn syndrome | DCLRE1C, RAG1, RAG2 |
| Wiskott-Aldrich syndrome | WAS |
| Gastrointestinal conditions | |
| Severe congenital diarrhea | |
| With tufting enteropathy, congenital | EPCAM |
| Secretory chloride, congenital | SLC26A3 |
| Secretory sodium, congenital, | SPINT2, SLC9A3 |
| Protein-losing enteropathy type | DGAT1 |
| Hepatic conditions | |
| Cholestasis, progressive familial intrahepatic | ABCB11, ABCB4, ATP8B1, TJP2 |
| Hepatic lipase deficiency | LIPC |
| Porphyria | ALAD, UROS |
| Liver failure, transient infantile | TRMU |
| Hypercholanaemia | TJP2 |
| Other gastrointestinal conditions | |
| Microvillus inclusion disease | MYO5B |
| Bile acid synthesis defect, congenital | AKR1D1, CYP7B1, HSD3B7 |
| Congenital short bowel syndrome | CLMP, FLNA |
| Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy | CD55 |
| Meconium ileus | GUCY2C |
| Mitchell-Riley syndrome | RFX6 |
| Chronic atrial and intestinal dysrhythmia | SGO1 |
| Inflammatory bowel disease, congenital, severe | IL10RA, IL10RB |
| Trichohepatoenteric syndrome | SKIV2L, TTC37 |
| Folate malabsorption, hereditary | SLC46A1 |
| Gastrointestinal defects and immunodeficiency syndrome | TTC7A |
| Hyperbilirubinemia, familial transient neonatal | UGT1A1 |
| Haematological conditions | |
| Anaemia | |
| Sideroblastic, with ataxia | ABCB7 |



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| Anaemia, sideroblastic, pyridoxine-refractory | SLC25A38 |
| Dyserythropoietic anaemia | SEC23B |
| Haemolytic anaemia due to hexokinase deficiency | HK1 |
| Fanconi anaemia | ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, UBE2T |

Clotting conditions

| | |
|---|---------------|
| Hypoprothrombinemia | F2 |
| Factor V deficiency | F5 |
| Factor VII deficiency | F7 |
| Haemophilia A | F8 |
| Haemophilia B | F9 |
| Afibrinogenemia Dysfibrinogenemia Hypodysfibrinogenemia Hypofibrinogenemia | FGA, FGB, FGG |
| Combined factor V and VIII deficiency | LMAN1, MCFD2 |
| Thrombotic thrombocytopenic purpura | ADAMTS13 |
| Thrombocytopenia, congenital amegakaryocytic | MPL |
| Thrombophilia | PROC, PROS1 |
| von Willebrand disease | VWF |
| Thrombocytopenia, X-linked | WAS |

Other haematological conditions

| | |
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| Vitamin K-dependent clotting factors, combined deficiency of | VKORC1 |
| Beta thalassemia | HBB |
| Sickle cell disease | HBB |
| Atransferrinemia | TF |

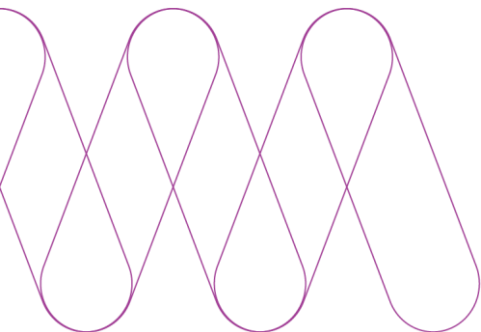
Cardiovascular conditions

Arrhythmias

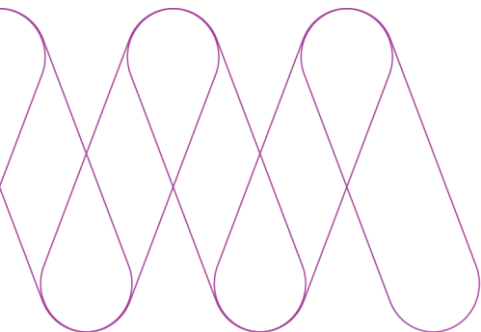
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| Ventricular tachycardia, catecholaminergic polymorphic | CASQ2 |
| Jervell and Lange-Nielsen syndrome | KCNQ1 |
| Ventricular tachycardia, catecholaminergic polymorphic with or without muscle weakness | TRDN |

