ACTG1 gene

actin gamma 1

Normal Function

The *ACTG1* gene provides instructions for making a protein called gamma (γ)-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, γ-actin and beta (β)-actin (produced from the *ACTB* gene), are found in cells throughout the body. These proteins play important roles in determining cell shape and controlling cell movement (motility).

γ-actin is particularly abundant in certain cells in the intestines and the inner ear. Within the inner ear, this protein is found in specialized cells called hair cells, which are essential for normal hearing.

Health Conditions Related to Genetic Changes

Baraitser-Winter syndrome

At least six mutations in the *ACTG1* 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 γ-actin. The most common mutation replaces the amino acid serine with the amino acid phenylalanine at protein position 155 (written as Ser155Phe or S155F). The mutations that cause Baraitser-Winter syndrome alter the function of γ-actin, which causes changes in the actin cytoskeleton that modify the structure and organization of cells and affect their ability to move. Because γ-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.

Nonsyndromic hearing loss

MedlinePlus Genetics provides information about Nonsyndromic hearing loss

Coloboma

MedlinePlus Genetics provides information about Coloboma

Other Names for This Gene

- ACT
- ACTG
- ACTG_HUMAN
- actin, cytoplasmic 2
- actin, gamma 1
- actin-like protein
- cytoskeletal gamma-actin
- deafness, autosomal dominant 20
- deafness, autosomal dominant 26
- DFNA20
- DFNA26
- gamma-actin

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ACTG1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=71[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28ACTG1%5BTIAB%5D%29+OR+%28DFNA20%5BTIAB%5D%29+OR+%28DFNA26%5BTIAB%5D%29%29+OR+%28%28gamma+1%5BTI%5D%29+AND+%28actin%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- ACTIN, GAMMA-1; ACTG1 (https://omim.org/entry/102560)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/71)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ACTG1[gene])

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Genomic Location

The *ACTG1* gene is found on chromosome 17 (https://medlineplus.gov/genetics/chromosome/17/).

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