

# Leukodystrophy and Leukoencephalopathy

Gene panel

## Gene panel information

<b>Gene panel</b>	<b>Leukodystrophy and Leukoencephalopathy</b>
<b>Version</b>	3
<b>Total genes</b>	325
<b>Activation date</b>	Thursday 04 april 2024
<b>Publisher</b>	Center for Medical Genetics, Ghent

## Genes

Gene	% coding region covered*	OMIM gene id	OMIM Phenotypes
<b>AARS1</b>	99.99 %	601065	Developmental and epileptic encephalopathy 29, 616339 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2N, 613287 (3), Autosomal dominant; ?Leukoencephalopathy, hereditary diffuse, with spheroids 2, 619661 (3), Autosomal dominant; Trichothiodystrophy 8, nonphotosensitive, 619691 (3), Autosomal recessive
<b>AARS2</b>	99.98 %	612035	Leukoencephalopathy, progressive, with ovarian failure, 615889 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 8, 614096 (3), Autosomal recessive
<b>ABAT</b>	99.98 %	137150	GABA-transaminase deficiency, 613163 (3), Autosomal recessive
<b>ABCD1</b>	99.98 %	300371	Adrenoleukodystrophy, 300100 (3), X-linked recessive; Adrenomyeloneuropathy, adult, 300100 (3), X-linked recessive
<b>ABHD16A</b>	100 %	142620	Spastic paraplegia 86, autosomal recessive, 619735 (3), Autosomal recessive
<b>ACBD5</b>	99.97 %	616618	Retinal dystrophy with leukodystrophy, 618863 (3), Autosomal recessive
<b>ACER3</b>	99.76 %	617036	?Leukodystrophy, progressive, early childhood-onset, 617762 (3), Autosomal recessive
<b>ACOX1</b>	99.98 %	609751	Mitchell syndrome, 618960 (3), Autosomal dominant; Peroxisomal acyl-CoA oxidase deficiency, 264470 (3), Autosomal recessive
<b>ACP5</b>	100 %	171640	Spondyloenchondrodysplasia with immune dysregulation, 607944 (3), Autosomal recessive
<b>ACTL6B</b>	99.9 %	612458	Developmental and epileptic encephalopathy 76, 618468 (3), Autosomal recessive; Intellectual developmental disorder with severe speech and ambulation defects, 618470 (3), Autosomal dominant
<b>ADAR</b>	99.84 %	146920	Dyschromatosis symmetrica hereditaria, 127400 (3), Autosomal dominant; Aicardi-Goutieres syndrome 6, 615010 (3), Autosomal recessive
<b>ADGRG1</b>	99.9 %	604110	Polymicrogyria, bilateral frontoparietal, 606854 (3), Autosomal recessive; Polymicrogyria, bilateral perisylvian, 615752 (3)
<b>ADSL</b>	99.93 %	608222	Adenylosuccinase deficiency, 103050 (3), Autosomal recessive
<b>AGA</b>	99.92 %	613228	Aspartylglucosaminuria, 208400 (3), Autosomal recessive
<b>AIFM1</b>	99.92 %	300169	Combined oxidative phosphorylation deficiency 6, 300816 (3), X-linked recessive; Cowchock syndrome, 310490 (3), X-linked recessive; Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 (3), X-linked recessive; Deafness, X-linked 5, 300614 (3), X-linked recessive
<b>AIMP1</b>	99.97 %	603605	Leukodystrophy, hypomyelinating, 3, 260600 (3), Autosomal recessive
<b>AIMP2</b>	99.99 %	600859	Leukodystrophy, hypomyelinating, 17, 618006 (3), Autosomal recessive
<b>ALDH3A2</b>	99.95 %	609523	Sjogren-Larsson syndrome, 270200 (3), Autosomal recessive
<b>ALDH5A1</b>	96.19 %	610045	Succinic semialdehyde dehydrogenase deficiency, 271980 (3), Autosomal recessive
<b>ALPK1</b>	99.92 %	607347	ROSAH syndrome, 614979 (3), Autosomal dominant
<b>AP1S2</b>	99.56 %	300629	Pettigrew syndrome, 304340 (3), X-linked recessive

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<b>AP4B1</b>	96.92 %	607245	Spastic paraplegia 47, autosomal recessive, 614066 (3), Autosomal recessive
<b>APP</b>	99.92 %	104760	Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants, 605714 (3), Autosomal dominant; Alzheimer disease 1, familial, 104300 (3), Autosomal dominant
<b>ARSA</b>	99.99 %	607574	Metachromatic leukodystrophy, 250100 (3), Autosomal recessive
<b>ASAH1</b>	99.9 %	613468	Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 (3), Autosomal recessive; Farber lipogranulomatosis, 228000 (3), Autosomal recessive
<b>ASPA</b>	99.98 %	608034	Canavan disease, 271900 (3), Autosomal recessive
<b>ATAD3A</b>	99.62 %	612316	Harel-Yoon syndrome, 617183 (3), Autosomal recessive, Autosomal dominant; Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810 (3), Autosomal recessive
<b>ATP11A</b>	99.81 %	605868	?Auditory neuropathy, autosomal dominant 2, 620384 (3), Autosomal dominant; ?Leukodystrophy, hypomyelinating, 24, 619851 (3), Autosomal dominant; Deafness, autosomal dominant 84, 619810 (3), Autosomal dominant
<b>ATP7A</b>	99.87 %	300011	Occipital horn syndrome, 304150 (3), X-linked recessive; Spinal muscular atrophy, distal, X-linked 3, 300489 (3), X-linked recessive; Menkes disease, 309400 (3), X-linked recessive
<b>ATP7B</b>	100 %	606882	Wilson disease, 277900 (3), Autosomal recessive
<b>AUH</b>	99.95 %	600529	3-methylglutaconic aciduria, type I, 250950 (3), Autosomal recessive
<b>B3GALNT2</b>	92.79 %	610194	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 11, 615181 (3), Autosomal recessive
<b>BCAP31</b>	99.95 %	300398	Deafness, dystonia, and cerebral hypomyelination, 300475 (3), X-linked recessive
<b>BCS1L</b>	99.99 %	603647	GRACILE syndrome, 603358 (3), Autosomal recessive; Mitochondrial complex III deficiency, nuclear type 1, 124000 (3), Autosomal recessive; Bjornstad syndrome, 262000 (3), Autosomal recessive
<b>BLOC1S1</b>	99.95 %	601444	<i>No OMIM phenotypes</i>
<b>BOLA3</b>	99.22 %	613183	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299 (3), Autosomal recessive
<b>BTD</b>	100 %	609019	Biotinidase deficiency, 253260 (3), Autosomal recessive
<b>C2orf69</b>	99.97 %	619219	Combined oxidative phosphorylation deficiency 53, 619423 (3), Autosomal recessive
<b>CBS</b>	17.79 %	613381	Thrombosis, hyperhomocysteinemic, 236200 (3), Autosomal recessive; Homocystinuria, B6-responsive and nonresponsive types, 236200 (3), Autosomal recessive
<b>CIC</b>	98.32 %	612082	Intellectual developmental disorder, autosomal dominant 45, 617600 (3), Autosomal dominant
<b>CLCN2</b>	100 %	600570	Leukoencephalopathy with ataxia, 615651 (3), Autosomal recessive; Hyperaldosteronism, familial, type II, 605635 (3), Autosomal dominant; {Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628 (3), Autosomal dominant; {Epilepsy, juvenile absence, susceptibility to, 2}, 607628 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628 (3), Autosomal dominant
<b>CLDN11</b>	100 %	601326	Leukodystrophy, hypomyelinating, 22, 619328 (3), Autosomal dominant
<b>CLDN5</b>	100 %	602101	<i>No OMIM phenotypes</i>
<b>CLN3</b>	99.92 %	607042	Ceroid lipofuscinosis, neuronal, 3, 204200 (3), Autosomal recessive
<b>CLN5</b>	100 %	608102	Ceroid lipofuscinosis, neuronal, 5, 256731 (3), Autosomal recessive
<b>CLN6</b>	100 %	606725	Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 6A, 601780 (3), Autosomal recessive
<b>CLN8</b>	100 %	607837	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 8, 600143 (3), Autosomal recessive

