

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

*Źródło

orphanet