PRENATAL SCREENING FOR CHROMOSOME CONDITIONS INCLUDING DOWN SYNDROME

PRACTICE RESOURCE JANUARY 2024

Down Syndrome Australia
Down Syndrome Queensland
Prenatal Screening
Queensland Government

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SECTION 1 INTRODUCTION

PURPOSE

The purpose of this practice resource is to provide evidence-based information to support health care professionals as they enable informed decision-making by parents about prenatal screening for aneuploidy. In particular, decisions about the future of their pregnancy.

AUDIENCE

The information in this resource is applicable to health care professionals providing both primary and tertiary prenatal care including, but not limited to, GPs, midwives and obstetricians.

OUTCOMES

The resource will support health care professionals to:

- Enable prospective parents to provide informed consent about participating or not participating in screening.
- Understand how the broader cultural context impacts informed decision-making and consent.
- Discuss the common prenatal screening tests for chromosome conditions including non-invasive prenatal screening.
- Describe the difference between screening and diagnostic tests.
- Accurately describe the most common chromosome conditions, including Down syndrome, which are the subject of screening.
- Use appropriate communication and language when discussing screening results and diagnosis.
- Support informed decision-making and access to appropriate care pathways at key decision points.

BACKGROUND

Prenatal screening technologies, such as non-invasive prenatal screening, are fast becoming a routine part of pregnancy care from as early as ten weeks gestation. The following guidance is provided to support clinical practice as health care professionals provide advice and support to prospective parents enabling informed decision-making both pre and post screening.

Research has shown that prenatal screening is not always presented as a choice to prospective parents (Lafarge et al, 2022; Down Syndrome Australia, 2021; Valentin et al, 2019). Prospective parents are often provided with minimal information about screening including the conditions screened for and the decisions they may be required to make following an increased chance screening result.

The information contained in this practice resource will assist the provision of support which respects diversity as well as a family's right to make informed choices, whatever those choices may be. Prospective parents have the right to access appropriate care pathways at key decision points including genetic counselling and the option to connect with condition specific support groups.

This practice resource is informed by and supports the implementation of relevant state and national guidelines including:

- The Pregnancy Care Clinical Practice Guidelines (Australian Government Department of Health, 2020)
- The Royal Australian and New Zealand College of Obstetricians and Gynaecologists and the Human Genetics Society of Australasia's statement on Prenatal screening and diagnostic testing for fetal chromosome and genetic conditions (2018)
- Genomics in General Practice 2nd edition (The Royal Australian College of General Practitioners, 2022)
- The Queensland Health Maternity and Neonatal Clinical Guideline: Standard Care (Queensland Government, 2018)

NOTES ON LANGUAGE

Terminology within this document has been chosen for accuracy and inclusivity.

Examples of language choices include:

Women and pregnant people
This term recognises that not all pregnant people identify as women.

Prospective parents
This term includes women and pregnant people and their partners. This recognises that decision-making about screening is often made by women and pregnant people in conjunction with their partners while prioritising the reproductive and bodily autonomy of the gestational parent.

Chromosome condition
This term includes chromosome differences such as Down syndrome (trisomy 21), Edwards syndrome (trisomy 18), Patau syndrome (trisomy 13) and sex chromosome differences.

Person with disability or person with Down syndrome
Person first language is used in this document while recognising that identity first (eg 'disabled person') language is also used in Australia. People with disability often prefer one term over another – follow their lead.

Down syndrome is the commonly used term for trisomy 21, which can also be referred to as T21, and is predominantly used throughout this document. When speaking with a person with Down syndrome or their family follow their language preferences.

The social model of disability is an internationally recognised way to view and address disability. In this document, the term 'disability' is used within the context of this model, recognising the physical, attitudinal, communication and social barriers that must be addressed to enable those living with impairments to participate in society on an equal basis with others.

Australia is a signatory to the United Nations Convention on the Rights of Persons with Disabilities (CRPD). As a party to the CRPD, Australia is required to promote, protect, and ensure the human rights of persons with disabilities. Australia is also a 'States party' to the CRPD convention. By virtue of being a 'States Party', whereby the Australian Government has ratified the document, Australia is legally bound to uphold the convention and also report to the monitoring Committee, as appropriate.

Process of selecting cited material
The cited material within this resource includes accepted state and national guidelines relevant to prenatal screening for chromosome conditions including Down syndrome. Other cited literature was gathered from electronic database searches focusing on current peer-reviewed evidence.

Material for inclusion was also nominated by members of the expert advisory group and members of the focus group who provided lived-experience perspectives of prenatal screening.

Diagnosis-specific advocacy and support groups were consulted to ensure cited material and content reflected the contemporary lived experience of people with chromosome differences.

Visit **www.prenatalscreening.org.au** to access the current version of this resource. Genetic technology is rapidly advancing and language evolving. Please contact **prenatal@downsyndromeqld.org.au** to suggest updates or additions.

NAVIGATING THIS PRACTICE RESOURCE

Prospective parents have the right to accurate, unbiased information and support as they make choices about screening, diagnostic tests and continuing or terminating a pregnancy.

The flow chart below is a simplified depiction representing in a general way the choices available to women and pregnant people in relation to prenatal screening at three key decision points:

1. Pre-screening
2. Following a screening result showing an increased chance of a chromosome condition
3. Following the diagnosis of a chromosome condition

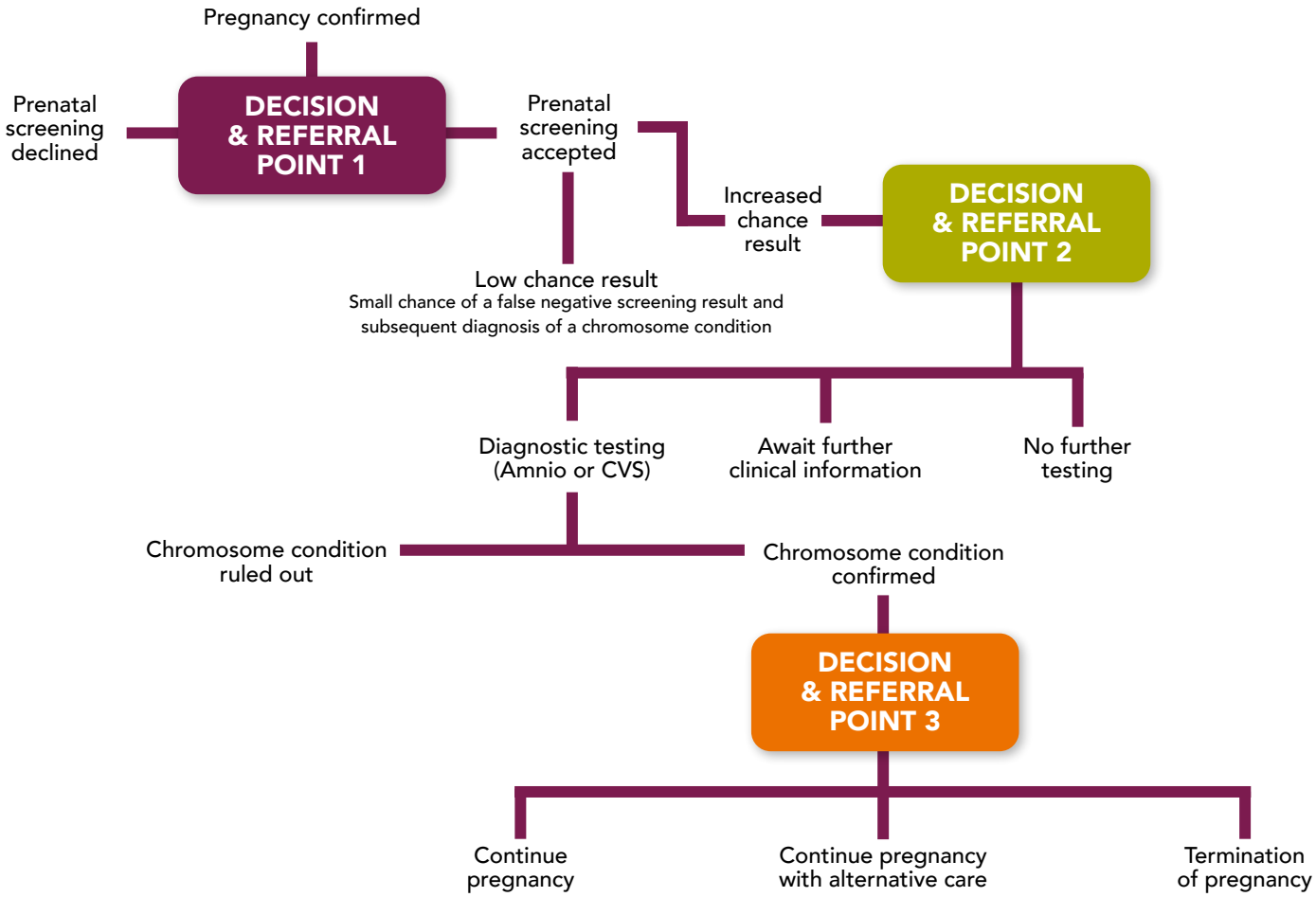
The practice resource builds the capacity of health care providers to support women and pregnant people at each of these points.

