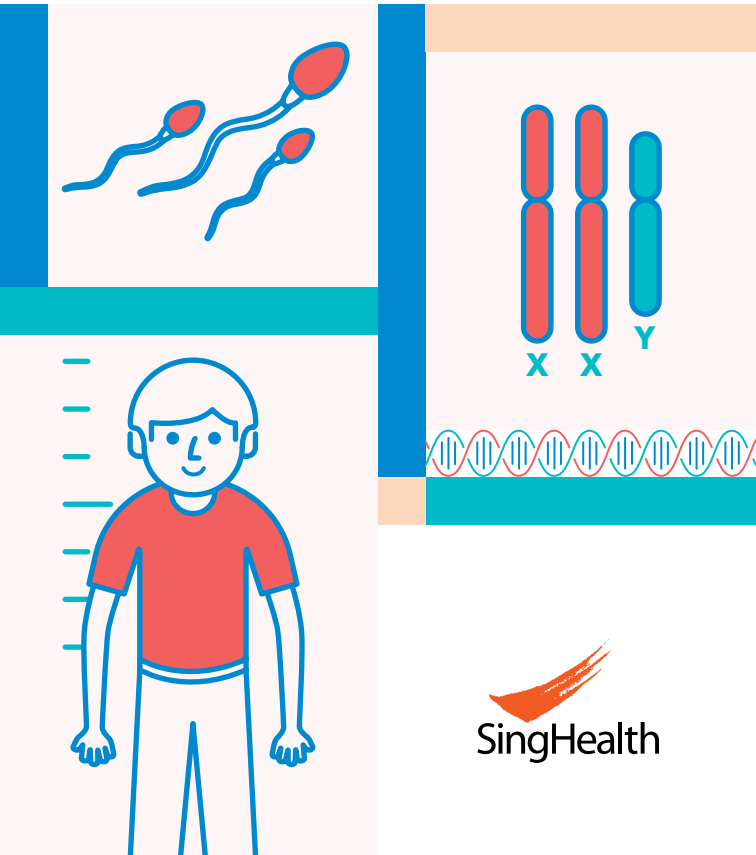
SingHealth DukeNUS
ACADEMIC MEDICAL CENTRE

Genomic
Medicine Centre

Klinefelter Syndrome

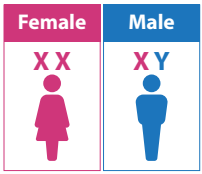


SingHealth

What is Klinefelter syndrome?

Klinefelter syndrome (KS) is a chromosomal condition that describes boys and men with common features, physical traits and medical conditions that are caused by the presence of an extra X chromosome (i.e., XXY).

Both males and females generally have 46 chromosomes. In females, this includes two X chromosomes, while males have one X and one Y chromosome.



How common is KS?

It is estimated that **one in 500 to 1,000 males** are born with KS.

What are the features of KS?

KS does not usually cause any obvious symptoms early in childhood, and many boys and men often do not realise they have KS until they go through puberty. In these instances, puberty may be late or incomplete. Sometimes, KS may only be diagnosed in adulthood due to infertility.

The features of KS can vary and it is important to note that not all the features described will be seen in your child.

Most boys with KS can expect to live long and healthy lives.

The typical features of KS in an adult (if untreated) are:

Growth. A tall and thin body with disproportionate long arms and legs.

Physical features. Breast tissue development (gynaecomastia) and female pattern of fat distribution. Reduced facial and body hair.

Small testicles. This feature is present in most people with KS.

Children and adolescents with KS may experience:

Learning difficulties.

- Delayed motor development
- Poor muscle tone
- Difficulties with speech and reading
- Lower attention span

Behavioural problems

In adulthood, in addition to the main typical features, untreated adults may have:

Hypogonadism. Individuals with KS often have this condition where men are unable to produce enough of the male hormone, testosterone, for the body's needs.

Testosterone is important for normal reproductive and sexual function, and physical changes that happen during male puberty such as development of the penis and testes, and facial and body hair. It also helps the growth of bone and muscles.

