# Leukodystrophy panel

| **versie** | V2 (265 genen) | Centrum voor Medische Genetica Gent |
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| Gene | OMIM gene ID | Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern |
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| *AARS2* | 612035 | Combined oxidative phosphorylation deficiency 8, 614096 (3), Autosomal recessive; Leukoencephalopathy, progressive, with ovarian failure, 615889 (3), Autosomal recessive |
| *ABAT* | 137150 | GABA-transaminase deficiency, 613163 (3), Autosomal recessive |
| *ABCD1* | 300371 | Adrenomyeloneuropathy, adult, 300100 (3), X-linked recessive; Adrenoleukodystrophy, 300100 (3), X-linked recessive |
| *ACBD5* | 616618 | Retinal dystrophy with leukodystrophy, 618863 (3), Autosomal recessive |
| *ACER3* | 617036 | ?Leukodystrophy, progressive, early childhood-onset, 617762 (3), Autosomal recessive |
| *ACOX1* | 609751 | Mitchell syndrome, 618960 (3); Peroxisomal acyl-CoA oxidase deficiency, 264470 (3), Autosomal recessive |
| *ACP5* | 171640 | Spondyloenchondrodysplasia with immune dysregulation, 607944 (3), Autosomal recessive |
| *ACTL6B* | 612458 | Intellectual developmental disorder with severe speech and ambulation defects, 618470 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 76, 618468 (3), Autosomal recessive |
| *ADAR* | 146920 | Dyschromatosis symmetrica hereditaria, 127400 (3), Autosomal dominant; Aicardi-Goutieres syndrome 6, 615010 (3), Autosomal recessive |
| *ADGRG1* | 604110 | Polymicrogyria, bilateral perisylvian, 615752 (3); Polymicrogyria, bilateral frontoparietal, 606854 (3), Autosomal recessive |
| *ADSL* | 608222 | Adenylosuccinase deficiency, 103050 (3), Autosomal recessive |
| *AGA* | 613228 | Aspartylglucosaminuria, 208400 (3), Autosomal recessive |
| *AIFM1* | 300169 | Cowchock syndrome, 310490 (3), X-linked recessive; Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 (3), X-linked recessive; Combined oxidative phosphorylation deficiency 6, 300816 (3), X-linked recessive; Deafness, X-linked 5, 300614 (3), X-linked recessive |
| *AIMP1* | 603605 | Leukodystrophy, hypomyelinating, 3, 260600 (3), Autosomal recessive |
| *AIMP2* | 600859 | Leukodystrophy, hypomyelinating, 17, 618006 (3), Autosomal recessive |
| *ALDH3A2* | 609523 | Sjogren-Larsson syndrome, 270200 (3), Autosomal recessive |
| *ALDH5A1* | 610045 | Succinic semialdehyde dehydrogenase deficiency, 271980 (3), Autosomal recessive |

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| *APP* | 104760 | Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants, 605714 (3), Autosomal dominant; Alzheimer disease 1, familial, 104300 (3), Autosomal dominant |
| *ARSA* | 607574 | Metachromatic leukodystrophy, 250100 (3), Autosomal recessive |
| *ASAH1* | 613468 | Farber lipogranulomatosis, 228000 (3), Autosomal recessive; Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 (3), Autosomal recessive |
| *ASPA* | 608034 | Canavan disease, 271900 (3), Autosomal recessive |
| *ATAD3A* | 612316 | Harel-Yoon syndrome, 617183 (3), Autosomal dominant, Autosomal recessive; Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810 (3), Autosomal recessive |
| *ATP7A* | 300011 | Occipital horn syndrome, 304150 (3), X-linked recessive; Menkes disease, 309400 (3), X-linked recessive; Spinal muscular atrophy, distal, X-linked 3, 300489 (3), X-linked recessive |
| *ATP7B* | 606882 | Wilson disease, 277900 (3), Autosomal recessive |
| *AUH* | 600529 | 3-methylglutaconic aciduria, type I, 250950 (3), Autosomal recessive |
| *B3GALNT2* | 610194 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181 (3), Autosomal recessive |
| *BCAP31* | 300398 | Deafness, dystonia, and cerebral hypomyelination, 300475 (3), X-linked recessive |
| *BCS1L* | 603647 | Leigh syndrome, 256000 (3), Autosomal recessive, Mitochondrial; GRACILE syndrome, 603358 (3), Autosomal recessive; Bjornstad syndrome, 262000 (3), Autosomal recessive; Mitochondrial complex III deficiency, nuclear type 1, 124000 (3), Autosomal recessive |
| *BOLA3* | 613183 | Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299 (3), Autosomal recessive |
| *BTD* | 609019 | Biotinidase deficiency, 253260 (3), Autosomal recessive |
| *CBS* | 613381 | Homocystinuria, B6-responsive and nonresponsive types, 236200 (3), Autosomal recessive; Thrombosis, hyperhomocysteinemic, 236200 (3), Autosomal recessive |
| *CIC* | 612082 | Mental retardation, autosomal dominant 45, 617600 (3), Autosomal dominant |
| *CLCN2* | 600570 | {Epilepsy, juvenile absence, susceptibility to, 2}, 607628 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628 (3), Autosomal dominant; {Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628 (3), Autosomal dominant; Leukoencephalopathy with ataxia, 615651 (3), Autosomal recessive; Hyperaldosteronism, familial, type II, 605635 (3), Autosomal dominant |
| *CLN3* | 607042 | Ceroid lipofuscinosis, neuronal, 3, 204200 (3), Autosomal recessive |
| *CLN5* | 608102 | Ceroid lipofuscinosis, neuronal, 5, 256731 (3), Autosomal recessive |