The three key decision and referral points are explored in more detail in Section 6 including key messages and referral options.

A comprehensive list of relevant resources for both health care providers and women and pregnant people is included in Section 7.

A low chance screening result does not mean there is no chance the baby will be born with a chromosome condition. A small number of families will receive a post-birth diagnosis of a screened-for chromosome condition following a low chance screening result. These families will benefit from attuned care as they make meaning of this unexpected news. The information and resources in this practice resource will be also largely relevant to supporting families following a post birth diagnosis.

KEY DECISION AND REFERRAL POINTS



SECTION 2 INFORMED DECISION-MAKING AND CONSENT

Informed decision-making is central to person-centred health care. It is a process which relies on two-way communication between a person and one or more health practitioners.

Informed decision-making reflects the principle that a person has the right to decide what is appropriate for them, taking into account their personal circumstances, beliefs and priorities. This includes the right to accept certain health care and to change that decision at any time. To exercise this right to decide, the person requires information that is relevant to them (Queensland Health, 2017).

Informed consent means a person has received the information relevant to them to allow them to make an informed decision and that they have given permission for a health care service to be provided (Queensland Health, 2017).

Pre-test counselling allows women and pregnant people to make an informed decision about whether or not they choose to participate in prenatal screening. Not all women and pregnant people will want to screen.

The woman or pregnant person has the right to make choices and decisions, even if the partner/co-parent does not agree with them. Follow the woman or pregnant person's lead in including or not including their partner in discussions and decision-making.

Qualified interpreter engagement is vital to enable informed decision-making by women and pregnant people from culturally and linguistically diverse backgrounds without, or with unknown, english language proficiency. Be aware of and sensitive to cultural contexts and beliefs about screening, disability and pregnancy outcomes which may impact decison-making.

RANZCOG's guidance on Prenatal Screening for Fetal Chromosome and Genetic Conditions (2018) states:

All women should be advised of the availability of investigations for prenatal screening and diagnosis as early as possible in pregnancy to allow time to discuss the options available and facilitate an informed choice. An informed choice is "based on relevant knowledge, consistent with the decision maker's values" (Marteau, 2001 in RANZCOG, 2018, p.10).

According to RANZCOG (2018) 'All such testing should be voluntary and only undertaken when the woman has been informed about the nature of the screening test, the possible results, and the options available to her'.

Choices about screening take place in the context of a society that displays bias and stigma against people with disability. A recent national survey about community attitudes towards people with disability found that 25% of the 2,000 respondents agreed with the statement that 'people with disability are a burden on their families' (Bollier et al, 2021). The narrative of people with disability 'suffering' or being a 'burden' on their families is at odds with the lived experience of most people with disability. When people with Down syndrome were asked about their lives as part of a US study, results showed 99% of people with Down syndrome were happy with their lives (Skotko et al, 2011).

The context and manner in which prenatal screening is presented to prospective parents, including the language used, has the potential to enable or compromise free and fully informed choice.

For example use the terms 'probability' or 'chance' rather than the more value-laden term, 'risk'.

When discussing prenatal screening with prospective parents, be aware of the following areas where there is potential for choice to be undermined (Shakespeare, 2014):

INFORMATION	Where no information is provided about a condition being screened for such as Down syndrome, it can carry the implication that 'it is obvious that someone would want to avoid these conditions and the primary focus becomes whether the test is effective in providing the diagnosis.' (Shakespeare, 2014, p. 131) Where prospective parents have access to full information about the lives of people with a particular condition and their families, they are enabled to make an informed decision about whether they believe the results from prenatal screening may be helpful, neutral or harmful for their family.
ATTITUDE	Displaying prejudice about disability and difference and a lack of support for people who decline screening, further testing or termination can compromise free choice. Avoid any implication that people have a duty to have screening and/or diagnostic tests and/or terminations.
ROUTINISATION	Sometimes referred to as 'screening creep', the autonomy of prospective parents is compromised where a decision to participate in screening is presumed as part of 'routine' pregnancy care.
BROADER CULTURAL CONTEXT	Prospective parents may hold negative attitudes about disability and fear the life of their child will be marked by suffering. Negative cultural stereotypes and messages about the benefits of genetic research and prenatal screening could reinforce these fears.

These are the key points that need to be covered in pre-test counselling to facilitate informed decision-making and consent:

- ***it is the woman or pregnant person's decision whether any testing takes place***
- ***an immediate decision is not required, they can take the time they need***
- ***the chromosome conditions for which testing is available and the differences between these conditions***
- ***the different pathways for testing (ie combined first trimester test alone, cell-free DNA (cfDNA) testing as first-tier or second-tier test or in a contingent model) and the risks and benefits of each approach and costs involved***
- ***the testing pathway, the decisions that need to be made at each point and their consequences and the support available***
- ***the need for accurate assessment of gestational age so that tests are conducted at the appropriate time***
- ***the sensitivity, specificity and positive predictive value for the woman's age of the test and a full explanation of the reporting format of the test (eg high probability/low probability, 1 in 10, 1 in 300,1 in 1,000)***

(adapted from Pregnancy Care Guidelines, 2020, p. 284)

Where a woman or pregnant person makes an informed decision to decline prenatal screening, that decision should be respected and recorded. If there are circumstances where a health care provider is required to revisit this choice, any conversation should note the previous decision and clarify the reason, for example new clinical information, why further discussion is deemed appropriate.

Where a woman or pregnant person makes an informed decision to participate in prenatal screening, assumptions should not be made about decisions such as diagnostic testing in the case of an increased chance result or termination of pregnancy following a diagnosis.

The information and resources contained in the following sections will equip health care providers with the knowledge required to enable informed choice throughout the screening process.

Screening tests are used to identify the chance of a baby having a chromosome condition such as Down syndrome. **They do not provide a definitive diagnosis.** In rare cases a low chance screening result will be returned and the baby will subsequently be diagnosed with a screened-for condition.

Where an increased chance result is returned on a screening test, a diagnostic test should be offered.

Prenatal screening tests include:

- Combined first trimester screening: Nuchal translucency ultrasound at 12-13^{+6} weeks combined with a blood test from the mother
- Non-invasive prenatal testing (NIPT): testing of the mother's blood
- Second trimester screening: testing of the mother's blood (maternal serum testing) between 15-20 weeks of pregnancy
- Ultrasound at 18-20 weeks

The following table summarises and differentiates screening and diagnostic tests.

TABLE 1 – Prenatal screening and diagnostic tests

	SCREENING TESTS			DIAGNOSTIC TESTS
	Combined first trimester screening	Second trimester serum screening	Cell-free DNA (non-invasive prenatal test, NIPT)	Chorionic villus sampling (CVS) or amniocentesis
Type of test	Blood test and ultrasound	Blood test	Blood test	Sample of placental cells or amniotic fluid
Timing of test (weeks)	Blood test 9-13 w Ultrasound 11-13^{+6} w	14-20 w	From 10 w	CVS 11-13 w Amnio from 15w
Conditions detected	Trisomy 21, 18, 13; Structural anomalies	Trisomy 21 and 18	Trisomy 21, 18, 13; sex chromosome conditions (optional)	Many chromosome conditions
Detection rate for trisomy 21	90%	75-80%	99%	99.99%
False positive rate for trisomy 21	3-5%	7-8%	<1%	<1%
Test failure rate	<1%	>1%	1-5%	<1%
Risk to pregnancy	None	None	None	Small risk of miscarriage
Medicare rebate available	Yes	Yes	No	Yes

Adapted from Murdoch Childrens Research Institute Prenatal Screening Decision Aid.

Further information about supporting informed decision-making following increased chance screening results, including diagnostic testing is included in Section 6.

The age of the woman or pregnant person is the most significant factor contributing to the likelihood a person will have a child with Down syndrome, with the prevalence of Down syndrome increasing as the woman or pregnant person's age increases (Nicolaides et al, 2004). Most babies with Down syndrome however, are born to mothers less than the age of 35 due to women under the age of 35 having more babies than those over 35. Down syndrome makes up around half of the chromosome conditions found prenatally with the next most common being trisomy 18 and 13 (Hui et al, 2017). Together, the three trisomies - 21, 18 and 13 make up 66% of aneuploidies identified by prenatal diagnosis (Hui et al, 2017).

PRENATAL SCREENING TESTS

COMBINED FIRST TRIMESTER SCREENING

In Australia, it is common practice to screen for chromosome conditions in the first trimester using combined first trimester screening.

Combined first trimester screening comprises:

- ultrasound measurement of fetal nuchal translucency thickness between 11 weeks and 13 weeks, 6 days gestation (when the fetus has a crown-rump length of 45–84 mm) combined with
- maternal plasma testing of pregnancy-associated placental protein-A (PAPP-A) and free beta-human chorionic gonadotrophin (ß-hCG) between 9 weeks and 13 weeks, 6 days gestation (Australian Government Department of Health, 2020)

Combined first trimester screening has a detection rate for Down syndrome of 90% and a false positive rate of 3-5% (Murdoch Children's Research Institute, 2018). Refer to Table 1 on page 9 for comparative data on screening and diagnostic tests.