Cardiomyopathies

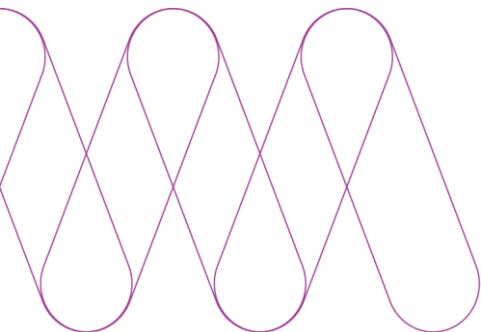
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| Cardiomyopathy, dilated, with woolly hair and keratoderma (Naxos disease) | DSP, JUP |
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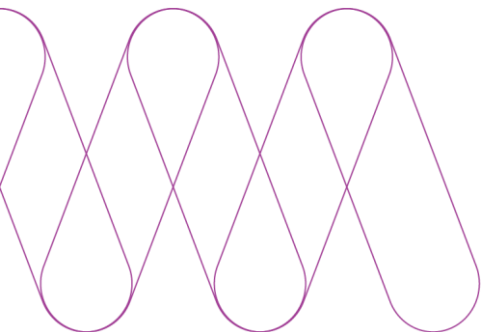
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| Dilated cardiomyopathy | FKTN |
| Structural cardiovascular conditions | |
| Arterial calcification of infancy | ENPP1 |
| Cardiac valvular dysplasia, X-linked | FLNA |
| Right atrial isomerism | GDF1 |
| Hypoplastic left heart syndrome | GJA1 |
| Arterial tortuosity syndrome | SLC2A10 |
| Heterotaxy, visceral | ZIC3, MMP21 |
| Congenital heart defects | ZIC3 |
| Other cardiovascular conditions | |
| Sudden cardiac failure, infantile | PPA2 |
| Renal conditions | |
| Syndromic renal conditions | |
| Alport syndrome | COL4A3, COL4A4, COL4A5 |
| Dent disease | OCRL, CLCN5 |
| Renal tubular acidosis with other abnormalities | ATP6V1B1, SLC4A4, SLC4A1 |
| Bartter syndrome | BSND, CLCNKB, KCNJ1, SLC12A1 |
| Renal-hepatic-pancreatic dysplasia | NPHP3, NEK8 |
| Polycystic kidney and hepatic disease | PKHD1 |
| Nephrotic syndrome | COQ8B, DGKE, LAMB2, NPHS1, NPHS2, NUP107, NUP93, PLCE1, SGPL1 |
| Tubular conditions | |
| Renal tubular dysgenesis | ACE, AGT, REN |
| Renal tubular acidosis | ATP6V0A4 |
| Other renal conditions | |
| Focal segmental glomerulosclerosis | CRB2 |
| Pseudohypoaldosteronism | SCNN1A, SCNN1B |
| Nephronophthisis and related conditions | ANKS6, DCDC2, INVS, MAPKBP1, NPHP1, NPHP3, NPHP4, TMEM67, TTC21B, WDR19 |
| Nephrogenic diabetes insipidus | AQP2 |
| Neuromuscular conditions | |
| Atrophy | |
| Spinal muscular atrophy with progressive myoclonic epilepsy | SAH1 |
| Spinal muscular atrophy | SMN1, UBA1 |



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| Spinal muscular atrophy with congenital bone fractures | ASCC1 |
| Arthrogryposis | |
| Arthrogryposis, distal | ECEL1, PIEZO2 |
| Arthrogryposis lethal with anterior horn cell disease | GLE1 |
| Arthrogryposis, renal dysfunction, and cholestasis | VIPAS39, VPS33B |
| Arthrogryposis multiplex congenita | LG14 |
| Dystrophy | |
| Limb-girdle muscular dystrophy | CAPN3, DYSF, PLEC, SGCA, SGCB, SGCD, SGCG, TCAP, TRAPPC11, TRIM32, TTN |
| Muscular dystrophy-dystroglycanopathy | B3GALNT2, CRPPA, FKR1, FKTN, GMPPB, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1 |
| Muscular dystrophy, congenital | CHKB, LAMA2 |
| Ullrich congenital muscular dystrophy | COL6A1, COL6A2, COL6A3 |
| Duchenne muscular dystrophy | DMD <i>*In VIC and NSW, most DMD carriers are unable to be detected due to limitations in testing technology</i> |
| Becker muscular dystrophy | DMD <i>*As above</i> |
| Emery-Dreifuss muscular dystrophy | EMD, FHL1, LMNA |
| Muscular dystrophy, rigid spine | SELENON |
| Myopathy | |
| Myopathy, congenital | ACTA1 |
| Nemaline myopathy | ACTA1, CFL2, KLHL40, KLHL41, LMOD3, NEB, TNNT1, TPM3 |
| Myopathy, centronuclear, autosomal recessive | BIN1, SPEG |
| Distal myopathy | DYSF |
| Myopathy with extrapyramidal signs | MICU1 |
| Myopathy, X-linked | FHL1 |
| Myopathy, X-linked, with excessive autophagy | VMA21 |
| Inclusion body myopathy | GNE |
| Myopathy, areflexia, respiratory distress, and dysphagia, early-onset | MEGF10 |
| Myotubular myopathy, X-linked | MTM1 |
| Minicore myopathy | RYR1 |
| Myopathy, myofibrillar | KY, PYROXD1 |
| Central core disease | RYR1 |



