Leukodystrophy panel

versie	16-Oct-2018 (179 genen)	Centrum voor Medische Genetica Gent

Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
AARS2	612035	Combined oxidative phosphorylation deficiency 8, 614096 (3), Autosomal recessive; Leukoencephalopathy, progressive, with ovarian failure, 615889 (3), Autosomal recessive
ABCD1	300371	Adrenoleukodystrophy, 300100 (3), X-linked recessive; Adrenomyeloneuropathy, adult, 300100 (3), X-linked recessive
ACOX1	609751	Peroxisomal acyl-CoA oxidase deficiency, 264470 (3), Autosomal recessive
ACP5	171640	Spondyloenchondrodysplasia with immune dysregulation, 607944 (3), Autosomal recessive
ADAR	146920	Aicardi-Goutieres syndrome 6, 615010 (3), Autosomal recessive; Dyschromatosis symmetrica hereditaria, 127400 (3), Autosomal dominant
ADGRG1	604110	Polymicrogyria, bilateral frontoparietal, 606854 (3), Autosomal recessive; Polymicrogyria, bilateral perisylvian, 615752 (3)
ADSL	608222	Adenylosuccinase deficiency, 103050 (3), Autosomal recessive
AGA	613228	Aspartylglucosaminuria, 208400 (3), Autosomal recessive
AIMP1	603605	Leukodystrophy, hypomyelinating, 3, 260600 (3), Autosomal recessive
ALDH3A2	609523	Sjogren-Larsson syndrome, 270200 (3), Autosomal recessive
ALDH5A1	610045	Succinic semialdehyde dehydrogenase deficiency, 271980 (3), Autosomal recessive
APP	104760	Alzheimer disease 1, familial, 104300 (3), Autosomal dominant; Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants, 605714 (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
ATP7A	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
ATP7B	606882	Wilson disease, 277900 (3), Autosomal recessive
BCAP31	300398	Deafness, dystonia, and cerebral hypomyelination, 300475 (3), X-linked 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
CLCN2	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
CLN3	607042	Ceroid lipofuscinosis, neuronal, 3, 204200 (3), Autosomal recessive
CLN5	608102	Ceroid lipofuscinosis, neuronal, 5, 256731 (3), Autosomal recessive
CLN6	606725	Ceroid lipofuscinosis, neuronal, 6, 601780 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300 (3), Autosomal recessive
CLN8	607837	Ceroid lipofuscinosis, neuronal, 8, 600143 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 (3), Autosomal recessive
COL4A1	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)
COL4A2	120090	{Hemorrhage, intracerebral, susceptibility to}, 614519 (3); Porencephaly 2, 614483 (3), Autosomal dominant
CSF1R	164770	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 (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
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
DLAT	608770	Pyruvate dehydrogenase E2 deficiency, 245348 (3), Autosomal recessive

DLD	238331	Dihydrolipoamide dehydrogenase deficiency, 246900 (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
EIF2B1	606686	Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive
EIF2B2	606454	Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive; Ovarioleukodystrophy, 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
EIF2B5	603945	Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive; Ovarioleukodystrophy, 603896 (3), Autosomal recessive
ERCC2	126340	?Cerebrooculofacioskeletal syndrome 2, 610756 (3), Autosomal recessive; Trichothiodystrophy 1, photosensitive, 601675 (3), Autosomal recessive; Xeroderma pigmentosum, group D, 278730 (3), Autosomal recessive
ERCC3	133510	Trichothiodystrophy 2, photosensitive, 616390 (3), Autosomal recessive; Xeroderma pigmentosum, group B, 610651 (3), Autosomal recessive
ERCC6	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
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	Combined oxidative phosphorylation deficiency 14, 614946 (3), Autosomal recessive; Spastic paraplegia 77, autosomal recessive, 617046 (3), Autosomal recessive

FHL1	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
FKTN	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
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
GFAP	137780	Alexander disease, 203450 (3), Autosomal dominant
GJA1	121014	Atrioventricular septal defect 3, 600309 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Erythrokeratodermia 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
GJC2	608803	Leukodystrophy, hypomyelinating, 2, 608804 (3), Autosomal recessive; Lymphedema, hereditary, IC, 613480 (3), Autosomal dominant; Spastic paraplegia 44, autosomal recessive, 613206 (3), Autosomal recessive
GLA	300644	Fabry disease, 301500 (3), X-linked; Fabry disease, cardiac variant, 301500 (3), X-linked
GLB1	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
GM2A	613109	GM2-gangliosidosis, AB variant, 272750 (3), Autosomal recessive

