Title: Leukodystrophy Overview *GeneReview* Table 10
Authors: Vanderver A, Tonduti D, Schiffmann R, Schmidt J, Van der Knaap M
Date: February 2014

Table 10. Disorders with Non-Calcifying Basal Ganglia Lesions on MRI

Feature	Inherited Disorder/Condition		Acquired Disorder/Condition
	Leukodystrophy	Not Leukodystrophy	
Non-calcifying basal ganglia lesions	• HDLS [1, 2] • Alexander disease • Canavan disease • CRMCC [3] • CTX [4] • CACH/VWM [2, 5] • Fucosidosis • HABC [6] • L-2-hydroxyglutaric aciduria • ODDD [7]	• DRPLA [8] • Disorders of branched-chain amino acids (BCAAS [9]) • LTBL • FAHN [10] • GM1 gangliosidoses • GM2 gangliosidoses • Glutaric aciduria type I • Mitochondrial encephalopathies • Other amino acidopathies & organic acidurias • MCT8-specific THCT [11] deficiency [2] • Mucolipidosis IV • SSADH [12] deficiency • Molybdenum cofactor deficiency & isolated sulfite oxidase deficiency • Urea cycle disorders • Wilson disease	• Histiocytosis • Vasculopathies

Note: Disorders are ordered alphabetically.

1. 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).

2. Rare

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

4. Cerebrotendinous xanthomatosis

5. Childhood ataxia with central nervous system hypomyelination / vanishing white matter

6. Hypomyelination with atrophy of the basal ganglia and cerebellum

7. Oculodentodigital dysplasia

8. Dentatorubropallidoluysian atrophy

9. Includes maple syrup urine disease [MSUD]

10. Fatty acid hydroxylase-associated neurodegeneration

11. Thyroid hormone cell-membrane transporter

12. Succinic semialdehyde dehydrogenase