

<b>Leukodystrophy panel</b>		
<b>versie</b>	16-Oct-2018 (179 genen)	Centrum voor Medische Genetica Gent
<b>Gene</b>	<b>OMIM gene ID</b>	<b>Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern</b>
<i>AARS2</i>	612035	Combined oxidative phosphorylation deficiency 8, 614096 (3), Autosomal recessive; Leukoencephalopathy, progressive, with ovarian failure, 615889 (3), Autosomal recessive
<i>ABCD1</i>	300371	Adrenoleukodystrophy, 300100 (3), X-linked recessive; Adrenomyeloneuropathy, adult, 300100 (3), X-linked recessive
<i>ACOX1</i>	609751	Peroxisomal acyl-CoA oxidase deficiency, 264470 (3), Autosomal recessive
<i>ACP5</i>	171640	Spondyloenchondrodysplasia with immune dysregulation, 607944 (3), Autosomal recessive
<i>ADAR</i>	146920	Aicardi-Goutieres syndrome 6, 615010 (3), Autosomal recessive; Dyschromatosis symmetrica hereditaria, 127400 (3), Autosomal dominant
<i>ADGRG1</i>	604110	Polymicrogyria, bilateral frontoparietal, 606854 (3), Autosomal recessive; Polymicrogyria, bilateral perisylvian, 615752 (3)
<i>ADSL</i>	608222	Adenylosuccinase deficiency, 103050 (3), Autosomal recessive
<i>AGA</i>	613228	Aspartylglucosaminuria, 208400 (3), Autosomal recessive
<i>AIMP1</i>	603605	Leukodystrophy, hypomyelinating, 3, 260600 (3), Autosomal recessive
<i>ALDH3A2</i>	609523	Sjogren-Larsson syndrome, 270200 (3), Autosomal recessive
<i>ALDH5A1</i>	610045	Succinic semialdehyde dehydrogenase deficiency, 271980 (3), Autosomal recessive
<i>APP</i>	104760	Alzheimer disease 1, familial, 104300 (3), Autosomal dominant; Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants, 605714 (3), Autosomal dominant
<i>ARSA</i>	607574	Metachromatic leukodystrophy, 250100 (3), Autosomal recessive
<i>ASAH1</i>	613468	Farber lipogranulomatosis, 228000 (3), Autosomal recessive; Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 (3), Autosomal recessive
<i>ASPA</i>	608034	Canavan disease, 271900 (3), Autosomal recessive
<i>ATP7A</i>	300011	Menkes disease, 309400 (3), X-linked recessive; Occipital horn syndrome, 304150 (3), X-linked recessive; Spinal muscular atrophy, distal, X-linked 3, 300489 (3), X-linked recessive
<i>ATP7B</i>	606882	Wilson disease, 277900 (3), Autosomal recessive
<i>BCAP31</i>	300398	Deafness, dystonia, and cerebral hypomyelination, 300475 (3), X-linked recessive
<i>BTD</i>	609019	Biotinidase deficiency, 253260 (3), Autosomal recessive

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<i>CBS</i>	613381	Homocystinuria, B6-responsive and nonresponsive types, 236200 (3), Autosomal recessive; Thrombosis, hyperhomocysteinemic, 236200 (3), Autosomal recessive
<i>CLCN2</i>	600570	{Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628 (3), Autosomal dominant; {Epilepsy, juvenile absence, susceptibility to, 2}, 607628 (3), Autosomal dominant; {Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628 (3), Autosomal dominant; Leukoencephalopathy with ataxia, 615651 (3), Autosomal recessive
<i>CLN3</i>	607042	Ceroid lipofuscinosis, neuronal, 3, 204200 (3), Autosomal recessive
<i>CLN5</i>	608102	Ceroid lipofuscinosis, neuronal, 5, 256731 (3), Autosomal recessive
<i>CLN6</i>	606725	Ceroid lipofuscinosis, neuronal, 6, 601780 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300 (3), Autosomal recessive
<i>CLN8</i>	607837	Ceroid lipofuscinosis, neuronal, 8, 600143 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 (3), Autosomal recessive
<i>COL4A1</i>	120130	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 (3), Autosomal dominant; Brain small vessel disease with or without ocular anomalies, 607595 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3); Porencephaly 1, 175780 (3), Autosomal dominant; ?Retinal arteries, tortuosity of, 180000 (3), Autosomal dominant; Schizencephaly, 269160 (3)
