Antenatal Screening for Down Syndrome and Other Conditions

Guidelines for health practitioners

Antenatal and newborn screening

The National Screening Unit (NSU) of the Ministry of Health is responsible for the development, implementation and management of three antenatal and newborn screening programmes:

- Universal Offer Antenatal HIV Screening Programme
- Newborn Metabolic Screening Programme
- Universal Newborn Hearing Screening and Early Intervention Programme.

The NSU is also responsible for the introduction of quality improvements to antenatal screening for Down syndrome and other conditions.

Quality improvements to antenatal screening for Down syndrome and other conditions have been introduced to bring this screening in New Zealand into line with international best practice. While all pregnant women are advised about this screening, it is optional. It is made available so that those women who wish to have this information about their baby are able to find out during their pregnancy and plan accordingly.

These guidelines replace the *Guidelines for maternity providers offering antenatal screening for Down syndrome and other conditions* in New Zealand dated November 2009.

Acknowledgements

The National Screening Unit would like to thank the many individuals and groups who contributed to the development of these guidelines.

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MINISTRY OF HEALTH
MANATŪ HAUORA

Contents

Key messages

1. Antenatal screening for Down syndrome and other conditions provides a risk estimate for Down syndrome (trisomy 21), trisomy 18 (Edwards syndrome), trisomy 13 (Patau syndrome) and some other rare genetic disorders. **Page 2**
2. Detection of fetal anomalies through this screening offers women information that may help them prepare for the birth of their child: the option of delivery in a setting that has access to specialist surgical or medical care; the possibility of considering termination; or palliative care in the newborn period. **Page 2**
3. Antenatal screening for Down syndrome and other conditions is optional for pregnant women. **Page 10**
4. The right to decline screening, decline tests or further investigations should be made clear by the health professional and any such decision by the woman, including withdrawal of consent, must be respected. **Page 10**
5. Women who are less than 20 weeks pregnant must be advised about the availability of antenatal screening for Down syndrome and other conditions. **Page 10**
6. Up-to-date information about antenatal screening for Down syndrome and other conditions must be provided to support the screening discussion, thus enabling women to make an informed decision whether to accept or decline. **Page 10**
7. Informed choice for this screening must include a discussion about the screened conditions and the decisions that may need to be made as a result of participation in this screening. **Page 10**
8. No single test checks for everything. No screening test finds all cases of a condition. **Page 10**
9. A thorough family history should be taken and where there is a family history of a genetic condition, a referral for a discussion with a specialist obstetrician or genetic services should be offered prior to screening. **Page 15**
10. First trimester combined screening should be completed between 9 weeks and 13 weeks 6 days gestation. **The recommended timing for the blood test is 9 to 10 weeks and for the Nuchal Translucency scan is at 12 weeks.** **Page 17**
11. Second trimester maternal serum screening should be completed between 14 weeks and 20 weeks gestation. **The recommended timing for this test is 14 to 18 weeks.** **Page 17**
12. A very high or very low level of the blood markers used in screening may indicate other conditions such as pre-eclampsia or pre-term birth. Fetal anomalies such as a heart condition or structural defect may be found on ultrasound. **Page 21**
13. Screening for neural tube defects (NTD) can be reported after 15 weeks of pregnancy using alpha fetoprotein (AFP) as the serum marker. It is noted that a better predictor of NTD is the 18 to 20 week anatomy scan. **Page 21**
14. The health professional requesting screening must fill in all sections of the screening request form to ensure an accurate risk assessment. **Page 18**
15. 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. **Page 9**

List of abbreviations

AFP	Alpha-fetoprotein
ASUM	Australasian Society for Ultrasound in Medicine
ßhCG	Beta-human chorionic gonadatrophin
BPD	Biparietal diameter
CRL	Crown–rump length
CVS	Chorionic villus sampling
DHB	District Health Board
EDD	Estimated date of delivery
FMF	Fetal Medicine Foundation
IVF	In vitro fertilisation
LMC	Lead maternity carer
LMP	Last menstrual period
MoM	Multiple of the median
NSU	National Screening Unit of the Ministry of Health
NTD	Neural tube defect
NT	Nuchal translucency
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
uE_3	Unconjugated oestriol

1. Introduction

These guidelines support health practitioners advising about the availability of services for antenatal screening for Down syndrome and other conditions. They are intended for all practitioners involved in aspects of antenatal screening for Down syndrome and other conditions including:

- lead maternity carers (LMCs)
- midwives
- general practitioners
- nurses
- sonologists, sonographers, radiologists
- obstetricians, fetal medicine specialists
- screening laboratory staff.

Health practitioners advising women about maternity care have an obligation under the Primary Maternity Services Notice 2007, issued pursuant to section 88 of the New Zealand Public Health and Disability Act 2000, to advise women of screening services available that are endorsed by the Ministry of Health, including antenatal screening for Down syndrome and other conditions.

During the screening process, the health practitioner is responsible for:

(a) providing information and education about antenatal screening to pregnant women

(b) supporting women to make an informed decision

(c) offering referrals as agreed with the woman

(d) communicating screening results

(e) ensuring documentation of screening discussions and choices in the clinical notes

(f) ensuring compliance with the:

- Privacy Act 1993 and Health Information Privacy Code 1994
- New Zealand Public Health and Disability Act 2000
- Code of Health and Disability Services Consumers' Rights 1996
- Health Act 1956
- Health Practitioners Competence Assurance Act 2003
- Public Records Act 2005
- Crimes Act 1961 (as amended).

It is strongly recommended that health practitioners complete the e-learning modules at www.learnonline.health.nz which have been approved as professional development by the Midwifery Council of New Zealand and the Royal Australasian College of General Practitioners (RNZGP).

Other screening resources are available for health practitioners at www.nsu.govt.nz

2. Background

Antenatal screening for Down syndrome and other conditions has been available to pregnant women in 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. The objective of the quality improvements initiative is to ensure that the screening tests available for pregnant women in New Zealand provide the best possible information, so that women can make an informed decision about the way their pregnancy is managed.

2.1 Overview of antenatal screening for Down syndrome and other conditions

Detection of fetal anomalies through this screening offers women information that may help them prepare for the birth of their child: the option of delivery in a setting that has access to specialist surgical or medical care; the possibility of considering termination; or palliative care in the newborn period.

Antenatal screening for Down syndrome and other conditions is a way of assessing the probability that a baby has Down syndrome or another genetic condition and offers women information and choice in the care and management of their pregnancy and baby's birth.

Antenatal screening for Down syndrome and other conditions has complex ethical and social implications. In addition there are technical considerations which involve a trade-off between the sensitivity (detection rate) and the specificity (false positive rate) of the screening tests. The combination of ultrasound and maternal serum markers increases detection rates (improves sensitivity) and/or reduces the number of women considered to be at increased risk (improves specificity), compared with previous first trimester screening practice.

These guidelines refer to the risk estimate calculations and reports. Health practitioners are encouraged to use different methods for communicating individual risk results to women and this may include words such as chance or likelihood. The use of risk in these guidelines is synonymous with chance used in the consumer information.

2.2 The screening options

Antenatal screening for Down syndrome and other conditions provides a risk estimate for Down syndrome (trisomy 21), trisomy 18 (Edwards syndrome), trisomy 13 (Patau syndrome) and some other rare genetic disorders.

This screening divides women into two groups based on risk, either increased risk or low risk. Table 1 outlines the screening options.

First trimester combined screening involves a nuchal translucency (NT) scan and maternal serum testing. The risk 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. Women will receive one combined result from their health practitioner, after they have had both the blood test and the NT scan. The incorporation of serum results in the risk calculation significantly increases the sensitivity and specificity of screening and provides a better risk assessment than NT scanning in isolation[1].

1. Wald NJ, Rodeck C, Hackshaw AK, Walters J, Chitty L, Mackinson AM. 2003. First and second trimester antenatal screening for Down's syndrome: the results of the Serum, Urine and Ultrasound Screening Study (SURUSS). *Health Technol Assess* 7(11): 1-77.

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 risk report.

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

- do not access maternity care early in their pregnancy
- do not have access to NT scanning for geographic, economic or other reasons
- have not completed first trimester combined screening
- prefer second trimester screening.

Table 1: The screening options available

First trimester combined screening (9 – 13w and 6 days)	OR	Second trimester maternal serum screening (14 – 20 weeks)	AND	Recommendations for practice
> Blood test that measures two maternal serum markers (PAPP-A and ßhCG) combined with an ultrasound scan to determine NT and CRL measurements. > Available to all women who present in the first trimester. > The blood test is fully funded. > Women may be required to make a co-payment for the ultrasound scan. (The Ministry of Health funds the ultrasound provider on a fee for service basis for each NT scan).		> Blood test that measures four maternal serum markers (ßhCG, AFP, uE_3 and inhibin A). > Available to women who present after the first trimester or who do not access first trimester combined screening. > The blood test is fully funded.		> The discontinuation of the use of maternal age and nuchal translucency as screening tools in isolation.
Provision of accurate, balanced information (both medical and non-medical).				
Support for decisions made by women throughout pregnancy, including the decision as to whether or not to participate in screening.				
Support for women who want their family/whānau to be actively involved.				

2.3 Screening performance

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.

In New Zealand, the screening cut-off is 1:300. As an example, a woman with a risk result of 1:250 will be increased risk and 1:350 will be low risk.

Based on international data, this screening finds approximately 85% of babies with Down syndrome. Approximately 5% of women will receive an increased risk result.

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

All pregnant women must be advised about the availability of antenatal screening for Down syndrome and other conditions, including the risks, benefits and harms of screening, so that they may make an informed decision to participate in screening or not. Screening poses different ethical considerations from those that arise when a person presents for medical care because they are unwell. Health practitioners have a special duty of care when advising women of screening.

The 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 risk results for the screened conditions
- reassurance associated with no abnormalities found through scanning.

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

- anxiety and stress associated with the screening process
- women having a poor understanding of the screening process. This may include a lack of understanding of risk estimates and what may or may not be detected
- anxiety and stress associated with an increased risk result which may be a false positive result
- false reassurance when a low risk result is given when the baby does have a condition ie, a false negative result
- a miscarriage resulting from diagnostic procedures following an increased risk result.

3. General requirements

3.1 Code of Health and Disability Services Consumers' Rights

The Code of Health and Disability Services Consumers' Rights provides that New Zealand health care consumers have a legal right to appropriate information to enable informed consent. Information about the Code can be obtained from the Health and Disability Commissioner's website, www.hdc.org.nz

3.2 Health Information Privacy Code

The Health Information Privacy Code 1994 (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.

