



## Overview of Important Information about 47,XXY (Klinefelter syndrome)

47,XXY occurs when a boy has an extra copy of an X chromosome in some or all of his cells.

It usually does not cause them to look physically different than their peers, but does cause a variety of subtle physical differences including:



Klinefelter syndrome  
(47,XXY)

- taller than average height,
- lower muscle tone, and
- impaired fertility.

Other features that can be seen include a curved little finger, difficulty fully straightening the elbows, flat feet, and a small depression in the chest.

Individuals with 47,XXY typically have intelligence in the normal range, although some learning disabilities and speech/expressive language delay are common. Some individuals with Klinefelter syndrome have more significant health and developmental concerns, while others may have few, if any, notable features, and may not even know that they have the condition.

### What Causes Klinefelter syndrome?

47,XXY occurs when either the sperm or egg that make a pregnancy has an extra sex chromosome, or when there are errors in cell division early in a pregnancy's development. The exact reason for these occurrences is not known.



## **What are the Health and Developmental Concerns Associated with Klinefelter syndrome?**

During pregnancy, the condition may be diagnosed through genetic testing such as a [chorionic villus sampling \(CVS\)](#) or [amniocentesis](#), or suspected through a blood test such as [cell-free DNA \(cfDNA\)](#) analysis. Keep in mind that [cfDNA](#) cannot tell for certain if a baby has a variation in their number of X and Y chromosomes. It is important to remember that the signs and symptoms of Klinefelter syndrome can vary quite a bit from person to person. While [prenatal diagnostic testing](#) can provide a 'yes' or 'no' answer about whether a pregnancy has Klinefelter syndrome, it can not predict the specific health concerns that a baby with Klinefelter might have due to the variability of the condition.

During the newborn period, boys with Klinefelter syndrome are often no different from other boys. During early childhood, low muscle tone and/or a delay in meeting developmental milestones may or may not be seen. Between the ages of 5 and 8 they may often have an increase in height above their peers, and may also have longer arms and legs. During school years, there may also be a lag in language skills and academic difficulties.

Many boys with Klinefelter syndrome appear to enter puberty normally but have a tendency for testosterone levels to decline in late adolescence and early adulthood. Therefore, testosterone supplements may be given to fully develop secondary sexual characteristics, such as muscle patterns, a deeper voice, and facial and body hair.

Most men with Klinefelter syndrome have infertility. However, there have been reports of men fathering natural pregnancies and pregnancies with assisted medical technologies. Most of these cases have occurred in men that have a mixture of 47,XXY and 46,XY cells (mosaicism).

As men with Klinefelter syndrome grow older, there is some higher risk for a range of conditions including osteoporosis, thyroid disorders, diabetes, and autoimmune diseases. They also have an increased risk of acquiring leg ulcers.

## **If I have a baby with Klinefelter syndrome, 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 Klinefelter syndrome:**

There is no “cure” for Klinefelter syndrome. Babies with Klinefelter syndrome often do not need any special treatment or intervention as infants. If any symptoms, such as learning disabilities, arise during childhood, they can be treated and managed with early intervention and support in school.

Many teens and adults with 47,XXY are given testosterone supplements to support full development of male secondary sex characteristics. Testosterone also helps increase and maintain bone density, but is unable to reverse infertility. Therefore, children who are reaching adolescence should work with a pediatric endocrinologist to determine if hormone therapy is recommended.

### **How common is Klinefelter syndrome?**

47,XXY (Klinefelter syndrome) is the most common [variation in X and Y chromosomes](#) and is estimated to occur in one of every 500 males.

### **Outlook for individuals with Klinefelter syndrome:**

Most men with Klinefelter syndrome will be taller than average and have infertility. Some may experience difficulty in learning in some areas, however many individuals with Klinefelter syndrome have success in school, including higher education. While many have issues with infertility some are able to have children with or without reproductive assistance, while others will go on to adopt children. Individuals with Klinefelter syndrome may need monitoring of their health throughout their life, but 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**

[Klinefelter Syndrome Information and Support](#)



47,XXY (Klinefelter Syndrome)

[Klinefelter Syndrome Global Support Group](#)

[The American Association for Klinefelter Syndrome Information and Support](#)

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

[The Focus Foundation](#)