

This fact sheet describes the chromosome condition trisomy 18 and includes the symptoms, cause and any treatment or testing which is available.

In summary

- Trisomy 18 is a chromosome condition also known as Edwards syndrome
- Babies with trisomy 18 usually have distinctive features, severe intellectual disability and other physical problems
- 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.

The combination of features present in babies affected with trisomy 18 can lead to many different developmental problems.

Some general symptoms include:

- Failure to grow and gain weight at the expected rate and severe feeding difficulties
- Diminished muscle tone and episodes in which there is temporary cessation of spontaneous breathing
- A prominent back portion of the head, low-set, malformed ears, an abnormally 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
- Developmental delays and intellectual disability
- Abnormalities in the bones of the hands and feet which may include overlapping flexed fingers, 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 abnormalities and structural heart (cardiac) defects at birth such as an abnormal opening in the partition dividing the lower chambers of the heart.
- These congenital heart defects and respiratory difficulties may lead to potential life-threatening complications during infancy or childhood.

WHAT CAUSES TRISOMY 18?

As mentioned above, trisomy 18 is caused by the presence of an extra copy of chromosome number 18.

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 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 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 38.1*).

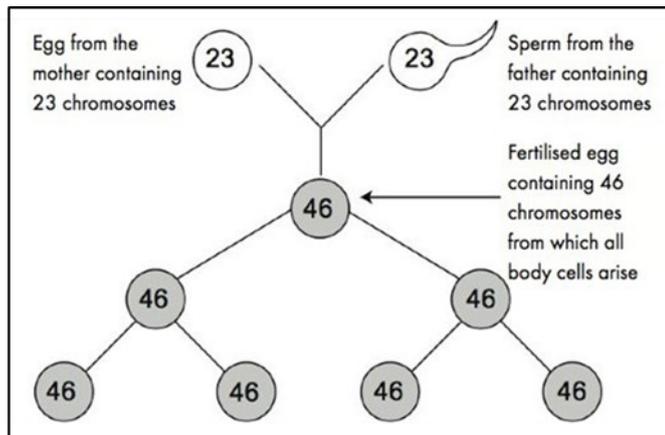


Figure 38.1: At conception the sperm and egg combine

Figure 38.2 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.

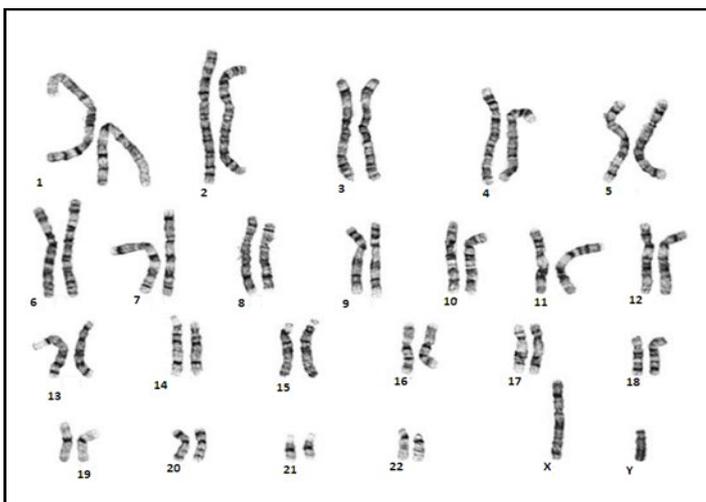


Figure 38.2: Chromosome picture (karyotype) from a male 46,XY.

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

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 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 an 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 and those with the usual 2 copies of chromosome 18.

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

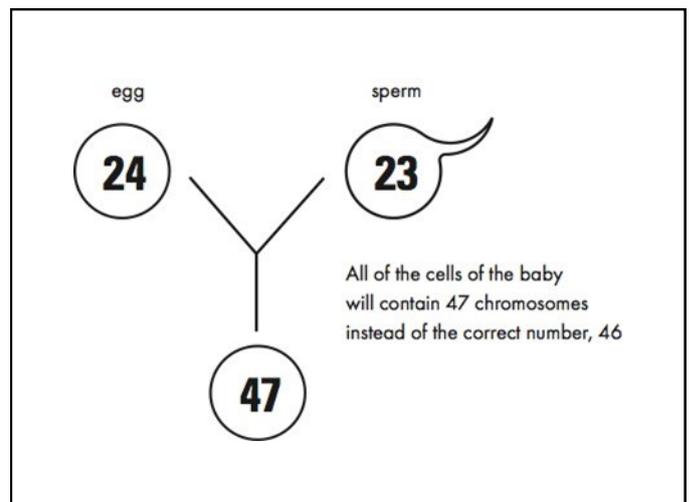


Figure 38.3: When the egg has 24 chromosomes, and the sperm has the usual 23, the baby's cells will contain 47 chromosomes instead of 46.

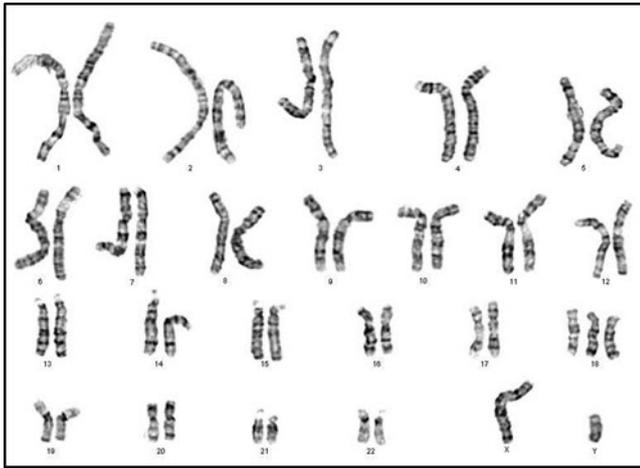


Figure 38.4: Chromosome picture (karyotype) from a male with trisomy 18. In this cell, there are 47 chromosomes including three copies of chromosome 18 instead of the usual two.

Figure 38.4 is a picture (karyotype) of the chromosomes from a male with trisomy 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 affected by the condition in that family. 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 being fertilised.

Older mothers have a higher chance of having these errors.

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 observing the characteristic features or symptoms associated with the syndrome.

Prenatal testing and PGD

Testing for trisomy 18 may be offered during pregnancy to some couples, especially for those women who are 35 years or older.

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 trisomy 18 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.