For the health sector, the HIPC takes the place of the information privacy principles set out in the Privacy Act 1993. The HIPC can be viewed at the Privacy Commissioner's website, www.privacy.org.nz

The 12 rules of the HIPC require agencies to be clear about the purpose for which they collect information, and open about those purposes to the health consumers they collect it from. 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.

3.3 Ensuring services meet the needs of women

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

Achieving Equitable Outcomes

Guidelines	Information
Health services should be attuned to the needs of individuals, families and communities.	Health services should be tailored to meet the health needs of all New Zealanders, including Māori, Pacific peoples and Asian populations. Health practitioners should recognise that what works for different populations varies. Health practitioners should familiarise themselves with Whānau Ora found at www.health.govt.nz and the NZ Disability Strategy found at www.odi.govt.nz
Health practitioners should offer additional support to women who have difficulty understanding information because of language difficulties, hearing impairment or intellectual disability.	Appropriate information that allows for informed consent includes using professional interpreter services, such as Language Line, a DHB interpreter or a NZ Sign Language interpreter where necessary. Using family members or friends as interpreters is not recommended practice. Women who have an intellectual disability may require extra support or the presence of family/whānau or other support people (eg independent advocate, welfare guardian) to understand the information and assist their decision making.
Health practitioners should recognise other barriers to access.	Barriers to accessing aspects of antenatal care and screening may include lack of knowledge, fear of health services, different cultural views of health, the location and cost of ultrasound services, the availability of transport, travel time and child care.

4. The screening pathway

Figure 1 details the screening pathway. It is the woman's choice whether to accept screening or not and she may also accept or decline any referral or test.

The points along the pathway where the woman needs to make an informed decision are:

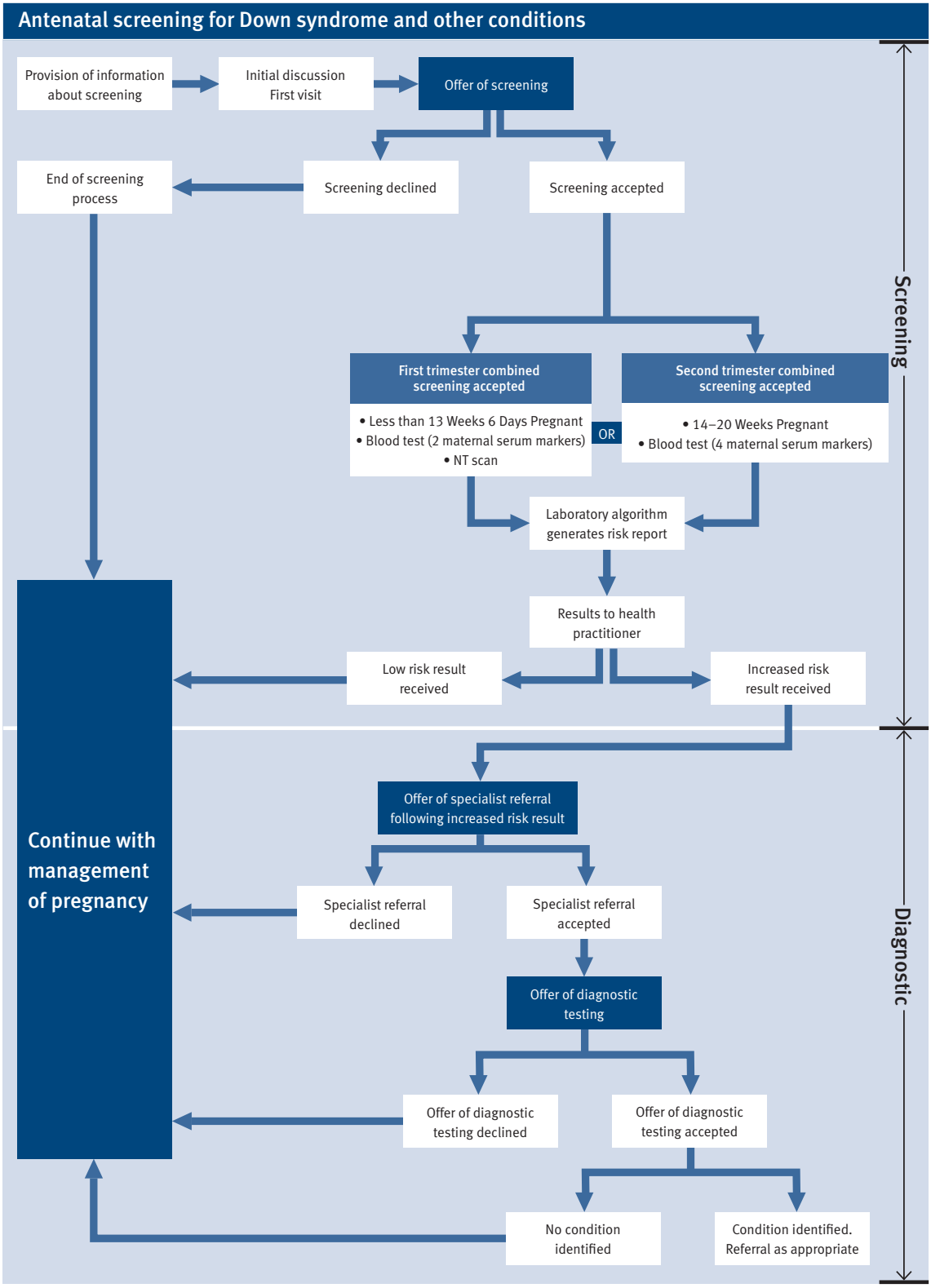
(a) whether or not to be screened

(b) whether to accept an offer of referral to a specialist obstetrician or specialist medical maternity service or Genetic Services to gather more information following an increased risk or abnormal result

(c) whether to have diagnostic testing following a referral and discussion

(d) when deciding the next step after receiving the results of the diagnostic test.

Figure 1: The screening pathway



5. Documentation

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.

Details of discussions and decisions must be documented by the health practitioner in the clinical records. Signed consent for antenatal screening for Down syndrome and other conditions is not required by the Code of Health and Disability Services Consumers' Rights.

Clinical Records

Guidelines	Information
Health practitioners must document discussions and decisions in the clinical notes.	Each stage of the screening process should be documented in the clinical records including: > the content of discussions with the woman > 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 the woman on results received > other support, resources or information offered or provided.
Health practitioners who are referring or handing over care to another health practitioner must 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.

6. Informed choice – consent or decline

Women who are less than 20 weeks pregnant must be advised about the availability of antenatal screening for Down syndrome and other conditions.

Antenatal screening for Down syndrome and other conditions is optional for pregnant women.

The right to decline screening, decline tests or further investigations should be made clear by the health professional and any such decision by the woman, including withdrawal of consent, must be respected.

Informed choice for this screening must include a discussion about the screened conditions and the decisions that may need to be made as a result of participation in this screening.

Up-to-date information about antenatal screening for Down syndrome and other conditions must be provided to support the screening offer, thus enabling women to make an informed decision whether to accept or decline.

No single test checks for everything. No screening test finds all cases of a condition.

Ensuring women make an informed decision about antenatal screening for Down syndrome and other conditions is a legal requirement under the Code of Health and Disability Services Consumers' Rights that is central to best practice in maternity care.

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

Information to Women

Guidelines	Information
Health practitioners must advise women about the availability of antenatal screening for Down syndrome and other conditions which is available to all pregnant women under 20 weeks gestation.	The discussion of screening must be made with sufficient information, advice and time to enable women to make an informed decision. The discussion with women should be initiated by the health practitioner as early as possible in the pregnancy, to allow the opportunity to ask questions, seek further information and consider participation. Women should be informed that only one screening option will be publicly funded in each pregnancy. Only one risk result will be issued. Women should be informed that screening provides a risk estimate and not a definitive result. Some women may wish to discuss their options with family/whānau.

Informed Consent or Decline

Guidelines	Information
Health practitioners must provide up-to-date, balanced information about the screened conditions. Health practitioners must ensure women are aware this screening is optional. Health practitioners must respect and support all screening choices the woman makes.	Information should include the Ministry of Health consumer pamphlet *Antenatal screening for Down syndrome and other conditions: optional screening – your choice, your decision*. Discussion about screening should include that there may be unexpected findings. Refer to Appendix 2 for more details. Informed consent is a process that must occur throughout the screening pathway. Ensuring informed choice includes: › provision of information about screening › offering screening in a non-directive manner › discussions about screening before and during the screening process › discussions about options following an increased risk screening result › giving sufficient time to consider options › documenting discussions and consent or decline to screening › assuring the woman that whatever choice she makes will be supported.

Communication

Guidelines	Information
Health practitioners must answer questions women ask regarding screening.	Women should be given the opportunity to ask questions about this screening and advised where they can find further information. Health practitioners may need to seek advice from other sources to assist them to answer questions from women for example from screening laboratories, radiology practices, obstetricians, paediatricians, Genetic Services and/or maternal fetal medicine specialists.

Communication *continued*

Guidelines	Information
Health practitioners should give women time to reflect and consider decisions.	Health practitioners should be cognisant of the important implications for pregnant women when screening is discussed. Where there is a history of pregnancy problems and/or genetic conditions in the family, women may have given careful thought to how they want to proceed with this pregnancy. A woman's decision whether or not to consent to screening will depend on her personal values and beliefs and the information available to her. Women will have a range of different views that may change over time. Some women are very clear what choices they may make if a condition is confirmed and others are not. This screening (and the implications of screening) involves complex ethical issues and women may want to explore these before giving consent to begin screening or to continue screening. For some women, screening is an opportunity to have information that enables them to choose the care and management of their pregnancy and birth. Screening and diagnostic testing can inform the choice of location for the birth, for instance a tertiary unit with specialist care. Women may wish to consider the choices following an increased risk result. This may include a discussion about: › diagnostic testing including the risks › what a positive diagnostic result may mean for her. If a woman does not wish to know further information about the pregnancy then she may choose not to have screening.

Screened Conditions

Guidelines	Information
Health practitioners must discuss the conditions that may be indicated as a result of participation in this screening.	Conditions include: > risk estimates for trisomy conditions: > T21 (Down syndrome) > T18 (Edwards syndrome) > T13 (Patau syndrome) > other rare genetic conditions for example: > Smith-Lemli-Opitz > Turner syndrome > Triploidy All the conditions above will require diagnostic testing for confirmation.
	Pregnancy conditions associated with poor placentation including: > stillbirth > miscarriage > growth restriction > preterm birth > pre-eclampsia. The ultrasound scan may show structural anomalies for instance: > cardiac > neural tube > renal > central nervous system. The ultrasound scan may also suggest a growth anomaly. The conditions above require referral as appropriate.
	As with any medical test, there is a chance that other conditions may be unexpectedly identified and a recommendation for further discussion with a specialist may be made. Screening is not able to detect all conditions that may be present. The results will provide a risk estimate for T21, T18 and T13 as well as incidental findings. Further advice can be obtained from the laboratory if required.

