

### Important points

- Trisomy 18 (also known as **Edward syndrome**) is a chromosomal condition in which all or a part of chromosome 18 is present in the cells of the body three times (*trisomy*) rather than the usual two times
- The syndrome impacts severely upon intellectual and physical development
  - Appears to affect females three to four times more frequently than males
  - Occurs in about 1 in 5,000 to 1 in 7,000 live births
  - About 1% of all recognised miscarriages occur in association with trisomy 18
- Of all babies born with the extra copy of chromosome 18, 50% do not survive one week of age and 90% do not survive past the first year of life
- The chromosomal problem in trisomy 18 is due to an egg cell or, rarely, a sperm cell that is formed containing one copy of each chromosome and an extra copy of chromosome 18, ie. 24 chromosomes rather than 23. When that egg combines with sperm containing the usual 23 chromosomes, the baby conceived has 47 chromosomes in the cell of their body rather than the usual 46
- When there are three copies of chromosome number 18 in **all** of the baby's cells, the condition is referred to as trisomy 18. The presence of the extra chromosome causes the mental and physical characteristics of the syndrome
- When the extra copy of chromosome 18 is only in **some** of the baby's cells due to a mistake in cell division occurring soon after conception, the chromosomes in the cells of the baby may show two different patterns: some cells with 46 chromosomes and some with 47: mosaic trisomy 18. The range and severity of symptoms depends on the number and distribution of cells containing the extra copy of chromosome 18. Mosaicism is discussed in more detail in Genetics Fact Sheet 13
- The chance for having a child with trisomy 18 increases with the mother's age
  - If a woman has had a child with trisomy 18 there may be a small additional increase in risk over her age risk for having another child with the condition
- Screening and diagnostic testing (where indicated) for trisomy 18 is available in pregnancy
- It is highly recommended that a pregnancy be made on an informed basis following counselling (see Genetics Fact Sheet 3)

Trisomy 18 (also known as **Edward syndrome**) is a chromosomal condition in which all or a part of chromosome 18 appears three times (*trisomy*) rather than twice in cells of the body (see later).

The syndrome

- Appears to affect females 3-4 times more frequently than males
- Occurs in about 1 in 5,000 to 1 in 7,000 live births
- About 1% of all recognised miscarriages occur in association with trisomy 18

The survival of infants born with trisomy 18 will depend on how severely they are affected. Studies conducted since the mid-1980s indicate that of all babies born with trisomy 18

- 50% died by one week of age
- 90% did not survive past the first year of life

Therefore while less than 10% survive the first year of life, some children live well past the first year.

### Characteristics of trisomy 18

A syndrome is a condition distinguished by a number of features that often occur together. The combination of features present in babies affected with trisomy 18 can lead to many different developmental and physical problems.

Symptoms may 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
- Developmental delays and intellectual disability
- 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
- 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 and a persistence of the fetal opening between the two major arteries (*aorta, pulmonary artery*) emerging from the heart
- These congenital heart defects and respiratory difficulties may lead to potential life-threatening complications during infancy or childhood

**The 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 chromosomes that 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 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.

When the egg and sperm are forming, a mistake may occur 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 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 (Figure 30.1).

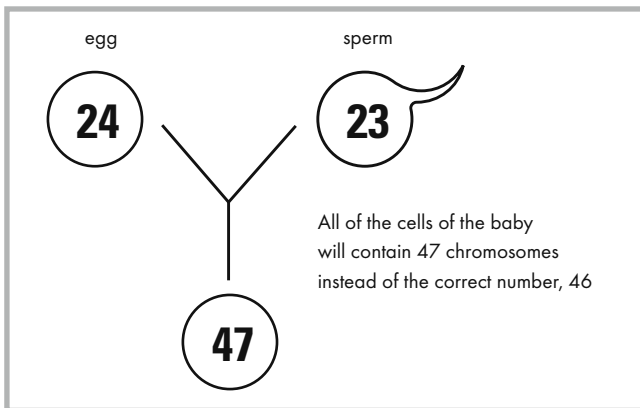
Thus there would be **three** copies of a particular chromosome in the cells rather than the usual two copies (see Genetics Fact Sheet 6). This is called **trisomy**.

