Choroid plexus cysts - These are fluid filled areas on a part of the brain known as the choroid plexus. The cysts themselves are harmless and occur in about 1 or 2% of pregnancies. On their own they might not suggest any medical problem, but they occur more often in babies with conditions such as Edwards syndrome.

Polyhydramnios - This is where the baby is surrounded by more amniotic fluid than usual. It occurs in about 2% of all pregnancies and can go away by itself or it might increase the risk of complications later on in pregnancy.

Intrauterine growth retardation or IUGR - The baby is smaller than expected on the basis of how many weeks into the pregnancy the mother is. IUGR occurs in 3 to 5% of pregnancies and often it is **not** associated with a chromosomal abnormality.

Microcephaly - This means the baby has a reduced head size compared to the rest of the body.

Non-immune hydrops fetalis - The baby shows enlarged liver, spleen, or heart, and fluid build-up in the abdomen.

Exomphalos - This means some of the organs of the body grow in a sac outside the abdomen.

Cleft lip and cleft palate - A cleft lip is an opening between the mouth and the nose and the lip looks "split". A cleft palate is an opening in the roof of the mouth.

Clenched hands and overlapping fingers

Tests during pregnancy

If Trisomy 18 is suspected during an ultrasound scan, the only way to get a definite diagnosis is to look at the baby's chromosomes and see if there is an extra copy of chromosome 18. 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.

In babies that are born and suspected of having Edwards syndrome, looking at the cells in a blood sample from the baby or from the umbilical cord can also check the baby's chromosomes.

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For more information:

If you need advice about any aspect of Edwards syndrome you are welcome to contact:

Clinical Genetics Departments

Northern Scotland (main base Aberdeen)
Tel: 01224 552120 Fax: 01224 559390
(Aberdeenshire, Moray, Highland, Western & Northern Isles)

Tayside (main base Dundee)
Tel: 01382 632035 Fax: 01382 645731
(Perth & Kinross, Angus, North East Fife)

South East Scotland (main base Edinburgh)
Tel: 0131 651 1012 Fax: 0131 651 1013
(Borders, Lothian, South West Fife)

West of Scotland (main base Glasgow)
Tel: 0141 201 0808 Fax: 0141 201 0361
(Glasgow, Argyll & Bute, Aryshire, Dumfries & Galloway, Stirling, Lanarkshire, Falkirk)

If you would like further information and the opportunity to talk to other parents you can contact:

Support Organisation for Trisomy 13/18 and Related Disorders (S.O.F.T)
Internet: www.soft.org.uk, Email: enquiries@soft.org.uk
Telephone: (0121) 351 3122

Contact a Family
209-211 City Road,
London EC1V 1JN
Internet: www.cafamily.org.uk, Email: info@cafamily.org.uk
Telephone: 020 7608 8700, Fax: 020 7608 8701

Antenatal Results & Choices (ARC). (Provide support and information for parents during antenatal screening/testing, awaiting results and diagnosis of fetal abnormality.)
73 Charlotte Street
W1T 4PN
Internet: www.arc-uk.org, Email: info@arc-uk.org
Telephone: 020 7631 0285

Seen in clinic by.....

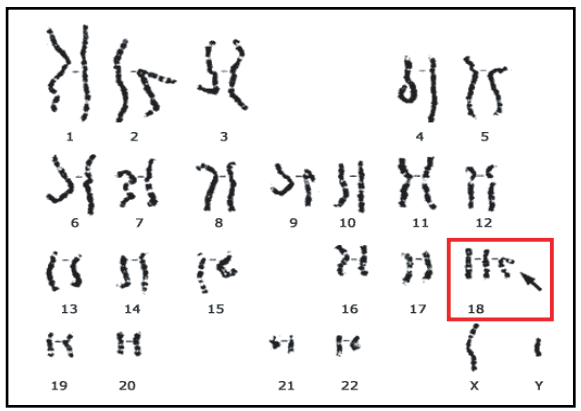
Edwards Syndrome (Trisomy 18)

CLINICAL GENETICS CLINICAL GENETICS

Information for parents of a baby or pregnancy with trisomy 18

Introduction

Humans 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 comes from our mother in the unfertilised egg and the other of the pair comes from our father in the sperm. Very occasionally, a baby is born with an extra copy of chromosome number 18. This condition is known as Edwards syndrome. Edwards syndrome happens more often in girls than boys, but it is not known why. The picture below is a photograph of the chromosomes of someone with Edwards syndrome.



About 1 in 3000 pregnancies/babies are diagnosed as having an extra copy of chromosome 18. 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 because there are 3 copies of chromosome 18.

What causes Edwards syndrome?

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. 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.

When Edwards syndrome is caused by an entire extra chromosome 18 this is called a "primary trisomy" (as shown opposite). This is a non-inherited version of Edwards syndrome.

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'. This can be discussed in a genetics clinic.

It is possible to distinguish between the 2 causes of Edwards syndrome by looking at the chromosomes of the baby. This can be done during pregnancy, or after pregnancy by testing the baby's blood, or blood from the umbilical cord. If the baby's chromosomes have not been checked then it is possible to look at the chromosomes of the parents to see if either of them carries a balanced translocation.

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 some parents do choose to have a test in a future pregnancy to check the chromosomes of the baby. The need for testing and how this is carried out can be discussed at the genetics clinic.

What are the features of Edwards syndrome?

Edwards syndrome is a serious condition and affected babies/pregnancies can have a range of severe medical problems. Sadly, most babies with Edwards syndrome die before the end of pregnancy or are stillborn. Of the babies that are born alive, about half survive the first month of life and less than 10% live longer than a year. For the most part they require specialised nursing in a hospital or hospice. However there are some infants who can live at home and be cared for by their parents.

Babies with Edwards syndrome tend to have a low birth weight, a small head ("microcephaly"), a small jaw ("micrognathia"), malformations of their heart and kidneys, clenched fists and malformed feet. They also characteristically have feeding and breathing problems in infancy and severe learning disability.

Mosaic form of 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. How a child/person is affected depends on how many cells have three copies of chromosome 18. If a couple have had one baby with mosaic Edwards syndrome, they are very unlikely to have another child with the same condition.

When is the diagnosis made?

Sometimes Edwards syndrome can be suspected during an ultrasound scan, where the main structures of the baby are looked at to pick up any potential problems. Detecting the condition early can help some parents to prepare themselves for how the condition will affect their lives. It also gives them the opportunity to make the personal choice of whether or not to continue with the pregnancy.

There are many things that could be seen during an ultrasound scan that increase the baby's chance of having Edwards syndrome. Some of these signs would suggest serious abnormalities. Other signs are not actually harmful on their own but do occur more often in babies with conditions like Edwards.

Here are a few of the more common signs that suggest a baby has Edwards syndrome:

Raised nuchal thickness - A scan may be done at 12 to 14 weeks to look for a small collection of fluid on the back of the baby's neck. The thickness of this fluid filled area is measured ("nuchal translucency" or "nuchal thickness") and if it is larger than average ("raised") it suggests the baby might have a condition such as Edwards syndrome.