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Edwards’ syndrome is almost always caused by a primary trisomy, so it is very unlikely that a future pregnancy will have this condition.

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**Contact us**
If you live in the South East Thames Region, contact Clinical Genetics Department, 7th Floor, Borough Wing, Guy’s Hospital, Great Maze Pond, London SE1 9RT, tel: 020 7188 1364, fax: 020 7188 1369, web: www.guysandstthomas.nhs.uk/genetics

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**Edwards’ syndrome (Trisomy 18)**
This leaflet gives information about Edwards’ syndrome, and the possible tests available to you.
Introduction
Humans are usually born with 46 chromosomes, which are arranged in 23 pairs. The chromosomes are numbered from 1 to 22 and the 23rd pair, known as X and Y, determine whether we are a boy (XY) or a girl (XX). One of 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 Edwards’ syndrome?
Very occasionally, a baby has an extra copy of chromosome number 18. The extra chromosome disrupts the normal course of development and can cause a range of problems. The name of this condition is Edwards’ syndrome.

The condition is also known as Trisomy 18 as there are 3 copies of chromosome 18.

What are the features of Edwards’ syndrome?
Edwards’ syndrome is a serious condition and affected babies can have a range of severe medical problems including:
- a low birth weight
- a small head (microcephaly)
- severe learning disability
- a small jaw (micrognathia)
- malformations of the heart
- malformations of kidneys
- clenched fists and malformed feet
- feeding and breathing problems
- cleft lip (an opening between the mouth and nose) and/or cleft palate (an opening in the roof of the mouth)

Sadly, most babies with Edwards’ syndrome die before the end of pregnancy or are stillborn.

Of the babies with Edwards’ syndrome that are born alive, about half survive the first month of life and less than 1 in 10 (10%) live longer than a year.

They usually require specialised nursing in a hospital or hospice. There are some infants who can live at home and be cared for by their parents.

When is the diagnosis made?
The diagnosis may be suspected in pregnancy during an ultrasound scan, but the only way to reach a definite diagnosis is to look at the baby's chromosomes.

There are 2 tests that can be done during pregnancy to look at the baby's chromosomes: CVS and amniocentesis. These tests can be discussed in the genetics clinic.

Detecting Edwards’ syndrome during a pregnancy gives parents the opportunity to make the personal choice of whether or not to continue with the pregnancy. It also allows time to come to terms with the diagnosis and prepare for the future.

If Edward’s syndrome is suspected at birth, a blood sample can be taken to look at the baby's chromosomes.

What causes Edwards’ syndrome?
In most cases, the extra chromosome is present due to a problem that occurred when the egg or sperm were made. It is not known why this happens, but it is slightly more likely to occur in babies of older mothers.

When Edwards’ syndrome is caused by an entire extra chromosome 18 this is called a primary trisomy. This type of Edwards’ syndrome is not inherited.

Chromosome translocation
Another (but rare) cause of Edwards’ syndrome is an unbalanced translocation. This happens when an extra part of chromosome 18 is attached to part of another chromosome. This can happen if one of the parents carries a balanced translocation. This can be discussed in more detail in the genetics clinic.

Mosaic Edwards’ syndrome
Mosaic Edwards' syndrome is a rare form of the condition where some cells in the body have 2 copies of chromosome 18 and others have 3 copies of 18.

Mosaic Edwards’ syndrome is very varied. Some babies are only mildly affected, while others have as many problems as babies with the ‘full’ form.
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