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<b>CLPP</b>	99.99 %	601119	Perrault syndrome 3, 614129 (3), Autosomal recessive
<b>CNP</b>	99.98 %	123830	?Leukodystrophy, hypomyelinating, 20, 619071 (3), Autosomal recessive
<b>CNTNAP1</b>	99.98 %	602346	Lethal congenital contracture syndrome 7, 616286 (3), Autosomal recessive; Hypomyelinating neuropathy, congenital, 3, 618186 (3), Autosomal recessive
<b>COA7</b>	99.91 %	615623	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387 (3), Autosomal recessive
<b>COA8</b>	99.94 %	616003	Mitochondrial complex IV deficiency, nuclear type 17, 619061 (3), Autosomal recessive
<b>COL4A1</b>	99.99 %	120130	?Retinal arteries, tortuosity of, 180000 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3); Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 (3), Autosomal dominant; Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564 (3), Autosomal dominant; Brain small vessel disease with or without ocular anomalies, 175780 (3), Autosomal dominant
<b>COL4A2</b>	99.98 %	120090	Brain small vessel disease 2, 614483 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3)
<b>COLGALT1</b>	99.82 %	617531	Brain small vessel disease 3, 618360 (3), Autosomal recessive
<b>COQ9</b>	99.62 %	612837	Coenzyme Q10 deficiency, primary, 5, 614654 (3), Autosomal recessive
<b>COX10</b>	99.99 %	602125	Mitochondrial complex IV deficiency, nuclear type 3, 619046 (3), Autosomal recessive
<b>COX15</b>	100 %	603646	Mitochondrial complex IV deficiency, nuclear type 6, 615119 (3), Autosomal recessive
<b>CPS1</b>	99.91 %	608307	Carbamoylphosphate synthetase I deficiency, 237300 (3), Autosomal recessive; {Pulmonary hypertension, neonatal, susceptibility to}, 615371 (3)
<b>CSF1R</b>	99.92 %	164770	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 (3), Autosomal recessive; Leukoencephalopathy, diffuse hereditary, with spheroids 1, 221820 (3), Autosomal dominant
<b>CST3</b>	100 %	604312	{Macular degeneration, age-related, 11}, 611953 (3); Cerebral amyloid angiopathy, 105150 (3), Autosomal dominant
<b>CTC1</b>	100 %	613129	Cerebroretinal microangiopathy with calcifications and cysts, 612199 (3), Autosomal recessive
<b>CTSA</b>	99.98 %	613111	Galactosialidosis, 256540 (3), Autosomal recessive
<b>CTSD</b>	100 %	116840	Ceroid lipofuscinosis, neuronal, 10, 610127 (3), Autosomal recessive
<b>CTSF</b>	99.96 %	603539	Ceroid lipofuscinosis, neuronal, 13 (Kufs type), 615362 (3), Autosomal recessive
<b>CYP27A1</b>	100 %	606530	Cerebrotendinous xanthomatosis, 213700 (3), Autosomal recessive
<b>CYP2U1</b>	99.99 %	610670	Spastic paraplegia 56, autosomal recessive, 615030 (3), Autosomal recessive
<b>CYP7B1</b>	99.82 %	603711	Spastic paraplegia 5A, autosomal recessive, 270800 (3), Autosomal recessive; Bile acid synthesis defect, congenital, 3, 613812 (3), Autosomal recessive
<b>D2HGDH</b>	100 %	609186	D-2-hydroxyglutaric aciduria, 600721 (3), Autosomal recessive
<b>DAG1</b>	100 %	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
<b>DARS1</b>	98.85 %	603084	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281 (3), Autosomal recessive
<b>DARS2</b>	98.31 %	610956	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105 (3), Autosomal recessive
<b>DCAF17</b>	99.84 %	612515	Woodhouse-Sakati syndrome, 241080 (3), Autosomal recessive
<b>DDHD2</b>	99.97 %	615003	Spastic paraplegia 54, autosomal recessive, 615033 (3), Autosomal recessive
<b>DEGS1</b>	99.99 %	615843	Leukodystrophy, hypomyelinating, 18, 618404 (3), Autosomal recessive

# Leukodystrophy and Leukoencephalopathy

Gene panel

Gene	% coding region covered*	OMIM gene id	OMIM Phenotypes
<b>DGUOK</b>	99.93 %	601465	Portal hypertension, noncirrhotic, 1, 617068 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880 (3), Autosomal recessive
<b>DLAT</b>	99.65 %	608770	Pyruvate dehydrogenase E2 deficiency, 245348 (3), Autosomal recessive
<b>DLD</b>	99.89 %	238331	Dihydrolipoamide dehydrogenase deficiency, 246900 (3), Autosomal recessive
<b>DMXL2</b>	99.86 %	612186	Developmental and epileptic encephalopathy 81, 618663 (3), Autosomal recessive; ?Deafness, autosomal dominant 71, 617605 (3), Autosomal dominant; ?Polyendocrine-polyneuropathy syndrome, 616113 (3), Autosomal recessive
<b>DNAJC5</b>	99.99 %	611203	Ceroid lipofuscinosis, neuronal, 4 (Kufs type), autosomal dominant, 162350 (3), Autosomal dominant
<b>DPYD</b>	94.53 %	612779	Dihydropyrimidine dehydrogenase deficiency, 274270 (3), Autosomal recessive; 5-fluorouracil toxicity, 274270 (3), Autosomal recessive
<b>EARS2</b>	99.96 %	612799	Combined oxidative phosphorylation deficiency 12, 614924 (3), Autosomal recessive
<b>ECHS1</b>	100 %	602292	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277 (3), Autosomal recessive
<b>EIF2AK1</b>	99.9 %	613635	?Leukoencephalopathy, motor delay, spasticity, and dysarthria syndrome, 618878 (3), Autosomal dominant
<b>EIF2AK2</b>	99.7 %	176871	Leukoencephalopathy, developmental delay, and episodic neurologic regression syndrome, 618877 (3), Autosomal dominant; Dystonia 33, 619687 (3), Autosomal recessive, Autosomal dominant
<b>EIF2B1</b>	99.98 %	606686	Leukoencephalopathy with vanishing white matter 1, with or without ovarian failure, 603896 (3), Autosomal recessive
<b>EIF2B2</b>	99.9 %	606454	Leukoencephalopathy with vanishing white matter 2, with or without ovarian failure, 620312 (3)
<b>EIF2B3</b>	97.26 %	606273	Leukoencephalopathy with vanishing white matter 3, with or without ovarian failure, 620313 (3)
<b>EIF2B4</b>	99.96 %	606687	Leukoencephalopathy with vanishing white matter 4, with or without ovarian failure, 620314 (3)
<b>EIF2B5</b>	99.98 %	603945	Leukoencephalopathy with vanishing white matter 5, with or without ovarian failure, 620315 (3)
<b>ELOVL1</b>	100 %	611813	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527 (3), Autosomal recessive, Autosomal dominant
<b>ENTPD1</b>	99.98 %	601752	Spastic paraplegia 64, autosomal recessive, 615683 (3), Autosomal recessive
<b>EPRS1</b>	99.53 %	138295	Leukodystrophy, hypomyelinating, 15, 617951 (3), Autosomal recessive
<b>ERCC2</b>	99.98 %	126340	Xeroderma pigmentosum, group D, 278730 (3), Autosomal recessive; Trichothiodystrophy 1, photosensitive, 601675 (3), Autosomal recessive; ?Cerebrooculofacioskeletal syndrome 2, 610756 (3), Autosomal recessive
<b>ERCC3</b>	99.9 %	133510	Trichothiodystrophy 2, photosensitive, 616390 (3), Autosomal recessive; Xeroderma pigmentosum, group B, 610651 (3), Autosomal recessive
<b>ERCC6</b>	99.6 %	609413	UV-sensitive syndrome 1, 600630 (3), Autosomal recessive; Cerebrooculofacioskeletal syndrome 1, 214150 (3), Autosomal recessive; ?De Sanctis-Cacchione syndrome, 278800 (3), Autosomal recessive; Cockayne syndrome, type B, 133540 (3), Autosomal recessive; {Macular degeneration, age-related, susceptibility to, 5}, 613761 (3); Premature ovarian failure 11, 616946 (3), Autosomal dominant; {Lung cancer, susceptibility to}, 211980 (3), Somatic mutation, Autosomal dominant
<b>ERCC8</b>	99.79 %	609412	UV-sensitive syndrome 2, 614621 (3), Autosomal recessive; Cockayne syndrome, type A, 216400 (3), Autosomal recessive

