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Klinefelter Syndrome

Klinefelter syndrome is a genetic condition in which a boy is born with an extra X chromosome. Instead of the typical XY chromosomes in men, they have XXY, so this condition is sometimes called XXY syndrome.

Men with Klinefelter usually don't know they have it until they run into problems trying to have a child. There's no cure, but it can be treated.

Other Names for This Condition

- 47,XXY syndrome
- Klinefelter syndrome (KS)
- Klinefelter's syndrome
- XXY syndrome
- XXY trisomy

Frequency

Klinefelter syndrome affects about 1 in 650 newborn boys. It is among the most common sex chromosome disorders, which are conditions caused by changes in the number of sex chromosomes (the X chromosome and the Y chromosome).

What Causes Klinefelter Syndrome?

You get the extra X chromosome by chance. Either the egg or the sperm that came together to create you had an extra X chromosome. Older women have a slightly higher chance of having a boy with XXY syndrome, but the chance is small.

Men with Klinefelter may have:

- An extra X chromosome in every cell, which is the most common
- An extra X chromosome in only some cells, called mosaic Klinefelter, in which you don't have as many symptoms
- More than one extra X chromosome, which is very rare and more severe.

Symptoms of Klinefelter Syndrome

Some men show symptoms of Klinefelter as children, but others don't know they have it until puberty or adulthood. Many men never realize that they have it because symptoms aren't always present.

Symptoms of Klinefelter vary with age and include:

Babies:

- Hernia
- More quiet than usual

- Slower to learn to sit up, crawl, and talk
- Testicles that haven't dropped into the scrotum
- Weaker muscles

Children:

- A hard time making friends and talking about feelings
- Low energy levels
- Problems learning to read, write, and do math
- Shyness and low confidence

Teenagers:

- Larger breasts than normal
- Less facial and body hair, and it comes in later
- Less muscle tone, and muscles grow slower than usual
- Longer arms and legs, wider hips, and a shorter torso than other boys their age
- Puberty never comes, comes later, or doesn't quite finish
- Small penis and small, firm testicles
- Taller than usual for the family

Adults:

- Infertility (can't have children because they can't make enough sperm)
- Low sex drive
- Low testosterone levels
- Problems getting or keeping an erection

When to see a doctor

See a doctor if you or your son has:

- **Slow development during infancy or boyhood.** Delays in growth and development can be the first sign of a number of conditions that need treatment — including Klinefelter syndrome. Though some variation in physical and mental development is normal, it's best to check with a doctor if you have any concerns.
- **Male infertility.** Many men with Klinefelter syndrome aren't diagnosed with infertility until they realize they're unable to father a child.

Treatment

A number of complications caused by Klinefelter syndrome are related to low testosterone (hypogonadism). Testosterone replacement therapy reduces the risk of certain health problems, especially when therapy is started at the beginning of puberty.