CATSPER1-related nonsyndromic male infertility

Description

CATSPER1-related nonsyndromic male infertility is a condition that affects the function of sperm, leading to difficulty conceiving children (a condition called infertility). People with this condition produce sperm that have decreased movement (motility). Affected individuals may also produce fewer sperm cells or sperm cells that are abnormally shaped. These sperm abnormalities prevent people with this condition from conceiving without assisted reproductive technologies.

Frequency

The prevalence of *CATSPER1*-related nonsyndromic male infertility is unknown.

Causes

Variants (also called mutations) in the *CATSPER1* gene cause *CATSPER1*-related nonsyndromic male infertility. The *CATSPER1* gene provides instructions for producing a protein that is found in the tail of sperm cells. The CATSPER1 protein is involved in the movement of the sperm tail, which propels the sperm forward and is required for sperm cells to push through the outside membrane of the egg cell during fertilization.

CATSPER1 gene variants cause sperm cells to produce altered versions of the CATSPER1 protein that are not functional or that can be quickly broken down (degraded) by the cell. Sperm cells that are missing a functional CATSPER1 protein have decreased motion in their tails and move more slowly than normal. These sperm cells cannot push through the outside membrane of the egg cell. As a result, the sperm cells cannot reach the inside of the egg cell to achieve fertilization.

Learn more about the gene associated with CATSPER1-related nonsyndromic male infertility

- CATSPER1

Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell must have a variant to cause the disorder. The parents of an

individual with an autosomal recessive condition each carry one copy of the altered gene, but they typically do not show signs and symptoms of the condition.

People with one Y chromosome (typical for males) who have two *CATSPER1* gene variants in each cell have *CATSPER1*-related nonsyndromic male infertility. People with two X chromosomes (typical for females) who have two *CATSPER1* gene variants in each cell have no symptoms because they do not produce sperm and the changes only affect sperm function.

Other Names for This Condition

- CATSPER-related nonsyndromic male infertility
- CATSPER1-related male infertility

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Spermatogenic failure 7 (https://www.ncbi.nlm.nih.gov/gtr/conditions/C2751811/)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- SPERMATOGENIC FAILURE 7; SPGF7 (https://omim.org/entry/612997)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28CATSPER1%5BALL%5D%29+AND+%28infertility%5BALL%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

References

- Avenarius MR, Hildebrand MS, Zhang Y, Meyer NC, Smith LL, Kahrizi K, NajmabadiH, Smith RJ. Human male infertility caused by mutations in the CATSPER1 channelprotein. Am J Hum Genet. 2009 Apr;84(4):505-10. doi: 10.1016/j.ajhg.2009.03.004.Epub 2009 Apr 2. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/19344877) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2667975/)

- Hildebrand MS, Avenarius MR, Smith RJH. CATSPER-Related MaleInfertility - RETIRED CHAPTER, FOR HISTORICAL REFERENCE ONLY. 2009 Dec 3 [updated2017 Mar 23]. In: Adam MP, Feldman J, Mirzaa GM, Pagon RA, Wallace SE, Amemiya A,editors. GeneReviews(R) [Internet]. Seattle (WA): University ofWashington, Seattle; 1993-2025. Available fromhttp://www.ncbi.nlm.nih.gov/books/NBK22925/ Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/20301780)
- Jalalabadi FN, Cheraghi E, Janatifar R, Momeni HR. The Detection of CatSper1and CatSper3 Expression in Men with Normozoospermia and Asthenoteratozoospermiaand Its Association with Sperm Parameters, Fertilization Rate, Embryo Quality.Reprod Sci. 2024 Mar;31(3):704-713. doi: 10.1007/s43032-023-01397-4. Epub 2023Nov 13. Citation on PubMed (https://www.ncbi.nlm.nih.gov/pubmed/37957468)
- Li HG, Liao AH, Ding XF, Zhou H, Xiong CL. The expression and significance ofCATSPER1 in human testis and ejaculated spermatozoa. Asian J Androl. 2006May;8(3):301-6. doi: 10.1111/j.1745-7262.2006.00132.x. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16625279)
- Singh AP, Rajender S. CatSper channel, sperm function and male fertility.Reprod Biomed Online. 2015 Jan;30(1):28-38. doi: 10.1016/j.rbmo.2014.09.014. Epub2014 Oct 8. Citation on PubMed (https://www.ncbi.nlm.nih.gov/pubmed/25457194)

Last updated April 30, 2024