Document No: **2773067**

TITLE: Amniocentesis

1. Purpose

To describe the process for the referral, triaging, counselling, and care of women who request diagnostic testing for fetal chromosomal conditions at Te Whatu Ora Lakes (Lakes).

In recognition of Te Tiriti o Waitangi (the Treaty of Waitangi) and the Crown's special relationship with Maori, Te Whatu Ora – Lakes, is committed to acknowledging the Treaty by working in partnership with Maori. Staff involved in implementing this policy should be aware of the Tiriti o Waitangi Policy (EDMS 40583).

2. Background

In Aotearoa, New Zealand, pregnant women/people are offered screening to assess whether their unborn baby may have Trisomy 21, or other rare chromosomal conditions. If a pregnant woman/person returns a high chance result (>1:300) they may choose to undergo diagnostic testing. They may also be offered diagnostic testing if there are other reasons to suspect a high chance of chromosomal or genetic difference (e.g. structural differences identified on ultrasound).

Currently available screening tests:

- Combined first trimester screening (CFTS): This test includes an ultrasound scan and maternal serum analytes between 9 weeks and 13^{+6} weeks. This test is fully funded, although women may be asked to pay a part-charge for the ultrasound component.

- Second trimester serum screening (MSS2): This test reviews maternal serum analytes between 14 and 20 weeks' gestation. This test is fully funded.

- Cell-free DNA (cfDNA), also known as Non-Invasive Prenatal Screening (NIPS): A screening test with very high sensitivity and specificity (98.8%, and 99.96%). This test is not currently funded. It is more accurate than the currently funded tests, such that 92% of women with a positive screen will have a pregnancy affected with a chromosomal difference. Women may choose to have this test as a first-line test instead of CFTS or MSS2, or as a second line test if the CFTS/MSS2 tests return a high chance result.

Diagnostic tests:

- Chorionic Villus Sampling (CVS): This test can be performed from 11+0 to 13+6 weeks' gestation. The additional risk of miscarriage is slightly higher than with amniocentesis. It is available through Maternal Fetal Medicine in Auckland and Wellington.

- Amniocentesis: This can be performed after 15+0 weeks' gestation, with an additional risk of miscarriage of 0.3%. This test is available through Lakes, Auckland, and Wellington.

  There are two types of laboratory tests available on amniocentesis fluid: Fluorescence In Situ Hybridisation (FISH), and a microarray. While FISH results will be available in 72 hours, it only targets specific areas on chromosomes 13, 18, 21, X, and Y. The microarray takes 2-3 weeks for a full result.

Contraindications to amniocentesis

There are very few situations where amniocentesis is absolutely contraindicated, however there are cases where the risks may outweigh the benefits. In such cases consultation with a tertiary service is recommended prior to amniocentesis. This may include referrals where:

- A referral is received prior to 13^{+6} (and CVS may be considered in the first instance)
- Multiple gestations (e.g. twins, triplets)
- Women who are HIV positive
- Women with Red Cell Antibodies implicated with Haemolytic Disease of the Newborn
- Women with a bleeding disorder/anticoagulation
- Gestations after 21 weeks
- Amniocentesis for other fetal-indicated conditions (e.g. structural anomaly)

3. Scope

All employed staff within Te Whatu Ora Lakes who provide care to pregnant women/people and Lead Maternity Carers (LMC's).

4. Definitions

ANC	Antenatal Clinic
CFTS	Combined First Trimester Screening
CVS	Chorionic Villus Sampling
EDD	Estimated Date of Delivery
FISH	Fluorescence In Situ Hybridisation
LMC	Lead Maternity Carer
NIPS	Non-Invasive Prenatal Screening
MSS2	Maternal Serum Screening Second Trimester
SMO	Senior Medical Officer

5. Procedure

5.1 Referral

All pregnant women/people with a high chance of chromosomal difference should be offered NIPS and diagnostic testing.

Women/people who wish to have a diagnostic test, or a consultation about their testing options should be referred to the Lakes Antenatal Clinic using the Amniocentesis referral form EDMS 2773053

Referrals may be accepted from out-of-area health care providers.

Referrals will be triaged in the Lakes Antenatal Clinic by an SMO Obstetrician.

Incomplete referrals will not be accepted. The minimum data-set includes:

- Patient NHI and contact details
- Referring clinician and contact details
- Reason for request, and copies of any relevant test results
- Blood Group

The woman/person and their referring clinician should receive a receipt of referral, and information on when they may expect to be seen.

The woman/person will be offered a telehealth appointment, booked in the Amniocentesis Clinic, prior to amniocentesis unless;

- The referral indicates they want/require face-to-face assessment
- They are over 17 weeks at the time of referral and a telehealth appointment will provide undue delay for an in-person review.

This initial telehealth consultation should be scheduled within 2 weeks of the receipt of referral, or at 15 weeks, whichever is later, to discuss testing options.

