

DISEASE:
Crigler-Najjar syndrome type 2

NAME:	Crigler-Najjar syndrome type 2
DESCRIPTION:	A form of Crigler Najjar syndrome (CNS), a rare hereditary disorder of bilirubin metabolism, characterized by unconjugated hyperbilirubinemia due to reduced and inducible activity of hepatic UDP-glucuronosyltransferase 1A1. The disorder clinically manifests with neonatal, isolated jaundice with a risk of developing bilirubin encephalopathy later in life due to triggers such as stress or infection.
ORPHACODE:	79235
SYNONYMS:	Bilirubin uridinediphosphate glucuronosyltransferase deficiency type 2 Bilirubin-UGT deficiency type 2
XREF(S):	Orphanet OMIM MeSH MedDRA ICD-10
ANALYTE(S):	UGT1A1
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