

## Opis choroby \*

### Definicja

A rare hereditary disorder of bilirubin metabolism characterized by unconjugated hyperbilirubinemia due to 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](#)

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