NZMFMN Statement on the use of Non-Invasive Prenatal Testing (NIPT)

New Zealand Maternal Fetal Medicine Network

Pregnancy care includes a number of options for women to screen the developing fetus for anomalies. These include the most common genetic disorders and structural disorders. The screening involves blood tests and scans and is usually completed by 20 weeks gestation. All screening is optional and it is important women understand what the screening is designed to detect so that she can decide whether she wishes to have the screening. It is also important women understand the limitations of screening and the potential pathways if there is a high risk result.

In New Zealand there is a fully funded option of screening for Trisomy 21 (Down Syndrome), Trisomy 18 (Edwards Syndrome) and Trisomy 13 (Patau Syndrome). This screening is called antenatal screening for Down syndrome and other conditions and there are two options available - first trimester combined screening and second trimester maternal serum screening. First trimester combined screening comprises an ultrasound measuring the nuchal translucency and crown rump length of the fetus and a blood test measuring the levels of chemicals released during pregnancy. This is completed by 14 weeks gestation. The two assessments are combined to give risk assessments for each of the trisomies. Second trimester maternal serum screening consists of a blood test measuring the levels of four chemicals released during pregnancy. Monitoring of this screening shows a sensitivity of 78% (i.e. ability to correctly identify cases of trisomy). The false positive rate is around 5% (i.e. the test incorrectly identifies a woman as high risk and the fetus does not have a trisomy). For more information on this screening see www.nsu.govt.nz

Non Invasive Prenatal Testing (NIPT) is a new method of screening a pregnancy for some genetic anomalies. NIPT relies on detecting minute quantities of cell free DNA (cfDNA) in the maternal blood arising from the placenta. It has been used in practice overseas (in particular in the US) for a number of years. The research in this area is moving very quickly and over the last two years NIPT has been shown to be a useful screening test in both low and high risk pregnancies. When NIPT is used to screen for Trisomy 21, 18 and 13 the sensitivity is 99.5%. The false positive rate is very low. So NIPT offers the advantage of very rarely missing a case of Trisomy 21, 18 and 13 and a very high chance it is correct if abnormal. This has resulted in a reduction in the number of invasive tests (amniocentesis and chorionic villus sampling) in geographical areas where it has been used. This is an advantage as invasive tests have a risk of miscarriage.

However there are more and more tests being developed using the cfDNA to diagnose problems in the developing fetus. These are for conditions which are rare and where there may be uncertainty about outcome. As they are very rare the screening is not as good and there is a much higher chance of a false positive result. This can result in more invasive procedures being performed and many of the benefits of NIPT being potentially lost.

NIPT screening is currently a user pays option for women in New Zealand. If a woman is considering NIPT in her pregnancy we recommend the following:

- **Clear pre-test counselling by a practitioner with a good understanding of the technology and issues that could arise**
- **Limit screening to the common trisomies and consideration of sex chromosomes if the woman wishes and understands the change in screening characteristics (increases false positive rate)**
- **Post-test counselling is available**
- **Invasive testing where there is a high risk result prior to making any decision to not continue the pregnancy**
- **Proceed with a scan at 11 to 14 weeks gestation as this has other benefits**

For more information from RANZCOG go to:

https://www.ranzcog.edu.au/womens-health/college-communiques/1357-dna-based-noninvasive-prenatal-testing-for-fetal-aneuploidy.html