ANALYTE:
EXT1

NAME:	exostosin glycosyltransferase 1
SYMBOL:	EXT1
VERSION OF ORPHANET:	2023-06-22 14:14:43
SYNONYMS:	Glucuronosyl-N-acetylglucosaminyl-proteoglycan 4-alpha-N- acetylglucosaminyltransferase N-acetylglucosaminyl-proteoglycan 4-beta-glucuronosyltransferase ttv
XREF(S):	Orphanet HGNC OMIM Reactome SwissProt Ensembl Genatlas
CREATED:	13 May 2019 - 01:01
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Congenital malformation (gene panel - 1721 genes)
- Intellectual disability (virtual gene panel)
- Metabolic disorders including disorders of glycosylation, peroxisomal disorders, organic acidurias, glycogenosis disorders, neurotransmitter disorders (213 genes)
- Multiple osteochondromas (2 genes)
- Neurodevelopmental disorders (1300 genes)
- Neurodevelopmental disorders gene panel
- Skeletal dysplasia (gene panel)
- Skeletal dysplasia (gene panel)
- Skeletal dysplasia (gene panel)

Related Diseases

- Chondrosarcoma
- Multiple osteochondromas
- Trichorhinophalangeal syndrome type 2

Related Gene Panels

- Congenital malformation (1721 genes) - ULB
- Intellectual disability (gene panel)
- Metabolic disorders (213 genes) - VUB
- Multiple osteochondromas (2 genes) - UZA
- Neurodevelopmental disorders (1300 genes) - ULB

- Neurodevelopmental disorders: developmental delay, intellectual disability, autistic disorders (1162 genes) - VUB
- Skeletal dysplasia (394 genes) - VUB
- Skeletal dysplasia (genepanel) - UZA
- Skeletal dysplasia - UGent
