Antenatal Screening for Down Syndrome and Other Conditions

Guidelines for health practitioners

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Te Kāwanatanga o Aotearoa
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Health New Zealand
Te Whatu Ora

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Manaakitia te māhuri

he tupuna kei roto

Nurture and take care of the future that grows within

Contents

Foreword

Health New Zealand |Te Whatu Ora[1] (Health NZ) is responsible for the development, implementation, and management of two newborn screening programmes:

- Newborn Metabolic Screening Programme.
- Universal Newborn Hearing Screening and Early Intervention Programme

Health NZ is also responsible for the introduction of quality improvements to Antenatal Screening for Down syndrome and other conditions.

While all wāhine are advised about all available screening, participation is optional.

Information about screening for Down syndrome and other conditions should be offered to all wāhine during pregnancy. This will enable people to make an informed decision and to receive screening results for their pēpi to help inform and plan accordingly.

These guidelines replace the *Antenatal Screening for Down Syndrome and Other Conditions: Guidelines for Health Practitioners* dated 2013.

Contact details for support services and sources of further information about Antenatal Screening for Down Syndrome and Other Conditions are listed in Appendix I – Resources and contacts.

Acknowledgements

These Guidelines were developed in 2012 and were updated in 2024.

Health NZ thanks the Review Panel and the many individuals and groups who contributed to the development of this document.

[1] What was previously known as the National Screening Unit (NSU) has now been incorporated into the Prevention Directorate within the National Public Health Service of Health New Zealand.

Key messages

1. Antenatal screening for Down syndrome and other conditions is optional for pregnant wāhine and information should be provided to support the screening discussion, thus enabling wāhine to make an informed decision whether to accept or decline. Cultural and linguistic diversity should be acknowledged, and care provision/resources should be responsive to the needs of the individual.
2. Antenatal screening is a way of assessing the chance that pēpi has Down syndrome (trisomy 21), Edwards syndrome (trisomy 18), Patau syndrome (trisomy 13) or other rare genetic conditions. It offers wāhine and whānau information and choice in the care and management of their pregnancy and birth.
3. No single test checks for everything. No screening test finds all cases of a condition.
4. A thorough family history should be taken and where there is a relevant family history a referral for a discussion with a specialist obstetrician or genetic services should be offered in addition to the offer of screening.
5. First trimester combined screening can be completed between 9 weeks and 13 weeks 6 days gestation. **The recommended timing for the blood test is 9-10 weeks and for the nuchal translucency scan it is 12-13 weeks.**
6. Second trimester maternal serum screening can be completed between 14 weeks and 20 weeks gestation. **The recommended timing for the blood test is 14 to 18 weeks.**
7. The referring practitioner is responsible for timely offer of screening, accurate completion of the request form and follow up of screening results. Clear documentation of the screening process must be kept in the clinical records including the discussion, consent or decline of tests or referrals and results of screening.

List of abbreviations

Abbreviation	Phrase
AFP	Alpha-fetoprotein
ASUM	Australasian Society for Ultrasound in Medicine
ßhCG	Beta-human chorionic gonadotropin
BPD	Biparietal diameter
cffDNA	Cell free fetal DNA
CRL	Crown–rump length
CVS	Chorionic villus sampling
DNA	Deoxyribonucleic acid
EDD	Estimated date of delivery
FMF	Fetal Medicine Foundation
FTCS	First trimester combined screening
IVF	In vitro fertilisation
LMC	Lead maternity carer
LMP	Last menstrual period
MoM	Multiple of the median
MSS1	Maternal serum screening 1 (1st trimester)
MSS2	Maternal serum screening 2 (2nd trimester)
NIPS	Non-invasive prenatal screening
NTD	Neural tube defect
NT	Nuchal translucency
NZCOM	New Zealand College of Midwives
NZDSA	New Zealand Down Syndrome Association
PAPP-A	Pregnancy-associated plasma protein A
RANZCOG	Royal Australian and New Zealand College of Obstetricians and Gynaecologists
RANZCR	Royal Australian and New Zealand College of Radiologists
uE3	Unconjugated oestriol

Glossary of Māori terms[2]

Te reo Māori	English
Aotearoa	New Zealand
Hapū	Subtribe, pregnant
Hapūtanga	pregnancy
Hui	gathering, meeting, discussion
Iwi	tribe
Kaitiakitanga	guardianship, including stewardship; the processes and practices of looking after the environment
Karakia	prayer, incantation, blessing
Kaupapa Māori	a philosophical doctrine incorporating the knowledge, skills, and values of Māori
Koha	gift, present, offering, donation
Kōpū	belly, womb, abdomen
Kōrero	conversation, narrative, speech, discourse
Kuia	elderly woman, grandmother, grand aunt
Kupu	word
Mana	inherent authority and dignity
Mana motuhake	an individual's authority to determine their own destiny, self-determination
Mana wāhine	an approach that privileges Māori women, heritage, and prestige
Mana whenua	the people of the land who have mana or customary authority; their historical, cultural, and genealogical heritage are attached to the land and sea
Māori	Indigenous New Zealander, Indigenous person of Aotearoa
Mātauranga	knowledge
Mauri	life force
Oriori	birth chants
Papatūānuku	Mother Earth
Purākau	stories

[2] Moorfield, J.C. (2024) Te Aka Māori Dictionary.

Te reo Māori	English
Pēpē	baby/babies (South of Puketutu (Bombay))
Pēpi	baby/babies (Northern regions)
Rangatahi	youth
Taonga	a treasure
Tangata whenua	Indigenous people of the land
Tapu	sacred
Te Tiriti o Waitangi	The Treaty of Waitangi, which is the document upon which the British and Māori agreed to found a nation state and build a government
Te reo Māori	the Māori language
Tikanga	cultural practice, Māori protocols
Tinana	the body, main part of something or someone
Tupuna/Tūpuna	ancestor(s), grandparent(s); has the same meaning as tipuna/tīpuna (pl)
Wahine	woman
Wāhine	women
Whakapapa	ancestral lineage
Whānau	family, the smallest social unit of Māori groupings; to be born/give birth (therefore includes pregnant women). Also used to refer to parents/guardians.
Whenua	land, country, earth, ground; placenta

Editorial language

In commitment to tāngata whenua and te Tiriti o Waitangi, te reo Māori is prioritised in this document. To maintain narrative flow, the editorial style will refer to wāhine, pēpē/pēpi, and whānau. These terms encompass all priority groups and ethnicities.

Health NZ acknowledges and respects gender diversity within the birthing population of Aotearoa New Zealand, including trans and non-binary people. Culturally safe practice includes health practitioners respecting and engaging with each individual receiving care and adapting their use of language accordingly in practice.

In a medicolegal context it is essential to have clarity about the specific individuals to whom the health practitioner is providing clinical care. For antenatal screening this is the wāhine hapū (pregnant woman) and the pēpi (baby/s). The foundational principle of informed consent requires the health practitioner to protect and support the right of the individual receiving care to exercise rangatiratanga (self-determination) and mana motuhake (bodily autonomy) when navigating health care decision making.

1. Introduction

1.1 Purpose

The purpose of this guideline is to provide health practitioners with clear, concise, and consistent guidance about antenatal screening for Down syndrome and other conditions in Aotearoa New Zealand. They are intended for all practitioners involved in aspects of antenatal screening for Down syndrome and other conditions.

1.2 Target audience

This guideline is written for health practitioners providing antenatal care in Aotearoa New Zealand. Health practitioners should use it to support clinical judgement, knowledge and expertise and provide for a timely, consistent, and effective approach to offering and providing antenatal screening for Down syndrome and other conditions. Screening participants and their whānau can use this guideline to understand the antenatal screening pathway.

The target audience includes but is not limited to:

- Midwives
- Lead Maternity Carers
- General Practitioners
- Nurses
- Obstetricians
- Fetal medicine specialists
- Radiologists, Sonographers and Sonologists
- Laboratory staff

1.3 Guiding principles

- **Informed choice:** The decision to engage in screening is an individual one and wāhine have the right to make an informed choice to accept or decline screening. Informed consent is a foundational principle of the quality initiative for Antenatal Screening for Down Syndrome and Other Conditions.
- **Health consumer rights:** The Code of Health and Disability Services Consumers' Rights provides that Aotearoa New Zealand healthcare consumers have a legal right to appropriate information to enable them to give informed consent. During the screening process, the health practitioner is responsible for providing information and education about antenatal screening to wāhine hapū (pregnant women), referring appropriately, and receiving and communicating screening results.
- **Cultural safety:** Health services should be tailored to meet the health needs of each individual, recognising the responsibility to support health equity for all,

including Māori and Pacific Peoples. Health practitioners should recognise that what works for different populations varies and can familiarise themselves with current health strategies and health plans developed by Health NZ and the Ministry of Health | Manatū Hauora (Ministry of Health). See also Te Tatau o te Whare Kahu | Midwifery Council Statement on Cultural Competence for Midwives.

- **Equity of access:** Health practitioners should be aware that barriers to accessing aspects of antenatal care and screening may include lack of knowledge, mistrust of health services, different cultural views of health, the location and cost of ultrasound services, access to funded maternity care, the availability of transport, travel time and childcare.
- **Rights of disabled persons:** Health practitioners should offer additional support to wāhine and whānau who have difficulty understanding information because of language difficulties, hearing impairment, intellectual disability, or other disabilities. Further information can be found on the Whaikaha (Ministry of Disabled People website).
- **Protecting privacy:** The Health Information Privacy Code 2020 (HIPC) sets specific rules for agencies in the health sector to ensure the protection of individual privacy. It addresses health information collected, used, held, and disclosed by health agencies. The HIPC requires agencies to be clear about the purpose for which they collect information, and open about those purposes to the health consumers from whom it is collected. Health information must be held securely to protect it against misuse, loss, or unauthorised disclosure. Health consumers can access their health information (with some minor exceptions) and seek its correction when it is wrong. Health information should only be used or disclosed for the purposes for which it was collected, unless one of the exceptions in the HIPC applies.
- **Professional accountability:** Health practitioners providing lead maternity care have an obligation under the Primary Maternity Services Notice 2021, issued pursuant to Section 94 of the Pae Ora (Healthy Futures) Act 2022, to advise wāhine of screening services available that are endorsed by Health New Zealand, including antenatal screening for Down syndrome and other conditions.
- **Legislation:** Health practitioners are responsible for documentation of screening discussions and choices in the clinical notes, ensuring compliance with the:
    - Privacy Act 2020 and Health Information Privacy Code 2020
    - Pae Ora (Healthy Futures) Act 2022
    - Code of Health and Disability Services Consumers' Rights 1996
    - Health Act 1956
    - Health Practitioners Competence Assurance Act 2003
    - Public Records Act 2005
    - Abortion Legislation Act 2020

Other screening resources are available for health practitioners at Health New Zealand | Te Whatu Ora.