NON-INVASIVE PRENATAL SCREENING/ TESTING (NIPT) (ALSO KNOWN AS CELL-FREE DNA TESTING)

No national data is collected on the use of prenatal screening in Australia however data from Victorian providers suggests around 20% of pregnant people who choose prenatal screening are using NIPT (Lindquist et al, 2020). Available data suggests that women and pregnant people accessing private obstetric care are significantly more likely to choose NIPT than those in the public system (Gadsboll et al, 2020).

Sensitivity and specificity

NIPT uses maternal plasma and can be performed from 10 weeks gestation. It has the highest sensitivity (or true positive rate) and specificity (or true negative rate) rates, both 99%, of all screening tests for Down syndrome (RANZCOG, 2018), however NIPT is not diagnostic. A sensitivity rate of 99% means that out of 100 increased chance results, 99 fetuses will actually have Down syndrome and 1 will not (known as a false positive). A specificity rate of 99% means that out of 100 low chance results, 99 fetuses will not have Down syndrome and 1 will (known as a false negative).

Positive predictive value

While NIPT has high sensitively and specificity, there is wide variation in the positive predictive value (PPV) (ie the probability that, following a positive screening result, the fetus actually has the chromosome condition identified). The PPV varies based on factors such as age, weight and how prevalent Down syndrome is in the general population.

Health care providers need to be able to accurately communicate the significance of the PPV to prospective parents and the implications for decision-making. For example, if a woman or pregnant person's PPV is given as 60%, it means that in 40% of cases, the baby is found (either at birth; or with subsequent diagnostic testing) not to have Down syndrome. The PPV decreases as prevalence decreases which means a younger woman or pregnant person's increased chance screening result is more likely to be a false positive. Non-invasive prenatal screening tests are commonly marketed as being 99% accurate. **It is important that women and pregnant people understand that this does not mean an increased chance result provides 99% certainty that the baby has Down syndrome.**

Sensitivity, specificity and positive predictive value differs depending on the chromosome difference.

Positive predictive values can be calculated here: **https://www.perinatalquality.org/Vendors/NSGC/NIPT/**

SECOND TRIMESTER SCREENING

Between 15-20 weeks, women and pregnant people may be offered maternal serum screening (or NIPT which is available at any gestation from 10 weeks). Prior to screening, gestational age should be confirmed by ultrasound. The 18-20 week morphology ultrasound has relatively poor sensitivity and specificity for Down syndrome and is not recommended as a screening test. Findings from this scan however provide more clinical information that may be relevant in decision-making.

The following table from the RANZCOG *Statement on Prenatal Screening and diagnostic testing for fetal chromosome and genetic conditions* (2018) provides a summary of the performance characteristics of current screening tests for Down syndrome.

TABLE 2: Screening Tests for Trisomy 21 currently in use in Australia and New Zealand

Test	Gestation for screening	Sensitivity	Specificity	Positive Predictive Value#
Combined first trimester screening: MA + NT+ ßhCG+PAPP-A	11^{+0} - 13^{+6} weeks	85%	95%	~ 7-10%
Second trimester serum screening: MA+AFP+ßhCG+UE3 +/- Inhibin	15-20 weeks	70-75%	93%	~ 2-3%
cfDNA - based screening*	$>10^{+}$ weeks	99%	99%*	~ 45%

*In a proportion (1-6%) of cases, cfDNA testing is unable to provide a result. These people should have follow up assessment including detailed ultrasound (if not already performed), and be offered the options of diagnostic testing, repeat cfDNA testing (successful in approximately 50%), or an alternative form of screening such as combined first trimester screening.

MA = maternal age; NT = nuchal translucency; ßhCG = free ß human chorionic gonadotrophin; PAPP-A = pregnancy associated plasma protein A; AFP = Alpha-fetoprotein; UE3 = oestriol.
# these positive predictive values are derived from test performance in the general pregnant population, but will vary according to the underlying prevalence of the condition. (RANZCOG, 2018)

ULTRASOUND
Typically two ultrasounds are performed during pregnancy, they are:

First Trimester or Nuchal Translucency scan
This ultrasound is performed between 11-13^{+6} weeks. During this scan, the thickness of the fluid filled space at the back of the baby's neck is measured. When this measurement is enlarged, this may mean an increased chance for certain conditions including Down syndrome.

Where a larger measurement is identified, follow-up diagnostic testing, if chosen, can confirm or exclude the presence of Down syndrome or another chromosome condition. This ultrasound can also confirm the estimated due date, identify the number of babies, including twins, identify if a miscarriage has occurred and examine anatomical features.

18-22 week ultrasound
This scan can provide information about the growth, development and position of the baby and placenta and detect a number of structural conditions, including heart anomalies and spina bifida. Some findings at this stage may also indicate a greater chance of a chromosome condition being present. Follow-up diagnostic testing, after informed consent, would be required.

In addition, an **early dating scan** may be performed between 7 and 11 weeks gestation. The explicit consent of the woman or pregnant person should be sought in relation to viewing and hearing and ultrasound, particularly in the first trimester.

The Australasian Society for Ultrasound in Medicine have produced Guidelines for Parent-Centred Communication in Obstetrics. The guidelines provide a framework for ultrasound practices, professional bodies and educational institutions when delivering news of unexpected or adverse outcomes. Read the guidelines at:
https://www.asum.com.au/files/public/SoP/curver/Obs-Gynae/Parent-centred-communication-in-obstetric-ultrasound.pdf

Find tips for GPs on prenatal screening and talking to parents about Down syndrome at:
https://www.downsyndrome.org.au/wp-content/uploads/2020/02/DSA_TipsForGPs_Prenatal_Screening_web.pdf

DIAGNOSTIC TESTS

Where screening results indicate a higher chance of Down syndrome or another chromosome difference, pregnant people should be offered diagnostic testing. A specialist referral is recommended to provide prospective parents with the information required about what is most appropriate in the clinical circumstances.

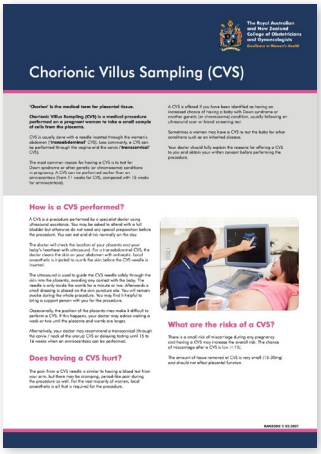
Women and pregnant people must be made aware that invasive diagnostic testing is the only way to confirm an increased chance screening result. Whether or not to progress to diagnostic testing is a choice and women and pregnant people may decline this testing. Some may choose to wait until birth to confirm the diagnosis.

Diagnostic testing with amniocentesis or chorionic villus sampling should be recommended prior to definitive management decisions such as termination of pregnancy (Standard 8, RANZCOG, 2018).

Where a woman or pregnant person chooses to have a diagnostic test, the choice of test should be based on gestational age (chorionic villus sampling before 14 weeks pregnancy and amniocentesis after 15 weeks), the preference of the woman or pregnant person (Pregnancy Care Guidelines, 2020) and the suitability of the test for individuals.

Diagnostic testing options should be presented with the most current statistics related to the miscarriage rates associated with invasive testing. Where a woman or pregnant person chooses a diagnostic option, health care providers should not presume that confirmation of a diagnosis would lead to a decision to terminate the pregnancy. Prospective parents may choose to pursue diagnostic testing to enable access to the right level of care and preparation during the remainder of the pregnancy or to have a definitive diagnosis on which to base a decision to terminate the pregnancy.

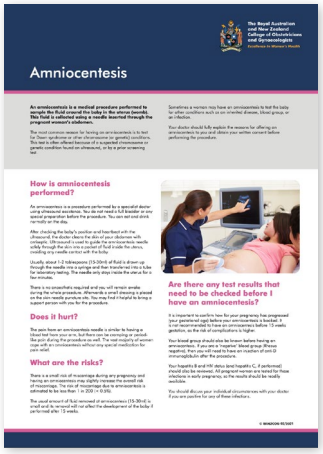
Find information leaflets to support patient understanding of diagnostic tests at **www.ranzcog.edu.au**


Chorionic Villus Sampling (CVS)

How is a CVS performed?

What are the risks of a CVS?

Does having a CVS hurt?


Amniocentesis

How is amniocentesis performed?

Are there any test results that need to be checked before I have an amniocentesis?

Does it hurt?

What are the risks?

Secondary consultation through the Down Syndrome Queensland support service is also available for any health care professional, community service, carer or family members supporting someone who has received unexpected news about their pregnancy.

Contact (07) 3356 6655 and ask for the Early Years Officer or use the online referral form at: **https://www.downsyndrome.org.au/qld/prenatal-form-2/**

SECTION 4 COMMON CHROMOSOME CONDITIONS

The information provided to prospective parents during the prenatal screening process should support families to make an informed choice about their pregnancy. **The language used has the potential to contribute to a more inclusive society or may reinforce stereotypes and stigma against people with disability.**

Many health professionals may not have extensive knowledge of screened-for conditions including Down syndrome. Understanding the contemporary experiences of people with screened-for conditions and their families will enhance the quality of the information and support provided to prospective parents considering their screening options and following increased chance screening results or diagnosis.

