Atlas of Genetics and Cytogenetics in Oncology and Haematology

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Gene Section
Mini Review

ACTB (Actin, beta)

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Identity

Other names: Beta cytoskeletal actin; Beta actin

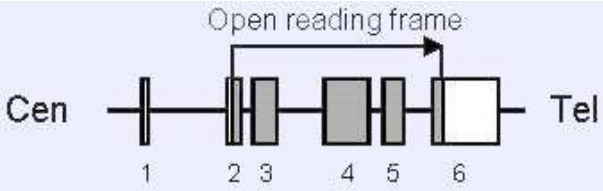
HGNC (Hugo): ACTB

Location: 7p22

Location (base pair): Position 5340026-5343462 on the chromosome 7 genomic sequence.

Note
Six actin isoforms are known: two sarcomeric (a-skeletal and a-cardiac), two smooth muscle actins (a and g), and two non-muscles, cytoskeletal actins (b and g).

DNA/RNA



Genomic organization of the ACTB gene.

Description

Six exons, spans approximately 3.4 kb of genomic DNA in the centromere-to-telomere orientation. The translation initiation codon ATG is located in exon 2, and the stop codon in exon 6.

Transcription

mRNA of approximately 1.8 kb.

Pseudogene

At least 19 processed, non-expressed, pseudogenes are dispersed throughout the genome.

Protein

Description

The open reading frame encodes a 374 amino acid protein, with an estimated molecular weight of approximately 41.7 kDa.

Expression

Abundantly expressed in all mammalian and avian non-muscle cells.

Localisation

Cytoplasm.

Function

Component (together with cytoplasmic g actin) of the cytoskeletal microfilaments. Involved in the transport of chromosomes and organelles as well as in cell motility.

Homology

The ACTB proteins are evolutionary conserved. Mammalian cytoplasmic actins (actin g and b) are remarkably similar to each other, but differ in at least 25 residues from the muscle actins.

Mutations

Somatic

ACTB is interrupted by the t(7;12)(p22;q13) detected in pericytoma with t(7;12).

Implicated in

Disease
Pericytoma with t(7;12).

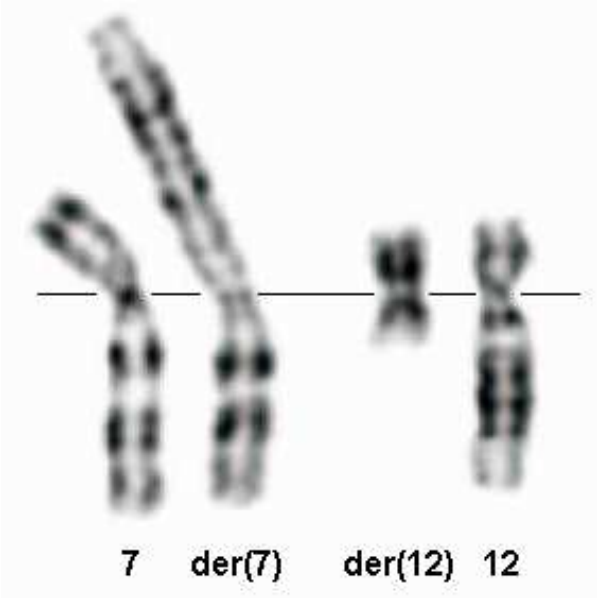
Prognosis
Benign or low-malignant.

Cytogenetics

t(7;12)(p22;q13).

Hybrid/Mutated gene

ACTB-GLI1 fusion gene. The breakpoints reported so far have been located to introns 1, 2 or 3 within the ACTB gene, and to introns 5 or 6 or to exon 7 within the GLI1 gene. Reciprocal GLI1-ACTB gene fusions have also been detected. The breakpoints have been located to introns 5 or 7 within the GLI1 gene, and to intron 3 of the ACTB gene.



Representative G-banded partial karyotype of the t(7;12)(p22;q13).

Abnormal protein

The ACTB-GLI1 fusion protein contains the N-terminal of ACTB and the C-terminal of GLI1, including the DNA-binding zink finger motifs (encoded by exons 7-10) and transactivating motifs (exon 12).

Oncogenesis

It is suggested that the strong ACTB promoter causes an overexpression of GLI1 sequences important for transcriptional activation of downstream target genes, akin to the oncogenic mechanisms of the COL1A1-PDGFB fusion gene detected in dermatofibrosarcoma protuberans.

References

Vandekerckhove J, Weber K. Mammalian cytoplasmic actins are the products of at least two genes and differ in primary structure in at least 25 identified positions from skeletal muscle actins. Proc Natl Acad Sci U S A. 1978 Mar;75(3):1106-10

Kedes L, Ng SY, Lin CS, Gunning P, Eddy R, Shows T, Leavitt J. The human beta-actin multigene family. Trans Assoc Am Physicians. 1985;98:42-6

Nakajima-Iijima S, Hamada H, Reddy P, Kakunaga T. Molecular structure of the human cytoplasmic beta-actin gene: interspecies homology of sequences in the introns. Proc Natl Acad Sci U S A. 1985 Sep;82(18):6133-7

Ng SY, Gunning P, Eddy R, Ponte P, Leavitt J, Shows T, Kedes L. Evolution of the functional human beta-actin gene and its multi-pseudogene family: conservation of noncoding regions and chromosomal dispersion of pseudogenes. Mol Cell Biol. 1985 Oct;5(10):2720-32

Ueyama H, Inazawa J, Nishino H, Ohkubo I, Miwa T. FISH localization of human cytoplasmic actin genes ACTB to 7p22 and ACTG1 to 17q25 and characterization of related pseudogenes. Cytogenet Cell Genet. 1996;74(3):221-4

Simon MP, Pedeutour F, Sirvent N, Grosgeorge J, Minoletti F, Coindre JM, Terrier-Lacombe MJ, Mandahl N, Craver RD, Blin N, Sozzi G, Turc-Carel C, O'Brien KP, Kedra D, Fransson I, Guilbaud C, Dumanski JP. Deregulation of the platelet-derived growth factor B-chain gene via fusion with collagen gene COL1A1 in dermatofibrosarcoma protuberans and giant-cell fibroblastoma. Nat Genet. 1997 Jan;15(1):95-8

Dahlén A, Fletcher CD, Mertens F, Fletcher JA, Perez-Atayde AR, Hicks MJ, Debiec-Rychter M, Sciot R, Wejde J, Wedin R, Mandahl N, Panagopoulos I. Activation of the GLI oncogene through fusion with the beta-actin gene (ACTB) in a group of distinctive pericytic neoplasms: pericytoma with t(7;12). Am J Pathol. 2004 May;164(5):1645-53

Dahlén A, Mertens F, Mandahl N, Panagopoulos I. Molecular genetic characterization of the genomic ACTB-GLI fusion in pericytoma with t(7;12). Biochem Biophys Res Commun. 2004 Dec 24;325(4):1318-23

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