<i>COL4A2</i>	120090	{Hemorrhage, intracerebral, susceptibility to}, 614519 (3); Porencephaly 2, 614483 (3), Autosomal dominant
<i>CSF1R</i>	164770	Leukoencephalopathy, diffuse hereditary, with spheroids, 221820 (3), Autosomal dominant
<i>CTC1</i>	613129	Cerebroretinal microangiopathy with calcifications and cysts, 612199 (3), Autosomal recessive
<i>CTSA</i>	613111	Galactosialidosis, 256540 (3), Autosomal recessive
<i>CTSD</i>	116840	Ceroid lipofuscinosis, neuronal, 10, 610127 (3), Autosomal recessive
<i>CTSF</i>	603539	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362 (3), Autosomal recessive
<i>CYP27A1</i>	606530	Cerebrotendinous xanthomatosis, 213700 (3), Autosomal recessive
<i>DAG1</i>	128239	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818 (3), Autosomal recessive
<i>DARS</i>	603084	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281 (3), Autosomal recessive
<i>DARS2</i>	610956	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105 (3), Autosomal recessive
<i>DCAF17</i>	612515	Woodhouse-Sakati syndrome, 241080 (3), Autosomal recessive
<i>DLAT</i>	608770	Pyruvate dehydrogenase E2 deficiency, 245348 (3), Autosomal recessive

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<i>DLD</i>	238331	Dihydrolipoamide dehydrogenase deficiency, 246900 (3), Autosomal recessive
<i>DNAJC5</i>	611203	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350 (3), Autosomal dominant
<i>EARS2</i>	612799	Combined oxidative phosphorylation deficiency 12, 614924 (3), Autosomal recessive
<i>EIF2B1</i>	606686	Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive
<i>EIF2B2</i>	606454	Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive; Ovarioleukodystrophy, 603896 (3), Autosomal recessive
<i>EIF2B3</i>	606273	Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive
<i>EIF2B4</i>	606687	Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive; Ovarioleukodystrophy, 603896 (3), Autosomal recessive
<i>EIF2B5</i>	603945	Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive; Ovarioleukodystrophy, 603896 (3), Autosomal recessive
<i>ERCC2</i>	126340	?Cerebrooculofacioskeletal syndrome 2, 610756 (3), Autosomal recessive; Trichothiodystrophy 1, photosensitive, 601675 (3), Autosomal recessive; Xeroderma pigmentosum, group D, 278730 (3), Autosomal recessive
<i>ERCC3</i>	133510	Trichothiodystrophy 2, photosensitive, 616390 (3), Autosomal recessive; Xeroderma pigmentosum, group B, 610651 (3), Autosomal recessive
<i>ERCC6</i>	609413	Cerebrooculofacioskeletal syndrome 1, 214150 (3), Autosomal recessive; Cockayne syndrome, type B, 133540 (3), Autosomal recessive; De Sanctis-Cacchione syndrome, 278800 (3), Autosomal recessive; {Lung cancer, susceptibility to}, 211980 (3), Autosomal recessive; {Macular degeneration, age-related, susceptibility to, 5}, 613761 (3); Premature ovarian failure 11, 616946 (3), Autosomal dominant; UV-sensitive syndrome 1, 600630 (3), Autosomal recessive
<i>ERCC8</i>	609412	Cockayne syndrome, type A, 216400 (3), Autosomal recessive; UV-sensitive syndrome 2, 614621 (3), Autosomal recessive
<i>FA2H</i>	611026	Spastic paraplegia 35, autosomal recessive, 612319 (3), Autosomal recessive
<i>FAM126A</i>	610531	Leukodystrophy, hypomyelinating, 5, 610532 (3), Autosomal recessive
<i>FAR1</i>	616107	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154 (3), Autosomal recessive
<i>FARS2</i>	611592	Combined oxidative phosphorylation deficiency 14, 614946 (3), Autosomal recessive; Spastic paraplegia 77, autosomal recessive, 617046 (3), Autosomal recessive

