

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

*Źródło

orphanet