**The chromosome pattern in trisomy 18**

When there are three copies of chromosome number 18 in the cells, the condition is referred to as trisomy 18. The presence of the extra chromosome causes the mental and physical characteristics of the syndrome.

Figure 30.2 is a picture (karyotype) of the chromosomes from a male with trisomy 18.

In some cases the mistake in the distribution of chromosomes in cell division occurs **after fertilisation of the egg by the sperm**, so the chromosomes in the cells of the baby may show two different patterns.



**Figure 30.1:** When the egg has 24 chromosomes, and the sperm has the usual 23, the baby’s cells will contain 47 chromosomes instead of 46. This may also happen in the reverse situation.

- This is called **chromosomal mosaicism** and means that some babies who have trisomy 18 have some of their body cells containing 47 chromosomes because of an extra copy of chromosome 18, while other cells in their body have the usual 46 chromosomes
- The baby is said to be mosaic for trisomy 18 (see Genetics Fact Sheet 13)

In individuals with mosaic trisomy 18, the range and severity of symptoms may depend on the percentage of cells containing the extra copy of chromosome 18.

In general, having all cells in the body containing the extra copy of chromosome 18 means that the symptoms will be more severe.

**Can a baby with trisomy 18 be cured?**

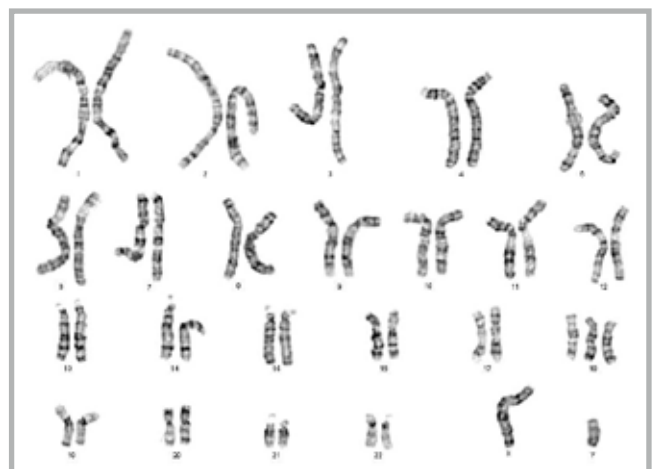
There is no cure for trisomy 18. Care is usually directed at making babies as comfortable as possible.

In individuals with mosaic trisomy 18, treatment is directed toward the specific symptoms that are apparent. Such treatment may require the coordinated efforts of a multidisciplinary team of medical professionals. In some cases, recommended treatment may include surgical correction of certain abnormalities associated with trisomy 18. The surgical procedures performed will depend upon the nature and severity of the abnormalities, their associated symptoms, and other factors.

**Who is at risk of having a child with trisomy 18?**

The extra copy of chromosome 18 can come from either the egg or the sperm. It has, however, been shown that a woman who is over the age of 35 is at slightly greater risk of having a child with trisomy 18, or any chromosomal abnormality, similar to the increased risks for Down syndrome (see Genetics Fact Sheets 6 & 28).

It is estimated that one out of every 3-4 fertilised eggs are chromosomally abnormal and this increases with the mother’s age. Therefore, most people have had at some time, a chromosomally



**Figure 30.2:** Chromosome picture (karyotype) from a boy with trisomy 18. In this cell, there are 47 chromosomes including three copies of chromosome 18 instead of the usual two. Other cells in this boy’s body may have the right chromosome number, 46 (SEALS Genetics, Prince of Wales Hospital, Randwick).

abnormal conception which may have miscarried or not even been recognised as a pregnancy because the miscarriage occurred very early.

### Can trisomy 18 be diagnosed before the baby is born?

There may be a number of indications that there is an increased risk for the baby having trisomy 18 including

- The mother's age
- A family history of trisomy 18
- The results of a screening test for this condition in pregnancy

There are several prenatal screening and diagnostic tests that can be done during pregnancy to determine if the baby is at risk of or definitely has trisomy 18. 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.

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

In addition, preimplantation genetic diagnosis (PGD) involves testing for trisomy 18 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 to try and achieve a pregnancy (see Genetics Fact Sheet 18).

**Other Genetics Fact Sheets referred to in this Fact Sheet: 1, 3, 6, 13, 17, 17B, 17C, 18, 28**

### Information in this Fact Sheet is sourced from:

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