

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

Hipermetyoninemia spowodowana niedoborem hydrolazy S-adenozylhomocysteiny

Kod ORPHA

88618

Kod OMIM

613752

Kod ICD10

E72.1

Kod ICD11

5C50.B

*Źródło

orphanet