Teaching *NeuroImages*: Megalencephalic leukoencephalopathy with subcortical cysts (Van der Knaap disease)

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Figure MRI features of megalencephalic leukoencephalopathy

(A) Axial T2-weighted image showing diffusely abnormal (hyperintense) and mildly swollen white matter with sparing of basal ganglia, thalami, and internal capsules. (B) Axial T2-weighted image showing subcortical cysts (arrows) in the bilateral anterior temporal lobes. Cerebellum and brainstem are normal.

A 9-month-old boy born at full term to consanguineous parents presented with seizures, macrocephaly, and inability to sit without support. MRI showed characteristic features^{1,2} (figure) establishing the diagnosis of megalencephalic leukoencephalopathy (MLC). MLC is an autosomal recessive, neurodegenerative disorder with macrocephaly in the first year of life but delayed onset of motor deterioration and cognitive decline despite markedly abnormal MRI findings.¹ The differential diagnosis of MLC includes Canavan disease, Alexander disease, L-2-

hydroxyglutaric aciduria, and merosin-deficient congenital muscular dystrophy. None of these disorders shares all the MRI features, clinical characteristics, and disease course of MLC.¹

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