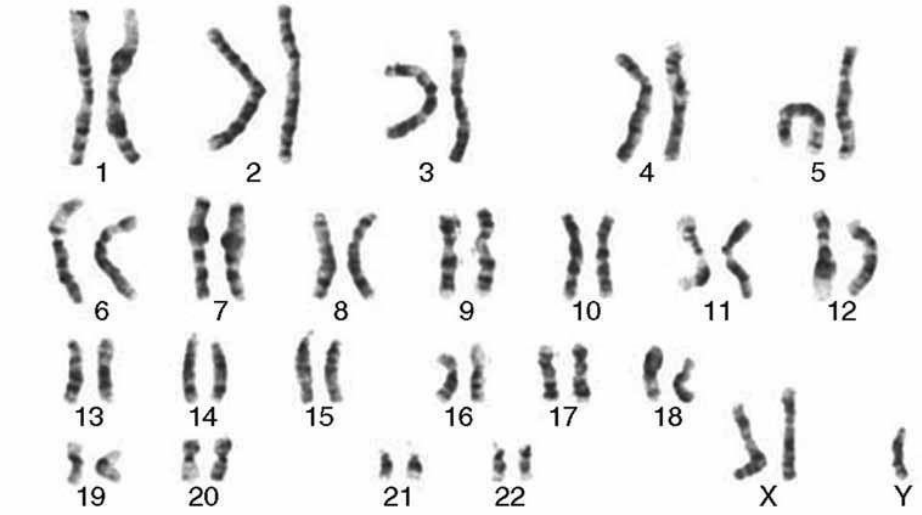
Saint Mary's Hospital
Manchester Centre for Genomic Medicine

Information for Patients

Klinefelter Syndrome (XXY)

Introduction

Humans are usually born with 46 chromosomes which are arranged in 23 pairs. One of these pairs determines whether a baby is male or female and these are known as the sex chromosomes. Boys are boys because they are born with the sex chromosomes XY and girls are girls because they are born with two X chromosomes (XX).



Occasionally, a boy is born with an extra X chromosome and this is known as Klinefelter Syndrome. The picture above is an enlarged photograph of chromosomes from someone with Klinefelter Syndrome.

About 1 in 1,000 boys has an extra X chromosome, but often they are unaware of it. In Britain, it is estimated that there are approximately 25,000 boys and men who have XXY. Even though these boys have an extra X sex chromosome, they are 100 percent male. They do, however, usually have fertility problems as they get older.

The condition is named after an American doctor called Dr Harry Klinefelter who first described the condition in 1942.

What causes Klinefelter Syndrome?

The vast majority of parents with a son with Klinefelter Syndrome have normal chromosomes themselves. The extra X chromosome is present due to a 'genetic mistake' occurring at the time of cell division to produce the egg or sperm that went to make that baby. In other words, the condition occurs by chance and the causes are not known. There is a slightly higher risk of the condition occurring in the children of older mothers.
This, however, only accounts for a minority of cases of boys with Klinefelter Syndrome.

Will it happen again?

This is very unlikely. Some parents do, however, opt to have a test in a future pregnancy to check the chromosomes of the baby. The need for testing and how this is carried out can be discussed at a genetics clinic.

How does it affect the child?

- ### Babies

Baby boys with Klinefelter Syndrome look completely normal at birth. Research has shown that they may be slightly smaller than babies with normal chromosomes. The genitals appear normal, although often one or both testicles will not have come down into the scrotum (an undescended testicle). This may need to be corrected by a minor operation in the first few years of life.

- ### Toddlers

Children with Klinefelter Syndrome usually sit, crawl and walk at the usual time, but they may be slower to learn to talk than other children. If this is a problem, then speech therapy will be helpful and can start when the child is between 3 and 4 years old.

- ### School children

Research has shown that the average intelligence (sometimes known as IQ) of boys with Klinefelter Syndrome is lower than that of boys with the usual number of chromosomes. In one study, the average IQ score was 98 for boys with Klinefelter compared with a value of 112 in the other boys. However, the range of IQ values in boys with Klinefelter Syndrome was 70 to 130, highlighting that IQ can vary between different individuals with Klinefelter Syndrome. Differences in IQ can be due to a variety of factors including the effect of hormone replacement treatment.

