C-Obs 35

Prenatal Screening for Fetal Genetic or Structural Conditions

1. General Principles

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.

1.1. Screening

Either pre-pregnancy, or early in the antenatal period, counselling should address:
- A detailed clinical assessment for any factors that might increase the chance of fetal structural or genetic conditions. Couples at increased chance of having a baby with a structural or genetic condition should receive individualised counselling from a health professional with appropriate expertise.
- Screening tests available include: cell-free DNA testing, combined first-trimester screening, second trimester serum screening, and second trimester fetal morphology ultrasound (at 18-22 weeks).
- Screening of the woman and her partner for gene changes that can result in their baby inheriting a specific genetic condition. This is called carrier screening. For further details, see C-Obs59.

1.2. Diagnostic testing

Where clinical assessment or screening tests identify an increased chance of fetal genetic or structural condition, timely access to diagnostic testing should be provided. These diagnostic tests may include:
- amniocentesis or chorionic villus sampling; or
- specialised fetal imaging (ultrasound/ MRI)
- Other specialised tests according to individual circumstances
2. Prenatal Screening Tests 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.

3. Clinical Assessment

A detailed medical history and clinical examination should be obtained in order that any particular needs of the couple can be addressed. Where possible, this should take place at a pre-pregnancy visit or alternatively at the earliest opportunity in pregnancy.

4. Counselling and Information

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) and the offer of a follow up diagnostic test if an ‘increased’ probability result is obtained.
- That continuing or not continuing the pregnancy are both options in the event that 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.
5. **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.

6. **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.

**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](http://www.fetalmedicine.com)

**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)
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.

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