



## C-Obs 5

# Joint HGSA/RANZCOG Prenatal diagnosis policy

*This document has been prepared by the Joint HGSA/RANZCOG Prenatal Diagnosis and Screening Committee.*

### Recommendations:

1. The increasing applications for and use of prenatal diagnosis make it necessary for all regions to have a specialised team for prenatal diagnosis for birth defects and genetic disorders. This team should consist of clinical and laboratory services.
2. There should be at least one specialised prenatal diagnostic service for all regions. Such services should be located in tertiary obstetric facilities available in the public sector. Independent private practitioners should be encouraged to make use of the regional specialised prenatal diagnostic service for consultation and referral and the service would expect to provide some support for and collaborative work with the private sector.
3. Each specialised prenatal diagnostic service requires the services of a multi-disciplinary team of health professionals, whose specialities may be dependent on the setting. Typically the team would comprise a clinical geneticist, genetic counsellor, midwife and/or nurse specialising in prenatal diagnosis, medical specialist in obstetric ultrasound, obstetrician specialising in prenatal diagnosis and management of fetal abnormality, paediatrician, social worker, a clinic coordinator, laboratory staff, and secretarial assistance. The specialised prenatal diagnostic service units should use state of the art ultrasound equipment that is well maintained and serviced. Appropriate space should be provided for counselling. The ultrasound and the counselling areas should be furnished so that consultants are seen in relaxing and pleasant surroundings. To practice effectively in the rapidly developing areas of genetics and prenatal diagnosis, library resources, Internet access and continuing medical education need to be available.
4. Laboratory units should consist of specialist biochemical, molecular genetic and cytogenetic facilities. Laboratory units should be NATA/IANZ accredited, with previous experience of carrying out the requested investigation and participate in appropriate external quality assurance programs. These laboratories should have documented experience in prenatal diagnosis and for rare disorders this may mean transfer of samples to laboratories interstate or overseas. Senior local laboratory staff should be considered part of the specialised prenatal diagnostic service team with a commitment to full flow of information between clinical and laboratory staff.
5. When a fetal anomaly is found and the implications are not clear, case management should include further opinions from relevant specialists (eg clinical geneticist, paediatric surgeon or neurosurgeon) and a multi-disciplinary group discussion should be encouraged to consider options for care and management.

6. All procedures should be performed by operators who have had appropriate training. There is evidence that the fetal loss rates for all invasive procedures are operator and experience dependent. Prenatal diagnostic service providers or pregnancy care providers who do not perform sufficient procedures per year to maintain skills should be encouraged to refer their cases to a specialised prenatal diagnostic service that does.
7. Amniocentesis should be performed or closely supervised by an operator who has appropriate training in the procedure, and performs sufficient numbers annually to maintain expertise. Concurrent ultrasound should be used. Amniocentesis is usually performed from 15 weeks gestation and should not routinely be performed before 14 weeks gestation because of the increased risk of adverse outcome.
8. CVS should not be performed before 10 weeks gestation because of the risk of transverse limb reduction defects. CVS should be performed or closely supervised by an operator using concurrent ultrasound. The operator should have adequate training and should perform sufficient numbers annually to maintain expertise.
9. Fetal blood sampling (direct ultrasound guided fetal blood sampling) should also be performed or closely supervised by operators trained in this procedure who perform a sufficient number of such samplings to ensure technical success (ie. sampling fetal blood), and to minimise the complication rate. (Maintenance of a procedure of this technical difficulty requires considerable and continuing experience).
10. Fetal magnetic resonance imaging (fetal MRI) is a new imaging modality used in the second and third trimesters. It may provide additional information relevant to the prognosis and recurrence risk of fetal anomalies, specifically of fetal brain anomalies. Fetal MRI is technically difficult. Its use is not known to pose a risk to the fetus, but its safety is not proven. Fetal MRI should only be performed in centres that have the appropriate equipment and where the radiologists and MRI technologists have training and experience in fetal MRI. Fetal MRI should only be used as an adjunct to expert ultrasound to answer specific clinical questions. Fetal MRI is not a screening test and it should not replace ultrasound as the imaging modality of choice for the midtrimester fetal morphology scan. Patients should be referred to a specialised prenatal diagnostic service for an assessment and ultrasound scan prior to fetal MRI.
11. All specialised prenatal diagnostic service should be closely associated with clinical genetic and diagnostic facilities in each State/Territory to allow effective counselling of the extended family if required. Facilities for amniocentesis or CVS should be available at least for the following:
  - Women of advanced maternal age who are not sufficiently confident to proceed with combined first trimester screening after receiving full information about such screening, or do not have such screening readily available to them. \*
  - Women who have a screening test for chromosomal abnormalities, the results of which suggest a risk greater than the accepted cut-off being used in the protocol.
  - All other women who have a high risk of a fetus with a diagnosable defect e.g. ultrasound-diagnosed abnormality, inborn error of metabolism, DNA identifiable condition, chromosome abnormality.
  - Maternal infectious diseases.
12. The choice between CVS, amniocentesis, first trimester combined testing (nuchal translucency with maternal serum screening), second trimester serum screening, integrated screening (nuchal translucency, first and second trimester maternal serum screening) should be on the basis of informed consent. This should take into consideration the risks of the test, timing, method of termination which may be considered (if affected) and accuracy of the test. All women should be offered a mid-trimester ultrasound scan.