<i>FHL1</i>	300163	Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 (3), X-linked recessive; Myopathy, X-linked, with postural muscle atrophy, 300696 (3), X-linked recessive; Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717 (3), X-linked dominant; Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718 (3), X-linked; Scapuloperoneal myopathy, X-linked dominant, 300695 (3), X-linked dominant; ?Uruguay faciocardiomusculoskeletal syndrome, 300280 (3), X-linked recessive
<i>FKTN</i>	607440	Cardiomyopathy, dilated, 1X, 611615 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 (3), Autosomal recessive
<i>FOLR1</i>	136430	Neurodegeneration due to cerebral folate transport deficiency, 613068 (3), Autosomal recessive
<i>FUCA1</i>	612280	Fucosidosis, 230000 (3), Autosomal recessive
<i>GALC</i>	606890	Krabbe disease, 245200 (3), Autosomal recessive
<i>GALT</i>	606999	Galactosemia, 230400 (3), Autosomal recessive
<i>GAN</i>	605379	Giant axonal neuropathy-1, 256850 (3), Autosomal recessive
<i>GFAP</i>	137780	Alexander disease, 203450 (3), Autosomal dominant
<i>GJA1</i>	121014	Atrioventricular septal defect 3, 600309 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Erythrokeratoderma variabilis et progressiva 3, 617525 (3), Autosomal dominant; Hypoplastic left heart syndrome 1, 241550 (3), Autosomal recessive; Oculodentodigital dysplasia, 164200 (3), Autosomal dominant; Oculodentodigital dysplasia, autosomal recessive, 257850 (3), Autosomal recessive; Palmoplantar keratoderma with congenital alopecia, 104100 (3), Autosomal dominant; Syndactyly, type III, 186100 (3), Autosomal dominant
<i>GJC2</i>	608803	Leukodystrophy, hypomyelinating, 2, 608804 (3), Autosomal recessive; Lymphedema, hereditary, IC, 613480 (3), Autosomal dominant; Spastic paraplegia 44, autosomal recessive, 613206 (3), Autosomal recessive
<i>GLA</i>	300644	Fabry disease, 301500 (3), X-linked; Fabry disease, cardiac variant, 301500 (3), X-linked
<i>GLB1</i>	611458	GM1-gangliosidosis, type I, 230500 (3), Autosomal recessive; GM1-gangliosidosis, type II, 230600 (3), Autosomal recessive; GM1-gangliosidosis, type III, 230650 (3), Autosomal recessive; Mucopolysaccharidosis type IVB (Morquio), 253010 (3), Autosomal recessive
<i>GM2A</i>	613109	GM2-gangliosidosis, AB variant, 272750 (3), Autosomal recessive

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<i>GNAS</i>	139320	ACTH-independent macronodular adrenal hyperplasia, 219080 (3), Isolated cases; 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; Pseudohypoparathyroidism Ib, 603233 (3), Autosomal dominant; Pseudohypoparathyroidism Ic, 612462 (3), Autosomal dominant; Pseudopseudohypoparathyroidism, 612463 (3), Autosomal dominant
<i>GPHN (GEPH)</i>	603930	Molybdenum cofactor deficiency C, 615501 (3), Autosomal recessive
<i>GRN</i>	138945	Aphasia, primary progressive, 607485 (3), Autosomal dominant; Ceroid lipofuscinosis, neuronal, 11, 614706 (3), Autosomal recessive; Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 (3), Autosomal dominant
<i>GTF2H5</i>	608780	Trichothiodystrophy 3, photosensitive, 616395 (3)
<i>HEPACAM</i>	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
<i>HEXA</i>	606869	GM2-gangliosidosis, several forms, 272800 (3), Autosomal recessive; [Hex A pseudodeficiency], 272800 (3), Autosomal recessive; Tay-Sachs disease, 272800 (3), Autosomal recessive
<i>HEXB</i>	606873	Sandhoff disease, infantile, juvenile, and adult forms, 268800 (3), Autosomal recessive
<i>HLCS</i>	609018	Holocarboxylase synthetase deficiency, 253270 (3), Autosomal recessive
<i>HMGCL</i>	613898	HMG-CoA lyase deficiency, 246450 (3), Autosomal recessive
