Trisomy 18
(also called Edwards’ syndrome or T18)

Information for parents
1. What is trisomy 18 (Edwards’ syndrome)?

Inside all of the cells of our bodies there are tiny structures called chromosomes. These chromosomes carry the genes that determine how we develop. There are 23 pairs of chromosomes in each cell. When our bodies produce sperm or egg cells, the pairs divide and rearrange themselves. Sometimes these pairs of chromosomes do not divide correctly.

In trisomy 18 there is an extra copy of chromosome 18 in each cell.

There are three forms of the syndrome as explained below. This information sheet focuses on complete trisomy 18, the most common and most serious form of trisomy 18.

<table>
<thead>
<tr>
<th>Complete trisomy 18</th>
<th>This is when every cell in the body has three copies of chromosome 18.</th>
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<tbody>
<tr>
<td>Mosaic trisomy 18</td>
<td>This is when some cells have the usual two copies of chromosome 18 and some have three copies.</td>
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<tr>
<td>Partial trisomy 18</td>
<td>This is when there is an extra part of some of chromosome 18 in all the body's cells.</td>
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Complete trisomy 18 is fatal. Babies with partial and mosaic trisomy 18 may survive to adulthood, but this is rare.

All babies with trisomy 18 will have a wide range of problems. Unfortunately these problems are usually extremely serious. These may include major brain abnormalities leading to learning difficulties. Many babies also have heart problems, abnormal head and facial features, growth problems, problems with their hands, legs and arms, and problems with their kidneys. It is difficult to say how serious the effects of partial and mosaic trisomy 18 will be before the baby is born.

Although women of any age can have a child with trisomy 18, the chance increases as a woman gets older.

2. How common is it?

Trisomy 18 affects about 3 of every 10,000 births.

3. How is it diagnosed and confirmed?

Trisomy 18 may be suspected from the early ‘combined’ test for Down’s syndrome because of an increased nuchal translucency (thickening at the back of the baby’s neck).
Signs that the baby may have trisomy 18 may also be noticed at the Fetal Anomaly ultrasound scan carried out between 18 weeks and 21 weeks of pregnancy.

If trisomy 18 is suspected, you will be offered another test called an amniocentesis or a chorionic villus sampling (or CVS). There is more information on CVS and amniocentesis in leaflets called: *Chorionic villus sampling (CVS) – information for parents* or *Amniocentesis test – information for parents*. These are available on our website at [www.fetalanomaly.screening.nhs.uk/publicationsandleaflets](http://www.fetalanomaly.screening.nhs.uk/publicationsandleaflets). You can also ask for a copy of these from your healthcare professional.

4. **Is there any treatment?**

Unfortunately, there is no cure for trisomy 18 as the extra chromosomes cannot be removed.

5. **What is the outlook for the baby?**

Most babies with complete trisomy 18 die before they are born or shortly after birth. Babies with partial or mosaic trisomy 18 can live beyond a year, but this is rare. Babies with these conditions still have complex physical and learning difficulties.

Treatment of babies born alive will focus on feeding, treating infections and managing heart abnormalities.

Many babies can be cared for at home with support. You will be offered emotional and practical support.

6. **What happens next?**

You will be given the chance to talk to specialists about your options. You will have the opportunity to discuss the possible implications of ending or continuing your pregnancy.

As babies with complete trisomy 18 die before or shortly after birth, you will be offered a termination to end the pregnancy. If you choose to have a termination, your health professional will talk to you about the procedure and support you through the process.

If you choose to continue your pregnancy, your healthcare team will help you plan how your care, including delivery, is managed. As there is no cure for this condition, doctors will discuss palliative care with you. Palliative care is care aimed at relieving the symptoms of a condition rather than treating or providing a cure. You may be referred to the children’s palliative care team and the local bereavement service.

Whatever you decide, your decision will be respected and you will be supported by your midwife and doctor.
7. How likely is it to happen in a future pregnancy?

Anyone can have a baby with trisomy 18. In most cases this condition does not run in the family. It is not due to anything you did or did not do.

You are much more likely to have a normal, healthy baby in your next pregnancy than to have another baby affected with trisomy 18.

For complete trisomy 18, the chance of having another baby with this condition is about 1% (1 in 100). The chance of having a baby with trisomy 18 does increase with age, so older mothers will have a higher chance of having another baby with this condition.

You may be offered a chromosome test to see whether this condition is linked to something called ‘a translocation’ of your chromosomes. If this is the case, you will be referred to a genetic counsellor to discuss the chances of this condition happening in another pregnancy. In a future pregnancy you would be offered scans by a specialist as well as diagnostic tests (chorionic villus sampling (CVS) or amniocentesis) for reassurance and to diagnose any problems at an early stage.

8. Where can I get more information and support?

You may feel you only want to talk to your family and friends, or a particular doctor or midwife from the hospital. However, there are a lot of other people and organisations that can provide information, help you make your decisions and support you in your pregnancy and afterwards. You can also talk things through with the hospital chaplain or your own minister or faith leader.

9. Further information, charities and support organisations

The organisations below can offer you support. There are details of other support organisations on our website at www.fetalanomaly.screening.nhs.uk. If you have any questions about the information in this leaflet or where the information came from, please email us at enquiries@ansnsc.co.uk.
Antenatal Results and Choices (ARC)
Email: info@arc-uk.org
Helpline: 0845 077 2290
Website: www.arc-uk.org

Antenatal Results and Choices (ARC) provides information and support to parents before, during and after antenatal screening and diagnostic tests, especially those parents making difficult decisions about testing, or about continuing or ending a pregnancy after a diagnosis. ARC offers ongoing support whatever decisions are made.

SOFT UK
Email: enquiries@soft.org.uk
Website: www.soft.org.uk

SOFT UK provides information booklets and support for those affected by Patau's syndrome or Edwards' syndrome (trisomy 13 and trisomy 18), and related disorders such as mosaicism, deletion, ring and partial trisomy:

• whether you have a termination or continue the pregnancy after a prenatal diagnosis;
• after the loss of an unborn baby, baby or child; and
• for parents and carers of a baby or child.

This information has been produced on behalf of the NHS Fetal Anomaly Screening Programme for the NHS in England. In other countries, check with a health professional to find out whether there are any differences in approaches to screening.

This leaflet has been developed through consultation with the NHS Fetal Anomaly Screening Programme expert groups.

All of our publications can be found online at www.fetalanomaly.screening.nhs.uk.

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