Prenatal assessment of fetal structural conditions

This statement was originally developed by the HGSA/RANZCOG Joint Committee on Prenatal Diagnosis and Screening

The principal authors of this statement were Dr Andrew McLennan and Professor Sue Walker.

A list of members of the HGSA/RANZCOG Joint Committee on Prenatal Diagnosis and Screening can be found in Appendix A and Women's Health Committee in Appendix B.

The Committee acknowledges contributing authorship in Appendix C.

Disclosure statements have been received from all authors and committee members.

First endorsed by RANZCOG: March 2015
Current: March 2018
Review due: March 2021, or as required

Objectives: This statement is intended to provide advice on the assessment of fetal structural conditions.

Outcomes: Improved understanding regarding effective assessment of fetal structural conditions that are performed at different stages in pregnancy.

Target audience: This statement is intended for use by health professionals providing antenatal care including: Obstetricians, Clinical Geneticists, Radiologists, Obstetricians, Sonologists, Sonographers and GPs, Midwives, Nurses and Genetic Counsellors.

Other audiences: This statement provides useful information for patients and carers, researchers, health policy makers, health regulators and those responsible for quality and safety of healthcare. This statement may also be a valuable resource to State and Federal Government bodies developing guidelines and other documents on prenatal screening and diagnosis.

Values: The evidence was reviewed by the HGSA/RANZCOG Joint Committee on Prenatal Diagnosis and Screening, and applied to local factors relating to Australia and New Zealand.

Background: This statement was originally developed by the HGSA/RANZCOG Joint Committee on Prenatal Diagnosis and Screening in 2015. It adds to the broad statement *Prenatal screening for fetal genetic or structural conditions (C-Obs 35).* More recently the statement was reviewed by Women's Health Committee and approved by RANZCOG Council.

Recommendation 5	Grade
Each practice should develop a protocol on the procedure to be followed when a condition is detected. This protocol should include guidelines for the immediate care of the patient and how the referring doctor will be informed.	Consensus Based Recommendation Reference [10]

Recommendation 6	Grade
All women should receive timely information following detection of a major fetal	

4. Discussion and recommendations

4.1 Consent standards and guidance

4.1.1 All pregnant women should be advised of the availability of prenatal ultrasound assessment for fetal structural conditions as early as possible in pregnancy to allow time to discuss the options available and facilitate an informed choice.

4.1.2 Some women may make an informed decision not to proceed with this assessment. Counselling should follow a shared decision-making model, where health professionals discuss information

4.2 Guidelines for first trimester fetal ultrasound (including structural condition)

Ultrasound-based first-trimester aneuploidy screening has resulted in some major structural conditions being diagnosed earlier in pregnancy. It is important that credentialled operators undertaking first trimester ultrasound assessment have a checklist of structures that are usually visualised at this time, and are aware of major structural conditions that should be diagnosed or excluded in the first trimester.

Australian Guidelines for the Performance of First Trimester Ultrasound have been published by the Australian Society for Ultrasound in Medicine (ASUM)[4] and this guideline provides a list of gestational ages at which various fetal structures may be visualised. The ISUOG first trimester fetal ultrasound guidelines[18] provide detailed information about the structures to be identified in first trimester when scanning for fetal structural conditions. Briefly, it is important to identify the following structures: the fetal head (cranium, falx and choroid plexus), chest (lungs, four

possible, patients with complex scanning needs should be referred to a practice with specific expertise in obstetric ultrasound. In addition, where a condition has been suspected, it is recommended that women are referred to a high risk or tertiary scanning service for confirmation, given the RADIUS study confirmed a relative detection rate of 2.7 (95% CI 1.3-5.8) in tertiary, compared to non-tertiary units.[21]

RANZCOG recommends that all practitioners involved in provision of mid-trimester fetal morphology ultrasound screening must undergo appropriate specific training in this critical and specialised area of practice. Service providers must participate in ongoing professional development, clinical audit, and multidisciplinary review of outcomes specific to their performance of mid-trimester fetal morphology ultrasound screening.

