

Histydynuria - wada kanalików nerkowych

Kod Orpha: 2158 Kod OMIM: 235830

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

-

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.