In the nucleus of a human cell, each DNA molecule is packaged into a long, thread-like structure called a chromosome. Most human cells contain 23 pairs of chromosomes. One half of each pair of chromosomes comes from one parent, while the other half comes from the other parent. The 23rd pair are the X and Y chromosomes, often called the sex chromosomes. The other 22 pairs are called autosomes.

The X and Y chromosomes are central for the process of sexual development, but other genes throughout the genome play a role. This complex process gives rise to the array of human sex characteristics found among male, female and intersex individuals. The Y chromosome is most commonly associated with male individuals, but the Y chromosome does not singularly define a person’s sex.

In fertilization, sperm can contribute either an X or a Y chromosome, while eggs always contribute an X chromosome.

One of the genes on the Y chromosome is the SRY gene. The protein produced by this gene turns on a set of other genes that cause the embryo to develop certain sex characteristics, such as testes. If the SRY gene doesn’t produce a functional protein, the embryo will not develop testes despite having a Y chromosome.

Variation in the number of sex chromosomes in a cell is not uncommon. Some individuals have more than two sex chromosomes in all of their cells, such as individuals with XXX, XYY or XXXY chromosomes. Some individuals have a single sex chromosome (X or 0X), or one of these chromosomes may be “incomplete.” Also, many people born with a Y chromosome can lose the Y chromosome from their cells as they age. Smoking may exacerbate this loss.

Some genes that were thought to be lost from the Y chromosome have actually relocated to other chromosomes.

About 66% of the Y chromosome is composed of repetitive DNA, which is particularly difficult for researchers to study. Specialized DNA sequencing and analysis techniques are needed to determine the arrangement of these highly similar segments.

Many health conditions are thought to be related to changes in genes on the Y chromosome. This is currently an active area of research!