- If this timeframe is not possible the Women's health clinic nurse/ administrator should discuss the case with the clinician performing amniocentesis
- If the timeframe is not possible due to the absence of a clinician performing amniocentesis, the Women's health clinic should re-direct the referral to the tertiary Maternal Fetal Medicine Service

The woman/person will be offered an appointment for amniocentesis after 15 weeks' gestation.

If a woman/person chooses to proceed with amniocentesis, a 1-hour appointment will be scheduled by the Women's Health Clinic Administrator in the Lakes Amniocentesis Clinic and a Prenatal Testing Information Booklet EDMS 2773055 will be sent to the woman/person.

Amniocentesis will be performed by a suitably skilled health professional with ongoing clinical audit in accordance with international guidelines.

5.2 Amniocentesis Procedure:

Nursing responsibility

- Confirm maternal blood group and rhesus factor
- Confirm the required equipment is available
- Confirm the minimum dataset is available for SMO Obstetrician to review
- Complete the Amniocentesis Record Sheet EDMS 2773060
- Send specimen to Pathlab Rotorua: and ensure it arrives by 3pm. N.B.: it **must not** be sent in the Lamson.

Obstetric responsibility

- Review for contraindications for amniocentesis in a secondary service
- Provide genetic and procedure related counselling
- Ensure questions are answered
- Confirm gestation
- Obtain written consent to perform the amniocentesis procedure
- Perform ultrasound scan and amniocentesis if the woman consents
- Document the procedure, outcome and any advice given or follow-up required
- Complete lab request form - noting a copy of the results to be sent to the referring clinician
- Complete ultrasound report
- Prescribe Anti-D, if indicated

Post-procedure

- If Rhesus Negative Women's Health Clinic Nurse is to give the prescribed Anti-D as per protocol
- Woman/person may be discharged home immediately after the procedure if comfortable
- Women's Health Clinic administrator is to send a copy of the amniocentesis report to the referrer.
- Women's Health Clinic Nurse is to inform the referrer of the outcome of the procedure
- Instruct the woman/person to phone their LMC, General Practitioner or local hospital if any of the following occur;

    - Pain not settling with paracetamol
    - Vaginal bleeding
    - Rupture of membranes

5.3 Results / Follow-Up:

Responsibility of the Amniocentesis Service**:**

- The amniocentesis service will contact the woman/person at 48-72 hours and 2-3 weeks post-procedure for a well-being check, and to discuss the test results.
- Provide information about what the test results mean and options available
- Refer to counselling services, if required/requested
- Arrange for a copy of the amniocentesis results to be sent to the referring clinician.

Responsibility of the Referring Clinician

- Arrange a formal 18 – 20 week anatomy ultrasound scan (only a limited scan is performed at the time of amniocentesis).
- The referring clinician is requested to inform the Lakes Women's Health Clinic of any adverse events in the two weeks following amniocentesis, to allow for robust monitoring and quality assurance: this includes
    - Threatened miscarriage or miscarriage
    - Preterm pre-labour rupture of membranes

6. Equipment to Be Used

- Sterile gloves
- Amniocentesis Record Sheet Ref. XXXXXX
- Signed consent form
- USS Machine
- Ultrasound Gel
- Sterile wash
- Sterile dressing pack
- 22 gauge Spinal needle (Quinke tip)
- Sterile Pottle
- Sterile 20ml Syringe
- Sterile USS probe covers
- Sterile gel
- Sterile drape for trolley
- Sterile dressing pack
- Sterile drape for patient
- Extension set
- Bandaid

7. Clinical Audit and Quality Assurance

The following aspects of this service may be audited for quality assurance purposed;

- Rate of pregnancy loss within 14 days of procedure (<0.5%)
- Rate of procedures requiring more than one needle insertion ('pass')
- Rate of procedures with failure to obtain adequate sample
- Proportion of lab failure to provide test result despite an adequate sample (<0.5%)
- Anti-D administration for women who are rhesus negative (100%)

8. Related Documentation

- Referral Form EDMS 2773053
- Amniocentesis Record Sheet EDMS 2773060
- Prenatal Testing Information Booklet EDMS 2773055

9. References

1. National Screening Unit. Antenatal Screening for Down Syndrome and Other Conditions. https://www.nsu.govt.nz/pregnancy-newborn-screening/antenatal-screening-down-syndrome-and-other-conditions/about-test [Accessed January 2023].
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4. Rose, Nancy C., et al. Systematic evidence-based review: The application of noninvasive prenatal screening using cell-free DNA in general-risk pregnancies. *Genetics in Medicine*. (2022).
5. Navaratnam, K., Z. Alfirevic, and Royal College of Obstetricians and Gynaecologists. "Amniocentesis and chorionic villus sampling: Green-top Guideline No. 8." BJOG: An International Journal of Obstetrics & Gynaecology 129.1 (2022): e1-e15.

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