Opis choroby *

Definicja

Progressive familial intrahepatic cholestasis type 2 (PFIC2), a type of progressive familial intrahepatic cholestasis (PFIC, see this term), is a severe, neonatal, hereditary disorder in bile formation that is hepatocellular in origin and not associated with extrahepatic features. Initially, PFIC2 was reported under the name Byler syndrome.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
BSEP deficiency
Niedobór BSEP
PFIC2
PFIC2

Kod ORPHA
79304

Kod OMIM
601847

Kod ICD10
K76.8

Kod ICD11
5C58.03

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*Źródło

orphanet