Screened Conditions *continued*

Guidelines	Information
Health practitioners must discuss with the woman that the 18 to 20 week anatomy scan is another screening test.	The 18 to 20 week anatomy scan is not included in this screening, however, it may identify potential markers for one of the screened conditions.

A thorough family history should be taken and where there is a family history of a genetic condition, a referral for a discussion with a specialist obstetrician or genetic service should be offered prior to screening.

Family History

Guidelines	Information
Health practitioners must discuss and document family history with the woman and advise the laboratory of any relevant details.	If a woman has previously been pregnant with or has had a child with a genetic condition, or a family history of a genetic condition, they may have a different risk status. Appropriate family history questions (on both sides of the family) may include: › previous recurrent miscarriage › previous pregnancy loss (still birth or neonatal death) › fetal or childhood abnormalities (for example neural tube defects) › developmental delay in other children › any genetic conditions including childhood diseases, cystic fibrosis, muscular dystrophy or neurodegenerative conditions. The practitioner may wish to consult with Genetic Services prior to determining a management plan. It may be appropriate that the woman is offered a referral to a specialist obstetrician or specialist medical maternity service or Genetic Services. Health practitioners should exercise particular sensitivity and be aware that women may have already given careful thought to having or not having screening or diagnostic testing.

A referral form for Genetic Services is available at www.nsu.govt.nz

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

6.1 Initial discussion

The discussion should include the following information.

(a) Screening information

- purpose of screening
- screening options available and what screening involves
- screened conditions
- detection of structural anomalies on the NT scan
- screening does not cover every condition
- incidental information may be found relating to the pregnancy or baby
- recommended timing for screening
- screening pathway and the decision points and options for screening
- screening provides a risk estimate only and is not diagnostic
- a further test would be required to determine whether a condition is present
- reliability of screening
- which tests may incur charges
- further information about the baby may be identified at the 18 to 20 week 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 a woman may choose to participate or not participate in first or second trimester screening and change her mind about this decision
- the woman may choose to participate in second trimester screening having declined the offer of first trimester combined screening
- women who choose not to participate in screening will not have their maternity care affected in any way
- if screening shows an increased risk result, diagnostic testing will be offered.

(d) Results

- how results are notified
- women will receive one risk result for first trimester combined screening combining NT scan measurements and serum markers
- when screening results are available
- the screening will provide an increased or low risk result and may also indicate other anomalies through ultrasound or serum markers
- for every 1000 women screened, about 50 will receive an increased risk result and 2 will be diagnosed with a baby with Down syndrome or another condition
- woman's preference for receiving her results.

(e) Data and information collection and monitoring

- information and data is collected and securely stored
- information is used for monitoring and quality improvements of this screening
- that 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.

7. The screening tests

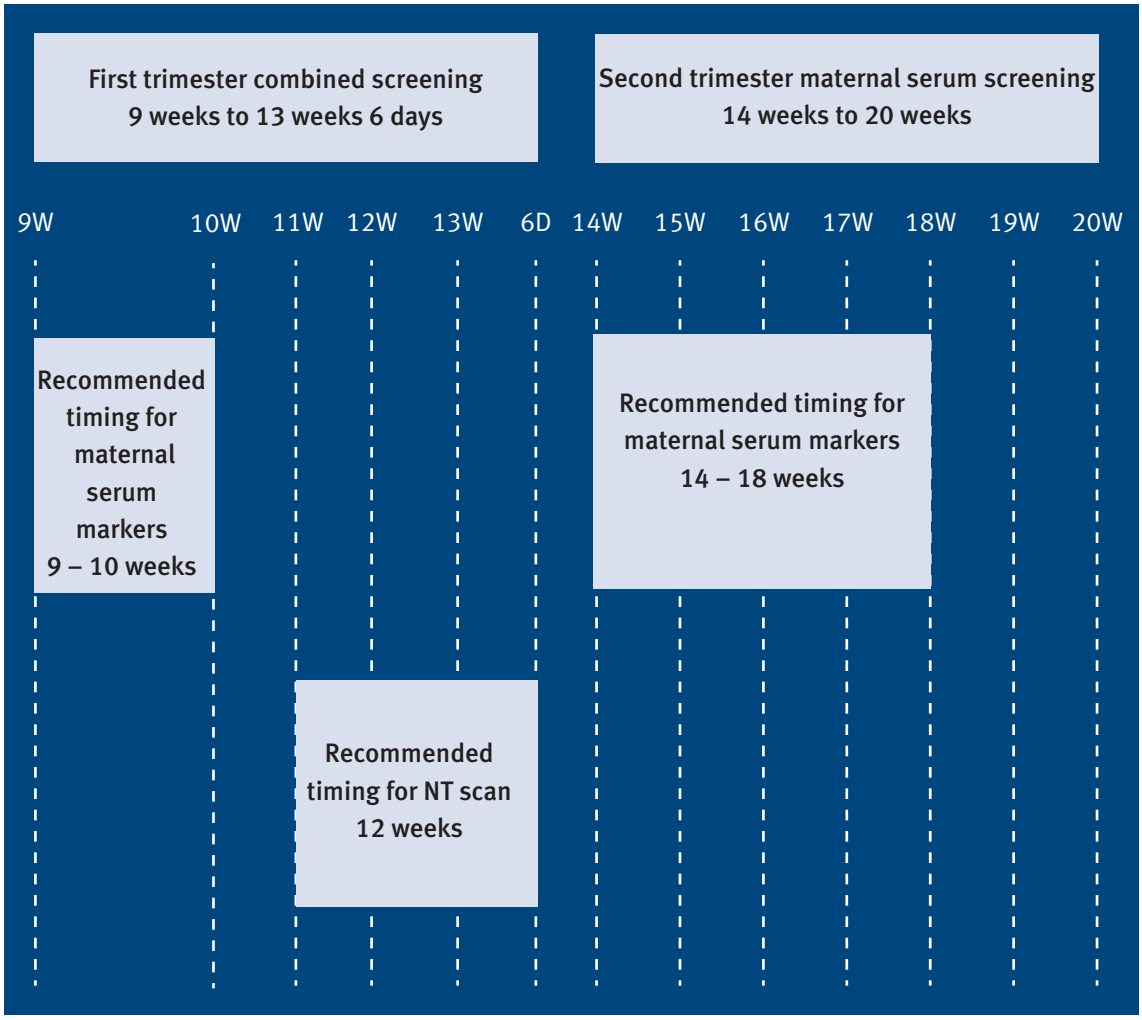
7.1 Timing of first trimester combined OR second trimester maternal screening tests

First trimester combined screening should be completed between 9 weeks and 13 weeks 6 days gestation. THE RECOMMENDED TIMING FOR THE BLOOD TEST IS 9 TO 10 WEEKS AND FOR THE NT SCAN AT 12 WEEKS.

Second trimester maternal serum screening should be completed between 14 weeks and 20 weeks gestation. THE RECOMMENDED TIMING FOR THIS TEST IS 14 TO 18 WEEKS.

Figure 2 shows when the different antenatal screening tests for Down syndrome and other conditions may be undertaken.

Figure 2: Timing of first trimester combined screening and second trimester serum screening



7.2 Ordering tests

If a woman accepts screening, the health practitioner (midwife or doctor) will complete the screening request form.

Eligibility criteria for publicly funded services can be found at www.moh.govt.nz

First Trimester Screening

Guidelines	Information
Health practitioners should inform women where they can go for their blood test and NT scan and the timing of each of these tests for first trimester combined screening. Referrals for the NT ultrasound must be made in accordance with Section 88 of the New Zealand Public Health and Disability Act 2000.	For first trimester combined screening, health practitioners must ensure the woman knows there are two components, blood test and ultrasound scan, and she needs to have each within certain timeframes. The woman is usually required to make a co-payment for the NT scan.

Second Trimester Screening

Guidelines	Information
Health practitioners should inform women where they can go for the blood test and the timing of the test for second trimester screening.	Second trimester serum screening is fully funded for eligible women.

7.3 Completing the screening request form

The health professional requesting screening must fill in all sections of the screening request form to ensure an accurate risk assessment.

Screening Request Form

Guidelines	Information
The screening request form must be completed with all the requested information.	All information requested on the laboratory request form is needed by the laboratory to ensure high quality testing. Screening results may be inaccurate if the information on the screening request form is not completed.

The current screening request form is a carbon copy, with the duplicate copy to be used for NT scan requests. Figure 3 provides further explanation.

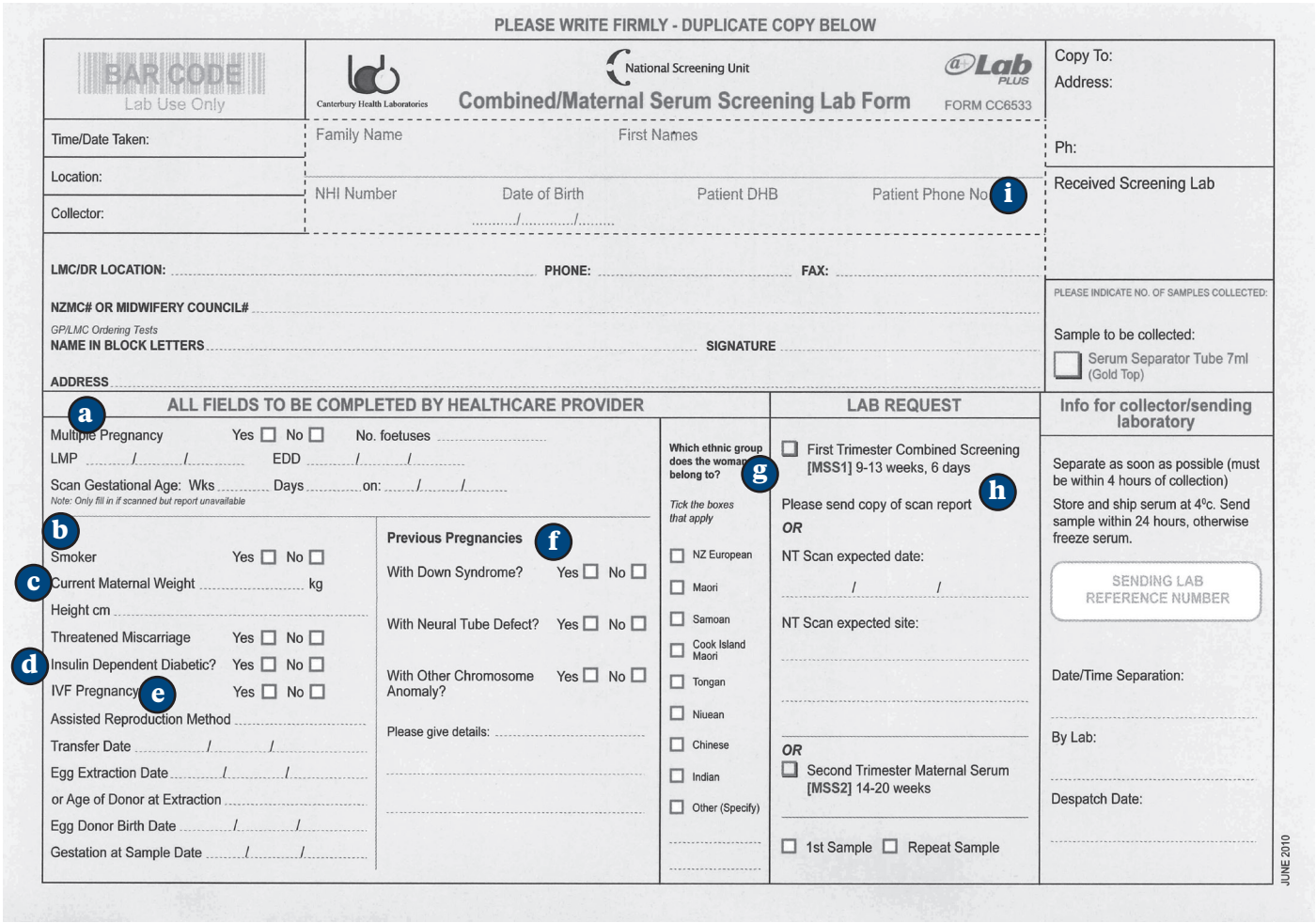
From time to time, the screening request form format may change, however, the information required for accurate risk assessment will stay the same.

