

## Important points

- Turner syndrome (also called XO syndrome) is a chromosomal condition where girls are missing one (or part of one) of their two X chromosome copies; they have a total of 45 chromosomes including one X chromosome (45,X), instead of the usual two copies (46,XX)
- Affects about 1 in every 2000 girls born in Australia
- Approximately 1.5% of all babies conceived have Turner syndrome but only 2% to 3% of these babies will survive to full term
- Girls with Turner syndrome have **normal intellectual development**. A range of other physical features may be present including failure to grow at the expected rate
- About 50% of women with Turner syndrome will have 45,X in all of their cells
- When there is a loss of the X chromosome in only **some** of the girl's cells due to a mistake in cell division occurring soon after conception, the chromosomes in the cells of their body may show two different patterns: some cells with 46 chromosomes and some with 45 ie. **mosaic**
- Mosaic Turner syndrome occurs in about 20% of cases
- About 30% of girls with Turner syndrome will have two copies of the X chromosome but with a number of possible re-arrangements of one of them
- The range and severity of symptoms depends on the number and distribution of cells containing the missing or re-arranged copy of the X chromosome
- Having a daughter with Turner syndrome does not appear to be related to either the age of the mother or the father at the time of conception
- There are a number of treatments that are aimed at reducing the impact of the symptoms of the condition
- Screening and diagnostic testing (where indicated) is available in pregnancy
- Decisions regarding screening and testing during pregnancy should only be made on an informed basis following counselling (see Genetics Fact Sheet 3)

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.

Turner syndrome is a condition that only affects girls:

- About 1 in every 2000 girls born in Australia are affected
- Approximately 1.5% of all babies conceived have Turner syndrome but only 2% to 3% of these babies will survive to full term

## Characteristics of Turner syndrome

A syndrome is a condition distinguished by a number of features that often occur together.

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.

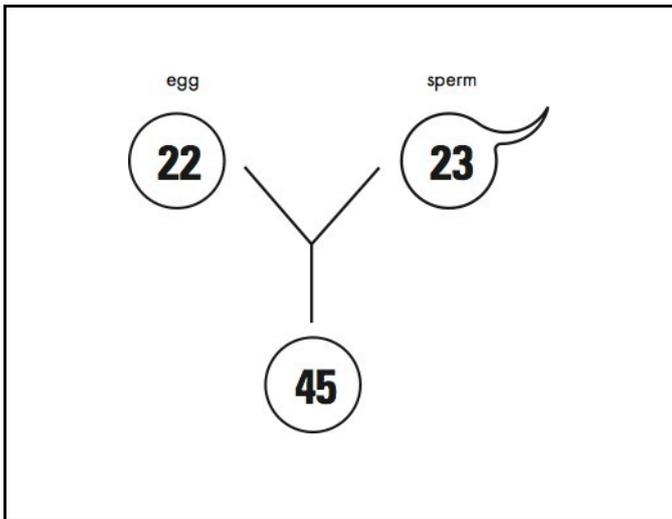
## Chromosomes

In each human cell, except the egg and sperm cells, there are 46 chromosomes, made up of 23 pairs (see Genetics Fact Sheet 1). There are

- 22 pairs of autosomes that scientists have numbered 1-22 according to their size from the largest to the smallest
- Two sex chromosomes: X and Y

When egg and sperm cells are formed, the chromosome pairs separate so that there is only one of each pair in these cells ie. 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.



**Figure 32.1:** When the egg has 22 chromosomes, and the sperm has the usual 23, the baby's cells will contain 45 chromosomes instead of 46. This may also happen in the reverse situation where the sperm has 22 and the egg has 23 chromosomes.



**Figure 32.2:** 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) (SEALS Genetics, Prince of Wales Hospital, Randwick).

One copy of each chromosome would have come from the mother and one copy from the father.

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 (Figure 32.1).

Thus there would be **only one** copy of a particular chromosome in the cells rather than the usual two copies (see Genetics Fact Sheet 6). This is referred to as **monosomy**.

### The chromosome pattern in Turner syndrome

In addition to 22 pairs of autosomes (chromosomes that are numbered 1-22), males usually have two **sex chromosomes**: one X chromosome copy that they receive from their mother and a Y chromosome copy that they receive from their father.

Every egg has an X chromosome and every sperm has either an X or a Y chromosome.

- If the sperm donates an X chromosome at conception, the baby will be a girl who will then have two X chromosomes, one from the mother and one from the father. The chromosome pattern is described as 46,XX
- If the sperm donates a Y chromosome, the baby will be a boy, with one X chromosome from the mother and a Y chromosome from father. The chromosome pattern is described as 46,XY

At the time of conception or in the period very soon afterwards, when the fertilised egg is dividing to form new cells in the body, a mistake can occur in which one X chromosome is lost, leaving 45 chromosomes including one X chromosome.