<i>HSD17B4</i>	601860	D-bifunctional protein deficiency, 261515 (3), Autosomal recessive; Perrault syndrome 1, 233400 (3), Autosomal recessive
<i>HSPD1</i>	118190	Leukodystrophy, hypomyelinating, 4, 612233 (3), Autosomal recessive; Spastic paraplegia 13, autosomal dominant, 605280 (3), Autosomal dominant
<i>HTRA1</i>	602194	CARASIL syndrome, 600142 (3), Autosomal recessive; Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779 (3), Autosomal dominant; {Macular degeneration, age-related, 7}, 610149 (3); {Macular degeneration, age-related, neovascular type}, 610149 (3)
<i>IFIH1</i>	606951	Aicardi-Goutieres syndrome 7, 615846 (3), Autosomal dominant; Singleton-Merten syndrome 1, 182250 (3), Autosomal dominant
<i>IKBKG</i>	300248	Ectodermal dysplasia, hypohidrotic, with immune deficiency, 300291 (3); Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 (3); Immunodeficiency 33, 300636 (3), X-linked recessive; Immunodeficiency, isolated, 300584 (3); Incontinentia pigmenti, 308300 (3), X-linked dominant; Invasive pneumococcal disease, recurrent isolated, 2, 300640 (3)

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<i>IVD</i>	607036	Isovaleric acidemia, 243500 (3), Autosomal recessive
<i>JAM3</i>	606871	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730 (3), Autosomal recessive
<i>L2HGDH (LHGD)</i>	609584	L-2-hydroxyglutaric aciduria, 236792 (3), Autosomal recessive
<i>LAMA2</i>	156225	Muscular dystrophy, congenital merosin-deficient, 607855 (3), Autosomal recessive; Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855 (3), Autosomal recessive
<i>LARGE1 (LARGE)</i>	603590	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840 (3), Autosomal recessive
<i>LMNB1</i>	150340	Leukodystrophy, adult-onset, autosomal dominant, 169500 (3), Autosomal dominant
<i>MAN2B1</i>	609458	Mannosidosis, alpha-, types I and II, 248500 (3), Autosomal recessive
<i>MANBA</i>	609489	Mannosidosis, beta, 248510 (3), Autosomal recessive
<i>MARS2</i>	609728	?Combined oxidative phosphorylation deficiency 25, 616430 (3), Autosomal recessive; Spastic ataxia 3, autosomal recessive, 611390 (3), Autosomal recessive
<i>MCOLN1</i>	605248	Mucopolipidosis IV, 252650 (3), Autosomal recessive
<i>MFSD8</i>	611124	Ceroid lipofuscinosis, neuronal, 7, 610951 (3), Autosomal recessive; Macular dystrophy with central cone involvement, 616170 (3), Autosomal recessive
<i>MLC1</i>	605908	Megalencephalic leukoencephalopathy with subcortical cysts, 604004 (3), Autosomal recessive
<i>MOCS1</i>	603707	Molybdenum cofactor deficiency A, 252150 (3), Autosomal recessive
<i>MOCS2</i>	603708	Molybdenum cofactor deficiency B, 252160 (3), Autosomal recessive
<i>MPLKIP</i>	609188	Trichothiodystrophy 4, nonphotosensitive, 234050 (3), Autosomal recessive
<i>MTHFR</i>	607093	Homocystinuria due to MTHFR deficiency, 236250 (3), Autosomal recessive; {Neural tube defects, susceptibility to}, 601634 (3), Autosomal recessive; {Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant; {Thromboembolism, susceptibility to}, 188050 (3), Autosomal dominant; {Vascular disease, susceptibility to} (3)
<i>MUT</i>	609058	Methylmalonic aciduria, mut(0) type, 251000 (3), Autosomal recessive
<i>NDUFS1</i>	157655	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
<i>NEU1</i>	608272	Sialidosis, type I, 256550 (3), Autosomal recessive; Sialidosis, type II, 256550 (3), Autosomal recessive
<i>NOTCH3</i>	600276	Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310 (3), Autosomal dominant; Lateral meningocele syndrome, 130720 (3), Autosomal dominant; ?Myofibromatosis, infantile 2, 615293 (3), Autosomal dominant
<i>NPC1</i>	607623	Niemann-Pick disease, type C1, 257220 (3), Autosomal recessive; Niemann-Pick disease, type D, 257220 (3), Autosomal recessive