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| Myopathy, early-onset, with fatal cardiomyopathy | TTN |
| CAP myopathy | TPM3 |
| Myasthenia | |
| Myasthenic syndrome | AGRN, ALG2, CHAT, CHRNA1, CHRND, CHRNE, COLQ, DOK7, DPAGT1, GFPT1, IGHMBP2, MUSK, RAPSN, SLC5A7 |
| Neuropathy | |
| Charcot-Marie-Tooth disease | FGD4, FIG4, GDAP1, LMNA, MFN2, MPZ, MTMR2, NDRG1, PRPS1, PRX, SBF2, SH3TC2 |
| Dysautonomia, familial | ELP1 |
| Insensitivity to pain, congenital | SCN9A, NTRK1 |
| Neuromyotonia and axonal neuropathy | HINT1 |
| Neuropathy, hereditary motor and sensory | HK1, IGHMBP2, KIF1A, SLC25A46 |
| Neuropathy, hereditary sensory and autonomic | NGF, PRDM12, RETREG1, WNK1 |
| Giant axonal neuropathy | GAN |
| Rhabdomyolysis | |
| Myoglobinuria, acute recurrent | LPIN1 |
| Spasticity | |
| Spastic paralysis, infantile onset ascending | ALS2 |
| Juvenile primary lateral sclerosis | ALS2 |
| Spastic paraplegia | AP4M1, AP4B1, AP4S1, ATP13A2, ALDH18A1, B4GALNT1, CYP2U1, CYP7B1, DDHD2, DSTYK, FA2H, FARS2, GBA2, GJC2, KIF1A, NT5C2, PLP1, PNPLA6, SPG11, VPS37A, ZFYVE26 |
| Connective tissue conditions | |
| Ehlers-Danlos syndrome (EDS) | |
| Ehlers-Danlos syndrome, progeroid type | ADAMTS2, B3GALT6, B4GALT7, PLOD1 |
| Ehlers-Danlos syndrome, musculocontractural type | CHST14 |
| Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss | FKBP14 |
| Vascular conditions | |
| Polyarteritis nodosa, childhood-onset | ADA2 |
| Meester-Loeys syndrome | BGN |



Ocular conditions

Albinism

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| Hermansky-Pudlak syndrome | HPS1, HPS3, HPS4, HPS5, HPS6 |
| Oculocutaneous albinism | GPR143, LRMDA, OCA2, SLC24A5, SLC45A2, TYR, TYRP1 |

Dystrophies

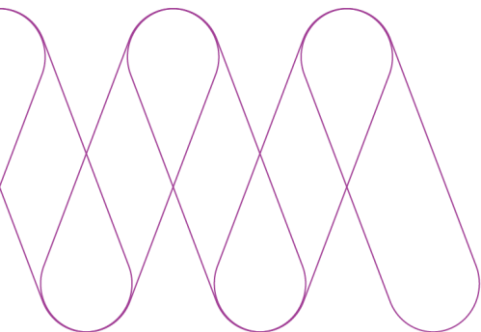
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| Retinal dystrophy, early-onset severe | LRAT, RCBTB1, CFAP410 |
| Macular dystrophy with central cone involvement | MFSD8 |
| Cone-rod dystrophy | AIPL1, C8orf37, CEP78, CNGB3, KCNV2, PDE6C, RPGRIP1, SEMA4A |

