

The BSPED is one of the affiliated speciality groups of the Royal College of Paediatrics and Child Health.

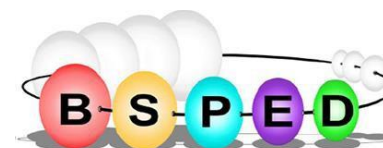
The society aims to improve the care of children and young people with endocrine disorders or diabetes mellitus, by bringing together professionals from a range of disciplines.

This leaflet has been written by members of the BSPED & reviewed by the Clinical Committee. It is designed to give you some general information about your child's condition and treatment. Your child's doctor or specialist nurse will be able to answer any further questions you have about your child.

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**British Society for
Paediatric Endocrinology
and Diabetes**

Klinefelter syndrome

Information for patients, parents and carers



Klinefelter syndrome

Klinefelter syndrome (KS) occurs when a boy is born with an extra X chromosome.

Chromosomes are structures in our cells that carry our genes. Genes are like a set of instructions for our body. Usually males have one X and one Y chromosome (XY) and females have two X chromosomes (XX).

Boys with KS have an extra X chromosome so giving them XXY.

Why does it happen?

The extra X chromosome comes from either the egg or the sperm that join to make the baby. It is just something that sometimes happens & there is nothing you could have done to prevent it.

How does it affect a child?

KS does not always have obvious effects and so many boys and men do not know they have the condition.

Boys may have some of the following features:

- Being later than others to reach milestones – such as sitting, crawling or walking.
- Starting to talk later than expected – the understanding of language is also often affected
- Sensitive or thoughtful personalities, and may be less confident than other boys their age

- Finding it difficult to pay attention - being easily distracted
- Normal range of IQ but may have difficulty learning and find it harder to process information quickly
- Starting puberty later than other boys
- Having a small penis and testes
- Developing breast tissue during puberty (gynaecomastia)

Those with fewer signs of KS are often not diagnosed until teenage years or adulthood.

The extra X chromosome can affect how well the testes work. The testes may grow slowly and produce less sperm and testosterone (the hormone that causes the changes seen in puberty).

Teenagers and adults with KS often have lower testosterone levels. They may not have enough to complete puberty. They may have little interest in sex and be unable to get an erection or have sexual intercourse.

Most adults with KS do not make enough sperm to be able to have children naturally.

How is it confirmed?

KS is confirmed by a blood test.

How often does this happen?

It is fairly common – about 1 in every 660 males has KS, only about 10% are diagnosed as children.

How is it treated?

It is not possible to get rid of the extra X chromosome so treatment is based on each young person's needs. If a boy's learning or behaviour is affected then it is important that this information is shared with nurseries, schools and colleges so the extra support can be put in place to allow the boy to achieve his full potential.

Treatment with testosterone will be given if testosterone levels are low. This is important for physical and emotional development.

Testosterone is usually given as an injection every 4 weeks, tablets or topical gel applied daily until the young adult is fully mature.

If a man with KS wants to have children, he will need to be seen by fertility specialist doctors for advice and treatment.

How long will this treatment be needed?

Testosterone is needed for all adult life. Testosterone is needed for physical development, sexual desire and function. It can make the young adult feel better and more positive. It is also needed for developing stronger bones and preventing osteoporosis.

Are there any long-term problems?

Generally men with KS have good health. There is a small risk of the following conditions, but these will be monitored for in clinic.

- Type 2 Diabetes
- Osteoporosis (weaker bones that break more easily)
- Heart disease and strokes
- Problems with blood vessels
- Anxiety & depression
- Auto-immune conditions (where the body's immune system attacks itself) such as under or overactive thyroid
- Breast cancer, lung cancer and lymphoma (very rare)

Suggested sites for further information

www.nhs.uk/conditions/klinefelters-syndrome/

www.ksa-uk.net

<https://ghr.nlm.nih.gov/condition/klinefelter-syndrome>