1.4 Te Tiriti o Waitangi statement

Our Te Tiriti o Waitangi obligations are fundamental to screening initiatives in Aotearoa New Zealand. This requires a strong focus on the principles found in Te Tiriti o Waitangi and must align with the transformations called for in the Wai 2575 Hauora report of the Waitangi Tribunal. As identified in the Hauora report, the Waitangi Tribunal proposed the framework of Te Tiriti o Waitangi principles be adopted for the primary health care system, inclusive of tino rangatiratanga, equity, active protection, options, and partnership. These principles can also be adopted for population-based screening programmes.

The Waitangi Tribunal concluded that persistent health inequities that Māori experience were the consequence of the failure to apply the principles of Te Tiriti at structural, organisational and health practitioner levels of the health and disability sector. Giving effect to Te Tiriti requires health practitioners to know the principles of Te Tiriti and to capably apply these in partnership with Māori in their day-to-day maternity clinical practice. Applying the principles to maternity service delivery is vital to enabling Māori to express their mana[3] and to receive high-quality, culturally safe care, and achieve equitable health outcomes.

How these principles apply to health services is supported by *Ngā paerewa* and, in particular, Pae ora healthy futures.

For the health and disability sector, the principles of Te Tiriti are as follows:

- **Tino rangatiratanga**: Health practitioners support the right of Māori to receive effective maternity care, conceptualising the decisions of the wāhine as a continuation of a much older, Māori collective-endorsed practice of self-determining one's own health and wellbeing and that of the whānau.
- **Equity**: Health practitioners can contribute to equitable health outcomes for Māori by ensuring that, at a minimum, maternity outcomes match those of other New Zealanders. Equitable maternity outcomes will be achieved when health practitioners work in ways that give effect to the principles of Te Tiriti and *Ngā paerewa*. The principle of equity requires the Crown to commit to achieving equitable health outcomes for Māori.
- **Active protection**: Health practitioners should share evidence-based information about maternity outcomes so that Māori can make decisions and prepare themselves to uphold their tikanga (for example, karakia, rongoā, support people). Health practitioners must actively support Māori to make decisions that are best for them.
- **Options**: Health practitioners ensure Māori have maternity care that enables them to uphold their tikanga regardless of where birth takes place. Processes must complement a Māori person's mana, support their tikanga, and be culturally safe as defined by Māori.

[3] See Ministry of Health's Te Tiriti o Waitangi Framework for the Ministry's four goals, each expressed in terms of mana. URL: https://www.health.govt.nz/system/files/documents/pages/whakamaua-tiriti-o-waitangi-framework-a3-aug20.pdf (accessed 2 February 2022).

- **Partnership**: Health practitioners work in partnership with Māori, including their whānau as defined by them. A partnered approach to the process and decision-making ensures Māori can enact their rangatiratanga while exercising mana Motuhake (authority over their bodies and reproductive health).

1.5 Equity

In Aotearoa New Zealand, people have differences in health outcomes that are not only avoidable but unfair and unjust.[4] Differences in the structural determinants of health and wellbeing (for example, disadvantages in income, employment, education, and housing, as well as multiple forms of discrimination) negatively impact people's health but people have little control over these. Health inequities are not about people; instead they are the result of avoidable structural determinants in our communities.[5] When health practitioners understand the structures that create inequitable maternity outcomes, they can use different approaches and resources to achieve equitable health outcomes.

Achieving equitable health outcomes for all happens when health service providers and health practitioners:

- understand the structures that create disadvantage for those groups
- are supported to work in ways that give effect to the principles of Te Tiriti, as well as meeting professional competencies and *Nga paerewa*.

Lastly, health practitioners should be aware that many people in Aotearoa New Zealand conceptualise anatomy, pregnancy, gender, sexuality, reproduction, contraception, and birth in diverse ways according to their worldviews. Therefore, health practitioners should use proven health literacy practices[6] to communicate effectively with everyone using their services (for sector guidance, see *Ngā paerewa* Standard 1.4 E whakautetia ana ahau | I am treated with respect and criteria 1.4.2).

[4] Ministry of Health. 2019. *Achieving Equity*. URL: https://www.health.govt.nz/about-ministry/what-we-do/work-programme-2019-20/achieving-equity (accessed 2 February 2022).
[5] Toi Te Ora Public Health. 2021. *Determinants of Health and Health Equity*. URL: https://toiteora.govt.nz/public/determinants-of-health-and-health-equity/ (accessed 2 February 2022).
[6] Ministry of Health. 2015. A framework for health literacy. URL: https://www.health.govt.nz/publication/framework-health-literacy (accessed 2 February 2022).

2. Background

Antenatal screening for Down syndrome and other conditions has been available to pregnant wāhine in Aotearoa New Zealand since 1968. In October 2007, the government agreed to implement quality improvements to antenatal screening for Down syndrome and other conditions to ensure consistency with international best practice at the time. The objective of the quality improvements initiative is to ensure that the screening programme provides information so that wāhine can make an informed decision about their pregnancy.

Antenatal screening for Down syndrome and other conditions is a way of assessing the chance that pēpi have Down syndrome or another genetic condition and offers wāhine and whānau information that enables choice in the care and management of their pregnancy and birth. Antenatal screening for Down syndrome and other conditions has complex ethical and social implications.

Detection of fetal anomalies through this screening offers whānau information that may help them prepare for the birth of their child, for example: the option of giving birth in a setting that has access to specialist surgical or medical care; the possibility of considering termination of pregnancy; or palliative care in the newborn period for pēpi with a poor prognosis.

People may have differing views on screening in pregnancy based on their cultural, ethical, and spiritual beliefs, and it is important to acknowledge this when supporting whānau with decision-making.

3. Te Ao Māori

Within a Māori worldview screening and the sampling of Māori DNA (and its association to whakapapa and te ao Māori) have important cultural values and hold a sacred significance for Māori. Wāhine and their ability to give birth have special significance in te ao Māori. This ability to continue whakapapa is celebrated, making childbirth one of the most important traditions in society. In Māoridom the practice of oriori while the child is in the womb is seen as an important aspect of connecting to whakapapa and acknowledging the past, present, and future aspirations of the child growing within the womb. Traditional Māori birthing practices focus on the importance of conveying ancestral journeys, stories, and achievements of ancestors through karakia and oriori. This practice gives the unborn child something to aspire to, and encourages a lifelong pursuit of learning, prosperity, and aroha for pēpi and whānau. This grounds the child through whakapapa to the land, and to Papatūānuku. The Tuku Iho app resource can be used for discovering oriori and kaupapa Māori antenatal resources.

Maternal figures are prominent in Māori cosmology and kōrero tuku iho (narratives passed down by Māori through oral tradition). These pūrākau (stories) form the basis of many tikanga pertaining to wāhine Māori and their birthing rites.

Health practitioners in Aotearoa New Zealand should ensure they are aware of the cultural importance of blood for Māori and its significant association to whakapapa. Blood is considered tapu (sacred) in te ao Māori and the storage or return of samples collected for screening must be fully explained with options provided for returning the sample to the whānau.

A Māori perspective on ‘disability’ may focus on the unique strengths of the person affected rather than a deficit mindset. Health practitioners should ensure they are aware of potential cultural differences for any whānau and choose their language carefully when counselling whānau.

For more information see:

Hakui website

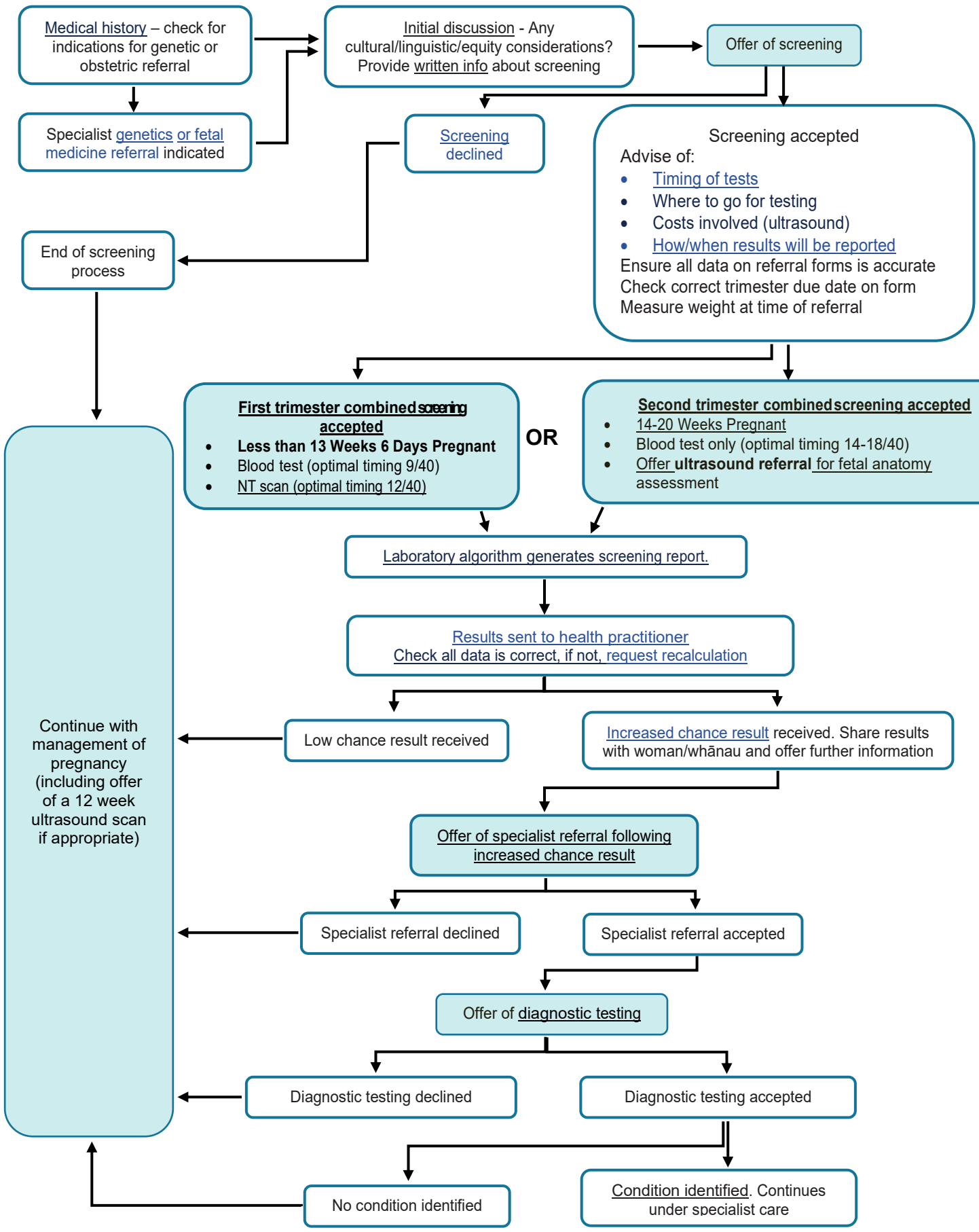
Turanga Kaupapa

Tuku Iho app

Whānau hauā: Reframing disability from an Indigenous perspective

4. The screening pathway

Figure 1: the Screening Pathway



Engaging in screening is a personal choice, and people may accept or decline any referral or test.