Two laboratories perform antenatal screening for Down syndrome and other conditions. LabPLUS (Auckland District Health Board) are responsible for screening women from Taupo north. Canterbury Health Laboratories (Canterbury District Health Board) are responsible for screening women south of Taupo.

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)

Figure 3: The screening request form



PLEASE WRITE FIRMLY - DUPLICATE COPY BELOW

BAR CODE
Lab Use Only

Canterbury Health Laboratories

National Screening Unit

Combined/Maternal Serum Screening Lab Form

FORM CC6533

Copy To:
Address:
Ph:

Time/Date Taken:
Location:
Collector:

Family Name First Names

NHI Number Date of Birth Patient DHB Patient Phone No (i)

Received Screening Lab

LMC/DR LOCATION: PHONE: FAX:

NZMC# OR MIDWIFERY COUNCIL#

GP/LMC Ordering Tests
NAME IN BLOCK LETTERS SIGNATURE

ADDRESS

PLEASE INDICATE NO. OF SAMPLES COLLECTED:

Sample to be collected:
Serum Separator Tube 7ml (Gold Top)

ALL FIELDS TO BE COMPLETED BY HEALTHCARE PROVIDER

(a) Multiple Pregnancy Yes No No. foetuses
LMP / / EDD / /
Scan Gestational Age: Wks Days on: / /
Note: Only fill in if scanned but report unavailable

(b) Smoker Yes No
(c) Current Maternal Weight kg
Height cm
Threatened Miscarriage Yes No
(d) Insulin Dependent Diabetic? Yes No
IVF Pregnancy (e) Yes No
Assisted Reproduction Method
Transfer Date / /
Egg Extraction Date / /
or Age of Donor at Extraction
Egg Donor Birth Date / /
Gestation at Sample Date / /

Previous Pregnancies (f)
With Down Syndrome? Yes No
With Neural Tube Defect? Yes No
With Other Chromosome Anomaly? Yes No
Please give details:

Which ethnic group does the woman belong to? (g)
Tick the boxes that apply
NZ European
Maori
Samoan
Cook Island Maori
Tongan
Niuean
Chinese
Indian
Other (Specify)

LAB REQUEST

First Trimester Combined Screening [MSS1] 9-13 weeks, 6 days
Please send copy of scan report (h)
OR
NT Scan expected date: / /
NT Scan expected site:
OR
Second Trimester Maternal Serum [MSS2] 14-20 weeks
1st Sample Repeat Sample

Info for collector/sending laboratory

Separate as soon as possible (must be within 4 hours of collection)
Store and ship serum at 4°c. Send sample within 24 hours, otherwise freeze serum.

SENDING LAB REFERENCE NUMBER

Date/Time Separation:
By Lab:
Despatch Date:

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(a) **Multiple pregnancies**: serum analyte levels come from the baby and placenta so will differ for multiple pregnancies.

(b) **Smoking**: affects placental function and serum analyte levels. The default if not provided is a non-smoker.

(c) **Weight**: smaller women have higher serum analyte levels and larger women have lower serum analyte levels. This has a significant impact on the risk calculation.

(d) **Diabetes**: serum analyte levels differ for diabetic women.

(e) **Gestation and IVF**: levels of serum analytes change through the pregnancy, so dating is important for accurate risk calculation. Essential information includes accurate gestation details including the method and age of the woman at the time of donation/ retrieval.

(f) **Previous pregnancy details**: these are added into the risk calculation as they affect the chance of the current pregnancy. Add any relevant family history.

(g) **Mother's ethnicity**: serum analyte levels vary with different ethnicities.

(h) **NT scan expected date and site**: this relates to which practice the woman is most likely to go to for her NT scan. The laboratory will contact the radiology practice if they have not received an NT scan report by 13 weeks.

(i) **Patient and health practitioner details**: this section ensures the right patient is linked to the right result and sent to the right health practitioner. Your phone number ensures you can be contacted to check details or provide results. Please provide alternative details if you will be away.

8. Receiving and communicating results

A very high or very low level of the blood markers used in screening may indicate other conditions such as pre-eclampsia or pre-term birth. Fetal anomalies such as a heart condition or structural defect may be found on ultrasound.

Screening for neural tube defects (NTD) can be reported after 15 weeks of pregnancy using alpha fetoprotein (AFP) as the serum marker. It is noted that a better predictor of NTD is the 18 to 20 week anatomy scan.

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 or scan information, whichever is the later (if first trimester combined screening). Screening results include information provided about the woman, details of the risks 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.

Receiving Screening Results

Guidelines	Information
The health practitioner is responsible for receiving screening results.	It is useful to ascertain the woman's preference for receiving results at the time that the screening offer is made. The woman may wish to be accompanied by family/whānau or support person. If the screening result is low risk the screening laboratory will dispatch the result to the health practitioner by mail or electronic means within 24 hours of the result being available. If the screening result is increased risk the screening laboratory will phone 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. There are a number of situations where the markers (both serum and the NT scan) may indicate other conditions. Refer to Appendix 2 for further information. If the screening result will indicate other conditions, the screening laboratory will phone the health practitioner within 24 hours of the result being available.

Preparing for Communicating Screening Results

Guidelines	Information
Health practitioners must communicate results to women in an appropriate and timely manner.	Prior to communicating with the woman, the health practitioner should consider discussing the result with the screening laboratory or Genetic Services. Communication to women 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 must not be sent via a text message. 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). Health practitioners should be able to present results in a clear and concise way to support women in their decision-making. This includes understanding statistical risk information. It can be useful to communicate the risk result in different ways to help women 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.' Further information about the risk result can be obtained from the screening laboratory.
The health practitioner should be prepared to discuss the results in detail and seek further information that supports them to inform women what their screening results may mean.	Discussion around the results may include: › the limitations of screening › that a low risk 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 the woman (and her family/whānau) to ask questions › providing information about other services, including community support agencies the woman (and her family/whānau) can contact. If a woman with a low risk result requests diagnostic testing, a referral to a specialist obstetrician may be made.

Communicating Increased Risk Results and Offering Referral

Guidelines	Information
Health practitioners must inform women in a timely manner of all screening results indicating an increased risk 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 any increased risk 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 risk of a genetic condition, women 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 1. Document the discussion and management plan in the clinical notes. Provide the woman with a copy of the results if requested.

Figures 4 and 5 provide an example of the screening reports produced by the laboratories.

