DISEASE:
Crigler-Najjar syndrome type 1

NAME:	Crigler-Najjar syndrome type 1
DESCRIPTION:	A form of Crigler Najjar syndrome (CNS), a hereditary disorder of hepatic bilirubin conjugation, characterized by severe neonatal unconjugated hyperbilirubinemia due to a complete absence of hepatic UDP-glucuronosyltransferase 1A1. The disorder clinically manifests with neonatal, isolated, severe and permanent jaundice with a permanent risk of bilirubin encephalopathy.
ORPHACODE:	79234
SYNONYMS:	Bilirubin uridinediphosphate glucuronosyltransferase deficiency type 1 Bilirubin-UGT deficiency type 1
XREF(S):	Orphanet MeSH MedDRA OMIM ICD-10
ANALYTE(S):	UGT1A1
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