# Leukodystrophy and Leukoencephalopathy

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Gene	% coding region covered*	OMIM gene id	OMIM Phenotypes
<b>ESAM</b>	99.99 %	614281	Neurodevelopmental disorder with intracranial hemorrhage, seizures, and spasticity, 620371 (3), Autosomal recessive
<b>FA2H</b>	99.98 %	611026	Spastic paraplegia 35, autosomal recessive, 612319 (3), Autosomal recessive
<b>FAM126A</b>	99.81 %	610531	Leukodystrophy, hypomyelinating, 5, 610532 (3), Autosomal recessive
<b>FAR1</b>	99.82 %	616107	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154 (3), Autosomal recessive; Cataracts, spastic paraparesis, and speech delay, 619338 (3), Autosomal dominant
<b>FARS2</b>	100 %	611592	Combined oxidative phosphorylation deficiency 14, 614946 (3), Autosomal recessive; Spastic paraplegia 77, autosomal recessive, 617046 (3), Autosomal recessive
<b>FASTKD2</b>	99.93 %	612322	Combined oxidative phosphorylation deficiency 44, 618855 (3), Autosomal recessive
<b>FBXL4</b>	100 %	605654	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471 (3), Autosomal recessive
<b>FDX2</b>	99.99 %	614585	Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900 (3), Autosomal recessive
<b>FHL1</b>	99.97 %	300163	Myopathy, X-linked, with postural muscle atrophy, 300696 (3), X-linked recessive; Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 (3), X-linked recessive; ?Uruguay faciocardiomusculoskeletal syndrome, 300280 (3), X-linked recessive; 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; Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717 (3), X-linked dominant
<b>FIG4</b>	99.83 %	609390	Yunis-Varon syndrome, 216340 (3), Autosomal recessive; ?Polymicrogyria, bilateral temporooccipital, 612691 (3), Autosomal recessive; Amyotrophic lateral sclerosis 11, 612577 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 4j, 611228 (3), Autosomal recessive
<b>FKTN</b>	99.94 %	607440	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 (3), Autosomal recessive; Cardiomyopathy, dilated, 1X, 611615 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital without impaired intellectual development), type B, 4, 613152 (3), Autosomal recessive
<b>FLVCR2</b>	100 %	610865	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790 (3), Autosomal recessive
<b>FOLR1</b>	100 %	136430	Neurodegeneration due to cerebral folate transport deficiency, 613068 (3), Autosomal recessive
<b>FUCA1</b>	98.72 %	612280	Fucosidosis, 230000 (3), Autosomal recessive
<b>GALC</b>	99.92 %	606890	Krabbe disease, 245200 (3), Autosomal recessive
<b>GALNT2</b>	99.44 %	602274	Congenital disorder of glycosylation, type IIt, 618885 (3), Autosomal recessive
<b>GALT</b>	100 %	606999	Galactosemia, 230400 (3), Autosomal recessive
<b>GAN</b>	99.98 %	605379	Giant axonal neuropathy-1, 256850 (3), Autosomal recessive
<b>GBE1</b>	99.73 %	607839	Glycogen storage disease IV, 232500 (3), Autosomal recessive; Polyglucosan body disease, adult form, 263570 (3), Autosomal recessive
<b>GCDH</b>	100 %	608801	Glutaricaciduria, type I, 231670 (3), Autosomal recessive
<b>GFAP</b>	99.99 %	137780	Alexander disease, 203450 (3), Autosomal dominant
<b>GFM1</b>	99.95 %	606639	Combined oxidative phosphorylation deficiency 1, 609060 (3), Autosomal recessive
<b>GFM2</b>	99.87 %	606544	Combined oxidative phosphorylation deficiency 39, 618397 (3), Autosomal recessive