WHAT IS DOWN SYNDROME?

Down syndrome is a chromosome condition – it is not an illness or disease and is not considered a life-limiting condition. It occurs at conception as a result of an extra copy of chromosome 21. In Australia, around 1 in 1,100 babies are born with Down syndrome. Down syndrome occurs across all ethnic and social groups and to parents of all ages and has nothing to do with anything the mother or father did before or during pregnancy.

Down syndrome almost always occurs randomly. People with Down syndrome have some characteristic physical features, some health and development challenges, and some level of intellectual disability. Because no two people are alike, each of these things will vary from one person to another. When a baby is born, there is no way to tell what level of intellectual disability the child may have or in what way this may affect a person's life. Just like everyone else, environmental, cultural and social factors shape the lives of people with Down syndrome.

While some aspects of life may be more challenging than for a typically developing person, such as health care and education, people with Down syndrome now commonly take part in mainstream school and post-school education, sports, performing and visual arts, community volunteering and the workplace. A growing number of people with Down syndrome live more independently and are choosing to form relationships and get married. Life for people with Down syndrome these days is very different from how it used to be and looks even more hopeful for the future.

Many parents worry about how a child with Down syndrome will affect their family. Of course, every family is different, but personal stories and research show that most families which include a child with Down syndrome are stable, successful and happy and that siblings often have greater compassion and empathy.

Babies with Down syndrome can be born without any health problems. Babies with Down syndrome will usually have lower muscle tone and meet milestones at a slower pace than their typically developing peers. People with Down syndrome will experience some level of intellectual disability and are more likely to be born with or develop a range of health conditions, many of which can be well managed. Regular health screening is important. The following health screening tool has been developed to support the care of people with Down syndrome from birth to adulthood:
https://www.downsyndrome.org.au/services-and-supports/professionals/health/health-screening-tool/

More information about living with Down syndrome can be found here:
https://www.downsyndrome.org.au/about-down-

syndrome/what-is-down-syndrome/

WHAT ARE TRISOMIES 13 AND 18?

Trisomy 13, or Patau syndrome occurs in approximately 1 in 10,000 or 1 in 20,000 live births (Carey, 2021) and is caused by an extra copy of chromosome 13.

A summary of the health and developmental impacts commonly associated with trisomy 13 can be found here: **https://trisomy.org/about-trisomy/trisomy-13/**

Trisomy 18, or Edwards syndrome occurs in approximately 1 in every 6,000 live births (Albizua et al, 2020) and is caused by an extra copy of chromosome 18.

A summary of the health and development impacts commonly associated with trisomy 18 can be found here: **https://trisomy.org/about-trisomy/trisomy-18/**

Trisomies 13 and 18 are life-limiting conditions. Where trisomy 13 has been prenatally diagnosed, US research found only 18.9% of pregnancies resulted in a live birth and for trisomy 18, 13.5% of pregnancies resulted in a live birth (Tonks et al, 2013 in Meyer 2015). Studies have shown that the majority of pregnancies diagnosed with trisomy 13 or 18 are electively terminated (Irving et al, 2011; Tonks et al, in Meyer 2015).

Babies born with trisomies 13 and 18 will often require specialist medical care and most will not live beyond their first year of life. A US study, the largest population-based study of survival among children with trisomies 13 and 18 (Meyer et al, 2015) found survival of children born with these trisomies to be higher than what has previously been reported. This is consistent with recent studies which reported improved survival following more aggressive medical intervention. Specifically, for children with trisomy 13, 5-year survival was found to be 9.7% and for those with trisomy 18, 5-year survival was 12.3% (Meyer et al, 2015). Gestational age was found to be an important predictor of survival in both trisomies.

Greatly improved survival has been observed for children with mosaic trisomy 13 and 18. One study reported 1 year survival rates of 29% and 70% for children with mosaic trisomy 13 and 18 respectively (Wue et al, in Meyer et al, 2015). This is contrasted with 1 year survival rates of 11.5% and 13.4% respectively when including non-mosaic trisomy 13 and 18 phenotypes (Meyer et al, 2015)

Support Organisation for Trisomy (SOFT) US has information relating to trisomies 13 and 18 specifically aimed at medical professionals **https://trisomy.org/blog/news/soft-welcomes-medical-professionals/**

Selective termination of fetuses with the poorest prognosis could be contributing to a higher proportion of less severely affected fetuses with trisomy 13 and trisomy 18 and an overall increase in the median and overall survival rates of fetuses with trisomies 13 and 18 (Meyer et al, 2015). Overall, survival rates were found to be higher for females, full term infants and for those whose mothers resided in a metropolitan area (Meyer et al, 2015).

Support Organisation for Trisomy in Australia, known as SOFT, is comprised of a network of families, professionals and friends who are dedicated to the support and education of parents, family, friends and health professionals regarding the many complex issues and decisions surrounding the diagnosis and care of children who are diagnosed with a trisomy and other related chromosome conditions.

For further information and non-directional support contact SOFT Australia **https://www.soft.org.au/support**

WHAT ARE SEX CHROMOSOME CONDITIONS?

One pair of the typical complement of 46 chromosomes are known as the sex chromosomes. Chromosomes do not necessarily coincide with gender identity, like everyone, people with sex chromosome conditions can grow up to express a range of gender identities.

Typically, females have two X sex chromosomes (XX) and males have one X and one Y sex chromosome (XY). Sex chromosome conditions occur when there is a missing X chromosome, an extra copy of the X chromosome or an extra copy of the Y chromosome.

These conditions affect individuals in a variety of ways. People with sex chromosome conditions can and do lead happy and fulfilling lives, contributing to society in a myriad of ways. Many remain undiagnosed in the general population.

For the majority of people, these chromosome changes will be evident in all cells however for a small percentage, cells with chromosome changes will be combined with cells that have a typical chromosome count – this is known as mosaicism.

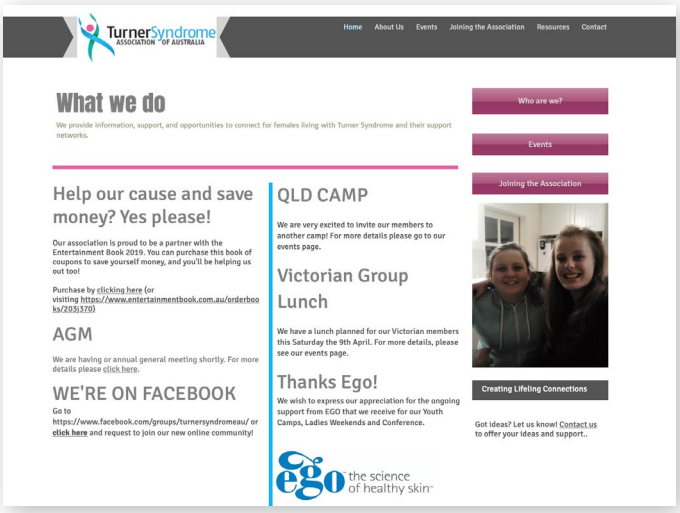
While prospective parents often approach non-invasive prenatal screening with a view to finding out the sex of their baby, they are often unaware that results may provide information about sex chromosome conditions.

Common sex chromosome conditions include:

Turner syndrome (also called monosomy X or 45, X) is associated with having one copy of the X chromosome in some or all cells of the body. Females typically have two copies of the X chromosome. People with Turner syndrome are usually female.

For more detail visit:
understandingturnersyndrome.org

The Turner Syndrome Association of Australia provides information, support, and opportunities to connect for people living with Turner syndrome and their support networks.
https://www.turnersyndrome.org.au/



Klinefelter syndrome (47,XXY) is associated with having an additional copy of the X chromosome in some or all cells of the body. Males typically have an X and a Y chromosome. People with Klinefelter syndrome are usually male.

Triple X (47,XXX) is associated with an additional copy of the X chromosome in some or all cells of the body. People with Triple X are usually female.

Jacob syndrome (47,XYY) is associated with an additional copy of the Y chromosome in some or all cells of the body. People with Jacob syndrome are usually male.

For further information visit Australian X and Y Spectrum Support (AXYS) at https://axys.org.au/ AXYS supports individuals and families living with Klinefelter Syndrome/XXY, XYY, XXX, XXYY, XXXY, XXXXY and associated variants.

Intersex Human Rights Australia (IHRA) is a national charity by and for people with innate variations of sex characteristics. IHRA promotes human rights, health and bodily autonomy. The goals of IHRA are to help create a society where people with innate variations of sex characteristics are not stigmatised, and where the rights of people with variations in sex characteristics are recognised.
https://ihra.org.au

Intersex Peer Support Australia (IPSA) is a peer support, information and advocacy group for people born with variations in sex characteristics and their families. IPSA tackles stigma and misconception through education, and advocating on issues affecting the wider intersex community. IPSA works towards improving affirmative healthcare, fostering intersexy pride, strengthening our community and deepening social culture.
https://isupport.org.au

Visit Intersex Human Rights Australia's Intersex for parents page at **https://ihra.org.au/parents/** for information, resources and personal stories.

