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EXT1 Rabbit pAb

Catalog # AP97711

Product Information

Application	IHC-P, IHC-F, IF, E
Primary Accession	Q16394
Other Accession	Q16394
Predicted	Rat, Baboon, Orangutan, Human, Mouse, Rabbit, Chicken, Horse
Host	Rabbit
Clonality	Polyclonal
Calculated MW	86255
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human EXT1
Epitope Specificity	651-746/746
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SIMILARITY	Belongs to the glycosyltransferase 47 family.
DISEASE	Defects in EXT1 are a cause of hereditary multiple exostoses type 1 (EXT1) [MIM:133700]. EXT is a genetically heterogeneous bone disorder caused by genes segregating on human chromosomes 8, 11, and 19 and designated EXT1, EXT2 and EXT3 respectively. EXT is a dominantly inherited skeletal disorder primarily affecting endochondral bone during growth. The disease is characterized by formation of numerous cartilage-capped, benign bone tumors (osteocartilaginous exostoses or osteochondromas) that are often accompanied by skeletal deformities and short stature. In a small percentage of cases exostoses have exhibited malignant transformation resulting in an osteosarcoma or chondrosarcoma. Osteochondromas development can also occur as a sporadic event. Defects in EXT1 are a cause of tricho-rhino-phalangeal syndrome type 2 (TRPS2) [MIM:150230]. A syndrome that combines the clinical features of trichorhinophalangeal syndrome type 1 and multiple exostoses type 1. Affected individuals manifest multiple dysmorphic facial features including large, laterally protruding ears, a bulbous nose, an elongated upper lip, as well as sparse scalp hair, winged scapulae, multiple cartilaginous exostoses, redundant skin, and mental retardation. Note=A chromosomal aberration resulting in the loss of functional copies of TRPS1 and EXT1 has been found in TRPS2 patients. Defects in EXT1 are a cause of chondrosarcoma (CHDSA) [MIM:215300]. It is a malignant neoplasm derived from cartilage cells. Chondrosarcomas range from slow-growing non-metastasizing lesions to highly aggressive metastasizing sarcomas.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	This gene encodes an endoplasmic reticulum-resident type II transmembrane glycosyltransferase involved in the chain elongation step of heparan sulfate biosynthesis. Mutations in this gene cause the type I form of multiple exostoses. [provided by RefSeq, Jul 2008]

Additional Information

Gene ID	2131
Other Names	EXT; LGCR; LGS; TRPS2; TTV; EXT1_CRIGR; EXT1; Heparan sulfate copolymerase; N-acetylglucosaminyl-proteoglycan 4-beta-glucuronosyltransferase; 2.4.1.225; EXT1_BOVIN; EXT1_HUMAN; Exostosin glycosyltransferase 1; Heparan sulfate co-polymerase subunit EXT1; Multiple exostoses protein 1; EXT1_MOUSE; Multiple exostoses protein 1 homolog; EXT1_PONAB; Langer-Giedion syndrome chromosome region; exostoses (multiple) 1; exostosin 1; Glucuronosyl-N-acetylglucosaminyl-proteoglycan 4-alpha-N-acetylglucosaminyltransferase
Dilution	IHC-P=1:100-500,IHC-F=1:100-500,ICC/IF=1:100-500,IF=1:100-500,ELISA=1:5000-10000
Storage	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.

Protein Information

Name	EXT1 (HGNC:3512)
Function	Glycosyltransferase forming with EXT2 the heterodimeric heparan sulfate polymerase which catalyzes the elongation of the heparan sulfate glycan backbone (PubMed:10639137, PubMed:22660413, PubMed:36402845, PubMed:36593275, PubMed:9620772). Glycan backbone extension consists in the alternating transfer of (1->4)-beta-D-GlcA and (1->4)-alpha-D-GlcNAc residues from their respective UDP-sugar donors. Both EXT1 and EXT2 are required for the full activity of the polymerase since EXT1 bears the N-acetylglucosaminyl-proteoglycan 4- beta-glucuronosyltransferase activity within the complex while EXT2 carries the glucuronosyl-N-acetylglucosaminyl-proteoglycan 4-alpha-N-acetylglucosaminyltransferase activity (PubMed:36402845, PubMed:36593275). Heparan sulfate proteoglycans are ubiquitous components of the extracellular matrix and play an important role in tissue homeostasis and signaling (PubMed:10639137, PubMed:11391482, PubMed:22660413, PubMed:9620772).
Cellular Location	Golgi apparatus membrane; Single-pass type II membrane protein. Golgi apparatus, cis-Golgi network membrane; Single-pass type II membrane protein. Endoplasmic reticulum membrane; Single-pass type II membrane protein. Note=The active heparan sulfate polymerase complex composed of EXT1 and EXT2 is localized to the Golgi apparatus. If both proteins are individually detected in the endoplasmic reticulum, the formation of the complex promotes their transport to the Golgi.
Tissue Location	Widely expressed..

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