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<b>GJA1</b>	100 %	121014	Erythrokeratoderma variabilis et progressiva 3, 617525 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Oculodentodigital dysplasia, 164200 (3), Autosomal dominant; Palmoplantar keratoderma with congenital alopecia, 104100 (3), Autosomal dominant; Syndactyly, type III, 186100 (3), Autosomal dominant; Oculodentodigital dysplasia, autosomal recessive, 257850 (3), Autosomal recessive
<b>GJB1</b>	100 %	304040	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800 (3), X-linked dominant
<b>GJC2</b>	100 %	608803	Lymphatic malformation 3, 613480 (3), Autosomal dominant; ?Spastic paraplegia 44, autosomal recessive, 613206 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 2, 608804 (3), Autosomal recessive
<b>GLA</b>	99.9 %	300644	Fabry disease, cardiac variant, 301500 (3), X-linked; Fabry disease, 301500 (3), X-linked
<b>GLB1</b>	100 %	611458	GM1-gangliosidosis, type I, 230500 (3), Autosomal recessive; GM1-gangliosidosis, type III, 230650 (3), Autosomal recessive; Mucopolysaccharidosis type IVB (Morquio), 253010 (3), Autosomal recessive; GM1-gangliosidosis, type II, 230600 (3), Autosomal recessive
<b>GLRX5</b>	100 %	609588	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 (3), Autosomal recessive; Spasticity, childhood-onset, with hyperglycinemia, 616859 (3), Autosomal recessive
<b>GM2A</b>	100 %	613109	GM2-gangliosidosis, AB variant, 272750 (3), Autosomal recessive
<b>GNAS</b>	100 %	139320	ACTH-independent macronodular adrenal hyperplasia, 219080 (3), Somatic mutation; Pituitary adenoma 3, multiple types, somatic, 617686 (3); Pseudohypoparathyroidism 1c, 612462 (3), Autosomal dominant; Pseudohypoparathyroidism 1a, 103580 (3), Autosomal dominant; Osseous heteroplasia, progressive, 166350 (3), Autosomal dominant; Pseudohypoparathyroidism 1b, 603233 (3), Autosomal dominant; McCune-Albright syndrome, somatic, mosaic, 174800 (3); Pseudopseudohypoparathyroidism, 612463 (3), Autosomal dominant
<b>GPHN</b>	99.94 %	603930	Molybdenum cofactor deficiency C, 615501 (3), Autosomal recessive
<b>GSN</b>	99.93 %	137350	Amyloidosis, Finnish type, 105120 (3), Autosomal dominant
<b>GTF2H5</b>	100 %	608780	Trichothiodystrophy 3, photosensitive, 616395 (3), Autosomal recessive
<b>HEPACAM</b>	100 %	611642	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 (3), Autosomal recessive; Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without impaired intellectual development, 613926 (3), Autosomal dominant
<b>HEXA</b>	99.99 %	606869	[Hex A pseudodeficiency], 272800 (3), Autosomal recessive; GM2-gangliosidosis, several forms, 272800 (3), Autosomal recessive; Tay-Sachs disease, 272800 (3), Autosomal recessive
<b>HEXB</b>	99.91 %	606873	Sandhoff disease, infantile, juvenile, and adult forms, 268800 (3), Autosomal recessive
<b>HIKESHI</b>	99.78 %	614908	Leukodystrophy, hypomyelinating, 13, 616881 (3), Autosomal recessive
<b>HLCS</b>	99.97 %	609018	Holocarboxylase synthetase deficiency, 253270 (3), Autosomal recessive
<b>HMBS</b>	99.97 %	609806	Porphyria, acute intermittent, nonerythroid variant, 176000 (3), Autosomal dominant; Porphyria, acute intermittent, 176000 (3), Autosomal dominant
<b>HMGCL</b>	99.31 %	613898	HMG-CoA lyase deficiency, 246450 (3), Autosomal recessive
<b>HPDL</b>	99.99 %	618994	Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026 (3), Autosomal recessive; Spastic paraplegia 83, autosomal recessive, 619027 (3), Autosomal recessive
<b>HSD17B4</b>	99.71 %	601860	D-bifunctional protein deficiency, 261515 (3), Autosomal recessive; Perrault syndrome 1, 233400 (3), Autosomal recessive

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Gene panel

Gene	% coding region covered*	OMIM gene id	OMIM Phenotypes
<b>HSPD1</b>	83.42 %	118190	Spastic paraplegia 13, autosomal dominant, 605280 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 4, 612233 (3), Autosomal recessive
<b>HTRA1</b>	100 %	602194	{Macular degeneration, age-related, neovascular type}, 610149 (3); {Macular degeneration, age-related, 7}, 610149 (3); CARASIL syndrome, 600142 (3), Autosomal recessive; Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779 (3), Autosomal dominant
<b>IBA57</b>	100 %	615316	Multiple mitochondrial dysfunctions syndrome 3, 615330 (3), Autosomal recessive; ?Spastic paraplegia 74, autosomal recessive, 616451 (3), Autosomal recessive
<b>IFIH1</b>	99.84 %	606951	Immunodeficiency 95, 619773 (3), Autosomal recessive; Aicardi-Goutieres syndrome 7, 615846 (3), Autosomal dominant; Singleton-Merten syndrome 1, 182250 (3), Autosomal dominant
<b>IKBKG</b>	57.34 %	300248	Incontinentia pigmenti, 308300 (3), X-linked dominant; Ectodermal dysplasia and immunodeficiency 1, 300291 (3), X-linked recessive; Immunodeficiency 33, 300636 (3), X-linked recessive; Autoinflammatory disease, systemic, X-linked, 301081 (3), X-linked
<b>ISCA1</b>	99.79 %	611006	Multiple mitochondrial dysfunctions syndrome 5, 617613 (3), Autosomal recessive
<b>ISCA2</b>	100 %	615317	Multiple mitochondrial dysfunctions syndrome 4, 616370 (3), Autosomal recessive
<b>ITM2B</b>	99.85 %	603904	?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079 (3), Autosomal dominant; Dementia, familial British, 176500 (3), Autosomal dominant; Dementia, familial Danish, 117300 (3), Autosomal dominant
<b>IVD</b>	100 %	607036	Isovaleric acidemia, 243500 (3), Autosomal recessive
<b>JAM3</b>	100 %	606871	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730 (3), Autosomal recessive
<b>KARS1</b>	99.98 %	601421	Deafness, autosomal recessive 89, 613916 (3), Autosomal recessive; Leukoencephalopathy, progressive, infantile-onset, with or without deafness, 619147 (3), Autosomal recessive; ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 (3), Autosomal recessive; Deafness, congenital, and adult-onset progressive leukoencephalopathy, 619196 (3), Autosomal recessive
<b>KIF5A</b>	99.91 %	602821	Myoclonus, intractable, neonatal, 617235 (3), Autosomal dominant; {Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921 (3), Autosomal dominant; Spastic paraplegia 10, autosomal dominant, 604187 (3), Autosomal dominant
<b>L2HGDH</b>	99.92 %	609584	L-2-hydroxyglutaric aciduria, 236792 (3), Autosomal recessive
<b>LAMA2</b>	99.95 %	156225	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 (3), Autosomal recessive; Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855 (3), Autosomal recessive
<b>LAMB1</b>	99.87 %	150240	Lissencephaly 5, 615191 (3), Autosomal recessive
<b>LARGE1</b>	100 %	603590	Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 6, 608840 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 (3), Autosomal recessive
<b>LARS2</b>	99.96 %	604544	Perrault syndrome 4, 615300 (3), Autosomal recessive; Hydrops, lactic acidosis, and sideroblastic anemia, 617021 (3), Autosomal recessive
<b>LIG3</b>	99.99 %	600940	Mitochondrial DNA depletion syndrome 20 (MNGIE type), 619780 (3), Autosomal recessive
<b>LIPT1</b>	99.89 %	610284	Lipoyltransferase 1 deficiency, 616299 (3), Autosomal recessive
<b>LIPT2</b>	99.99 %	617659	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668 (3), Autosomal recessive
<b>LMNB1</b>	99.73 %	150340	Leukodystrophy, adult-onset, autosomal dominant, 169500 (3), Autosomal dominant; Microcephaly 26, primary, autosomal dominant, 619179 (3), Autosomal dominant