Recommendation 4	Grade and references
All pregnant women should be offered the opportunity to undertake a mid-trimester fetal anomaly ultrasound assessment. Second trimester ultrasound should screen for the number of fetuses, the gestational age, cervical length, the location of the placenta and should screen for fetal conditions.	Consensus-based recommendation Reference [9]
Recommendation 5	**Grade and references**
RANZCOG recommends that all practitioners involved in provision of mid-trimester fetal morphology ultrasound screening must undergo appropriate training and ongoing professional development in this area of practice.	Consensus-based recommendation

4.4 Pathways in the event of a condition

While normal findings promote positive feelings about a pregnancy and improve bonding, the finding of isolated or multiple serious conditionson prenatal ultrasound examination is inevitably stressful.[22] Families require prompt and accurate information, including confirmation of the condition, the likely aetiology, relevant associations, further investigations required (which may involve further imaging or genetic testing), implications for the remainder of the pregnancy, labour and delivery, and the outlook for their unborn baby as a newborn and beyond. This often involves referral to a tertiary unit and a multidisciplinary team. Women regard the speed at which they obtain information regarding the findings as extremely important, even if it means seeing another caregiver with whom they are not familiar.[23] The principles of management include timely review, multidisciplinary input in diagnostic evaluation, detailed counselling of the woman and her family, plans for ongoing care in the pregnancy and timely communication back to the referring practitioner.

Individual units will have their own specific protocols but the following is a suggested approach to the management of families following diagnosis of a structural condition in the fetus;

1. Clinical review

Once a fetal structural condition has been identified, a thorough pregnancy, family and medical history should be performed.

2. Further imaging

Confirmation of the condition in a tertiary setting is recommended. Other imaging modalities, such as 3D ultrasound, magnetic resonance imaging (MRI) and parental ultrasound may be appropriate.

6.

pulsed Doppler should not be used routinely during the first trimester. If it is necessary, the TI should be <1.0 and exposure time minimised (ideally to 5-10 minutes).[12]

Recommendation 9	Grade and references
Routine clinical scanning of every woman during pregnancy using real time B-mode imaging is not contraindicated. The ALARA principle for duration of ultrasound exposure ('as low as reasonably achievable') should be observed.	Consensus Based Recommendation Reference [11]
Recommendation 10	**Grade and references**
Pulsed Doppler ultrasound should not be used routinely in first trimester. If it is necessary, the TI should be <1.0 and exposure time minimised (ideally to 5-10 minutes).	Consensus Based Recommendation Reference [12]

4.6 Non-medi

6. References

1. Whitworth M, Bricker L, Neilson JP, Dowswell T. Ultrasound for fetal assessment in early pregnancy. Cochrane Database Syst Rev. 2010(4):Cd007058.
2. Kirwan D, and NHS Fetal Anomaly Screening Programme in collaboration with the Royal College of Obstetricians and Gynaecologists (RCOG), British Maternal and Fetal Medicine Society (BMFMS) and the Society and College of Radiographers (SCoR). 18+0 to 20+6 Weeks Fetal Anomaly Scan. National Standards and Guidance for England. 2010 ISBN 978-0-9562084-1-5.
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6. Syngelaki A, Chelemen T, Dagklis T, Allan L, Nicolaides KH. Challenges in the diagnosis of fetal non-chromosomal abnormalities at 11-13 weeks. Prenat Diagn. 2011;31(1):90-102.
7. Grande M, Arigita M, Borobio V, Jimenez JM, Fernandez S, Borrell A. First-trimester detection of structural

Appendices

Appendix A

Human Genetics Society of Australia (HGSA) and the Royal Australian and New Zealand College of Obstetricians and Gynaecologists (RANZCOG) Joint Committee on Prenatal Diagnosis and Screening Membership

