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Megalencephalic leukoencephalopathy with subcortical cysts

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base.

Megalencephalic leukoencephalopathy with subcortical cysts. ORPHA:2478

Megalencephalic leukoencephalopathy with subcortical cysts (MLC) is a form of leukodystrophy that is characterized by infantile-onset macrocephaly, often with mild neurologic signs at presentation (such as mild motor delay), which worsen with time, leading to poor ambulation, falls, ataxia, spasticity, increasing seizures and cognitive decline. Brain magnetic resonance imaging reveals diffusely abnormal and mildly swollen white matter as well as subcortical cysts in the anterior temporal and frontoparietal regions.