13. Couples should receive appropriate counselling, ideally well before the diagnostic procedures are performed. This must include, at least, the risk of occurrence or recurrence, the risks and benefits of the procedure, and exploration of the couple's response to an abnormal result. Educational material should be supplied.
14. Interpretation of results should be a team responsibility. The results should be communicated to the referring doctor and patient as soon as possible and in a manner that ensures clear understanding. The action to be taken on the basis of abnormal results is a decision for the couple concerned based on the information given with full counselling support. Where termination of pregnancy is undertaken because of an abnormal test the managing doctor must first sight a written report.
15. Each specialised prenatal diagnostic service should be expected to monitor its own performance of prenatal diagnostic procedures and make statistics available about their sampling success rate, proportion of abnormalities detected, fetal loss rate and other complications. The outcome of all pregnancies where prenatal diagnosis has taken place should be monitored in a standard manner.
16. The pathological examination of an aborted fetus and placenta should be done by a pathologist experienced in fetal pathology with due regard to confirmation of the prenatal diagnostic test result and including fetal radiography, cytogenetic analysis and any biochemical or molecular investigation which is indicated.
17. Pathological examinations should not be commenced until written consent has been obtained from the parents. Limited pathological examination should be made available if requested, customised to the request of the parents.
18. Preimplantation genetic diagnosis is an evolving method of very early prenatal diagnosis and is not discussed in any detail here. An appropriate level of assessment, counselling support and collaborating health professionals are required for preimplantation genetic diagnosis as listed here for prenatal diagnosis.
19. Paternity testing is not a medical indication alone for prenatal diagnosis, but it is recognised that molecular genetic testing may allow paternity to be implied. There may be legal implications in this testing. Where prenatal diagnosis is used for paternity testing the patient requesting this, and preferably the putative father of the pregnancy, should receive counselling about the implications of the testing, and the implications for the family. This should include discussion of the likely outcome for the pregnancy when the actual father is identified. Paternity testing is sometimes useful in a family consulting the genetics clinic for a genetic disorder and these families should receive counselling both for the disorder and for the implications of paternity testing.
20. To ensure ease and equity of access to specialised prenatal diagnostic services by the public, appropriate funding should be provided from Government sources for the provision of these services as well as staff training and the purchase and maintenance of equipment.
21. The local health planning authority should make provision for publicising specialised prenatal diagnostic services and informing health professionals and the community of their availability.

- \* Some States/Territories/Regions offer prenatal diagnosis by invasive procedures at the age of 35 years and over. This policy statement is not meant to alter this practice but has stated 37 years as a minimum standard. This policy does NOT specifically recommend

such invasive procedures in place of screening, but supports the option of an invasive procedure without screening as a choice for women.

### **Links to other related College statements**

The following statement prepared by the Joint HGSA/RANZCOG Prenatal Diagnosis and Screening Committee, should be read in conjunction with this policy:

[C-Obs 4 Best Practice Guidelines on Antenatal screening for Down Syndrome \(DS\) and other Fetal aneuploidy](#)

[C-Gen 2 Guidelines for Consent and the provision of information regarding proposed treatment](#)

### **Patient Resources**

RANZCOG patient information pamphlets:

- Antenatal care and routine tests during pregnancy - a guide for women (July 2002)
- Prenatal Screening tests for Down syndrome and other conditions (July 2002)

### **Disclaimer**

This College Statement is intended to provide general advice to Practitioners. The statement should never be relied on as a substitute for proper assessment with respect to the particular circumstances of each case and the needs of each patient.

The statement 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, and the application of this statement in each case. In particular, clinical management must always be responsive to the needs of the individual patient and the particular circumstances of each case.

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

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