Klinefelter's 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 slower than others to reach milestones – such as late sitting, crawling or walking.
- Starting to talk later than expected – the understanding of language is also often affected
- Being shy with less confidence
- Finding it difficult to pay attention - being easily distracted
- They have the normal range of IQ but may have difficulty learning as they 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 are very small in childhood and start to grow as the boy starts puberty. The testes make testosterone (the hormone that causes the changes seen in puberty), and sperm.

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 very 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 boys 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 at 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 over active thyroid g
- 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

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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