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<i>NPC2</i>	601015	Niemann-pick disease, type C2, 607625 (3), Autosomal recessive
<i>NUBPL</i>	613621	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
<i>OCLN</i>	602876	Pseudo-TORCH syndrome 1, 251290 (3), Autosomal recessive
<i>OCRL</i>	300535	Dent disease 2, 300555 (3), X-linked recessive; Lowe syndrome, 309000 (3), X-linked recessive
<i>PAH</i>	612349	[Hyperphenylalaninemia, non-PKU mild], 261600 (3), Autosomal recessive; Phenylketonuria, 261600 (3), Autosomal recessive
<i>PCCA</i>	232000	Propionicacidemia, 606054 (3), Autosomal recessive
<i>PCCB</i>	232050	Propionicacidemia, 606054 (3), Autosomal recessive
<i>PDGFB</i>	190040	Basal ganglia calcification, idiopathic, 5, 615483 (3), Autosomal dominant; Dermatofibrosarcoma protuberans, 607907 (3); Meningioma, SIS-related, 607174 (3), Autosomal dominant
<i>PDGFRB</i>	173410	Basal ganglia calcification, idiopathic, 4, 615007 (3), Autosomal dominant; Kosaki overgrowth syndrome, 616592 (3), Autosomal dominant; Myeloproliferative disorder with eosinophilia, 131440 (4), Autosomal dominant; Myofibromatosis, infantile, 1, 228550 (3), Autosomal dominant; Premature aging syndrome, Penttinen type, 601812 (3), Autosomal dominant
<i>PDHA1</i>	300502	Pyruvate dehydrogenase E1-alpha deficiency, 312170 (3), X-linked dominant
<i>PDHB</i>	179060	Pyruvate dehydrogenase E1-beta deficiency, 614111 (3)
<i>PDHX</i>	608769	Lacticacidemia due to PDX1 deficiency, 245349 (3), Autosomal recessive
<i>PDP1</i>	605993	Pyruvate dehydrogenase phosphatase deficiency, 608782 (3), Autosomal recessive
<i>PEX1</i>	602136	Heimler syndrome 1, 234580 (3), Autosomal recessive; Peroxisome biogenesis disorder 1A (Zellweger), 214100 (3), Autosomal recessive; Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 (3), Autosomal recessive
<i>PEX10</i>	602859	Peroxisome biogenesis disorder 6A (Zellweger), 614870 (3), Autosomal recessive; Peroxisome biogenesis disorder 6B, 614871 (3), Autosomal recessive
<i>PEX11B</i>	603867	?Peroxisome biogenesis disorder 14B, 614920 (3), Autosomal recessive
<i>PEX12</i>	601758	Peroxisome biogenesis disorder 3A (Zellweger), 614859 (3), Autosomal recessive; Peroxisome biogenesis disorder 3B, 266510 (3), Autosomal recessive
<i>PEX13</i>	601789	Peroxisome biogenesis disorder 11A (Zellweger), 614883 (3), Autosomal recessive; Peroxisome biogenesis disorder 11B, 614885 (3), Autosomal recessive
<i>PEX14</i>	601791	Peroxisome biogenesis disorder 13A (Zellweger), 614887 (3), Autosomal recessive
<i>PEX16</i>	603360	Peroxisome biogenesis disorder 8A (Zellweger), 614876 (3), Autosomal recessive; Peroxisome biogenesis disorder 8B, 614877 (3), Autosomal recessive

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<i>PEX19</i>	600279	Peroxisome biogenesis disorder 12A (Zellweger), 614886 (3), Autosomal recessive
<i>PEX2</i>	170993	Peroxisome biogenesis disorder 5A (Zellweger), 614866 (3), Autosomal recessive; Peroxisome biogenesis disorder 5B, 614867 (3), Autosomal recessive
<i>PEX26</i>	608666	Peroxisome biogenesis disorder 7A (Zellweger), 614872 (3), Autosomal recessive; Peroxisome biogenesis disorder 7B, 614873 (3), Autosomal recessive
<i>PEX3</i>	603164	Peroxisome biogenesis disorder 10A (Zellweger), 614882 (3), Autosomal recessive; ?Peroxisome biogenesis disorder 10B, 617370 (3), Autosomal recessive
<i>PEX5</i>	600414	Peroxisome biogenesis disorder 2A (Zellweger), 214110 (3), Autosomal recessive; Peroxisome biogenesis disorder 2B, 202370 (3), Autosomal recessive; Rhizomelic chondrodysplasia punctata, type 5, 616716 (3), Autosomal recessive
