Can Turner Syndrome be cured or treated?

Klinefelter syndrome is a chromosomal disorder and hence there is no direct cure for the condition. However, a number of treatment approaches can help:

- **Testosterone replacement therapy**
  - Testosterone replacement therapy could help to boost the male hormone levels.
  - Your medical team will be monitoring the pubertal progression and testicular function of your boy. When suboptimal pubertal progression is noted, testosterone replacement therapy (monthly intramuscular injection or daily oral pills) will be initiated to stimulate the development of secondary sexual characteristics. It also helps with increasing the energy level, building up more muscle mass, as well as protecting against 'thin bones' (osteoporosis).
- **Reproductive technologies**
  - After puberty, your medical team can arrange semen analysis to check if there is any viable sperm in the ejaculates. These sperms can be frozen for future use. In boys with no viable sperm in the ejaculate, viable sperm may be found from a biopsy of the testes, which is an invasive procedure. In vitro fertilization is usually required whether sperms are found in the ejaculate or biopsy.
- **Developmental and Learning issues**
  - Most boys with Klinefelter syndrome have normal development, but some of them could have various degree of developmental issues and learning challaneges. They may need extra assistance in learning. With early diagnosis and timely assessment, it could facilitate early intervention strategies.

Boys with Klinefelter syndrome need ongoing medical care from a variety of specialists. Nevertheless, the condition is rather well-understood in the medical field and standardized surveillance protocol in taking care of this group of boys is available worldwide. With regular follow-ups and appropriate care, most boys with Klinefelter syndrome can lead happy, healthy, independent lives!

Should I proceed to confirmatory testing? And when?

Options of confirmatory tests

	Timing	Pros :)	Cons :(
Chorionic villus sampling (CVS)	11-14 weeks of pregnancy	Can be done earliest	Procedure-related risk of miscarriage 0.1-0.2% Chance of confined placental mosaicism, result may not represent the fetus
Amniocen-tesis	16-20 weeks of pregnancy	More accurate than CVS Can be done earlier than post-natal testing	Procedure-related risk of miscarriage 0.1-0.2%
Post-natal blood test	After birth, can use cord blood or baby's blood	Most accurate No risk of miscarriage	Only know about the condition after delivery

Which confirmatory test should I choose?

Your medical team will discuss with you on your best option based on the best available information. The decision will be affected by the following:

- Will the confirmatory test affect your immediate management during pregnancy?
- Any other special findings based on the morphology ultrasound that may warrant an invasive prenatal test?
- The medical issues and potential challenges ahead for boys with Klinefelter syndrome

Please contact your doctor for further discussion.

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Klinefelter syndrome

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What is Klinefelter syndrome?

Klinefelter syndrome is a common genetic condition occurring in males. It affects around 1 in every 500-1000 boys.

Klinefelter syndrome is caused by an additional X chromosome.

Usually, a person has 46 chromosomes in each cell, divided into 23 pairs, which includes two sex chromosomes. For boys, they typically have one X chromosome and one Y chromosome (or known as 46,XY). However, boys with Klinefelter syndrome have an additional X chromosome (47,XXY).

The extra X chromosome may either be carried in every cell in the body or it may only affect some cells (known as mosaic Klinefelter syndrome).

This condition is the result of a chromosomal abnormality and it is usually not inherited. Instead, it occurs as a random event during reproduction.

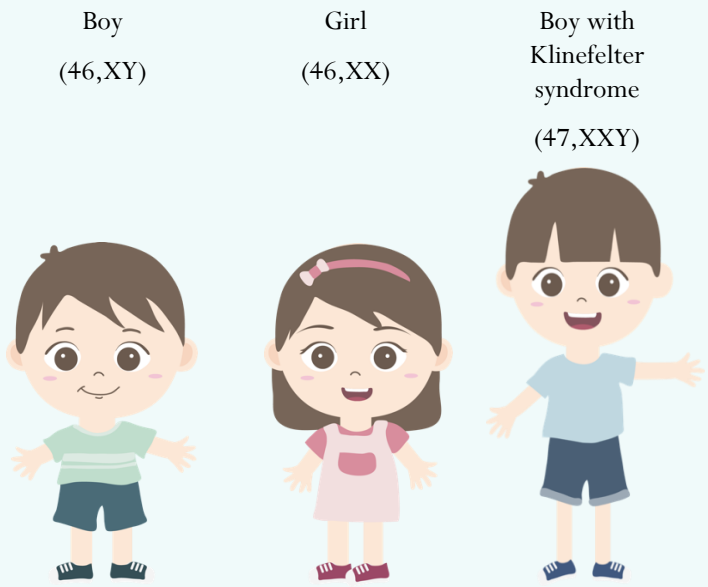
If you have a son with the condition, the chances of this happening again are very small.

How is Klinefelter syndrome diagnosed?

Klinefelter syndrome may be diagnosed before birth (antenatally), during childhood, at time of puberty, or in adulthood.

Klinefelter syndrome would be suspected based on certain unique clinical characteristics. However, these characteristics can be subtle, and many boys with Klinefelter syndrome do not realize they are affected. Boys born with the condition usually come to medical attention during puberty when they fail to complete puberty normally or when they develop breast enlargement (gynaecomastia). Some present with infertility when they grow up. Occasionally, it is detected incidentally from other investigations, e.g. non-invasive prenatal testing (NIPT) for fetal aneuploidy screening.

To confirm the diagnosis of Klinefelter syndrome, your medical team will order a test that looks at the chromosomes — a karyotype.



I was told that my baby may have Klinefelter syndrome from NIPT; how accurate is it?

A positive result from NIPT for 47,XXY indicates an approximately 70% chance that the fetus could actually have Klinefelter syndrome. To confirm this, you may consider to do an invasive prenatal procedure for karyotype.

What is the implication and what issues can happen?

Klinefelter syndrome can cause a variety of medical and developmental issues. Signs and symptoms of Klinefelter syndrome include:

- Incomplete puberty or suboptimal pubertal progression, related to reduced testicular function
- Small testicles
- Reduced facial or body hair
- Breast enlargement (gynecomastia)
- Reduced fertility or infertility (Klinefelter syndrome is the etiology of infertility in 10% of men without any sperm in the ejaculate and accounts for up to 2% of infertility)
- Developmental issues e.g. gross motor delay or motor clumsiness, delayed speech and language development
- Learning difficulties