

Opis choroby *

Definicja

Histidinemia is a rare metabolic disorder characterized by elevated histidine levels in blood, urine, and cerebrospinal fluid, generally with no clinical repercussions.

Dane

Klasyfikacja

Choroba

Synonimy

HAL deficiency

Hiperhistydynemia

Histydynuria

Niedobór amoniakolizy histydyny

Niedobór HAL

Niedobór HIS

Niedobór histydazy

HIS deficiency

Histidase deficiency

Histidine ammonia-lyase deficiency

Histidinuria

Hyperhistidinemia

Kod ORPHA

2157

Kod OMIM

235800

Kod ICD10

E70.8

Kod ICD11

5C50.20

*Źródło

orphanet