<i>PEX6</i>	601498	Heimler syndrome 2, 616617 (3), Autosomal recessive; Peroxisome biogenesis disorder 4A (Zellweger), 614862 (3), Autosomal recessive; Peroxisome biogenesis disorder 4B, 614863 (3), Autosomal recessive, Autosomal dominant
<i>PEX7</i>	601757	Peroxisome biogenesis disorder 9B, 614879 (3); Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive
<i>PLP1</i>	300401	Pelizaeus-Merzbacher disease, 312080 (3), X-linked recessive; Spastic paraplegia 2, X-linked, 312920 (3), X-linked recessive
<i>POLG</i>	174763	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 (3), Autosomal recessive; Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 (3), Autosomal recessive; Progressive external ophthalmoplegia, autosomal dominant 1, 157640 (3), Autosomal dominant; Progressive external ophthalmoplegia, autosomal recessive 1, 258450 (3), Autosomal recessive
<i>POLR1C</i>	610060	Leukodystrophy, hypomyelinating, 11, 616494 (3), Autosomal recessive; Treacher Collins syndrome 3, 248390 (3), Autosomal recessive
<i>POLR3A</i>	614258	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3), Autosomal recessive
<i>POLR3B</i>	614366	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381 (3), Autosomal recessive



<i>POMGNT1</i>	606822	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 (3), Autosomal recessive; 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
<i>POMT1</i>	607423	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; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 (3), Autosomal recessive
<i>POMT2</i>	607439	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; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 (3), Autosomal recessive
<i>PPT1</i>	600722	Ceroid lipofuscinosis, neuronal, 1, 256730 (3), Autosomal recessive
<i>PRF1</i>	170280	Aplastic anemia, 609135 (3); Hemophagocytic lymphohistiocytosis, familial, 2, 603553 (3), Autosomal recessive; Lymphoma, non-Hodgkin, 605027 (3)
<i>PSAP</i>	176801	Combined SAP deficiency, 611721 (3), Autosomal recessive; Gaucher disease, atypical, 610539 (3); Krabbe disease, atypical, 611722 (3), Autosomal recessive; Metachromatic leukodystrophy due to SAP-b deficiency, 249900 (3), Autosomal recessive
<i>PSAT1</i>	610936	Neu-Laxova syndrome 2, 616038 (3), Autosomal recessive; ?Phosphoserine aminotransferase deficiency, 610992 (3), Autosomal recessive
<i>PSPH</i>	172480	Phosphoserine phosphatase deficiency, 614023 (3), Autosomal recessive
<i>PYCR2</i>	616406	Leukodystrophy, hypomyelinating, 10, 616420 (3), Autosomal recessive
<i>QDPR</i>	612676	Hyperphenylalaninemia, BH4-deficient, C, 261630 (3), Autosomal recessive
<i>RARS</i>	107820	Leukodystrophy, hypomyelinating, 9, 616140 (3), Autosomal recessive
<i>RNASEH2A</i>	606034	Aicardi-Goutieres syndrome 4, 610333 (3), Autosomal recessive
<i>RNASEH2B</i>	610326	Aicardi-Goutieres syndrome 2, 610181 (3), Autosomal recessive
<i>RNASEH2C</i>	610330	Aicardi-Goutieres syndrome 3, 610329 (3), Autosomal recessive
<i>RNASET2</i>	612944	Leukoencephalopathy, cystic, without megalencephaly, 612951 (3), Autosomal recessive
<i>SAMHD1</i>	606754	Aicardi-Goutieres syndrome 5, 612952 (3), Autosomal recessive; ?Chilblain lupus 2, 614415 (3), Autosomal dominant
<i>SCP2</i>	184755	?Leukoencephalopathy with dystonia and motor neuropathy, 613724 (3), Autosomal recessive

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<i>SDHA</i>	600857	Cardiomyopathy, dilated, 1GG, 613642 (3); Leigh syndrome, 256000 (3), Autosomal recessive, Mitochondrial; Mitochondrial respiratory chain complex II deficiency, 252011 (3), Autosomal recessive; Parangliomas 5, 614165 (3), Autosomal dominant
