

This fact sheet describes the chromosome condition Klinefelter syndrome and includes the symptoms, cause and any treatment or testing which is available.

In summary

- **Klinefelter syndrome is also called XXY syndrome and only affects males**
- **The condition comes about because instead of males having the usual one copy of the X chromosome, they have two or more copies**
- **Klinefelter syndrome is thought to occur in 1 in every 500 or 1 in every 1000 males born in Australia each year.**

WHAT IS KLINEFELTER SYNDROME?

Klinefelter syndrome (also called XXY syndrome), was first described in 1942 by Dr Harry Klinefelter and is one of the more common chromosomal conditions affecting males. An additional X chromosome is found in the cells of these affected boys, giving them two X chromosomes instead of the usual one (see later).

A syndrome is a condition distinguished by a number of features that often occur together. There are a number of features that can be present in Klinefelter syndrome.

Some affected boys will have more features of the syndrome than others and there will be a difference in the degree of severity of the symptoms between affected boys. In some cases, a diagnosis of Klinefelter syndrome is not made until a boy approaches puberty and some men may never be diagnosed with the condition.

Features include:

- Learning difficulties which may be the greatest developmental challenge. IQ however, is in the normal range, although it may be 10-15 points lower than that of their siblings
- As children, affected boys are often quieter, less assertive and more passive than other boys and more so than their siblings
- They may be slower to develop language, motor (movement) skills and emotional maturity than their peers. This does not mean that they will not achieve these things, but will do so at a slower rate
- As boys with Klinefelter syndrome grow older, shyness, unassertiveness and relative immaturity may persist
- Problems with language, speech and verbal skills generally are the major learning problems for these boys and they may need some remedial intervention at school
- There is a tendency to grow at a slightly quicker rate than their peers so that their final height may be more than expected, but is usually within the normal range
- In around 50% of boys there is some growth of breast tissue at puberty (*gynaecomastia*). Plastic surgery may be considered by some men to reduce breast tissue
- Body hair, beard growth and testicular size are all reduced in males with Klinefelter syndrome. The use of the male hormone testosterone has been effective in increasing strength and developing a more masculine appearance. Treatment usually begins around the age of 11-12 years and can be given in the form of patches. Consultation with an endocrinologist (a hormone specialist) will provide further information about testosterone treatment
- Sexuality is normal although men with Klinefelter syndrome are unable to produce sperm and are usually infertile.

WHAT CAUSES KLINEFELTER SYNDROME?

As mentioned above, Klinefelter syndrome is caused by the presence of an extra copy of the X chromosome.

In each human cell, except the egg and sperm cells, there are 46 chromosomes. Chromosomes are found in pairs and each pair varies in size.

Thus there are 23 pairs of chromosomes, one of each pair being inherited from each parent.

- There are 22 numbered chromosomes from the largest to the smallest: i.e. 1-22. These are called autosomes
- There are also two sex chromosomes, called X and Y.

In females, cells in the body usually have 46 chromosomes (44 autosomes plus two copies of the X chromosome). They are said to have a 46,XX karyotype. Eggs (female reproductive cells) are different as they only contain half of the chromosomes (23 made up of 22 numbered chromosomes and an X chromosome).

In males, cells in the body usually have 46 chromosomes (44 autosomes plus an X and a Y chromosome). They are said to have a 46,XY karyotype. Sperm (male reproductive cells) are different as they only contain half of the chromosomes (23 made up of 22 numbered chromosomes and an X chromosome or a Y chromosome).

When egg and sperm cells are formed, the chromosome pairs separate so that there is only one of each pair in these cells i.e. 23 chromosomes instead of 46. A baby is conceived when the egg from the mother and the sperm from the father come together.

The baby would then have two copies of each chromosome (46 chromosomes in total) just like the parents. One copy of each chromosome would have come from the mother and one copy from the father. (see *Figure 39.1*)

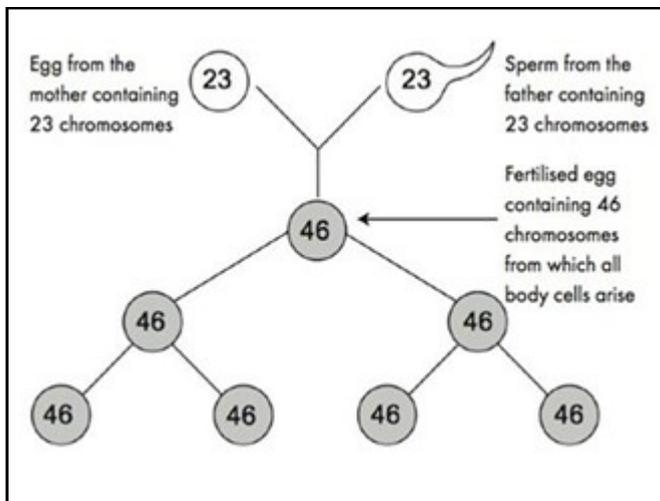


Figure 39.1: At conception the sperm and egg combine

Figure 39.2 shows a chromosome picture (karyotype) set from a male where each chromosome has been numbered from the largest (chromosome number 1) to the smallest (chromosome number 22) and arranged in pairs in order of size.

We know these chromosomes are from a male because of the X and Y.