# Leukodystrophy and Leukoencephalopathy

Gene panel

Gene	% coding region covered*	OMIM gene id	OMIM Phenotypes
<b>LYRM7</b>	99.98 %	615831	Mitochondrial complex III deficiency, nuclear type 8, 615838 (3), Autosomal recessive
<b>MAL</b>	99.87 %	188860	No OMIM phenotypes
<b>MAN2B1</b>	99.99 %	609458	Mannosidosis, alpha-, types I and II, 248500 (3), Autosomal recessive
<b>MANBA</b>	99.81 %	609489	Mannosidosis, beta, 248510 (3), Autosomal recessive
<b>MARS2</b>	100 %	609728	?Combined oxidative phosphorylation deficiency 25, 616430 (3), Autosomal recessive; Spastic ataxia 3, autosomal recessive, 611390 (3), Autosomal recessive
<b>MCOLN1</b>	100 %	605248	Mucopolipidosis IV, 252650 (3), Autosomal recessive
<b>MEF2C</b>	99.57 %	600662	Chromosome 5q14.3 deletion syndrome, 613443 (4), Autosomal dominant; Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language, 613443 (3), Autosomal dominant
<b>MFSD8</b>	99.7 %	611124	Macular dystrophy with central cone involvement, 616170 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 7, 610951 (3), Autosomal recessive
<b>MLC1</b>	99.99 %	605908	Megalencephalic leukoencephalopathy with subcortical cysts 1, 604004 (3), Autosomal recessive
<b>MMUT</b>	99.68 %	609058	Methylmalonic aciduria, mut(0) type, 251000 (3), Autosomal recessive
<b>MOCS1</b>	99.95 %	603707	Molybdenum cofactor deficiency A, 252150 (3), Autosomal recessive
<b>MOCS2</b>	99.96 %	603708	Molybdenum cofactor deficiency B, 252160 (3), Autosomal recessive
<b>MPLKIP</b>	99.99 %	609188	Trichothiodystrophy 4, nonphotosensitive, 234050 (3), Autosomal recessive
<b>MPV17</b>	99.98 %	137960	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810 (3), Autosomal recessive
<b>MRPS22</b>	99.87 %	605810	Ovarian dysgenesis 7, 618117 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 5, 611719 (3), Autosomal recessive
<b>MRPS34</b>	100 %	611994	Combined oxidative phosphorylation deficiency 32, 617664 (3), Autosomal recessive
<b>MTFMT</b>	99.98 %	611766	Combined oxidative phosphorylation deficiency 15, 614947 (3), Autosomal recessive; Mitochondrial complex I deficiency, nuclear type 27, 618248 (3), Autosomal recessive
<b>MTHFR</b>	99.97 %	607093	{Vascular disease, susceptibility to} (3); Homocystinuria due to MTHFR deficiency, 236250 (3), Autosomal recessive; {Thromboembolism, susceptibility to}, 188050 (3), Autosomal dominant; {Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant; {Neural tube defects, susceptibility to}, 601634 (3), Autosomal recessive
<b>MTO1</b>	90.25 %	614667	Combined oxidative phosphorylation deficiency 10, 614702 (3), Autosomal recessive
<b>MYORG</b>	99.99 %	618255	Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317 (3), Autosomal recessive
<b>NAXD</b>	99.99 %	615910	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321 (3), Autosomal recessive
<b>NAXE</b>	99.99 %	608862	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186 (3), Autosomal recessive
<b>NDUFA2</b>	99.95 %	602137	Mitochondrial complex I deficiency, nuclear type 13, 618235 (3), Autosomal recessive
<b>NDUFA9</b>	100 %	603834	Mitochondrial complex I deficiency, nuclear type 26, 618247 (3), Autosomal recessive
<b>NDUFAF1</b>	100 %	606934	Mitochondrial complex I deficiency, nuclear type 11, 618234 (3), Autosomal recessive
<b>NDUFAF3</b>	100 %	612911	Mitochondrial complex I deficiency, nuclear type 18, 618240 (3), Autosomal recessive
<b>NDUFAF4</b>	99.95 %	611776	Mitochondrial complex I deficiency, nuclear type 15, 618237 (3), Autosomal recessive
<b>NDUFAF6</b>	99.86 %	612392	Mitochondrial complex I deficiency, nuclear type 17, 618239 (3), Autosomal recessive; Fanconi renotubular syndrome 5, 618913 (3), Autosomal recessive
<b>NDUFS1</b>	99.79 %	157655	Mitochondrial complex I deficiency, nuclear type 5, 618226 (3), Autosomal recessive
<b>NDUFS2</b>	99.66 %	602985	Mitochondrial complex I deficiency, nuclear type 6, 618228 (3), Autosomal recessive
<b>NDUFS4</b>	99.99 %	602694	Mitochondrial complex I deficiency, nuclear type 1, 252010 (3), Autosomal recessive



# Leukodystrophy and Leukoencephalopathy

Gene panel

Gene	% coding region covered*	OMIM gene id	OMIM Phenotypes
<b>NDUFS7</b>	99.99 %	601825	Mitochondrial complex I deficiency, nuclear type 3, 618224 (3), Autosomal recessive
<b>NDUFS8</b>	100 %	602141	Mitochondrial complex I deficiency, nuclear type 2, 618222 (3), Autosomal recessive
<b>NDUFV1</b>	99.99 %	161015	Mitochondrial complex I deficiency, nuclear type 4, 618225 (3), Autosomal recessive
<b>NDUFV2</b>	99.98 %	600532	Mitochondrial complex I deficiency, nuclear type 7, 618229 (3), Autosomal recessive
<b>NEU1</b>	99.98 %	608272	Sialidosis, type II, 256550 (3), Autosomal recessive; Sialidosis, type I, 256550 (3), Autosomal recessive
<b>NFU1</b>	99.48 %	608100	Multiple mitochondrial dysfunctions syndrome 1, 605711 (3), Autosomal recessive
<b>NKX6-2</b>	100 %	605955	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560 (3), Autosomal recessive
<b>NOTCH3</b>	99.99 %	600276	Lateral meningocele syndrome, 130720 (3), Autosomal dominant; ?Myofibromatosis, infantile 2, 615293 (3), Autosomal dominant; Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310 (3), Autosomal dominant
<b>NPC1</b>	99.99 %	607623	Niemann-Pick disease, type C1, 257220 (3), Autosomal recessive; Niemann-Pick disease, type D, 257220 (3), Autosomal recessive
<b>NPC2</b>	100 %	601015	Niemann-pick disease, type C2, 607625 (3), Autosomal recessive
<b>NTNG2</b>	99.98 %	618689	Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718 (3), Autosomal recessive
<b>NUBPL</b>	99.62 %	613621	Mitochondrial complex I deficiency, nuclear type 21, 618242 (3), Autosomal recessive
<b>NUP188</b>	99.88 %	615587	Sandestig-Stefanova syndrome, 618804 (3), Autosomal recessive
<b>OCLN</b>	82.91 %	602876	Pseudo-TORCH syndrome 1, 251290 (3), Autosomal recessive
<b>OCRL</b>	99.89 %	300535	Dent disease 2, 300555 (3), X-linked recessive; Lowe syndrome, 309000 (3), X-linked recessive
<b>PAFAH1B1</b>	99.96 %	601545	Subcortical laminar heterotopia, 607432 (3), Autosomal dominant; Lissencephaly 1, 607432 (3), Autosomal dominant
<b>PAH</b>	99.96 %	612349	[Hyperphenylalaninemia, non-PKU mild], 261600 (3), Autosomal recessive; Phenylketonuria, 261600 (3), Autosomal recessive
<b>PARS2</b>	99.99 %	612036	Developmental and epileptic encephalopathy 75, 618437 (3), Autosomal recessive
<b>PCCA</b>	99.9 %	232000	Propionicacidemia, 606054 (3), Autosomal recessive
<b>PCCB</b>	99.97 %	232050	Propionicacidemia, 606054 (3), Autosomal recessive
<b>PDGFB</b>	99.99 %	190040	Meningioma, SIS-related, 607174 (3), Autosomal dominant; Basal ganglia calcification, idiopathic, 5, 615483 (3), Autosomal dominant; Dermatofibrosarcoma protuberans, 607907 (3)
<b>PDGFRB</b>	99.99 %	173410	Premature aging syndrome, Penttinen type, 601812 (3), Autosomal dominant; Kosaki overgrowth syndrome, 616592 (3), Autosomal dominant; Myofibromatosis, infantile, 1, 228550 (3), Autosomal dominant; Basal ganglia calcification, idiopathic, 4, 615007 (3), Autosomal dominant; Myeloproliferative disorder with eosinophilia, 131440 (4), Autosomal dominant
<b>PDHA1</b>	99.04 %	300502	Pyruvate dehydrogenase E1-alpha deficiency, 312170 (3), X-linked dominant
<b>PDHB</b>	99.94 %	179060	Pyruvate dehydrogenase E1-beta deficiency, 614111 (3), Autosomal recessive
<b>PDHX</b>	99.64 %	608769	Lacticacidemia due to PDX1 deficiency, 245349 (3), Autosomal recessive
<b>PDP1</b>	100 %	605993	Pyruvate dehydrogenase phosphatase deficiency, 608782 (3), Autosomal recessive
<b>PEX1</b>	98.8 %	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
<b>PEX10</b>	100 %	602859	Peroxisome biogenesis disorder 6A (Zellweger), 614870 (3), Autosomal recessive; Peroxisome biogenesis disorder 6B, 614871 (3), Autosomal recessive
<b>PEX11B</b>	99.62 %	603867	Peroxisome biogenesis disorder 14B, 614920 (3), Autosomal recessive

