

Gene Section

Mini Review

CTNNB1 (Catenin, beta-1)

Brigitte Debuire, Antoinette Lemoine, Raphaël Saffroy

Service de Biochimie et Biologie moléculaire, Hôpital Universitaire Paul Brousse, UPRES 1596-Faculté de Médecine Paris-Sud, 14 avenue Paul Vaillant Couturier, 94804 Villejuif Cedex, France (BD, AL, RS)

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Identity

Other names: Cadherin-associated protein, beta

HGNC (Hugo): CTNNB1

Location: 3p22-p21.3



CTNNB1 (3p22) - Courtesy Mariano Rocchi, Resources for Molecular Cytogenetics.

DNA/RNA

Description

The gene encompasses 23.2 kb of DNA; 16 exons (the first is non-coding).

Transcription

3362 nucleotides mRNA; 2343 bp open reading frame. Alternative splicing within exon 16 produces a splice variant that is 159 bp shorter in the 3' untranslated region.

Protein

Description

781 amino acids; 92 kDa protein. Can be

phosphorylated ; contains from N-term to C-term, a phosphorylation site by the serine-threonine glycogen synthase kinase -3b (GSK-3b), an α -catenin binding site, 13 armadillo repeats and a transactivating domain.

Expression

Widely expressed.

Localisation

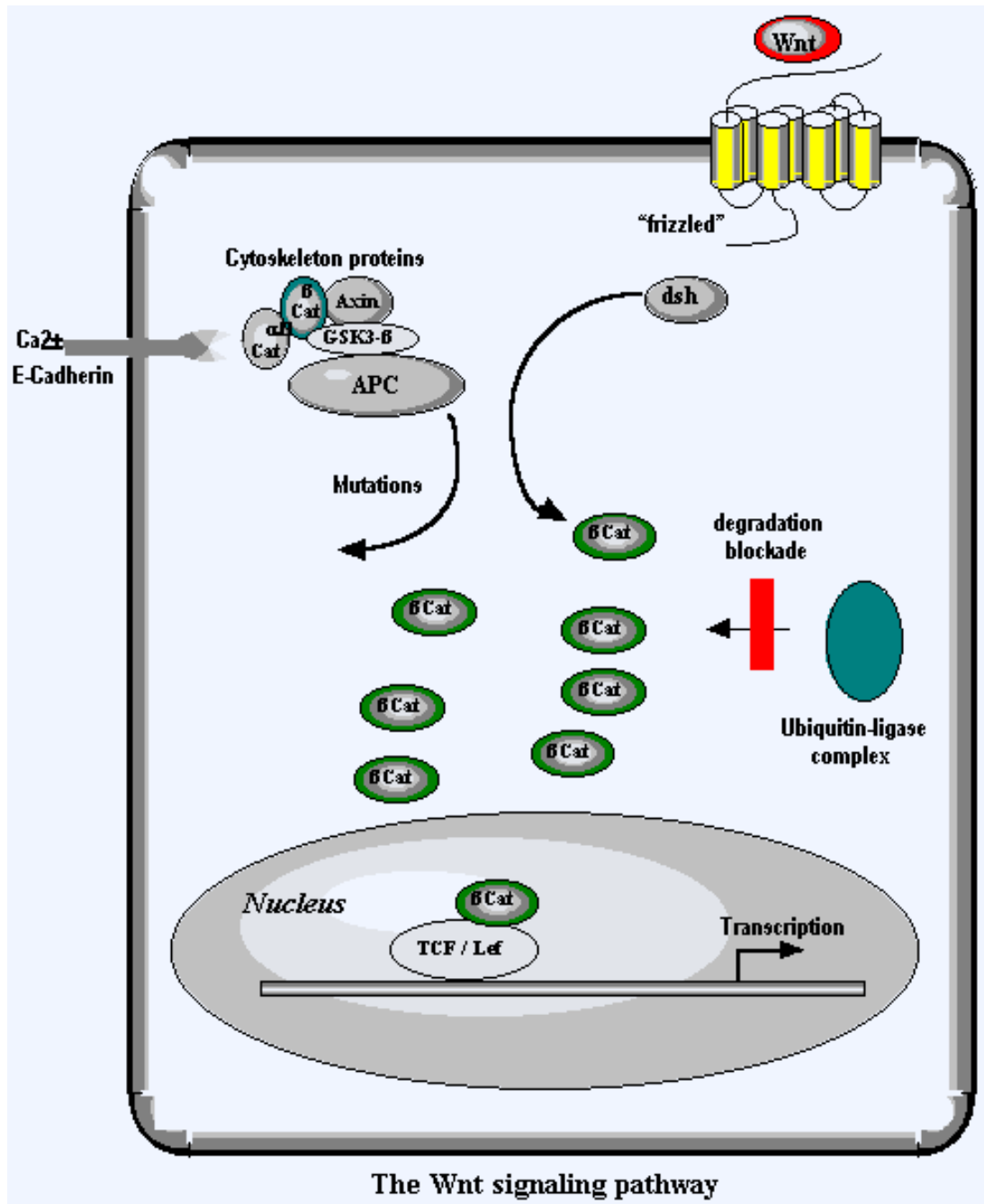
Cytoplasm and nucleus.

