Opis choroby *

Definicja

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.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
Bilirubin uridinediphosphate glucuronosyltransferase deficiency type 1
Dziedziczna niesprzężona hiperbilirubinemia typu 1
Niedobór bilirubiny-UGT typu 1
Niedobór UGT typu 1
Niedobór urydynodifosforan-glukonylotransferazy bilirubiny typu 1
Bilirubin-UGT deficiency type 1

Kod ORPHA
79234

Kod OMIM
218800

Kod ICD10
E80.5

Kod ICD11
5C58.00

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*Źródło

orphanet