Opis choroby *

Definicja

A rare autosomal recessive urea cycle defect characterized clinically by recurring episodes of hyperammonemia and associated neuropsychiatric symptoms in the adult-onset form (citrullinemia type II), and by transient cholestasis and variable hepatic dysfunction in the neonatal form (neonatal intrahepatic cholestasis due to citrin deficiency).

Dane

Klasyfikacja
Kategoria

Kod ORPHA
247582

Kod OMIM
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Kod ICD10
E72.2

Kod ICD11
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*Źródło

orphanet