Opis choroby *

Definicja

Hypermethioninemia due to glycine N-methyltransferase deficiency is a rare, genetic inborn error of metabolism characterized by a relatively benign clinical phenotype, with only mild to moderate hepatomegaly reported, in addition to laboratory studies revealing permanent, greatly increased hypermethioninemia, mild to moderate elevation of aminotransferases and highly elevated plasma S-adenosyl-methionine with normal S-adenosylhomocysteine and total homocysteine.

Dane

Klasyfikacja
Choroba

Synonimy
Glycine N-methyltransferase deficiency
Hipermetioninemia z powodu niedoboru GNMT
Niedobór N-metylotransferazy glicyny
Hypermethioninemia due to GNMT deficiency

Kod ORPHA
289891

Kod OMIM
606664

Kod ICD10
E72.1

Kod ICD11
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*Źródło

orphanet