

Opis choroby *

Definicja

Hypermethioninemia due to methionine adenosyltransferase deficiency is a very rare metabolic disorder resulting in isolated hepatic hypermethioninemia that is usually benign due to partial inactivation of enzyme activity. Rarely patients have been found to have an odd odor or neurological disorders such as brain demyelination.

Dane

Klasyfikacja

Choroba

Synonimy

MAT I/III deficiency

Niedobór adenozylotransferazy metioniny

Niedobór MAT

Niedobór MAT I/III

Niedobór adenozylotransferazy metioninowej
I/III

Mudd's disease

Kod ORPHA

168598

Kod OMIM

250850

Kod ICD10

E72.1

Kod ICD11

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*Źródło

orphanet