ANALYTE:
KMT2D

NAME:	lysine methyltransferase 2D
SYMBOL:	KMT2D
VERSION OF ORPHANET:	2023-06-22 14:14:43
SYNONYMS:	ALR CAGL114 MLL4 histone-lysine N-methyltransferase 2D
XREF(S):	Orphanet Ensembl HGNC OMIM Reactome SwissProt Genatlas
CREATED:	13 May 2019 - 01:01
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RELATED CONTENT

Related Genetic Tests

- Congenital malformation (gene panel - 1721 genes)
- Congenital malformation gene panel
- Congenital structural heart defects (gene panel)
- Dermatogenetic panel, severe, rare and hereditary genodermatoses (gene panel - 394 genes)
- Early onset epileptic encephalopathy (gene panel - 845 genes)
- Epilepsy gene panel
- Epilepsy, seizures (gene panel)
- Inherited Kidney Diseases (Gene Panel)
- Intellectual disability & Epilepsy (gene panel)
- Intellectual disability (gene panel)
- Intellectual disability (virtual gene panel)
- Kabuki syndrome (gene panel)
- Microphthalmia / Anophthalmia / Coloboma-Anterior Segment Dysgenesis (MAC-ASD) (gene panel)
- Neurodevelopmental disorders (1300 genes)
- Neurodevelopmental disorders gene panel
- Onco-endocrine pathologies (gene panel)
- Paraganglioma and pheochromocytoma (gene panel)
- Primary immune deficiencies (gene panel)
- Primary immune deficiencies (gene panel)
- Short Stature (gene panel)
- Short stature/ Growth retardation/ (gene panel)
- Skeletal dysplasia (gene panel)
- Thyroid disgenesis (38 genes)
- cleft lip with/whitout cleft palate (virtual gene panel)

Related Diseases

- Choanal atresia-athelia-hypothyroidism-delayed puberty-short stature syndrome
- Kabuki syndrome

Related Gene Panels

- Cleft lip and palate / dysmorphic facial features / craniofacial anomalies (255 genes)) - UCL
- Congenital malformation (1721 genes) - ULB
- Congenital malformation gene panel - VUB
- Congenital structural heart defects - UGent
- Dermatogenetic / severe, rare and hereditary genodermatoses (394 genes) - ULB
- Early onset epileptic encephalopathy (845 genes) - ULB
- Epilepsy gene panel - VUB
- Epilepsy, seizures (196 genes) - IPG
- Growth retardation/short stature (genepanel) - UZA
- Intellectual disability & Epilepsy - UGent
- Intellectual disability (gene panel)
- Intellectual disability/Epilepsy (1091 genes) - ULG
- Kabuki (7 genes) - IPG
- Microphthalmia/Anophthalmia/Coloboma - Anterior Segment Dysgenesis - UGent
- Neurodevelopmental disorders (1300 genes) - ULB
- Neurodevelopmental disorders: developmental delay, intellectual disability, autistic disorders (1162 genes) - VUB
- Onco-endocine pathologies (50 genes) - UCL
- Panel Nephro-ULG-V1
- Paraganglioma and pheochromocytoma (29 genes) - UCL
- Primary immune deficiencies (444 genes) - KUL
- Primary immune deficiencies - UGent
- Short Stature (46 genes) - IPG
- Skeletal dysplasia - UGent
- Thyroid disgenesis (38 genes) - VUB
