

Y chromosome infertility

Description

Y chromosome infertility is a condition that affects the production of sperm and causes male infertility, which means it is difficult or impossible for affected men to father children. An affected man's body may produce no mature sperm cells (azoospermia), fewer than the usual number of sperm cells (oligospermia), or sperm cells that are abnormally shaped or that do not move properly. Men with Y chromosome infertility do not have any other signs or symptoms related to the condition.

Some men with Y chromosome infertility who have mild to moderate oligospermia may eventually father a child naturally. Men with oligospermia may also be helped with assisted reproductive technologies; most men with Y chromosome infertility have some sperm cells in the testes that can be extracted for this purpose.

Frequency

Y chromosome infertility occurs in approximately 1 in 2,000 to 1 in 3,000 males of all ethnic groups. This condition accounts for about 13 percent of cases of azoospermia and 5 percent of severe oligospermia.

Causes

As its name suggests, this form of infertility is caused by changes in the Y chromosome. People normally have 46 chromosomes in each cell. Two of the 46 chromosomes are sex chromosomes, called X and Y. Females have two X chromosomes (46,XX), and males have one X chromosome and one Y chromosome (46,XY).

Many genes on the Y chromosome are involved in male sex determination and development. In particular, genes in areas of the Y chromosome called azoospermia factor (AZF) regions provide instructions for making proteins that are involved in sperm cell production and development, although the specific functions of these proteins are not well understood.

Y chromosome infertility is caused by deletions of genes in the AZF regions. These deletions remove several genes, or in rare cases, a single gene. Loss of this genetic material likely prevents the production of one or more proteins needed for normal sperm cell development. As a result, either few sperm develop or sperm do not develop at all, leading to Y chromosome infertility.

[Learn more about the chromosome associated with Y chromosome infertility](#)

- y chromosome

Additional Information from NCBI Gene:

- CDY1
- DAZ1
- DAZ2
- DAZ3
- DAZ4
- DDX3Y
- HSFY1
- HSFY2
- KDM5D
- PRY
- RBMY1A1
- RPS4Y2
- TSPY1
- USP9Y
- VCY

Inheritance

Because Y chromosome infertility impedes the ability to father children, this condition is usually not inherited. Most cases of this condition result from new (de novo) deletions on the Y chromosome that occur during formation of sperm cells in an affected individual's father who is not himself infertile. These cases occur in men with no history of the disorder in their family.

When men with Y chromosome infertility do father children, either naturally or with the aid of assisted reproductive technologies, they pass the genetic changes on the Y chromosome to all their sons. As a result, the sons will also have Y chromosome infertility. This form of inheritance is called Y-linked. Daughters do not inherit the Y chromosome and are not affected.

Other Names for This Condition

- Spermatogenic failure, Y-linked
- Y chromosome-related azoospermia

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Spermatogenic failure, Y-linked, 2 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1839071/>)
- Genetic Testing Registry: Spermatogenic failure, Y-linked, 1 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C4551960/>)

Genetic and Rare Diseases Information Center

- Y chromosome infertility (<https://rarediseases.info.nih.gov/diseases/185/index>)

Patient Support and Advocacy Resources

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

Clinical Trials

- ClinicalTrials.gov ([https://clinicaltrials.gov/search?cond=%22Y chromosome infertility%22](https://clinicaltrials.gov/search?cond=%22Y%20chromosome%20infertility%22))

Catalog of Genes and Diseases from OMIM

- SPERMATOGENIC FAILURE, Y-LINKED, 1; SPGFY1 (<https://omim.org/entry/400042>)
- SPERMATOGENIC FAILURE, Y-LINKED, 2; SPGFY2 (<https://omim.org/entry/415000>)

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28y+chromosome%5BTIAB%5D%29+AND+%28infertility%5BTIAB%5D%29+AND+%28AZF%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>)

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