Microphthalmia

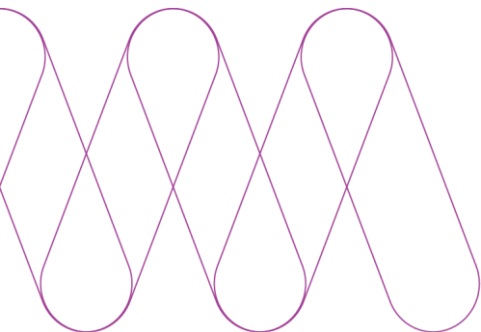
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| Isolated | ALDH1A3, RAX, VSX2 |
| With coloboma | STRA6, VSX2 |
| Syndromic | STRA6, RARB |

Other ocular conditions

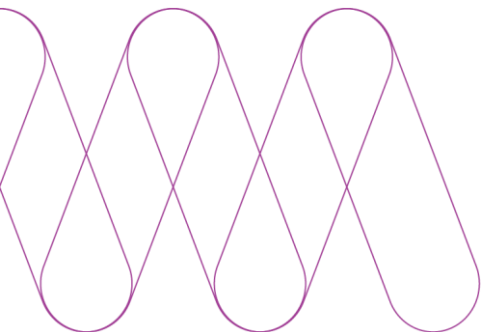
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| Achromatopsia | ATF6, CNGA3, CNGB3, GNAT2 |
| Aphakia | FOXE3 |
| Congenital cataracts | AGK, FYCO1, NHS, TDRD7 |
| Cone-rod synaptic disorder, congenital non-progressive | CABP4 |
| Choroideremia | CHM |
| Congenital stationary night blindness | GPR179, NYX |
| Persistent hyperplastic primary vitreous | ATOH7 |
| Macular degeneration (congenital) | CNGB3, RPGR |
| Leber congenital amaurosis | AIPL1, CEP290, CRB1, GUCY2D, LCA5, LRAT, NMNAT1, RD3, RDH12, RPE65, RPGRIP1, SPATA7, TULP1 |
| Glaucoma (congenital) | CYP1B1 |
| Peters anomaly | CYP1B1 |
| Retinal arterial macroaneurysm with supra-valvular pulmonic stenosis | IGFBP7 |
| Retinitis pigmentosa | AGBL5, AIPL1, C8orf37, CRB1, DHDDS, IFT172, LRAT, MERTK, REEP6, RP2, SEMA4A, SPATA7, TULP1, USH2A |
| Progressive external ophthalmoplegia | POLG |
| Brittle cornea syndrome | PRDM5 |
| Corneal opacification and other ocular anomalies | PXDN |
| Gaze palsy, horizontal, with progressive scoliosis | ROBO3 |



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| Foveal hypoplasia, with or without optic nerve misrouting and/or anterior segment dysgenesis | SLC38A8 |
| Optic atrophy | TMEM126A |
| Skeletal conditions | |
| Dysplasias | |
| Spondyloepiphyseal dysplasia with other abnormalities | CHST3, CCN6 |
| Anauxetic dysplasia | POP1, RMRP |
| Spondyloepimetaphyseal dysplasia | B3GALT6, NANS |
| Desbuquois dysplasia | CANT1, XYLT1 |
| Schneckenbecken dysplasia | SLC35D1 |
| Short-rib thoracic dysplasia with or without polydactyly | CEP120, DYNC2H1, DYNC2I1*, DYNC2I2^, DYNC2LI1, KIAA0586, TTC21B, WDR35, IFT140, IFT172, IFT80, NEK1 <i>*Formerly known as WDR60</i> <i>^Formerly known as WDR34</i> |
| Spondylometaphyseal dysplasia, short limb-hand type | DDR2 |
| Spondylo-megaepiphyseal-metaphyseal dysplasia | NKX3-2 |
| Chondrodysplasia, Grebe type | GDF5 |
| Oculodentodigital dysplasia | GJA1 |
| Smith-McCort dysplasia | DYM, RAB33B |
| Omodysplasia | GPC6 |
| Dyssegmental dysplasia, Silverman-Handmaker type | HSPG2 |
| Cranioectodermal dysplasia | IFT122 |
| Opsismodysplasia | INPPL1 |
| Otospondylomegaepiphyseal dysplasia | COL11A2 |
| Greenberg skeletal dysplasia | LBR |
| Cleft lip/palate-ectodermal dysplasia syndrome | NECTIN1 |
| Spondylometaphyseal dysplasia with additional abnormalities | PCYT1A, CFAP410 |
| Chondrodysplasia, Blomstrand type | PTH1R |
| Metaphyseal dysplasia without hypotrichosis | RMRP |
| Cranioleptocrotaphic dysplasia | SEC23A |
| Langer mesomelic dysplasia | SHOX |
| De la Chapelle dysplasia | SLC26A2 |
| Diastrophic dysplasia | SLC26A2 |
| Craniofrontonasal dysplasia | EFNB1 |
| Chondrodysplasia punctata, rhizomelic | AGPS, GNPAT, PEX7 |
| Mandibuloacral dysplasia | LMNA |



