

Smad2, mothers against decapentaplegic homolog 2 (Drosophila)

Identity

| | |
|-------------|--|
| Other names | MADH2 MADR2 JV18-1 JV18 |
| Hugo | <u>SMAD2</u> |
| Location | 18q21.1 |

DNA/RNA

Description The gene encompasses 90 kb of DNA; 12 exons.
Transcription 2285 nucleotides mRNA. Alternative splicing was described.

Protein

Description 467 amino acids; 52 kDa protein. A short 438 amino acids isoform was also described. Smad2 belongs to the Darwin proteins family which are composed of two conserved amino- and carboxyl-terminal domains known as MH1 and MH2, respectively.

Expression High expression levels in skeletal muscle, heart and placenta.

Function Smad2 is an intracellular mediator of TGF-beta family and activin type 1 receptor. Smad2 mediate TGF-beta signaling to regulate cell growth and differentiation. Smad2 is released from cytoplasmic retention by TGF-beta receptor-mediated phosphorylation. The phosphorylated Smad2 then forms a heterodimeric complex with [Smad4](#), and this complex translocates from cytoplasm into nucleus. By interacting with DNA-binding proteins, Smad complexes then positively or negatively regulate the transcription of target genes.

Homology With the other members of the Darwin/Smad family.

Implicated in

Disease [Colorectal cancers](#)

Oncogenesis Smad2 was proposed to be a tumor suppressor gene that may function to disrupt TGF-beta signaling. Inactivating mutations in Smad2 have been found in various cancer including colorectal carcinomas. The majority of tumor-derived mutations cluster in the carboxy-terminal MH2 domain, and some of these have been shown to disrupt TGF-beta signaling by blocking receptor-dependent phosphorylation or by preventing heterodimeric interactions between Smads. A mutation at position 133 in the amino-terminal MH1 domain has also been associated with colorectal carcinoma. Nevertheless, loss of Smad2 activation and/or expression was related to occur in less than 10% of colorectal cancers.

To be noted

Smad2 gene has also been found altered in [lung carcinomas](#), [cervical](#)

carcinomas and hepatocellular carcinomas.

External links

[Hugo](#)
[GDB](#)
[Entrez Gene](#)

Nomenclature

[SMAD2](#)
[SMAD2](#)
[SMAD2 4087](#) SMAD, mothers against DPP homolog 2 (Drosophila)

[Atlas](#)
[GeneCards](#)
[Ensembl](#)
[CancerGene](#)
[Genatlas](#)
[GeneLynx](#)
[eGenome](#)
[euGene](#)

Cards

[SMAD2ID370](#)

[SMAD2](#)
[SMAD2](#)
[MADH2](#)
[SMAD2](#)
[SMAD2](#)
[SMAD2](#)
[SMAD2](#)
[4087](#)

[GoldenPath](#)
[Ensembl](#)
[NCBI](#)
[OMIM](#)
[HomoloGene](#)

Genomic and cartography

[SMAD2 - 18q21.1](#) [chr18:43618435-43711221 - 18q21.1](#) (hg17-May_2004)
[SMAD2 - 18q21.1 \[CytoView\]](#)
[Genes Cyto](#) [Gene Seq](#) [Map View - NCBI]
[Disease map \[OMIM\]](#)
[SMAD2](#)

[Genbank](#)
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[RefSeq](#)
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[TRASER](#)
[Unigene](#)

Gene and transcription

[U78733](#) [SRS] [U78733](#) [ENTREZ]
[AA081871](#) [SRS] [AA081871](#) [ENTREZ]
[AA418737](#) [SRS] [AA418737](#) [ENTREZ]
[AF027964](#) [SRS] [AF027964](#) [ENTREZ]
[AI087928](#) [SRS] [AI087928](#) [ENTREZ]
[NM_001003652](#) [SRS] [NM_001003652](#) [ENTREZ]
[NM_005901](#) [SRS] [NM_005901](#) [ENTREZ]
[NT_086889](#) [SRS] [NT_086889](#) [ENTREZ]
[SMAD2](#) AceView - NCBI
[SMAD2](#) Traser - Stanford
[Hs.465061](#) [SRS] [Hs.465061](#) [NCBI] [HS465061](#) [spliceNest]

[SwissProt](#)
[Interpro](#)
[Interpro](#)
[Interpro](#)
[CluSTr](#)
[Pfam](#)
[Pfam](#)
[Smart](#)
[Smart](#)
[Blocks](#)
[PDB](#)
[PDB](#)

Protein : pattern, domain, 3D structure

[Q15796](#) [SRS] [Q15796](#) [EXPASY] [Q15796](#) [INTERPRO]
[IPR001132 Dwarfin](#) [SRS] [IPR001132 Dwarfin](#) [EBI]
[IPR003619 Dwarfin_A](#) [SRS] [IPR003619 Dwarfin_A](#) [EBI]
[IPR008984 SMAD_FHA](#) [SRS] [IPR008984 SMAD_FHA](#) [EBI]
[Q15796](#)
[PF03165 MH1](#) [SRS] [PF03165 MH1](#) [Sanger] [pfam03165](#) [NCBI-CDD]
[PF03166 MH2](#) [SRS] [PF03166 MH2](#) [Sanger] [pfam03166](#) [NCBI-CDD]
[SM00523 DWA](#) [EMBL]
[SM00524 DWB](#) [EMBL]
[Q15796](#)
[1DEV](#) [SRS] [1DEV](#) [PdbSum], [1DEV](#) [IMB]
[1KHX](#) [SRS] [1KHX](#) [PdbSum], [1KHX](#) [IMB]

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Polymorphism : SNP, mutations, diseases

[601366](#) [map]
[601366](#)

[SNP](#) [SMAD2](#) [dbSNP-NCBI]
[SNP](#) [NM_001003652](#) [SNP-NCI]
[SNP](#) [NM_005901](#) [SNP-NCI]
[SNP](#) [SMAD2](#) [GeneSNPs - Utah] [SMAD2](#) [SNP - CSHL] [SMAD2](#) [HGBase - SRS]

General knowledge

[Family Browser](#) [SMAD2](#) [UCSC Family Browser]
[SOURCE](#) [NM_001003652](#)
[SOURCE](#) [NM_005901](#)
[SMD](#) [Hs.465061](#)
[SAGE](#) [Hs.465061](#)
[Amigo](#) [component|nucleus](#)
[Amigo](#) [function|protein binding](#)
[Amigo](#) [process|regulation of transcription, DNA-dependent](#)
[Amigo](#) [process|signal transduction](#)
[BIOCARTA](#) [TGF beta signaling pathway](#)
[PubGene](#) [SMAD2](#)

Other databases

Probes

[Probe](#) [SMAD2 Related clones \(RZPD - Berlin\)](#)

PubMed

[PubMed](#) [47 Pubmed reference\(s\) in LocusLink](#)

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Medline [8752209](#)

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Medline [10781087](#)

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Medline [12967141](#)

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URL : <http://www.infobiogen.fr/services/chromcancer/Genes/SMAD2ID370.html>

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