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