Deafness-infertility syndrome

Description

Deafness-infertility syndrome is a condition characterized by hearing loss and difficulty conceiving children (a condition called infertility). Affected individuals have moderate to severe sensorineural hearing loss, which is caused by abnormalities in the inner ear. The hearing loss is typically diagnosed in early childhood and does not worsen over time. Individuals with this condition produce sperm that have decreased movement (motility). As a result, they cannot conceive without assisted reproductive technologies.

Frequency

The prevalence of deafness-infertility syndrome is unknown. It is likely under diagnosed in people with hearing loss.

Causes

Deafness-infertility syndrome is caused by a deletion of genetic material on the long (q) arm of chromosome 15. The signs and symptoms of deafness-infertility syndrome are related to the loss of multiple genes in this region. The size of the deletion varies among affected individuals. Researchers have determined that the loss of two specific genes on chromosome 15 is responsible for the main features of this condition. The loss of the *STRC* gene, which plays a role in the generation of nerve impulses that get interpreted as sound, is responsible for hearing loss. The loss of another gene, *CATSPER2*, which plays a role in sperm motility, is responsible for the sperm abnormalities. Researchers are working to determine how the loss of additional genes in the deleted region affects people with deafness-infertility syndrome.

Learn more about the genes and chromosome associated with Deafness-infertility syndrome

- CATSPER2
- STRC
- chromosome 15

Inheritance

Deafness-infertility syndrome is inherited in an autosomal recessive pattern, which means both copies of chromosome 15 in each cell have a deletion. The parents of an individual with deafness-infertility syndrome each carry one copy of the chromosome 15 deletion, but they typically do not show symptoms of the condition.

People with one Y chromosome (typical for males) who have two chromosome 15 deletions in each cell have deafness-infertility syndrome. People with two X chromosomes (typical for females) who have two chromosome 15 deletions in each cell have sensorineural deafness as their only symptom. They do not produce sperm and so are not affected by the *CATSPER2* gene deletions.

Other Names for This Condition

- Chromosome 15q15.3 deletion syndrome
- DIS
- Sensorineural deafness and infertility
- Sensorineural deafness and male infertility

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Deafness-infertility syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1970187/)

Genetic and Rare Diseases Information Center

- Deafness-infertility syndrome (https://rarediseases.info.nih.gov/diseases/11911/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Catalog of Genes and Diseases from OMIM

- DEAFNESS-INFERTILITY SYNDROME; DIS (https://omim.org/entry/611102)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=(deafness+infertility+syndrome))

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