

Product datasheet for **TA327231**

EXT1 Rabbit Polyclonal Antibody

Product data:

Product Type:	Primary Antibodies
Applications:	ICC/IF, WB
Recommended Dilution:	WB 1:500 - 1:2000;IF 1:10 - 1:100
Reactivity:	Human, Mouse, Rat
Host:	Rabbit
Isotype:	IgG
Clonality:	Polyclonal
Immunogen:	Recombinant protein of human EXT1
Formulation:	Store at -20C or -80C. Avoid freeze / thaw cycles. Buffer: PBS with 0.02% sodium azide, 50% glycerol, pH7.3
Concentration:	lot specific
Purification:	Affinity purification
Conjugation:	Unconjugated
Storage:	Store at -20°C as received.
Stability:	Stable for 12 months from date of receipt.
Predicted Protein Size:	86 kDa
Gene Name:	exostosin glycosyltransferase 1
Database Link:	NP_000118 Entrez Gene 14042 Mouse Entrez Gene 299907 Rat Entrez Gene 2131 Human Q16394



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Background:

Hereditary multiple exostoses (EXT) is an autosomal dominant disorder characterized by the formation of cartilage-capped tumors (exostoses) that develop from the growth plate of endochondral bone. This condition can lead to skeletal abnormalities, short stature and malignant transformation of exostoses to chondrosarcomas or osteosarcomas. Linkage analyses have identified three different genes for EXT, EXT1 on 8q24.1, EXT2 on 11p11-13 and EXT3 on 19p, a family of tumor suppressor genes. Most EXT cases have been attributed to missense or frameshift mutations, which lead to loss of function of the EXT genes. EXT1 is an ER-resident type II transmembrane glycoprotein and a heparan sulphate polymerase with both D-glucuronyl and N-acetyl-D-glucosaminoglycan transferase activities. Expression of EXT1 in cells results in the alteration of the synthesis and display of cell surface heparan sulfate glycosaminoglycans. EXT1 mutations have been identified in multiple types of human tumors.

Synonyms:

EXT; LGCR; LGS; TRPS2; TTV

Protein Families:

Druggable Genome, Transmembrane

Protein Pathways:

Heparan sulfate biosynthesis, Metabolic pathways

Product images: