

Opis choroby *

Definicja

A mild subtype of citrin deficiency characterized clinically by low birth weight, failure to thrive, transient intrahepatic cholestasis, multiple aminoacidemia, galactosemia, hypoproteinemia, hepatomegaly, decreased coagulation factors, hemolytic anemia, variable but mostly mild liver dysfunction, and hypoglycemia.

Dane

Klasyfikacja

Choroba

Synonimy

NICCD

Cholestaza wewnątrzwątrobowa noworodków
wywołana przez Niedobór cytrynianu

NICCD

Neonatal intrahepatic cholestasis caused by
citrin deficiency

Kod ORPHA

247598

Kod OMIM

605814

Kod ICD10

E72.2

Kod ICD11

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*Źródło

orphanet