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| *CLN6* | 606725 | Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 6, 601780 (3), Autosomal recessive |
| *CLN8* | 607837 | Ceroid lipofuscinosis, neuronal, 8, 600143 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 (3), Autosomal recessive |
| *CNP* | 123830 | No OMIM phenotype |
| *CNTNAP1* | 602346 | Lethal congenital contracture syndrome 7, 616286 (3), Autosomal recessive; Hypomyelinating neuropathy, congenital, 3, 618186 (3), Autosomal recessive |
| *COA8 (APOPT1)* | 616003 | Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial |
| *COL4A1* | 120130 | Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 (3), Autosomal dominant; Brain small vessel disease with or without ocular anomalies, 175780 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3); ?Retinal arteries, tortuosity of, 180000 (3), Autosomal dominant; Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564 (3), Autosomal dominant |
| *COL4A2* | 120090 | Brain small vessel disease 2, 614483 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3) |
| *COX10* | 602125 | Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial; Leigh syndrome due to mitochondrial COX4 deficiency, 256000 (3), Autosomal recessive, Mitochondrial |
| *COX15* | 603646 | Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 (3), Autosomal recessive; Leigh syndrome due to cytochrome c oxidase deficiency, 256000 (3), Autosomal recessive, Mitochondrial |
| *CPS1* | 608307 | Carbamoylphosphate synthetase I deficiency, 237300 (3), Autosomal recessive; {Pulmonary hypertension, neonatal, susceptibility to}, 615371 (3) |
| *CSF1R* | 164770 | Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 (3), Autosomal recessive; Leukoencephalopathy, diffuse hereditary, with spheroids, 221820 (3), Autosomal dominant |
| *CTC1* | 613129 | Cerebroretinal microangiopathy with calcifications and cysts, 612199 (3), Autosomal recessive |
| *CTSA* | 613111 | Galactosialidosis, 256540 (3), Autosomal recessive |
| *CTSD* | 116840 | Ceroid lipofuscinosis, neuronal, 10, 610127 (3), Autosomal recessive |
| *CTSF* | 603539 | Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362 (3), Autosomal recessive |
| *CYP27A1* | 606530 | Cerebrotendinous xanthomatosis, 213700 (3), Autosomal recessive |
| *DAG1* | 128239 | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818 (3), Autosomal recessive; Muscular dystrophy- |

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| | | dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 (3), Autosomal recessive |
| *DARS1 (DARS)* | 603084 | Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281 (3), Autosomal recessive |
| *DARS2* | 610956 | Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105 (3), Autosomal recessive |
| *DCAF17* | 612515 | Woodhouse-Sakati syndrome, 241080 (3), Autosomal recessive |
| *DDHD2* | 615003 | Spastic paraplegia 54, autosomal recessive, 615033 (3), Autosomal recessive |
| *DEGS1* | 615843 | Leukodystrophy, hypomyelinating, 18, 618404 (3), Autosomal recessive |
| *DGUOK* | 601465 | Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 (3), Autosomal recessive; Portal hypertension, noncirrhotic, 617068 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880 (3), Autosomal recessive |
| *DLAT* | 608770 | Pyruvate dehydrogenase E2 deficiency, 245348 (3), Autosomal recessive |
| *DLD* | 238331 | Dihydrolipoamide dehydrogenase deficiency, 246900 (3), Autosomal recessive |
| *DMXL2* | 612186 | ?Deafness, autosomal dominant 71, 617605 (3), Autosomal dominant; ?Polyendocrine-polyneuropathy syndrome, 616113 (3), Autosomal recessive; Epileptic encephalopathy, early infantile, 81, 618663 (3), Autosomal recessive |
| *DNAJC5* | 611203 | Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350 (3), Autosomal dominant |
| *EARS2* | 612799 | Combined oxidative phosphorylation deficiency 12, 614924 (3), Autosomal recessive |
| *ECHS1* | 602292 | Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277 (3), Autosomal recessive |
| *EIF2AK1* | 613635 | ?Leukoencephalopathy, motor delay, spasticity, and dysarthria syndrome, 618878 (3) |
| *EIF2AK2* | 176871 | Leukoencephalopathy, developmental delay, and episodic neurologic regression syndrome, 618877 (3), Autosomal dominant |
| *EIF2B1* | 606686 | Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive |
| *EIF2B2* | 606454 | Ovarioleukodystrophy, 603896 (3), Autosomal recessive; Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive |
| *EIF2B3* | 606273 | Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive |
| *EIF2B4* | 606687 | Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive; Ovarioleukodystrophy, 603896 (3), Autosomal recessive |

