**NF2 (neurofibromatosis type 2)**

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**Identity**

Other names: SCH  
HGNC (Hugo): NF2  
Location: 22q12.1-12.2  
Local order: 22q12.1-12.2 junction, incidentally not far from EWS.

**DNA/RNA**

Description  
Axons 17 exons (1-15, 17 constitutive, 16 alternatively spliced); spans 120 kb; open reading frame: 1.8 kb.

Transcription  
Alternate splicing, in particular after exon 15.

**Protein**

Description  
Called merlin, schwannomin, or SCH; isoform 1 595 amino acids, isoform 2 590 amino acids (due to inclusion of exon 16 in transcript) : 66 KDa; NH2 -- FERM domain -- large a helix domain – COOH.

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

Localisation  
Membrane associated interacts with integral membrane proteins and actin-cytoskeleton.

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, radixin, moesin, members of the erythrocytes band 4.1 family, especially in the N-terminal FERM domain.

**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. that severe phenotype 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 tumor prone disease; neurofibromatosis type 2 (NF2: the same symbol is used for the disease neurofibromatosis type 2 and the gene) is an hamartoneoplastic syndrome.
**Prognosis**

Hamartomas have a potential towards neoplasia; those, in NF2, are the tumors of NF2 are slow-growing benign schwannomas which do not progress to malignancy and meningiomas.

**Sporadic meningioma**

**Sporadic schwannoma**

**Other tumours**: ependymoma; mesothelioma

**References**


McClatchey AI, Saotome I, Ramesh V, Gusella JF, Jacks T. The NF2 tumor suppressor gene product is essential for extraembryonic development immediately prior to gastrulation. Genes Dev. 1997 May 15;11(10):1253-65


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