GNAS	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
GPHN (GEPH)	603930	Molybdenum cofactor deficiency C, 615501 (3), Autosomal recessive
GRN	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
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; [Hex A pseudodeficiency], 272800 (3), Autosomal recessive; Tay-Sachs disease, 272800 (3), Autosomal recessive
HEXB	606873	Sandhoff disease, infantile, juvenile, and adult forms, 268800 (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	Leukodystrophy, hypomyelinating, 4, 612233 (3), Autosomal recessive; Spastic paraplegia 13, autosomal dominant, 605280 (3), Autosomal dominant
HTRA1	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)
IFIH1	606951	Aicardi-Goutieres syndrome 7, 615846 (3), Autosomal dominant; Singleton-Merten syndrome 1, 182250 (3), Autosomal dominant
IKBKG	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)

IVD	607036	Isovaleric acidemia, 243500 (3), Autosomal recessive
JAM3	606871	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730 (3), Autosomal recessive
L2HGDH (LHGD)	609584	L-2-hydroxyglutaric aciduria, 236792 (3), Autosomal recessive
LAMA2	156225	Muscular dystrophy, congenital merosin-deficient, 607855 (3), Autosomal recessive; Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855 (3), Autosomal recessive
LARGE1 (LARGE)	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
LMNB1	150340	Leukodystrophy, adult-onset, autosomal dominant, 169500 (3), Autosomal dominant
MAN2B1	609458	Mannosidosis, alpha-, types I and II, 248500 (3), Autosomal recessive
MANBA	609489	Mannosidosis, beta, 248510 (3), Autosomal recessive
MARS2	609728	?Combined oxidative phosphorylation deficiency 25, 616430 (3), Autosomal recessive; Spastic ataxia 3, autosomal recessive, 611390 (3), Autosomal recessive
MCOLN1	605248	Mucolipidosis IV, 252650 (3), Autosomal recessive
MFSD8	611124	Ceroid lipofuscinosis, neuronal, 7, 610951 (3), Autosomal recessive; Macular dystrophy with central cone involvement, 616170 (3), Autosomal recessive
MLC1	605908	Megalencephalic leukoencephalopathy with subcortical cysts, 604004 (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
MTHFR	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)
MUT	609058	Methylmalonic aciduria, mut(0) type, 251000 (3), Autosomal recessive
NDUFS1	157655	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NEU1	608272	Sialidosis, type I, 256550 (3), Autosomal recessive; Sialidosis, type II, 256550 (3), Autosomal recessive
NOTCH3	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
NPC1	607623	Niemann-Pick disease, type C1, 257220 (3), Autosomal recessive; Niemann-Pick disease, type D, 257220 (3), Autosomal recessive

NPC2	601015	Niemann-pick disease, type C2, 607625 (3), Autosomal recessive
NUBPL	613621	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
OCLN	602876	Pseudo-TORCH syndrome 1, 251290 (3), Autosomal recessive
OCRL	300535	Dent disease 2, 300555 (3), X-linked recessive; Lowe syndrome, 309000 (3), X-linked recessive
PAH	612349	[Hyperphenylalaninemia, non-PKU mild], 261600 (3), Autosomal recessive; Phenylketonuria, 261600 (3), Autosomal recessive
PCCA	232000	Propionicacidemia, 606054 (3), Autosomal recessive
PCCB	232050	Propionicacidemia, 606054 (3), Autosomal recessive
PDGFB	190040	Basal ganglia calcification, idiopathic, 5, 615483 (3), Autosomal dominant; Dermatofibrosarcoma protuberans, 607907 (3); Meningioma, SIS-related, 607174 (3), Autosomal dominant
PDGFRB	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
PDHA1	300502	Pyruvate dehydrogenase E1-alpha deficiency, 312170 (3), X-linked dominant
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 1A (Zellweger), 214100 (3), Autosomal recessive; Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 (3), Autosomal recessive
PEX10	602859	Peroxisome biogenesis disorder 6A (Zellweger), 614870 (3), Autosomal recessive; Peroxisome biogenesis disorder 6B, 614871 (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 2A (Zellweger), 214110 (3), Autosomal recessive; Peroxisome biogenesis disorder 2B, 202370 (3), Autosomal recessive; Rhizomelic chondrodysplasia punctata, type 5, 616716 (3), Autosomal recessive
PEX6	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
PEX7	601757	Peroxisome biogenesis disorder 9B, 614879 (3); Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive
PLP1	300401	Pelizaeus-Merzbacher disease, 312080 (3), X-linked recessive; Spastic paraplegia 2, X-linked, 312920 (3), X-linked recessive
POLG	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
POLR1C	610060	Leukodystrophy, hypomyelinating, 11, 616494 (3), Autosomal recessive; Treacher Collins syndrome 3, 248390 (3), Autosomal recessive
POLR3A	614258	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3), Autosomal recessive
POLR3B	614366	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381 (3), Autosomal recessive

