

ACTB (Actin, beta)

Identity

Note Six actin isoforms are known: two sarcomeric (α -skeletal and α -cardiac), two smooth muscle actins (α and γ), and two non-muscle, cytoskeletal actins (β and γ).

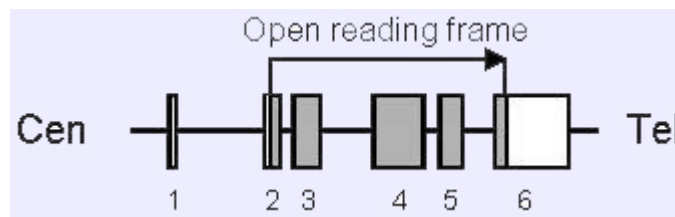
Other names **Beta cytoskeletal actin**

Beta actin

Hugo [ACTB](#)

Location 7p22.

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 γ 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 α and β) are remarkably similar to each other.

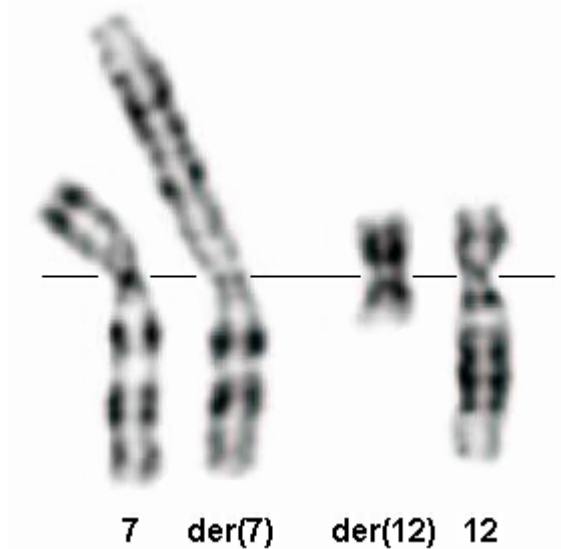
but differ in at least 25 residues from the muscle actins.

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)



Representative G-banded partial karyotype of the 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.

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.

External links

Nomenclature

[Hugo](#) [ACTB](#)
[GDB](#) [ACTB](#)
[Entrez_Gene](#) [ACTB_60](#) actin, beta

Cards

[Atlas](#) [ACTBID42959ch7p22](#)
[GeneCards](#) [ACTB](#)
[Ensembl](#) [ACTB](#)
[Genatlas](#) [ACTB](#)
[GeneLynx](#) [ACTB](#)
[eGenome](#) [ACTB](#)
[euGene](#) [60](#)

Genomic and cartography

[GoldenPath](#) [ACTB - 7p22.1](#) [chr7:5340027-5343462 - 7p22.1](#) (hg17-May_2004)
[Ensembl](#) [ACTB - 7p22.1 \[CytoView\]](#)
[NCBI](#) [Genes Cyto](#) [Gene Seq](#) [Map View - NCBI]
[OMIM](#) [Disease map \[OMIM\]](#)
[HomoloGene](#) [ACTB](#)

Gene and transcription

[Genbank](#) [AC006483](#) [SRS] [AC006483](#) [ENTREZ]
[Genbank](#) [AY582799](#) [SRS] [AY582799](#) [ENTREZ]
[Genbank](#) [M10277](#) [SRS] [M10277](#) [ENTREZ]
[Genbank](#) [AK025375](#) [SRS] [AK025375](#) [ENTREZ]
[Genbank](#) [AK058019](#) [SRS] [AK058019](#) [ENTREZ]
[RefSeq](#) [NM_001101](#) [SRS] [NM_001101](#) [ENTREZ]
[RefSeq](#) [NT_086701](#) [SRS] [NT_086701](#) [ENTREZ]
[AceView](#) [ACTB](#) AceView - NCBI
[TRASER](#) [ACTB](#) Traser - Stanford
[Unigene](#) [Hs.520640](#) [SRS] [Hs.520640](#) [NCBI] [HS520640](#) [spliceNest]

Protein : pattern, domain, 3D structure

Polymorphism : SNP, mutations, diseases

[OMIM](#) [102630](#) [[map](#)]
[GENECLINICS](#) [102630](#)
[SNP](#) [ACTB](#) [dbSNP-NCBI]
[SNP](#) [NM_001101](#) [SNP-NCI]
[SNP](#) [ACTB](#) [GeneSNPs - Utah] [ACTB](#) [SNP - CSHL] [ACTB](#) [HGBASE - SRS]

General knowledge

[Familv](#) [ACTB](#) [UCSC Family Browser]

Browser	
SOURCE	NM_001101
SMD	Hs.520640
SAGE	Hs.520640
Amigo	component TIP60 histone acetyltransferase complex
Amigo	component actin filament
Amigo	component cytoskeleton
Amigo	function motor activity
Amigo	function protein binding
Amigo	function structural constituent of cytoskeleton
BIOCARTA	Chromatin Remodeling by hSWI/SNF ATP-dependent Complexes
PubGene	ACTB

Other databases

Probes

[Probe](#) [probes RP11-1275H24 \(AC092171; substantially larger than the 85 kb reported by the NCBI\) and RP11-93G19 \(BAC ends: AQ321651, AQ321650\)](#)

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

PubMed

[PubMed](#) [79 Pubmed reference\(s\) in LocusLink](#)

Bibliography

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Proc Natl Acad Sci USA 1978; 75: 1106-1110.

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Am J Pathol 2004; 164: 1645-1653.
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Molecular genetic characterization of the ACTB-GLI fusion in pericytoma with t(7;12).

Dahlén A, Mertens F, Mandahl N, Panagopoulos I.
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Medline [15555571](#)

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URL :

<http://www.infobiogen.fr/services/chromcancer/Genes/ACTBID42959ch7p22.html>

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