The points along the pathway where wāhine need to make an informed decision are:

a) when there is a relevant family or obstetric history: whether to accept an offer of referral to a specialist or Genetic Services to gather more information

b) whether to accept the offer of screening

c) when there is an increased chance screening result, whether to accept an offer of referral to a specialist or Genetic Services to gather more information. This should occur following the provision and explanation of screening results by the LMC (or the referring health practitioner)

d) whether to have diagnostic testing following specialist referral and discussion

e) when deciding the next step after receiving the results of the diagnostic test

4.1 The screening options

Antenatal screening for Down syndrome and other conditions is a way of assessing the chance that pēpi have Down syndrome (trisomy 21), trisomy 18 (Edwards syndrome), trisomy 13 (Patau syndrome) and some other rare genetic disorders.

First trimester combined screening involves an ultrasound scan to measure the nuchal translucency (NT) and maternal serum testing and is often referred to as MSS1 (Maternal Serum Screening 1). The report is calculated by the screening laboratory from the NT measurement, the serum marker levels and other factors including crown-rump length, maternal age, and weight. Wāhine will receive one combined result from their health practitioner after they have had **both** the blood test and the NT scan. The result is presented as a natural frequency which describes the chance that pēpi has a certain condition, for example 1 in 500.

Second trimester screening involves maternal serum testing only. The results of the serum tests are incorporated with other parameters such as maternal age, weight, and gestation to provide a natural frequency estimate as for first trimester screening.

Both first and second trimester screening will provide one of two results: either increased chance or low chance.

The option of screening during the second trimester means screening can be offered to wāhine who:

- did not access maternity care early in their pregnancy
- have not completed first trimester combined screening
- prefer second trimester screening
- were unable to access ultrasound scanning in the first trimester

In Aotearoa New Zealand, the threshold for increased chance is 1:300. As an example, wāhine with a result of 1:250 will be noted as having an increased chance of having a baby with the screened for condition and 1:350 will be low chance of having a pēpi with the screened for condition.

In 2020, 4.2% of wāhine screened in Aotearoa New Zealand received an increased chance result[7]. Of those with an increased chance result, 6.1% were true positives. So, if a woman received an increased chance result, there was a 6.1% probability that she was carrying a fetus with trisomy 21, 18 or 13.

In 2020 this screening's detection rate (sensitivity) for identifying pēpi with Down syndrome was 84%. This meant that 16% of pēpi with Down syndrome were not detected through this screening.

Figure 2: Screening options

Screen	Description	Funding	Availability	Optimal timing
MSS1 **First trimester combined screening** **(9 weeks – 13 weeks and 6 days)**	Blood test that measures two maternal serum markers (PAPP-A and ßhCG) combined with an ultrasound scan to determine NT and CRL measurements	Fully funded blood test Surcharge may apply for ultrasound scan	Available to all eligible wāhine who present in the first trimester	The optimal timing for the first trimester blood test is **9-10 weeks** The optimal timing for the nuchal translucency scan is **12-13 +6 weeks**
MSS2 **Second trimester maternal serum screening** **(14 - 20 weeks)**	Blood test that measures four maternal serum markers (ßhCG, AFP, uE3 and inhibin A)	Fully funded blood test	Available to eligible wāhine who present after the first trimester or who do not access first trimester combined screening	The optimal timing for the second trimester blood test is **14 - 18 weeks**

[7] https://www.nsu.govt.nz/system/files/page/antenatal-screening-for-down-syndrome-and-other-conditions-2019-report.pdf

4.2 Non-Invasive Prenatal Screening (NIPS)

As a primary screening test non-invasive prenatal screening (NIPS) is currently a user pays, non-scheduled test. Health NZ funds first trimester combined screening (MSS1) and second trimester maternal serum screening (MSS2) for all eligible wāhine (note there may be a surcharge for ultrasound screening).

NIPS is a way of determining the chance of pēpi having certain chromosomal abnormalities such as trisomy 21 (Down syndrome), trisomy 18 (Edwards syndrome) and trisomy 13 (Patau syndrome) as well as other conditions. Most NIPS tests offer fetal sex and sex chromosome aneuploidy detection in addition to trisomies 21, 18 and 13. The accuracy of the test changes depending on the disorder in question.

Non-invasive prenatal screening analyses small fragments of circulating fetal DNA in the maternal blood. These fragments are free floating and not within cells (unlike most DNA) and are therefore known as cell-free fetal DNA (cffDNA). During pregnancy, cffDNA from the placenta are found in the maternal blood. Evaluation of the cffDNA from the placenta allows screening for certain genetic anomalies.

NIPS has the highest sensitivity and specificity of all the screening tests for Down syndrome and can be performed reliably from 10 weeks of pregnancy. NIPS performs better than MSS1 for aneuploidy detection therefore **concurrent MSS1 screening is not recommended**. A wāhine may choose to have NIPS (non-funded) following combined screening assessment.

NIPS is considered a screening test. It does not replace current diagnostic testing using amniocentesis or chorionic villus sampling. **Specialist referral** is recommended when:

- there is known fetal abnormality
- there is a significantly raised nuchal translucency (NT)
- an increased chance result from MSS1 or MSS2 is reported

In these situations NIPS may be offered by specialist services following initial consultation with whānau, however **a private NIPS without specialist input is not appropriate.** LMC's should check with their local specialist service (ie, Maternal Fetal Medicine) to determine whether funded NIPS is offered to whānau who receive an increased chance screening result.

If wāhine choose to have NIPS (non-funded) as their primary screening test, knowledge of dates is required. **They should still be offered an ultrasound scan** at 12 to 13 weeks gestation for an early structural assessment, as 50 percent of major abnormalities can now be detected at this gestation. However, NT measurement (and combined screening assessment) is not recommended in wāhine with a previous NIPS result (Note: **The NT should still be measured if it appears increased**, and Fetal Medicine or other local equivalent specialist referral should be offered if NT is ≥ 3.5 mm.).

In a proportion (1-6%) of cases, NIPS is unable to provide a result. These “no call” results appear to have a higher chance of fetal abnormalities (eg, triploidy). These wāhine should be offered follow up specialist assessment including detailed ultrasound (if not already performed), and be offered the options of diagnostic testing, repeat NIPS testing (successful in approximately 50% of cases), or an alternative form of screening such as combined first trimester screening.

For further information, see:

- NIPS information in Appendix 5 of the New Zealand Obstetric Ultrasound Guidelines
- Health NZ’s Position Statement on Non-Invasive Prenatal Screening (NIPS)
- the NZMFMN Statement on the use of Non-Invasive Prenatal Testing (NIPT) (PDF, 259 KB) (NZMFMN 2016)
- What is non-invasive prenatal testing (NIPT) and what disorders can it screen for?
- RANZCOG statement: Prenatal screening and diagnostic testing for fetal chromosomal and genetic conditions

4.3 Initial discussion

Initial discussion should include all of the following points:

(a) Screening information

- purpose of screening
- screening options available and what screening involves
- screened conditions
- the NT scan will include early anatomy and structural development assessment
- screening does not cover every condition
- incidental information may be found relating to the pregnancy or pēpi
- recommended timing for screening
- the screening pathway and the decision points and options for screening
- the screening result is presented as the estimated chance your pēpi may have one of the conditions and further testing is required for diagnosis if there is an increased chance result
- reliability of screening - the screening can detect approximately 8 to 9 out of every 10 pēpi with Down syndrome but will miss 1 to 2 in 10. Screening may result in a false positive result.
- which tests may incur charges
- further information about pēpi may be identified at the second trimester anatomy scan.

(b) Resources

- the consumer resource, Antenatal screening for Down syndrome and other conditions: Optional screening – your choice – your decision should be given at this time.

(c) Consent

- screening is optional and wāhine may choose to participate or not participate in first or second trimester screening and may change their mind about this decision
- wāhine may choose to participate in second trimester screening having declined the offer of first trimester combined screening
- those who choose not to participate in screening will not have their maternity care affected in any way
- if screening shows an increased chance result, specialist referral and diagnostic testing will be offered and only made with consent.

(d) Results

- how results are notified
- preference for method of receiving results
- when screening results are available
- wāhine will receive one result for first trimester combined screening utilising the NT scan measurements and serum markers
- the screening will provide an increased or low chance result and may also indicate other anomalies identified through ultrasound or serum markers
- maternal age at time of screening impacts on the result (ie, in 2020, less than 20 per 1000 wāhine who were under 29 years received an increased chance result whereas for those over 40 years, over 250 wāhine per 1000 received an increase chance result). The overall increased chance result across the population was 4.2%.

(e) Data and information collection and monitoring

- information and data are collected and securely stored
- information is used for monitoring and quality improvements of this screening
- this screening is monitored at a national level including monitoring and evaluating pregnancy and birth outcomes
- monitoring reports or any public information will present summary information only and will not be identifiable.

