Support for parents and carers

Having a baby with Trisomy 18 can be a difficult situation for parents to cope with. However, you should never feel that you are alone. Great support and advice is available for parents who have a pregnancy diagnosed with Trisomy 18.
You will be able to access pregnancy counselling through your Fetal Medicine Unit for you and your whanau.

You may also like to contact:

Parent to Parent New Zealand

www.parent2parent.org.nz

Parent to Parent New Zealand is an information and support network for parents of children with special needs ranging from the very common to the most rare conditions. The service is free to families.

SOFT - Support Group for Trisomy 13/18

trisomy.org
Email: soft.info@nzord.org.nz

References

http://www.britannica.com
www.trisomy18.org
http://www.pathology.leedsth.nhs.uk/pathology/Portals/0/PDFs/Edwards%20Syndrome%20%28Trisomy%2018%29.pdf
TRISOMY 18.doc

For more information please contact your local NZMFMN Unit

AUCKLAND DISTRICT HEALTH BOARD
Te Toka Tumai

Auckland: 09 307 4949 ext 24951

Capital & Coast District Health Board
ŪPOKO KI TE URU HAUORA

Wellington: 04 806 0774

Canterbury District Health Board
Te Poari Hauora ō Waitaha

Christchurch: 03 364 4557

New Zealand Maternal Fetal Medicine Network
NZMFMN@adhb.govt.nz

Trisomy 18 (Edward's Syndrome)

New Zealand Maternal Fetal Medicine Network

Reviewed: September 2015

What is a chromosome?

Inside our cells there are microscopic, threadlike parts called chromosomes. They carry hereditary information from one generation to the next in the form of genes. A chromosome is made up of protein and deoxyribonucleic acid (DNA) which determines a person's inherited traits such as eye colour or left/right handedness.

You inherit 23 chromosomes from your mother and 23 from your father for a total of 46 chromosomes arranged in pairs (one from each parent). They are numbered in pairs from 1 to 22. The last pair (23) is the sex chromosomes and determine if the developing baby is a boy or a girl.

What is Trisomy 18?

Trisomy 18 or Edward's Syndrome results when there are three copies of chromosome 18 instead of the usual two.

There are three forms of the syndrome:

- Complete - this is when every cell in the body has 3 copies of chromosome 18
- Mosaic - this is when some cells have the usual 2 copies of chromosome 18 and some have 3 copies
- Partial - this is when there is an extra part of chromosome 18 in every cell

The survival of infants born with Trisomy 18 will depend on how severely they are affected. In one study, of all the babies born with Trisomy 18, 50% died by one week of age and 90% did not survive past the first year of life. Therefore while less than 10% survive the first year of life, some children live well past the first year.

How common is Trisomy 18?

Trisomy 18 affects about 3 of every 10,000 births and is one of the most common chromosomal abnormalities. Although women of any age can have a child with Trisomy 18, the chance increases as a woman gets older.

How is Trisomy 18 diagnosed?

If you have come to our Fetal Medicine Unit it is likely that you have already had the screening test for Trisomy 18 (MSS1 or MSS2) and that it shows an increased risk.

It is important to understand that this screening test does not give a definitive answer as to whether your baby does have Trisomy 18- it just gives a risk.

If your screening test shows an increased risk there is a test available in the private sector called a non-invasive (blood test) prenatal test (NIPT). It is currently not publicly funded and will cost you approximately $1000. If this test was low risk it would virtually exclude the risk of Trisomy 18, however, if it was positive you would still be offered invasive testing for a definitive answer.

There are two prenatal diagnostic tests that can be done- amniocentesis and chorionic villus sampling (CVS). These tests carry a small risk of miscarriage (approximately 0.1% for amniocentesis and 0.2% for CVS). An amniocentesis/CVS is an antenatal test that allows us to detect chromosomal abnormalities from a sample of the fluid from around your baby or a sample of tissue from the placenta. It is obtained by inserting a thin needle through your abdomen and into your uterus.

Is there any treatment for Trisomy 18?

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

Features

All babies with Trisomy 18 will have a wide range of problems. Unfortunately these problems are usually extremely serious and can include:

- Low birth weight
- Microcephaly (small head)
- Developmental delay and intellectual disabilities
- Micrognathia (small jaw)
- Heart defects
- Kidney defects
- Clenched fists
- Malformed feet
- Feeding problems
- Breathing problems
- Cleft lip (an opening between the mouth and nose)
- Cleft palate (an opening in the roof of the mouth)

What happens next?

When the diagnosis is confirmed, you will be given time to make a decision about what happens next. You may choose to continue your pregnancy. If this is the case the Fetal Medicine team will make a plan for your care which will continue under your LMC (Lead Maternity Carer) with obstetrician input through your local hospital. You are at a much greater risk of developing pre-eclampsia during your pregnancy if your baby has Trisomy 18 so you will be carefully monitored for this by your midwife and doctor.

Treatment of your baby will focus on feeding, treating infections and managing heart abnormalities. Many babies can be cared for at home with support which will be planned and implemented prior to discharge from hospital.

If there is a Palliative Care team in your area, they can be involved to help you make memories of your baby.

If you decide not to carry on with your pregnancy a plan will be made with you by the Fetal Medicine team and you will be able to deliver at your local tertiary hospital. Whatever you decide, your decision will be respected and you will be supported by the Fetal Medicine team.