Opis choroby *

Definicja

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.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
Bilirubin uridinediphosphate glucuronosyltransferase deficiency type 2
Dziedziczna niesprzężona hiperbilirubinemia typu 2
Niedobó UGT typu 2
Niedobór bilirubiny-UGT typu 2
Niedobór urydynodifosforan-glukuronosylotransferazy bilirubiny typu 2
Zespół Ariasa
Bilirubin-UGT deficiency type 2

Kod ORPHA
79235

Kod OMIM
606785

Kod ICD10
E80.5

Kod ICD11
5C58.00

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*Źródło

orphanet