

NF2 (neurofibromatosis type 2) (updated: old version not available)

Identity

Other names **SCH**
 Hugo **NF2**
 Location 22q12.1-12.2
 22q12.1-12.2 junction, incidentally not far from [EWS](#)

DNA/RNA

Description exons 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

Entity [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

Entity [sporadic meningioma](#)

Entity sporadic schwannoma

Entity other tumours: [ependymoma](#); mesothelioma

External links

Nomenclature	
Hugo	NF2
GDB	NF2
Entrez_Gene	NF2_4771 neurofibromin 2 (bilateral acoustic neuroma)
Cards	
Atlas	NF2117
GeneCards	NF2
Ensembl	NF2
CancerGene	NF2
Genatlas	NF2
GeneLynx	NF2
eGenome	NF2
euGene	4771
Genomic and cartography	
GoldenPath	NF2 - chr22:28324119-28419137 + 22q12.2 (hg17-May_2004)
Ensembl	NF2 - 22q12.2 [CytoView]
NCBI	Genes Cyto Gene Seq [Map View - NCBI]
OMIM	Disease map [OMIM]
HomoloGene	NF2
Gene and transcription	
Genbank	AF165426 [SRS] AF165426 [ENTREZ]
Genbank	X72655 [SRS] X72655 [ENTREZ]
Genbank	X72670 [SRS] X72670 [ENTREZ]
Genbank	AF113694 [SRS] AF113694 [ENTREZ]
Genbank	AF122827 [SRS] AF122827 [ENTREZ]
RefSeq	NM_000268 [SRS] NM_000268 [ENTREZ]
RefSeq	NM_016418 [SRS] NM_016418 [ENTREZ]
RefSeq	NM_181825 [SRS] NM_181825 [ENTREZ]
RefSeq	NM_181826 [SRS] NM_181826 [ENTREZ]
RefSeq	NM_181827 [SRS] NM_181827 [ENTREZ]
RefSeq	NM_181828 [SRS] NM_181828 [ENTREZ]
RefSeq	NM_181829 [SRS] NM_181829 [ENTREZ]
RefSeq	NM_181830 [SRS] NM_181830 [ENTREZ]
RefSeq	NM_181831 [SRS] NM_181831 [ENTREZ]
RefSeq	NM_181832 [SRS] NM_181832 [ENTREZ]
RefSeq	NM_181833 [SRS] NM_181833 [ENTREZ]
RefSeq	NM_181834 [SRS] NM_181834 [ENTREZ]
RefSeq	NM_181835 [SRS] NM_181835 [ENTREZ]
RefSeq	NT_086921 [SRS] NT_086921 [ENTREZ]
AceView	NF2 AceView - NCBI
TRASER	NF2 Traser - Stanford
Unigene	Hs.187898 [SRS] Hs.187898 [NCBI] HS187898 [spliceNest]
Protein : pattern, domain, 3D structure	
SwissProt	P35240 [SRS] P35240 [EXPASY] P35240 [INTERPRO]

[CluSTr](#) [P35240](#)
[Blocks](#) [P35240](#)

Polymorphism : SNP, mutations, diseases

[OMIM](#) [607379](#) [[map](#)]

[GENECLINICS](#) [607379](#)

[SNP](#) [NF2](#) [dbSNP-NCBI]

[SNP](#) [NM_000268](#) [SNP-NCI]

[SNP](#) [NM_016418](#) [SNP-NCI]

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[SNP](#) [NM_181834](#) [SNP-NCI]

[SNP](#) [NM_181835](#) [SNP-NCI]

[SNP](#) [NF2](#) [GeneSNPs - Utah] [NF2](#) [SNP - CSHL] [NF2](#) [HGBASE - SRS]

General knowledge

[Family Browser](#) [NF2](#) [UCSC Family Browser]

[SOURCE](#) [NM_000268](#)

[SOURCE](#) [NM_016418](#)

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[SOURCE](#) [NM_181833](#)

[SOURCE](#) [NM_181834](#)

[SOURCE](#) [NM_181835](#)

[SMD](#) [Hs.187898](#)

[SAGE](#) [Hs.187898](#)

[Amigo](#) [component|cytoplasm](#)

[Amigo](#) [function|cytoskeletal protein binding](#)

[Amigo](#) [component|cytoskeleton](#)

[Amigo](#) [component|cytoskeleton](#)

[Amigo](#) [process|negative regulation of cell cycle](#)

[Amigo](#) [process|negative regulation of cell proliferation](#)

[Amigo](#) [process|perception of sound](#)

[Amigo](#) [component|plasma membrane](#)

[Amigo](#) [function|structural molecule activity](#)

[PubGene](#) [NF2](#)

Other databases

Probes

[Probe](#) [NF2 Related clones \(RZPD - Berlin\)](#)

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Medline [8453669](#)

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Medline [10992163](#)

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