UK National Screening Committee

NHS Screening Programmes

Trisomy 18 (Edward) and Trisomy 13 (Patau) – Frequently asked questions

What are Edward's Syndrome and Patau's Syndrome?

Edward's Syndrome (trisomy 18 or T18) and Patau's Syndrome (trisomy 13 or T13) are rare, but very serious conditions which affect a small number of babies every year. They are both genetic conditions caused by 'chromosomal abnormalities' known as 'trisomies'. This is when there are three copies of a particular chromosome rather than the usual two.

What is a chromosomal defect?

Chromosomes exist in all of the cells in our bodies; they contain the genes that determine how we develop. There are 23 pairs of chromosomes in each cell. Problems can occur when the sperm or egg cells are produced which can lead to a baby having an extra chromosome.

In most cases of Edward's Syndrome there is an extra copy of chromosome 18 in each cell. Likewise, in most cases of Patau's Syndrome there is an extra copy of chromosome 13 in each cell.

What can happen to children affected by these syndromes?

Most babies affected by these conditions die before they are born or shortly after birth.

Some babies may survive to adulthood but this is rare. Many will be stillborn. While it is difficult to say how seriously those who survive will be affected it is likely these babies will have a wide range of problems. These may include major brain abnormalities.

Babies affected by Edward's Syndrome (trisomy 18) may:

- have heart problems, unusual head and facial features, and growth problems;
- be unable to stand or walk, unable to talk; and
- have problems with their kidneys.

Babies affected by Patau's Syndrome (trisomy 13) can:

- have heart problems;
- have a cleft lip and palate;
- have growth problems;

- be unable to stand or walk; and
- have poorly formed eyes and ears.

Who is at risk and how many are affected?

Although women of any age can have a child with Edward's Syndrome or Patau's Syndrome, the chance increases as a woman gets older. In most cases it does not run in the family.

Edward's Syndrome affects about 3 of every 10,000 births and Patau's Syndrome affects about 2 of every 10,000 births.

What is the UK NSC screening policy for these syndromes?

Although Edward's Syndrome and Patau's Syndrome may be identified through the current 18 – 20 week scan, the UK NSC has reviewed the evidence and has recommended screening for these conditions earlier in pregnancy.

First trimester screening helps provide an earlier diagnosis allowing the woman to make decisions about her pregnancy at an earlier stage. The screening test is part of the NHS Fetal Anomaly Screening Programme (FASP).

How will babies be identified as at risk of having these conditions?

The risk of a baby having either Edward's Syndrome or Patau's Syndrome can be identified through a blood test and an ultrasound scan during the first trimester of pregnancy.

The blood test will check the levels of certain hormones and the first trimester scan will measure the amount of fluid at the back of the baby's neck (nuchal translucency). When these are combined, the test can identify whether the woman has an increased risk of having a baby with one of these conditions. This is called the 'combined test'.

If Edward's Syndrome or Patau's Syndrome are suspected, the mother will be offered another test called an amniocentesis or a chorionic villus sampling (or CVS) to make the diagnosis.

What treatment options are available?

Most babies with Edward's Syndrome and Patau's Syndrome die before they are born or shortly after birth. Some babies live beyond a year, but this is rare. Babies with these conditions will have complex physical and learning disabilities.

Treatment would focus on managing heart abnormalities, feeding and treating infections. Many of these babies can be cared for at home with support.

Before the baby is born the mother/parents would be given the chance to talk to specialists about their options.

Whatever decision the mother makes will be respected and appropriate support and care will be offered.

How will the test be implemented?

The combined test will be offered between 10 and 14 weeks of pregnancy, alongside the current first trimester screening for Down's Syndrome.

A working group is now looking at the most effective method for implementing the screening test in England as part of the NHS Fetal Anomaly Screening Programme.

As part of this the group will assess what training and education resources are needed, and agree a standardised process for carrying out the screening test.