Opis choroby *

Definicja

A rare hereditary disorder of bilirubin metabolism characterized by unconjugated hyperbilirubinemia due to a either a complete (type 1) or partial and inducible (type 2) hepatic deficit of UDP-glucuronosyltransferase 1A1 activity. The disorder manifests with neonatal jaundice with a risk of developing bilirubin encephalopathy.

Dane

Klasyfikacja
Choroba

Synonimy
Bilirubin uridinediphosphate glucuronosyltransferase deficiency
Dziedziczna nieskoniugowana hiperbilirubinemia
Niedobór bilirubiny-UGT
Niedobór UGT
Niedobór urydynodifosforanu glukuronylotransferazy bilirubiny
Bilirubin-UGT deficiency

Kod ORPHA
205

Kod OMIM
606785

Kod ICD10
E80.5

Kod ICD11
5C58.00

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*Źródło

orphanet