DISEASE:
Joubert syndrome with oculorenal defect

NAME:	Joubert syndrome with oculorenal defect
DESCRIPTION:	A rare subtype of Joubert syndrome (JS) and related disorders (JSRD) characterized by the neurological features of JS associated with both renal and ocular disease.
ORPHACODE:	2318
SYNONYMS:	Arima syndrome CORS Cerebellooculorenal syndrome Dekaban-Arima syndrome JS type B JS-OR Joubert syndrome with Senior-Loken syndrome

XREF(S):	Orphanet OMIM OMIM OMIM OMIM OMIM OMIM OMIM ICD-10
ANALYTE(S):	CEP290 CC2D2A TMEM216 TMEM237 TMEM138 ZNF423 TMEM231
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RELATED CONTENT

Related Genetic Tests

- Ciliopathy (gene panel)
- Ciliopathy / polycystic kidney and liver diseases / ADTKD/ nephronophtisis / Bardet-Biedl syndromes and kidney cancers (gene panel)
- Early-onset severe obesity

Related Laboratories

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

Related Analytes

- coiled-coil and C2 domain containing 2A
- centrosomal protein 290
- transmembrane protein 138
- transmembrane protein 216
- transmembrane protein 231
- transmembrane protein 237
- zinc finger protein 423

Related Gene Panels

- 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