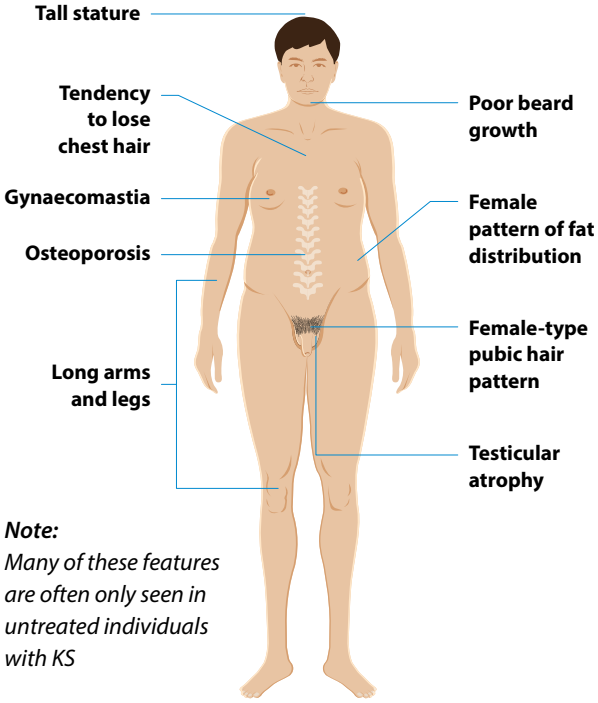
Hence, men with KS can have underdeveloped testes, low libido (sex drive), infertility and osteoporosis (thinning of the bones).

Almost all men with KS will be infertile (sterile), which means they will be unable to father a child without medical help.

Physical features. A physique which is less muscular than other men.

Intelligence. Most men with KS have normal intelligence, though some may have their intelligence affected.

Anxiety and depression



Note:
Many of these features are often only seen in untreated individuals with KS

Other medical concerns

The following problems can also be seen in individuals with KS, and need to be monitored for and treated if present:

- Cardiovascular diseases such as ischaemic heart disease, peripheral vascular disease and pulmonary embolism
- Metabolic syndromes such as dyslipidaemia and diabetes mellitus
- Breast cancer
- Thromboembolism (blood clots in blood vessels)
- Autoimmune diseases such as systemic lupus erythematosus, rheumatoid arthritis and Sjogren's syndrome

Can KS be cured?

While it is not possible to cure KS, much can be done to make sure your child has the best possible outcome.

How can KS be managed?

The multidisciplinary medical team looking after your child will be able to address specific medical concerns and routine KS-related healthcare issues.

Below are some related medical issues and how they can be managed:

Medical issue	Treatment
Developmental delay	Early intervention therapies
Hypogonadism	Testosterone replacement
Infertility	Assisted reproductive therapy

Testosterone therapy

Testosterone can be given in the form of injections or tablets. It is usually started from puberty and continued through adulthood.

Testosterone replacement can help to:

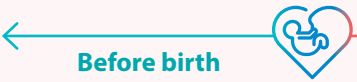
- Increase strength and build a more muscular body type
- Increase facial and pubic hair growth
- Increase libido (sex drive)
- Promote feelings of well-being

What causes KS?

KS occurs when an extra X chromosome is present before or soon after the time of conception. In most instances, this occurs spontaneously and is not inherited from either parent.

How is KS diagnosed?

The diagnosis of KS is made by looking at the complete set of chromosomes of the individual. This is called a karyotype. A karyotype shows the number and visual appearance of the chromosomes found in the cells of a person.



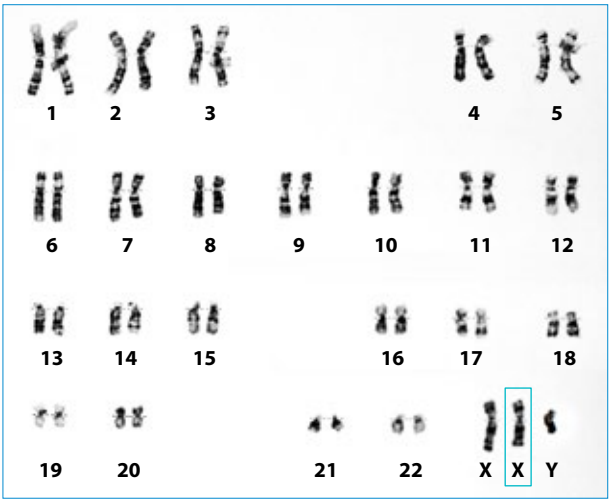
Before birth

Diagnosis of KS can be made by taking a sample of amniotic fluid, or other foetal tissue, to look at the foetal karyotype.

After birth

Diagnosis of KS is confirmed by taking a sample of blood or other tissue to obtain a karyotype.

Karyotype showing KS



How likely will I have another child with KS?

Recurrence risk is low (<1%). However, risk has been shown to increase with increasing maternal age.

KS is a lifelong condition. Should you require financial assistance or emotional support, please approach your doctor for referral to a medical social worker.

Support Group

Club Rainbow Singapore

Club Rainbow Singapore supports and empowers children with chronic illnesses and their families by providing relevant compassionate services in their journey.
Tel: 6377 1789
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www.clubrainbow.org

Acknowledgements

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