

Title: Leukodystrophy Overview *GeneReview* Tables 5 and 6

Authors: Vanderver A, Tonduti D, Schiffmann R, Schmidt J, Van der Knaap M

Date: February 2014

**Table 5. Disorders with Contrast Enhancement on MRI**

Feature	Inherited Disorder/Condition		Acquired Disorder / Condition
	Leukodystrophy	Not Leukodystrophy	
<b>Contrast enhancement</b>	<ul style="list-style-type: none"> <li>• <a href="#">Alexander disease</a></li> <li>• CRMCC <sup>1</sup></li> <li>• <a href="#">X-ALD</a> <sup>2</sup></li> <li>• <a href="#">HDLS</a> <sup>3</sup></li> <li>• Peroxisomal Acyl-CoA oxidase deficiency</li> </ul>	<ul style="list-style-type: none"> <li>• <a href="#">FHLH</a> <sup>4</sup></li> <li>• <a href="#">Mitochondrial disorders</a></li> <li>• <a href="#">MS</a> <sup>45</sup> or ADEM <sup>6</sup></li> <li>• Peroxisomal biogenesis disorder</li> </ul>	<ul style="list-style-type: none"> <li>• Infectious disorders</li> <li>• Vascular / perivascular malignancies</li> <li>• Vasculitis</li> </ul>

Note: Disorders are ordered alphabetically.

1. Cerebroretinal microangiopathy w/calcifications & cysts; this disorder now appears to be distinct from Coats plus caused by mutations in CTC1, encoding conserved telomere maintenance component 1.

2. X-linked adrenoleukodystrophy

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. Familial hemophagocytic lymphohistiocytosis

5. Multiple sclerosis

6. Acute disseminated encephalomyelitis

**Table 6. Disorders with Macrocephaly and Leukoencephalopathy on MRI**

Feature	Inherited Disorder/Condition	
	Leukodystrophy	Not Leukodystrophy
<b>Megalencephalic leukoencephalopathy</b>	<ul style="list-style-type: none"> <li>• <a href="#">Alexander disease</a></li> <li>• <a href="#">Canavan disease</a></li> <li>• L-2-hydroxyglutaric aciduria (inconstant)</li> <li>• <a href="#">MLC</a> <sup>1</sup></li> </ul>	<ul style="list-style-type: none"> <li>• <a href="#">Hexosaminidase A deficiency</a> (inconstant)</li> <li>• Infantile lysosomal storage disorders (inconstant)</li> <li>• <a href="#">Neurocutaneous syndromes</a></li> <li>• <a href="#">PHTS</a> <sup>2</sup></li> <li>• Hypomelanosis of Ito <sup>3</sup></li> </ul>

Note: Disorders are ordered alphabetically.

1. Megalencephalic leukoencephalopathy with subcortical cysts

2. *PTEN* hamartoma tumor syndromes

3. Hypomelanosis of Ito is not a specific disorder, but rather a nonspecific cutaneous finding often associated with mosaicism for a genetic alteration such as chromosomal mosaicism.