Opis choroby *

Definicja

A rare mitochondrial disease characterized by a distinctive MRI pattern of cavitating leukodystrophy, predominantly in the posterior region of the cerebral hemispheres. The clinical picture varies widely between acute neurometabolic decompensation in infancy with loss of developmental milestones, seizures, and pyramidal signs rapidly evolving into spastic tetraparesis, to subtle neurological symptoms presenting in adolescence. The disease course tends to stabilize over time in most patients, and marked recovery of milestones may be observed.

Dane

Klasyfikacja
Choroba

Kod ORPHA
436271

Kod OMIM
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Kod ICD10
G93.4

Kod ICD11
-

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*Źródło

orphanet