Figure 4: Example laboratory report: increased risk for Down syndrome

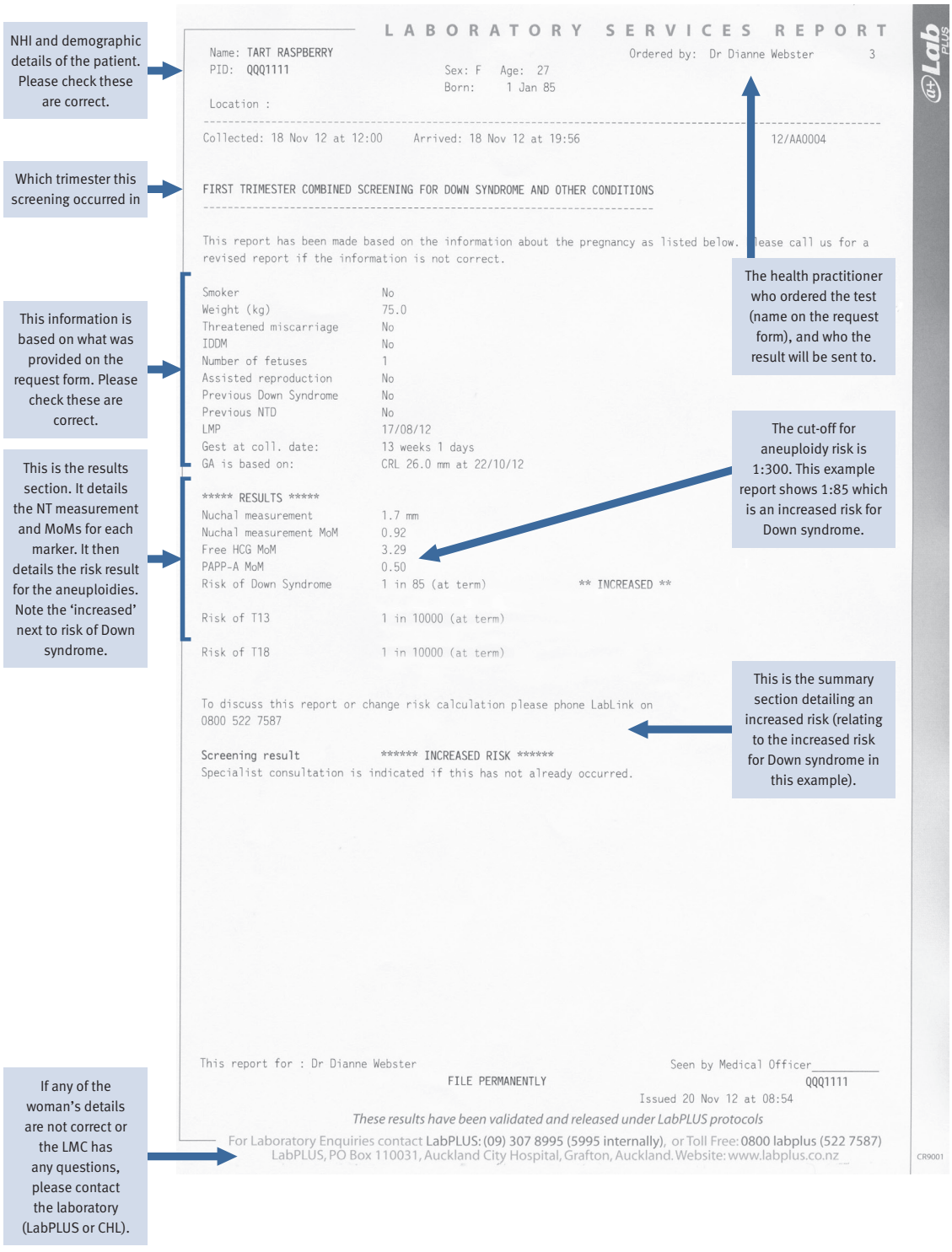
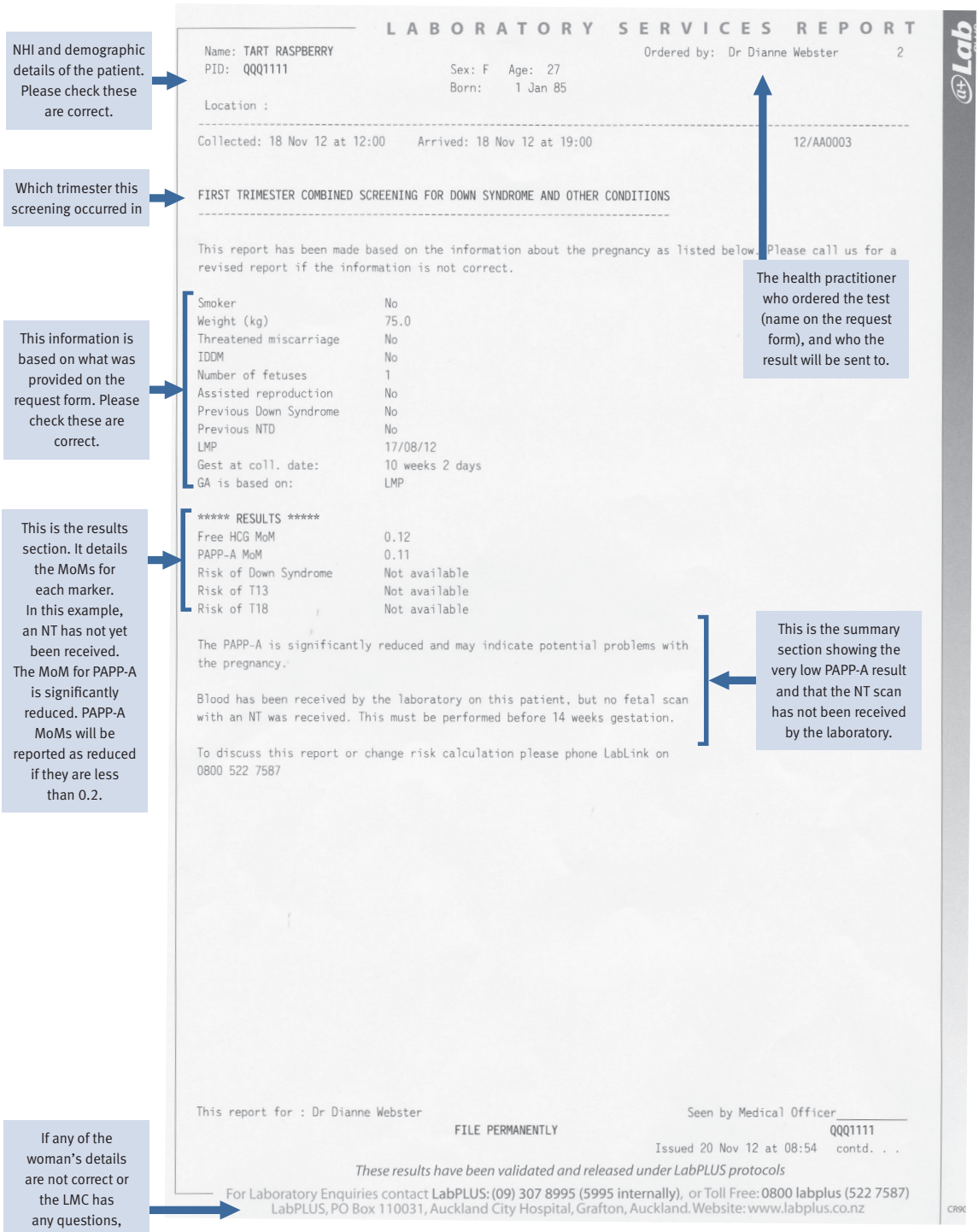


Figure 5: Example laboratory report: low PAPP-A (unusual analyte)



LABORATORY SERVICES REPORT

Name: TART RASPBERRY
PID: QQQ1111
Sex: F Age: 27
Born: 1 Jan 85
Ordered by: Dr Dianne Webster 2
Location :

Collected: 18 Nov 12 at 12:00 Arrived: 18 Nov 12 at 19:00 12/AA0003

FIRST TRIMESTER COMBINED SCREENING FOR DOWN SYNDROME AND OTHER CONDITIONS

This report has been made based on the information about the pregnancy as listed below. Please call us for a revised report if the information is not correct.

Smoker	No
Weight (kg)	75.0
Threatened miscarriage	No
IDDM	No
Number of fetuses	1
Assisted reproduction	No
Previous Down Syndrome	No
Previous NTD	No
LMP	17/08/12
Gest at coll. date:	10 weeks 2 days
GA is based on:	LMP

***** RESULTS *****

Free HCG MoM	0.12
PAPP-A MoM	0.11
Risk of Down Syndrome	Not available
Risk of T13	Not available
Risk of T18	Not available

The PAPP-A is significantly reduced and may indicate potential problems with the pregnancy.

Blood has been received by the laboratory on this patient, but no fetal scan with an NT was received. This must be performed before 14 weeks gestation.

To discuss this report or change risk calculation please phone LabLink on 0800 522 7587

This report for : Dr Dianne Webster
FILE PERMANENTLY
Seen by Medical Officer_____
QQQ1111
Issued 20 Nov 12 at 08:54 contd. . .

These results have been validated and released under LabPLUS protocols

For Laboratory Enquiries contact LabPLUS: (09) 307 8995 (5995 internally), or Toll Free: 0800 labplus (522 7587)
LabPLUS, PO Box 110031, Auckland City Hospital, Grafton, Auckland. Website: www.labplus.co.nz

NHI and demographic details of the patient. Please check these are correct.

Which trimester this screening occurred in

This information is based on what was provided on the request form. Please check these are correct.

This is the results section. It details the MoMs for each marker. In this example, an NT has not yet been received. The MoM for PAPP-A is significantly reduced. PAPP-A MoMs will be reported as reduced if they are less than 0.2.

If any of the woman's details are not correct or the LMC has any questions, please contact the laboratory (LabPLUS or CHL).

The health practitioner who ordered the test (name on the request form), and who the result will be sent to.

This is the summary section showing the very low PAPP-A result and that the NT scan has not been received by the laboratory.

Referral to Specialist

Guidelines	Information
Health practitioners must offer a timely referral to all women with increased risk results or unusual analytes.	Following an increased risk result a woman may be undecided about her next steps. She may require further information from other sources about: › what the increased risk 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 referral should include details of: › gestation › screening results › any issues identified which require further discussion › any relevant family history. 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. The woman may wish to seek further information from sources listed in Appendix 1. The woman must be given time to reflect and to consider her decision.
Health practitioners must make clear the woman's right to decline a referral following an increased risk result. Health practitioners must respect and support any decision made by the woman throughout the screening process.	The provision of links to community organisations to enable the gathering of more information and to access support may also be helpful. These can be found in Appendix 1.

9. 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 aneuploidy testing.

CVS can be performed from 11 to 14 weeks of pregnancy but is typically performed between 10 and 13 weeks. CVS is only offered in a few centres. CVS results may take one to three weeks.

Amniocentesis can be performed between 15 and 20 weeks. Amniocentesis results may take one to three weeks.

A more detailed scan may be required following an abnormal finding on ultrasound or from unusual analytes or positive NTD screen.

Diagnostic testing is publicly funded for women who have:

- an increased risk result
- an abnormal ultrasound scan (structural abnormalities)
- previously had a baby with a congenital anomaly
- maternal anxiety
- a family history of Down syndrome and/or other conditions, if recommended by Genetic Services.

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

Diagnostic Testing

Guidelines	Information
Health practitioners must provide information on diagnostic testing and inform the woman that diagnostic testing is optional.	Health practitioners should outline the following choices the woman has available: › 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 must explain: › what information the diagnostic tests can provide › the risks associated with a diagnostic test › the decisions that the woman 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. Fetal cells can be analysed in a number of ways. There is an option of more rapid tests which can provide a result in 24 to 48 hours. These tests are accurate but do not test for as many abnormalities as other testing. Further information is available from the laboratories. A charge may apply for these tests. If diagnostic testing is undertaken, the sample can be used for chromosome testing and for other specific genetic tests that may be indicated in the family history. These will need to be specified on the diagnostic testing form sent to the cytogenetics laboratory.

Diagnostic Testing *continued*

Guidelines	Information
Health practitioners must inform women of the risk of diagnostic testing procedures.	The risk of miscarriage after amniocentesis is about one miscarriage in every 100–200 women tested. The risk of miscarriage after CVS is about one miscarriage in every 50–100 women tested. Other rare complications include: › leaking of amniotic fluid from the vagina › infection of the uterus or fetus › some research has suggested that development of arms, fingers, legs or toes may be disrupted if CVS is performed before nine weeks gestation: for this reason, a CVS procedure is done after 10 weeks gestation. › development of Rhesus factor incompatibility. All women who have Rh-negative blood group are given an injection of anti-D to prevent this complication.
Specialist obstetricians who are performing diagnostic testing must specify the genetic tests required on the diagnostic testing form. This may include requests due to familial conditions.	If diagnostic testing is undertaken, the sample can be used for chromosome testing and for other specific genetic tests that may be indicated in family history. These will need to be specified on the diagnostic testing form sent to the cytogenetic laboratory.

