



EXT1

Antibody

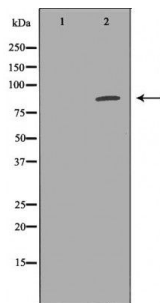
#24286

**Catalog Number:** 24286-1, 24286-2**Amount:** 50µg/50µl, 100µg/100µl**Swiss-Prot No. :**Q16394**Form of Antibody:** Rabbit IgG in phosphate buffered saline (without Mg<sup>2+</sup> and Ca<sup>2+</sup>), pH 7.4, 150mM NaCl, 0.02% sodium azide and 50% glycerol.**Storage/Stability:** Store at -20°C/1 year**Immunogen:** The antiserum was produced against synthesized peptide derived from Human EXT1**Purification:** The antibody was affinity-purified from rabbit antiserum by affinity-chromatography using epitope-specific immunogen.**Specificity/Sensitivity:** EXT1 Antibody detects endogenous levels of total EXT1**Reactivity:** Human, Mouse, Rat**Applications:**

Predicted MW:86kd

WB:1:500-2000

IHC:1:50-200



Western blot analysis of extracts of various celllines, using EXT1 antibody.

**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.