



X and Y chromosome variations are conditions that result when a baby has a different number of sex chromosomes, also known as X & Y chromosomes, than we would expect to see in some or all of the cells in their body. Typically, girls have two X chromosomes while boys have one X and one Y chromosome. Many variations from this number can occur, and are also sometimes referred to as “sex chromosome anomaly,” “sex chromosome abnormality”, or “sex chromosome aneuploidy.”

The most common variations involve a trisomy, which means three sex chromosomes instead of the typical two. Girls who are born with an extra X chromosome are referred to as having Triple X or [Trisomy X](#). Boys who are born with an extra X chromosome have [47,XXY](#), also known as [Klinefelter syndrome](#). And boys who are born with an extra Y chromosome have [47,XYY](#), occasionally referred to as [Jacob’s syndrome](#).

In addition, there are a number of other X and/or Y conditions including 48 or 49 chromosomes. These include 48,XXXX, 48XXXY, 48XXYY and 48XYYY; and although increasingly rare, also 49XXXXX, 49XXXXY, 49XXXYY, 49XXYYY and 49XYYY. Some individuals may have two cell lines in their body, called mosaicism, such as 46,XY/[47,XXY](#).

There is only one X and Y chromosome variation that involves a monosomy (missing sex chromosome): [Turner syndrome](#). [Turner syndrome](#) is caused when a girl has one X chromosome and no second sex chromosome ([45,X](#)).

What Causes X and Y Chromosome Variations?

We know that X and Y chromosome variations occur when there is an error in the cell division early on in development, or if the sperm or egg that make the baby had an extra sex chromosome. The exact reason why this happens is currently unknown.

What are the Health and Developmental Concerns Associated with X and Y Chromosome Variations?

It is important to remember that the signs and symptoms of X and Y chromosome variations can vary quite a bit from person to person. During pregnancy, the conditions may be diagnosed through genetic testing such as a [chorionic villus sampling \(CVS\)](#) or [amniocentesis](#) or may be suspected through a blood test such as [cell-free DNA \(cfDNA\)](#) analysis. Keep in mind that [cfDNA](#) analysis cannot tell for certain if a baby has a variation in their number of X and Y chromosomes.

Please click on the links below for information known about specific chromosome variations:



[47,XXX \(Trisomy X syndrome\)](#)

[47,XXY \(Klinefelter syndrome\)](#)

[47,XYY \(Jacobs syndrome\)](#)

[45,X \(Turner syndrome\)](#)

If I have a baby with an X and Y chromosome variation, what is the chance I will have another baby with this condition?

In most cases, the chance that a woman will have another baby with an X and Y chromosome variation is thought to be less than 1%, as the majority of cases of X and Y chromosome variation are not inherited.

Treatment for X and Y Chromosome Variations

There is no “cure” for X and Y chromosome variations. Because many individuals with X and Y chromosome variations do not have significant medical issues, treatment is generally determined based on the specific patient. For example, if an individual with an X or Y chromosome variation is having learning difficulties, early intervention and extra support in school should be considered. Some people with sex chromosome differences may also have a difference in height and development during puberty, which hormone therapy can help with. It is important to work with your doctor if you have an X or Y chromosome variation to determine the care plan that is right for you.

How common are X and Y Chromosome Variations?

Variations in the number of X and Y chromosomes are estimated to occur in one of every 400 to 500 newborns.

Outlook and Prognosis

Most individuals with X and Y chromosome variations will have differences in final adult height and have infertility. Some will experience difficulties in learning, however, many have success in school, including higher education. Men and women with X and Y chromosome disorders work in a variety of career settings. Some may have children on their own or with the help of assisted reproductive technologies. While others may choose to adopt. Individuals with X and Y chromosome variations often benefit from careful monitoring of their health throughout their life. However, most can expect to live full and



productive lives.

Click [here](#) to learn more about scheduling a genetic counseling appointment for pregnancy-related questions.

Click [here](#) to learn more about scheduling a genetic counseling appointment for infertility or preconception questions.

Click [here](#) to learn more about scheduling a genetic counseling appointment for questions about pediatric or adult genetic conditions.

Resources

[The Association for X and Y Variations \(AXYS\)](#)

[The Focus Foundation](#)

Related Articles

- [Down Syndrome](#)

Down syndrome, also known as trisomy 21, is a genetic condition caused by an extra copy of the 21st chromosome. Each individual with Down syndrome is unique. Individuals with Down syndrome have variable health issues, including developmental delays and some degree of intellectual disability. What is life like for people...

- [Trisomy 18 \(Edwards Syndrome\)](#)

Trisomy 18, also known as Edwards syndrome, is a genetic condition caused by an extra chromosome 18. Babies with trisomy 18 generally have many complex medical complications, including heart defects, growth restriction, a small abnormally shaped head, and clenched fingers with overlapping fingers. Because of the severity of medical complications...

- [Trisomy 13 \(Patau Syndrome\)](#)

Trisomy 13, also known as Patau syndrome, is a genetic condition caused by an extra chromosome 13. Babies with trisomy 13 generally have many complex medical complications, including heart defects, brain and spinal cord abnormalities, very small or poorly developed eyes, cleft lip and/or cleft palate, and low muscle tone...

- [Open Neural Tube Defects](#)

Open neural tube defects (ONTDs), which includes spina bifida and anencephaly, is a



range of birth defects that can include the brain and/or spinal cord. This is generally something that happens very early in pregnancy, and can vary widely in severity and expected outcome. Although the cause of most cases...

- [Microdeletion Syndromes](#)

There are many different kinds of genetic conditions. Some are caused by whole extra or missing chromosomes (such as Down syndrome), while others are caused by changes in a specific gene (such as sickle cell anemia). There is a third group of genetic conditions called microdeletion syndromes. A microdeletion is...