Sensorineural deafness and male infertility

Sensorineural deafness and male infertility is a condition characterized by hearing loss and an inability to father children. 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. Males with this condition produce sperm that have decreased movement (motility), causing affected males to be infertile.

Frequency

The prevalence of sensorineural deafness and male infertility is unknown.

Causes

Sensorineural deafness and male infertility is caused by a deletion of genetic material on the long (q) arm of chromosome 15. The signs and symptoms of sensorineural deafness and male infertility 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 a particular gene on chromosome 15, the \textit{STRC} gene, is responsible for hearing loss in affected individuals. The loss of another gene, \textit{CATSPER2}, in the same region of chromosome 15 is responsible for the sperm abnormalities and infertility in affected males. Researchers are working to determine how the loss of additional genes in the deleted region affects people with sensorineural deafness and male infertility.

Inheritance Pattern

Sensorineural deafness and male infertility 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 sensorineural deafness and male infertility each carry one copy of the chromosome 15 deletion, but they do not show symptoms of the condition. Males with two chromosome 15 deletions in each cell have sensorineural deafness and infertility. Females with two chromosome 15 deletions in each cell have sensorineural deafness as their only symptom because the \textit{CATSPER2} gene deletions affect sperm function, and women do not produce sperm.

Other Names for This Condition

- chromosome 15q15.3 deletion syndrome
- deafness-infertility syndrome
- DIS
Diagnosis & Management

Genetic Testing Information

• What is genetic testing? /primer/testing/genetictesting

• Genetic Testing Registry: Deafness-infertility syndrome

Research Studies from ClinicalTrials.gov

• ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22Infertility%2C+Male%22+OR+%22sensorineural+deafness+and+male+infertility%22+OR+%22Male+Infertility%22

Other Diagnosis and Management Resources

• GeneReview: CATSPER-Related Male Infertility
  https://www.ncbi.nlm.nih.gov/books/NBK22925

• MedlinePlus Health Topic: Assisted Reproductive Technology
  https://medlineplus.gov/assistedreproductivetechnology.html

• RESOLVE: The National Infertility Association: Semen Analysis
  https://resolve.org/infertility-101/infertility-faq/male-fertility-workup/

Additional Information & Resources

Health Information from MedlinePlus

• Encyclopedia: Sensorineural Deafness
  https://medlineplus.gov/ency/article/003291.htm

• Health Topic: Assisted Reproductive Technology
  https://medlineplus.gov/assistedreproductivetechnology.html

• Health Topic: Hearing Disorders and Deafness
  https://medlineplus.gov/hearingdisordersanddeafness.html

• Health Topic: Male Infertility
  https://medlineplus.gov/maleinfertility.html

Genetic and Rare Diseases Information Center

• Deafness-infertility syndrome
Educational Resources

- Boston Children's Hospital: Sensorineural Hearing Loss
  http://www.childrenshospital.org/conditions-and-treatments/conditions/s/sensorineural-hearing-loss

- Boys Town National Research Hospital: The Normal Ear
  https://www.boystownhospital.org/knowledge-center/the-normal-ear

- MalaCards: deafness-infertility syndrome
  https://www.malacards.org/card/deafness_infertility_syndrome

- Merck Manual Professional Version: Sperm Disorders
  https://www.merckmanuals.com/professional/gynecology-and-obstetrics/infertility/sperm-disorders

- Orphanet: Deafness-infertility syndrome
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=94064

Patient Support and Advocacy Resources

- Medical Home Portal: Hearing Loss and Deafness
  https://www.medicalhomeportal.org/diagnoses-and-conditions/hearing-loss-and-deafness

- RESOLVE: The National Infertility Association
  https://resolve.org/

- University of Kansas Medical Center Resource List: Hard of Hearing/Deafness
  http://www.kumc.edu/gec/support/hearing.html

- University of Kansas Medical Center Resource List: Infertility
  http://www.kumc.edu/gec/support/inferti.html

Clinical Information from GeneReviews

- CATSPER-Related Male Infertility
  https://www.ncbi.nlm.nih.gov/books/NBK22925

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28sensorineural+deafness+and+male+infertility%29+AND+english%5Bl%5D+AND+human%5Bm%5D+AND+LAST+3600+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- DEAFNESS-INFERTILITY SYNDROME
  http://omim.org/entry/611102
Sources for This Summary

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