



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 enabling men to father biological children (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. In the translocation that causes 46,XX testicular disorder of sex development, the *SRY* gene, which is normally found on the Y chromosome, is misplaced, almost always onto an X chromosome. An individual with an X chromosome that carries the *SRY* gene will develop as a male despite not having a Y chromosome, but will not be able to produce sperm to father biological children.

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. An extra copy of the genes contained in the pseudoautosomal region of the Y chromosome may explain the tall stature and other features that can affect boys and men with this condition.

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, a condition that leads to infertility, developmental and behavioral disorders, and other health problems in males, 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 chromosomes contribute to the signs and symptoms of 48,XXYY syndrome; however, the specific genes have not been identified.

Y chromosome infertility

Deletions of small amounts of genetic material in certain areas of the Y chromosome lead to a condition called Y chromosome infertility. This condition affects the production of sperm and makes it difficult or impossible for affected men to father children. The deletions occur in areas of the Y chromosome called azoospermia factor (AZF) regions. Genes in these regions provide instructions for making proteins thought to be involved in sperm cell development, although the specific functions of these proteins are unknown.

Deletions in the AZF regions remove all or part of 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, sperm develop abnormally or do not develop at all, leading to Y chromosome infertility.

Other chromosomal conditions

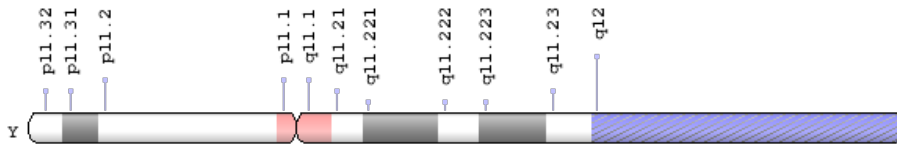
Chromosomal conditions involving the sex chromosomes often affect sex determination (whether a person has the sexual characteristics of a male or a female), sexual development, and fertility. The signs and symptoms of these conditions vary widely and range from mild to severe. They can be caused by missing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

Health Information from 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

Clinical Information from 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%5BM AJR%5D%29+AND+%28Y+Chromosome%5BTI%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D>

Research Resources

- Cancer Genetics Web
<http://www.cancerindex.org/geneweb/clinkc24.htm>
- Database of Genomic Variants
http://projects.tcag.ca/variation/cgi-bin/tbrowse/tbrowse?source=hg17&table=Locus&show=table&keyword=&flop=AND&fcol=_C19&fcomp==&fkwd=chrY&cols=
- Ensembl Human Map View
http://www.ensembl.org/Homo_sapiens/Location/Chromosome?chr=Y;r=Y:1-57227415
- The male-specific region of the human Y chromosome is a mosaic of discrete sequence classes. *Nature*. 2003 Jun 19;423(6942):825-37.
<https://www.nature.com/articles/nature01722.pdf>
- U.S. Department of Energy: Human Genome Project Information Archive
https://web.ornl.gov/sci/techresources/Human_Genome/posters/chromosome/chromoY.shtml
- Y Chromosome Haplotype Reference Database (YHRD)
<https://yhrd.org/>

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