Y chromosome

The Y chromosome is one of the two sex chromosomes in humans (the other is the X chromosome). The sex chromosomes form one of the 23 pairs of human chromosomes in each cell. The Y chromosome spans more than 59 million building blocks of DNA (base pairs) and represents almost 2 percent of the total DNA in cells.

Each person normally has one pair of sex chromosomes in each cell. The Y chromosome is present in males, who have one X and one Y chromosome, while females have two X chromosomes.

Identifying genes on each chromosome is an active area of genetic research. Because researchers use different approaches to predict the number of genes on each chromosome, the estimated number of genes varies. The Y chromosome likely contains 50 to 60 genes that provide instructions for making proteins. Because only males have the Y chromosome, the genes on this chromosome tend to be involved in male sex determination and development. Sex is determined by the *SRY* gene, which is responsible for the development of a fetus into a male. Other genes on the Y chromosome are important for male fertility.

Many genes are unique to the Y chromosome, but genes in areas known as pseudoautosomal regions are present on both sex chromosomes. As a result, men and women each have two functional copies of these genes. Many genes in the pseudoautosomal regions are essential for normal development.

Health Conditions Related to Chromosomal Changes

The following chromosomal conditions are associated with changes in the structure or number of copies of Y chromosome.

46,XX testicular disorder of sex development

In most individuals with 46,XX testicular disorder of sex development, the condition results from an abnormal exchange of genetic material between chromosomes (translocation). This exchange occurs as a random event during the formation of sperm cells in the affected person's father. The translocation affects the gene responsible for development of a fetus into a male (the *SRY* gene). The *SRY* gene, which is normally found on the Y chromosome, is misplaced in this disorder, almost always onto an X chromosome. A fetus with an X chromosome that carries the *SRY* gene will develop as a male despite not having a Y chromosome.

47,XYY syndrome

Males with 47,XYY syndrome have one X chromosome and two Y chromosomes in each cell, for a total of 47 chromosomes. It is unclear why an extra copy of the Y chromosome is associated with tall stature, learning problems, and other features in some boys and men.

Some males with 47,XYY syndrome have an extra Y chromosome in only some of their cells. This phenomenon is called 46,XY/47,XYY mosaicism.

48,XXYY syndrome

48,XXYY syndrome is caused by the presence of an extra X chromosome and an extra Y chromosome in a male's cells. Extra genetic material from the X chromosome interferes with male sexual development, preventing the testes from functioning normally and reducing the levels of testosterone (a hormone that directs male sexual development) in adolescent and adult males. Extra copies of genes from the pseudoautosomal region of the extra X and Y chromosome contribute to the signs and symptoms of 48,XXYY syndrome; however, the specific genes have not been identified.

Y chromosome infertility

Y chromosome infertility is usually caused by deletions of genetic material in regions of the Y chromosome called azoospermia factor (AZF) A, B, or C. Genes in these regions are believed to provide instructions for making proteins involved in sperm cell development, although the specific functions of these proteins are unknown.

Deletions in the AZF regions may affect several genes. The missing genetic material likely prevents production of a number of proteins needed for normal sperm cell development, resulting in an inability to father children.

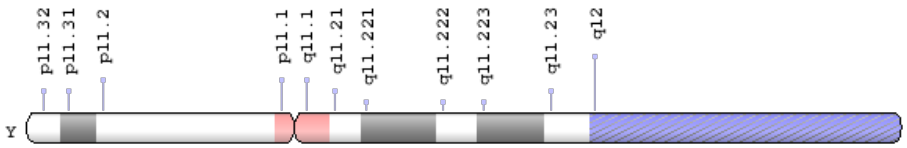
other chromosomal conditions

Chromosomal conditions involving the sex chromosomes often affect sex determination (whether a person has the sexual characteristics of a male or a female), sexual development, and the ability to have children (fertility). The signs and symptoms of these conditions vary widely and range from mild to severe. They can be caused by missing or extra copies of the sex chromosomes or by structural changes in these chromosomes.

Rarely, males may have more than one extra copy of the Y chromosome in every cell (polysomy Y). For example, the presence of two extra Y chromosomes is written as 48,XYYY. The extra genetic material in these cases can lead to skeletal abnormalities, decreased IQ, and delayed development, but the features of these conditions are variable.

Chromosome Diagram

Geneticists use diagrams called idiograms as a standard representation for chromosomes. Idiograms show a chromosome's relative size and its banding pattern, which is the characteristic pattern of dark and light bands that appears when a chromosome is stained with a chemical solution and then viewed under a microscope. These bands are used to describe the location of genes on each chromosome.



Credit: Genome Decoration Page/NCBI

Additional Information & Resources

MedlinePlus

- Encyclopedia: Chromosome
  https://medlineplus.gov/ency/article/002327.htm

Additional NIH Resources

- National Human Genome Research Institute: Chromosome Abnormalities
  https://www.genome.gov/11508982/
- National Human Genome Research Institute: Researchers Discover Use of Novel Mechanism Preserves Y Chromosome Genes (June 18, 2003)
  https://www.genome.gov/11007628/

Educational Resources

- Genome News Network: Sex and "The Y" (June 27, 2003)
  http://www.genomenewsnetwork.org/articles/06_03/y_chrom.shtml

GeneReviews

- Y Chromosome Infertility
  https://www.ncbi.nlm.nih.gov/books/NBK1339

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28Chromosomes,+Human,+Y%5BMAJR%5D%29+AND+%28Y+Chromosome%5BTI%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D

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