

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

### In summary

- **Turner syndrome is also called 45,X or XO syndrome and only affects females**
- **The condition comes about because instead of females having the usual two copies of the X chromosome, they have only one copy**
- **Turner syndrome is thought to occur in 1 in every 2000 girls born in Australia each year.**

### WHAT IS TURNER SYNDROME?

Turner syndrome (also known as 45,X syndrome) was first described in 1938 by Dr Henry Turner who noticed a pattern in some women who had decreased height and a lack of breast development, menstruation and sexual hair growth.

Some 20 years later, in 1959, it was discovered that women with this pattern of symptoms were missing all or part of an X chromosome.

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 Turner syndrome.

Some girls and women 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 females. In some cases, a diagnosis of Turner syndrome is not made until a girl approaches puberty.

The average intellectual performance of girls with Turner syndrome is within the normal range. Other features which may be present include:

- At birth, puffiness (*oedema*) of the back of the hands and top of the feet. This puffiness generally disappears within a couple of months but may persist on the feet for some years. Some girls (about 45%) may also have skin folds in the neck that are harmless and usually disappear soon after birth. In a few cases, however, there are more permanent skin folds that give the appearance of a broad neck or webbing on both sides of the neck

- Sucking and feeding problems due to a high-arched palate in the first months of life during which time regurgitation of food and vomiting are common
- Failure to grow at the expected rate occurs during the first few years. It is often during this period that the diagnosis of Turner syndrome may be made
- Without treatment, the average full height of a girl with Turner syndrome will be about 20 cm below the average for other women and will range from around 133-162 cm
- Chronic middle ear infections; a constriction or stenosis of the aorta, present in about 1 in 15 girls with Turner syndrome; heart valve abnormalities; emotional immaturity; kidney changes which generally do not cause any problems, and learning problems in the area of non-verbal or spatial learning such as mathematics
- Infertility; nearly all women with 45,X Turner syndrome have underdeveloped ovaries, which means that they do not spontaneously menstruate or develop secondary sexual characteristics such as developed breasts or sexual hair. About 5% of these women will menstruate but the period of fertility will be short and pregnancy is very rare. Women with other arrangements of the X chromosome can occasionally be fertile. The rate of miscarriage and birth abnormalities, however, is likely to be higher in the children of women with Turner syndrome than is the average.

### WHAT CAUSES TURNER SYNDROME?

As mentioned above, Turner syndrome is caused by a chromosome change where instead of two X chromosomes, girls are born with only one

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 40.1*).

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).

Sometimes, when the egg and sperm are forming, a mistake occurs so that the chromosome pairs do not separate in an ordered fashion.

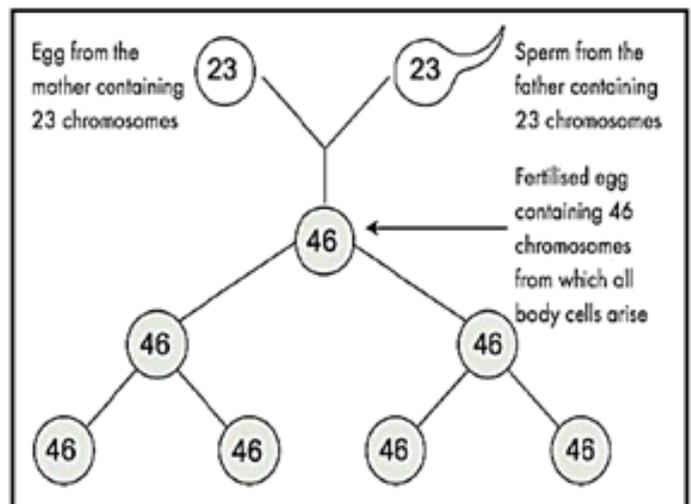
- The result is an egg or sperm cell that has only 22 chromosomes while others have 24 chromosomes.

If an egg or sperm carrying 22 chromosomes combines with an egg or sperm carrying the usual 23 chromosomes, the result would be an individual with cells in which there are 45 chromosomes instead of the usual 46 (See *Figure 40.2*).

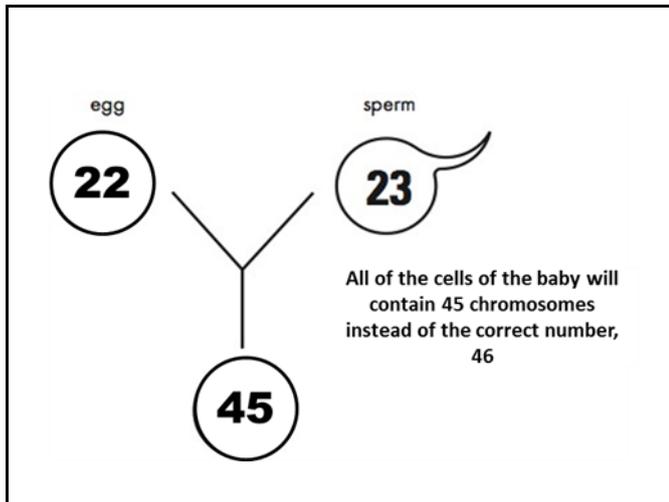
Thus there would be **only one** copy of a particular chromosome in the cells rather than the usual two copies. This is referred to as **monosomy**.

The chromosome pattern in females with Turner syndrome shows a missing copy of the X chromosome. This missing chromosome causes an imbalance and is the reason why there are differences in females with this condition.

*Figure 40.3* is a chromosome picture (karyotype) from a female with Turner syndrome.



*Figure 40.1: At conception the sperm and egg combine*



**Figure 40.2:** When the egg has 22 chromosomes, and the sperm has the usual 23, the baby's cells will contain 45 chromosomes instead of 46.



**Figure 40.3:** A chromosome picture (karyotype) from a woman with Turner syndrome. In this cell, the number of chromosomes is 45 with only one copy of the X chromosome (45,X) though other cells in her body may have had the correct chromosome number (46,XX).

### HOW IS TURNER SYNDROME INHERITED?

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

### IS THERE ANY TESTING AVAILABLE FOR TURNER SYNDROME?

Chromosome testing can be done using a blood sample. A doctor may suspect a baby 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 or missing 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, the tests also may cause a loss of the baby in a small number of cases. 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.