Boys with Klinefelter Syndrome tend to do less well at school than their brothers and sisters and may feel discouraged by this. It is important to try not to compare children within a family.

Approximately 75% of boys with Klinefelter have difficulty learning to read and will require special help on a one-to-one basis at school. This is possibly related to the particular difficulties that many of them have with short attention span, poor memory and difficulty in keeping things in the right order.

These types of problem are not unique to boys with Klinefelter Syndrome, but are found more often in this group of boys than in other children. They can be helped by patience and an appreciation that much repetition may be required in order to retain what has been learnt. In addition, boys with Klinefelter Syndrome may experience problems with the social side of school as they are often quite sensitive and may be moved to tears quite easily.

- ### Growth in Childhood and Adolescence

Boys with Klinefelter Syndrome often grow faster than usual, especially in the length of their legs. On average, a man with Klinefelter Syndrome will be about 186cm (6ft 2in) tall. If it looks as if a boy is going to be exceptionally tall, his growth can be slowed down by a course of injections. Such treatment is usually organised through a growth specialist, and needs to be commenced in plenty of time, before his height has become a problem. Muscle development in Klinefelter Syndrome is sometimes poor, but this can be improved with exercise. Approximately 75% of affected boys put on extra fat from about 7 years of age, particularly around the waistline, so it is important to be aware of this possibility and try to maintain a healthy diet to prevent this.

- ### Puberty

Puberty usually occurs at the normal time and usually progresses normally. However, in some cases the growth of the penis may slow down towards the end of puberty, and the facial, pubic and underarm hair may be quite sparse. This can be treated with supplements of the male hormone, testosterone, which are often given towards the end of puberty (15 to 16 years).

A mild degree of breast development (gynaecomastia) occurs in about two-thirds of boys with Klinefelter Syndrome. In the majority of boys it resolves without any treatment, but very rarely can persist and cause embarrassment. In such cases, the excess breast tissue can be removed.

Fertility

Men with Klinefelter Syndrome do not usually produce sperm, and so they have always been considered to be infertile, except in very rare cases. Many males with Klinefelter Syndrome are only diagnosed when they are found to be infertile, having been unaware of any problems before. The lack of sperm does not affect the ability to have sexual intercourse and the sex life of men with Klinefelter Syndrome is usually normal. Many men marry and couples are able to have children with the help of donor insemination, which can be arranged through a fertility clinic or assisted conception unit. Recently, new techniques of sperm retrieval and fertilisation have succeeded in finding living sperm in a few men with Klinefelter syndrome and they have been able to become fathers.

When should a boy with Klinefelter Syndrome be told about it?

Generally, it is considered best to begin to explain things in simple terms that the boy can understand from the age of about 10 or 11 years. Statements like 'the blood test showed a change in the cells that could mean you grow too tall and put on weight easily' can be used to explain the need for hospital visits and other tests. If testosterone supplements are required, then the explanation can include the fact that it will help the muscles to develop, as well as increase the size of the penis and scrotum.

The issue of fertility can be introduced at whatever time seems sensible. It is worth mentioning that, although he may not be able to have children, he will be able to have sexual intercourse like any other man.

For more information

If you need more advice about any aspect of Klinefelter Syndrome, you are welcome to contact:

Manchester Centre for Genomic Medicine
Sixth Floor
Saint Mary's Hospital
Oxford Road
Manchester
M13 9WL

Telephone: (0161) 276 6506 (Reception)
Facsimile: (0161) 276 6145

The department is staffed Monday–Friday, 9.00 am to 5.00 pm.

Website: www.mangen.co.uk

Further information and support is available from:

Klinefelter Syndrome Association
Sue Cook, National Co-ordinator
56 Little Yeldham Road
Little Yeldham
Halstead
Essex
CO9 4QT

Telephone: 0300 111 47 48
Website: www.ksa-uk.net

Please let us know if you would like this leaflet in another format (e.g. large print, Braille, audio).

The information in this leaflet was collated from various sources including the work of Dr Shirley Ratcliffe in the UK and the American Klinefelter Syndrome Association. Thanks also to the Kennedy-Galton Centre.

We would like to acknowledge our Clinical Genetics colleagues at Guy's and St Thomas' Hospital NHS Trust who originally wrote this leaflet.

Seen in clinic by (doctor): _____

And (Genetic Counsellor): _____

Telephone number: _____

Family reference number: _____