Opis choroby *

Definicja

A rare, multisystemic inherited metabolic diseases characterized clinically, by a variable spectrum of severity, primarily comprised of psychomotor delay, myopathy and liver dysfunction. Most patients present in infancy, but the onset can be already in utero or in adult age. Hypermethioninemia is frequent, but often absent in infancy. Creatine kinase is elevated in most patients.

Dane

Klasyfikacja
Choroba

Synonimy
Hipermetioninemia spowodowana niedoborem hydrolazy S-adenozylolhomocysteiny

Kod ORPHA
88618

Kod OMIM
613752

Kod ICD10
E72.1

Kod ICD11
5C50.B

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*Źródło

orphanet