This is called Turner syndrome and is written as 45,X.

- Figure 32.2 shows a picture of the chromosomes from a woman with Turner syndrome (45,X)
- About 50% of females with Turner syndrome will have this chromosome pattern in all of their cells
- When a mistake in cell division occurs soon after conception, the chromosomes in the cells of the girls may show two different patterns. This is called **chromosomal mosaicism**
- Some females with Turner syndrome have some cells with the usual 46,XX pattern and other cells will have 45,X. These individuals are said to be mosaic for Turner syndrome (see Genetics Fact Sheet 6)
- About 30% of females with Turner syndrome will have two copies of the X chromosome (46,XX) but with a number of possible re-arrangements of the second X chromosome. These variations can lead to varying symptoms of Turner syndrome and include having
  - Missing (*deleted*) portions of the short ('p'), or the long ('q') arm of the X chromosome
  - One of the X chromosomes arranged in a ring form
  - The two long ('q') arms of the X chromosomes joined together in an arrangement called *isochromosome Xq*
  - Very rarely, cells that contain part of the Y chromosome may be present

### Who is at risk of having a girl with Turner syndrome?

Turner syndrome results from an error during the division of the sex chromosomes in either the egg or the sperm. Such an event is said to occur sporadically (spontaneously for unknown reasons) and it is highly unlikely to affect further children.

Having a girl with Turner syndrome does not appear to be related to either the age of the mother or the father at the time of conception.

## Can Turner syndrome be inherited?

Girls with Turner syndrome are almost invariably born to women who themselves have a normal chromosome pattern.

## Is there any treatment for Turner syndrome?

The X chromosomes carry many important genes, including genes for the development of ovaries, sex hormone production, sexual development and height. The loss of the X chromosome in some or most cells explains the range of features of the condition.

There is no cure for Turner syndrome. There are, however, a number of treatments that are aimed at reducing the impact of the symptoms of the condition.

Cardiac and hearing assessments need to be regularly made for girls with Turner syndrome, approximately every two or three years. In some cases skin folds that give the appearance of a broad neck or webbing on both sides of the neck persist into childhood. The folds can be surgically corrected by a plastic surgeon. Scarring can occur due to surgery. The decision to operate needs to be carefully considered.

Girls with Turner syndrome do not grow at the same rate as girls who do not have this condition. They also do not achieve the same final height as other girls. Treatment with growth hormone from an early age until around the time of puberty, has been shown to improve the final height to around the lowest limits of normal for other girls. There will be a variation in the final height of girls with Turner syndrome as there is for all people, as height of individuals is very much determined by the height of their parents.

At around the time of puberty, the growth hormone treatment is stopped and hence height development also stops. Female sex hormones are introduced over a period of time and breast and sexual hair development occur. Menstruation will occur but this is not an indication of fertility.

Most women with Turner syndrome who have had children have a **mosaic Turner** pattern in their chromosomes ie. they have two types of cells including some normal 46,XX cells.

Other women with Turner syndrome have achieved a pregnancy using assisted reproductive technologies (ART) such as *in vitro* fertilisation (IVF).

Girls with Turner syndrome who have part of the Y chromosome in their chromosome pattern, may be more susceptible to developing tumours in their unformed ovarian tissue. Often removal of this tissue is recommended for these girls.

While at school, girls with Turner syndrome may need some remedial help with mathematics and other spatial problems. Psychosocial development may be assisted by encouraging girls to participate in groups and in other social activities.

## Can Turner syndrome be diagnosed before the baby is born?

There are several prenatal screening and diagnostic tests that can be done during pregnancy to determine if the baby is at risk of having, or definitely has Turner syndrome.

- Genetics Fact Sheet 17 provides a summary of the most common prenatal diagnostic and screening tests available
- The prenatal screening tests are detailed in Genetics Fact Sheet 17B and the prenatal diagnostic tests in Genetics Fact Sheet 17C

In addition, preimplantation genetic diagnosis (PGD) allows for testing for Turner syndrome on an embryo that has been created using assisted reproductive technology (ART) such as *in vitro* fertilisation (IVF). If the embryo does not have the condition, it is transferred to the uterus and allowed to develop normally (see Genetics Fact Sheet 18).

Any consideration of testing before or during pregnancy should only be made on an informed basis following counselling (see Genetics Fact Sheet 3).

## Other Genetics Fact Sheets referred to in this Fact Sheet: 1, 3, 6, 17, 17B, 17C, 18