

EXT1 Polyclonal antibody

Catalog Number: 10117-1-AP

1 Publications

Basic Information

Catalog Number:

10117-1-AP

Size:

130 µg/ml

Source:

Rabbit

Isotype:

IgG

Immunogen Catalog Number:

AG0171

GenBank Accession Number:

BC001174

GeneID (NCBI):

2131

UNIPROT ID:

Q16394

Full Name:

exostoses (multiple) 1

Calculated MW:

86 kDa

Purification Method:

Antigen affinity purification

Applications

Tested Applications:

ELISA

Cited Applications:

IF, IHC

Species Specificity:

human, mouse, rat

Cited Species:

mouse

Background Information

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.

Notable Publications

Author	Pubmed ID	Journal	Application
Nozaki Koji K	19359424	Am J Physiol Gastrointest Liver Physiol	IHC, IF

Storage

Storage:

Store at -20°C. Stable for one year after shipment.

Storage Buffer:

PBS with 0.02% sodium azide and 50% glycerol pH 7.3.

Aliquoting is unnecessary for -20°C storage

For technical support and original validation data for this product please contact:

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