Opis choroby *

Definicja

A rare disorder of histidine metabolism characterized by histidinuria without histidinemia due to impaired intestinal and renal tubular absorption of histidine. Developmental delay, intellectual disability, seizures, and mild dysmorphic features have been reported in association. There have been no further descriptions in the literature since 1992.

Dane

Klasyfikacja
Choroba

Kod ORPHA
2158

Kod OMIM
235830

Kod ICD10
E70.8

Kod ICD11
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*Źródło

orphanet