Title: Leukodystrophy Overview *GeneReview* Table 2
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Table 2. Heritable Disorders with Significant White Matter Involvement that are not Leukodystrophies

3-hydroxy-3-methylglutaryl-CoA lyase deficiency
Adenylosuccinase deficiency
ACAN-related disorders
AIMP1-related disorders
Aspartylglucosaminuria
Band-like intracranial calcification with simplified gyration and polymicrogyria , caused by mutations in *OCLN,* encoding a tight junction protein expressed in endothelia in the brain
Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL)
Cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy (CARASIL)
Cockayne syndrome and trichothiodystrophy
COL4A1-related disorders
Congenital muscular dystrophy (CMD) (including defects of O-glycan synthesis and *LAMA2*-related muscular dystrophy [merosin-deficient congenital muscular dystrophy type 1A])
Defects of N-glycan synthesis including congenital disorders of glycosylation
Dentatorubropallidoluysian atrophy (DRPLA)
Disorders of branched chain amino acids (BCAAS) and other amino acid disorders (including untreated propionic aciduria, methylmalonic aciduria, isovaleric aciduria, maple syrup urine disease [MSUD]; excluding E3 subunit deficiency)
BH4-deficient hyperphenylalaninemia C
Disorders of glycoprotein degradation (including alpha-mannosidosis, beta mannosidosis and sialidosis; excluding fucosidosis)
Fabry disease
Familial hemophagocytic lymphohistiocytosis
Fatty acid hydroxylase-associated neurodegeneration (*FA2H*-related disorders)
Fragile X-associated tremor/ataxia syndrome (FXTAS)
Fumarate hydratase deficiency
Galactosemia type I
Giant axonal neuropathy
Glutaric aciduria type I (GA-I)
Glutaric aciduria type II (GA-II; multiple acyl-CoA dehydrogenase deficiency; MADD)
Glycine encephalopathy
GM1 gangliosidosis, infantile onset
GM2 gangliosidosis, infantile onset
GPR56-related disorders
Hereditary homocystinurias
HSPD1-related disorders
Hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome
Hypomelanosis of Ito (HMI; incontinentia pigmenti achromians)

Incontinentia pigmenti
JAM3-related disorders
L-2-hydroxyglutaric aciduria
Lowe syndrome
MCT8-specific disorders
Menkes disease
Mitochondrial neurogastrointestinal encephalopathy (MNGIE)
Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke (MELAS)
Molybdenum cofactor deficiency and isolated sulfite oxidase deficiency
Mucolipidosis IV
Mucopolysaccharidosis including MPS type II (Hunter syndrome)
Multiple carboxylase deficiency, including biotinidase deficiency and holocarboxylase synthase deficiency
Myotonic dystrophy type I
Neuronal ceroid-lipofuscinoses (NCL), infantile onset
Neuronopathic form of malignant infantile osteopetrosis
Niemann-Pick disease type C
Phenylketonuria (PKU)
POLG-related disorders
Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy (PLOSL; Nasu Hakola disease)
Pyruvate carboxylase (PC) deficiency
Pyruvate dehydrogenase (PDH) deficiency
Serine synthesis defects
SPG 11 and SPG 15
Spondyloenchondrodysplasia
Succinic semialdehyde dehydrogenase (SSADH) deficiency (4-hydroxybutyric aciduria)
Urea cycle disorders:
- Carbamoylphosphate synthetase I deficiency
- Ornithine transcarbamylase (OTC) deficiency
- Citrullinemia type I
- Argininosuccinic aciduria
- Arginase deficiency
- N-acetylglutamate synthase (NAGS) deficiency

Wilson disease
Woodhouse-Sakati syndrome (WSS)

Note: Disorders are ordered alphabetically.