Opis choroby *

Definicja

A rare, genetic, neurometabolic disease characterized by early onset encephalopathy with progressive microcephaly, severe global development delay, seizures, hypotonia, feeding difficulties, variable cardiac abnormalities, and cataracts. Brain MRI shows distinct pattern with high T2 signal and restricted diffusion in the posterior limb of the internal capsule in combination with delayed myelination and progressive cerebral atrophy. The disease is typically fatal.

Dane

Klasyfikacja
Choroba

Synonimy
Martsolf-like syndrome
Martsolf-like syndrome

Kod ORPHA
457375

Kod OMIM
616647

Kod ICD10
G40.4

Kod ICD11
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*Źródło

orphanet