

Title: Leukodystrophy Overview *GeneReview* Table 3

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Table 3. Disorders with White Matter Rarefaction and Cysts Observed on MRI

Feature		Inherited Disorder/Condition		Acquired Disorder/Condition
		Leukodystrophy	Not Leukodystrophy	
Diffuse white matter rarefaction		<ul style="list-style-type: none"> • Alexander disease • CACH/VWM¹ • L-2-hydroxyglutaric aciduria² • LBSL³ 	<ul style="list-style-type: none"> • Mitochondrial defects 	
Cysts	Anterior temporal	<ul style="list-style-type: none"> • AGS⁴ (inconstant) • MLC⁵ (MLC1 & MLC2) • RNAse T2-deficient leukoencephalopathy 	<ul style="list-style-type: none"> • Dystroglycanopathies • LAMA2 MD⁶ • Menkes disease 	<ul style="list-style-type: none"> • Congenital cytomegalovirus • Perinatal HSV
	Other locations	<ul style="list-style-type: none"> • AGS⁴ • Alexander disease • CRMCC⁷ • Krabbe disease • L-2-hydroxyglutaric aciduria (advanced stages) • MLC⁵ (MLC1 & MLC2) 	<ul style="list-style-type: none"> • Dystroglycanopathies • Incontinentia pigmenti • LAMA2 MD⁶ • Molybdenum cofactor deficiency & isolated sulfite oxidase deficiency • Neonatal energy depletion (inborn error or exogenous), including hypoglycemia • Menkes disease • Mitochondrial defects 	<ul style="list-style-type: none"> • Infections, especially in the neonatal period

Note: Disorders are ordered alphabetically.

1. Childhood ataxia with central nervous system hypomyelination / vanishing white matter
2. Characterized by less significant rarefaction
3. Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation
4. Aicardi-Goutières syndrome
5. Megalencephalic leukoencephalopathy with subcortical cysts
6. *LAMA2*-related muscular dystrophy
7. Cerebroretinal microangiopathy with calcifications and cysts