ACTB gene

actin beta

Normal Function

The *ACTB* gene provides instructions for making a protein called beta (β)-actin, which is part of the actin protein family. Proteins in this family are organized into a network of fibers called the actin cytoskeleton, which makes up the structural framework inside cells. There are six types of actin; four are present only in muscle cells, where they are involved in the tensing of muscle fibers (muscle contraction). The other two actin proteins, β-actin and gamma (γ)-actin (produced from the *ACTG1* gene), are found in cells throughout the body. These proteins play important roles in determining cell shape and controlling cell movement (motility). Studies suggest that β-actin may also be involved in relaying chemical signals within cells.

Health Conditions Related to Genetic Changes

Baraitser-Winter syndrome

Several mutations in the *ACTB* gene have been found to cause Baraitser-Winter syndrome, a rare condition that affects the development of the brain, eyes, and other facial features. The known mutations change single protein building blocks (amino acids) in β-actin. The most common mutation replaces the amino acid arginine with the amino acid histidine at protein position 196 (written as Arg196His or R196H). The mutations that cause Baraitser-Winter syndrome alter the function of β-actin, which causes changes in the actin cytoskeleton that modify the structure and organization of cells and affect their ability to move. Because β-actin is present in cells throughout the body and is involved in many cell activities, problems with its function likely impact many aspects of development. These changes underlie the variety of signs and symptoms associated with Baraitser-Winter syndrome.

Coloboma

MedlinePlus Genetics provides information about Coloboma

Other Names for This Gene

- ACTB_HUMAN
- actin, beta

- actin, cytoplasmic 1
- beta cytoskeletal actin
- BRWS1
- PS1TP5-binding protein 1
- PS1TP5BP1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ACTB (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=60[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28ACTB%5BTI%5D%29+OR+%28beta+actin%5BTI%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- ACTIN, BETA; ACTB (https://omim.org/entry/102630)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/60)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ACTB[gene])

References

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- Su Y, Kondrikov D, Block ER. Beta-actin: a regulator of NOS-3. Sci STKE. 2007Sep 18;2007(404):pe52. doi: 10.1126/stke.4042007pe52. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17878410)

Genomic Location

The *ACTB* gene is found on chromosome 7 (https://medlineplus.gov/genetics/chromosome/7/).

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