

Opis choroby *

Definicja

A rare neurometabolic disease, due to a lipoic acid biosynthesis defect, with a highly variable phenotype, typically characterized by early-onset acute or subacute developmental delay or regression frequently associated with feeding difficulties. Clinical severity is variable and may range from mild cases which present a later onset with slow neurological deterioration and general improvement over time to severe cases with clinical signs since birth and leading to early death. Associated manifestations include hypotonia, vision loss, respiratory failure, seizures, and intellectual disability. Brain magnetic resonance imaging frequently shows cavitating leukoencephalopathy with lesions in the periventricular/central white matter and parieto-occipital lobes.

Dane

Klasyfikacja

Choroba

Synonimy

IBA57 deficiency

Śmiertelny zespół złożonej dysfunkcji
mitochondrialnej typu 3

Wrodzone zaburzenie glikozylacji typu II_m

MMDS3

Kod ORPHA

363424

Kod OMIM

615330

Kod ICD10

E88.8

Kod ICD11

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*Źródło

orphanet