# CATSPER1-related nonsyndromic male infertility

## Description

*CATSPER1*-related nonsyndromic male infertility is a condition that affects the function of sperm, leading to an inability to father children. Males with this condition produce sperm that have decreased movement (motility). Affected men may also produce a smaller than usual number of sperm cells or sperm cells that are abnormally shaped. Men with *CATSPER1*-related nonsyndromic male infertility do not have any other symptoms related to this condition.

## Frequency

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

## Causes

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 mutations result in the production of a CATSPER1 protein that may be altered, nonfunctional, or quickly broken down (degraded) by the cell. Sperm cells missing a functional CATSPER1 protein have decreased motion in their tails and move more slowly than normal. Sperm cells lacking functional CATSPER1 protein cannot push through the outside membrane of the egg cell. As a result, 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 have mutations. The parents of an individual with an autosomal

recessive condition each carry one copy of the mutated gene, but they typically do not show symptoms of the condition.

Males with two *CATSPER1* gene mutations in each cell have *CATSPER1*-related nonsyndromic male infertility. Females with two *CATSPER1* gene mutations in each cell have no symptoms because the mutations only affect sperm function, and women do not produce sperm.

## 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


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