Congenital bilateral absence of the vas deferens

Description

Congenital bilateral absence of the vas deferens occurs in males when the tubes that carry sperm out of the testes (the vas deferens) fail to develop properly. Although the testes usually develop and function normally, sperm cannot be transported through the vas deferens to become part of semen. As a result, men with this condition are unable to father children (infertile) unless they use assisted reproductive technologies. This condition has not been reported to affect sex drive or sexual performance.

This condition can occur alone or as a sign of cystic fibrosis, an inherited disease of the mucus glands. Cystic fibrosis causes progressive damage to the respiratory system and chronic digestive system problems. Many men with congenital bilateral absence of the vas deferens do not have the other characteristic features of cystic fibrosis; however, some men with this condition may experience mild respiratory or digestive problems.

Frequency

This condition is responsible for 1 percent to 2 percent of all infertility in men.

Causes

Mutations in the *CFTR* gene cause congenital bilateral absence of the vas deferens.

More than half of all men with this condition have mutations in the *CFTR* gene. Mutations in this gene also cause cystic fibrosis. When congenital bilateral absence of the vas deferens occurs with *CFTR* mutations and without other features of cystic fibrosis, the condition is considered a form of atypical cystic fibrosis.

The protein made from the *CFTR* gene forms a channel that transports negatively charged particles called chloride ions into and out of cells. The flow of chloride ions helps control the movement of water in tissues, which is necessary for the production of thin, freely flowing mucus. Mucus is a slippery substance that lubricates and protects the linings of the airways, digestive system, reproductive system, and other organs and tissues.

Mutations in the *CFTR* gene disrupt the function of the chloride channels, preventing them from regulating the flow of chloride ions and water across cell membranes. As a result, cells in the male genital tract produce mucus that is abnormally thick and sticky.

This mucus clogs the vas deferens as they are forming, causing them to deteriorate before birth.

In instances of congenital bilateral absence of the vas deferens without a mutation in the *CFTR* gene, the cause of this condition is often unknown. Some cases are associated with other structural problems of the urinary tract.

Learn more about the gene associated with Congenital bilateral absence of the vas deferens

- CFTR

Inheritance

When this condition is caused by mutations in the *CFTR* gene, it is inherited in an autosomal recessive pattern. This pattern of inheritance means that both copies of the gene in each cell have a mutation. Men with this condition who choose to father children through assisted reproduction have an increased risk of having a child with cystic fibrosis. If congenital absence of the vas deferens is not caused by mutations in *CFTR*, the risk of having children with cystic fibrosis is not increased.

Other Names for This Condition

- Absence of vas deferens
- Absent vasa
- CAVD
- CBAVD
- Congenital absence of vas deferens
- Congenital aplasia of vas deferens
- Congenital bilateral absence of vas deferens

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Congenital bilateral aplasia of vas deferens from CFTR mutation (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0403814/)

Genetic and Rare Diseases Information Center

- Congenital bilateral absence of vas deferens (https://rarediseases.info.nih.gov/diseases/5461/index)

Patient Support and Advocacy Resources

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

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Congenital bilateral absence of the vas deferens%22)

Catalog of Genes and Diseases from OMIM

- VAS DEFERENS, CONGENITAL BILATERAL APLASIA OF; CBAVD (https://omim.org/entry/277180)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28absence+of+vas+deferens%5BTIAB%5D%29+OR+%28absent+vasa%5BTIAB%5D%29+OR+%28cavd%5BTIAB%5D%29+OR+%28cbavd%5BTIAB%5D%29+OR+%28congenital+absence+of+vas+deferens%5BTIAB%5D%29+OR+%28congenital+aplasia+of+vas+deferens%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D)

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