Support for parents and carers

Having a baby with Trisomy 13 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 13.

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

http://www.patient.co.uk/doctor/pataus-syndrome-trisomy-13

http://brochures.mater.org.au/Home/Brochures/Mater-Mothers-Hospital/Patau-Syndrome-Trisomy-13

For more information please contact your local NZMFMN Unit

Auckland: 09 307 4949 ext 24951

Wellington: 04 806 0774

Christchurch: 03 364 4557

New Zealand Maternal Fetal Medicine Network

NZMFMN@adhb.govt.nz

Trisomy 13 (Patau Syndrome)

New Zealand Maternal Fetal Medicine Network

Reviewed: August 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 13?

Trisomy 13 results when there are three copies of chromosome 13 instead of the usual two.

The extra genetic material disrupts the normal course of development in the fetus. Babies diagnosed with Trisomy 13 have multiple severe abnormalities that are often not compatible with life. Sadly, many babies do not survive to the end of the pregnancy or full-term, and are stillborn.

Of all the babies born with an extra copy of chromosome 13 in all the cells of their body, around 50% die in the first month, and the rest within the first year.

How common is Trisomy 13?

The prevalence of Trisomy 13 is approximately 1 in 10,000 live births and is one of the most common chromosomal abnormalities. Although women of any age can have a child with Trisomy 13, the chance increases as a woman gets older.

How is Trisomy 13 diagnosed?

If you have come to our Fetal Medicine Unit it is more than likely that you have already had the screening test for Trisomy 13 (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 13- 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 13, 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- an amniocentesis and a 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 13?

Unfortunately, there is no cure for Trisomy 13 as the extra chromosomes cannot be removed.

Features

Your baby can have some or all of these:

- Intrauterine growth restriction and low birth weight
- Heart defects
- Holoprosencephaly - a condition whereby the brain doesn't divide in half. This can be associated with facial defects such as midline cleft lip and palate, small/absent eyes or problems with your baby's nose
- Neural tube defects (spine and brain)
- Severe learning disabilities
- Breathing problems
- Microcephaly (abnormally small head)
- Deformities of the ear and deafness
- Problems with your baby's gut such as an omphalocele or hernia
- Problems with your baby's kidneys and genitals such as polycystic kidneys, micropenis or hypertrophy of the clitoris
- Abnormalities of the hands and feet such as extra fingers or toes and rocker-bottom feet (rounded sole that can indicate a chromosomal abnormality)

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. Once your baby is born the Neonatologists/Paediatricians will care for your baby with you until he/she passes away.

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 hospital.

Whatever you decide, your decision will be respected and you will be fully supported by the Fetal Medicine team.