DISEASE:
Neonatal intrahepatic cholestasis due to citrin deficiency

NAME:	Neonatal intrahepatic cholestasis due to citrin deficiency
DESCRIPTION:	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.
ORPHACODE:	247598
SYNONYMS:	NICCD Neonatal intrahepatic cholestasis caused by citrin deficiency
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	SLC25A13
CREATED:	13 May 2019 - 01:02
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