LESS COMMON CHROMOSOME CONDITIONS

Beyond testing for the most common chromosome conditions, Down syndrome, Edwards syndrome and Patau syndrome, many NIPT providers also assess all 23 pairs of chromosomes. Other conditions may be identified as a result of this including:

- conditions caused by changes to the number of other chromosomes or
- conditions caused by missing or extra pieces of chromosomes.

A specialist review should be considered where results indicate a chance of a less common chromosome condition.

SECTION 5 COMMUNICATION AND LANGUAGE

Good clinical communication means providing information that is 'relevant, respectful, timely, non-judgemental and non-biased' (Queensland Clinical Guidelines, 2018, p.15)

Unfortunately, for many prospective parents, the way in which they received their increased chance screening results did not reflect good clinical communication and compromised their ability to make informed decisions (Down Syndrome Australia, 2021).

A national survey conducted by Down Syndrome Australia in 2021 found many examples of bias and stigma about Down syndrome and disability expressed by health professionals in the context of prenatal screening. Common themes which emerged from the survey included:

- Low expectations, such as being told the child would never walk, talk, go to school, would die young or not have a meaningful life.
- A focus on potential medical complications and a lack of information about treatment and support options.
- Negative and inaccurate comments about impacts on relationships with partners or siblings
- Inaccurate stereotypes.
- Inaccurate information about the potential costs of therapy and support.

The survey found:

- 42 per cent of families were told information about Down syndrome they now know to be untrue.
- 47 per cent of families felt that they did not get the information they needed to understand Down syndrome during their pregnancy.
- 69 per cent of families felt that the information provided did not give them an understanding of the lived experiences of people with Down syndrome and their families.

Research has shown that the way in which parents receive news about unexpected findings can have implications for their immediate and longer-term psychological wellbeing. Where communication is poor or there are delays in receiving information, a sense of mistrust can develop towards the health system and the relationship between the prospective parent and health care provider can be undermined (ASUM, 2020).

Carefully consider how you share news of an increased chance screening result with prospective parents. The exact wording used by health care professionals to convey screening or diagnostic results is often recalled by prospective parents many years later.

Communicate using clear, simple and consistent language and take time to confirm and to document that the information you have conveyed has been understood (Australian Government Department of Health, 2020). Ensure an interpreter service is available if required.

SHARING INCREASED CHANCE RESULTS

An increased chance screening result will often be experienced by expectant parents as unexpected or shocking (Hodgson & McClaren, 2018). Recognise that receiving this news often marks the beginning of a highly emotional and morally challenging decision making process (Hodgson & McClaren, 2018). Where possible, arrangements should be made to share the news face-to-face and the woman or pregnant person should have the opportunity for their partner or another support person to be present. Arrange for a quiet space for the conversation where the woman or pregnant person and their support person will have the opportunity to remain and absorb the information if they wish.

Ideally, phone contact with the woman or pregnant person to arrange the in-person conversation should be made by a health professional with whom they are familiar. Health professionals should prioritise immediate availability to the woman or pregnant person to provide further information and support, including psychosocial support as required.

MEETING DIVERSE NEEDS

Consider how best to meet the diverse needs of women and pregnant people including those who may identify as Aboriginal and/or Torres Strait Islander, culturally and linguistically diverse and/or living with disability. Be mindful of the impact of cultural contexts, variation in health literacy and beliefs around prenatal screening and disability.

Where women or pregnant people have lower health literacy, adapt communication accordingly and take the time to check their understanding of all the concepts and meaning. The use of a qualified interpreter is vital where language proficiency is not clear.

Visit **www.disabilitymaternitycare.com** for information, training and resources aimed at health care providers supporting women with disability to become parents. Some people with intellectual disability might need additional support to make their own decisions. This is called Supported Decision Making. Find out more at: **www.inclusionaustralia.org.au**

REFLECTING ON LIVED EXPERIENCE

Prospective parents reflect on their contemporary experiences of having received an increased chance screening result for Down syndrome (Down Syndrome Australia, 2021, p.14):

"It felt like the room went dark and it felt like they told us our baby died. That's how they acted as if they were delivering terrible news."

"I was told my child was not really human, she would have a body like a human but nothing inside. I was told she would never be able to show she loved us. But we would love her like a family pet."

"We were given all the negatives about Down syndrome and that it would negatively impact our lives and I was given huge pressure to terminate. No support for my decision to continue."

"I had quite a bit of fear regarding having a child with Down syndrome. Would have been great to talk with someone about how wonderful life can be with a child with Down syndrome."

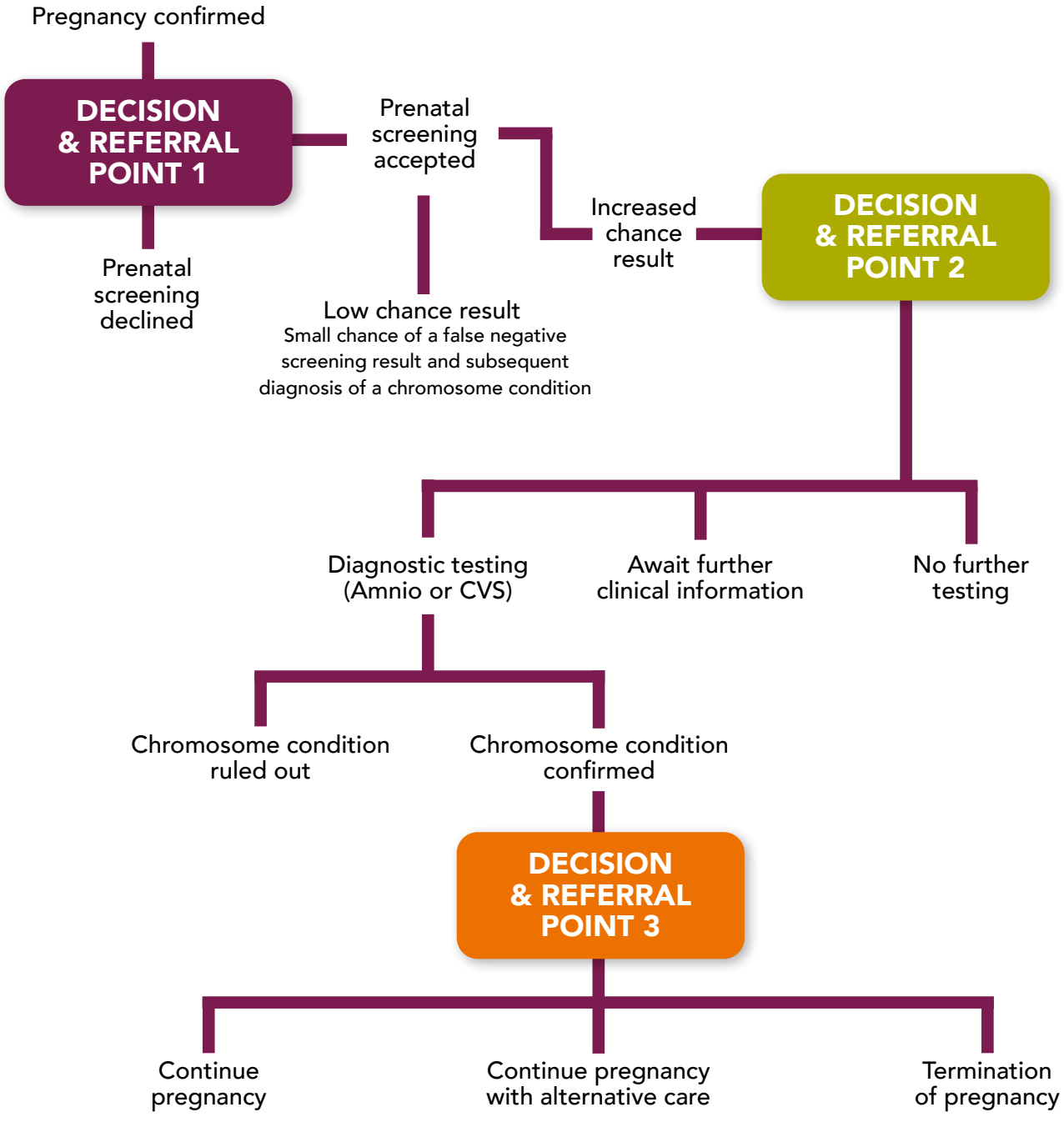
The following table (adapted from Australian Society for Ultrasound in Medicine, 2022) provides possible lead-in phrases and terminology to support conversations with prospective parents about an increased chance screening result or diagnosis. These are presented as a guide only and it is important to adapt your communication to each situation, always utilise active listening skills and take the lead from the woman or pregnant person.

Verbal information should be supplemented with clear written information as parents may not remember specific details due to the impact of shock. Information may need to be repeated and follow up appointments made.

