extra chromosome 18 this is called a “primary trisomy” (as shown the diagram). This type of Edwards syndrome is not inherited.

**Chromosome translocations**

An alternative (*but rare*) cause of Edwards syndrome is an “unbalanced translocation”. This happens when an extra portion of chromosome 18 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”.

**Mosaic Edwards syndrome**

Mosaic Edwards syndrome is a rare form of the condition where some cells in the body have two copies of chromosome 18 and others have three.

Mosaic Edwards syndrome is very variable. Some babies are only mildly affected, while others have as many problems as babies with the “full” form.

**Will it happen again?**

Edwards syndrome is almost always caused by a primary trisomy and therefore it is very unlikely that a future pregnancy will have this condition. However it would be possible to have testing in a future pregnancy to check the chromosomes of the baby. This possibility can be discussed at the genetics clinic.

**For more information:**

If you need more information please contact your local Genetics Department. If you live in the Yorkshire region please contact:

Department of Clinical Genetics
Ward 10
Chapel Allerton Hospital
Harehills Lane
Leeds
LS7 4SA
Telephone: 0113 392 4432

Seen in clinic by..............................
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 a boy (XY) or a girl (XX). One of each pair of chromosomes is inherited from our mother in the egg and the other of the pair is inherited from our father in the sperm.

What is Edwards Syndrome?
Very occasionally, a baby boy or girl 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 picture below is a photograph of the chromosomes of a baby with Edwards syndrome.

Edwards syndrome is named after Dr. John Edwards who discovered that the extra chromosome causes the condition. The condition is also known as Trisomy 18 as there are three 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.

For the most part they require specialised nursing in a hospital or hospice. However, there are some babies 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 two tests that can be done during pregnancy to look at the baby’s chromosomes. 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 Edwards syndrome is suspected at birth then 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