Receipt of a Positive Result Following Diagnostic Testing

Guidelines	Information
The health practitioner must explain the meaning of any test results and provide information about any diagnosis.	Health practitioners can seek further information from the cytogenetic laboratory or other sources as required. See Appendix 2 for more information on the conditions. Women may wish to have time to consider the results and what they may mean for her and her baby.

After receiving the results of the diagnostic test, health practitioners should support women to make an informed decision.
 	
Health practitioners must support the woman's decision.
	The following could be discussed:
	> information about the condition
	> options available which include:
	  > continuing with the pregnancy
	  > termination of the pregnancy.
	
	If the woman chooses to continue with the pregnancy, the options for antenatal care such as specialist care and support, and postnatal options should be discussed. If the baby has a condition which has a very short life expectancy, consideration should be given to offering antenatal or postnatal palliative care for the baby and counselling services to the women (and her family).
	
	Women may need to be provided with support, which may include access to groups in Appendix 1.

Health practitioners must provide women with opportunities to access additional information and support.	This may include referral to a: > paediatrician > health social worker > Genetic Services > counsellor. The woman and/or her family/whānau may seek information from sources listed in Appendix 1 to find out what living with a specific condition may mean. Written resources for example, *Living With Down* and web links are available (see Appendix 1).

10. Genetic services and other referrals

Genetic Services

Guidelines	Information
Health practitioners should advise women with increased risk results about the availability of Genetic Services.	Staff from Genetic Services can provide information and support for families with, or at risk of, a genetic disorder. Referrals to Genetic Services should come from the GP or LMC in the first instance. A copy of the screening report should be included. Initial discussions with the health practitioner will be conducted with the cover (on call) genetic associate. If the health practitioner wishes a staff member (genetic associate or clinical geneticist) to subsequently talk to the woman, this should be handled as a formal referral. Please use the Genetic Services Referral form available from the NSU website at www.nsu.govt.nz/files/ANNB/Referral_form_final120511.pdf Genetic Services are physically located in Auckland, Wellington, and Christchurch. Telephone or in-person consultations will be negotiated based on the woman's location and circumstances. Any queries about the screening laboratory analytical process and the result algorithm can be referred back to the designated specialists in LabPLUS and CHL.

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

Other Referrals

Guidelines	Information
Health practitioners must provide information about medical and non-medical services the woman may access to support her to make decisions about the management of her pregnancy.	Health practitioners should find out about services in their area and how to access them. Referrals may include: > obstetrician > specialist maternity services > maternal fetal medicine specialist > paediatrician > general medical practitioner > health social worker > counsellor > disability support services > parent support groups. Refer to Appendix 1 for resources and contacts.

11. Nuchal Translucency (NT) scan

The NSU supports continual improvement in the quality of this screening initiative. 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.

If the woman agrees to first trimester combined screening the ultrasound/radiology provider would then complete the NT and CRL measurements and send the results to the screening laboratory.

Referral for NT Scan

Guidelines	Information
Specialist medical maternity services, including NT scans, may only be provided to women on written referral from another practitioner (midwife or doctor).	This requires a written referral, stating the clinical reason, either on the screening request form or radiology request form.

Risk Calculation

Guidelines	Information
The risk calculation will be performed by the screening laboratories.	The two screening laboratories are LabPLUS at Auckland District Health Board and Canterbury Health Laboratories at Canterbury District Health Board. The screening laboratories use a single database for risk calculation. This ensures consistent risk calculation for all women across New Zealand.

Discussions With Women

Guidelines	Information
Ultrasound/radiology providers may discuss the scan findings with the woman. However, ultrasound/radiology providers should not provide a risk assessment for Down syndrome and other conditions to the woman based on NT alone (for example, 1:300 risk).	It is expected that practitioners may discuss the findings of the NT scan with the woman, but will not calculate a risk result. In all but exceptional circumstances, the risk result will be communicated to the woman by the referring practitioner. There may be very limited circumstances where the radiology/ultrasound provider contacts the screening laboratory informing them of the NT and CRL measurements and requesting a risk assessment (if serum has already been taken). This would generally be on the rare occasion where a significant anomaly is found on the ultrasound scan.

Reporting Requirements

Guidelines	Information
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 risk calculation > significant abnormalities which may change the management of the pregnancy > name of the practice > name of the radiologist > name of the practitioner performing the scan.	An NT scan is usually performed between 11 weeks 2 days and 13 weeks 6 days. For acceptance for first trimester combined screening, at the time the NT scan is performed, the fetus must have a CRL between 45–84mm. The CRL is used by the laboratory to determine gestational age from the Robinson equation (see Appendix 3). Precise measurement of CRL and NT is essential in the interpretation and final risk assessment provided to the pregnant woman. The combined screening test is highly sensitive to errors in CRL measurement and NT measurement. The name of the scanning practitioner in addition to the reporting radiologist or obstetrician on the report will provide audit at an individual level. Best practice report templates can be found at www.nsu.govt.nz

Nasal Bone Assessment

Guidelines	Information
Nasal bone will be included in the risk calculation if it is reported to the screening laboratory at the same time as the NT measurement.	Nasal bone assessment is not currently required to provide a risk result for first trimester combined screening. When an NT measurement is provided to the screening laboratory and a risk has been issued, no further risk calculation will be done if the nasal bone is assessed at a subsequent scan.
Nasal bone is to be reported as: > not looked for > present > absent > not able to be visualised for technical reasons	If a result states that a nasal bone is hypoplastic, this will not be included in the risk calculation.

CRL Out of Range for First Trimester Combined Screening

Guidelines	Information
If the CRL is greater than 84mm, the ultrasound/radiology provider is to inform the woman that the fetus is outside the range for first trimester screening and an NT scan cannot be completed. The ultrasound/radiology provider will refer the woman back to her health practitioner for other options including second trimester maternal serum screening.	For first trimester combined screening, the CRL should be between 45 – 84mm. Where the CRL is greater than 84mm, the fetus is outside the range for first trimester combined screening.

Specialist Referral Following Abnormal NT

Guidelines	Information
If an NT scan shows an obvious anomaly, for instance structural/anatomical anomaly, the radiologist should: (a) inform the referring health practitioner in a timely manner; (b) provide information to the woman at the time of the scan.	The referring health practitioner should offer the woman referral to a specialist obstetrician.

NT Greater Than 3.5 mm

Guidelines	Information
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 the woman referral to a specialist obstetrician, with the expectation she 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.

NT for Twins

Guidelines	Information
For twin pregnancies, an NT and CRL for each fetus must be measured at the same time to ensure accurate risk assessment.	The screening laboratory uses CRL to date the pregnancy and hence calculate the marker MoMs. 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 risk calculation for each fetus should be included in the report to the screening laboratory. If an NT measurement is only able to be completed on one twin, a subsequent NT scan must include NT and CRL for both fetuses (if more than 2 days has elapsed).

Transmitting Scan Results to the Screening Laboratories

Guidelines	Information
Ultrasound/radiology providers will transmit copies of the report results in a timely manner direct to LabPLUS for Taupo north or Canterbury Health Laboratories (CHL) for south of Taupo.	Ultrasound/radiology providers are to have a system in place to send the ultrasound report to the screening laboratory and to confirm the report has been sent.

Loss of One Twin

Guidelines	Information
For pregnancies where one twin has died, a NT and CRL measurement is to be sent to the screening laboratory and will be used to calculate the risk assessment 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 to calculate a risk assessment. The screening laboratory will provide a risk assessment based on NT without biochemistry.

NT for Multiple Pregnancies – Triplets or Greater Multiples

Guidelines	Information
For pregnancies with three or more fetuses, an NT alone can be used for risk assessment.	The screening laboratory software is not able to provide a risk assessment for pregnancies where there are triplets or greater multiples.

The NSU supports International Accreditation New Zealand (IANZ) radiology accreditation. This occurs for the majority of practices around New Zealand and provides assurance that the practice operates to established standards.

Individual Certification and Standards Requirements

Guidelines	Information
Practitioners must follow the requirements of Section 88 of the New Zealand Public Health and Disability Act 2000. Practitioners should follow the standards of practice for diagnostic and interventional radiology (version 9.1) of the Royal Australian and New Zealand College of Radiologists. Practitioners should follow the statements from the Royal Australian and New Zealand College of Obstetricians and Gynaecologists. Practitioners should follow the requirements of ASUM.	The NSU recommends radiology practices have IANZ accreditation. The NSU is developing initiatives to audit and monitor individual operators. In 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. Further policies and statements on fetal ultrasound can be found at: www.ranzcog.edu.au www.ranzcr.edu.au www.asum.com.au www.fetalmedicine.com While ultrasound/radiology providers are no longer required to provide the risk calculation for women, the requirements of ultrasound practitioners providing services remain unchanged. This means ongoing participation in certification and audit programmes continues to be a requirement.

Nasal Bone Certification

Guidelines	Information
Practitioners assessing nasal bone must be certified for the assessment of nasal bone.	Nasal bone evaluation requires additional certification over and above that for NT assessment. The certification is provided by the FMF programme.

12. Screening laboratory processes

Reporting Information to the Health Practitioner

Guidelines	Information

The screening laboratory must provide a first trimester combined screening or second trimester maternal serum screening result to the health practitioner who referred the woman for screening, by electronic means and/or hard copy reporting.	The screening laboratory report must include:
	› screening result ('increased risk' or 'low risk')
	› multiple of the median (MoM) of each analyte
	› NT measurement and MoM
	› individual risk assessments for:
	  › trisomy 21 (Down syndrome)
	  › trisomy 18 (Edwards syndrome)
	  › trisomy 13 (Patau syndrome)
	  › neural tube defects (after 15 weeks).
	The screening laboratory report may include other information, for example that specialist obstetric referral is recommended, that a rare genetic disorder is indicated or that the marker levels indicate other problems.
	The screening cut-off is 1:300 for aneuploidies.

Provision of Specialist Laboratory Advice

Guidelines	Information
The screening laboratory must provide specialist laboratory advice to the health practitioner, when requested. The screening laboratory must ensure that health practitioners have screening information required to inform women of their screening results.	The screening laboratory will phone all increased risk results and abnormal analyte results and discuss these with the health practitioner. Health practitioners are welcome to contact the screening laboratory if required.