4.4 Medical history

Prior to referral for antenatal screening the health practitioner should discuss medical and family history with wāhine and whānau. The presence of some conditions in the medical history will indicate referral for genetics and/or obstetric specialist consultation. Examples of conditions indicating a referral to genetic services include:

1. Personal or family history of a known genetic condition (eg, Cystic fibrosis, Spinal Muscular Atrophy, Polycystic kidney disease.)
2. Personal or family history of a chromosome translocation.
3. Personal or family history of multiple congenital anomalies, or significant intellectual disability of unknown cause.

See the indications for referral to specialist obstetric services for guidance on when specialist referral is indicated.

5. Informed choice

Informed consent is a foundational principle of antenatal screening.

Wāhine who are less than 20 weeks pregnant should be advised about the availability of antenatal screening for Down syndrome and other conditions and that these screens are optional. The discussion of screening should be made with sufficient information, advice, and time to enable wāhine and whānau to make an informed decision. This discussion should be initiated by the health practitioner as early as possible in pregnancy.

Participation in antenatal screening for Down syndrome and other conditions is entirely the woman's choice. Wāhine also have the option to accept or decline further testing or referrals within the screening pathway. For instance, wāhine may decline first trimester combined screening, but later change their mind and accept second trimester screening. All choices that wāhine make must be respected and supported by health practitioners providing their care.

No single test checks for everything. No screening test finds all cases of a condition. Wāhine and whānau should be informed that screening states the estimated chance of a chromosomal abnormality but will not confirm the presence of the abnormality (this would require further (diagnostic) testing). Discussion about screening should include that there may be unexpected findings.

Up-to-date information about antenatal screening for Down syndrome and other conditions should be provided to support the screening offer. Informed choice for this screening should include a discussion about the screened conditions and the options and decisions that may need to be considered as a result of participation in this screening.

6. Interpreter services

Health services should be tailored to meet the needs of the individuals receiving them and should enable people to take responsibility for managing their own health. This helps to ensure equity of access and outcomes.

Health practitioners should offer additional support to whānau who have difficulty understanding information because of the language they speak and understand, hearing impairment or intellectual disability. Interpreter services are funded by Health NZ using Connecting Now. To access Connecting Now's interpreting service the number is 0800 854 737 (PIN 14059) or you can email support@connectingnow.com.au.

7. Potential benefits and harms of antenatal screening for Down syndrome and other conditions

Potential benefits of antenatal screening for Down syndrome and other conditions include:

- access to information that may provide more choice in the care and management of the pregnancy and birth
- reassurance associated with low chance results for the screened conditions
- reassurance associated with no abnormalities found through scanning.

Potential harms of antenatal screening for Down syndrome and other conditions include:

- anxiety and stress associated with the screening process
- wāhine and/or whānau having a limited understanding of the screening process. This may include a lack of understanding of screening results and what may or may not be detected
- anxiety and stress associated with an increased chance result which is very likely to be a false positive result
- false reassurance when a low chance result is given when pēpi does have a condition ie, a false negative result
- false reassurance when a low chance result is given but pēpi has a condition not screened for
- a very small chance of miscarriage (0.3%) resulting from diagnostic procedures following an increased chance result.

8. Documentation

Clear documentation of the screening process should be kept in the clinical records including the discussion, consent or decline of tests or referrals, and results of screening.

Each stage of the process should be documented in the clinical records, including:

- the content of discussions with wāhine
- the use of interpreters or other services
- consent or decline for screening and procedures or further testing
- details of results, follow up, or referral
- discussions with wāhine on results received
- other support, resources or information offered or provided

Health practitioners who refer wāhine for MSS blood tests should be aware that they will receive the screening results. If they are not the LMC, they should consider copying the screening result to the LMC (if available/known).

Health practitioners referring or handing over care to another health practitioner should provide appropriate documentation.

Referral information should include:

- consent or decline for screening
- details of screening tests ordered
- results of screening
- any follow-up from screening results
- any relevant family history
- referrals made to other services

9. The screening tests

Health NZ offers two options for screening for Down syndrome and other conditions in pregnancy; first trimester combined screening (ultrasound assessment and blood test) and second trimester maternal serum screening (blood test only). These are known as MSS1 (first trimester) and MSS2 (second trimester) screening tests. Referrals for this screening can be made by a midwife, GP, obstetrician, or other health provider.

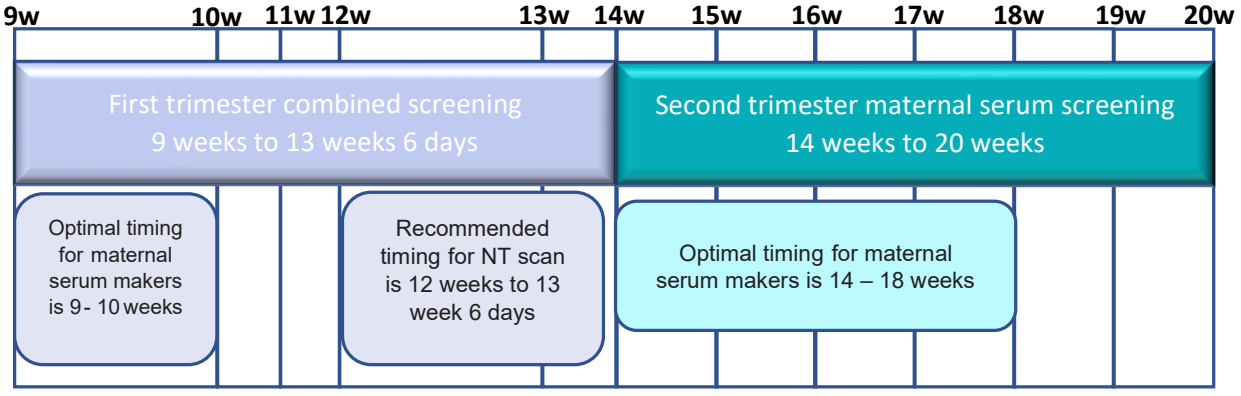
9.1 Timing of first trimester combined OR second trimester maternal serum screening tests

The recommended timing for the first trimester blood test is 9 to 10 weeks and for the NT scan 12 weeks, although it is also acceptable to do both blood test and ultrasound at 12 weeks. First trimester combined screening can be completed between 9 weeks and 13 weeks 6 days gestation.

The recommended timing for the second trimester blood test is 14 to 18 weeks. Second trimester maternal serum screening can be completed between 14 weeks and 20 weeks gestation.

Here is a link to an online gestational age calculator.

Figure 3: Optimal timing for different antenatal screening tests for Down syndrome and other conditions.



9.2 Dating of pregnancy

Screening accuracy is affected by correct estimation of gestational age. The order of precision for various methods of estimating gestational age is as follows:

1. IVF

2. Ultrasound assessment (of Crown to Rump Length)
3. Last Menstrual Period
4. Clinical assessment

The laboratory will use gestational age to normalise serum markers for comparison to multiples of the population median and calculate accordingly. The more accurate the gestational age the more accurate the screening results will be.

The most accurate screening result will be achieved when the best available dating method is used (CRL in most cases). Practitioners should identify which trimester the woman is in (by selecting either MSS1 or MSS2 on the request form) and can send the blood referral form **before** the NT scan has been done. The laboratory will add the dating based on CRL when they receive the ultrasound report.

An uncomplicated pregnancy does not generally require additional first trimester (eg, dating) scans. If the woman knows the approximate date of her LMP (within 1-2 weeks) it is most appropriate to arrange the ultrasound scan for 12 weeks gestation according to the LMP date.

9.3 Ordering tests

If wāhine accept screening, the health practitioner (midwife or doctor) will complete the screening request form.

Eligibility criteria for publicly funded services can be found on the Health NZ website.

When referring wāhine for tests the health practitioner should inform them:

- There are two components to first trimester combined screening: a blood test and an ultrasound scan
- Where they can go for their blood test and their ultrasound scan
- They need to have each component within certain timeframes
- They are usually required to pay a surcharge for an ultrasound scan

Referrals for the NT ultrasound must be made in accordance with Section 94 of the Pae Ora (Healthy Futures) Act 2022.

9.4 Completing the screening request form

The screening test relies on accurate and full information being provided by the health practitioner. The screening request form must include details of gestation, IVF, weight, smoking status, ethnicity, and relevant family history.

- **The screening request form must be completed with all the requested information** (except CRL).
- All information on the request form is needed by the laboratory to ensure high quality testing. Screening results will be inaccurate if the information on the screening request form is not completed/correct.

- Practitioners do not need to wait for a scan report to fill in the MSS1 request form – the CRL will be provided to the laboratory by the radiologist after nuchal scan completion, or referrers may provide a CRL measurement from an earlier dating scan if available.
- In pregnancies resulting from assisted reproductive technologies (ie, IVF) it is important that complete and accurate information is provided on the screening referral form.

Two laboratories perform antenatal screening for Down syndrome and other conditions. LabPLUS (Health NZ Te Toka Tumai) service areas from Taupō north and Canterbury Health Laboratories (Health NZ Waitaha) service areas south of Taupō.

Laboratory contact details for enquiries and screening request form orders:

Laboratory	Phone number
LabPLUS	0800 LABPLUS (0800 522 7587)
Canterbury Health Laboratories	0800 THE LAB (0800 843 522)

The screening request form can be found at Antenatal screening for Down syndrome and other conditions: Resources – Health New Zealand | Te Whatu Ora

9.5 Impact of accurate referral information

High quality screening results rely on complete and accurate information which informs the calculation. This includes details of smoking status, ethnicity, weight, IVF, twins, and gestational age.

As an example, obesity dilutes the analytes measured, and IVF pregnancies have a higher ßHCG. The calculation takes these factors into account when they are provided.

Importance of accurate maternal weight data

Referring practitioners should undertake weight measurement for women **at the time of screening referral**. The accuracy of the weight metric is an important factor in the accuracy of the screening calculation as blood volume will differ according to body weight. Weight influences the calculation by means of a dilution effect on the analytes. As such, an incorrect weight reported on the MSS laboratory request form could lead to the over or underestimation of chance (Table 1). If wāhine are assessed as having an increased chance of trisomy (ie, greater than 1:300) they will be offered diagnostic tests which come with a risk of miscarriage.

Table 1. An example of the impact of weight on the chance calculation for Trisomy 21 (Down syndrome)

	Referral form data	Actual measurement
Weight (kg)	70	80
MSS1 result	Increased chance	Low chance

The table below provides some other scenarios which affect the calculation and therefore the result.

