



# Prenatal screening for fetal genetic or structural conditions

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This statement has been developed and reviewed by the Women's Health Committee and approved by the RANZCOG Board and Council.

A list of Women's Health Committee Members can be found in [Appendix A](#).

Disclosure statements have been received from all members of this committee.

**Disclaimer** This information is intended to provide general advice to practitioners. This information should not be relied on as a substitute for proper assessment with respect to the particular circumstances of each case and the needs of any patient. This document reflects emerging clinical and scientific advances as of the date issued and is subject to change. The document has been prepared having regard to general circumstances.

First endorsed by RANZCOG: March 2010  
Current: July 2019  
Review due: July 2022

**Objectives:** To provide advice on prenatal screening for fetal genetic and structural conditions.

**Target audience:** All health practitioners providing pre-pregnancy and antenatal care.

**Values:** The evidence was reviewed by the Women's Health Committee (RANZCOG), and applied to local factors relating to Australia and New Zealand.

**Background:** This statement was first developed by Women's Health Committee March 2010 and most recently reviewed in July 2019.

**Funding:** The development and review of this statement was funded by RANZCOG.

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## 1. Plain language summary

Prenatal screening is offered in maternity care to provide the pregnant woman with more information about her unborn baby. All women, irrespective of their geographical location, resources or chosen model of antenatal care, should have access to prenatal screening and diagnostic testing for fetal structural or genetic conditions that may impact on the future health of their baby.

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

## 2. Summary of recommendations

Either pre-pregnancy, or early in the antenatal period, women (or couples) should be counselled about the availability of screening tests in pregnancy for fetal structural and genetic conditions.

Counselling should address:

- Detailed clinical assessment (and referral of high-probability couples as appropriate).
- Available screening tests including cell-free DNA testing, combined First Trimester Screening, second trimester serum screening and second trimester fetal morphology ultrasound (College Statements [Prenatal screening and diagnostic testing for fetal chromosomal and genetic conditions C-Obs 59](#) and [Prenatal assessment of fetal structural conditions C-Obs 60](#)).
- Carrier screening for common inheritable genetic conditions (see College Statement [Genetic Carrier Screening \(C-Obs 63\)](#))
- The use of diagnostic tests including amniocentesis or CVS, specialised fetal imaging (ultrasound and MRI) and other tests as indicated for high probability results.

## 3. Introduction

Most babies are born healthy. However, a small number of newborns have genetic, chromosomal or structural changes that have an impact on their wellbeing, varying from minor, correctable problems to lethal conditions. Prenatal knowledge of the presence of such conditions allows changes to the management of the pregnancy to permit delivery in an appropriate location, rapid access to neonatal treatment, appropriate antenatal care or, in some circumstances, the option of abortion. It may also help parents understand the condition before the baby's birth, to help them prepare.

It is accepted that not all conditions will be detected antenatally. Prenatal screening programs aim to identify women with pregnancies at high probability of fetal abnormalities. Where clinical assessment or screening tests identify an increased chance of fetal genetic or structural conditions, timely access to appropriate diagnostic testing should be provided.

## 4. Discussion and recommendations

### 4.1 Prenatal screening test for fetal genetic and structural conditions

In Australia and New Zealand, maternal serum screening and obstetric ultrasound are widely used to identify pregnancies with an increased chance of fetal genetic and structural conditions. Initial screening tests may lead to an offer of further testing (tertiary ultrasound, chorionic villus sampling or amniocentesis) for a definitive diagnosis. In the event of a diagnosis of a fetal genetic or structural condition, the woman and her partner may choose either to continue or not continue with the pregnancy.

Prenatal screening is best implemented in the context of a comprehensive program that coordinates pre-test counselling and information, biochemical and ultrasound measurements, post- test interpretation, counselling and support during decision-making and, where indicated, follow-up consultations and diagnostic testing.

#### Good Practice Point

Prenatal screening is best implemented in the context of a comprehensive program that coordinates pre-test counselling and information, biochemical and ultrasound measurements, post- test interpretation, counselling and support during decision-making and, where indicated, follow-up consultations and diagnostic testing.

