

Opis choroby *

Definicja

Fatal infantile lactic acidosis with methylmalonic aciduria is a rare neurometabolic disease characterized by infantile onset of severe encephalomyopathy, lactic acidosis and elevated methylmalonic acid urinary excretion. Clinically it manifests with severe psychomotor delay, hypotonia, failure to thrive, feeding difficulties and dystonia. Epilepsy and multiple congenital anomalies may be associated.

Dane

Klasyfikacja

Choroba

Kod ORPHA

17

Kod OMIM

245400

Kod ICD10

E71.1

Kod ICD11

5C53.20

*Źródło

orphanet