

**ANALYTE:**  
**XYLT1**

|                             |                                                                                                                                                                                          |
|-----------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>NAME:</b>                | xylosyltransferase 1                                                                                                                                                                     |
| <b>SYMBOL:</b>              | XYLT1                                                                                                                                                                                    |
| <b>VERSION OF ORPHANET:</b> | 2023-06-22 14:14:43                                                                                                                                                                      |
| <b>SYNONYMS:</b>            | PXYLT1<br>XT-I<br>protein xylosyltransferase 1                                                                                                                                           |
| <b>XREF(S):</b>             | <a href="#">Orphanet</a><br><a href="#">Ensembl</a><br><a href="#">Genatlas</a><br><a href="#">HGNC</a><br><a href="#">OMIM</a><br><a href="#">SwissProt</a><br><a href="#">Reactome</a> |
| <b>CREATED:</b>             | 13 May 2019 - 01:01                                                                                                                                                                      |
| <b>CHANGED:</b>             | 22 Jun 2023 - 16:14                                                                                                                                                                      |

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## RELATED CONTENT

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### Related Genetic Tests

- [Congenital malformation \(gene panel - 1721 genes\)](#)
- [Ehlers-Danlos syndroom, EDS \(gene panel\)](#)
- [Intellectual disability & Epilepsy \(gene panel\)](#)
- [Intellectual disability \(gene panel\)](#)
- [Intellectual disability \(virtual gene panel\)](#)
- [Metabolic disorders including disorders of glycosylation, peroxisomal disorders, organic acidurias, glycogenosis disorders, neurotransmitter disorders \(213 genes\)](#)
- [Myopia \(early onset high myopia\)](#)
- [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

- [Desbuquois syndrome](#)
- [XYLT1-CDG](#)

### Related Gene Panels

- [Cleft lip and palate / dysmorphic facial features / craniofacial anomalies \(255 genes\)\) - UCL](#)
- [Congenital malformation \(1721 genes\) - ULB](#)

- Ehlers-Danlos syndrome -UGent
- Intellectual disability & Epilepsy - UGent
- Intellectual disability (gene panel)
- Intellectual disability/Epilepsy (1091 genes) - ULG
- Metabolic disorders (213 genes) - VUB
- Myopia gene panel - UGent
- Neurodevelopmental disorders (1300 genes) - ULB
- Neurodevelopmental disorders: developmental delay, intellectual disability, autistic disorders (1162 genes) - VUB
- Recessive Ehlers-Danlos Syndrome (11 genes) - UGent
- Skeletal dysplasia (394 genes) - VUB
- Skeletal dysplasia (genepanel) - UZA
- Skeletal dysplasia - UGent

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