Title: Leukodystrophy Overview GeneReview Table 9

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Table 9. Disorders with Thinning of the Corpus Callosum on MRI

Feature	Inherited Disorder/Condition		Acquired Disorder /
	Leukodystrophy	Not Leukodystrophy	Condition
Thinning of the corpus callosum	 Free sialic acid storage disorders ¹ Fucosidosis HCC ² HDLS ³ Pol III-related leukodystrophies ⁴ 	 Adenylosuccinase deficiency FAHN ⁵ Fumarate hydratase deficiency GPR56-related disorders Glycine encephalopathy Molybdenum cofactor deficiency & isolated sulfite oxidase deficiency Mucolipidosis IV Neuronopathic form of malignant infantile osteopetrosis Peroxisomal disorders such as peroxisome biogenesis disorders and single enzyme deficiencies (excluding X-ALD) SPG 11 ⁶ SPG 15 	

Note: Disorders are ordered alphabetically.

- 1. Includes Salla disease; infantile sialic acid storage disease, intermediate form
- 2. Hypomyelination and congenital cataract
- 3. Hereditary diffuse leukoencephalopathy with spheroids. Also known as adult-onset leukodystrophy w/ neuroaxonal spheroids & pigmented glia; may include hereditary diffuse pigmentary type of orthochromatic leukodystrophy w/pigmented glia (POLD).
- 4. Includes hypomyelination, hypodontia, hypogonadotropic hypogonadism (4H syndrome); ataxia, delayed dentition, and hypomyelination (ADDH); tremor-ataxia with central hypomyelination (TACH); leukodystrophy with oligodontia (LO); and hypomyelination with cerebellar atrophy and hypoplasia of the corpus callosum (HCAHC).
- 5. Fatty acid hydroxylase-associated neurodegeneration
- 6. SPG = spastic paraplegia