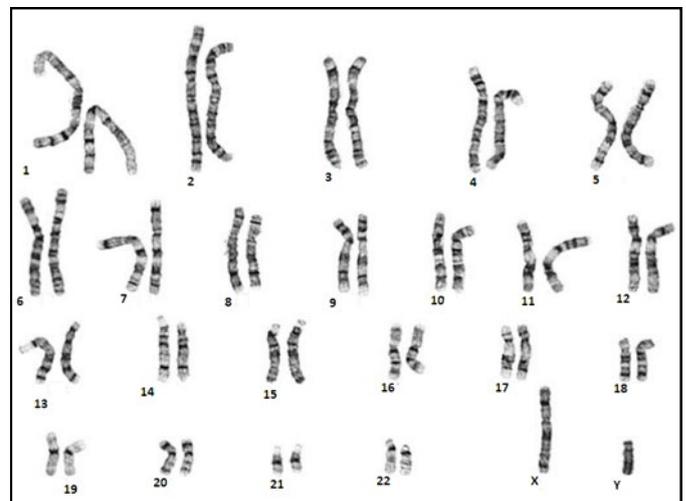


Figure 39.2: Chromosome picture (karyotype) from a male 46,XY

Sometimes, when the egg and sperm are forming, a mistake can occur so that the chromosome pairs do not separate in the usual way. The result is an egg or sperm cell that has only 22 chromosomes while others have 24 chromosomes.

If an egg or sperm carrying 24 chromosomes combines with an egg or sperm carrying the usual 23 chromosomes, the result would be an individual with cells in which there are 47 chromosomes instead of the usual 46. (See *Figure 39.3*)

There would be three copies of a particular chromosome in the cells rather than two. This is called **trisomy**.

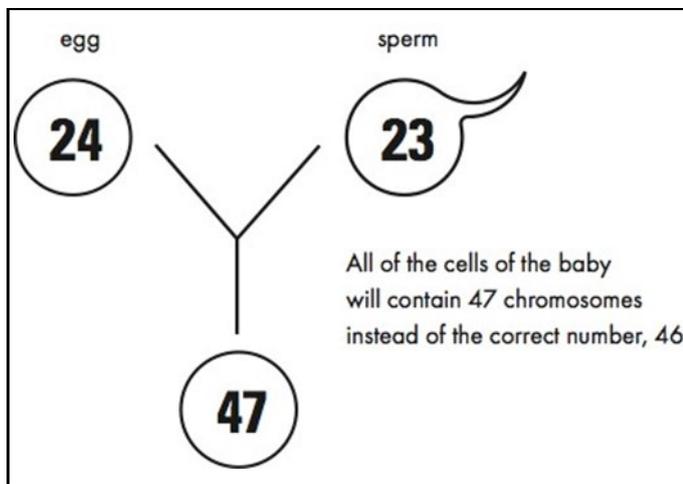


Figure 39.3: When the egg has 24 chromosomes, and the sperm has the usual 23, the baby's cells will contain 47 chromosomes instead of 46.

The chromosome pattern in males with Klinefelter syndrome includes an extra copy of the X chromosome. This extra chromosome causes an imbalance and is the reason why there are differences in males with this condition.

Figure 39.4 is a picture (karyotype) of the chromosomes from a male with Klinefelter syndrome.

HOW IS KLINEFELTER SYNDROME INHERITED?

In most cases a male with Klinefelter syndrome will be the first and only person affected by the condition in that family.

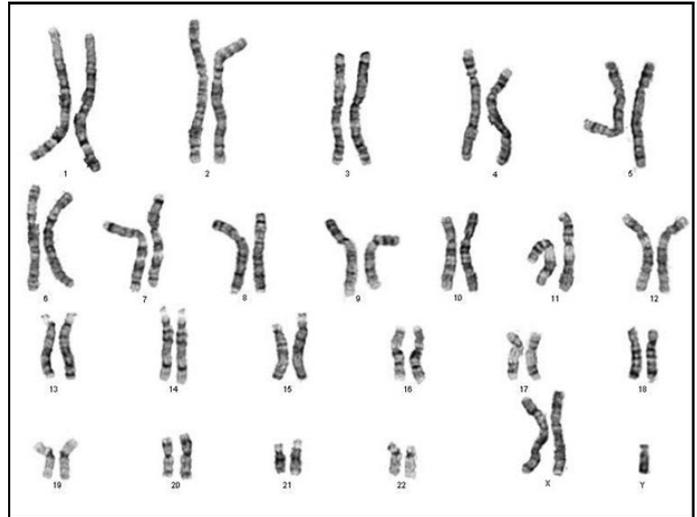


Figure 39.4: Chromosome picture (karyotype) from a male with Klinefelter syndrome. In this cell, there are 47 chromosomes including two copies of the X chromosome instead of one.

IS THERE ANY TESTING AVAILABLE FOR KLINEFELTER SYNDROME?

Chromosome testing can be done using a blood sample. A doctor may suspect a boy has the condition based on observing the characteristic features or symptoms associated with the syndrome.

Prenatal testing and PGD

Chromosome testing may be offered during pregnancy to some couples, especially for those women who are 35 years or older. The presence of an extra chromosome may be detected.

Prenatal tests can occur in a number of different ways and at certain stages of the pregnancy. In general, there are two main types of prenatal test – a screening test and a diagnostic test.

Screening tests give a risk or estimate of the probability that a baby has a health problem. These tests do not generally look directly at a sample from the developing baby and are therefore very safe. Included in the group of screening tests are ultrasounds, non-invasive prenatal testing, first trimester screening and second trimester screening.

Diagnostic tests provide a more accurate result since they are directly testing the baby. Because of this, in a very small number of cases, the test may also cause a miscarriage. Included in the group of diagnostic tests are ultrasounds, chorionic villus sampling (CVS) and amniocentesis.

Testing during pregnancy is optional and should be discussed in full with your doctor, midwife or genetic counsellor. Making a decision to have a test or not is always up to you.

It may also be possible to undergo pre-implantation genetic diagnosis (PGD) screening for chromosome conditions on an embryo created using in vitro fertilisation (IVF). These options are best discussed and considered before pregnancy, when possible, in order to ensure all possible risks, benefits and outcomes can be explored.