<i>SDHAF1</i>	612848	Mitochondrial complex II deficiency, 252011 (3), Autosomal recessive
<i>SDHAF2</i>	613019	Parangliomas 2, 601650 (3), Autosomal dominant
<i>SDHB</i>	185470	Cowden syndrome 2, 612359 (3), Autosomal dominant; Gastrointestinal stromal tumor, 606764 (3), Autosomal dominant, Isolated cases; Paranglioma and gastric stromal sarcoma, 606864 (3); Parangliomas 4, 115310 (3), Autosomal dominant; Pheochromocytoma, 171300 (3), Autosomal dominant
<i>SDHD</i>	602690	Carcinoid tumors, intestinal, 114900 (3), Autosomal dominant; Cowden syndrome 3, 615106 (3); Merkel cell carcinoma, somatic (3); Mitochondrial complex II deficiency, 252011 (3), Autosomal recessive; Paranglioma and gastric stromal sarcoma, 606864 (3); Parangliomas 1, with or without deafness, 168000 (3), Autosomal dominant; Pheochromocytoma, 171300 (3), Autosomal dominant
<i>SLC16A2</i>	300095	Allan-Herndon-Dudley syndrome, 300523 (3), X-linked
<i>SLC17A5</i>	604322	Salla disease, 604369 (3), Autosomal recessive; Sialic acid storage disorder, infantile, 269920 (3), Autosomal recessive
<i>SLC20A2</i>	158378	Basal ganglia calcification, idiopathic, 1, 213600 (3), Autosomal dominant
<i>SLC25A12</i>	603667	Epileptic encephalopathy, early infantile, 39, 612949 (3), Autosomal recessive
<i>SLC7A2</i>	601872	No OMIM phenotype
<i>SOX10</i>	602229	PCWH syndrome, 609136 (3), Autosomal dominant; Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 (3), Autosomal dominant; Waardenburg syndrome, type 4C, 613266 (3), Autosomal dominant
<i>SPG11</i>	610844	Amyotrophic lateral sclerosis 5, juvenile, 602099 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2X, 616668 (3), Autosomal recessive; Spastic paraplegia 11, autosomal recessive, 604360 (3), Autosomal recessive
<i>STX11</i>	605014	Hemophagocytic lymphohistiocytosis, familial, 4, 603552 (3), Autosomal recessive
<i>STXBP2</i>	601717	Hemophagocytic lymphohistiocytosis, familial, 5, 613101 (3)
<i>SUMF1</i>	607939	Multiple sulfatase deficiency, 272200 (3), Autosomal recessive
<i>SUOX</i>	606887	Sulfite oxidase deficiency, 272300 (3), Autosomal recessive
<i>TMEM106B</i>	613413	Leukodystrophy, hypomyelinating, 16, 617964 (3), Autosomal dominant
<i>TPP1</i>	607998	Ceroid lipofuscinosis, neuronal, 2, 204500 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 7, 609270 (3), Autosomal recessive
<i>TREM2</i>	605086	Nasu-Hakola disease, 221770 (3), Autosomal recessive

<i>TREX1</i>	606609	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 (3), Autosomal recessive, Autosomal dominant; Chilblain lupus, 610448 (3), Autosomal dominant; {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant; Vasculopathy, retinal, with cerebral leukodystrophy, 192315 (3), Autosomal dominant
<i>TTR</i>	176300	Amyloidosis, hereditary, transthyretin-related, 105210 (3), Autosomal dominant; Carpal tunnel syndrome, familial, 115430 (3), Autosomal dominant; [Dystransthyretinemic hyperthyroxinemia], 145680 (3), Autosomal dominant
<i>TUBB4A</i>	602662	Dystonia 4, torsion, autosomal dominant, 128101 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 6, 612438 (3), Autosomal dominant
<i>TYMP</i>	131222	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041 (3), Autosomal recessive
<i>TYROBP</i>	604142	Nasu-Hakola disease, 221770 (3), Autosomal recessive
<i>UNC13D</i>	608897	Hemophagocytic lymphohistiocytosis, familial, 3, 608898 (3)
<i>VPS11</i>	608549	Leukodystrophy, hypomyelinating, 12, 616683 (3), Autosomal recessive
<i>XPR1</i>	605237	Basal ganglia calcification, idiopathic, 6, 616413 (3), Autosomal dominant
<i>ZFYVE26</i>	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: July 04, 2018

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.