DISEASE:
Meckel syndrome

NAME:	Meckel syndrome
DESCRIPTION:	A rare, lethal, genetic, multiple congenital anomaly disorder characterized by the triad of brain malformation (mainly occipital encephalocele), large polycystic kidneys, and polydactyly, as well as associated abnormalities that may include cleft lip/palate, cardiac and genital anomalies, central nervous system (CNS) malformations, liver fibrosis, and bone dysplasia.
ORPHACODE:	564
SYNONYMS:	Dysencephalia splanchnocystica Meckel-Gruber syndrome

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

ANALYTE(S):	TXNDC15 TMEM107 TCTN1 TMEM237 RPGRIP1 CEP290 TMEM67 MKS1 RPGRIP1L CC2D2A TMEM216 TCTN3 TCTN2 B9D1 B9D2 TMEM231 CSPP1
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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)
- 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

- B9 domain containing 1
- B9 domain containing 2
- coiled-coil and C2 domain containing 2A
- centrosomal protein 290
- centrosome and spindle pole associated protein 1
- MKS transition zone complex subunit 1

- RPGR interacting protein 1
- RPGRIP1 like
- tectonic family member 1
- tectonic family member 2
- tectonic family member 3
- transmembrane protein 107
- transmembrane protein 216
- transmembrane protein 231
- transmembrane protein 237
- transmembrane protein 67
- thioredoxin domain containing 15

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
- Cleft lip and palate / dysmorphic facial features / craniofacial anomalies (255 genes)) - UCL
- Early-onset severe obesity (44 genes) - ULG
- Hepatorenal disorders (13 genes) - UCL