Table 2: Effect of different scenarios on screening results

Scenario	Result 1	Change in Details	Result 2	Comment
Change in gestational age	Gestational age calculated at 15.1 T21 result: 1:220 chance	Ultrasound scan calculated gestational age at 14.1	Revised calculation T21 result: 1:910 chance	The analytes change over the pregnancy and therefore the calculation changes depending on the gestational age.
Change from singleton to twin pregnancy	Assumed singleton pregnancy T21 result: 1:250 chance	Ultrasound scan shows twins	Revised calculation T21 result: 1:500 chance	The analytes are divided for a twin pregnancy.
Non-smoker to smoker	Assumed non-smoker T21 result: 1:210 chance	Health practitioner informs the screening laboratory that the woman is a smoker	Revised calculation T21 result: 1:300 chance	Smoking affects placental function and inhibin levels are higher in wāhine who smoke.
Compounding effect of many changes	42 year old wāhine using LMP dating at 18.2. No scan data, singleton pregnancy reported, no weight, no smoking information T21 result: 1:520 chance	Age of wāhine incorrect – found she is 32 years of age, LMP was wrong, and it is actually 14.1. Twins, smoker and weighs 45 kg	Revised calculation T21 result: 1:13,000 chance	There is a compounding effect when many of the variables are incorrect.

9.6 Blood sampling

Blood samples for First Trimester Combined Screening (MSS1) should be taken in an SST (gold top) tube.

Blood samples for Second Trimester Screening (MSS2) should be taken in an SST (gold top) tube.

Samples need to be spun and separated by laboratory staff **within four hours of collection**. Once separated the sample is relatively stable and in many cases will be frozen prior to transporting to the main screening laboratory (in Auckland or Christchurch). Sample takers should ensure that MSS samples are delivered to the laboratory as soon as possible after collection. At busy times registration and centrifugation may be delayed by the volume of samples coming to the laboratory.

Practitioners may need to check with local collection centres if they have the ability to separate samples as this may vary by location.

10. Nuchal translucency scan

If wāhine agree to first trimester combined screening, the ultrasound provider will complete the nuchal translucency (NT) and CRL measurements and send the results to the screening laboratory. An NT scan is optimally performed between 12 weeks and 13 weeks 6 days.

10.1 Referral for NT scan

Specialist medical maternity services, including NT scans, may only be provided to wāhine on referral from another practitioner (midwife or doctor).

This requires a written or electronic radiology referral.

Referrals for the NT ultrasound must be made in accordance with Section 94 of the Pae Ora (Healthy Futures) Act 2022.

10.2 Screening calculation algorithm

The screening calculation will be performed by the laboratories.

The screening laboratories use a single database for probability calculation. This ensures consistent calculation of chance for all wāhine across Aotearoa New Zealand.

10.3 Discussions with wāhine at time of ultrasound scan

Ultrasound practitioners may discuss the findings of the NT scan with wāhine but should not offer a calculation on the chance of Down syndrome and other conditions based on NT alone (for example, 1:300 chance).

In all but exceptional circumstances, the screening result will be communicated to wāhine by the referring practitioner.

10.4 Reporting requirements

Reporting templates for radiologists can be found within the NZ Obstetric Ultrasound Guidelines.

Precise measurement of CRL and NT is essential in the interpretation and final assessment provided via ultrasound reporting. The CRL is used by the laboratory to determine gestational age using the ASUM guidelines.

The following information from the NT scan must be provided for this screening:

- National Health Index (NHI) number
- demographic information (DOB, name)
- referrer's name
- date of NT scan

- the CRL measurement
- the NT measurement
- multiple pregnancy (chorionicity and amnionicity)
- other details that may inform the screening calculation
- significant abnormalities which may change the management of the pregnancy
- name of the practice
- name of the radiologist
- name and FMF number of the practitioner performing the scan

10.5 CRL out of range for first trimester combined screening

For acceptance for first trimester combined screening, at the time the NT scan is performed, pēpi must have a CRL between 45–84mm.

If the CRL is greater than 84mm it is too late for NT assessment. A second trimester maternal serum screening report will be provided if the blood sample was collected in the second trimester, otherwise a new blood sample will be needed.

10.6 Specialist referral following abnormal NT scan

If an NT scan shows an obvious anomaly, for instance structural/anatomical anomaly, the radiologist should inform the referring health practitioner in a timely manner.

The referring health practitioner should discuss the results with wāhine and offer referral to a specialist obstetrician. Practitioners can review the indications for referral to specialist obstetric services.

10.7 NT 3.5 mm or more

If the NT scan shows a NT measurement greater than or equal to 3.5mm, the ultrasound/radiology provider should communicate with the referring health practitioner to discuss the scan results.

The referring health practitioner should offer wāhine referral to a Fetal Medicine specialist (or local equivalent), with the expectation that they will be seen in a timely manner.

Completion of first trimester combined screening is still recommended. This will assist the specialist to develop a care pathway with a full clinical picture.

An early detailed fetal heart scan should be considered at 16 weeks gestation.

10.8 NT for twins

For twin pregnancies, an NT and CRL for each twin must be measured at the same time to ensure accurate screening calculation.

The screening laboratory uses CRL to date the pregnancy and interpret the measured marker values. Both NT measurements should be made at the time of the CRL measurements. If they are different in twins, the larger will be used.

Any information that may assist screening calculation for each pēp should be included in the report to the screening laboratory.

If a scan is performed and an NT measurement is only able to be completed for one twin, a subsequent NT scan should be offered. The subsequent scan should reassess NT and CRL for **both** pēpi if more than 2 days has elapsed.

10.9 Transmitting scan results to the screening laboratories

Ultrasound/radiology providers will transmit copies of the report results in a timely manner direct to LabPLUS for Taupō north, or Canterbury Health Laboratories (CHL) for south of Taupō.

Ultrasound/radiology providers should have a system in place to send the ultrasound report to the screening laboratory and to confirm the report has been sent.

10.10 Loss of one twin

For pregnancies where there has been demise of one twin, a NT and CRL measurement for the surviving twin should be sent to the screening laboratory and will be used to provide a calculation without serum levels.

If the NT scan identifies a sac showing fetal demise, it is possible that there could be a contribution to the maternal biochemical markers for many weeks. Therefore, serum analytes are not used in the calculation. The screening laboratory may provide an assessment based on NT without biochemistry.

10.11 NT for multiple pregnancies – triplets or greater multiples

For pregnancies with three or more pēpi, an NT alone can be used for assessment. Specialist referral is indicated for all multiple pregnancies.

The screening laboratory is not able to provide a probability calculation for pregnancies where there are triplets or greater multiples.

10.12 Individual certification and standards requirements

Health NZ oversees a quality and monitoring programme for NT practitioners in Aotearoa New Zealand. This includes working with the health sector to monitor and develop ultrasound screening practices. Quality improvements will continue to evolve. This may result in changes in delivery of service, monitoring, and audit.

Health NZ recommends International Accreditation New Zealand (IANZ) radiology accreditation for radiology providers. This occurs for the majority of practices around Aotearoa New Zealand and provides assurance that the practice operates to established standards.

In Aotearoa New Zealand, the quality of service requirements relates to appropriate education and training for the measurement of NT. Appropriate certification is recognised through FMF London and the Australian Nuchal Translucency – Ultrasound, Education and Monitoring Program.

For further guidance on fetal ultrasound in Aotearoa New Zealand, see the New Zealand Obstetric Ultrasound Guidelines.

11. Screening laboratory processes

11.1 Laboratory communication of results and reports

- All laboratory reports are sent according to the preference of the requesting practitioner (mail, email, EDI). To change preference, contact Lablink@adhb.govt.nz and labinfo@cdhb.health.nz
- Some communication is by SMS (text message). The messages are computer generated so can be replied to, but the number cannot be called back. The message will contain a number to ring to speak to a person.
- Requests for additional information to enable calculation of a screening result (eg, weight) may be made by SMS (text) message or by phone.
- Reports of incomplete screening are sent by SMS message in addition to reporting as outlined above.
- Increased chance results may be sent by SMS message or phoned.
  - If results are sent by SMS: the message includes a request for acknowledgement (text back “ok” to indicated message received).
  - If the laboratory receives no reply the message is sent again the next day.
  - If there is still no reply the number is phoned (which may indicate for example that the requestor is on holiday and the name of the backup is then provided to the laboratory).
  - This communication does not replace formal reporting as outlined above.
  - Increased chance results are not communicated after noon on the day before a weekend or public holiday as this does not allow time for the requestor to arrange follow-up before contacting the family.
- It is very important to check the parameters used for the screening calculation as detailed on the report (ie, weight, age) and contact the laboratory for recalculation of the chance if items are incorrect.
- Reporting is done within three business days after all information needed to calculate the chance is obtained (blood results, scan, weight etc).

11.2 Reporting information to the health practitioner

The screening laboratory will provide a first trimester combined screening or second trimester maternal serum screening result to the health practitioner who referred the wāhine for screening.

The screening laboratory report will include:

- screening result (‘increased chance’ or ‘low chance’)
- woman’s demographics and pregnancy dating information
- NT and CRL measurement

Individual probability assessments for:

- trisomy 21 (Down syndrome)
- trisomy 18 (Edwards syndrome)
- trisomy 13 (Patau syndrome)

The screening laboratory report may include other information, for example that specialist obstetric referral is recommended or that a rare genetic disorder is indicated.

The screening threshold for reporting increased chance of aneuploidy is 1:300.

11.3 Provision of specialist laboratory advice

The screening laboratory will communicate all increased chance results to the health practitioner.

The screening laboratory will provide specialist laboratory advice to the health practitioner, when requested, and will ensure that health practitioners have screening information required to inform wāhine of their screening results.

Health practitioners are welcome to contact the screening laboratory if required.

11.4 Incomplete screening

The screening laboratory will provide an incomplete report to the health practitioner when a blood sample for first trimester combined screening has been received, but no NT, CRL measurement, or maternal weight, **OR** if a scan report has been received by the laboratory but no blood sample.

The screening laboratory will accept a CRL of 45–84 mm for first trimester combined screening. If the CRL result is above 84mm, the screening laboratory will advise the health practitioner that first trimester combined screening cannot be completed because the woman is in the second trimester. If the blood has also been taken in the second trimester, second trimester screening will be performed.

