

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 vary and may include:

- **Infertility** - the condition may be diagnosed when an apparently normal male is seen because they have fertility problems.
- **Reduced testicle size** - the testicle may not develop properly.
- **Late puberty** - not enough of the male hormone testosterone is produced and puberty may be late. 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 500 male babies, which makes it one of the most common abnormalities of the sex chromosomes. This condition is also known as XXY syndrome. The additional X chromosome does not influence sexual orientation.

Symptoms at birth

The condition isn't usually diagnosed at birth, because the baby looks healthy and normal. However, certain physical characteristics of 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 at puberty

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

- Small penis
- Small testicles (hypogonadism)
- Difficulties with sexual functioning
- Lack of facial, pubic and underarm hair
- Taller than average height
- Enlarged breasts (gynaecomastia)
- Slim physique
- Disproportionately long legs compared to the length of the body
- Single crease in the palm (simian crease)
- Slightly impaired IQ
- Sometimes, the boy may have speech and hearing difficulties.

The cause 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; 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 are smaller than average, and why most men with Klinefelter syndrome are infertile. Some researchers suspect that advanced maternal age may be a risk factor.

Possible complications

Klinefelter syndrome is associated with an increased risk of certain diseases and conditions, including:

- Breast cancer
- Diabetes mellitus
- Infertility
- Leukaemia
- Non-Hodgkin's lymphoma
- Osteoporosis
- Pulmonary disease
- Thyroid disease
- Tooth decay
- Varicose veins.

Diagnosis methods

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

- **Physical examination** - including rectal exam of the prostate gland (most males with Klinefelter syndrome have an enlarged prostate).
- **Chromosome analysis** - to confirm diagnosis.
- **Blood tests** - to check for hormone levels.
- **Semen examination** - to check fertility.

Treatment options

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

- **Cosmetic surgery** - to remove enlarged breast tissue.
- **Hormone therapy** - such as testosterone, to help bring on the normal changes of puberty. Hormone therapy will need to be lifelong.
- **Counselling** - to help the boy come to terms with his condition.
- **Educational support** - if necessary.
- **Speech therapy** - if necessary.
- **Frequent screening tests** - to ensure the early diagnosis of any associated complications (such as diabetes or breast cancer).
- **Reproductive technologies** - such as IVF, to help men with Klinefelter syndrome become fathers of their own biological children.

Where to get help

- Your doctor
- Klinefelter Syndrome Support Group Tel. (02) 9836 2970 or email: klinefeltersaus@hotmail.com
- XXY Support Group Tel. (08) 9279 6440
- Genetic Support Network Victoria Tel. (03) 8341 6315

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 and other characteristics such as slim build and development of breast tissue.
- The chromosomes are present in every cell of the body and the extra X chromosome cannot be removed.
- Treatment options include cosmetic surgery to remove enlarged breast tissue and lifelong testosterone therapy.

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

Genetic Health Services Victoria

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