# Leukodystrophy and Leukoencephalopathy

Gene panel

Gene	% coding region covered*	OMIM gene id	OMIM Phenotypes
<b>PEX12</b>	100 %	601758	Peroxisome biogenesis disorder 3B, 266510 (3), Autosomal recessive; Peroxisome biogenesis disorder 3A (Zellweger), 614859 (3), Autosomal recessive
<b>PEX13</b>	99.36 %	601789	Peroxisome biogenesis disorder 11A (Zellweger), 614883 (3), Autosomal recessive; Peroxisome biogenesis disorder 11B, 614885 (3), Autosomal recessive
<b>PEX14</b>	100 %	601791	Peroxisome biogenesis disorder 13A (Zellweger), 614887 (3), Autosomal recessive
<b>PEX16</b>	99.94 %	603360	Peroxisome biogenesis disorder 8B, 614877 (3), Autosomal recessive; Peroxisome biogenesis disorder 8A (Zellweger), 614876 (3), Autosomal recessive
<b>PEX19</b>	99.25 %	600279	Peroxisome biogenesis disorder 12A (Zellweger), 614886 (3), Autosomal recessive
<b>PEX2</b>	100 %	170993	Peroxisome biogenesis disorder 5A (Zellweger), 614866 (3), Autosomal recessive; Peroxisome biogenesis disorder 5B, 614867 (3), Autosomal recessive
<b>PEX26</b>	100 %	608666	Peroxisome biogenesis disorder 7B, 614873 (3), Autosomal recessive; Peroxisome biogenesis disorder 7A (Zellweger), 614872 (3), Autosomal recessive
<b>PEX3</b>	99.85 %	603164	Peroxisome biogenesis disorder 10A (Zellweger), 614882 (3), Autosomal recessive; ?Peroxisome biogenesis disorder 10B, 617370 (3), Autosomal recessive
<b>PEX5</b>	99.89 %	600414	Peroxisome biogenesis disorder 2B, 202370 (3), Autosomal recessive; Peroxisome biogenesis disorder 2A (Zellweger), 214110 (3), Autosomal recessive; Rhizomelic chondrodysplasia punctata, type 5, 616716 (3), Autosomal recessive
<b>PEX6</b>	99.99 %	601498	Peroxisome biogenesis disorder 4B, 614863 (3), Autosomal recessive, Autosomal dominant; Peroxisome biogenesis disorder 4A (Zellweger), 614862 (3), Autosomal recessive; Heimler syndrome 2, 616617 (3), Autosomal recessive
<b>PEX7</b>	99.72 %	601757	Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive; Peroxisome biogenesis disorder 9B, 614879 (3), Autosomal recessive
<b>PI4KA</b>	99.76 %	600286	Spastic paraplegia 84, autosomal recessive, 619621 (3), Autosomal recessive; Gastrointestinal defects and immunodeficiency syndrome 2, 619708 (3), Autosomal recessive; Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531 (3), Autosomal recessive
<b>PLEKHG2</b>	99.99 %	611893	Leukodystrophy and acquired microcephaly with or without dystonia, 616763 (3), Autosomal recessive
<b>PLP1</b>	99.98 %	300401	Pelizaeus-Merzbacher disease, 312080 (3), X-linked recessive; Spastic paraplegia 2, X-linked, 312920 (3), X-linked recessive
<b>PLPBP</b>	99.99 %	604436	Epilepsy, early-onset, vitamin B6-dependent, 617290 (3), Autosomal recessive
<b>PMPCB</b>	99.94 %	603131	Multiple mitochondrial dysfunctions syndrome 6, 617954 (3), Autosomal recessive
<b>PNPT1</b>	99.56 %	610316	Spinocerebellar ataxia 25, 608703 (3), Autosomal dominant; Deafness, autosomal recessive 70, with or without adult-onset neurodegeneration, 614934 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 13, 614932 (3), Autosomal recessive
<b>POLG</b>	100 %	174763	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 (3), Autosomal recessive; Progressive external ophthalmoplegia, autosomal dominant 1, 157640 (3), Autosomal dominant; Progressive external ophthalmoplegia, autosomal recessive 1, 258450 (3), Autosomal recessive
<b>POLR1C</b>	100 %	610060	Leukodystrophy, hypomyelinating, 11, 616494 (3), Autosomal recessive; Treacher Collins syndrome 3, 248390 (3), Autosomal recessive
<b>POLR3A</b>	99.97 %	614258	Wiedemann-Rautenstrauch syndrome, 264090 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3), Autosomal recessive

