

Opis choroby *

Definicja

A rare histidine metabolism disorder characterized by urocanic aciduria and other variable manifestations including intellectual deficit and intermittent ataxia.

Dane

Klasyfikacja

Choroba

Synonimy

Encephalopathy due to urocanase deficiency

Encefalopatia z powodu niedoboru urokanazy

Kod ORPHA

210128

Kod OMIM

276880

Kod ICD10

E70.8

Kod ICD11

5C50.21

*Źródło

orphanet