

Opis choroby *

Definicja

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.

Dane

Klasyfikacja

Choroba

Synonimy

MLC

Leukodystrofia z wielkogłowie

MLC

Wielkogłowie - leukodystrofia torbielowata

Wielkogłowie z leukodystrofią wakuolizującą i torbielami podkorowymi

Zespół Van der Knaapa

Megalencephalic leukodystrophy

Megalencephaly-cystic leukodystrophy syndrome

Vacuolating megalencephalic

leukoencephalopathy with subcortical cysts

Van der Knaap syndrome

Kod ORPHA

2478

Kod OMIM

613926

Kod ICD10

E75.2

Kod ICD11

8A44.3

[*Źródło](#)

orphanet