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 is involved in hearing.

Stereocilin is associated with hairlike structures called stereocilia, which project from specialized cells called hair cells in the inner ear. Stereocilin links the tips of neighboring stereocilia to one another. Stereocilia must be physically connected to carry out certain auditory functions, such as making quiet sounds louder and detecting sound frequency.

Health Conditions Related to Genetic Changes

Nonsyndromic hearing loss

Researchers have identified *STRC* gene variants (also called mutations) in individuals with nonsyndromic hearing loss, which is loss of hearing that is not associated with other signs and symptoms. Variants 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 variants 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 variant deletes a piece of chromosome 15 that includes the entire *STRC* gene. Variants 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 connecting to one another. As a result, certain auditory functions are impaired, which leads to hearing loss in people with DFNB16.

Deafness-infertility syndrome

Deafness-infertility syndrome 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 difficulty conceiving children (a condition called infertility).

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, no stereocilin protein is produced. A lack of stereocilin likely interferes with the normal function of stereocilia and impairs certain auditory functions, resulting in hearing loss in people with deafness-infertility syndrome.

The loss of another gene, *CATSPER2*, in the same region of chromosome 15 results in the impaired movement of sperm. This causes the infertility seen in people with deafness-infertility syndrome. Researchers are working to determine how the loss of additional genes in the deleted region affects people with deafness-infertility syndrome.

Other Names for This Gene

- deafness, autosomal recessive 16
- DFNB16
- STRC_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of STRC (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=161497[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?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; STRC (https://omim.org/entry/606440)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/161497)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=STRC[gene])

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Genomic Location

The *STRC* gene is found on chromosome 15 (https://medlineplus.gov/genetics/chromosome/15/).

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