
Klinefelter syndrome

Summary

- Klinefelter syndrome is a chromosome disorder that affects males. It is not an inherited disorder.
 - A male with Klinefelter syndrome has an additional X chromosome, which causes infertility, low testosterone and other characteristics such as development of breast tissue.
 - The chromosomes are present in every cell of the body and the extra X chromosome cannot be removed.
 - Treatment includes lifelong testosterone therapy and cosmetic surgery to remove enlarged breast tissue. Options for men with Klinefelter syndrome to have their own children are becoming more widely available.
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Klinefelter syndrome is a chromosome disorder that affects males. Usually, a male has two chromosomes that determine his sex: an X inherited from his mother and a Y inherited from his father. A male with Klinefelter syndrome has an additional X chromosome.

The effects can vary greatly and may include:

- infertility – the condition may be diagnosed when fertility problems are being investigated
 - reduced testicle size – in all males with Klinefelter syndrome, the testicles do not develop properly and are noticeably smaller from early adolescence
 - incomplete puberty – not enough of the male hormone testosterone is produced and puberty may appear to be delayed. These boys may be given testosterone treatment so they develop usual male physical characteristics like facial hair and a deeper voice
 - language and learning problems – in many males with Klinefelter syndrome, difficulties with speech, writing, or understanding and processing speech may be noticed.
- Klinefelter syndrome occurs in around one in every 450 male babies, which makes it one of the most common variations of the chromosomes. However, only around a quarter of these males have a diagnosis of Klinefelter syndrome, also known as XXY syndrome. The additional X chromosome does not influence sexual orientation.

Signs of Klinefelter syndrome at birth

The condition isn't usually diagnosed at birth, because the baby boy looks healthy and unaffected. However, certain physical characteristics sometimes associated with Klinefelter syndrome may be apparent, including:

- small penis
- undescended testicles
- hypospadias (the urethra is located on the underside of the penis instead of the tip).

Signs of Klinefelter syndrome at childhood

The appearance of signs of Klinefelter syndrome in childhood can vary greatly and range from mild to those that are more obvious. Many of the signs of Klinefelter syndrome also occur in children with other conditions, or children who do not have an underlying diagnosis, which makes detecting Klinefelter syndrome in childhood difficult.

Furthermore, many boys show no signs of Klinefelter syndrome during childhood. However, for those that do, early intervention provides the opportunity to overcome difficulties and for each boy to reach their individual potential.

Signs that may be apparent in childhood include:

- delays in starting to talk or walk
 - learning difficulties, including trouble listening or concentrating in class
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- poor motor development or coordination (sometimes leading to shyness or an avoidance of rough sports)
- tiredness and fatigue
- reduced muscle strength or tone (sometimes leading to other complications such as sleep apnoea or chronic constipation).

Signs of Klinefelter syndrome at puberty

Klinefelter syndrome is often diagnosed at puberty, when the expected physical changes don't occur. Some of the signs and symptoms of the condition include:

- small testicles (hypogonadism) – males with Klinefelter syndrome always have small testicles from puberty
- lack of facial, pubic and underarm hair
- poor muscle development
- breast tissue development (gynaecomastia)
- unexplained weight gain, especially on the stomach or trunk
- tiredness and fatigue
- small penis
- disproportionately long arms and legs compared to the length of the body
- taller than average height
- poor concentration
- single crease in the palm (simian crease)
- difficulties with sexual functioning
- speech and learning difficulties – in some cases this can lead to behavioural problems
- sadness, lowered mood or depression.

Boys may become particularly self-conscious in puberty when they notice that their body is not undergoing the same changes as their peers. Testosterone replacement therapy can help the body to undergo these changes.

Most males with Klinefelter syndrome are not diagnosed until they are well into adulthood, when diagnosis can come as a major shock. An earlier diagnosis provides opportunities for the child and his family to access treatments and interventions that can help overcome any difficulties associated with the condition.

Cause of Klinefelter syndrome

While the cause of Klinefelter syndrome is genetic, it is not inherited. Humans have 46 paired chromosomes, made up of two sex chromosomes that determine gender and 44 chromosomes that determine other factors. The mother always passes on an X chromosome. If the father provides an X chromosome, the child will be female, while a Y chromosome makes the child male.

A boy with Klinefelter syndrome has an additional X chromosome. This is thought to be caused by an error within the fertilised egg or the dividing cells as the baby develops.

The presence of the Y chromosome ensures male sexual characteristics but, because the testicles are underdeveloped, there may not be enough testosterone production. This is why the penis and testicles may be smaller than average, and why most men with Klinefelter syndrome do not produce sperm. Some researchers suspect that advanced parental age may be a risk factor.

Complications of Klinefelter syndrome

Compared with males in the general population, Klinefelter syndrome is associated with an increased risk of certain diseases and conditions, including:

- osteoporosis
- diabetes mellitus
- lung disease
- breast cancer

- testicular cancer
- anaemia
- infertility
- depression or other psychiatric conditions
- leukaemia
- non-Hodgkin lymphoma
- thyroid disease
- sleep apnoea
- high cholesterol
- tooth decay
- varicose veins.

It is currently thought that testosterone treatment from puberty, which helps with bone and muscle development among other things, may reduce the risk of many of the above conditions.

Diagnosis of Klinefelter syndrome

Klinefelter syndrome is diagnosed using a number of tests, including:

- physical examination – all males with Klinefelter syndrome have small testicles, which can be detected by genital examination
- chromosome analysis – will confirm the diagnosis
- blood tests – can check for hormone levels
- semen examination – checks fertility.

Treatment of Klinefelter syndrome

There is no cure for Klinefelter syndrome. Treatment options can improve some aspects of the condition and provide emotional support. Sample treatments include:

- hormone therapy
- reproductive technologies to aid fertility
- educational support (including: speech therapy, physical or occupational therapy) – if necessary
- cosmetic surgery
- counselling
- frequent screening tests – to ensure the early diagnosis of any associated complications.

Further information about Klinefelter syndrome can be found on the [**Andrology Australia website**](#).

Where to get help

- Your doctor
- Endocrinologist
- **Genetic Support Network Victoria** Tel. **(03) 8341 6315**
- **Andrology Australia** Tel. **1300 303 878**

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