DISEASE:
Joubert syndrome with hepatic defect

NAME:	Joubert syndrome with hepatic defect
DESCRIPTION:	Joubert syndrome with hepatic defect is a very rare subtype of Joubert syndrome and related disorders (JSRD, see this term) characterized by the neurological features of JS associated with congenital hepatic fibrosis (CHF).
ORPHACODE:	1454
SYNONYMS:	COACH syndrome Cerebellar vermis hypoplasia-oligophrenia-congenital ataxia-coloboma-hepatic fibrosis Gentile syndrome JS-H Joubert syndrome with congenital hepatic fibrosis
XREF(S):	Orphanet OMIM ICD-10 OMIM OMIM

ANALYTE(S):	TMEM67 RPGRIP1L CC2D2A INPP5E
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/657

RELATED CONTENT

Related Genetic Tests

- Cholestasis (gene panel)
- Ciliopathy (gene panel)
- Ciliopathy / polycystic kidney and liver diseases / ADTKD/ nephronophtisis / Bardet-Biedl syndromes and kidney cancers (gene panel)
- Early-onset severe obesity
- Hepatorenal disorders (gene panel)
- Renal or urinary tract malformation (CAKUT) (gene panel)

Related Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman
- Centre de Génétique Médicale UCL
- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Gent

Related Analytes

- coiled-coil and C2 domain containing 2A
- inositol polyphosphate-5-phosphatase E
- RPGRIP1 like
- transmembrane protein 67

Related Gene Panels

- Cakut (congenital anomalies of the kidney and urinary tract-1) (69 genes) - IPG
- Cholestasis (40 genes) - UCL
- Ciliopathy (120 genes) - UGent
- Ciliopathy, polycystic kidney and liver diseases, ADTKD, nephronophtisis, Bardet-Biedl syndromes and kidney cancers (146 genes) - IPG
- Early-onset severe obesity (44 genes) - ULG
- Hepatorenal disorders (13 genes) - UCL
