ANALYTE:
INPP5E

NAME:	inositol polyphosphate-5-phosphatase E
SYMBOL:	INPP5E
VERSION OF ORPHANET:	2023-06-22 14:14:43
SYNONYMS:	CORS1 PPI5PIV pharbin
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)
- 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 epileptic encephalopathy (gene panel - 845 genes)
- Early-onset severe obesity
- Epilepsy gene panel
- Hereditary Spastic Paraplegia (gene panel)
- Intellectual disability & Epilepsy (gene panel)
- Intellectual disability (gene panel)
- Intellectual disability (virtual gene panel)
- Nephrogenetics / Nephropathy (gene panel)
- Neurodevelopmental disorders (1300 genes)
- Neurodevelopmental disorders gene panel
- Retinal dystrophy / RETNET (gene panel)

Related Diseases

- Joubert syndrome
- Joubert syndrome with hepatic defect
- Joubert syndrome with ocular defect
- MORM syndrome

Related Gene Panels

- Ataxia (348 genes) - ULB
- Ciliopathy (120 genes) - UGent
- Ciliopathy, polycystic kidney and liver diseases, ADTKD, nephronophtisis, Bardet-Biedl syndromes and kidney cancers (146 genes) - IPG
- Congenital malformation (1721 genes) - ULB
- Congenital malformation gene panel - VUB
- Early onset epileptic encephalopathy (845 genes) - ULB
- Early-onset severe obesity (44 genes) - ULG
- Epilepsy gene panel - VUB
- Hereditary Spastic Paraplegia & ataxia (genepanel) - UZA
- Intellectual disability & Epilepsy - UGent
- Intellectual disability (gene panel)
- Intellectual disability/Epilepsy (1091 genes) - ULG
- Nephropathy panel - UGent
- Neurodevelopmental disorders (1300 genes) - ULB
- Neurodevelopmental disorders: developmental delay, intellectual disability, autistic disorders (1162 genes) - VUB
- Retinal dystrophy - UGent