Name	Expertise	Role
Dr Agnes Wilson – RANZCOG member	RANZCOG Guideline developer Research Scientist	Committee Chair. RANZCOG Senior Coordinator, Guideline development and Women's Health
A/Professor Michael Gabbett – HGSA member	Paediatrics and Epidemiology	Senior Staff Specialist in Clinical Genetics, Genetic Health Queensland, Associate Professor, Griffith University, Senior Lecturer, The University of Queensland
Professor Jane Halliday – HGSA member	Epidemiology and Research	Head, Public Health Genetics Genetics Theme, Murdoch Childrens Research Institute
Clinical Professor Jon Hyett – RANZCOG member	Obstetrics and Gynaecology	Head of High Risk Obstetrics, Royal Prince Alfred Women and Babies. Clinical Professor, Obstetrics and Gynaecology University of Sydney
Dr Natalie Kiesey-Calding – RANZCOG member	Obstetrics and Gynaecology	Private Consultant, Cairns Obstetrics & Gynaecology
Ms Pauline McGrath – HGSA member	Genetic Counselling and Prenatal Screening and Diagnosis	HGSA Certified Genetic Counsellor at Queensland Health
Dr Andrew McLennan – RANZCOG member	Obstetrics and Gynaecology	Consultant to the Maternal Fetal Medicine Unit at Royal North Shore Hospital and a Partner at Sydney Ultrasound for Women
A/Professor Ricardo Palma-Dias – RANZCOG member	Obstetrics and Gynaecology	Clinical Director - Ultrasound Services, Royal Women's Hospital, Victoria. Clinical Associate Professor at University of Melbourne
Dr Jason Pinner – HGSA member	Medical Geneticist	University of Sydney (member to January 2014)
Professor Peter Stone – RANZCOG member	Obstetrics and Gynaecology Professor of Maternal Fetal Medicine	The University of Auckland
Dr Marleen Susman – HGSA member	Public Health Geneticist	Murdoch Childrens Research Institute (member to January 2014)
Professor Susan Walker – RANZCOG member	Obstetrics and Gynaecology Professor of Maternal Fetal Medicine	Shiela Handbury Chair of Maternal Fetal Medicine, Director Perinatal Medicine, Mercy Hospital for Women
Dr Dianne Webster – HGSA member	Laboratory Science	Lead Clinical Scientist, LabPlus, Auckland City Hospital, New Zealand

Appendix B Women's Health Committee Membership

Name	Position on Committee
Professor Yee Leung	Chair
Dr Joseph Sgroi	Deputy Chair, Gynaecology
Associate Professor Lisa Hui	Member
Associate Professor Ian Pettigrew	EAC Representative
Dr Tal Jacobson	Member
Dr Ian Page	Member
Dr John Regan	Member
Dr Craig Skidmore	Member
Associate Professor Janet Vaughan	Member
Dr Bernadette White	Member
Dr Scott White	Member
Associate Professor Kirsten Black	Member
Dr Greg Fox	College Medical Officer
Dr Marilyn Clarke	Chair of the ATSI WHC
Dr Martin Byrne	GPOAC Representative
Ms Catherine Whitby	Community Representative
Ms Sherryn Elworthy	Midwifery Representative
Dr Amelia Ryan	Trainee Representative

Appendix C Contributing Author

Dr Anna Lee	COGU Subspecialist and Consultant, Fetal Diagnostic Unit, Monash Medical Centre

Appendix D Overview of the Development and Review Process for this Statement

i. Steps in developing and updating this statement

This statement was originally developed in August 1991 and was re-developed during 2015. The statement was reviewed by the Women's Health Committee in March 2018. The WHC 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 March 2018 face-to-face committee meeting, 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

Good Practice Note	Practical advice and information based on clinical opinion and expertise

Appendix E 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.

Appendix F Considerations for Indigenous and Culturally and Linguistically Diverse Populations

4.2.1 There should be appropriate communication with all women. Particular care should be taken to ensure that communication is clear and understood by women who are from culturally and linguistically diverse populations (including women from an Indigenous background).

4.2.2 In Australia, the Department of Immigration and Citizenship offers Free Interpreting Services through TIS National for private medical practitioners (defined as General Practitioners and Medical Specialists) providing Medicare rebate-able services and their reception staff to arrange appointments and provide results of medical tests. Free interpreters are also available in New Zealand.

4.2.3 A resource developed especially for Indigenous women by the Menzies School of Health Research is available on line at this link: - Fetal Anomaly Screening Resource "Take Home Booklet" Menzies School of Health Research.