EXT2 (exostoses (multiple) 2)
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Published in Atlas Database: January 2000
Online updated version: http://AtlasGeneticsOncology.org/Genes/EXT2ID213.html
DOI: 10.4267/2042/37576

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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
Germ line 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.

References


Bridge JA, Nelson M, Orndal C, Bhatia P, Neff JR. Clonal karyotypic abnormalities of the hereditary multiple exostoses chromosomal loci 8q24.1 (EXT1) and 11p11-12 (EXT2) in patients with sporadic and hereditary osteochondromas. Cancer. 1998 May 1;82(9):1657-63


This article should be referenced as such: