## 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