

What is Klinefelter syndrome?

Klinefelter syndrome refers to a collection of characteristics in males that are caused by having two or more X chromosomes.

The collection of chromosomes in an organism's cells is known as its karyotype. The usual human karyotype is made up of 22 pairs of chromosomes called autosomes (which are the same in males and females) and a pair of sex chromosomes (resulting in a total of 46 chromosomes). Normally, women have 44 autosomes and two X chromosomes (denoted 46,XX) and men have 44 autosomes and one X and one Y chromosome (46,XY).

The most common karyotype associated with Klinefelter syndrome is 47,XXY, which occurs in 80-90% of males with the condition². This is why the condition is sometimes referred to as 'XXY syndrome'.

The number of males with Klinefelter syndrome is not known². About 1 in 100 male human embryos are 47,XXY, and 1-2 in 1000 newborn baby boys are diagnosed with the condition².

Symptoms of Klinefelter syndrome

The effects of Klinefelter syndrome vary between males with the condition¹. Symptoms of Klinefelter syndrome can be so mild in some individuals that they, their parents, and their doctors may not even realise they have the condition.

Severely affected baby boys may be born with:

- [Undescended testes](#)
- A smaller-than-normal penis
- Hypospadias.

Symptoms in childhood include:

- Small testicles
- Long legs and tall stature
- More body fat than normal
- Difficulties with speaking, learning, behavior, and socialising.

Boys with Klinefelter syndrome might start puberty but then it stops. They may have:

- Signs of [low testosterone](#), such as [gynaecomastia](#) (the growth of breast tissue)
- Reduced growth of the testes and penis
- A less masculine appearance (e.g. reduced facial and body hair growth, reduced muscle and bone development).

The effects of Klinefelter syndrome observed in children continue into adulthood.

For some men with Klinefelter syndrome, the condition goes undiagnosed until they try to start a family. This is because even for those with mild symptoms, most men with Klinefelter syndrome produce very few or no sperm and are [infertile](#).

Causes of Klinefelter syndrome

Even though Klinefelter syndrome is a genetic condition, it's not inherited. Rather, it results from failure of the sex chromosomes to separate during egg or sperm development².

The symptoms of Klinefelter syndrome stem from a lower-than-normal production of [testosterone](#), and the effects the extra X chromosome(s) have on the body's development and function.

Diagnosis of Klinefelter syndrome

Klinefelter syndrome is diagnosed by examining someone's karyotype, which is done using a small sample of blood or other tissue.

Treatment of Klinefelter syndrome

Infants and children with Klinefelter syndrome should be examined by their doctor at least every two years to monitor their physical development. They may need support from specialists to manage any problems with speech, learning, behaviour or psychiatric issues. Testosterone treatment might be prescribed for boys with Klinefelter syndrome who have a very small penis.

Monitoring the growth and [hormone](#) function of boys with Klinefelter syndrome in the lead-up to puberty helps guide decisions about testosterone treatment, which may become necessary.

Testosterone treatment is recommended to many adults with Klinefelter syndrome. If you have Klinefelter syndrome and are not receiving testosterone treatment, your hormonal function should be checked every 12 months.

Health effects of Klinefelter syndrome

In addition to its impact on development and fertility, Klinefelter syndrome is associated with many health problems, mainly due to reduced testosterone levels.

Men with Klinefelter syndrome are more likely than men without to have:

- Psychosexual and social problems
- Obesity
- Metabolic disease (e.g. type 2 [diabetes](#))
- [Cardiovascular disease](#)
- Some forms of cancer
- Autoimmune disease (e.g. multiple sclerosis, lupus)
- Poor vision

- Dental problems
- Blood clots.

Testosterone treatment to achieve levels like those in men without Klinefelter syndrome minimises the risk of health problems³.

If you have Klinefelter syndrome and want to start a family, it might be possible for a fertility specialist to help you by using assisted reproductive technology (ART).

What to do about Klinefelter syndrome

Although there's no cure for Klinefelter syndrome, there are things you can do to minimise the effects of the condition on your health and wellbeing, including:

- Monitoring your health and development
- Seeking appropriate care if necessary
- Maintaining a normal testosterone level.

If you're a parent of a child with symptoms of Klinefelter syndrome, a confirmed diagnosis will allow doctors to provide care that achieves the best possible outcomes for your child's health and wellbeing.

Similarly, if you're a man with symptoms of Klinefelter syndrome, an accurate diagnosis and ongoing specialist treatment managed by your doctor can help.

Questions to ask your doctor about Klinefelter syndrome

- What health specialists will I need to see to help me manage Klinefelter syndrome?
- What happens next if my child is suspected to have Klinefelter syndrome?
- Are there lifestyle changes I can make to help manage my Klinefelter syndrome?
- Are there any limits to my activity I should make?

References

[1] Wray et al., 2020. Balanitis. In: StatPearls Available from <https://www.ncbi.nlm.nih.gov/books/NBK537143/>

[2] Edwards, 1996. Balanitis and balanoposthitis: a review. Sexually Transmitted Infection

[3] Morris & Krieger, 2017. Penile Inflammatory Skin Disorders and the Preventive Role of Circumcision. International Journal of Preventive Medicine

[4] Charlton & Smith, 2019. Balanitis xerotica obliterans: a review of diagnosis and management. International Journal of Dermatology

[5] Pow-Sang et al., 2010. Epidemiology and Natural History of Penile Cancer. Urology

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