# Leukodystrophy and Leukoencephalopathy

Gene panel

Gene	% coding region covered*	OMIM gene id	OMIM Phenotypes
<b>POLR3B</b>	99.94 %	614366	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381 (3), Autosomal recessive; Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742 (3), Autosomal dominant
<b>POLR3K</b>	100 %	606007	Leukodystrophy, hypomyelinating, 21, 619310 (3), Autosomal recessive
<b>POMGNT1</b>	99.69 %	606822	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 3, 613151 (3), Autosomal recessive; Retinitis pigmentosa 76, 617123 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 (3), Autosomal recessive
<b>POMT1</b>	99.96 %	607423	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 1, 613155 (3), Autosomal recessive
<b>POMT2</b>	99.98 %	607439	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 2, 613156 (3), Autosomal recessive
<b>PPFIBP1</b>	99.11 %	603141	Neurodevelopmental disorder with seizures, microcephaly, and brain abnormalities, 620024 (3), Autosomal recessive
<b>PPT1</b>	97.48 %	600722	Ceroid lipofuscinosis, neuronal, 1, 256730 (3), Autosomal recessive
<b>PRF1</b>	100 %	170280	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 (3), Autosomal recessive; Aplastic anemia, 609135 (3); Lymphoma, non-Hodgkin, 605027 (3)
<b>PRNP</b>	100 %	176640	Spongiform encephalopathy with neuropsychiatric features, 606688 (3), Autosomal dominant; Gerstmann-Strausler disease, 137440 (3), Autosomal dominant; Huntington disease-like 1, 603218 (3), Autosomal dominant; Insomnia, fatal familial, 600072 (3), Autosomal dominant; {Kuru, susceptibility to}, 245300 (3); Cerebral amyloid angiopathy, PRNP-related, 137440 (3), Autosomal dominant; Creutzfeldt-Jakob disease, 123400 (3), Autosomal dominant
<b>PSAP</b>	99.94 %	176801	Combined SAP deficiency, 611721 (3), Autosomal recessive; Krabbe disease, atypical, 611722 (3), Autosomal recessive; Metachromatic leukodystrophy due to SAP-b deficiency, 249900 (3), Autosomal recessive; Gaucher disease, atypical, 610539 (3); {Parkinson disease 24, autosomal dominant, susceptibility to}, 619491 (3), Autosomal dominant
<b>PSAT1</b>	99.98 %	610936	Neu-Laxova syndrome 2, 616038 (3), Autosomal recessive; ?Phosphoserine aminotransferase deficiency, 610992 (3), Autosomal recessive
<b>PSEN1</b>	100 %	104311	Pick disease, 172700 (3), Autosomal dominant; Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 (3), Autosomal dominant; Dementia, frontotemporal, 600274 (3), Autosomal dominant; ?Acne inversa, familial, 3, 613737 (3), Autosomal dominant; Cardiomyopathy, dilated, 1U, 613694 (3), Autosomal dominant; Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 (3), Autosomal dominant; Alzheimer disease, type 3, 607822 (3), Autosomal dominant
<b>PSEN2</b>	99.97 %	600759	Alzheimer disease-4, 606889 (3), Autosomal dominant; Cardiomyopathy, dilated, 1V, 613697 (3), Autosomal dominant
<b>PSPH</b>	99.09 %	172480	Phosphoserine phosphatase deficiency, 614023 (3), Autosomal recessive
<b>PYCR2</b>	99.95 %	616406	Leukodystrophy, hypomyelinating, 10, 616420 (3), Autosomal recessive
<b>QDPR</b>	99.92 %	612676	Hyperphenylalaninemia, BH4-deficient, C, 261630 (3), Autosomal recessive

# Leukodystrophy and Leukoencephalopathy

Gene panel

Gene	% coding region covered*	OMIM gene id	OMIM Phenotypes
<b>RAB11B</b>	100 %	604198	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807 (3), Autosomal dominant
<b>RARS1</b>	99.76 %	107820	Leukodystrophy, hypomyelinating, 9, 616140 (3), Autosomal recessive
<b>RNASEH2A</b>	99.95 %	606034	Aicardi-Goutieres syndrome 4, 610333 (3), Autosomal recessive
<b>RNASEH2B</b>	99.94 %	610326	Aicardi-Goutieres syndrome 2, 610181 (3), Autosomal recessive
<b>RNASEH2C</b>	99.99 %	610330	Aicardi-Goutieres syndrome 3, 610329 (3), Autosomal recessive
<b>RNASET2</b>	99.99 %	612944	Leukoencephalopathy, cystic, without megalencephaly, 612951 (3), Autosomal recessive
<b>RNF216</b>	99.99 %	609948	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840 (3), Autosomal recessive
<b>RNF220</b>	99.68 %	616136	Leukodystrophy, hypomyelinating, 23, with ataxia, deafness, liver dysfunction, and dilated cardiomyopathy, 619688 (3), Autosomal recessive
<b>RPIA</b>	99.84 %	180430	Ribose 5-phosphate isomerase deficiency, 608611 (3), Autosomal recessive
<b>RRM2B</b>	99.97 %	604712	Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 (3), Autosomal recessive; Rod-cone dystrophy, sensorineural deafness, and Fanconi-type renal dysfunction, 268315 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 (3), Autosomal dominant
<b>SAMD9L</b>	99.95 %	611170	Ataxia-pancytopenia syndrome, 159550 (3), Autosomal dominant; Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270 (3), Autosomal dominant; Spinocerebellar ataxia 49, 619806 (3), Autosomal dominant
<b>SAMHD1</b>	99.98 %	606754	?Chilblain lupus 2, 614415 (3), Autosomal dominant; Aicardi-Goutieres syndrome 5, 612952 (3), Autosomal recessive
<b>SCAF4</b>	99.91 %	616023	No OMIM phenotypes
<b>SCO1</b>	99.98 %	603644	Mitochondrial complex IV deficiency, nuclear type 4, 619048 (3), Autosomal recessive
<b>SCO2</b>	100 %	604272	Myopia 6, 608908 (3), Autosomal dominant; Mitochondrial complex IV deficiency, nuclear type 2, 604377 (3), Autosomal recessive
<b>SCP2</b>	94.94 %	184755	?Leukoencephalopathy with dystonia and motor neuropathy, 613724 (3), Autosomal recessive
<b>SDHA</b>	99.98 %	600857	Cardiomyopathy, dilated, 1GG, 613642 (3), Autosomal recessive; Mitochondrial complex II deficiency, nuclear type 1, 252011 (3), Autosomal recessive; Neurodegeneration with ataxia and late-onset optic atrophy, 619259 (3), Autosomal dominant; Paragangliomas 5, 614165 (3), Autosomal dominant
<b>SDHAF1</b>	99.99 %	612848	Mitochondrial complex II deficiency, nuclear type 2, 619166 (3), Autosomal recessive
<b>SDHAF2</b>	99.96 %	613019	Paragangliomas 2, 601650 (3), Autosomal dominant
<b>SDHB</b>	97.32 %	185470	Paragangliomas 4, 115310 (3), Autosomal dominant; Mitochondrial complex II deficiency, nuclear type 4, 619224 (3), Autosomal recessive; Gastrointestinal stromal tumor, 606764 (3), Isolated cases, Autosomal dominant; Pheochromocytoma, 171300 (3), Autosomal dominant; Paraganglioma and gastric stromal sarcoma, 606864 (3)
<b>SDHD</b>	82.93 %	602690	Paragangliomas 1, with or without deafness, 168000 (3), Autosomal dominant; Paraganglioma and gastric stromal sarcoma, 606864 (3); Mitochondrial complex II deficiency, nuclear type 3, 619167 (3), Autosomal recessive; Pheochromocytoma, 171300 (3), Autosomal dominant
<b>SEPSECS</b>	99.78 %	613009	Pontocerebellar hypoplasia type 2D, 613811 (3), Autosomal recessive
<b>SGO1</b>	99.9 %	609168	Chronic atrial and intestinal dysrhythmia, 616201 (3), Autosomal recessive
<b>SHANK3</b>	98.45 %	606230	Phelan-McDermid syndrome, 606232 (3), Autosomal dominant; {Schizophrenia 15}, 613950 (3), Autosomal dominant

