2. Mosaic Patau syndrome
Mosaic Patau syndrome is a rare form of the condition where some cells in the body have two copies of chromosome 13 and others have three copies. Mosaic Patau syndrome is very variable. Some babies are only mildly affected, while others have many problems.

3. Chromosome translocation
An alternative (but rarer) cause of Patau syndrome is an ‘unbalanced translocation’. This happens when an extra portion of chromosome 13 is attached to part of another chromosome. This can occur because one of the baby's parents carries what is known as a 'balanced translocation'.

It is possible to find out whether a baby has a chromosome translocation or a primary trisomy by looking at their chromosomes.

Will it happen again?
Patau syndrome is almost always caused by a primary trisomy, so it is very unlikely that a future pregnancy will be affected.

However some parents do choose to have a test in a future pregnancy to check the chromosomes of the baby. This possibility can be discussed at the genetics clinic.

Further information
SOFT UK
Support Organisation for Trisomy 13, 18 and related disorders
Website: www.soft.org.uk
Email: enquiries@soft.org.uk

Feedback
We appreciate and encourage feedback. If you need advice or are concerned about any aspect of your care or treatment please speak to a member of staff or contact the Patient Advice and Liaison Service (PALS):

Freephone: 0800 183 0204
From a mobile or abroad: 0115 924 9924 ext. 65412 or 62301
E-mail: pals@nuh.nhs.uk
Letter: NUH NHS Trust, c/o PALS, Freepost NEA 14614, Nottingham NG7 1BR
www.nuh.nhs.uk

If you require a full list of references for this leaflet please email patientinformation@nuh.nhs.uk or phone 0115 924 9924 ext. 67184.

The Trust endeavours to ensure that the information given here is accurate and impartial.

Adapted from leaflet by Clinical Genetics at Guy’s and St Thomas’ NHS Foundation Trust.

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Ref: 1042/v3/0318/AR.
Introduction
People are usually born with 46 chromosomes, which are arranged in 23 pairs. The chromosomes are numbered from 1 to 22 and the last pair, known as X and Y, determine whether we are male (XY) or female (XX). One from each pair of chromosomes comes from our mother in the egg and the other of the pair comes from our father in the sperm.

What is Patau syndrome?
Very occasionally, a baby boy or girl has an extra copy of chromosome number 13. The extra chromosome disrupts the normal course of development and can cause a range of problems. The name of this condition is Patau syndrome. The picture below shows the chromosomes of a baby with Patau syndrome.

About one in 12,000 babies are diagnosed as having an extra copy of chromosome 13. Patau syndrome was first described by Dr Klaus Patau. The condition is also known as trisomy 13 because there are three copies of chromosome 13.

What are the features of Patau syndrome?
Patau syndrome is a serious condition with a range of medical problems. Some of the problems that can occur are:
- Major malformations of the brain causing delays in development and seizures
- Heart defects
- Malformations of the eyes which can cause blindness
- Extra fingers or toes ('polydactyly')
- Abdominal defects
- Cleft lip (an opening between the mouth and the nose, where the lip looks 'split') and cleft palate (an opening in the roof of the mouth)
- Kidney problems
- Deafness
- Feeding difficulties

Sadly, most babies with Patau syndrome die before the end of a pregnancy or are stillborn. Babies that are born alive usually die shortly after birth, or in the first few months of life.

Most babies born with Patau syndrome need specialised nursing in hospital.

When is the diagnosis made?
The diagnosis may be suspected in the pregnancy during an ultrasound scan but the only way to reach a definite diagnosis is to look at the baby's chromosomes and see if there is an extra copy of chromosome 13.

There are tests that can be done during pregnancy to look at the baby's chromosomes. These tests can be discussed with your midwife.

Detecting Patau syndrome in a pregnancy gives parents the opportunity to make the personal choice of whether or not to continue with the pregnancy. It can also help parents to prepare themselves for how the condition will affect their lives.

When Patau syndrome is suspected at birth the baby's chromosomes can be checked by taking a blood sample from the baby or from the umbilical cord.

What causes Patau syndrome?
Patau syndrome is usually caused by the presence of an extra chromosome 13. In most cases, the extra chromosome is present due to a 'genetic mistake' that occurred in either the egg or the sperm that went to make that baby. There are three different ways that the extra chromosome can be present.

1. A third copy of chromosome 13 (trisomy 13)
The majority of babies with Patau syndrome have an extra copy of chromosome number 13 (ie. three copies). This is called a 'primary trisomy' (as shown in the diagram). This is a non-inherited form of Patau syndrome.

The parents usually have normal chromosomes themselves. It is not known why the 'genetic mistake' happens, but it is slightly more likely to occur in babies of older mothers.