

Opis choroby *

Definicja

A rare inborn error of metabolism characterized by abnormal accumulation of plasma cystathionine and subsequent increased urinary excretion due to cystathionine gamma-lyase deficiency. The condition is considered benign without pathological relevance. Mode of inheritance is autosomal recessive.

Dane

Klasyfikacja

Choroba

Synonimy

Cystathionase deficiency

Niedobór gamma-cystationazy

Niedobór cystationazy

Niedobór liazy gamma - cystationu

Cystathionine gamma-lyase deficiency syndrome

Gamma-cystathionase deficiency

Kod ORPHA

212

Kod OMIM

219500

Kod ICD10

E72.1

Kod ICD11

5C50.B

*Źródło

orphanet