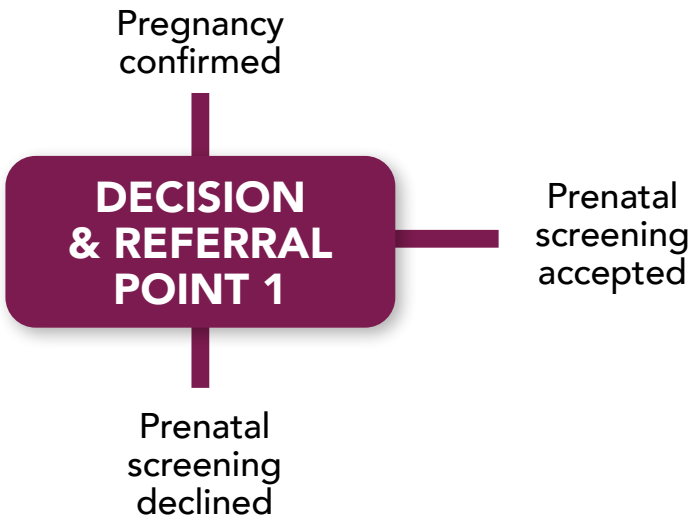
POSSIBLE LEAD-IN PHRASES AND TERMINOLOGY	CONSIDERATIONS
When discussing results use terminology such as 'anomaly', 'difference' and 'variation'.	Avoid stigmatising terms like 'abnormality', 'malformation', 'problem', 'mistake', 'wrong', 'defect' and 'adverse finding' in the case of variations.
We've just received your prenatal screening results and there have been some unexpected findings. I was hoping you could come into the clinic so we could talk about what the results mean. Would you like to bring in a support person?	Aim for value-free and honest communication. Avoid prefacing your comments with 'I'm sorry' or 'I have bad news' which immediately assigns a negative meaning to the condition identified. Empathise with parents and let them know you have time put aside to talk to them in more detail face-to-face. Ensure when you make the telephone call, you have considered your availability to meet.
I am here to make sure you have the information and the care you need. I will support you and give you the time you need to make decisions that are right for you and your family.	It is important that women and pregnant people are reassured that they will not be rushed into a decision and that whatever their choices, they will be supported.
The results of these tests alone indicate the probability or chance of some fetal chromosome conditions but nothing else.	It is important to manage expectations about what a pre-natal diagnosis can provide in terms of future health and abilities.
I can give you information from a clinical perspective about Down syndrome (or another condition) but my understanding of the lived experience of people with Down syndrome is limited.	Health care providers are not expected to be experts on the lived experience of people with Down syndrome and their families.
People often find it useful to connect with people who have lived experience of Down syndrome (or another condition). If this is something you are interested in I can refer you to Down Syndrome Queensland (or another diagnosis specific group).	Be open and transparent about the limits of clinical knowledge and provide opportunities for prospective parents to connect with condition specific groups.

SECTION 6 KEY DECISION AND REFERRAL POINTS

This section provides key messages, information and referral options for each of the three decision points shown in the flow chart below.



DECISION AND REFERRAL POINT 1



Key messages for health professionals to convey at this decision point:

- **Participating in prenatal screening is a choice**
- **Evidence based, contemporary information about the most common screened for chromosome conditions such as Down syndrome**
- **The testing pathway including decisions that need to be made at each point, consequences and support available**
- **The difference between screening and diagnostic tests**
- **Any out of pocket costs (particularly in relation to NIPT)**
- **The sensitivity, specificity and positive predictive value for the woman or pregnant person's age of the test and a full explanation of the reporting format of the test**

(adapted from Pregnancy Care Guidelines, 2020, p. 284)

THE IMPORTANCE OF TIME AND SPACE

At key decision points, women and pregnant people need to be offered time, space and the right support to help them make meaning out of the news they have received.

INFORMATION AND REFERRAL OPTIONS

PRENATAL SCREENING INFORMATION

Information about prenatal screening for prospective parents (page 29) lists resources to support informed decision-making.

The Queensland Health funded website **www.prenatalscreening.org.au** brings together information to support health care professionals and better enable prospective parents to make informed choices about screening, diagnostic testing and continuing or terminating a pregnancy.

GENETIC COUNSELLING

Genetic counsellors are specialists in supporting prospective parents to make informed decisions about prenatal screening in keeping with their beliefs and values.

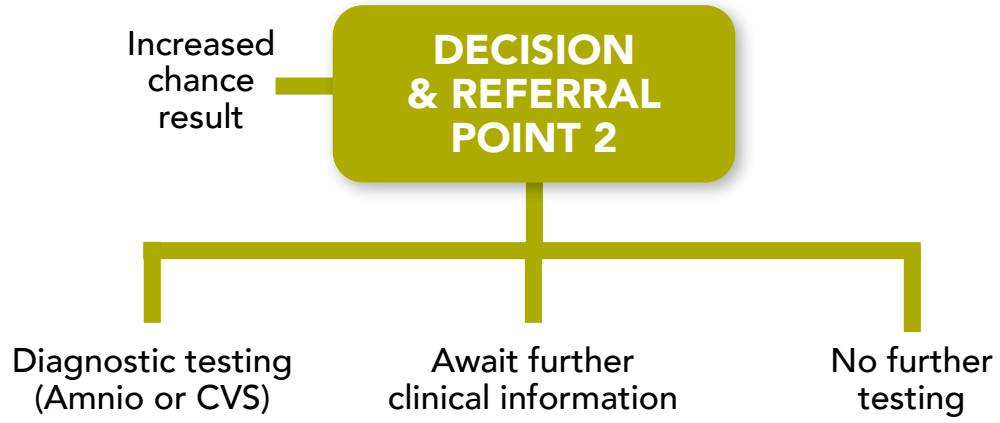
The capacity of public genetic counselling services does not usually extend to pre-test counselling.

Prospective parents can access a private provider through the Human Genetics Society of Australasia website: **https://www.hgsa.org.au/resources/queensland** (keep in mind that providers listed in other states can provide telehealth services).

In relation to NIPT, pre-test genetic counselling can also be accessed directly through some NIPT providers including the not-for-profit laboratory Victorian Clinical Genetics Services, contact:
https://www.vcgs.org.au/tests/perceptnipt
Phone: (03) 9936 6402
Email: perceptNIPT@vcgs.org.au

See page 32 for more information about genetic counselling available through NIPT providers.

DECISION AND REFERRAL POINT 2



A referral to a maternal fetal medicine service is recommended at this decision point where the following key messages should be conveyed to prospective parents:

- **the chromosome conditions that may be diagnosed**
- **the available tests, the gestational stage at which they should be undertaken, the process of the procedure and the risks involved**
- **the possibility that the procedure may not be successful or the result may not accurately reflect the fetal status**
- **the possibility of other fetal conditions that are not identified by the test**
- **the timeframe for receiving results and making further decisions if necessary**
- **options to consider if a chromosome condition is identified (eg continuation of the pregnancy or termination), the need for additional care if the pregnancy continues (eg specialist management of the pregnancy and the baby) and options for adoption or alternative care arrangements**
- **accurate and balanced information about health and developmental outcomes for children diagnosed with this condition including support available**
- **the impact on the woman or pregnant person and their family of a false negative or false positive result (eg anxiety following a false positive may remain) (Green et al 2004, Kristjansdottir and Gottfredsdottir, 2014)**
- **costs involved and how they are to be met**

WHERE DIAGNOSTIC TESTING IS DECLINED

Where an informed decision has been made for no further testing, this choice should be respected and recorded. Routine antenatal care should continue to be offered and a referral made to a maternal fetal medicine service for tertiary support including ultrasound is recommended. If there are clinical indications that it may be appropriate to revisit the decision to decline diagnostic testing this should be discussed sensitively with the woman or pregnant person.

INFORMATION AND REFERRAL OPTIONS

GENETIC COUNSELLING

Genetic counsellors are specialists in supporting women and pregnant people to make informed decisions about prenatal screening in keeping with their beliefs and values.

The capacity of public genetic counselling services does not usually extend to counselling following an increased chance prenatal screening result.

In relation to NIPT, genetic counselling may be accessed at no cost through many NIPT providers. See links on page 32 to information about the genetic counselling services offered by a range of providers.

Women and pregnant people can access a private provider through the Human Genetics Society of Australasia website: **https://www.hgsa.org.au/resources/queensland** (keep in mind that providers listed in other states can provide telehealth services).

THE DOWN SYNDROME QUEENSLAND SUPPORT SERVICE

This service is available to support people following increased chance screening results for Down syndrome. Secondary consultation is also available for any health care professional, community service, carer or family member supporting someone who has received unexpected news about their pregnancy.