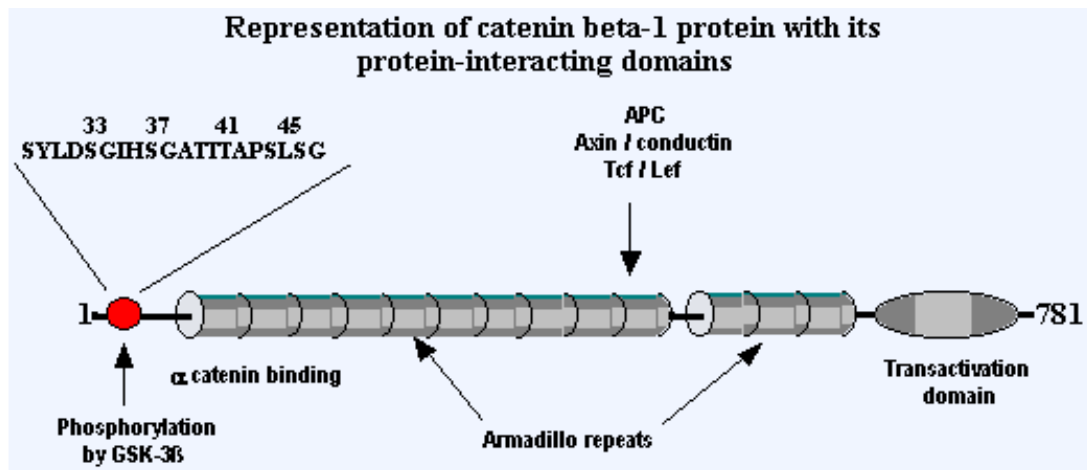
Function

Important functions in the E-cadherin-mediated cell-cell adhesion system and also as a downstream signaling molecule in the Wnt pathway. Cytoplasmic accumulation of b-catenin allows it to translocate to the nucleus to form complexes with transcription factors of the T cell factor-lymphoid enhancer factor (Tcf-Lef) family. b-catenin is assumed to transactivate mostly unknown target genes, which may stimulate cell proliferation (acts as an oncogene) or inhibit apoptosis. The b-catenin level in the cell is regulated by its association with the adenomatous polyposis coli (APC) tumor suppressor protein, axin and GSK-3b. Phosphorylation of b-catenin by the APC-axin-GSK-3b complex leads to its degradation by the ubiquitin-proteasome system.

Homology

The b-catenin protein shares 70% amino acid identity with both plakoglobin (intracellular junction in desmosomes) and the product of the Drosophila segment polarity gene "armadillo".





Mutations

Somatic

Two mechanisms underlying the increase in β -catenin levels by stabilizing β -catenin are known. One is inactivating mutation in the APC gene, the other is activating mutation at the GSK-3 β phosphorylation sites within exon 3 of the β -catenin gene.

β -catenin plays a key role in the development of colorectal cancer and has been found mutated in colorectal cancer cell lines. β -catenin aberration is a frequent event in the development of hepatocellular carcinoma and may facilitate its development in the course of chronic hepatitis. β -catenin has also been found mutated in hepatoblastoma, ovarian carcinoma, medulloblastoma, pilomatricoma as well as in melanoma cell lines.

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