If the combined screening tests (blood and scan) have not been performed by 13 weeks 6 days, the screening laboratory will:

- advise that first trimester combined screening cannot be completed because the scan data or blood sample was not available
- recommend that wāhine are offered second trimester maternal serum screening

The health practitioner should advise that first trimester combined screening has not been completed and provide information about second trimester maternal serum screening.

11.5 Further serum received

The screening laboratory will advise the health practitioner if a serum sample has been received after a result has already been issued.

The screening laboratory will advise the health practitioner that a further result will not be issued unless there are clinical indications to do so. The health practitioner can phone the laboratory to discuss as required. Decisions regarding recalculating assessments will be made on a case by case basis.

Screening laboratories contact details:

Laboratory	Contact details	Contact details
LabPLUS	www.labplus.co.nz Phone: 0800 LABPLUS (522 7587) Lead Clinical Scientist: Dr Dianne Webster diannew@adhb.govt.nz	**Taupō and North** (Northland, Waitemata, Counties Manukau, Auckland, Waikato, Lakes, and Bay of Plenty)
Canterbury Health Laboratories	www.chl.co.nz Phone: 0800 THE LAB Chemical Pathologist: Dr Richard King	**South of Taupō** (Tairāwhiti, Hawke's Bay, Whanganui, Taranaki, MidCentral, Wairarapa, Capital & Coast, Hutt Valley, Nelson Marlborough, West Coast, Canterbury, South Canterbury, Southern)

12. Results

Blood samples are generally received by the screening laboratory two to three days after collection. Screening results will be completed by the screening laboratory within three business days after the receipt of the blood sample, scan information or any other information required (ie, maternal weight), whichever is the latter (if first trimester combined screening). Screening results include information provided about the wāhine, details of the chance of various trisomies, and recommendations.

The screening laboratory will send a report to the health practitioner if both parts of screening (ie, NT scan and blood tests) have not been received by 13 weeks 6 days.

The combination of ultrasound and maternal serum markers increases detection rates (improves sensitivity) and/or reduces the number of wāhine considered to be at increased chance (improves specificity).

12.1 Receiving screening results

The referring health practitioner is responsible for receiving screening results.

When reviewing screening results, LMC's should check the report thoroughly for any data errors (ie, incorrect smoking status, incorrect maternal weight etc) and contact the laboratory to request a recalculation of risk if required.

If the screening result is **low chance** the screening laboratory will dispatch the result to the health practitioner by mail or electronic means within 48 hours of the result being available.

If the screening result is **increased chance** the screening laboratory will contact the health practitioner within 24 hours of the result being available. The result will also be dispatched to the health practitioner by mail or electronically.

It is useful to ascertain women's preference for receiving results at the time that the screening offer is made. Wāhine may wish to be accompanied by whānau or a support person when receiving results.

Referrers should have a system in place to ensure that women who gave consent for screening have undertaken both the blood test and the NT scan (for MSS1). Referrers should also have a system for checking that results have been received when women have undertaken screening tests to enable timely discussion and referral in the event of an increased chance result.

12.2 Communicating screening results

Health practitioners should communicate results to wāhine in an appropriate and timely manner, and should consider the following:

- Prior to communicating with wāhine, the health practitioner can discuss the result with the screening laboratory or Genetic Services (if further information will be useful or results need to be clarified).
- Communication to wāhine and whānau needs to occur through reliable methods such as face to face or telephone, taking into account appropriate timing (such as the need for timely referral or follow up). Results or professional advice should not be sent via a text message.
- Health practitioners should be able to present results in a clear and concise way to support wāhine and whānau in their decision-making. This includes understanding statistical information.

It can be useful to communicate the screening result in different ways to help wāhine and whānau better understand.

For example:

'You have a 1 in 4 chance of…, or put another way, you have a 25 percent chance of….'

'You have a 1 in 20 chance of having a baby with one of the conditions, this means there is a 19 in 20 chance of having a baby without the condition'

- Discussion around the results may include:
  - the limitations of screening
  - that a low chance result means that the baby is unlikely to be born with one of the conditions screened for, but it does not mean they will definitely not be born with one of the conditions (or another condition not indicated by screening)
  - providing an opportunity for wāhine and whānau to ask questions
  - providing information about other services, including community support agencies wāhine and whānau can contact. If wāhine with a low chance result request diagnostic testing, a referral to a specialist obstetrician may be made.

12.3 Communicating increased chance results and offering referral

Health practitioners should inform wāhine and whānau in a timely manner of all screening results indicating an increased chance of Down syndrome or another condition.

Consideration should be given to the timing of giving results and whether access to timely support services or further information is available (for example, on public holidays or Friday afternoons).

Anxiety following an increased chance result is normal. Anxiety includes the stress and worry experienced while waiting for decisions about diagnostic testing, and the possibility of a higher level of anxiety for the remainder of the pregnancy.

If screening shows an increased chance of a genetic condition, wāhine and whānau may require more information to enable them to make an informed decision about the ongoing management of their pregnancy; one which they feel is best for themselves and their families.

Sources of information and support are listed in Appendix I.

Document the discussion and management plan in the clinical notes.

Provide wāhine with a copy of the results if requested.

Upon receiving an increased chance result, health practitioners should:

- offer a timely specialist referral to all wāhine with increased chance results
- make clear to wāhine that they have the right to decline a referral following an increased chance result
- respect and support any decision made by wāhine throughout the screening process
- provide links to community organisations to enable the gathering of more information and to access support which may also be helpful. These can be found in Appendix I.

12.4 Referral to Specialist

Following an increased chance result, wāhine may be undecided about their next steps. Practitioners can review the indications for referral to specialist obstetric services to determine whether referral is indicated.

Wāhine may require further information about:

- what the increased chance result may mean and how it may affect the on-going management of the pregnancy and birth
- the difference between treatable conditions (for instance heart defects) and non-treatable conditions (for instance trisomy 13).
- the likely process with specialist referral and offer of diagnostic testing
- antenatal care plan if specialist referral is declined

The referral to a specialist should include details of:

- gestation
- screening results
- past obstetric history
- any issues identified which require further discussion
- any relevant family history.
- booking bloods including blood group

A referral to Genetic Services may provide the opportunity to gather information to make or confirm a diagnosis of a genetic disorder.

Other referrals may also be considered. These include:

- a paediatrician
- a health social worker
- a counsellor
- a kaiawhina or Māori health worker

Wāhine must be given time to reflect and to consider their decision.

13. Diagnostic testing

Diagnostic testing includes a procedure to collect a sample of fetal cells either by chorionic villus sampling (CVS) or amniocentesis. The sample collected is sent for chromosome analysis.

CVS can be performed from 11 to 14 weeks of pregnancy but is typically performed between 11 and 13 weeks. CVS is only offered in a few locations. CVS requires access to the placenta and is not possible in all cases.

Amniocentesis can be performed between 15 and 20 weeks (or later by agreement).

In both cases samples may be analysed in one or two ways. A quicker test (FISH) is utilised in certain situations and takes around 2-4 working days. The comprehensive test (microarray) is generally performed and takes 2-3 weeks.

Both procedures carry a risk of miscarriage of around 0.3% (3 per 1000 procedures performed).

A more detailed scan may be required following an abnormal finding on ultrasound.

Diagnostic testing is publicly funded for eligible wāhine who have:

- an increased chance result from a prenatal screening test
- an abnormal ultrasound scan (structural abnormalities)
- previously had a baby with a congenital anomaly

International best practice does not support direct referral to diagnostic testing based on maternal age alone.

13.1 Discussing diagnostic testing with whānau

Health practitioners should provide information on diagnostic testing and inform wāhine that diagnostic testing is optional.

Health practitioners should outline the following choices available to wāhine:

- whether or not to accept a referral and diagnostic testing
- referral to another service for example a paediatrician, Genetic Services, health social worker or counsellor.

The health practitioner should explain:

- what information the diagnostic tests can provide
- the risks associated with a diagnostic test
- the decisions that wāhine may need to consider
- the anxiety that may be experienced while waiting for results and possibly for the remainder of the pregnancy
- the support services that can be accessed

The obstetric specialist should inform wāhine of the risk of diagnostic testing procedures. Cells obtained at CVS and/or amniocentesis will be analysed as appropriate for the result.

The risk of miscarriage after CVS and amniocentesis is about three miscarriages in every 1000 wāhine tested.
Other rare complications include:

- rupture of membranes
- CVS procedure is offered after 11 weeks gestation as some research has suggested that development of arms, fingers, legs, or toes may be disrupted if CVS is performed before nine weeks gestation
- development of Rhesus factor incompatibility. All wāhine who have Rh-negative blood group are offered an injection of anti-D to prevent this complication

13.2 Receipt of a positive result following diagnostic testing

The specialist should explain the meaning of any test results and provide information about any diagnosis. Health practitioners can seek further information from genetic services and other sources as required. See Appendix II for more information on the conditions. Wāhine and whānau may wish to have time to consider the results and what they may mean for their pēpi.

After receiving the results of the diagnostic test, health practitioners should support wāhine to make an informed decision.

The following may be discussed:

- information about the condition
- options available which include:
    - continuing with the pregnancy
    - termination of the pregnancy

If wāhine choose to continue with the pregnancy, the options for antenatal care such as specialist care and support, and postnatal options should be discussed. If the pēpi has a condition which has a very short life expectancy, consideration should be given to offering antenatal or postnatal palliative care for pēpi and counselling services to wāhine (and their whānau).

Health practitioners must provide wāhine with opportunities to access additional information and support.

This may include referral to a:

- paediatrician
- health social worker

- genetic services
- counsellor
- kaiawhina/Māori health worker

Whānau may seek information from sources listed in Appendix I to find out what living with a specific condition may mean.

14. Genetic services

Health practitioners should advise wāhine with increased chance results about the availability of Genetic Services. Genetic Services can provide information and support for whānau with, or with increased chance of, a genetic disorder. Initial discussions with the health practitioner will be with the on call genetic counsellor. If the health practitioner wishes a staff member (genetic counsellor or clinical geneticist) to subsequently talk to wāhine, this should be handled as a formal referral.

Genetic Services are physically located in Auckland, Wellington, and Christchurch. Telephone, video or in-person consultations are available.

Contact details can be accessed via the website: About Genetic Health Service NZ – Health New Zealand | Te Whatu Ora.

Referrals to Genetic Services should come from the GP or LMC in the first instance. A copy of the screening report should be included. Please use the Genetic Services Referral form available from the Health NZ website.

