

## NF1 (neurofibromin 1)

### Identity

Hugo [NF1](#)  
Location 17q11.2

### DNA/RNA

Description 60 exons (57 constitutive, 3 alternative); spans 280 kb; presence of 3 cryptic genes: OMGP, EVI2A, and EVI2B ('overlapping genes'), hidden (!) within NF1 intron 27b with an opposite transcription direction.  
Transcription at least 4 alternate splicings; ~ 9.0 kb mRNA complete cds; coding sequence: CDS 198..8717

### Protein

Description the protein has been called neurofibromin; 2818 and 2839 amino acids (type-1 and type-2 isoform)  
Expression is tissue and development stage specific  
Function GTPase activating protein (GAP) interacting with p21RAS -> tumour suppressor.  
Homology other (GAP); IRA1 and 2, the yeast inhibitors of p21RAS

### Mutations

Germinal large submicroscopic deletions in 5-10% of cases, translocations rare and point mutations in app. 85-90% of cases; widely dispersed, with no clustering, unusual splicing mutations yield difficulties in molecular genetic testing, truncating effect in large majority of cases.  
Somatic second inactivating mutation occurs in the Schwann cell of benign neurofibromas; additional genetic alterations in this cell lead to malignant transformation; the spectrum of inactivating somatic mutation not fully elucidated, LOH owing to copy number loss and mitotic recombination, point mutations; another inactivating process may involve RNA editing (for the second allele), which gives rise to a truncated neurofibromin having lost it's GAP activity.

### Implicated in

Entity [neurofibromatosis type 1](#)  
Disease autosomal dominant cancer prone disease; neurofibromatosis type 1 (NF1: the same symbol is used for the disease neurofibromatosis type 1 and the gene neurofibromin 1) is an hamartoneoplastic syndrome.

**Entity** Watson syndrome

**Disease** autosomal dominant disease with cardiac malformations, and, as is found in von Recklinghausen neurofibromatosis, low normal intelligence, café-au-lait spots, and neurofibromas but to a lesser extend.

**Oncogenesis** in accordance with the two-hit model for neoplasia, as is found in retinoblastoma.

## External links

### Nomenclature

[Hugo](#) [NF1](#)

[GDB](#) [NF1](#)

[Entrez Gene](#) [NF1 4763](#) neurofibromin 1 (neurofibromatosis, von Recklinghausen disease, Watson disease)

### Cards

[Atlas](#) [NF1ID134](#)

[GeneCards](#) [NF1](#)

[Ensembl](#) [NF1](#)

[Genatlas](#) [NF1](#)

[GeneLynx](#) [NF1](#)

[eGenome](#) [NF1](#)

[euGene](#) [4763](#)

### Genomic and cartography

[GoldenPath](#) [NF1 - 17q11.2 chr17:26446243-26725590 + 17q11.2](#) (hg18-Mar\_2006)

[Ensembl](#) [NF1 - 17q11.2 \[CytoView\]](#)

[NCBI](#) [Genes Cyto](#) [Gene Seq](#) [Map View - NCBI]

[OMIM](#) [Disease map \[OMIM\]](#)

[HomoloGene](#) [NF1](#)

### Gene and transcription

[Genbank](#) [AB209336](#) [ENTREZ]

[Genbank](#) [AF055023](#) [ENTREZ]

[Genbank](#) [AF086346](#) [ENTREZ]

[Genbank](#) [AK026658](#) [ENTREZ]

[Genbank](#) [BP271334](#) [ENTREZ]

[RefSeq](#) [NM\\_000267](#) [SRS] [NM\\_000267](#) [ENTREZ]

[RefSeq](#) [NM\\_001042492](#) [SRS] [NM\\_001042492](#) [ENTREZ]