Contact (07) 3356 6655 and ask for the Early Years Officer or complete an online form to request a call-back: **https://www.downsyndrome.org.au/qld/prenatal-form-2/**

RESOURCES ABOUT DOWN SYNDROME AND OTHER SCREENED-FOR CONDITIONS

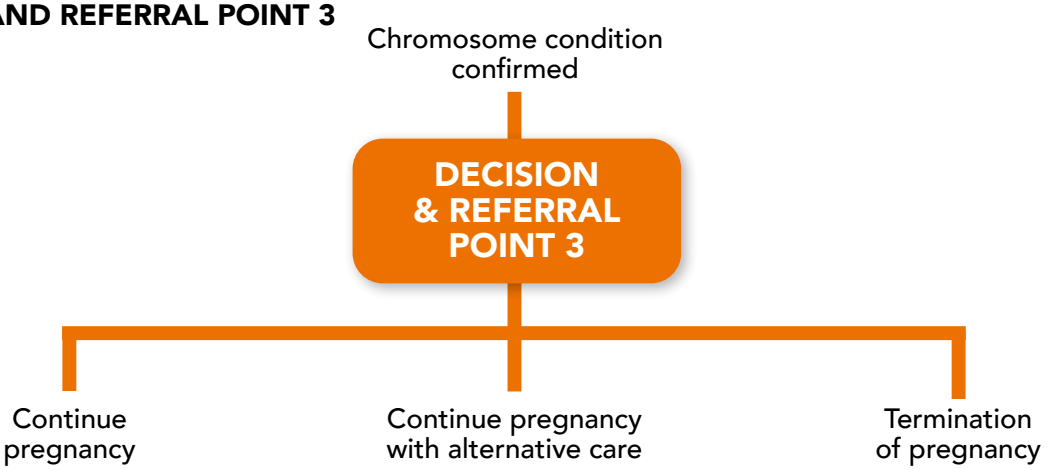
Resources about Down syndrome and other screened-for conditions can be found from page 29 of this practice resource. These resources can be used to support discussions with prospective parents to enable informed decision-making.

MENTAL HEALTH SUPPORT

Through the Unexpected – offers resources to inform and empower expectant parents through an unexpected diagnosis, decision making and the time that follows.

Centre of Perinatal Excellence e-COPE Directory – a directory of services and professionals who have a focus on emotional and mental health in pregnancy. **https://www.cope.org.au/getting-help/e-cope-directory/**

DECISION AND REFERRAL POINT 3



A maternal fetal medicine specialist may be providing care at this decision point, however, given limited specialist availability, the continued involvement of primary health care professionals is likely. Ensure you communicate using clear, simple and consistent language and take time to confirm and document that the information you have conveyed has been understood (Australian Government Department of Health, 2020). Ensure an interpreter service is available if required.

For further detail, refer to Section 5 'Communication and language' (page 19).

Key messages for tertiary health care professionals to convey at this decision point:

- **that the result has confirmed Down syndrome (or another chromosome condition)**
- **accurate and balanced information about health and developmental outcomes for children diagnosed with this condition including support available**
- **assurance they will be provided with the information, support and time they need to make a decision that is right for them and their family**
- **where prospective parents would like to connect with people with lived experience of Down syndrome, or another chromosome condition, this can be arranged**
- **options to consider include continuing the pregnancy (which may include alternative care or, in the case of life limiting conditions, perinatal palliative care) or termination of pregnancy.**

It is very important that there is no negative commentary on the condition diagnosed or personal opinions attached to the pregnancy options presented. Prospective parents require accurate information about the chromosome condition and information about relevant support organisations (Australian Government Department of Health, 2020). Prospective parents should be supported to access trusted resources and websites.

Where the diagnosed condition is life-limiting, prospective parents may consider a pathway of perinatal palliative care. Support and information should be provided.

Following diagnostic testing results, prospective parents require timely support and referral to appropriate health professionals which can include obstetricians, midwives experienced in genetic counselling, genetic counsellors and clinical geneticists (Australian Government Department of Health, 2020).

As part of their decision-making process, or following a decision to continue the pregnancy, prospective parents should be made aware of diagnosis specific support organisations such as Down Syndrome Queensland.

Where an informed decision has been made to continue the pregnancy, this choice should be respected and recorded. Routine antenatal care should continue to be offered together with continued engagement with specialist tertiary support. Where new clinical findings emerge, it may be appropriate to check in with the woman or pregnant person about their decision to continue the pregnancy. This should be discussed sensitively with the woman or pregnant person ensuring they do not perceive any pressure to change their decision.

Where a woman or pregnant person chooses termination of pregnancy, refer to Queensland Clinical Guidelines, Termination of pregnancy (Queensland Health, 2020).

INFORMATION AND REFERRAL OPTIONS

GENETIC COUNSELLING

Genetic counsellors are specialists in supporting women and pregnant people to make informed decisions about prenatal screening and diagnosis in keeping with their beliefs and values.

The capacity of public genetic counselling services may not extend to counselling following a prenatal diagnosis for one of the more common chromosome conditions including Down syndrome.

Women and pregnant people can access a private provider through the Human Genetics Society of Australasia website: **https://www.hgsa.org.au/resources/queensland** (keep in mind that providers listed in other states can provide telehealth services)

THE DOWN SYNDROME QUEENSLAND SUPPORT SERVICE

This service is available to support people following increased chance screening results and diagnosis. Secondary consultation is also available for any health care professional, community service, carer or family member supporting someone who has received unexpected news about their pregnancy. Contact (07) 3356 6655 and ask for the Early Years Officer or complete an online form to request a call-back: **https://www.downsyndrome.org.au/qld/prenatal-form-2/**

RESOURCES ABOUT DOWN SYNDROME AND OTHER SCREENED-FOR CONDITIONS

Resources about Down syndrome and other screened-for conditions (from page 31) can be used to support discussions with prospective parents to enable informed decision-making.

MENTAL HEALTH SUPPORT, NON-DIRECTIVE COUNSELLING AND PEER SUPPORT

Through the Unexpected – offers resources to inform and empower expectant parents through an unexpected diagnosis, decision making and the time that follows.

Centre of Perinatal Excellence e-COPE Directory – a directory of services and professionals who have a focus on emotional and mental health in pregnancy**. https://www.cope.org.au**

Non-directive pregnancy counselling (available through medicare) Find out more at:
https://www.cope.org.au/getting-help/self-help/pregnancy-support-counselling/

Pregnancy Counselling Link - counsellors provide non-directive counselling, helpful information and emotional support to those who are experiencing difficult or unforeseen circumstances. **https://www.pcl.org.au/**

Red Nose – offers peer-to-peer support for those who have experienced early pregnancy loss, medically advised termination, stillbirth or newborn death. 24/7 telephone support is available on 1300 308 307

Children by Choice offer pregnancy options counselling ph:1800 177 725 or **www.childrenbychoice.org.au**

ALTERNATIVE CARE

Contact the Queensland Government, Adoption and Permanent Care services for further information:
https://www.qld.gov.au/community/caring-child/adoption/considering-adoption-for-your-child

TERMINATION OF PREGNANCY

Resources about termination of pregnancy can be found on page 32.

Find an abortion service using the Children by Choice Abortion and Contraception Services Map
https://findaservice.childrenbychoice.org.au

Marie Stopes Australia offers access to termination of pregnancy, bookings can be made online and no referral is required in Queensland: **https://www.msiaustralia.org.au**

INFORMATION ABOUT PRENATAL SCREENING
The Queensland Health funded prenatal screening awareness website **www.prenatalscreening.org.au** brings together information to support health care professionals and better enable prospective parents to make informed choices about screening, diagnostic testing and continuing or terminating a pregnancy.

FOR PROSPECTIVE PARENTS:
• *Your choice – Prenatal screening tests in pregnancy*
A booklet designed by Murdoch Children's Research Institute to help prospective parents make decisions about prenatal screening. Useful for people who want to learn more about prenatal screening; are undecided whether or not to have screening or are unsure about which test to have. A web based app is available at:
https://yourchoice.mcri.edu.au/

• *Prenatal testing for Down syndrome fact sheet*
https://www.downsyndrome.org.au/wp-content/uploads/2020/02/DSA-prenatal-factsheet-C06.pdf

• *One screened every minute* – this podcast presents a series of interviews with people across Australia who have received increased chance screening results for a range of chromosome conditions, including Down syndrome. The interviews incorporate the experiences of prospective parents who have continued the pregnancy, as well as those who have chosen termination. **www.onescreenedeveryminute.com**

• *Through the Unexpected* works to protect the mental wellbeing of people who receive unexpected news regarding the health, development or genetics of their unborn baby. The website offers links to parent stories, support organisations and information resources for parents who have received a prenatal diagnosis and are decision making or moving forward through the unexpected. **www.throughtheunexpected.org.au**

FOR HEALTH CARE PROVIDERS:
• *Tips for GPs on prenatal screening and talking to parents about Down syndrome*
https://www.downsyndrome.org.au/wp-content/uploads/2020/02/DSA_TipsForGPs_Prenatal_Screening_web.pdf

• *Through the Unexpected* provides a list of supports for professionals with the aim of supporting the workforce that supports people through prenatal screening and diagnosis experiences. This list can be found here:
https://throughtheunexpected.org.au/find/support-for-professionals/

• *Genetics in Pregnancy* Professional development opportunity via this online genetics course designed for maternity health professionals by clinicians with expertise in prenatal screening and diagnosis.
https://study.unimelb.edu.au/find/short-courses/genetics-in-pregnancy/

FOR BOTH HEALTH CARE PROVIDERS AND PROSPECTIVE PARENTS
The Upside – ABC iview
Host Julia Hales explores people's thoughts when they face a decision about having a baby with Down Syndrome and re-examines her experience as she confronts parents' fears and celebrates the joys of living with Down Syndrome.
https://iview.abc.net.au/show/upside

INFORMATION ABOUT SCREENED-FOR CONDITIONS
Through the Unexpected has an online directory of Patient Support Organisations including condition and diagnosis-specific formal and informal groups and disabled persons and family organisations:
https://throughtheunexpected.org.au and **www.raisingchildren.net.au**

INFORMATION ABOUT DOWN SYNDROME

The *Down Syndrome Australia* website is constantly being updated and provides professionals and prospective parents with accurate and contemporary information about Down syndrome - **www.downsyndrome.org.au**
Find the answer to the question: What is Down syndrome?**https://www.downsyndrome.org.au/about-down-syndrome/what-is-down-syndrome/**

Additional information from outside Australia can be found here:

- *Down Syndrome Diagnosis Network* - **https://www.dsdiagnosisnetwork.org/**
- *Lettercase* - **lettercase.org**
- National Down Syndrome Congress *Information for new and expectant parents* - **www.ndsccenter.org/programsresources/new-and-expectant-parents**

DOWN SYNDROME QUEENSLAND

Down Syndrome Queensland (DSQ) can support prospective parents and health care providers in a number of ways.