POMGNT1	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
POMT1	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
POMT2	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
PPT1	600722	Ceroid lipofuscinosis, neuronal, 1, 256730 (3), Autosomal recessive
PRF1	170280	Aplastic anemia, 609135 (3); Hemophagocytic lymphohistiocytosis, familial, 2, 603553 (3), Autosomal recessive; Lymphoma, non-Hodgkin, 605027 (3)
PSAP	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
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
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	Aicardi-Goutieres syndrome 5, 612952 (3), Autosomal recessive; ?Chilblain lupus 2, 614415 (3), Autosomal dominant
SCP2	184755	?Leukoencephalopathy with dystonia and motor neuropathy, 613724 (3), Autosomal recessive

SDHA	600857	Cardiomyopathy, dilated, 1GG, 613642 (3); Leigh syndrome, 256000 (3), Autosomal recessive, Mitochondrial; Mitochondrial respiratory chain complex II deficiency, 252011 (3), Autosomal recessive; Paragangliomas 5, 614165 (3), Autosomal dominant
SDHAF1	612848	Mitochondrial complex II deficiency, 252011 (3), Autosomal recessive
SDHAF2	613019	Paragangliomas 2, 601650 (3), Autosomal dominant
SDHB	185470	Cowden syndrome 2, 612359 (3), Autosomal dominant; Gastrointestinal stromal tumor, 606764 (3), Autosomal dominant, Isolated cases; Paraganglioma and gastric stromal sarcoma, 606864 (3); Paragangliomas 4, 115310 (3), Autosomal dominant; Pheochromocytoma, 171300 (3), Autosomal dominant
SDHD	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; Paraganglioma and gastric stromal sarcoma, 606864 (3); Paragangliomas 1, with or without deafness, 168000 (3), Autosomal dominant; Pheochromocytoma, 171300 (3), Autosomal dominant
SLC16A2	300095	Allan-Herndon-Dudley syndrome, 300523 (3), X-linked
SLC17A5	604322	Salla disease, 604369 (3), Autosomal recessive; Sialic acid storage disorder, infantile, 269920 (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
SLC7A2	601872	No OMIM phenotype
SOX10	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
SPG11	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
STX11	605014	Hemophagocytic lymphohistiocytosis, familial, 4, 603552 (3), Autosomal recessive
STXBP2	601717	Hemophagocytic lymphohistiocytosis, familial, 5, 613101 (3)
SUMF1	607939	Multiple sulfatase deficiency, 272200 (3), Autosomal recessive
SUOX	606887	Sulfite oxidase deficiency, 272300 (3), Autosomal recessive
TMEM106B	613413	Leukodystrophy, hypomyelinating, 16, 617964 (3), Autosomal dominant
TPP1	607998	Ceroid lipofuscinosis, neuronal, 2, 204500 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 7, 609270 (3), Autosomal recessive
TREM2	605086	Nasu-Hakola disease, 221770 (3), Autosomal recessive

TREX1	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
TTR	176300	Amyloidosis, hereditary, transthyretin-related, 105210 (3), Autosomal dominant; Carpal tunnel syndrome, familial, 115430 (3), Autosomal dominant; [Dystransthyretinemic hyperthyroxinemia], 145680 (3), Autosomal dominant
TUBB4A	602662	Dystonia 4, torsion, autosomal dominant, 128101 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 6, 612438 (3), Autosomal dominant
TYMP	131222	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041 (3), Autosomal recessive
TYROBP	604142	Nasu-Hakola disease, 221770 (3), Autosomal recessive
UNC13D	608897	Hemophagocytic lymphohistiocytosis, familial, 3, 608898 (3)
VPS11	608549	Leukodystrophy, hypomyelinating, 12, 616683 (3), Autosomal recessive
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: 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.