

Klinefelter syndrome

Klinefelter syndrome is a chromosome disorder that affects males. Normally, 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 an apparently normal male is seen because they have fertility problems.
- **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 delayed. These boys may be given testosterone treatment so they develop normal male physical characteristics like facial hair and a deeper voice.

Klinefelter syndrome occurs in around one in every 450 male babies, which makes it one of the most common variations of the chromosomes. This condition is 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 looks healthy and normal. 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 moderate to mild. 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 and/or walk
- Learning difficulties, including trouble listening or concentrating in class
- Poor motor development and/or coordination (sometimes leading to shyness or an avoidance of rough sports)
- Tiredness and fatigue.

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)
- Tiredness and fatigue
- Small penis
- Disproportionately long arms and legs compared to the length of the body
- Taller than average height
- Poor concentration, depression
- Single crease in the palm (simian crease)
- Difficulties with sexual functioning
- Sometimes, speech and learning difficulties. In some cases this can lead to behavioural problems.

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 assist the body in undergoing these typical pubertal 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 their family to access treatments and interventions that can help overcome any difficulties associated with the condition.

The cause of Klinefelter syndrome is unknown and not inherited

Humans have 46 paired chromosomes, made up of two sex chromosomes that determine gender and 44 chromosomes that dictate 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.

Possible 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
- Pulmonary disease
- Breast cancer
- Infertility
- Leukaemia
- Non-Hodgkin's lymphoma
- Thyroid disease
- 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 diagnosis.
- **Blood tests** – can check for hormone levels.
- **Semen examination** – checks fertility.

Treatment of Klinefelter syndrome

There is no cure for Klinefelter syndrome. Treatment aims to correct some of the abnormalities and provide emotional support. Options may include:

- **Hormone therapy** – such as testosterone, to help bring on the normal changes of puberty. Hormone therapy will need to be lifelong, however it can have a broad range of benefits including increased energy levels and physical strength.
- **Reproductive technologies** – such as IVF, to help men with Klinefelter syndrome become fathers of their own children. New advances in reproductive technologies mean that up to a quarter of men with Klinefelter syndrome are now able to have their own biological children
- **Educational support** – if necessary
- **Speech therapy** – if necessary
- **Cosmetic surgery** – to remove enlarged breast tissue
- **Counselling** – to help the male come to terms with his condition
- **Frequent screening tests** – to ensure the early diagnosis of any associated complications (such as diabetes or breast cancer).

Where to get help

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

Things to remember

- 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 available.

This page has been produced in consultation with, and approved by:

Genetic Health Services Victoria

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