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| *EIF2B5* | 603945 | Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive; Ovarioleukodystrophy, 603896 (3), Autosomal recessive |
| *EPRS1 (EPRS)* | 138295 | Leukodystrophy, hypomyelinating, 15, 617951 (3), Autosomal recessive |
| *ERCC2* | 126340 | Trichothiodystrophy 1, photosensitive, 601675 (3), Autosomal recessive; ?Cerebrooculofacioskeletal syndrome 2, 610756 (3), Autosomal recessive; Xeroderma pigmentosum, group D, 278730 (3), Autosomal recessive |
| *ERCC3* | 133510 | Xeroderma pigmentosum, group B, 610651 (3), Autosomal recessive; Trichothiodystrophy 2, photosensitive, 616390 (3), Autosomal recessive |
| *ERCC6* | 609413 | {Macular degeneration, age-related, susceptibility to, 5}, 613761 (3); {Lung cancer, susceptibility to}, 211980 (3), Autosomal dominant, Somatic mutation; Cerebrooculofacioskeletal syndrome 1, 214150 (3), Autosomal recessive; Cockayne syndrome, type B, 133540 (3), Autosomal recessive; Premature ovarian failure 11, 616946 (3), Autosomal dominant; UV-sensitive syndrome 1, 600630 (3), Autosomal recessive; De Sanctis-Cacchione syndrome, 278800 (3), Autosomal recessive |
| *ERCC8* | 609412 | Cockayne syndrome, type A, 216400 (3), Autosomal recessive; UV-sensitive syndrome 2, 614621 (3), Autosomal recessive |
| *FA2H* | 611026 | Spastic paraplegia 35, autosomal recessive, 612319 (3), Autosomal recessive |
| *FAM126A* | 610531 | Leukodystrophy, hypomyelinating, 5, 610532 (3), Autosomal recessive |
| *FAR1* | 616107 | Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154 (3), Autosomal recessive |
| *FARS2* | 611592 | Spastic paraplegia 77, autosomal recessive, 617046 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 14, 614946 (3), Autosomal recessive |
| *FASTKD2* | 612322 | Combined oxidative phosphorylation deficiency 44, 618855 (3), Autosomal recessive |
| *FBXL4* | 605654 | Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471 (3), Autosomal recessive |
| *FDX2* | 614585 | Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900 (3), Autosomal recessive |
| *FHL1* | 300163 | Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717 (3), X-linked dominant; Scapuloperoneal myopathy, X-linked dominant, 300695 (3), X-linked dominant; Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718 (3), X-linked; ?Uruguay faciocardiomusculoskeletal syndrome, 300280 (3), X-linked recessive; Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 (3), X-linked recessive; Myopathy, X-linked, with postural muscle atrophy, 300696 (3), X-linked recessive |

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| *FIG4* | 609390 | Yunis-Varon syndrome, 216340 (3), Autosomal recessive; ?Polymicrogyria, bilateral temporooccipital, 612691 (3), Autosomal recessive; Charcot-Marie-Tooth disease, type 4J, 611228 (3), Autosomal recessive; Amyotrophic lateral sclerosis 11, 612577 (3), Autosomal dominant |
| *FKTN* | 607440 | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 (3), Autosomal recessive; Cardiomyopathy, dilated, 1X, 611615 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 (3), Autosomal recessive |
| *FLVCR2* | 610865 | Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790 (3), Autosomal recessive |
| *FOLR1* | 136430 | Neurodegeneration due to cerebral folate transport deficiency, 613068 (3), Autosomal recessive |
| *FUCA1* | 612280 | Fucosidosis, 230000 (3), Autosomal recessive |
| *GALC* | 606890 | Krabbe disease, 245200 (3), Autosomal recessive |
| *GALT* | 606999 | Galactosemia, 230400 (3), Autosomal recessive |
| *GAN* | 605379 | Giant axonal neuropathy-1, 256850 (3), Autosomal recessive |
| *GBE1* | 607839 | Polyglucosan body disease, adult form, 263570 (3), Autosomal recessive; Glycogen storage disease IV, 232500 (3), Autosomal recessive |
| *GFAP* | 137780 | Alexander disease, 203450 (3), Autosomal dominant |
| *GFM1* | 606639 | Combined oxidative phosphorylation deficiency 1, 609060 (3), Autosomal recessive |
| *GFM2* | 606544 | Combined oxidative phosphorylation deficiency 39, 618397 (3), Autosomal recessive |
| *GJA1* | 121014 | Erythrokeratodermia variabilis et progressiva 3, 617525 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Atrioventricular septal defect 3, 600309 (3), Autosomal dominant; Oculodentodigital dysplasia, 164200 (3), Autosomal dominant; Syndactyly, type III, 186100 (3), Autosomal dominant; Oculodentodigital dysplasia, autosomal recessive, 257850 (3), Autosomal recessive; Hypoplastic left heart syndrome 1, 241550 (3), Autosomal recessive; Palmoplantar keratoderma with congenital alopecia, 104100 (3), Autosomal dominant |
| *GJB1* | 304040 | Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800 (3), X-linked dominant |
| *GJC2* | 608803 | Spastic paraplegia 44, autosomal recessive, 613206 (3), Autosomal recessive; Lymphatic malformation 3, 613480 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 2, 608804 (3), Autosomal recessive |
| *GLA* | 300644 | Fabry disease, 301500 (3), X-linked; Fabry disease, cardiac variant, 301500 (3), X-linked |

