

This fact sheet talks about the chromosome condition trisomy 18 and includes the symptoms, cause, treatment and available testing.



IN SUMMARY

- Trisomy 18 is a chromosome condition also known as Edwards syndrome
- Babies with trisomy 18 usually have distinctive features, severe learning disability and other physical developmental concerns
- Trisomy 18 is caused by having an extra copy of chromosome number 18.

WHAT IS TRISOMY 18?

Trisomy 18 is also known as Edwards syndrome. It is a condition which is considered very serious and most babies with trisomy 18 do not survive to birth.

Some general signs and symptoms include:

- Developmental delays and severe learning disability
- Slow to grow and gain weight and severe feeding difficulties
- Low muscle tone (floppy) and episodes where breathing may stop
- A prominent back part of the head, low-set, differently shaped ears, an unusually small jaw, a small mouth with an unusually narrow roof, an upturned nose, narrow eyelid folds, widely spaced eyes, and drooping of the upper eyelids and undescended testes in boys
- Unusually developed hands and feet which may include overlapping fingers and clenched fist, webbing of the toes, a deformity causing the heels to turn inwards and the soles flexed (clubfeet)
- A small pelvis with limited hip movement and a short breastbone

- Kidney differences and structural heart changes at birth such as an abnormal opening in the partition dividing the lower chambers of the heart
- Heart differences and respiratory difficulties may lead to potential life-threatening complications during infancy or childhood.

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

There are therefore 23 pairs of chromosomes, one of each pair being inherited from each parent.

- There are 22 numbered chromosomes from roughly 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 typically 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 typically 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). *Figure 38.1* shows a chromosome picture (karyotype) from a typical male (46,XY). The usual way a sperm and egg combine at conception is shown in *Figure 38.2*.

Figure 38.1:
Chromosome picture (karyotype) from a male (46,XY).

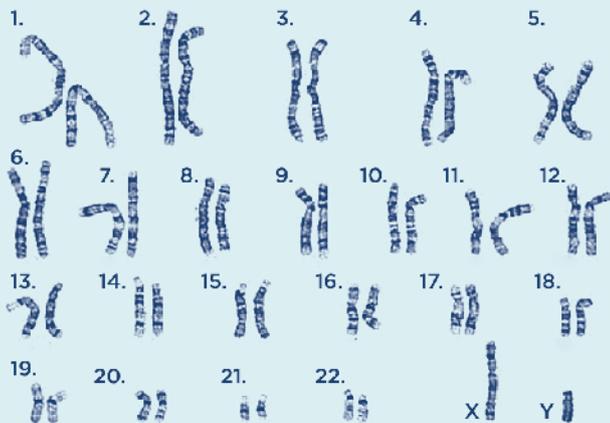
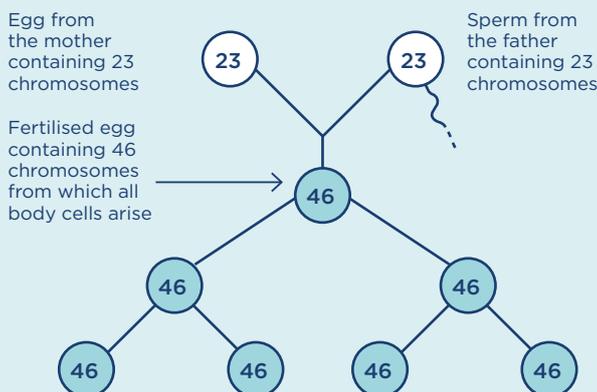


Figure 38.2:
At conception the sperm and egg combine



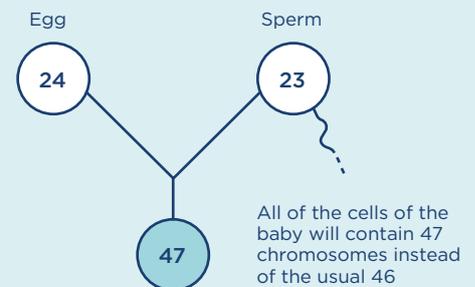
If an egg or sperm carrying 24 chromosomes combines with an egg or sperm carrying the usual 23 chromosomes, the result would be a person with 47 chromosomes instead of the usual 46 (See *Figure 38.3*).

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

The chromosome pattern in people with trisomy 18 includes a whole extra copy of chromosome number 18. This extra chromosome causes an imbalance and is the reason why there are differences in people with 3 copies of chromosome 18 material compared with people with the usual pair.

