

Opis choroby *

Definicja

Hyperlysinaemia is a lysine metabolism disorder characterised by elevated levels of lysine in the cerebrospinal fluid and blood. Variable degrees of saccharopinuria are also present.

Dane

Klasyfikacja

Choroba

Synonimy

Hyperlysinemia type I

Hiperlizynemia typu I

Niedobór reduktazy lizyny alfa-keto glutarowej

Lysine alpha-ketoglutarate reductase deficiency

Kod ORPHA

2203

Kod OMIM

238710

Kod ICD10

E72.3

Kod ICD11

5C50.4

*Źródło

orphanet