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| *GLB1* | 611458 | GM1-gangliosidosis, type III, 230650 (3), Autosomal recessive; GM1-gangliosidosis, type I, 230500 (3), Autosomal recessive; Mucopolysaccharidosis type IVB (Morquio), 253010 (3), Autosomal recessive; GM1-gangliosidosis, type II, 230600 (3), Autosomal recessive |
| *GM2A* | 613109 | GM2-gangliosidosis, AB variant, 272750 (3), Autosomal recessive |
| *GNAS* | 139320 | ACTH-independent macronodular adrenal hyperplasia, 219080 (3), Somatic mutation; Pseudohypoparathyroidism Ic, 612462 (3), Autosomal dominant; Pseudohypoparathyroidism Ib, 603233 (3), Autosomal dominant; Pseudopseudohypoparathyroidism, 612463 (3), Autosomal dominant; McCune-Albright syndrome, somatic, mosaic, 174800 (3); Osseous heteroplasia, progressive, 166350 (3), Autosomal dominant; Pituitary adenoma 3, multiple types, somatic, 617686 (3); Pseudohypoparathyroidism Ia, 103580 (3), Autosomal dominant |
| *GPHN* | 603930 | Molybdenum cofactor deficiency C, 615501 (3), Autosomal recessive |
| *GRN* | 138945 | Ceroid lipofuscinosis, neuronal, 11, 614706 (3), Autosomal recessive; Aphasia, primary progressive, 607485 (3), Autosomal dominant; Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 (3), Autosomal dominant |
| *GTF2H5* | 608780 | Trichothiodystrophy 3, photosensitive, 616395 (3) |
| *HEPACAM* | 611642 | Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 (3), Autosomal recessive; Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926 (3), Autosomal dominant |
| *HEXA* | 606869 | GM2-gangliosidosis, several forms, 272800 (3), Autosomal recessive; Tay-Sachs disease, 272800 (3), Autosomal recessive; [Hex A pseudodeficiency], 272800 (3), Autosomal recessive |
| *HEXB* | 606873 | Sandhoff disease, infantile, juvenile, and adult forms, 268800 (3), Autosomal recessive |
| *HIKESHI* | 614908 | Leukodystrophy, hypomyelinating, 13, 616881 (3), Autosomal recessive |
| *HLCS* | 609018 | Holocarboxylase synthetase deficiency, 253270 (3), Autosomal recessive |
| *HMGCL* | 613898 | HMG-CoA lyase deficiency, 246450 (3), Autosomal recessive |
| *HSD17B4* | 601860 | D-bifunctional protein deficiency, 261515 (3), Autosomal recessive; Perrault syndrome 1, 233400 (3), Autosomal recessive |
| *HSPD1* | 118190 | Spastic paraplegia 13, autosomal dominant, 605280 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 4, 612233 (3), Autosomal recessive |
| *HTRA1* | 602194 | {Macular degeneration, age-related, neovascular type}, 610149 (3); {Macular degeneration, age-related, 7}, 610149 (3); Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779 (3), Autosomal dominant; CARASIL syndrome, 600142 (3), Autosomal recessive |

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| *IBA57* | 615316 | ?Spastic paraplegia 74, autosomal recessive, 616451 (3), Autosomal recessive; Multiple mitochondrial dysfunctions syndrome 3, 615330 (3), Autosomal recessive |
| *IFIH1* | 606951 | Aicardi-Goutieres syndrome 7, 615846 (3), Autosomal dominant; Singleton-Merten syndrome 1, 182250 (3), Autosomal dominant |
| *IKBKG* | 300248 | Immunodeficiency 33, 300636 (3), X-linked recessive; Incontinentia pigmenti, 308300 (3), X-linked dominant; Ectodermal dysplasia and immunodeficiency 1, 300291 (3), X-linked recessive |
| *ISCA2* | 615317 | Multiple mitochondrial dysfunctions syndrome 4, 616370 (3), Autosomal recessive |
| *IVD* | 607036 | Isovaleric acidemia, 243500 (3), Autosomal recessive |
| *JAM3* | 606871 | Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730 (3), Autosomal recessive |
| *KARS1 (KARS)* | 601421 | ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 (3), Autosomal recessive; Deafness, autosomal recessive 89, 613916 (3), Autosomal recessive |
| *L2HGDH* | 609584 | L-2-hydroxyglutaric aciduria, 236792 (3), Autosomal recessive |
| *LAMA2* | 156225 | Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 (3), Autosomal recessive; Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855 (3), Autosomal recessive |
| *LARGE1* | 603590 | Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 (3), Autosomal recessive |
| *LARS2* | 604544 | Perrault syndrome 4, 615300 (3), Autosomal recessive; ?Hydrops, lactic acidosis, and sideroblastic anemia, 617021 (3), Autosomal recessive |
| *LIPT1* | 610284 | Lipoyltransferase 1 deficiency, 616299 (3), Autosomal recessive |
| *LIPT2* | 617659 | Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668 (3), Autosomal recessive |
| *LMNB1* | 150340 | Leukodystrophy, adult-onset, autosomal dominant, 169500 (3), Autosomal dominant |
| *LYRM7* | 615831 | Mitochondrial complex III deficiency, nuclear type 8, 615838 (3), Autosomal recessive |
| *MAN2B1* | 609458 | Mannosidosis, alpha-, types I and II, 248500 (3), Autosomal recessive |
| *MANBA* | 609489 | Mannosidosis, beta, 248510 (3), Autosomal recessive |
| *MARS2* | 609728 | Spastic ataxia 3, autosomal recessive, 611390 (3), Autosomal recessive; ?Combined oxidative phosphorylation deficiency 25, 616430 (3), Autosomal recessive |
| *MCOLN1* | 605248 | Mucolipidosis IV, 252650 (3), Autosomal recessive |
| *MFSD8* | 611124 | Macular dystrophy with central cone involvement, 616170 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 7, 610951 (3), Autosomal recessive |

