

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\[geneid\]](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%29>)

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\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=CATSPER2[gene]))

References

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Genomic Location

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

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