Any queries about the screening laboratory analytical process and the result algorithm can be referred back to the designated specialists in LabPLUS **and** Canterbury Health Laboratories.

Genetic Services contact details:

Genetic Services	Phone Number
Northern and Midland Region	0800 476 123
Central and Southern Region	0508 364 436

14.1 Other referrals

Wāhine and whānau may access information to support decision making about the management of their pregnancy. Health practitioners should identify services in their region which may provide additional support.

This may include:

- obstetrician
- specialist maternity services
- maternal fetal medicine specialist
- paediatrician
- general medical practitioner
- health social worker
- counsellor
- kaiawhina/Māori health worker

- disability support services
- parent support groups

Refer to Appendix I for resources and contacts.

15. Data information and monitoring

15.1 Data and information collection

Antenatal screening for Down syndrome and other conditions collects, creates, and retains indefinitely the following data and information. Table 3 outlines what information is collected.

Table 3: Data and information collected for antenatal screening for Down syndrome and other conditions

Information about wāhine as collected on request form	Sample data	Health practitioner data
• name (in full) • National Health Index (NHI) number • date of birth • gestation at time of sampling • ethnicity • weight • information about the pregnancy • estimated date of delivery • relevant family history	• date and time of samples/ scans • collection and screening laboratory assigned ID# • screening results • diagnostic results and outcomes • information about what has been reported and to whom including any clinical information provided	• name • midwifery/medical council number • radiology practice and practitioner • telephone numbers • address • other contact information

This data and information are held indefinitely by NZ laboratories on behalf of Health NZ.

15.2 Uses of data and information

Only authorised personnel have access to the identifiable information and data for the purposes of screening, quality assurance, monitoring, and evaluation.

Data and information are collected and held securely to:

- interpret screening results
- make sure that results can be provided to health practitioners
- monitor and evaluate this screening including the results of diagnostic testing and outcomes of pregnancies.

From time to time, there may be requests for access to screening data. This may include data requests for research, or other requests from individuals, committees, groups, or organisations.

Any requests regarding this data must be forwarded to, and authorised by, Health NZ using the official data request form.

15.3 Monitoring

Antenatal screening for Down syndrome and other conditions is overseen by Health NZ. To maintain the quality of this screening, it is closely monitored on a regular basis, with evaluation undertaken periodically. Monitoring is dependent on the information collected as set out in Table 3 (above).

Health NZ publishes reports on this screening. These reports are summary information only and do not contain identifiable data or information.

Appendix I – Resources and contacts

Sources of further information and contact details for support services are listed here.

This list should be supplemented by the local or regional services within your own networks.

Health NZ is responsible for oversight of antenatal screening for Down syndrome and other conditions. Health NZ produces consumer and practitioner resources and audits and monitors this screening initiative.

Consumer resource *Antenatal screening for Down syndrome and other conditions: optional screening – your choice, your decision*

- Hard copies are available free of charge and can be ordered at www.healthed.govt.nz or by contacting the Authorised Provider of Health Education Resources in your area. A full list of who these are (by region) is available on the HealthEd website.

On-line education for health practitioners who provide services within the antenatal and newborn screening programmes can be accessed at www.learnonline.health.nz.

Information for whānau can be found at Screening for Down syndrome and other conditions (info.health.nz).

For questions or comments:

Antenatal and Childhood Screening Team

Email: Antenatalnewbornscreening@tewhatuora.govt.nz

There are two Screening Laboratories:

1. LabPLUS at Te Toka Tumai Auckland
2. Canterbury Health Laboratories at Health NZ Waitaha

Laboratory request forms can be ordered from the screening laboratories:

Laboratory	Contact details	Areas covered
LabPLUS	www.labplus.co.nz Phone: 0800 LABPLUS (522 7587) Lead Clinical Scientist: Dr Dianne Webster diannew@adhb.govt.nz	Taupō and North (Northland, Waitematā, Counties Manukau, Auckland, Waikato, Lakes, and Bay of Plenty)

Canterbury Health Laboratories	www.chl.co.nz Phone: 0800 THE LAB Chemical Pathologist: Dr Richard King	South of Taupō (Tairawhiti, Hawkes Bay, Whanganui, Taranaki, MidCentral, Wairarapa, Capital & Coast, Hutt Valley, Nelson Marlborough, West Coast, Canterbury, South Canterbury, Southern)

NZ Maternal Fetal Medicine Network

www.nzmfm.health.nz

Genetic Services, New Zealand

www.geneticshealthservice.org.nz

- Northern and Midland Region, phone: 0800 476 123
- Central and Southern Region, phone: 0508 364 436

Health and Disability Commissioner

www.hdc.org.nz/

Office of the Privacy Commissioner

www.privacy.org.nz

Ministry of Health

www.health.govt.nz

Primary Maternity Services Notice 2021

www.tewhatuora.govt.nz

Other resources

Aotearoa New Zealand
CCS Disability Action

www.ccsdisabilityaction.org.nz

Phone: 0800 227 200

Enable New Zealand

www.enable.co.nz

Phone: (04) 472 2247

IHC

www.ihc.org.nz

Phone: (04) 472 2247

Kiwi Families

Links to disability support articles

www.kiwifamilies.co.nz

Midwifery Council of New Zealand

www.midwiferycouncil.health.nz

New Zealand College of Midwives

www.midwife.org.nz

New Zealand Down Syndrome Association

www.nzdsa.org.nz

Phone: 0800 693 724

Email: national.coordinator@nzdsa.org.nz

New Zealand Federation of Disability Information Centres

The Federation of Disability Information Centres

Pacific Information Advocacy Support Services

www.vakatautau.co.nz

Parent and Family Resource Centre (engage Aotearoa)

www.parentandfamily.org.nz

Parent to Parent

www.parent2parent.org.nz

People First (Nga Tāngata Tuatahi)

www.peoplefirst.org.nz

Rare Disorders NZ

www.raredisorders.org.nz

Royal Australian and New Zealand College of Obstetricians and Gynaecologists

www.ranzcog.edu.au

Prenatal screening and diagnostic testing for fetal chromosomal and genetic conditions

Royal New Zealand College of General Practitioners

www.rnzcgp.org.nz

Royal Australian and New Zealand College of Radiologists

www.ranzcr.edu.au

Sands New Zealand

www.sands.org.nz

Starship Hospital

Down Syndrome: Management of the neonate (guideline)

Turner Syndrome Association of New Zealand

www.turnersydrome.co.nz

UpsideDowns Education Trust

www.upsidedowns.co.nz

Australia

Australian Centre for Genetics Education

Changes to Chromosomes

Mosaicism

Down Syndrome Fact Sheet

Trisomy 13 – Patau Syndrome

Trisomy 18 – Edwards Syndrome

Turners Syndrome

Kleinefelter Syndrome Human Genetic Society of Australasia (HGSA)

www.hgsa.com.au

United Kingdom

Antenatal Results and Choices (UK)

www.arc-uk.org/

Down's Syndrome Association (UK)

A New Parent's Guide

Down syndrome online

www.down-syndrome.org

Fetal Medicine Foundation UK

The 11–13+6 Weeks Scan

Fetal abnormalities

National Health Service (UK)

Trisomy screening

What is Down syndrome

Other

Spina Bifida Association of America

Spina Bifida Association - Research, Advocacy, Education, and Support

International Mosaic Down Syndrome Association

Booklet for professionals

Appendix II – Screened conditions

Antenatal screening for Down Syndrome and other conditions primarily reports on chromosomal trisomies 21, 18, and 13. The list of 'other conditions' cannot be exhaustive as it is unknown what other conditions may be indicated through this screening. This is the same for blood tests taken in any health setting where unanticipated findings may be identified.

Chromosomes are located in the nucleus of each cell, and contain the genetic material that, in combination with environmental influences, determines a person's individual characteristics. Some genetic conditions are caused by extra or missing chromosomes. These are collectively referred to as aneuploidy and the screening programme will provide an estimate of likelihood for several of these aneuploidies.

Down syndrome (Trisomy 21)

Down syndrome (also known as Trisomy 21) is a genetic disorder caused by an extra copy of chromosome 21. In Down syndrome, instead of a pair there are three copies of chromosome 21 inside each of the body's cells. This extra genetic material is responsible for the physical and intellectual disabilities which are the characteristics of Down syndrome.

The average life expectancy of people with Down syndrome has increased with improved healthcare, better education, greater opportunities, and a shift in societal attitudes. Studies indicate that average life expectancy in the UK was estimated to be 9 years of age in 1929 and 12 years in 1949. Subsequent reports have shown a marked increase in life expectancy that began in the 1950s. By the year 2000 the median life expectancy for people with Down syndrome in Australia was 60 years[8].

Research shows that whānau appreciate information about the abilities and potential of people with Down syndrome (eg, participation in community sports, activities, inclusion in mainstream education classes, employment, independent living, life expectancy to 50–60s and having friends); as well as clinical details[9]. Balancing clinical information (eg cause, chance of recurrence for future pregnancies, physical features, associated medical conditions, intellectual ability, and developmental delay) with a better understanding of the information parents consider most important, may enable health practitioners to provide the information that satisfies the needs of whānau about Down syndrome.

The New Zealand Down Syndrome Association note that:

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[8] Torr, J., Strydom, A., Patti, P. and Jokinen, N. (2010). Aging in Down Syndrome: Morbidity and Mortality Journal of Policy and Practice in Intellectual Disabilities, March 2010, Volume 7:1, pp 70–81.

[9] Sheets KB, Best RG, Brasington CK, Will MC. 2011. Balanced information about Down syndrome: What is essential? Am J Med Genet Part A 155: 1246–1257.

'People with Down syndrome are all unique individuals and vary in their abilities and achievements. They do have features in common, but they also closely resemble their parents and family. Many characteristics are associated with Down syndrome, but any one person will only have some of them. Thus each person is an individual, with a unique appearance, personality and set of abilities. The extent to which a child shows the physical characteristics of the syndrome is no indication of his or her abilities and achievements'[10]

Similarly, the organisation Upside of Down report that:

'Today people with Down syndrome live at home with their families and are active participants in the educational, vocational, social, and recreational activities of the community. They are integrated into the regular education system, and take part in sports, camping, music, art programs and all the other activities of their communities. In addition, they are socializing with people with and without disabilities, and as adults are obtaining employment and living in group homes and other independent housing arrangements'[11].

