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Important points

- Trisomy 13 (also known as **Patau syndrome**) is a chromosomal condition in which there are three copies instead of the usual two copies of all, or a part of chromosome 13 in the cells of the body
- Trisomy 13 severely impacts on intellectual and physical development
 - Appears to affect females slightly more frequently than males
 - Occurs in about 1 in 5,000 to 1 in 12,000 live births
 - About 1% of all recognised miscarriages occur in association with trisomy 13
- Of all babies born with the extra copy of chromosome 13 in all the cells of their body, around 50% die in the first month, and the rest within the first year
- The chromosomal problem in trisomy 13 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 13, 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 13 in **all** of the baby's cells, the condition is referred to as trisomy 13
- When the extra copy of chromosome 13 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 13. The range and severity of symptoms depends on the number and distribution of cells containing the extra copy of chromosome 13
- The chance of having a child with trisomy 13 increases with the mother's age
 - If a woman has had a child with trisomy 13 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 13 is available in pregnancy
- Decisions regarding screening and testing during pregnancy should only be made on an informed basis following appropriate counselling (see Genetics Fact Sheet 3)

Trisomy 13 (also known as **Patau syndrome**) is a chromosomal condition in which all or a part of chromosome 13 appears three times (*trisomy*) rather than twice in cells of the body (see later). The syndrome:

- Appears to affect females slightly more frequently than males
- Occurs in about 1 in 5,000 to 1 in 12,000 live births
- Of all babies born with the extra copy of chromosome 13 in all the cells of their body, around 50% die in the first month, and the rest within the first year
- About 1% of all recognised miscarriages occur in association with trisomy 13

Characteristics of trisomy 13

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 13 can lead to many different developmental problems.

Some general symptoms may include:

- Developmental delays, profound intellectual disability, unusually small eyes, an abnormal groove or split in the upper lip (cleft lip), an incomplete closure of the roof of the mouth (cleft palate), undescended testes in boys, and the presence of extra fingers and toes

- Incomplete development of certain parts of the brain (eg. the forebrain), kidney abnormalities, structural heart defects at birth such as an abnormal opening in the partition dividing the upper or lower chambers of the heart or the persistence of the fetal opening between the two major arteries (aorta, pulmonary artery) emerging from the heart
- A relatively small head with a sloping forehead, a broad, flat nose, widely set eyes, vertical skin folds covering the eyes inner corners, scalp defects and low-set ears
- 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
- Life-threatening complications which may develop during infancy or early 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 been numbered 1-22 according to their size from the largest to the smallest
- Two sex chromosomes: X and Y

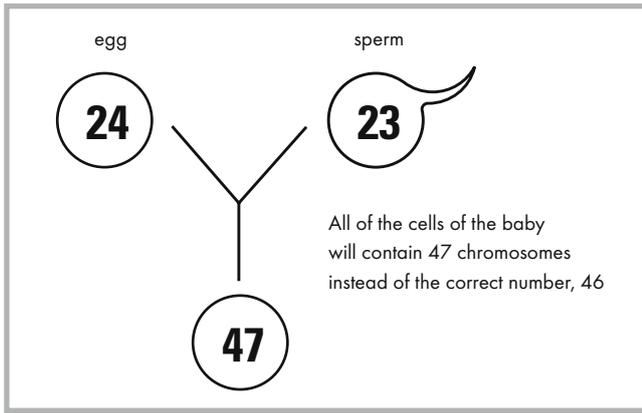


Figure 29.1: When the mother's egg has 24 chromosomes, and the father's sperm has the usual 23, the baby's cells will contain 47 chromosomes instead of 46. This may also happen in the reverse situation.

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.

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 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 29.1).

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 13

When there are three copies of chromosome number 13 in the cells, the condition is referred to as trisomy 13. The presence of the extra chromosome causes the mental and physical characteristics of the syndrome.

Figure 29.2 is a picture (*karyotype*) of the chromosomes from a female with trisomy 13.

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.

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

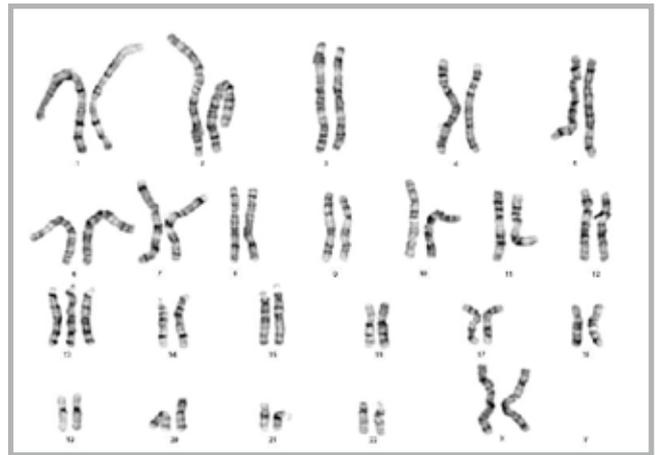


Figure 29.2: Chromosome picture (*karyotype*) from a female with trisomy 13. In this cell, there are 47 chromosomes including three copies of chromosome 13 instead of the usual two. Other cells in this female's body may have the correct chromosome number, 46 (SEALS Genetics, Prince of Wales Hospital, Randwick).

In individuals with mosaic trisomy 13, the range and severity of symptoms depends on the number and distribution of cells containing the extra copy of chromosome 13.

In general, having all cells in the body containing the extra copy of chromosome 13 means that the symptoms will be more severe.

Can a baby with trisomy 13 be cured?

There is no cure for trisomy 13. Care is usually directed at making babies as comfortable as possible.

In individuals with mosaic trisomy 13, 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 13. 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 13?

The extra copy of chromosome 13 can come from either the egg or the sperm. It has been shown however, that a woman who is over the age of 35 is at greater risk of having a child with trisomy 13, or any chromosomal abnormality (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 abnormal conception, which may have miscarried or not even have been recognised as a pregnancy because the miscarriage occurred so early.

Can trisomy 13 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 13 including:

- The mother's age
- A family history of trisomy 13
- 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 having, or definitely has trisomy 13.

- 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 trisomy 13 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, 13, 17, 17B, 17C, 18, 28

Information in this Fact Sheet is sourced from:

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