STRC gene

stereocilin

Normal Function

The *STRC* gene provides instructions for making a protein called stereocilin. This protein is found in the inner ear and appears to be involved in hearing.

Stereocilin is associated with hairlike structures called stereocilia, which project from specialized cells called hair cells in the inner ear. Specifically, stereocilin helps to maintain the structure of stereocilia by linking their tips to one another. Stereocilia bend in response to sound waves, triggering a series of reactions within hair cells that generate a nerve impulse. Such nerve impulses are transmitted via the auditory nerve to the brain, where they are interpreted as sound.

Health Conditions Related to Genetic Changes

Nonsyndromic hearing loss

Researchers have identified a few *STRC* gene mutations in individuals with nonsyndromic hearing loss, which is loss of hearing that is not associated with other signs and symptoms. Mutations in this gene cause a form of nonsyndromic hearing loss called DFNB16. This form of hearing loss can either be present before a child learns to speak (prelingual) or begin after a child learns to speak (postlingual). The hearing loss ranges from mild to profound and particularly affects the ability to hear high-frequency sounds.

The *STRC* gene mutations that cause nonsyndromic hearing loss add a small amount of DNA to the *STRC* gene or delete DNA from the gene. In many cases, the mutation deletes a piece of chromosome 15 that includes the entire *STRC* gene. Mutations in this gene lead to the production of a nonfunctional version of stereocilin or prevent any of this protein from being produced. A loss of functional stereocilin likely alters the structure of stereocilia, preventing them from reacting normally to sound waves. As a result, hair cells cannot convert sound into electrical impulses, which leads to hearing loss in people with DFNB16.

Sensorineural deafness and male infertility

Sensorineural deafness and male infertility is a condition caused by a deletion of genetic material on the long (q) arm of chromosome 15. This condition is characterized by the combination of hearing loss and an inability to father children.

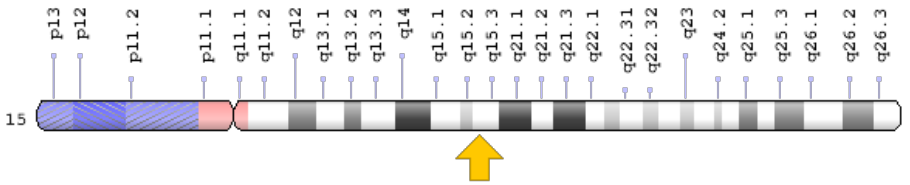
The chromosomal region that is typically deleted contains multiple genes, including the *STRC* gene. People with this condition have the deletion in both copies of chromosome 15 in each cell. As a result of the deletion, affected individuals are

missing both copies of the *STRC* gene, and no stereocilin protein is produced. A lack of stereocilin likely interferes with the normal function of stereocilia and impairs how these structures respond to sound waves, resulting in hearing loss. The loss of another gene, *CATSPER2*, in the same region of chromosome 15 is responsible for infertility in affected males.

Chromosomal Location

Cytogenetic Location: 15q15.3, which is the long (q) arm of chromosome 15 at position 15.3

Molecular Location: base pairs 43,599,563 to 43,618,800 on chromosome 15 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- deafness, autosomal recessive 16
- DFNB16
- STRC_HUMAN

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): Hair Cells Use a Connected Bundle of Stereocilia to Detect Tiny Motions
  https://www.ncbi.nlm.nih.gov/books/NBK22542/#A4621
- Molecular Biology of the Cell (fourth edition, 2002): Auditory Hair Cells Have to Last a Lifetime
  https://www.ncbi.nlm.nih.gov/books/NBK26868/#A4104
- Neuroscience (second edition, 2001): Hair Cells and the Mechanoelectrical Transduction of Sound Waves
  https://www.ncbi.nlm.nih.gov/books/NBK10867/

Clinical Information from GeneReviews

- CATSPER-Related Male Infertility
  https://www.ncbi.nlm.nih.gov/books/NBK22925
- Hereditary Hearing Loss and Deafness Overview
  https://www.ncbi.nlm.nih.gov/books/NBK1434

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28STRC%5BTIAB%5D%29+OR+%28stereocilin%5BTIAB%5D%29%29+OR+%28DFNB16%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- STEREOCILIN
  http://omim.org/entry/606440

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=STRC%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:16035
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:161497
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/161497
- The Hereditary Hearing Loss Homepage
  https://hereditaryhearingloss.org/
- UniProt
  https://www.uniprot.org/uniprot/Q7RTU9

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