

ANALYTE:
CREBBP

NAME:	CREB binding protein
SYMBOL:	CREBBP
VERSION OF ORPHANET:	2023-06-22 14:14:43
SYNONYMS:	CBP KAT3A RTS
XREF(S):	<u>Orphanet</u> <u>Ensembl</u> <u>Genatlas</u> <u>HGNC</u> <u>OMIM</u> <u>Reactome</u> <u>SwissProt</u>
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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 \(gene panel - 394 genes\)](#)
- [Disorders of sex development - Primary Ovarian insufficiency - Hypogonadotropic Hypogonadism \(gene panel\)](#)
- [Early-onset severe obesity](#)
- [Glaucoma \(gene panel\)](#)
- [Intellectual Disability \(gene panel\)](#)
- [Intellectual disability & Epilepsy \(gene panel\)](#)
- [Intellectual disability \(gene panel\)](#)
- [Intellectual disability \(virtual gene panel\)](#)
- [Neurodevelopmental disorders \(1300 genes\)](#)
- [Neurodevelopmental disorders gene panel](#)
- [Pediatric oncopredisposition \(gene panel\)](#)
- [Primary immune deficiencies \(gene panel\)](#)
- [Short Stature \(gene panel\)](#)
- [Short stature/ Growth retardation/ \(gene panel\)](#)
- [Skeletal dysplasia \(gene panel\)](#)
- [Skeletal dysplasia \(gene panel\)](#)
- [Skeletal dysplasia \(gene panel\)](#)

Related Diseases

- [Acute myeloid leukemia with t\(8;16\)\(p11;p13\) translocation](#)

- Menke-Hennekam syndrome
- Rubinstein-Taybi syndrome due to 16p13.3 microdeletion
- Rubinstein-Taybi syndrome due to CREBBP mutations

Related Gene Panels

- Congenital malformation (1721 genes) - ULB
- Congenital malformation gene panel - VUB
- Congenital structural heart defects - UGent
- Dermatogenetic / severe, rare and hereditary genodermatoses (394 genes) - ULB
- Disorders of Sex Development - Primary Ovarian Insufficiency - Hypogonadotropic Hypogonadism - UGent
- Early-onset severe obesity (44 genes) - ULG
- Glaucoma - UGent
- Growth retardation/short stature (genepanel) - UZA
- Intellectual Disability (104 genes) - IPG
- Intellectual disability & Epilepsy - UGent
- Intellectual disability (gene panel)
- Intellectual disability/Epilepsy (1091 genes) - ULG
- Neurodevelopmental disorders (1300 genes) - ULB
- Neurodevelopmental disorders: developmental delay, intellectual disability, autistic disorders (1162 genes) - VUB
- Pediatric oncopredisposition - UGent
- Primary immune deficiencies - UGent
- Short Stature (46 genes) - IPG
- Skeletal dysplasia (394 genes) - VUB
- Skeletal dysplasia (genepanel) - UZA
- Skeletal dysplasia - UGent

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