CATSPER1-related nonsyndromic male infertility

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.

Inheritance Pattern

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
Diagnosis & Management

Genetic Testing Information
• What is genetic testing?
  https://primer/testing/genetictesting

• Genetic Testing Registry: CATSPER-Related Male Infertility

Research Studies from ClinicalTrials.gov
• ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22CATSPER1-related+male+infertility%22+OR+%22Infertility%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: Male Fertility Workup
  https://resolve.org/infertility-101/infertility-faq/male-fertility-workup/

Additional Information & Resources

Health Information from MedlinePlus
• Encyclopedia: Semen Analysis
  https://medlineplus.gov/ency/article/003627.htm

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

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

Educational Resources
• MalaCards: catsper1-related nonsyndromic male infertility
  https://www.malacards.org/card/catsper1_related_nonsyndromic_male_infertility

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

- RESOLVE: The National Infertility Association
  https://resolve.org/
- 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=%28%28CATSPER1%5BALL%5D%29+AND+%28infertility%5BALL%5D%29%29+AND+english%5Blanguage%5D+AND+human%5Bspecies%5D+AND+%22last+3600+days%22

Catalog of Genes and Diseases from OMIM

- SPERMATOGENIC FAILURE 7
  http://omim.org/entry/612997

Sources for This Summary


Reprinted from Genetics Home Reference:

Reviewed: April 2010
Published: March 3, 2020