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| *MLC1* | 605908 | Megalencephalic leukoencephalopathy with subcortical cysts, 604004 (3), Autosomal recessive |
| *MMUT (MUT)* | 609058 | Methylmalonic aciduria, mut(0) type, 251000 (3), Autosomal recessive |
| *MOCS1* | 603707 | Molybdenum cofactor deficiency A, 252150 (3), Autosomal recessive |
| *MOCS2* | 603708 | Molybdenum cofactor deficiency B, 252160 (3), Autosomal recessive |
| *MPLKIP* | 609188 | Trichothiodystrophy 4, nonphotosensitive, 234050 (3), Autosomal recessive |
| *MPV17* | 137960 | Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810 (3), Autosomal recessive |
| *MRPS34* | 611994 | Combined oxidative phosphorylation deficiency 32, 617664 (3), Autosomal recessive |
| *MTFMT* | 611766 | Combined oxidative phosphorylation deficiency 15, 614947 (3), Autosomal recessive; Mitochondrial complex I deficiency, nuclear type 27, 618248 (3), Autosomal recessive |
| *MTHFR* | 607093 | {Vascular disease, susceptibility to} (3); {Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant; Homocystinuria due to MTHFR deficiency, 236250 (3), Autosomal recessive; {Neural tube defects, susceptibility to}, 601634 (3), Autosomal recessive; {Thromboembolism, susceptibility to}, 188050 (3), Autosomal dominant |
| *MTO1* | 614667 | Combined oxidative phosphorylation deficiency 10, 614702 (3), Autosomal recessive |
| *NAXD* | 615910 | Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321 (3), Autosomal recessive |
| *NAXE* | 608862 | Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186 (3), Autosomal recessive |
| *NDUFA2* | 602137 | ?Mitochondrial complex I deficiency, nuclear type 13, 618235 (3), Autosomal recessive |
| *NDUFA9* | 603834 | Mitochondrial complex I deficiency, nuclear type 26, 618247 (3), Autosomal recessive |
| *NDUFAF1* | 606934 | Mitochondrial complex I deficiency, nuclear type 11, 618234 (3), Autosomal recessive |
| *NDUFAF3* | 612911 | Mitochondrial complex I deficiency, nuclear type 18, 618240 (3), Autosomal recessive |
| *NDUFAF4* | 611776 | Mitochondrial complex I deficiency, nuclear type 15, 618237 (3), Autosomal recessive |
| *NDUFAF6* | 612392 | Fanconi renotubular syndrome 5, 618913 (3), Autosomal recessive; Mitochondrial complex I deficiency, nuclear type 17, 618239 (3), Autosomal recessive |
| *NDUFS1* | 157655 | Mitochondrial complex I deficiency, nuclear type 5, 618226 (3), Autosomal recessive |

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| *NDUFS2* | 602985 | Mitochondrial complex I deficiency, nuclear type 6, 618228 (3), Autosomal recessive |
| *NDUFS4* | 602694 | Mitochondrial complex I deficiency, nuclear type 1, 252010 (3), Autosomal recessive |
| *NDUFS7* | 601825 | Mitochondrial complex I deficiency, nuclear type 3, 618224 (3), Autosomal recessive |
| *NDUFS8* | 602141 | Mitochondrial complex I deficiency, nuclear type 2, 618222 (3), Autosomal recessive |
| *NDUFV1* | 161015 | Mitochondrial complex I deficiency, nuclear type 4, 618225 (3), Autosomal recessive |
| *NEU1* | 608272 | Sialidosis, type II, 256550 (3), Autosomal recessive; Sialidosis, type I, 256550 (3), Autosomal recessive |
| *NKX6-2* | 605955 | Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560 (3), Autosomal recessive |
| *NOTCH3* | 600276 | ?Myofibromatosis, infantile 2, 615293 (3), Autosomal dominant; Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310 (3), Autosomal dominant; Lateral meningocele syndrome, 130720 (3), Autosomal dominant |
| *NPC1* | 607623 | Niemann-Pick disease, type D, 257220 (3), Autosomal recessive; Niemann-Pick disease, type C1, 257220 (3), Autosomal recessive |
| *NPC2* | 601015 | Niemann-pick disease, type C2, 607625 (3), Autosomal recessive |
| *NTNG2* | 618689 | Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718 (3), Autosomal recessive |
| *NUBPL* | 613621 | Mitochondrial complex I deficiency, nuclear type 21, 618242 (3), Autosomal recessive |
| *OCLN* | 602876 | Pseudo-TORCH syndrome 1, 251290 (3), Autosomal recessive |
| *OCRL* | 300535 | Lowe syndrome, 309000 (3), X-linked recessive; Dent disease 2, 300555 (3), X-linked recessive |
| *PAH* | 612349 | [Hyperphenylalaninemia, non-PKU mild], 261600 (3), Autosomal recessive; Phenylketonuria, 261600 (3), Autosomal recessive |
| *PARS2* | 612036 | Epileptic encephalopathy, early infantile, 75, 618437 (3), Autosomal recessive |
| *PCCA* | 232000 | Propionicacidemia, 606054 (3), Autosomal recessive |
| *PCCB* | 232050 | Propionicacidemia, 606054 (3), Autosomal recessive |
| *PDGFB* | 190040 | Dermatofibrosarcoma protuberans, 607907 (3); Basal ganglia calcification, idiopathic, 5, 615483 (3), Autosomal dominant; Meningioma, SIS-related, 607174 (3), Autosomal dominant |
| *PDGFRB* | 173410 | Myeloproliferative disorder with eosinophilia, 131440 (4), Autosomal dominant; Basal ganglia calcification, idiopathic, 4, 615007 (3), Autosomal dominant; Kosaki overgrowth syndrome, 616592 (3), Autosomal dominant; Premature aging syndrome, Penttinen type, 601812 (3), Autosomal dominant; Myofibromatosis, infantile, 1, 228550 (3), Autosomal dominant |
| *PDHA1* | 300502 | Pyruvate dehydrogenase E1-alpha deficiency, 312170 (3), X-linked dominant |