# Leukodystrophy and Leukoencephalopathy

Gene panel

Gene	% coding region covered*	OMIM gene id	OMIM Phenotypes
<b>SLC13A3</b>	99.98 %	606411	Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate, 618384 (3), Autosomal recessive
<b>SLC16A2</b>	99.97 %	300095	Allan-Herndon-Dudley syndrome, 300523 (3), X-linked
<b>SLC17A5</b>	99.71 %	604322	Salla disease, 604369 (3), Autosomal recessive; Sialic acid storage disorder, infantile, 269920 (3), Autosomal recessive
<b>SLC20A2</b>	99.95 %	158378	Basal ganglia calcification, idiopathic, 1, 213600 (3), Autosomal dominant
<b>SLC25A12</b>	99.72 %	603667	Developmental and epileptic encephalopathy 39, 612949 (3), Autosomal recessive
<b>SLC35B2</b>	100 %	610788	Leukodystrophy, hypomyelinating, 26, with chondrodysplasia, 620269 (3), Autosomal recessive
<b>SLC44A1</b>	99.9 %	606105	Neurodegeneration, childhood-onset, with ataxia, tremor, optic atrophy, and cognitive decline, 618868 (3), Autosomal recessive
<b>SNAP29</b>	99.85 %	604202	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528 (3), Autosomal recessive
<b>SNORD118</b>	100 %	616663	Leukoencephalopathy, brain calcifications, and cysts, 614561 (3), Autosomal recessive
<b>SOX10</b>	100 %	602229	Waardenburg syndrome, type 4C, 613266 (3), Autosomal dominant; PCWH syndrome, 609136 (3), Autosomal dominant; Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 (3), Autosomal dominant
<b>SPART</b>	99.98 %	607111	Troyer syndrome, 275900 (3), Autosomal recessive
<b>SPG11</b>	99.89 %	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
<b>SPG7</b>	99.99 %	602783	Spastic paraplegia 7, autosomal recessive, 607259 (3), Autosomal recessive, Autosomal dominant
<b>STN1</b>	99.88 %	613128	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341 (3), Autosomal recessive
<b>STX11</b>	100 %	605014	Hemophagocytic lymphohistiocytosis, familial, 4, 603552 (3), Autosomal recessive
<b>STXBP2</b>	100 %	601717	Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease, 613101 (3), Autosomal recessive
<b>SUCLA2</b>	99.96 %	603921	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 (3), Autosomal recessive
<b>SUMF1</b>	99.95 %	607939	Multiple sulfatase deficiency, 272200 (3), Autosomal recessive
<b>SUOX</b>	100 %	606887	Sulfite oxidase deficiency, 272300 (3), Autosomal recessive
<b>SURF1</b>	100 %	185620	Charcot-Marie-Tooth disease, type 4K, 616684 (3), Autosomal recessive; Mitochondrial complex IV deficiency, nuclear type 1, 220110 (3), Autosomal recessive
<b>TACO1</b>	100 %	612958	Mitochondrial complex IV deficiency, nuclear type 8, 619052 (3), Autosomal recessive
<b>TMEM106B</b>	99.92 %	613413	Leukodystrophy, hypomyelinating, 16, 617964 (3), Autosomal dominant
<b>TMEM163</b>	100 %	618978	Leukodystrophy, hypomyelinating, 25, 620243 (3), Autosomal dominant
<b>TMEM63A</b>	99.91 %	618685	Leukodystrophy, hypomyelinating, 19, transient infantile, 618688 (3), Autosomal dominant
<b>TPP1</b>	99.99 %	607998	Ceroid lipofuscinosis, neuronal, 2, 204500 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 7, 609270 (3), Autosomal recessive
<b>TPP2</b>	99.89 %	190470	Immunodeficiency 78 with autoimmunity and developmental delay, 619220 (3), Autosomal recessive
<b>TREM2</b>	100 %	605086	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193 (3), Autosomal recessive

# Leukodystrophy and Leukoencephalopathy

Gene panel

Gene	% coding region covered*	OMIM gene id	OMIM Phenotypes
<b>TREX1</b>	100 %	606609	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 (3), Autosomal dominant; Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 (3), Autosomal recessive, Autosomal dominant; {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant; Chilblain lupus, 610448 (3), Autosomal dominant
<b>TSEN54</b>	100 %	608755	Pontocerebellar hypoplasia type 2A, 277470 (3), Autosomal recessive; Pontocerebellar hypoplasia type 4, 225753 (3), Autosomal recessive; ?Pontocerebellar hypoplasia type 5, 610204 (3), Autosomal recessive
<b>TTR</b>	100 %	176300	Amyloidosis, hereditary, transthyretin-related, 105210 (3), Autosomal dominant; Carpal tunnel syndrome, familial, 115430 (3), Autosomal dominant; [Dystransthyretinemic hyperthyroxinemia], 145680 (3), Autosomal dominant
<b>TUBB4A</b>	100 %	602662	Dystonia 4, torsion, autosomal dominant, 128101 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 6, 612438 (3), Autosomal dominant
<b>TUFM</b>	100 %	602389	Combined oxidative phosphorylation deficiency 4, 610678 (3), Autosomal recessive
<b>TWNK</b>	100 %	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
<b>TYMP</b>	99.87 %	131222	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041 (3), Autosomal recessive
<b>TYROBP</b>	99.95 %	604142	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770 (3), Autosomal recessive
<b>UBTF</b>	99.99 %	600673	Neurodegeneration, childhood-onset, with brain atrophy, 617672 (3), Autosomal dominant
<b>UFM1</b>	99.25 %	610553	Leukodystrophy, hypomyelinating, 14, 617899 (3), Autosomal recessive
<b>UNC13D</b>	100 %	608897	Hemophagocytic lymphohistiocytosis, familial, 3, 608898 (3), Autosomal recessive
<b>USP18</b>	93.05 %	607057	Pseudo-TORCH syndrome 2, 617397 (3), Autosomal recessive
<b>VPS11</b>	99.99 %	608549	?Dystonia 32, 619637 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 12, 616683 (3), Autosomal recessive
<b>WARS2</b>	97.72 %	604733	Parkinsonism-dystonia 3, childhood-onset, 619738 (3), Autosomal recessive; Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710 (3), Autosomal recessive
<b>WT1</b>	99.99 %	607102	Mesothelioma, somatic, 156240 (3); Meacham syndrome, 608978 (3), Autosomal dominant; Frasier syndrome, 136680 (3), Somatic mutation, Autosomal dominant; Nephrotic syndrome, type 4, 256370 (3), Autosomal dominant; Denys-Drash syndrome, 194080 (3), Somatic mutation, Autosomal dominant; Wilms tumor, type 1, 194070 (3), Somatic mutation, Autosomal dominant
<b>XPR1</b>	99.13 %	605237	Basal ganglia calcification, idiopathic, 6, 616413 (3), Autosomal dominant
<b>ZFYVE26</b>	100 %	612012	Spastic paraplegia 15, autosomal recessive, 270700 (3), Autosomal recessive

# Leukodystrophy and Leukoencephalopathy

Gene panel

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## Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2023-07-31**

Gene symbols used are according to the HGNC guidelines (corresponding to Ensembl database release 105).

Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.

Possible phenotype mapping keys

- (1) the disorder is placed on the map based on its association with a gene, but the underlying defect is not known
- (2) the disorder has been placed on the map by linkage; no mutation has been found
- (3) the molecular basis for the disorder is known; a mutation has been found in the gene
- (4) a contiguous gene deletion or duplication syndrome, multiple genes are deleted or duplicated causing the phenotype

Brackets, "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values (e.g., dysalbuminemic euthyroidal hyperthyroxinemia).

Braces, "{ }", indicate mutations that contribute to susceptibility to multifactorial disorders (e.g., diabetes, asthma) or to susceptibility to infection (e.g., malaria).

A question mark, "?", before the phenotype name indicates that the relationship between the phenotype and gene is provisional. More details about this relationship are provided in the comment field of the map and in the gene and phenotype OMIM entries.

\* Exome panels:  $\geq 20x$ , HyperCap panels:  $\geq 30x$