Criteria and Pathway for Funded NIPT

Te Whatu Ora
Health New Zealand
Te Toka Tumai Auckland

Government-funded antenatal screening for Down Syndrome (Trisomy 21) and other conditions (mainly Trisomy 18 and Trisomy 13) is available in Aotearoa to all pregnant women eligible for funded care. There are two Government-funded screening options – first trimester combined (also known as MSS1) which is performed prior to 14 weeks, and second trimester maternal serum screening (also known as MSS2) which is performed between 14 and 20 weeks of pregnancy. First trimester combined screening involves a maternal blood test (taken between 9 weeks and 13 weeks and 6 days of pregnancy) to examine maternal hormone levels, along with an ultrasound scan assessing the nuchal translucency (performed between 11 weeks and 2 days and 13 weeks and 6 days of pregnancy). Second trimester maternal serum screening involves a blood test only to examine maternal hormone levels.

Non-invasive prenatal testing or screening (NIPT or NIPS) is an alternative to the Government-funded antenatal screening for Down Syndrome and other conditions, and its use in Aotearoa to date has largely been as a self-funded test. NIPT is an established technology, which extracts placental cell-free DNA from a maternal blood sample. It can be performed from 10 weeks gestation onwards. As placental DNA and fetal DNA are usually identical, this test can be used to screen for chromosome differences with a much higher sensitivity than MSS1 and MSS2. NIPT is not currently part of the Government-funded screening programme and this has created inequity in care, as not every pregnant person can afford this option.

From June 2023 it has been agreed that National Women's Health will fund NIPT for a cohort of women who have been screened through the government-funded pathway and have received an '***increased chance'*** result. NIPT will be funded for the following:

- Women who have received an 'increased chance' result from MSS1 of between 1: 20 to 1:300, with an NT measurement of <3.5mm
- Women who have received an 'increased chance' result from MSS2 of between 1:20 to 1:300
- Women with an 'increased chance' result from MSS1 or MSS2 with a chance higher than 1:20 would be recommended to proceed to diagnostic testing with chorionic villous (CVS) or amniocentesis, however those that decline diagnostic testing will still be able to access funded NIPT.

In order to access funded NIPT women will need to be referred to the Auckland Fetal Medicine Service via the usual pathway. This pathway applies to women in the catchment areas covered by the Auckland Fetal Medicine Service (currently Northland, Auckland and Waikato areas – this may change if boundary areas are redefined).

Women who have an NT measurement ≥3.5mm will continue to be offered an appointment in Fetal Medicine Clinic for detailed review of early anatomy and to discuss appropriate testing options, including diagnostic testing.

Women with an increased chance MSS1 with an NT<3.5mm, or increased chance MSS2 result will receive care via the following pathway:

- Lead maternity carer (LMC) or general practitioner to refer to the Fetal Medicine Service via the usual pathway.
- The referral will be triaged to a Midwife Telehealth Clinic.
- Appointments in Midwife Telehealth Clinic will involve a phone discussion of available testing options. Outcome from this discussion will be one of the following:
    - No further testing wanted by the woman; LMC to arrange routine anatomy scan in the community at around 20 weeks.
    - Diagnostic testing requested; Fetal Medicine midwife refers to the CVS/amniocentesis clinic in National Women's Ultrasound.
    - NIPT requested; Fetal Medicine midwife arranges an appointment for the blood sample to be taken or refers to a local collection centre.
- Results of CVS/amniocentesis/NIPT will be followed up by Fetal Medicine team. Women with low chance/normal results will be discharged following discussion of the results with the woman. Women with high chance NIPT results will be brought to a Fetal Medicine Specialist Clinic for an ultrasound scan and further discussion about diagnostic testing options. Abnormal CVS/amniocentesis results will be discussed with the woman and her LMC and follow up arranged depending on individual circumstances and woman's wishes (this may involve referral to Genetics, Fetal Medicine Specialist Clinic or a termination service).

Referral Pathway for Funded NIPT

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