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| *PDHB* | 179060 | Pyruvate dehydrogenase E1-beta deficiency, 614111 (3) |
| *PDHX* | 608769 | Lacticacidemia due to PDX1 deficiency, 245349 (3), Autosomal recessive |
| *PDP1* | 605993 | Pyruvate dehydrogenase phosphatase deficiency, 608782 (3), Autosomal recessive |
| *PEX1* | 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* | 602859 | Peroxisome biogenesis disorder 6B, 614871 (3), Autosomal recessive; Peroxisome biogenesis disorder 6A (Zellweger), 614870 (3), Autosomal recessive |
| *PEX11B* | 603867 | ?Peroxisome biogenesis disorder 14B, 614920 (3), Autosomal recessive |
| *PEX12* | 601758 | Peroxisome biogenesis disorder 3A (Zellweger), 614859 (3), Autosomal recessive; Peroxisome biogenesis disorder 3B, 266510 (3), Autosomal recessive |
| *PEX13* | 601789 | Peroxisome biogenesis disorder 11A (Zellweger), 614883 (3), Autosomal recessive; Peroxisome biogenesis disorder 11B, 614885 (3), Autosomal recessive |
| *PEX14* | 601791 | Peroxisome biogenesis disorder 13A (Zellweger), 614887 (3), Autosomal recessive |
| *PEX16* | 603360 | Peroxisome biogenesis disorder 8A (Zellweger), 614876 (3), Autosomal recessive; Peroxisome biogenesis disorder 8B, 614877 (3), Autosomal recessive |
| *PEX19* | 600279 | Peroxisome biogenesis disorder 12A (Zellweger), 614886 (3), Autosomal recessive |
| *PEX2* | 170993 | Peroxisome biogenesis disorder 5A (Zellweger), 614866 (3), Autosomal recessive; Peroxisome biogenesis disorder 5B, 614867 (3), Autosomal recessive |
| *PEX26* | 608666 | Peroxisome biogenesis disorder 7A (Zellweger), 614872 (3), Autosomal recessive; Peroxisome biogenesis disorder 7B, 614873 (3), Autosomal recessive |
| *PEX3* | 603164 | Peroxisome biogenesis disorder 10A (Zellweger), 614882 (3), Autosomal recessive; ?Peroxisome biogenesis disorder 10B, 617370 (3), Autosomal recessive |
| *PEX5* | 600414 | Peroxisome biogenesis disorder 2B, 202370 (3), Autosomal recessive; Rhizomelic chondrodysplasia punctata, type 5, 616716 (3), Autosomal recessive; Peroxisome biogenesis disorder 2A (Zellweger), 214110 (3), Autosomal recessive |
| *PEX6* | 601498 | Peroxisome biogenesis disorder 4B, 614863 (3), Autosomal dominant, Autosomal recessive; Heimler syndrome 2, 616617 (3), Autosomal recessive; Peroxisome biogenesis disorder 4A (Zellweger), 614862 (3), Autosomal recessive |

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| *PEX7* | 601757 | Peroxisome biogenesis disorder 9B, 614879 (3), Autosomal recessive; Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive |
| *PLEKHG2* | 611893 | Leukodystrophy and acquired microcephaly with or without dystonia, 616763 (3), Autosomal recessive |
| *PLP1* | 300401 | Pelizaeus-Merzbacher disease, 312080 (3), X-linked recessive; Spastic paraplegia 2, X-linked, 312920 (3), X-linked recessive |
| *PLPBP* | 604436 | Epilepsy, early-onset, vitamin B6-dependent, 617290 (3), Autosomal recessive |
| *PMPCB* | 603131 | Multiple mitochondrial dysfunctions syndrome 6, 617954 (3), Autosomal recessive |
| *PNPT1* | 610316 | Deafness, autosomal recessive 70, 614934 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 13, 614932 (3), Autosomal recessive |
| *POLG* | 174763 | Progressive external ophthalmoplegia, autosomal dominant 1, 157640 (3), Autosomal dominant; Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 (3), Autosomal recessive; Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 (3), Autosomal recessive; Progressive external ophthalmoplegia, autosomal recessive 1, 258450 (3), Autosomal recessive |
| *POLR1C* | 610060 | Treacher Collins syndrome 3, 248390 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 11, 616494 (3), Autosomal recessive |
| *POLR3A* | 614258 | Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3), Autosomal recessive; Wiedemann-Rautenstrauch syndrome, 264090 (3), Autosomal recessive |
| *POLR3B* | 614366 | Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381 (3), Autosomal recessive |
| *POLR3K* | 606007 | No OMIM phenotype |
| *POMGNT1* | 606822 | Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 (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 |
| *POMT1* | 607423 | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 (3), Autosomal recessive |