Incomplete Screening

Guidelines	Information
The screening laboratory will provide a report to the health practitioner when a blood sample for first trimester combined screening has been received, but no NT or CRL measurement.	The screening laboratory will inform the health practitioner if the NT scan has not been received. The screening laboratory will advise the health practitioner to contact the woman about the need to have her scan performed before 13 weeks 6 days for a first trimester combined screening risk assessment to be possible. If the scan has not been performed by 13 weeks 6 days, the screening laboratory will: › issue a report to the health practitioner that provides the MoMs of the serum analytes only › advise that first trimester combined screening cannot be completed because the scan data was not available › recommend that the woman is offered second trimester maternal serum screening. The health practitioner should advise the woman that first trimester combined screening has not been completed and provide her with information about second trimester maternal serum screening.
The screening laboratory will advise the health practitioner if first trimester combined screening cannot be completed because the CRL is above 84mm.	The screening laboratory will accept a CRL of 45–84 mm. If the result is above 84mm, the screening laboratory will advise the health practitioner that 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 blood has been taken in the first trimester, the health practitioner will be informed that first trimester combined screening cannot be completed and provide her with information about second trimester maternity screening. The screening laboratory will indicate on the reports the dating used to calculate the risk. Preference will be given to CRL or BPD over LMP.

Further Serum Received

Guidelines	Information
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 risk result has already been provided and a further result will not be issued unless there are clinical indications. The health practitioner must phone the laboratory to discuss as required. Decisions regarding recalculating risk assessments will be made on a case by case basis.

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	Taupo and North (Northland, Waitemata, Counties Manukau, Auckland, Waikato, Lakes and Bay of Plenty)
Canterbury Health Laboratories	www.chl.co.nz Phone: 0800 THE LAB (843 522 x 80484) Chemical Pathologist: Dr Richard MacKay richard.mackay@cdhb.health.nz	South of Taupo (Tairawhiti, Hawkes Bay, Whanganui, Taranaki, Mid Central, Wairarapa, Capital & Coast, Hutt Valley, Nelson Marlborough, West Coast, Canterbury, South Canterbury, Southern)

13. Data, information and monitoring

13.1 Data and information collection

Antenatal screening for Down syndrome and other conditions collects, creates and retains indefinitely the following data and information. Table 2 outlines what information is collected.

Table 2: Data and information collected for antenatal screening for Down syndrome and other conditions

Information About the Woman as Collected on the 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

This data and information is held indefinitely by the screening laboratories on behalf of the National Screening Unit.

13.2 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 is 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.

Data and information may also be used in research studies.

From time to time, there may be requests for 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, the National Screening Unit. The data access request form can be found at www.nsu.govt.nz

Data Requests

Guidelines	Information
All requests for screening data must be forwarded to, and authorised by, the National Screening Unit.	To maintain consistency, all screening data requests will be managed by the National Screening Unit. This includes data requests for research, or from other individuals, committees, groups or organisations.

13.3 Monitoring

Antenatal screening for Down syndrome and other conditions is overseen by the National Screening Unit of the Ministry of Health. 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 2 which includes the numbers of screened women, practitioner information, results and outcomes (for instance comparing screening results with diagnostic results or pregnancy outcomes).

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

Appendix 1: 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.

The National Screening Unit (NSU), of the Ministry of Health is responsible for oversight of antenatal screening for Down syndrome and other conditions. The NSU produces consumer and practitioner resources and audits and monitors this screening initiative.

The **consumer resource** *Antenatal screening for Down syndrome and other conditions: optional screening – your choice, your decision* can be downloaded in pdf form at www.nsu.govt.nz 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 at www.healthed.govt.nz/contact-us

These **practitioner guidelines** can be downloaded from www.nsu.govt.nz Hard copies are available from Wickliffe on (04) 496 2277, Ministry of Health Publications, c/- Wickliffe Press, PO Box 932, Dunedin, or email moh@wickliffe.co.nz Please quote Code: HP5409.

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

Consumer Questions and Answers and other support information can be found at www.nsu.govt.nz

For questions or comments: screening@moh.govt.nz

Mailing address:
National Screening Unit
Private Bag 92522
Wellesley Street
Auckland

There are two Screening Laboratories. LabPLUS at Auckland District Health Board and Canterbury Health Laboratories at Canterbury District Health Board.

Laboratory request forms can be ordered from the Screening Laboratories.

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	Taupo and North (Northland, Waitemata, Counties Manukau, Auckland, Waikato, Lakes and Bay of Plenty)
Canterbury Health Laboratories	www.chl.co.nz Phone: 0800 THE LAB (843 522 x 80484) Chemical Pathologist: Dr Richard MacKay richard.mackay@cdhb.health.nz	South of Taupo (Tairawhiti, Hawkes Bay, Whanganui, Taranaki, Mid Central, 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
Northern and Midland Region
Phone: 0800 476 123

Central and Southern Region
Phone: 0508 364 436
www.geneticshealthservice.org.nz

Health and Disability Commissioner
www.hdc.org.nz/

Office of the Privacy Commissioner
www.privacy.org.nz

Ministry of Health
Primary Maternity Services Notice 2007
www.health.govt.nz

Other resources

New Zealand

Auckland District Health Board
Management of Babies with Down syndrome
www.adhb.govt.nz/newborn/Guidelines/Anomalies/DownSyndrome.htm

ASUM
www.asum.com.au

College of Midwives
www.midwife.org.nz

CCS Disability Action
www.ccs.org.nz
Phone: 0800 227 200

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 Down Syndrome Association
www.nzdsa.org.nz
Tel. 0800 693 724
Email: national.coordinator@nzdsa.org.nz

New Zealand Federation of Disability Information Centres
www.nzfdic.org.nz

New Zealand Organisation for Rare Disorders
www.nzord.org.nz

Pacific Information Advocacy Support Services
www.vakatautau.co.nz

Parent and Family Resource Centre
www.parentandfamily.org.nz

Parent to Parent
www.parent2parent.org.nz

People First
www.peoplefirst.org.nz/Home/tabid/36/Default.aspx

Prenatal screening tests for trisomy 21 (Down syndrome), trisomy 18 (Edwards syndrome) and neural tube defects
www.ranzcog.edu.au/publications/statements/C-obs4.pdf

Royal Australian and New Zealand College of Obstetricians and Gynaecologists
www.ranzcog.edu.au

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

Spina Bifida Association of New Zealand

What Everyone Keeps Asking
www.weka.net.nz

Australia

Australian Centre for Genetics Education
Changes to Chromosomes – Number, Size and Structure Fact sheet
www.genetics.com.au/pdf/factsheets/fs06.pdf

Mosaicism – Complex Patterns of Inheritance Fact Sheet
www.genetics.edu.au/pdf/factsheets/fs13.pdf

Down syndrome Fact Sheet
www.genetics.com.au/pdf/factsheets/fs28.pdf

Trisomy 13 – Patau Syndrome
www.genetics.edu.au/factsheet/fs29

Trisomy 18 – Edwards Syndrome
www.genetics.edu.au/pdf/factsheets/fs30.pdf

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
www.downs-syndrome.org.uk

Down syndrome online
www.down-syndrome.org

Fetal Medicine Foundation, London
www.fetalmedicine.com/FMF

The 11–13+6 Weeks Scan
www.studiolift.com/fetal/site/FMF-English.pdf

International Mosaic Down Syndrome Association
Booklet for professionals
www.imdsa.org/Information/professional.htm

National Health Service (UK)
Antenatal Screening: Introduction
www.nhs.uk/conditions/Antenatal-screening/Pages/Introduction.aspx

Brief descriptions of other trisomies (13 Patau; 18 Edwards), monosomy (Turner) and also Klinefelters, XXX, YY and features which might identify them antenatally
www.perinatal.nhs.uk/car/anomaly/chromosome/chromosome.htm

National Screening Committee (UK)
Fetal Anomaly Screening Programme – Screening for Down's Syndrome: UK NSC Policy recommendations 2007–2010: Model of Best Practice
www.dh.gov.uk/en/Publicationsandstatistics/Publications/PublicationsPolicyAndGuidance/ DH_084732

Appendix 2: Screened conditions

The list of 'other conditions' cannot be exhaustive as it is unknown what the 'condition' for a particular woman or her baby may be. This is the same for blood tests taken in any health setting where unanticipated findings may be identified.

The following provides details of the aneuploidies and unusual analytes.

Trisomy 21

Trisomy 21, also known as Down syndrome is a genetic disorder caused by an extra copy of chromosome 21 inside each of the body's cells. The 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. In Down syndrome, instead of a pair there are three copies of chromosome 21. The extra genetic material from the extra chromosome results in the physical and intellectual attributes 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 during the past 20 to 30 years. 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[2].

Recent research shows that parents 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[3]. Balancing clinical information (eg cause, recurrence risk for future pregnancies, physical features, associated medical conditions, intellectual disability 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 families about Down syndrome.

The New Zealand Down Syndrome Association note that:

> '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[4].

2. Bittles AH, Glasson EJ. 2004. Clinical, social and ethical implications of changing life expectancy in Down syndrome. Dev.Med.Ch.Neurol. 46(4): 282-6.
3. 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.
4. www.nzdsa.org.nz (accessed 9 September 2009).

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'[5].

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
- thyroid disorders, most commonly hypothyroidism, in up to 40 percent
- gastrointestinal tract congenital malformations, such as duodenal atresia and Hirschsprung's disease
- cataracts and visual refractive errors
- childhood leukaemia in about 2 percent
- early onset Alzheimer's disease.

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

Trisomy 18

Trisomy 18, also known as Edwards syndrome is a chromosomal condition caused by the presence of all or part of an extra 18th chromosome.

The syndrome 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 about one in 5,000 to 7,000 live births[6]. 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 apnea and heart abnormalities. It is impossible to predict the exact prognosis of a child with Edwards syndrome during pregnancy or post birth. The median lifespan is 5–15 days. A small percentage of babies with the full Edwards syndrome who survive birth and early infancy may live to adulthood. Children with mosaic or partial forms of this trisomy may have a different morbidity and mortality statistics. In mosaic forms there are some cells in the body where the chromosome number and structure is different from other cells.

5. www.upsideofdown.org/about_t21 (accessed 13 May 2012).
6. www.rarediseases.org/rare-disease-information/rare-diseases/byID/217/viewFullReport (accessed 13 May 2012).

Trisomy 13

Trisomy 13, also known as Patau syndrome, is a rare chromosomal condition, in which there is an additional chromosome 13. 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 means that a 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 babies born alive, most are likely to die in the hours or days following birth. A few babies with triploidy have lived five months or longer, but this is rare and usually the babies who survive longer have mosaic triploidy rather than full triploidy. Babies with triploidy usually have multiple genetic problems and severe growth restriction.

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 baby's 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. Babies with anencephaly are likely to be stillborn or will die shortly after birth.

Adequate maternal intake of folate/folic acid (400 micrograms per day) commencing antenatally and continued through the first trimester of pregnancy significantly reduces the probability of NTD.

First trimester combined screening cannot estimate the risk for NTDs. Second trimester screening performed after 15 weeks includes alpha fetoprotein (AFP) which may indicate a risk of NTDs at this time. However, more severe forms may be detected at the nuchal translucency ultrasound.

