NF2 (neurofibromin 2)
Jean-Loup Huret

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.

Implied 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.

References
Rouleau GA, Merel P, Lutchman M, Sanson M, Zucman J, Marineau C, Hoang-Xuan K, Demczuk S, Desmaze C,


This article should be referenced as such: