Opis choroby *

Definicja

A rare mitochondrial disease characterized by infantile onset of severe regression after a period of normal development, epileptic encephalopathy, hypotonia, movement disorder, cardiomyopathy, hyperglycinemia, and lactic acidosis. Optic atrophy may also be present. Brain imaging findings are highly variable and include white matter abnormalities. The disease is typically fatal in infancy.

Dane

Klasyfikacja
Choroba

Synonimy
BOLA3 deficiency
Niedobór BOLA3
MMDS2

Kod ORPHA
401874

Kod OMIM
614299

Kod ICD10
E88.8

Kod ICD11
5C53.21

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*Źródło

orphanet