### 4.2 Clinical assessment

A detailed medical history, family history and clinical examination should be obtained to allow specific needs of the couple to be addressed. Where possible, this should take place at a pre-pregnancy visit or alternatively at the earliest opportunity in pregnancy. Couples at high probability of a genetic or structural problem in their fetus should be referred to a specialist with the necessary skills for further counselling and management.

### 4.3 Counselling and information on screening

All pregnant women should be advised of the availability of prenatal screening at pre-pregnancy counselling or as early as possible in the pregnancy to allow time to discuss the options available and facilitate an informed choice.

The clinician providing the counselling should do so in the appropriate context including:

- Local resources
- The patient's clinical circumstances (e.g. gestational age).
- Financial costs

Information should be given in a way that is easily understood and culturally appropriate. Written information is particularly valuable for many patients.

Information provided should include:

- The difference between screening and diagnostic testing.
- The relative advantages and disadvantages of the available screening tests.
- Details of the nature, purpose, limitations and consequences of screening.
- That the decision whether to undertake screening or not is entirely that of the woman.
- Practical aspects of screening including the conditions that are being screened for, the type of tests, the timing of tests and the approximate costs involved.
- The possibility of diagnosing fetal genetic or structural conditions other than those for which the screening programs are designed.
- The nature of results (often expressed as a numerical probability estimate), the occurrence of false positive and false negative results, and the offer of a follow up diagnostic test if an

- 'increased' probability' result is obtained.
- That not continuing the pregnancy may be an option (subject to local law) if a fetal genetic or structural condition is diagnosed.
- An assurance that continuation of the pregnancy is a valid option should a fetal genetic or structural condition be diagnosed, and that couples will receive appropriate counselling and care in preparation for birth.

Recommendation 1	Grade
All pregnant women should be advised of the availability of prenatal screening at pre-pregnancy counselling or as early as possible in the pregnancy to allow time to discuss the options available and facilitate an informed choice.	Consensus-based Recommendation
Recommendation 2	Grade
<p>The clinician providing the counselling should do so in the appropriate context including:</p> <ul style="list-style-type: none"> <li>• Local resources</li> <li>• The patient's clinical circumstances (e.g. gestational age).</li> <li>• Financial costs</li> </ul>	Consensus-based Recommendation

#### 4.4 Referral for further evaluation and counselling

In some circumstances, access to further evaluation or counselling may be sought from one or more of the following: an obstetrician (general practitioner, specialist or subspecialist), a sub-specialist radiologist, a paediatrician, a paediatric surgeon, a geneticist or a genetics counsellor.

Mechanisms should be in place for timely referral when a fetal genetic or structural condition is suggested on initial screening for the following reasons:

- To take into consideration the expected time required for the primary referrer to receive the initial report, discuss the findings and refer for a second opinion where required.
- To allow time for additional genetic investigations to be performed where necessary; in some cases testing requires transfer of samples to laboratories interstate or overseas and requires specific genetic counselling concerning the findings.
- New and advanced imaging modalities (i.e. advanced ultrasound and MRI) used in prenatal diagnosis of fetal structural conditions will commonly require specific timing for complex booking arrangements and sub-specialty expertise.
- Timely evaluation to allow full counselling of the results allows women and partners a full range of options, considering the local legislation for termination of pregnancy.

#### 4.5 Education, Training and Continuing Professional Development

Health professionals caring for pregnant women should:

- Have had education and training with respect to the clinical assessment and testing that is available for prenatal screening.
- Participate in Continuing Professional Development (CPD) and through seminars, courses, journals and other printed material, maintain an awareness of the most up to date evidenced-based practice.

## 5. Useful links

Nuchal Translucency  
Ultrasound, Education and Monitoring Program  
[www.nuchaltrans.edu.au](http://www.nuchaltrans.edu.au)

The Fetal Medicine Foundation (FMF)  
Credentials ultrasound operators and provides ongoing quality assurance for operators working outside Australia.  
<http://www.fetalmedicine.com>

## 6. Links to other College statements

[Prenatal screening and diagnosis of chromosomal and genetic conditions in the fetus in pregnancy \(C-Obs 59\)](#)

[Prenatal assessment of fetal structural conditions \(C-Obs 60\)](#)

