Fact sheet **37**

TRISOMY 13 – **PATAU SYNDROME**

This fact sheet talks about the chromosome condition trisomy 13 and includes the symptoms, cause, treatment and available testing.

IN SUMMARY

- Trisomy 13 is a chromosome condition also known as Patau syndrome
- Babies with trisomy 13 usually have distinctive features, severe learning disability and other physical developmental concerns
- Trisomy 13 is caused by having an extra copy of chromosome number 13.

WHAT IS TRISOMY 13?

Trisomy 13 is also known as Patau syndrome. It is a condition which is considered very serious and most babies with trisomy 13 do not survive to birth.

Some general signs and symptoms include:

- Developmental delays and severe learning 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 (e.g. the forebrain), kidney abnormalities, structural heart differences at birth such as an unusual opening in the partition dividing the upper or lower chambers of the heart.
- A relatively small head with an unusual sloping forehead, a broad, flat nose, widely set eyes, vertical skin folds covering the eyes inner corners, scalp differences and low-set ears
- Slow to grow and gain weight and severe feeding difficulties, low muscle tone (floppy) and episodes where breathing may stop
- Life-threatening complications may happen before birth, during early life of a baby or a child.

WHAT CAUSES TRISOMY 13?

Trisomy 13 is caused by the presence of an extra full (or partial) copy of chromosome number 13.

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 37.1* 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 37.2*.

This information is not a substitute for professional medical advice. Always consult a qualified health professional for personal advice about genetic risk assessment, diagnosis and treatment. Knowledge and research into genetics and genetic conditions can change rapidly. While this information was considered current at the time of publication, knowledge and understanding may have changed since.

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Figure 37.1:
Chromosome picture (karyotype) from a male (46,XY)

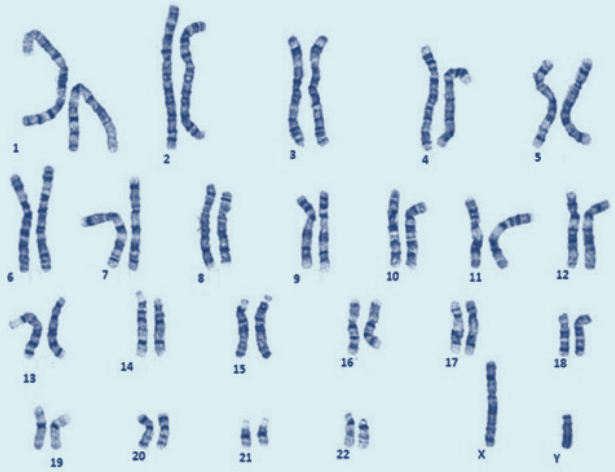
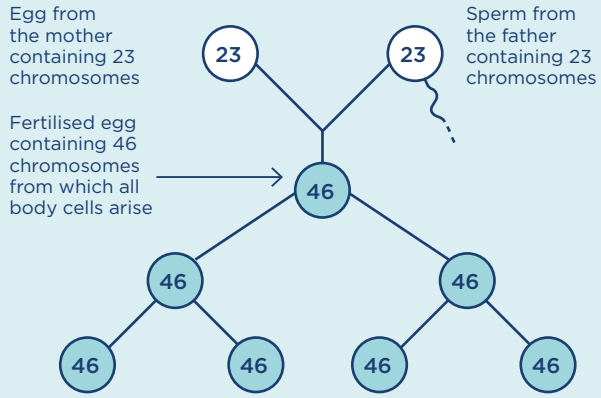


Figure 37.2:
At conception the sperm and egg combine



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.

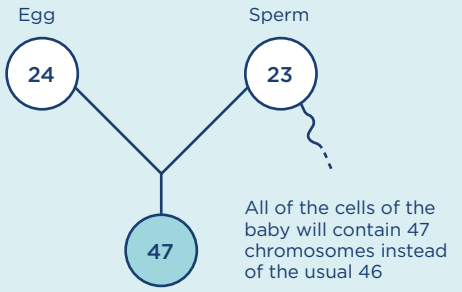
If an egg or sperm carrying 24 chromosomes combines with an egg or sperm carrying the usual 23 chromosomes, the result would be a person with 47 chromosomes instead of the usual 46 (See *Figure 37.3*).

- 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 13 usually includes a whole extra copy of chromosome number 13. This extra chromosome causes an imbalance and is the reason why there are differences in people with 3 copies of chromosome 13 material compared with people with the usual pair.

The presence of the extra copy of chromosome 13 causes the learning disability and physical features of trisomy 13. People with this condition usually have three whole copies of chromosome number 13, i.e. 47 chromosomes in their cells instead of 46. **Trisomy** means *three bodies*.

Figure 37.3:
When the egg has 24 chromosomes, and the sperm has the usual 23, the baby's cells will contain 47 chromosomes instead of 46.



Some people have trisomy 13 as a result of a chromosome rearrangement

Trisomy 13 due to a Robertsonian translocation occurs when chromosome 13 material is *stuck onto* or *translocated* onto another chromosome. Rarely, a chromosome translocation may happen when sections of chromosome 13 are rearranged with sections of another chromosome, so there is too much of only a part of chromosome 13 (partial trisomy). Signs and symptoms may be different from those found in full trisomy 13. Sometimes this can be inherited from a parent even if the parent does not have a health or developmental concern.

Figure 37.4:
Chromosome picture (karyotype) from a female with trisomy 13 (47,XX+13). In this cell, there are 47 chromosomes including three copies of chromosome 13 instead of the usual two.

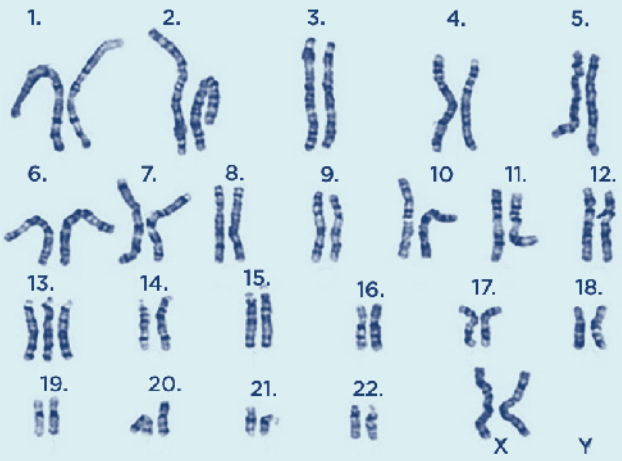


Figure 37.4 is a picture (karyotype) of the chromosomes from a female with trisomy 13 (47,XX+13).

Some people have trisomy 13 as a result of a mosaicism

Most individuals have the same chromosome makeup in all the cells in their body. People with trisomy 13 as a result of mosaicism have some cells in the body with the usual two copies of chromosome 13, and other cells with three copies of chromosome 13. 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.

HOW IS TRISOMY 13 INHERITED?

In most cases where trisomy 13 is caused by a complete extra copy of chromosome 13, that person will be the first and only person affected by the condition in that family. This is also the case where trisomy 13 is the result of mosaicism.

It is usually assumed that if the parents of a person with trisomy 13 have the usual two copies of chromosome 13, then the extra 13 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 13, 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 13 can vary.

IS THERE ANY TESTING AVAILABLE FOR TRISOMY 13?

Chromosome testing in a baby who is suspected of having trisomy 13 can be done using a blood sample. A doctor may suspect a baby has the condition if there are features or symptoms linked with the condition.

Testing for pregnancy

Testing for trisomy 13 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 13 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.