DISEASE:
Joubert syndrome with ocular defect

NAME:	Joubert syndrome with ocular defect
DESCRIPTION:	Joubert syndrome with ocular defect is, along with pure JS, the most frequent subtype of Joubert syndrome and related disorders (JSRD, see these terms) characterized by the neurological features of JS associated with retinal dystrophy.
ORPHACODE:	220493
SYNONYMS:	JS-O Joubert syndrome with retinopathy
XREF(S):	Orphanet ICD-10 OMIM OMIM OMIM OMIM ICD-10 OMIM

ANALYTE(S):	AHI1 MKS1 INPP5E CEP41 CEP120
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RELATED CONTENT

Related Genetic Tests

- Ciliopathy / polycystic kidney and liver diseases / ADTKD/ nephronophtisis / Bardet-Biedl syndromes and kidney cancers (gene panel)
- Early-onset severe obesity
- cleft lip with/whitout cleft palate (virtual 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)

Related Analytes

- Abelson helper integration site 1
- centrosomal protein 120
- centrosomal protein 41
- inositol polyphosphate-5-phosphatase E
- MKS transition zone complex subunit 1

Related Gene Panels

- Ciliopathy, polycystic kidney and liver diseases, ADTKD, nephronophtisis, Bardet-Biedl syndromes and kidney cancers (146 genes) - IPG
- Cleft lip and palate / dysmorphic facial features / craniofacial anomalies (255 genes)) - UCL

- Early-onset severe obesity (44 genes) - ULG

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