[Evidence-based Medicine, Obstetrics and Gynaecology \(C-Gen 15\)](#)

## Appendices

### Appendix A Women's Health Committee Membership

Name	Position on Committee
Professor Yee Leung	Chair and Board Member
Dr Gillian Gibson	Deputy Chair, Gynaecology
Dr Scott White	Deputy Chair, Obstetrics and Subspecialties Representative
Associate Professor Ian Pettigrew	Member and EAC Representative
Dr Kristy Milward	Member and Councillor
Dr Will Milford	Member and Councillor
Dr Frank O'Keeffe	Member and Councillor
Professor Sue Walker	Member
Dr Roy Watson	Member and Councillor
Dr Susan Fleming	Member and Councillor
Dr Sue Belgrave	Member and Councillor
Dr Marilyn Clarke	ATSI Representative
Associate Professor Kirsten Black	Member
Dr Thangeswaran Rudra	Member
Dr Nisha Khot	Member and SIMG Representative
Dr Judith Gardiner	Diplomate Representative
Dr Angela Brown	Midwifery Representative
Ms Ann Jorgensen	Community Representative
Dr Rebecca Mackenzie-Proctor	Trainee Representative
Prof Caroline De Costa	Co-opted member (ANZJOG member)

### Appendix B Overview of the development and review process for this statement

#### *i. Steps in developing and updating this statement*

This statement was originally developed in March 2010 and was most recently reviewed in July 2019. The Women's Health Committee carried out the following steps in reviewing this statement:

- Declarations of interest were sought from all members prior to reviewing this statement.
- Structured clinical questions were developed and agreed upon.
- An updated literature search to answer the clinical questions was undertaken.
- At the June 2016 teleconference, the existing consensus-based recommendations were reviewed and updated (where appropriate) based on the available body of evidence and clinical expertise. Recommendations were graded as set out below in Appendix B part iii)

#### *ii. Declaration of interest process and management*

Declaring interests is essential in order to prevent any potential conflict between the private interests of members, and their duties as part of the Women's Health Committee.

A declaration of interest form specific to guidelines and statements was developed by RANZCOG and approved by the RANZCOG Board in September 2012. The Women's Health Committee members were required to declare their relevant interests in writing on this form prior to participating in the review of this statement.

Members were required to update their information as soon as they become aware of any changes to their interests and there was also a standing agenda item at each meeting where declarations of interest were called for and recorded as part of the meeting minutes.

There were no significant real or perceived conflicts of interest that required management during the process of updating this statement.

### *iii. Grading of recommendations*

Each recommendation in this College statement is given an overall grade as per the table below, based on the National Health and Medical Research Council (NHMRC) Levels of Evidence and Grades of Recommendations for Developers of Guidelines. Where no robust evidence was available but there was sufficient consensus within the Women's Health Committee, consensus-based recommendations were developed or existing ones updated and are identifiable as such. Consensus-based recommendations were agreed to by the entire committee. Good Practice Notes are highlighted throughout and provide practical guidance to facilitate implementation. These were also developed through consensus of the entire committee.

Recommendation category		Description
Evidence-based	A	Body of evidence can be trusted to guide practice
	B	Body of evidence can be trusted to guide practice in most situations
	C	Body of evidence provides some support for recommendation(s) but care should be taken in its application
	D	The body of evidence is weak and the recommendation must be applied with caution
Consensus-based		Recommendation based on clinical opinion and expertise as insufficient evidence available
Good Practice Note		Practical advice and information based on clinical opinion and expertise



### Appendix C Full Disclaimer

This information is intended to provide general advice to practitioners and should not be relied on as a substitute for proper assessment with respect to the particular circumstances of each case and the needs of any patient.

This information has been prepared having regard to general circumstances. It is the responsibility of each practitioner to have regard to the particular circumstances of each case. Clinical management should be responsive to the needs of the individual patient and the particular circumstances of each case.

This information has been prepared having regard to the information available at the time of its preparation, and each practitioner should have regard to relevant information, research or material which may have been published or become available subsequently.

Whilst the College endeavours to ensure that information is accurate and current at the time of preparation, it takes no responsibility for matters arising from changed circumstances or information or material that may have become subsequently available.