DISEASE:
Joubert syndrome

NAME:	Joubert syndrome
DESCRIPTION:	A rare, autosomal recessive congenital cerebellar ataxia characterized by congenital malformation of the brainstem and agenesis or hypoplasia of the cerebellar vermis leading to an abnormal respiratory pattern, nystagmus, hypotonia, ataxia, and delay in achieving motor milestones.
ORPHACODE:	475
SYNONYMS:	CPD IV Cerebelloparenchymal disorder IV Classic Joubert syndrome Joubert syndrome type A Joubert-Boltshauser syndrome Pure Joubert syndrome

XREF(S):	Orphanet OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM ICD-10 OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM

ANALYTE(S):	CEP120 PIBF1 OFD1 TCTN3 PDE6D KIAA0753 IFT74 B9D2 TCTN2 AHI1 TMEM67 MKS1 ARL13B INPP5E TCTN1 B9D1 TMEM237 CEP41 CPLANE1 CSPP1 KIAA0586 CEP104 KATNIP SUFU ARMC9 CBY1 HYLS1 TOGARAM1 TMEM218 ARL3
CREATED:	13 May 2019 - 01:02

CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/521

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)
- 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)
- Centrum Medische Genetica - UZ Gent

Related Analytes

- Abelson helper integration site 1
- ADP ribosylation factor like GTPase 13B
- ADP ribosylation factor like GTPase 3
- armadillo repeat containing 9
- B9 domain containing 1
- B9 domain containing 2
- chibby family member 1, beta catenin antagonist

- centrosomal protein 104
- centrosomal protein 120
- centrosomal protein 41
- ciliogenesis and planar polarity effector complex subunit 1
- centrosome and spindle pole associated protein 1
- HYLS1 centriolar and ciliogenesis associated
- intraflagellar transport 74
- inositol polyphosphate-5-phosphatase E
- katanin interacting protein
- KIAA0586
- KIAA0753
- MKS transition zone complex subunit 1
- OFD1 centriole and centriolar satellite protein
- phosphodiesterase 6D
- progesterone immunomodulatory binding factor 1
- SUFU negative regulator of hedgehog signaling
- tectonic family member 1
- tectonic family member 2
- tectonic family member 3
- transmembrane protein 218
- transmembrane protein 237
- transmembrane protein 67
- TOG array regulator of axonemal microtubules 1

Related Gene Panels

- Ciliopathy (120 genes) - UGent
- 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
- Hepatorenal disorders (13 genes) - UCL

Source URL: http://gentest.healthdata.be/disease/521