

ANALYTE:
NSD1

NAME:	nuclear receptor binding SET domain protein 1
SYMBOL:	NSD1
VERSION OF ORPHANET:	2023-06-22 14:14:43
SYNONYMS:	ARA267 FLJ22263 KMT3B
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 \(gene panel - 1721 genes\)](#)
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- [Intellectual disability \(gene panel\)](#)
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Related Diseases

- [5q35 microduplication syndrome](#)
- [Beckwith-Wiedemann syndrome due to NSD1 mutation](#)
- [Deletion 5q35](#)

- Sotos syndrome
- Weaver syndrome

Related Gene Panels

- Congenital malformation (1721 genes) - ULB
- Congenital malformation gene panel - VUB
- Congenital structural heart defects - UGent
- Early onset epileptic encephalopathy (845 genes) - ULB
- Epilepsy gene panel - VUB
- Heterotaxie PCD - UGent
- 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
- Overgrowth (24 genes) - IPG
- Pediatric oncopredisposition - UGent
- Skeletal dysplasia (394 genes) - VUB
- Skeletal dysplasia (genepanel) - UZA
- Skeletal dysplasia - UGent

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