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| *POMT2* | 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 mental retardation), type B, 2, 613156 (3), Autosomal recessive |
| *PPT1* | 600722 | Ceroid lipofuscinosis, neuronal, 1, 256730 (3), Autosomal recessive |
| *PRF1* | 170280 | Hemophagocytic lymphohistiocytosis, familial, 2, 603553 (3), Autosomal recessive; Aplastic anemia, 609135 (3); Lymphoma, non-Hodgkin, 605027 (3) |
| *PSAP* | 176801 | Gaucher disease, atypical, 610539 (3); Krabbe disease, atypical, 611722 (3), Autosomal recessive; Combined SAP deficiency, 611721 (3), Autosomal recessive; Metachromatic leukodystrophy due to SAP-b deficiency, 249900 (3), Autosomal recessive |
| *PSAT1* | 610936 | Neu-Laxova syndrome 2, 616038 (3), Autosomal recessive; ?Phosphoserine aminotransferase deficiency, 610992 (3), Autosomal recessive |
| *PSPH* | 172480 | Phosphoserine phosphatase deficiency, 614023 (3), Autosomal recessive |
| *PYCR2* | 616406 | Leukodystrophy, hypomyelinating, 10, 616420 (3), Autosomal recessive |
| *QDPR* | 612676 | Hyperphenylalaninemia, BH4-deficient, C, 261630 (3), Autosomal recessive |
| *RARS1 (RARS)* | 107820 | Leukodystrophy, hypomyelinating, 9, 616140 (3), Autosomal recessive |
| *RNASEH2A* | 606034 | Aicardi-Goutieres syndrome 4, 610333 (3), Autosomal recessive |
| *RNASEH2B* | 610326 | Aicardi-Goutieres syndrome 2, 610181 (3), Autosomal recessive |
| *RNASEH2C* | 610330 | Aicardi-Goutieres syndrome 3, 610329 (3), Autosomal recessive |
| *RNASET2* | 612944 | Leukoencephalopathy, cystic, without megalencephaly, 612951 (3), Autosomal recessive |
| *SAMHD1* | 606754 | ?Chilblain lupus 2, 614415 (3), Autosomal dominant; Aicardi-Goutieres syndrome 5, 612952 (3), Autosomal recessive |
| *SCP2* | 184755 | ?Leukoencephalopathy with dystonia and motor neuropathy, 613724 (3), Autosomal recessive |
| *SDHA* | 600857 | Cardiomyopathy, dilated, 1GG, 613642 (3), Autosomal recessive; Leigh syndrome, 256000 (3), Autosomal recessive, Mitochondrial; Paragangliomas 5, 614165 (3), Autosomal dominant; Mitochondrial respiratory chain complex II deficiency, 252011 (3), Autosomal recessive |
| *SDHAF1* | 612848 | Mitochondrial complex II deficiency, 252011 (3), Autosomal recessive |
| *SDHAF2* | 613019 | Paragangliomas 2, 601650 (3), Autosomal dominant |
| *SDHB* | 185470 | Gastrointestinal stromal tumor, 606764 (3), Isolated cases, Autosomal dominant; Pheochromocytoma, 171300 (3), Autosomal dominant; Paragangliomas 4, 115310 (3), Autosomal dominant; Paraganglioma and gastric stromal sarcoma, 606864 (3) |

