

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