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Gene Section
Mini Review

EXT2 (exostoses (multiple) 2)

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Identity

Location: 11p11-p12

DNA/RNA

Description

Sixteen exons across the EXT2 locus were identified, two of which (1a and 1b) are alternatively spliced; spans approximately 108 kb of genomic DNA.

Transcription

3.5 and 3.7 kb.

Protein

Description

718 amino acids; 82.2 kDa.

Expression

mRNA is ubiquitously expressed.

Localisation

Endoplasmic reticulum.

Function

A tumour suppressor function is suggested; EXT2 is a glycosyltransferase, suggested to be involved in chain polymerization of heparan sulphate.

Homology

Human EXT1, EXTL1, EXTL2 and EXTL3, mouse Ext2.

Mutations

Germinal

Germline mutations in EXT2 are causative for hereditary multiple exostoses, a heterogeneous autosomal dominant disorder; mutations include nucleotide substitutions (57%), small deletions (19%) and small insertions (24%), of which the majority is predicted to result in a truncated or non-functional protein.

Somatic

No somatic mutations were found in 34 sporadic and hereditary osteochondromas and secondary peripheral chondrosarcomas tested.

Implicated in

Hereditary multiple exostoses

Prognosis

The main complication in hereditary multiple exostoses is malignant transformation of an osteochondroma (exostosis) into chondrosarcoma, which is estimated to occur in 1-5% of the HME cases.

Cytogenetics

11p rearrangement was found in 1 sporadic osteochondroma (exostosis) using cytogenetic analysis; loss of heterozygosity at the EXT2 locus was absent in 14 osteochondromas.

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