Y chromosome

Description

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 70 to 200 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 difference of sex development

In most individuals with 46,XX testicular difference 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 difference 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

Individuals 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 those individuals with this condition.

Some people 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, is caused by the presence of an extra X chromosome and an extra Y chromosome in cells. Extra genetic material from the X chromosome interferes with sex development, though affected individuals are often assigned male gender at birth. Small testes that do not function normally lead to a reduction in the levels of testosterone (a hormone that directs male sex development). Extra copies of genes from the pseudoautosomal regions of the extra X and Y chromosomes likely contribute to the signs and symptoms of 48,XXYY syndrome.

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 individuals 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

Other chromosomal conditions involving the sex chromosomes can also often affect sex 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, more than one extra copy of the Y chromosome can occur 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.

Additional Information & Resources

Additional NIH Resources

- National Human Genome Research Institute: Chromosome Abnormalities (https://www.genome.gov/about-genomics/fact-sheets/Chromosome-Abnormalities-Fact-Sheet)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28Chromosomes,+Human,+Y%5BMAJR%5D%29+AND+%28Y+Chromosome%5BTI%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D)

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