

ANALYTE:
NIPBL

NAME:	NIPBL cohesin loading factor
SYMBOL:	NIPBL
VERSION OF ORPHANET:	2023-06-22 14:14:43
SYNONYMS:	DKFZp434L1319 FLJ11203 FLJ12597 FLJ13354 FLJ13648 IDN3 Scc2 sister chromatid cohesion 2 homolog (yeast)
XREF(S):	Orphanet Ensembl Genatlas HGNC OMIM Reactome SwissProt
CREATED:	13 May 2019 - 01:01

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RELATED CONTENT

Related Genetic Tests

- [Autism \(gene panel\)](#)
- [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](#)
- [Inherited Kidney Diseases \(Gene Panel\)](#)
- [Intellectual Disability \(gene panel\)](#)
- [Intellectual disability & Epilepsy \(gene panel\)](#)
- [Intellectual disability \(gene panel\)](#)
- [Intellectual disability \(virtual gene panel\)](#)
- [Malformations of cortical development \(235 genes\)](#)
- [Neurodevelopmental disorders \(1300 genes\)](#)
- [Neurodevelopmental disorders gene panel](#)
- [Skeletal dysplasia \(gene panel\)](#)
- [Skeletal dysplasia \(gene panel\)](#)
- [Skeletal dysplasia \(gene panel\)](#)
- [cleft lip with/without cleft palate \(virtual gene panel\)](#)

Related Diseases

- 5p13 microduplication syndrome
- Cornelia de Lange syndrome

Related Gene Panels

- Autism (57 genes) - IPG
- 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
- Intellectual Disability (104 genes) - IPG
- Intellectual disability & Epilepsy - UGent
- Intellectual disability (gene panel)
- Intellectual disability/Epilepsy (1091 genes) - ULG
- Malformations of cortical development (235 genes) - VUB
- Neurodevelopmental disorders (1300 genes) - ULB
- Neurodevelopmental disorders: developmental delay, intellectual disability, autistic disorders (1162 genes) - VUB
- Panel Nephro-ULG-V1
- Skeletal dysplasia (394 genes) - VUB
- Skeletal dysplasia (genepanel) - UZA
- Skeletal dysplasia - UGent

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