For NTDs, including spina bifida, the 18 to 20 week anatomy scan is the best available screening tool. 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.

Conditions identified by ultrasound

Ultrasound scans undertaken as part of this screening 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 is associated with trisomy 21, Turner syndrome and other chromosomal defects as well as other fetal malformations and genetic conditions.

There are instances where babies have increased NT measurements but at diagnostic testing have normal chromosomes. These babies may have an increased risk 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.

Unusual analytes

First trimester combined screening looks at two markers, pregnancy-associated plasma protein A (PAPP-A) and Beta-Human Chorionic Gonadatrophin (ßhCG).

Second trimester maternal serum looks at four markers, Alpha-fetoprotein (AFP), Beta-Human Chorionic Gonadatrophin (ßhCG), Unconjugated Oestriol (uE_3) and Inhibin A.

The markers used in screening come from the fetus and the placenta. Very abnormal levels may indicate a possibility of poor placentation or fetal growth restriction.

For example, if PAPP-A is reduced and/or inhibin-A and/or ßhCG is elevated it has been shown that there is a possibility of conditions related to poor placentation such as pre-eclampsia , preterm birth or intrauterine growth restriction.

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

Table 3 indicates the analytes measured. It is noted that there may be combinations of these markers that indicate increased pregnancy concerns, for instance low PAPP-A and low ßhCG or high ßhCG and high AFP.

The correlation of low levels of one or two markers with adverse pregnancy outcomes may not be strong, however very low level results can provide information which may be combined with ongoing assessment to assist with pregnancy management.

Table 3: Analytes measured during first or second trimester serum testing

What is Measured	When is it Measured	Where Does it Come From	Other Information
Pregnancy associated plasma protein-A (PAPP-A)	1st trimester	Produced by the baby and placenta	Very low levels of PAPP-A in the mother's blood can indicate poor placentation.
Beta human chorionic gonadotrophin (ßhCG)	1st and 2nd trimester	Produced by the baby	High levels of ßhCG in the mother's blood can indicate problems with the pregnancy (for instance fetal growth restriction).
Unconjugated oestriol (uE_3)	2nd trimester	Produced by the placenta	Very low levels of uE_3 can indicate the biochemical disorders Smith Lemli Optiz syndrome and steroid sulphatase deficiency.
Alpha fetoprotein (AFP)	2nd trimester	Produced by the baby	Very high levels of AFP in the absence of structural anomalies can indicate problems with the pregnancy (for instance fetal growth restriction).
Inhibin A	2nd trimester	Produced by the placenta	Very high levels of Inhibin-A can indicate poor placentation.

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 standardize test results a median value for test results in normal pregnancies is determined for each week of gestation. The woman's individual 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. The multiple of the median is used as a standard reporting tool for antenatal screening.

Figure 6 shows that an analyte level of 5 may be increased or decreased depending on the stage of pregnancy. At 10 weeks, the median value is 2.5, hence a level of 5 is 2 MoMs (elevated) but at 14 weeks, the median value is 10 hence a level of 5 is 0.5 MoMs (decreased).

Figure 6: Serum marker levels by gestational age

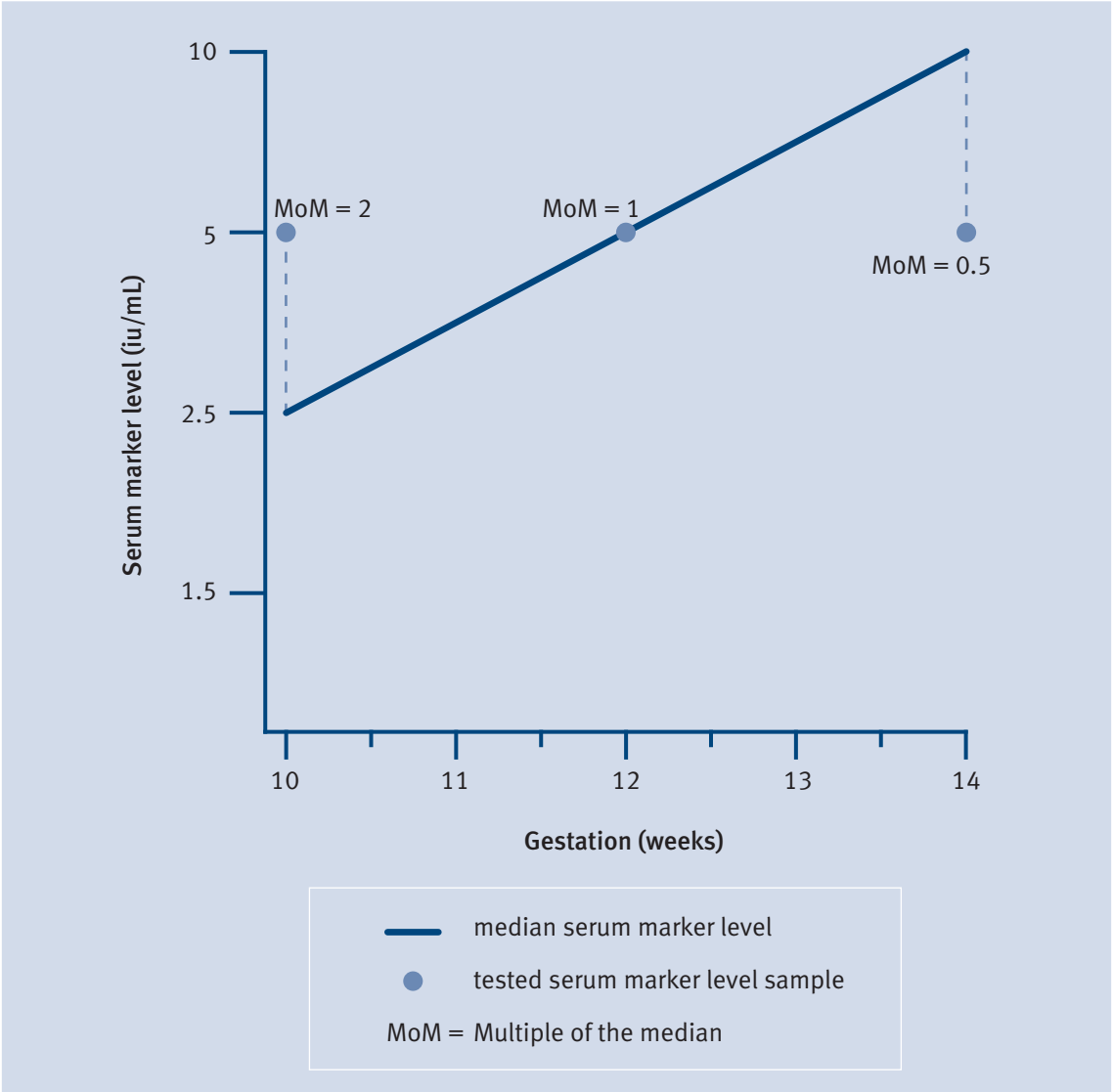


Table 4 show the analyte relationship with aneuploidy. These are not absolute. Most will have these patterns but not all.

Table 4: First and second trimester analytes and the relationship with aneuploidy

First Trimester				
	PAPP-A	Free ßHCG	NT measurement	
Trisomy 21	↓	↑	↑	
Trisomy 18	↓	↓	↑	
Trisomy 13	↓	↓	↑	
Second Trimester				
	Free ßHCG	uE_3	AFP	inhibin-A
Trisomy 21	↑	↓	↓	↑
Trisomy 18	↓	↓	↓	
Trisomy 13	↓	↓	↓	

Impact of accurate information

High quality screening results rely on complete and accurate information which informs the risk 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 risk calculation takes these factors into account when they are provided.

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

Table 5: Effect of different scenarios on pregnancy risk results

Scenario	Result 1	Change in Details	Result 2	Comment
Change in gestational age	Gestational age calculated at 15.1 **T21 risk result: 1:220**	Ultrasound scan calculated gestational age at 14.1	Revised risk calculation **T21 risk result: 1:910**	The analytes change over the pregnancy and therefore the risk calculation changes depending on the gestational age.
Change from singleton to twin pregnancy	Assumed singleton pregnancy **T21 risk result: 1:250**	Ultrasound scan shows twins	Revised risk calculation **T21 risk result: 1:500**	The analytes are divided when there is more than one fetus.
Non-smoker to smoker	Assumed non-smoker **T21 risk result: 1:210**	Health practitioner informs the Screening Laboratory that the woman is a smoker	Revised risk calculation **T21 risk result: 1:300**	Smoking affects placental function and inhibin levels are higher in women who smoke.
Compounding effect of many changes	42 year old woman using LMP dating at 18,2. No scan data, singleton pregnancy reported, no weight, no smoking information **T21 risk result: 1:520**	Woman's age incorrect – found she is 32 years of age, LMP was wrong and it is actually 14,1. Twins, Smoker and weighs 45 kg	Revised risk calculation **T21 risk result: 1:13,000**	There is a compounding effect when many of the variables are incorrect.

Appendix 3: Robinson Equation

The table below outlines the Robinson equation that is used by the screening laboratories to measure gestational age. Ranges for NT acceptance are highlighted in the darker cells.

Table 6: The Robinson equation for measuring gestational age

CRL	GA	
in mm	weeks	days
5	7	1
10	7	5
15	8	2
20	8	6
25	9	2
28	9	4
30	9	5
35	10	0
35	10	2
40	10	5
41	10	6
42	10	6
43	11	0
45	11	0
45.5	11	1
46	11	2
48	11	3
49.5	11	4
51	11	5
53	11	6

CRL	GA	
in mm	weeks	days
54.5	12	0
56.5	12	1
58.5	12	2
60	12	3
62	12	4
64	12	5
66	12	6
68	13	0
70	13	1
72	13	2
74	13	3
76	13	4
79	13	5
81	13	6
82.6	14	0
83	14	0
84	14	0
85	14	1
86	not available	

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 including NTDs after 15 weeks of pregnancy.

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.

Cut-off point – The point that divides people into a group at lower risk or increased risk for the condition being screened for. In New Zealand the cut-off point in screening for Down syndrome and other conditions is 1:300 at term.

False negative result – when a woman receives a low risk screening result but the baby does have the condition screened for.

False positive result – when a woman receives an increased risk screening result but the baby 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.

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.

Pre-eclampsia – a condition that occurs in some women during the second half of pregnancy, commonly associated with a rise in blood pressure and proteinuria.

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.

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

Screening – a way of identifying a group of 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 the conditions being present in the fetus.

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.

Triploidy – an extremely rare chromosomal disorder in which a baby 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.

Further terms can be found at www.nsu.govt.nz