

## 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 niesprężona 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