ANALYTE:
FGFR1

NAME:	fibroblast growth factor receptor 1
SYMBOL:	FGFR1
VERSION OF ORPHANET:	2023-06-22 14:14:43
SYNONYMS:	BFGFR CD331 CEK FLG H2 H3 H4 H5 N-SAM Pfeiffer syndrome

XREF(S):	Orphanet Ensembl Genatlas HGNC OMIM Reactome SwissProt
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RELATED CONTENT

Related Genetic Tests

- Brain malformations (gene panel)
- Congenital malformation (gene panel - 1721 genes)
- Congenital malformation gene panel
- Congenital structural heart defects (gene panel)
- Craniosynostosis (gene panel)
- Craniosynostosis syndrome (hot spot mutation - p.Pro252Arg)
- Dermatogenetic panel, severe, rare and hereditary genodermatoses (gene panel - 394 genes)
- Disorders of sex development - Primary Ovarian insufficiency - Hypogonadotropic Hypogonadism (gene panel)
- Hypogonadotropic hypogonadism (33 genes)
- Intellectual disability & Epilepsy (gene panel)
- Intellectual disability (gene panel)
- Intellectual disability (virtual gene panel)
- Kallmann syndrome / Hypogonadotropic Hypogonadism (FGFR1 gene)
- Maffucci syndrome (gene panel)
- Nephrogenetics / Nephropathy (gene panel)
- Neurodevelopmental disorders (1300 genes)
- Neurodevelopmental disorders gene panel
- Overgrowth & vascular anomalies (gene panel)
- Skeletal dysplasia (gene panel)
- Skeletal dysplasia (gene panel)
- Skeletal dysplasia (gene panel)
- Skin disorders (gene panel)
- Sturge-Weber syndrome (gene panel)
- cleft lip with/whitout cleft palate (virtual gene panel)

Related Diseases

- Encephalocraniocutaneous lipomatosis
- Giant cell glioblastoma
- Gliosarcoma
- Hartsfield syndrome
- Holoprosencephaly
- Kallmann syndrome
- Lobar holoprosencephaly
- Microform holoprosencephaly
- Myeloid/lymphoid neoplasm associated with FGFR1 rearrangement
- NON RARE IN EUROPE: Hypodontia
- Non-syndromic metopic craniosynostosis
- Normosmic congenital hypogonadotropic hypogonadism
- Oligodontia
- Osteoglosphonic dysplasia
- Pfeiffer syndrome type 1
- Pilomyxoid astrocytoma
- Primary hypereosinophilic syndrome
- Semilobar holoprosencephaly
- Septo-optic dysplasia spectrum

Related Gene Panels

- Brain malformations (34 genes) - ULB
- 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
- Craniosynostosis (32 genes) - KUL
- Dermatogenetic / severe, rare and hereditary genodermatoses (394 genes) - ULB

- Disorders of Sex Development - Primary Ovarian Insufficiency - Hypogonadotropic Hypogonadism - UGent
- Hypogonadotropic Hypogonadism/Kallmann (61 genes) - ULG
- Hypogonadotropic hypogonadism (33 genes) - VUB
- Intellectual disability & Epilepsy - UGent
- Intellectual disability (gene panel)
- Intellectual disability/Epilepsy (1091 genes) - ULG
- Maffucci syndrome (65 genes) - KUL
- Nephropathy panel - UGent
- Neurodevelopmental disorders (1300 genes) - ULB
- Neurodevelopmental disorders: developmental delay, intellectual disability, autistic disorders (1162 genes) - VUB
- Overgrowth & vascular anomalies (65 genes) - KUL
- Skeletal dysplasia (394 genes) - VUB
- Skeletal dysplasia (genepanel) - UZA
- Skeletal dysplasia - UGent
- Skin disorders - UGent
- Sturge-Weber syndrome (65 genes) - KUL
