What Is Klinefelter Syndrome?


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**Also known as XXY syndrome, this chromosomal condition affects only males.**

A common genetic chromosomal condition, Klinefelter syndrome occurs when a boy is born with an extra copy of the X chromosome.

Klinefelter syndrome can affect both physical and cognitive development.

According to the National Human Genome Research Institute, Klinefelter syndrome is found in about 1 out of every 500 to 1,000 newborn males.

**Klinefelter Syndrome Causes**

Klinefelter syndrome occurs as the result of a random genetic error during the formation of the egg or sperm or after conception.

The syndrome is not the result of something the parents did or did not do.

However, women who become pregnant after 35 years of age have a slightly increased risk of having a boy with Klinefelter syndrome.

People normally have 46 chromosomes in each cell. Two of those 46 chromosomes, known as X and Y, are called sex chromosomes because they help determine whether a person will develop male or female sex characteristics.

Females usually have two X chromosomes, while males have one X chromosome and one Y chromosome.

However, Klinefelter syndrome occurs because of one extra copy of the X chromosome in each cell (XXY).

Some males with Klinefelter syndrome have the extra X chromosome only in some of their cells. When this occurs it is called mosaic Klinefelter syndrome.

While rare, a more severe form of Klinefelter can occur if a male has more than one extra copy of the X chromosome.

**Klinefelter Syndrome Symptoms**

The most common symptom of Klinefelter syndrome is infertility, which often isn't noticed until adulthood.

Klinefelter syndrome adversely affects testicular growth, resulting in smaller than normal testicles, which often leads to low production of testosterone.

Although most men with Klinefelter syndrome produce little or no sperm, reproductive procedures may make it possible for some men with the condition to father children.
The effects of Klinefelter syndrome vary from person to person, and signs and symptoms of the syndrome can appear differently at different ages.

**Symptoms in babies:**
- Weak muscles
- Slow motor development of things like sitting up, crawling, and walking
- Speech delay
- Docile personality
- Testicles that haven't descended into the scrotum

**Symptoms in boys and teenagers:**
- Taller than average stature
- Longer legs, shorter torso, and broader hips
- Absent, delayed, or incomplete puberty
- Less-muscular bodies and less facial and body hair after puberty
- Small, firm testicles
- Small penis
- Enlarged breast tissue
- Weak bones
- Low energy levels
- Shyness
- Difficulty expressing feelings or socializing
- Problems with reading, writing, spelling, or math
- Difficulty being attentive

**Symptoms in men:**
- Infertility
- Small testicles and penis
- Taller than average stature
- Weak bones (osteoporosis)
- Decreased facial and body hair
- Enlarged breast tissue
- Decreased sex drive

**Complications**

Men with Klinefelter syndrome may be at increased risk for the following:

- Varicose veins and other problems with blood vessels
- Breast cancer and cancers of the blood, bone marrow, or lymph nodes
- Lung disease
- Autoimmune disorders, such as type 1 diabetes and lupus
- Belly fat, which can lead to other health problems
- Testosterone replacement therapy can reduce the risk of the health problems listed above, especially when therapy is started at the onset of puberty.

**Klinefelter Syndrome Diagnosis**
If you suspect that you or your son may have Klinefelter syndrome, your doctor will examine your (or your son's) genital area and chest, as well as conduct tests to check reflexes and mental functioning.

Additionally, the following tests are used to diagnose Klinefelter Syndrome:

**Chromosomal analysis:** Also called karyotype, this involves drawing a small sample of blood, which is sent to a lab to check the shape and number of chromosomes.

**Hormone testing:** This can be done through blood or urine samples and can detect if abnormal sex hormone levels — a sign of Klinefelter syndrome — are present.

**Klinefelter Syndrome Treatment**

While there is no cure for sex chromosome changes caused by Klinefelter syndrome, treatments can help minimize effects, especially when they're started early. Treatment may include the following.

**Testosterone replacement therapy:** Since males with Klinefelter syndrome don't produce enough testosterone, undergoing testosterone replacement therapy at the onset of puberty can allow a boy to undergo the body changes that normally occur at puberty, such as developing a deeper voice, growing facial and body hair, and increasing muscle mass and penis size.

Testosterone therapy can also improve bone density and reduce the risk of fractures, but it will not enlarge the testicles or help with infertility.

Testosterone may be given as an injection or with a gel or patch on the skin.

**Fertility treatments:** Intracytoplasmic sperm injection (ICSI) can help men with minimal sperm production by removing sperm from the testicle and injecting it directly into a woman's egg.

**Breast tissue removal:** For those who develop enlarged breasts, excess breast tissue can be removed with plastic surgery.

**Educational support:** Teachers, school counselors, or school nurses may be able to help with finding extra support for learning.

**Speech and physical therapy:** These can help overcome problems with speech, language, and muscle weakness.

**Psychological counseling:** Whether from a family therapist, counselor, or psychologist, counseling can help people with Klinefelter syndrome work through emotional issues related to puberty, young adulthood, and infertility.

Sources

- Klinefelter syndrome; [National Institutes of Health](https://www.nationalhealth.gov).
- Klinefelter syndrome; [Mayo Clinic](https://www.mayoclinic.org).
- Learning About Klinefelter Syndrome; [National Human Genome Research Institute](https://www.nhgri.nih.gov).