People with Down syndrome experience varying degrees of delay in their learning and development and may have additional health needs. They will almost always learn to walk, speak, read, and write but commonly require support in using money, negotiating public transport, and building skills for appropriate social behaviour.

Some of the health issues associated with Down syndrome include:

- hearing loss in up to 50 percent of people with Down syndrome
- congenital heart disease in up to 50 percent of people with Down syndrome
- thyroid disorders, most commonly hypothyroidism, in up to 40 percent of cases
- gastrointestinal tract congenital malformations, such as duodenal atresia and Hirschsprung's disease
- cataracts and visual refractive errors
- childhood leukaemia in about two percent of cases
- early onset Alzheimer's disease.

Children with mosaic or partial forms of this trisomy are likely to be less severely affected.

Trisomy 18 (Edwards syndrome)

Trisomy 18, also known as Edwards' syndrome, is a genetic disorder caused by an extra copy of chromosome 18 inside each of the body's cells.

Trisomy 18 appears to affect females more frequently than males by a ratio of approximately three or four to one. Large population surveys indicate that it occurs in

[10] www.nzdsa.org.nz
[11] www.upsideofdown.org/about_t21

about one in 5,000 to 7,000 live births[12]. The incidence increases as the mother's age increases. The syndrome has a very low rate of survival, resulting from heart abnormalities, kidney malformations, and other internal organ disorders.

About 50 percent of live born infants with trisomy 18 live to 2 months, and 5–10 percent survive their first year of life. Major causes of death include apnoea and heart abnormalities. It is impossible to predict the exact prognosis of a child with Trisomy 18 during pregnancy or post birth. The average lifespan is between 5–15 days. A small percentage of babies with the full trisomy 18 who survive birth and early infancy may live to adulthood.

Children with mosaic or partial forms of a trisomy may have different morbidity and mortality statistics. In mosaic forms there are some cells in the body where the chromosome number and structure are different from other cells.

Trisomy 13 (Patau syndrome)

Trisomy 13, also known as Patau syndrome, is a genetic disorder caused by an extra copy of chromosome 13 inside each of the body's cells. The extra chromosome 13 disrupts the normal course of development. It causes severe neurological, heart and kidney defects which make it difficult for infants to survive. Newborns with trisomy 13 share common physical characteristics including: extra fingers or toes (polydactyly), small head (microcephaly), facial defects such as small eyes (microphthalmia), absent or malformed nose, cleft lip and/or cleft palate.

Many infants have difficulty surviving the first few days or weeks due to severe neurological problems or complex heart defects. Surgery may be necessary to repair heart defects or cleft lip and cleft palate. Physical, occupational, and speech therapy will help those individuals with trisomy 13 who live beyond the first few weeks/months.

Triploidy

Triploidy is a genetic disorder caused by an extra copy of each chromosome inside each of the body's cells, so the baby has three copies of each chromosome in each cell rather than two, making a total of 69 chromosomes rather than 46. The majority (more than 99 percent) of babies with triploidy will miscarry or be stillborn. Of those pēpi born alive, most are likely to die in the hours or days following birth. A few pēpi with triploidy have lived five months or longer, but this is rare and usually pēpi who survive longer have mosaic triploidy rather than full triploidy. Pēpi with triploidy usually have multiple genetic problems and severe growth restriction.

[12] www.rarediseases.org/rare-disease-information/rare-diseases/byID/217/viewFullReport

Conditions identified by ultrasound

Ultrasound scans undertaken as part of this screening (or any pregnancy ultrasound assessment) may detect some major fetal structural anomalies, such as skeletal anomalies, brain and neural tube defects, congenital heart defects, and abnormalities of the renal tract, gastrointestinal system, and abdominal wall. These will be mentioned in the ultrasound scan report.

An increased NT measurement (≥3.5mm) is associated with trisomy 21, Turner syndrome and other chromosomal defects as well as other fetal malformations and genetic conditions.

There are instances where pēpi have increased NT measurements but at diagnostic testing have normal chromosomes. These pēpi may have an increased chance of a number of abnormalities. Conditions associated with increased NT measurement include cardiac malformation, diaphragmatic hernia, omphalocoele, body stalk anomaly, skeletal anomalies, Noonan syndrome, Smith-Lemli-Opitz syndrome and spinal muscular dystrophy.

All information identified through screening will be included in the report to the health practitioner.

Neural tube defects

Neural tube defects (NTDs) are birth defects of the brain and spinal cord. The two most common neural tube defects are spina bifida and anencephaly. In spina bifida, the spinal column does not close completely during the first month of pregnancy. There is usually nerve damage that causes at least some paralysis of the legs. In anencephaly, much of the brain does not develop. Pēpi with anencephaly are likely to be stillborn or will die shortly after birth.

Adequate maternal intake of folate/folic acid (800 micrograms per day) commencing antenatally (or pre-conceptually) and continued through the first trimester of pregnancy significantly reduces the probability of NTD.

First trimester combined screening cannot estimate the probability of NTDs however more severe forms may be detected at the nuchal translucency ultrasound.

For NTDs, including spina bifida, the second trimester anatomy scan (19+ weeks) is the best available screening tool (although may sometimes be detected at the 12 week scan). The use of AFP as a screening tool for NTDs is neither very sensitive nor specific and is not considered best practice internationally and should not be ordered specifically with the intention of screening for NTDs.

Multiple of the median (MoM)

Serum marker levels used in antenatal screening change by gestational age. Therefore, for accurate interpretation of the test results, a different reference range must be used for each week of gestation, depending on when the test is drawn. To

avoid the multiple reference range problems and also to standardise test results a median value for test results in normal pregnancies is determined for each week of gestation. The individual maternal analyte levels are compared with the median for that analyte at the appropriate gestational age and expressed as a multiple. This is the multiple of the median. Although serum analytes will continue to form part of the screening algorithm, they are no longer included on screening reports.

Appendix III – Glossary of Terms

Alpha-fetoprotein (AFP) − a protein that is normally produced by the fetus. Maternal serum AFP levels can be used as a biochemical marker in the detection of certain fetal abnormalities.

Amniocentesis − a procedure involving the withdrawal of a small amount of amniotic fluid by needle and syringe through the abdomen guided by ultrasound performed at the same time. The tests performed on fetal cells found in the sample can detect a range of chromosomal and genetic disorders.

Analyte − a substance that is undergoing analysis or being measured. Analytes measured in antenatal screening include pregnancy associated plasma protein-A, beta-human chorionic gonadotrophin, unconjugated oestriol, alpha fetoprotein, and inhibin A.

Aneuploidy – is the condition of having less than or more than the normal diploid number of chromosomes. For instance, Down syndrome has 47 (not 46) chromosomes with an extra chromosome 21.

Beta-human chorionic gonadotropin (ßhCG) − a hormone produced during pregnancy and present in maternal blood and urine. It is used as a biochemical marker for Down syndrome and other conditions in first trimester combined and second trimester maternal serum screening.

Biparietal Diameter (BPD) – the measurement of the distance between the fetal parietal bones at their widest point. This is used for dating in the second trimester.

Chorionic villus sampling (CVS) – a procedure involving the withdrawal of a small amount of placental tissue by needle and syringe through the abdomen guided by ultrasound performed at the same time. Tests performed on the placental cells can detect a range of chromosomal and genetic disorders.

Crown rump length (CRL) − the measurement from the fetal crown to the prominence of the buttocks or breech. This is used for dating in the first trimester.

Chromosome − an organised structure of DNA and protein found in all living cells that carries the genes determining heredity.

False negative result − when a wāhine receives a low chance screening result but pēpi does have the condition screened for.

False positive result − when a woman receives an increased chance screening result but pēpi does not have the condition screened for.

Inhibin A − a hormone secreted by the ovary that is used as a biochemical marker in second trimester maternal serum screening for Down syndrome and other conditions.

Mosaic – the presence of two populations of cells with different genotypes in one patient, where usually one of the two is affected by a genetic disorder.

Multiple of the median (MoM) − a measure which compares the values of a biochemical marker in an individual sample with the median value of that biochemical marker in other women at the same gestation.

Neural tube defect (NTD) − a congenital anomaly involving the brain and spinal cord caused by failure of the neural tube to close properly during embryonic development. Open NTDs occur when the brain and/or spinal cord are exposed at birth through a defect in the skull or vertebrae. Examples of open NTDs are spina bifida (myelomeningocele), anencephaly, and encephalocele.

Non-invasive prenatal screening (NIPS) – a screening test that is determines the chance of pēpi having certain chromosomal abnormalities such as trisomy 21 (Down syndrome), trisomy 18 (Edwards syndrome) and trisomy 13 (Patau syndrome) as well as other conditions. Most NIPS tests offer fetal sex and sex chromosome aneuploidy detection in addition to trisomies 21, 18 and 13. The accuracy of the test changes depending on the disorder in question.

Nuchal translucency (NT) − sonographic appearance of the collection of fluid under the skin at the back of the fetal neck. NT is a marker for chromosomal and other anomalies and can be measured in the first trimester of pregnancy.

Pregnancy-associated plasma protein A (PAPP-A) − a protein originating from the placenta used as a biochemical marker in first trimester combined screening for Down syndrome and other conditions.

Screening calculation algorithm − an explicit protocol (in this case computer based) that combines a number of factors in determining overall chance of a particular outcome or condition.

Screening − a way of identifying people who are more likely than others to have a particular condition. The screening process involves testing people for the presence of the condition and predicting the likelihood that they have the condition. Antenatal screening for Down syndrome and other conditions predicts the likelihood of pēpi having the conditions.

Sensitivity − the ability of screening to identify individuals with the condition screened for. A test with high sensitivity will have few false negative results.

Specificity − the ability of screening to identify individuals who do not have the condition screened for. A test with high specificity will have few false positive results.

Threshold − the point that divides people into a group at lower chance or increased chance for the condition being screened for. In Aotearoa New Zealand the threshold is 1:300.

Triploidy − an extremely rare chromosomal disorder in which pēpi has three of every chromosome making a total of sixty-nine rather than the normal forty-six chromosomes.

Trisomy − a group of chromosomal disorders in which there are three copies, instead of the normal two, of a particular chromosome present in the cell nuclei. The most common trisomies in newborns are trisomy 21 (Down syndrome), trisomy 18 (Edwards syndrome) and trisomy 13 (Patau syndrome).

Unconjugated oestriol (uE_3) − a hormone produced by the placenta and used as a biochemical marker in second trimester maternal serum screening for Down syndrome and other conditions.