Belgian Genetic Tests database

ANALYTE:
CC2D2A

NAME:	coiled-coil and C2 domain containing 2A
SYMBOL:	CC2D2A
VERSION OF ORPHANET:	2023-06-22 14:14:43
SYNONYMS:	JBTS9 KIAA1345 MKS6 Meckel syndrome, type 6
XREF(S):	Orphanet Reactome SwissProt Ensembl Genatlas HGNC OMIM
CREATED:	13 May 2019 - 01:01
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RELATED CONTENT

Related Genetic Tests

- Ataxia (autosomic dominant and recessive / except expansion of triplets) (gene panel - 722 genes)
- Ataxia Spasticity (gene panel)
- 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 epileptic encephalopathy (gene panel - 845 genes)
- End-stage renal disease, ESRD (gene panel)
- Epilepsy gene panel
- 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)
- Microphthalmia / Anophthalmia / Coloboma-Anterior Segment Dysgenesis (MAC-ASD) (gene panel)
- Nephrogenetics / Nephropathy (gene panel)
- Neurodevelopmental disorders (1300 genes)
- Neurodevelopmental disorders 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

- Joubert syndrome with hepatic defect
- Joubert syndrome with oculorenal defect
- Meckel syndrome
- Retinitis pigmentosa

Related Gene Panels

- Ataxia (348 genes) - ULB
- Ataxia Spasticity - UGent
- 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 epileptic encephalopathy (845 genes) - ULB
- End-stage renal disease (106 genes) - IPG
- Epilepsy gene panel - VUB
- Hepatology panel - UGent
- Hepatorenal disorders (13 genes) - UCL
- Intellectual disability & Epilepsy - UGent
- Intellectual disability (gene panel)
- Intellectual disability/Epilepsy (1091 genes) - ULG
- Microphthalmia/Anophthalmia/Coloboma - Anterior Segment Dysgenesis - UGent
- Nephropathy panel - UGent
- Neurodevelopmental disorders (1300 genes) - ULB
- Neurodevelopmental disorders: developmental delay, intellectual disability, autistic disorders (1162 genes) - VUB
- Panel Nephro-ULG-V1
- Retinal dystrophy - UGent

- Skeletal dysplasia (394 genes) - VUB
- Skeletal dysplasia (genepanel) - UZA
- Skeletal dysplasia - UGent

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