DISEASE:
Progressive familial intrahepatic cholestasis type 1

NAME:	Progressive familial intrahepatic cholestasis type 1
DESCRIPTION:	PFIC1, a type of progressive familial intrahepathic cholestasis (PFIC, see this term), is an infantile hereditary disorder in bile formation that is hepatocellular in origin and associated with extrahepatic features.
ORPHACODE:	79306
SYNONYMS:	Byler disease FIC1 deficiency PFIC1
XREF(S):	Orphanet ICD-10 MeSH OMIM
ANALYTE(S):	MYO5B ATP8B1
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

Related Genetic Tests

- Cholestasis (gene panel)
- Cholestasis, intrahepatic (1 gene among ATP8B1, ABCB11,ABCB4)
- Cholestasis, intrahepatic (2 genes among ATP8B1, ABCB11, ABCB4)
- Cholestasis, progressive familial intrahepatic (gene panel)

Related Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman
- Centre de Génétique Médicale UCL

Related Analytes

- ATPase phospholipid transporting 8B1
- myosin VB

Related Gene Panels

- Cholestasis (40 genes) - UCL