Title: Leukodystrophy Overview *GeneReview* Table 11
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**Table 11. Disorders with Brain Stem Involvement on MRI**

| Feature | | Inherited Disorder/Condition | | Acquired Disorder/Condition |
|---|---|---|---|---|
| | | **Leukodystrophy** | **Not Leukodystrophy** | |
| **Brain stem involvement** | **Common** | • Alexander disease<br>• CTX [1]<br>• Krabbe disease<br>• LBSL [2]<br>• LTBL [3]<br>• MLD [4] and its biochemical variants<br>• ODDD [5]<br>• PMD [6]<br>• PMLD [7]<br>• PBD, ZSS [8]<br>• PGBD [9] | • DRPLA [10]<br>• Disorders of branched-chain amino acids (BCAAS [11])<br>• FAHN [12] (atrophy)<br>• FXTAS [13]<br>• *GPR56*-related disorders<br>• Mitochondrial encephalopathies<br>• Molybdenum cofactor deficiency & isolated sulfite oxidase deficiency<br>• Wilson disease | |
| | **More rare** | • X-ALD [14]<br>• HDLS [15]<br>• AGS [16]<br>• Canavan disease<br>• CACH/VWM [17]<br>• MLC [18]<br>• Sjögren-Larsson syndrome | • 3-hydroxy-3-methylglutaryl-CoA lyase deficiency<br>• Kearns-Sayre syndrome | • Histiocytosis |

Note: Disorders are ordered alphabetically.

'Brain stem involvment' usually refers to brain stem white matter abnormalities, but in some instances can also refer to brain stem atrophy

1. Cerebrotendinous xanthomatosis

2. Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation

3. Leukoencephalopathy with thalamus and brain stem involvement and lactate elevation

4. Metachromatic leukodystrophy

5. Oculodentodigital dysplasia

6. Pelizaeus-Merzbacher disease

7. Pelizaeus-Merzbacher like-disease

8. Peroxisome biogenesis disorders, Zellweger syndrome spectrum; includes neonatal adrenoleukodystrophy; infantile Refsum disease

9. Polyglucosan body disease

10. Dentatorubropallidoluysian atrophy

11. Includes maple syrup urine disease [MSUD]

12. Fatty acid hydroxylase-associated neurodegeneration

13. Fragile X-associated tremor/ataxia syndrome

14. X-linked adrenoleukodystrophy

15. 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).

16. Aicardi-Goutières syndrome

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

18. Megalencephalic leukodystrophy with subcortical cysts