| Acromesomelic dysplasia | |
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| Hunter-Thompson type | GDF5 |
| Maroteaux type | NPR2 |
| Demirhan type | BMPR1B |
| Arthropathies | |
| Arthropathy, progressive pseudorheumatoid | CCN6 |
| Cranioosteoarthropathy | HPGD |
| Hypertrophic osteoarthropathy | HPGD |
| Multicentric osteolysis, nodulosis, and arthropathy | MMP2 |
| Camptodactyly-arthropathy-coxa vara-pericarditis syndrome | PRG4 |
| Short stature and dwarfism | |
| Multiple joint dislocations, short stature, craniofacial dysmorphism, and congenital heart defects | B3GAT3 |
| Amelogenesis imperfecta and short stature | LTBP3 |
| Microcephalic osteodysplastic primordial dwarfism | PCNT, RNU4ATAC |
| Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis | POC1A |
| Short stature, optic nerve atrophy, and Pelger-Huet anomaly | NBAS |
| Mulibrey nanism | TRIM37 |
| Other skeletal conditions | |
| 3-M syndrome | CCDC8, OBSL1, CUL7 |
| Antley-Bixler syndrome | POR |
| Hypophosphatasia, infantile | ALPL |
| Diaphanospondylodysostosis | BMPER |
| Meier-Gorlin syndrome | CDT1, CDC45, ORC1, ORC6 |
| Osteopetrosis, infantile | CA2, CLCN7, OSTM1, TCIRG1, TNFRSF11A, TNFSF11 |
| Fibrochondrogenesis | COL11A1, COL11A2 |
| Osteogenesis imperfecta, recessive type | CRTAP, FKBP10, P3H1, PPIB, SERPINF1, WNT1 |
| Pycnodysostosis | CTSK |
| Spondylocostal dysostosis | DLL3, HES7, MESP2 |
| Ellis-van Creveld syndrome | EVC, EVC2 |
| Raine syndrome | FAM20C |
| Bruck syndrome | FKBP10, PLOD2 |
| Spondylocarpotarsal synostosis syndrome | FLNB |



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| Brachydactyly | GDF5 |
| Geroderma osteodysplasticum | GORAB |
| Craniosynostosis | IL11RA |
| Alazami syndrome | LARP7 |
| Schwartz-Jampel syndrome | HSPG2 |
| Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome | LIFR |
| Acheiropody | LMBR1 |
| Cenani-Lenz syndactyly syndrome | LRP4 |
| Sclerosteosis | LRP4, SOST |
| Osteoporosis-pseudoglioma syndrome | LRP5 |
| Orofacial cleft | NECTIN1 |
| Brachyolmia 4 with mild epiphyseal and metaphyseal change | PAPSS2 |
| Carpenter syndrome | RAB23, MEGF8 |
| Baller-Gerold syndrome | RECQL4 |
| RAPADILINO syndrome | RECQL4 |
| Cartilage-hair hypoplasia | RMRP |
| Robinow syndrome | ROR2 |
| Van den Ende-Gupta syndrome | SCARF2 |
| Frank-ter Haar syndrome | SH3PXD2B |
| Achondrogenesis | SLC26A2, TRIP11 |
| Atelosteogenesis | SLC26A2 |
| Van Buchem disease | SOST |
| Kenny-Caffey syndrome | TBCE |
| Paget disease of bone | TNFRSF11B |
| Ulna and fibula, absence of, with severe limb deficiency | WNT7A |
| Fuhrmann syndrome | WNT7A |
| CODAS syndrome | LONP1 |
| Keutel syndrome | MGP |
| Steel syndrome | COL27A1 |