Atlas of Genetics and Cytogenetics in Oncology and Haematology

OPEN ACCESS JOURNAL AT INIST-CNRS

Gene Section
Short Communication

NF2 (neurofibromin 2)

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Published in Atlas Database: March 1998

Online updated version: http://AtlasGeneticsOncology.org/Genes/NF2117.html
DOI: 10.4267/2042/37406

Identity

Other names: SCH (schwannoma)
Location: 22q12.1-12.2 junction, incidentally not far from EWS.

DNA/RNA

Description

16 exons; spans 120 kb; open reading frame: 1.8 kb.

Transcription

Alternate splicings, in particular after exon 15.

Protein

Description

Called merlin, schwannomin, or SCH; 590 or 595 amino acids; 66 kDa; NH2 -- membrane binding -- large a helix domain binding to actin of the cytoskeleton -- COOH

Expression

Wide: in lung, kidney, ovary, breast, placenta, neuroblasts; high in fetal brain.

Localisation

Membrane associated.

Function

Membrane-cytoskeleton anchor (as APC also appears to be); role in the development of extraembryonic structures before gastrulation; has characteristics of a tumour suppressor, as has been found in sporadic as well as neurofibromatosis type 2 induced schwannomas and meningiomas.

Homology

Ezrin, talin, radixin, moesin, members of the erythrocytes band 4.1 family, especially in the N-term.

Mutations

Germinal

Inborn condition of neurofibromatosis type 2 patients: protein truncations due to various frameshift deletions or insertions or nonsense mutations; splice-site or missense mutations are also found; phenotype-genotype correlations are observed (i.e. those severe phenotypes are found in cases with protein truncations rather than those with amino acid substitution).

Somatic

Mutation and allele loss events in tumours in neurofibromatosis type 2 and in sporadic schwannomas and meningiomas are in accordance with the two-hit model for neoplasia, as is found in retinoblastoma.

Implicated in

Neurofibromatosis type 2

Disease

Autosomal dominant cancer prone disease; neurofibromatosis type 2 (NF2: the same symbol is used for the disease neurofibromatosis type 2 and the gene neurofibromin 2) is a hamartoneoplastic syndrome.

Prognosis

Hamartomas have a potential towards neoplasia; those, in NF2, are schwannomas and meningiomas.

Sporadic meningioma

Sporadic schwannoma

Other tumours

Ependymoma; mesothelioma.

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This article should be referenced as such:

Huret JL. NF2 (neurofibromin 2). Atlas Genet Cytogenet Oncol Haematol.1998;2(2):39-40.