[AceView](#) [NF1](#) AceView - NCBI

[TRASER](#) [NF1](#) Traser - Stanford

<a href="#">Unigene</a>	<a href="#">Hs.567266</a> [SRS] <a href="#">Hs.567266</a> [NCBI] <a href="#">HS567266</a> [spliceNest]
<b>Protein : pattern, domain, 3D structure</b>	
<a href="#">SwissProt</a>	<a href="#">P21359</a> [SRS] <a href="#">P21359</a> [EXPASY] <a href="#">P21359</a> [INTERPRO]
<a href="#">Prosite</a>	<a href="#">PS50191 CRAL_TRIO</a> [SRS] <a href="#">PS50191 CRAL_TRIO</a> [Expasy]
<a href="#">Prosite</a>	<a href="#">PS00509 RAS_GTPASE_ACTIV_1</a> [SRS] <a href="#">PS00509 RAS_GTPASE_ACTIV_1</a> [Expasy]
<a href="#">Prosite</a>	<a href="#">PS50018 RAS_GTPASE_ACTIV_2</a> [SRS] <a href="#">PS50018 RAS_GTPASE_ACTIV_2</a> [Expasy]
<a href="#">Interpro</a>	<a href="#">IPR001251 CRAL_bd_TRIO_C</a> [SRS] <a href="#">IPR001251 CRAL_bd_TRIO_C</a> [EBI]
<a href="#">Interpro</a>	<a href="#">IPR001936 RasGAP</a> [SRS] <a href="#">IPR001936 RasGAP</a> [EBI]
<a href="#">Interpro</a>	<a href="#">IPR008936 Rho_GAP</a> [SRS] <a href="#">IPR008936 Rho_GAP</a> [EBI]
<a href="#">CluSTr</a>	<a href="#">P21359</a>
<a href="#">Pfam</a>	<a href="#">PF00616 RasGAP</a> [SRS] <a href="#">PF00616 RasGAP</a> [Sanger] <a href="#">pfam00616</a> [NCBI-CDD]
<a href="#">Smart</a>	<a href="#">SM00323 RasGAP</a> [EMBL]
<a href="#">Smart</a>	<a href="#">SM00516 SEC14</a> [EMBL]
<a href="#">Blocks</a>	<a href="#">P21359</a>
<a href="#">PDB</a>	<a href="#">1NF1</a> [SRS] <a href="#">1NF1</a> [PdbSum], <a href="#">1NF1</a> [IMB] <a href="#">1NF1</a> [RSDB]
<a href="#">HPRD</a>	<a href="#">P21359</a>
<b>Protein Interaction databases</b>	
<a href="#">DIP</a>	<a href="#">P21359</a>
<a href="#">IntAct</a>	<a href="#">P21359</a>
<b>Polymorphism : SNP, mutations, diseases</b>	
<a href="#">OMIM</a>	<a href="#">162200;162210;193520;601321;607785</a> [map]
<a href="#">GENECLINICS</a>	<a href="#">162200;162210;193520;601321;607785</a>
<a href="#">SNP</a>	<a href="#">NF1</a> [dbSNP-NCBI]
<a href="#">SNP</a>	<a href="#">NM_000267</a> [SNP-NCI]
<a href="#">SNP</a>	<a href="#">NM_001042492</a> [SNP-NCI]
<a href="#">SNP</a>	<a href="#">NF1</a> [GeneSNPs - Utah] <a href="#">NF1</a> [HGBASE - SRS]
<a href="#">HAPMAP</a>	<a href="#">NF1</a> [HAPMAP]
<b>General knowledge</b>	
<a href="#">Family Browser</a>	<a href="#">NF1</a> [UCSC Family Browser]
<a href="#">SOURCE</a>	<a href="#">NM_000267</a>
<a href="#">SOURCE</a>	<a href="#">NM_001042492</a>
<a href="#">SMD</a>	<a href="#">Hs.567266</a>
<a href="#">SAGE</a>	<a href="#">Hs.567266</a>
<a href="#">Amigo</a>	<a href="#">biological process unknown</a>

[Amigo](#) [enzyme inhibitor activity](#)  
[Amigo](#) [Ras GTPase activator activity](#)  
[Amigo](#) [molecular function unknown](#)  
[Amigo](#) [intracellular](#)  
[Amigo](#) [cytoplasm](#)  
[Amigo](#) [cell cycle](#)  
[Amigo](#) [Ras protein signal transduction](#)  
[Amigo](#) [negative regulation of cell proliferation](#)  
[Amigo](#) [cellular component unknown](#)  
[Amigo](#) [negative regulation of progression through cell cycle](#)  
[Amigo](#) [regulation of small GTPase mediated signal transduction](#)

[BIOCARTA](#) [Chromatin Remodeling by hSWI/SNF ATP-dependent Complexes](#) [[Genes](#)]

[PubGene](#) [NF1](#)

#### Other databases

#### Probes

[Probe](#) [NF1 Related clones \(RZPD - Berlin\)](#)

#### PubMed

[PubMed](#) [118 Pubmed reference\(s\) in LocusLink](#)

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Medline [16380919](#)

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URL : <http://AtlasGeneticsOncology.org/Genes/NF1ID134.html>

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