CATSPER2 gene

cation channel sperm associated 2

Normal Function

The *CATSPER2* gene provides instructions for producing a protein that is found in the tail (flagellum) of sperm cells. The CATSPER2 protein plays a role in sperm cell movement (motility). It is one of four proteins that together form a hole (pore) that allows for the movement of charged calcium atoms (Ca^{2+}) across the sperm cell membrane. Ca^{2+} is needed for a type of sperm motility called hyperactivation. Hyperactivation is characterized by vigorous movements of the sperm tail, allow the sperm to push through the membrane of the egg cell during fertilization.

Health Conditions Related to Genetic Changes

Deafness-infertility syndrome

Deafness-infertility syndrome is 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 in people with this condition contains multiple genes, including the *CATSPER2* gene. People with this condition have the deletion in both copies of chromosome 15 in each cell. As a result of the deletion, no CATSPER2 protein is produced. A lack of CATSPER2 protein impairs Ca^{2+} entry into the sperm cell and prevents hyperactivation. Without hyperactivation, sperm have decreased motility and are unable to push through the membrane of the egg cell and achieve fertilization.

Loss of another gene on chromosome 15, the *STRC* gene, is responsible for the hearing loss in affected individuals. 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

- cation channel, sperm associated 2
- CTSR2_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CATSPER2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=117155[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28CATSPER2%5BTIAB%5D%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

- CATION CHANNEL, SPERM-ASSOCIATED, 2; CATSPER2 (https://omim.org/entry/607249)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/117155)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=CATSPER2[gene])

References

- Avidan N, Tamary H, Dgany O, Cattan D, Pariente A, Thulliez M, Borot N, MoatiL, Barthelme A, Shalmon L, Krasnov T, Ben-Asher E, Olender T, Khen M, Yaniv I, Zaizov R, Shalev H, Delaunay J, Fellous M, Lancet D, Beckmann JS. CATSPER2, ahuman autosomal nonsyndromic male infertility gene. Eur J Hum Genet. 2003Jul; 11(7):497-502. doi: 10.1038/sj.ejhg.5200991. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/12825070)
- Luo T, Chen HY, Zou QX, Wang T, Cheng YM, Wang HF, Wang F, Jin ZL, Chen Y, Weng SQ, Zeng XH. A novel copy number variation in CATSPER2 causes idiopathicmale infertility with normal semen parameters. Hum Reprod. 2019 Mar1;34(3):414-423. doi: 10.1093/humrep/dey377. Citation on PubMed (https://www.ncbi.nlm.nih.gov/pubmed/30629171)
- Maurya S, Bhoi NR, Kesari KK, Roychoudhury S, Kumar D. In Silico Analysis ofCatSper Family Genes and APOB Gene Regulation in Male Infertility. Adv Exp MedBiol. 2022;1391:323-332. doi: 10.1007/978-3-031-12966-7_18. Citation on PubMed (https://www.ncbi.nlm.nih.gov/pubmed/36472830)
- Quill TA, Sugden SA, Rossi KL, Doolittle LK, Hammer RE, Garbers DL. Hyperactivated sperm motility driven by CatSper2 is required for fertilization.Proc Natl Acad Sci U S A. 2003 Dec 9;100(25):14869-74. doi:10.1073/pnas.2136654100.

Epub 2003 Dec 1. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/14657366) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC299835/)

- Zhang Y, Malekpour M, Al-Madani N, Kahrizi K, Zanganeh M, Lohr NJ, Mohseni M, Mojahedi F, Daneshi A, Najmabadi H, Smith RJ. Sensorineural deafness and maleinfertility: a contiguous gene deletion syndrome. J Med Genet. 2007Apr;44(4):233-40. doi: 10.1136/jmg.2006.045765. Epub 2006 Nov 10. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17098888) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2598039/)

Genomic Location

The *CATSPER2* gene is found on chromosome 15 (https://medlineplus.gov/genetics/chromosome/15/).

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