The presence of the extra copy of chromosome 18 causes the learning disability and physical features of trisomy 18. People with this condition usually have three whole copies of chromosome number 18, i.e. 47 chromosomes in their cells instead of 46. **Trisomy** means three bodies.

Figure 38.3:
When the egg has 24 chromosomes, and the sperm has the usual 23, the baby's cells will contain 47 chromosomes instead of 46.



WHAT CAUSES TRISOMY 18?

Trisomy 18 is caused by the presence of an extra full (or partial) copy of chromosome number 18.

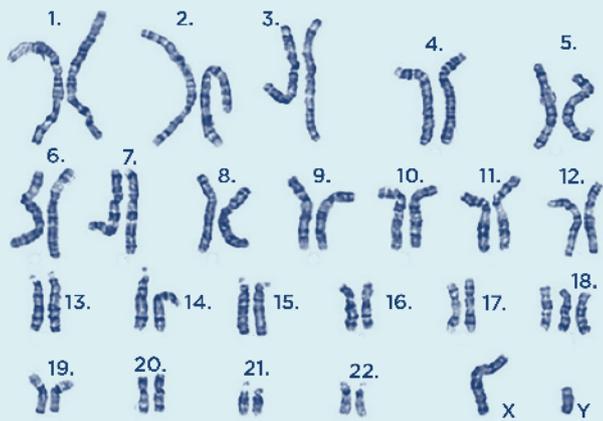
Sometimes, when the egg and sperm are forming, the chromosome pairs do not separate in the usual way. The result is an egg or sperm cell that has only 22 chromosomes while others have 24 chromosomes.

Some people have trisomy 18 as a result of a chromosome rearrangement

Rarely, a chromosome translocation happens when a section of chromosome 18 is rearranged with sections of another chromosome so there is too much of only a part of chromosome 18 (partial trisomy). Signs and symptoms may be different from those found in full trisomy 18. Sometimes this can be inherited from a parent even if the parent does not have a health or developmental concern.

Figure 38.4:

Chromosome picture (karyotype) from a male with trisomy 18 (47,XY+18). In this cell, there are 47 chromosomes including three copies of chromosome 18 instead of the usual two.



Some people have trisomy 18 as a result of a mosaicism

Most people have the same chromosome makeup in all the cells in their body. People with trisomy 18 as a result of mosaicism have some cells in the body with the usual two copies of chromosome 18, and other cells with three copies of chromosome 18. Someone who is mosaic for a chromosome change therefore has a mixture of cells in their body. Although, signs and symptoms tend to be milder in those with a lower proportion of cells with trisomy, it may be difficult to predict how signs and symptoms will show up, just from a blood test.

Figure 38.4 is a picture (karyotype) of the chromosomes from a male with trisomy 18 (47,XY+18).

HOW IS TRISOMY 18 INHERITED?

In most cases where trisomy 18 is caused by a complete extra copy of chromosome 18, that person will be the first and only person with the condition in that family. This is also the case where trisomy 18 is the result of mosaicism.

It is usually assumed that if the parents of a person with trisomy 18 have the usual two copies of chromosome 18, then the extra 18 in their child was a result of an egg or sperm with 24 instead of 23 chromosomes.

As mothers get older, errors in chromosome number are more likely to happen in their eggs. For parents who have a child with a translocation form of trisomy 18, there may be more tests needed to check if the chromosome rearrangement in the child has happened as a new change or not. Depending on the results from additional tests (in the child and parents), the chance of another child having trisomy 18 can vary.

IS THERE ANY TESTING AVAILABLE FOR TRISOMY 18?

Chromosome testing of a baby who is suspected of having trisomy 18 can be done using a blood sample. A doctor may suspect a baby has the condition based on seeing the characteristic features or symptoms linked with the condition.

Testing for pregnancy

Testing for trisomy 18 may be offered during pregnancy.

Prenatal tests can happen 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 chance that a baby has a health condition. 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 generally directly testing the baby. Because of this, in a very small number of cases, a 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 talked about 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 have pre-implantation genetic diagnosis (PGD) to look for trisomy 18 in an embryo made using in vitro fertilisation (IVF). When planning a family, options for testing are best talked about and considered before pregnancy.

More support and information is available for individuals and families through support organisations including Genetic Alliance Australia.