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| *SDHD* | 602690 | Paragangliomas 1, with or without deafness, 168000 (3), Autosomal dominant; Mitochondrial complex II deficiency, 252011 (3), Autosomal recessive; Paraganglioma and gastric stromal sarcoma, 606864 (3); Pheochromocytoma, 171300 (3), Autosomal dominant |
| *SEPSECS* | 613009 | Pontocerebellar hypoplasia type 2D, 613811 (3), Autosomal recessive |
| *SGO1* | 609168 | Chronic atrial and intestinal dysrhythmia, 616201 (3), Autosomal recessive |
| *SHANK3* | 606230 | {Schizophrenia 15}, 613950 (3), Autosomal dominant; Phelan-McDermid syndrome, 606232 (3), Autosomal dominant |
| *SLC13A3* | 606411 | Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate, 618384 (3), Autosomal recessive |
| *SLC16A2* | 300095 | Allan-Herndon-Dudley syndrome, 300523 (3), X-linked |
| *SLC17A5* | 604322 | Sialic acid storage disorder, infantile, 269920 (3), Autosomal recessive; Salla disease, 604369 (3), Autosomal recessive |
| *SLC20A2* | 158378 | Basal ganglia calcification, idiopathic, 1, 213600 (3), Autosomal dominant |
| *SLC25A12* | 603667 | Epileptic encephalopathy, early infantile, 39, 612949 (3), Autosomal recessive |
| *SLC44A1* | 606105 | Neurodegeneration, childhood-onset, with ataxia, tremor, optic atrophy, and cognitive decline, 618868 (3), Autosomal recessive |
| *SLC7A2* | 601872 | No OMIM phenotype |
| *SNAP29* | 604202 | Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528 (3), Autosomal recessive |
| *SNORD118* | 616663 | Leukoencephalopathy, brain calcifications, and cysts, 614561 (3), Autosomal recessive |
| *SOX10* | 602229 | Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 (3), Autosomal dominant; PCWH syndrome, 609136 (3), Autosomal dominant; Waardenburg syndrome, type 4C, 613266 (3), Autosomal dominant |
| *SPG11* | 610844 | Charcot-Marie-Tooth disease, axonal, type 2X, 616668 (3), Autosomal recessive; Spastic paraplegia 11, autosomal recessive, 604360 (3), Autosomal recessive; Amyotrophic lateral sclerosis 5, juvenile, 602099 (3), Autosomal recessive |
| *STN1* | 613128 | Cerebroretinal microangiopathy with calcifications and cysts 2, 617341 (3), Autosomal recessive |
| *STX11* | 605014 | Hemophagocytic lymphohistiocytosis, familial, 4, 603552 (3), Autosomal recessive |
| *STXBP2* | 601717 | Hemophagocytic lymphohistiocytosis, familial, 5, 613101 (3) |
| *SUCLA2* | 603921 | Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 (3), Autosomal recessive |
| *SUMF1* | 607939 | Multiple sulfatase deficiency, 272200 (3), Autosomal recessive |
| *SUOX* | 606887 | Sulfite oxidase deficiency, 272300 (3), Autosomal recessive |

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| *SURF1* | 185620 | Leigh syndrome, due to COX IV deficiency, 256000 (3), Autosomal recessive, Mitochondrial; Charcot-Marie-Tooth disease, type 4K, 616684 (3), Autosomal recessive |
| *TACO1* | 612958 | Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial |
| *TMEM106B* | 613413 | Leukodystrophy, hypomyelinating, 16, 617964 (3), Autosomal dominant |
| *TMEM63A* | 618685 | Leukodystrophy, hypomyelinating, 19, transient infantile, 618688 (3), Autosomal dominant |
| *TPP1* | 607998 | Spinocerebellar ataxia, autosomal recessive 7, 609270 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 2, 204500 (3), Autosomal recessive |
| *TREM2* | 605086 | Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193 (3), Autosomal dominant |
| *TREX1* | 606609 | {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant; Vasculopathy, retinal, with cerebral leukodystrophy, 192315 (3), Autosomal dominant; Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 (3), Autosomal dominant, Autosomal recessive; Chilblain lupus, 610448 (3), Autosomal dominant |
| *TSEN54* | 608755 | Pontocerebellar hypoplasia type 4, 225753 (3), Autosomal recessive; Pontocerebellar hypoplasia type 2A, 277470 (3), Autosomal recessive; ?Pontocerebellar hypoplasia type 5, 610204 (3), Autosomal recessive |
| *TTR* | 176300 | Amyloidosis, hereditary, transthyretin-related, 105210 (3), Autosomal dominant; [Dystransthyretinemic hyperthyroxinemia], 145680 (3), Autosomal dominant; Carpal tunnel syndrome, familial, 115430 (3), Autosomal dominant |
| *TUBB4A* | 602662 | Leukodystrophy, hypomyelinating, 6, 612438 (3), Autosomal dominant; Dystonia 4, torsion, autosomal dominant, 128101 (3), Autosomal dominant |
| *TWNK* | 606075 | Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 (3), Autosomal dominant; Perrault syndrome 5, 616138 (3), Autosomal recessive |
| *TYMP* | 131222 | Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041 (3), Autosomal recessive |
| *TYROBP* | 604142 | Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770 (3), Autosomal recessive |
| *UBTF* | 600673 | Neurodegeneration, childhood-onset, with brain atrophy, 617672 (3), Autosomal dominant |
| *UFM1* | 610553 | Leukodystrophy, hypomyelinating, 14, 617899 (3), Autosomal recessive |
| *UNC13D* | 608897 | Hemophagocytic lymphohistiocytosis, familial, 3, 608898 (3), Autosomal recessive |

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| *VPS11* | 608549 | Leukodystrophy, hypomyelinating, 12, 616683 (3), Autosomal recessive |
| *WARS2* | 604733 | Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710 (3), Autosomal recessive |
| *WT1* | 607102 | Mesothelioma, somatic, 156240 (3); Wilms tumor, type 1, 194070 (3), Autosomal dominant, Somatic mutation; Frasier syndrome, 136680 (3), Autosomal dominant, Somatic mutation; Denys-Drash syndrome, 194080 (3), Autosomal dominant, Somatic mutation; Meacham syndrome, 608978 (3); Nephrotic syndrome, type 4, 256370 (3), Autosomal dominant |
| *XPR1* | 605237 | Basal ganglia calcification, idiopathic, 6, 616413 (3), Autosomal dominant |
| *ZFYVE26* | 612012 | Spastic paraplegia 15, autosomal recessive, 270700 (3), Autosomal recessive |

Gene symbols used are according to the HGNC guidelines. For some genes a previously HGNC-approved symbol is in brackets.
Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.
OMIM release used for OMIM disease identifiers and descriptions: Aug 20, 2020

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.