Opis choroby *

Definicja

A rare mitochondrial disease characterized by a variable phenotype comprising delayed psychomotor development or neurodevelopmental regression, hypotonia, seizures, microcephaly, optic atrophy, pyramidal signs, and peripheral neuropathy, among others. Age of onset and disease severity are also variable with some cases taking a fatal course in early infancy. Serum lactate levels may be elevated. Reported brain imaging findings include abnormal signals in the basal ganglia, cerebral and/or cerebellar atrophy, and white matter abnormalities.

Dane

Klasyfikacja
Choroba

Kod ORPHA
527276

Kod OMIM
-

Kod ICD10
E88.8

Kod ICD11
-

---

*Źródło

orphanet