ANALYTE:
MKS1

NAME:	MKS transition zone complex subunit 1
SYMBOL:	MKS1
VERSION OF ORPHANET:	2023-06-22 14:14:43
SYNONYMS:	BBS13 FLJ20345 POC12 POC12 centriolar protein homolog (Chlamydomonas)
XREF(S):	Orphanet Ensembl Genatlas HGNC OMIM Reactome SwissProt
CREATED:	13 May 2019 - 01:01
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Ataxia (autosomic dominant and recessive / except expansion of triplets) (gene panel - 722 genes)
- Cholestasis (gene panel)
- Ciliopathy (gene panel)
- Ciliopathy / polycystic kidney and liver diseases / ADTKD/ nephronophtisis / Bardet-Biedl syndromes and kidney cancers (gene panel)
- Congenital malformation (gene panel - 1721 genes)
- Congenital malformation gene panel
- Early-onset severe obesity
- Hepatology (gene panel)
- Hepatorenal disorders (gene panel)
- Inherited Kidney Diseases (Gene Panel)
- Intellectual disability & Epilepsy (gene panel)
- Intellectual disability (gene panel)
- Intellectual disability (virtual gene panel)
- Nephrogenetics / Nephropathy (gene panel)
- Primary ciliary dyskinesia (PCD) Heterotaxyies (gene panel)
- Retinal dystrophy / RETNET (gene panel)
- Skeletal dysplasia (gene panel)
- Skeletal dysplasia (gene panel)
- Skeletal dysplasia (gene panel)
- cleft lip with/whitout cleft palate (virtual gene panel)

Related Diseases

- Bardet-Biedl syndrome

- Joubert syndrome
- Joubert syndrome with ocular defect
- Meckel syndrome

Related Gene Panels

- Ataxia (348 genes) - ULB
- 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
- Congenital malformation (1721 genes) - ULB
- Congenital malformation gene panel - VUB
- Early-onset severe obesity (44 genes) - ULG
- Hepatology panel - UGent
- Hepatorenal disorders (13 genes) - UCL
- Heterotaxie PCD - UGent
- Intellectual disability & Epilepsy - UGent
- Intellectual disability (gene panel)
- Intellectual disability/Epilepsy (1091 genes) - ULG
- Nephropathy panel - UGent
- Panel Nephro-ULG-V1
- Retinal dystrophy - UGent
- Skeletal dysplasia (394 genes) - VUB
- Skeletal dysplasia (genepanel) - UZA
- Skeletal dysplasia - UGent
