

Supplemental Table 7: 48 leukodystrophy genes

Leukodystrophies	Inheritance	Gene(s)	Source
18q deletion syndrome	de novo	<i>MBP</i>	GeneReviews
AD adult-onset leukodystrophy(ADLD)	AD	<i>LMNB1</i>	GeneReviews
Adult polyglucosan body disease(APBD)	AR	<i>GBE1</i>	GeneReviews
		<i>ADAR</i>	GeneReviews
		<i>RNASEH2A</i>	GeneReviews
Aicardi–Goutières syndrome (AGS)	AR>AD	<i>RNASEH2B</i>	GeneReviews
		<i>RNASEH2C</i>	GeneReviews
		<i>SAMHD1</i>	GeneReviews
		<i>TREX1</i>	GeneReviews
Alexander disease	AD	<i>GFAP</i>	GeneReviews
Canavan disease	AR	<i>ASPA</i>	GeneReviews
Cerebroretinal microangiopathy w/calcifications & cysts (CRMCC) 2	Likely AR	<i>CTC1</i>	GeneReviews
Cerebrotendinous xanthomatosis(CTX)	AR	<i>CYP27A1</i>	GeneReviews
Free sialic acid storage disorders 3	AR	<i>SLC17A5</i>	GeneReviews
Fucosidosis	AR	<i>FUCA1</i>	GeneReviews
Galactocerebrosidase deficiency (Krabbe disease)	AR	<i>GALC</i>	GeneReviews
Hereditary diffuse leukoencephalopathy w/spheroids(HDLS) 6	AD	<i>CSF1R</i>	GeneReviews
Hypomyelination and congenital cataract (HCC)	AR	<i>FAM126A</i>	GeneReviews
Hypomyelination w/atrophy of the basal ganglia & cerebellum (H-ABC)	Likely AD	<i>TUBB4A</i>	GeneReviews
L-2-hydroxyglutaric aciduria	AR	<i>L2HGDH</i>	GeneReviews
Leukoencephalopathy w/brain stem & spinal cord involvement & lactate elevation (LBSL)	AR	<i>DARS2</i>	GeneReviews
Leukoencephalopathy w/thalamus and brain stem involvement & lactate elevation (LTBL)	AR	<i>EARS2</i>	GeneReviews
Megalencephalic leukodystrophy w/subcortical cysts (MLC)	AR	<i>HEPACAM</i>	GeneReviews
		<i>MLC1</i>	GeneReviews
Metachromatic leukodystrophy(MLD)	AR	<i>ARSA</i>	GeneReviews
Multiple sulfatase deficiency (MSD)	AR	<i>SUMF1</i>	GeneReviews
Neurofibromatosis	AD	<i>NF1</i>	GeneReviews
Oculodentodigital dysplasia (ODDD)	AD>AR	<i>GJA1</i>	GeneReviews
Pelizaeus-Merzbacher disease (PMD)	XL	<i>PLP1</i>	GeneReviews
Pelizaeus-Merzbacher-like disease 1 (PMLD1)	AR	<i>GJC2</i>	GeneReviews
		<i>PEX1</i>	GeneReviews
Peroxisome Biogenesis Disorders, Zellweger Syndrome Spectrum	AR	<i>PEX6</i>	GeneReviews
		<i>PEX26</i>	GeneReviews
		<i>POLR3A</i>	GeneReviews
Pol III-related leukodystrophies 8	AR	<i>POLR3B</i>	GeneReviews
PSAP-related MLD 5	AR	<i>PSAP</i>	GeneReviews
RNAse T2-deficient leukoencephalopathy	AR	<i>RNASET2</i>	GeneReviews
Single-enzyme deficiencies of peroxisomal fatty acid beta oxidation: Dibifunctional protein deficiency	AR	<i>HSD17B4</i>	GeneReviews
Single-enzyme deficiencies of peroxisomal fatty acid beta oxidation: Peroxisomal acyl-CoA-oxidase deficiency	AR	<i>ACOX1</i>	GeneReviews
Single-enzyme deficiencies of peroxisomal fatty acid beta oxidation: SCPx deficiency	AR	<i>SCP2</i>	GeneReviews
Sjögren-Larsson syndrome	AR	<i>ALDH3A2</i>	GeneReviews
SOX10-associated disorders	AD	<i>SOX10</i>	GeneReviews
		<i>EIF2B2</i>	GeneReviews
Vanishing White Matter Disease, Childhood Ataxia with Central Nervous System Hypomyelination	AR	<i>EIF2B3</i>	GeneReviews
		<i>EIF2B4</i>	GeneReviews
		<i>EIF2B5</i>	GeneReviews
		<i>ALDP</i>	GeneReviews
X-linked adrenoleukodystrophy (X-ALD)	XL	<i>ABCD1</i>	GeneReviews