People often contact DSQ to better understand their screening results. Prospective parents are offered information and support as they explore what raising a child with Down syndrome may mean for them and their family. DSQ can also assist prospective parents understand more about a diagnosis of Down syndrome and provide information on options available if they are undecided about continuing with the pregnancy.

DSQ believes families have the right to feel supported in their choices no matter which path they choose. DSQ provides balanced, evidence-based information so that people can feel empowered to move forward.

In conjunction with support offered from health care professionals including GPs, genetic counsellors or specialist teams, DSQ can offer free non-directive assistance to prospective parents to:

- Understand the difference between screening and diagnostic results and what these mean
- Provide current information about Down syndrome
- Discuss what this news may mean for their pregnancy, and/or for their family
- Explore options, including time frames to consider
- Support them through the decision-making process
- As the peak body for people living with Down syndrome and their support networks in Queensland, DSQ offers balanced, current and evidence-based information and follow up support.

service, with support offered flexibly (phone, email, Zoom, in-person where location allows). Online contact and referral forms are available at **https://www.downsyndrome.org.au/qld/services-and-supports/expectant-parents/** or contact 1300 881 935 and ask for the Early Years Officer.

There are a range of resources available online through Down Syndrome Australia aimed at expectant and new parents including:

- Prenatal testing for Down syndrome fact sheet **https://www.downsyndrome.org.au/wp-content/uploads/2020/02/DSA-prenatal-factsheet-C06.pdf**
- The new parent information pack **https://www.downsyndrome.org.au/new-parent-information/**

Following an increased chance screening result or diagnosis, many prospective parents describe the value they found connecting with people with Down syndrome and their families. DSQ can facilitate – if desired – the opportunity to connect with people with Down syndrome, and/or parents and families for perspectives on lived experience.

PEOPLE WITH DOWN SYNDROME TALKING ABOUT THEIR LIVES:

• *You can't ask that* – Down syndrome - **iview.abc.net.au/programs/you-cant-ask-that/LE1617H002S00#pageloaded**

• *Down Syndrome Answers* – The best people to answer questions about Down syndrome are people with Down syndrome. The Canadian Down Syndrome Society found the most-asked questions on Google and asked 10 Canadians with Down syndrome to give their answers **https://cdss.ca/awareness/down-syndrome-answers/**

• *I have one more chromosome than you. So what?* Karen Gaffney - **www.youtube.com/watch?v=HwxjoBQdn0s**

• *Dear Future Mom* - **https://www.youtube.com/watch?v=Ju-q4OnBtNU**

• BBC Three – *Things people with Down's syndrome are tired of hearing* - **www.youtube.com/watch?v=AAPmGW-GDHA**

Opportunities for connection

Support groups

• State based Down syndrome organisations – contact your local organisation on 1300 881 935.

Online Groups

• Many state based Down syndrome organisations have online groups contact your local organisation on 1300 881 935.

• *321 Pregnancy Care* is a Facebook page run by Down Syndrome Australia offering a safe place for women and pregnant people who have received a potential or confirmed diagnosis for their baby.

• *T21 Mum Australia Network* – A network of Facebook groups designed to create meaningful connections for mums of children with Down syndrome in the early years.

OTHER RESOURCES RELATING TO DOWN SYNDROME:

21 GIFTS is a kindness project aimed at rewriting the narrative surrounding a child diagnosis. 21 Gifts is a not-for-profit organisation providing educational resources and free suitcases which health professionals can provide to families at the point of diagnosis which are filled with hope, inspiration and lived experiences. **www.twentyonegifts.com**

Celebrate T21 –The Celebrate T21 books are a collaborative annual project with wonderful families and amazing photographers across various countries. **www.celebratet21.com**

The Super Power Baby Project – An internationally celebrated book featuring portraits of children with chromosome and genetic conditions and challenging the deficit language so often reached for when describing disability. **www.superpowerbabyproject.org**

GENETIC COUNSELLING

Most NIPT providers offer genetic counselling for prospective parents who have received an increased chance result from the provider's particular test.

Victorian Clinical Genetics Service
https://www.vcgs.org.au/tests/perceptnipt
Phone: (03) 9936 6402
Email: perceptNIPT@vcgs.org.au
VCGS provides a comprehensive NIPT service called perceptNIPT which is available across Australia. Their expert team, including genetic counsellors and clinical geneticists, can support health care providers and patients before, during and after screening. For people who receive a result suggesting an increased chance for a chromosome condition, telehealth or in person genetic counselling appointments are available. The team can also arrange referrals to other specialists and/or support services as needed. This clinical support is provided free of charge for anyone accessing their NIPT through VCGS.

Additional providers listed on the following pages, all have pathology collection points in Queensland, follow the links for information about access to their genetic counselling service.

While there is some variation, generally, a single counselling session within business hours is available to people accessing NIPT through these providers. Please note this is not an exhaustive list.

Sonic Healthcare
https://www.sonicgenetics.com.au/clinicians/featured-tests/nipt/nipt-genetic-counselling/
https://www.sonicgenetics.com.au/media/14515/shg-fm-0002001-reproductive-counselling-referral-form.pdf

Genomic Diagnostics
https://www.genomicdiagnostics.com.au/genetic-counselling/?_sft_resource_category=nsw

Invitae
https://www.invitae.com/en-au/here-for-you
Phone: 1800 961 370

Harmony/Australian Clinical Labs
https://antenatal.clinicallabs.com.au/doctor/images/Harmony%20Genetic%20Counselling%20A4%20-%20ACLMAR-BF-NAT-0209.6%20-.pdf

Nest
https://nestscreen.com.au/genetic-counselling/

INFORMATION ABOUT PREGNANCY OPTIONS AND TERMINATION OF PREGNANCY

FOR WOMEN AND PREGNANT PEOPLE:

Red nose – article containing information and support relating to termination for medical reasons.
https://rednosegriefandloss.org.au/support/article/medical-termination-of-pregnancy

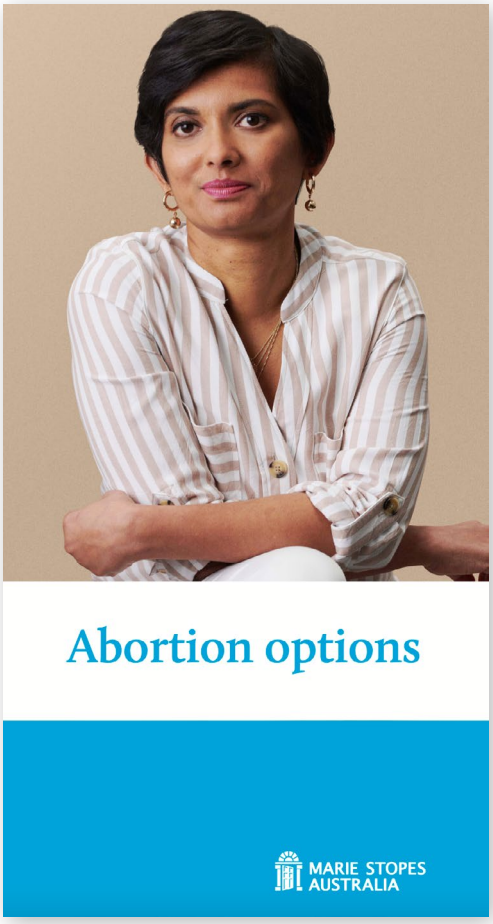
Abortion Options – a brochure developed by Marie Stopes which supports women and pregnant people to consider their options and make an informed choice
resources.msiaustralia.org.au/Abortion-Options-DL-Flyer.pdf

Abortion in Queensland – Answers to frequently asked questions by Children by Choice
https://resources.msiaustralia.org.au/Abortion-Options-DL-Flyer.pdf

FOR HEALTH CARE PROVIDERS:

The Royal Australian and New Zealand College of Obstetricians and Gynaecologists have produced a statement on Abortion (updated March 2019)
https://ranzcog.edu.au/wp-content/uploads/2022/05/Abortion.pdf

Queensland clinical guidelines, Termination of pregnancy
https://www.health.qld.gov.au/__data/assets/pdf_file/0029/735293/g-top.pdf



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