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| Policy Facilitator: Jeremy Tuohy<br>Authorised by: Executive Director Clinical Surgery, Women & Children's Directorate | Version no 4<br>Issue Date: 5th March 2012<br>Review date: 5th March 2015 | Policy no. W&CHD<br>**WC UT-01** |

# Antenatal diagnostic screening and testing for aneuploidy

## Related documents

W&CHD PPPG documents:

- Anti-D Immunoglobulin administration and Kleihauer testing

Other:
New Zealand Genetic Services - *Maternal serum test information sheet*
New Zealand Genetic Services - *Maternal serum test request form* (for bloods)
The Royal Australian and New Zealand College of Obstetricians and Gynaecologists – *Amniocentesis and chorionic villus sampling information sheet.*

# Policy

Chromosomal aneuploidy describes any variation in chromosome number that involves individual chromosomes as opposed to entire sets. This is a major cause of perinatal morbidity and mortality and the diagnosis can have significant long-term consequences for both the infant and their family.

Prenatal non-invasive screening and invasive diagnostic testing are carefully targeted to identify the majority of these cases. Non-invasive screening is necessary in order to facilitate timely information and/or intervention in those pregnancies which are affected.

This policy is to:

1. Inform all providers of maternity care about the current screening and diagnostic services which are available.

2. Enable all pregnant women who have a positive non-invasive screening test to be appropriately counselled by their lead maternity carer (LMC), hospital-based midwife or medical personnel. The woman must then be offered referral to the Maternal Fetal Medicine (MFM) Service at Capital and Coast District Health Board in a timely manner.

# Scope

- All WHS Obstetricians, Registrars, and Senior House Officers
- All WHS Midwives
- All Access holders
- All Ultrasonographers

- All Cytogenetics laboratory personnel

# Indications for diagnostic testing

Antenatal diagnostic testing for aneuploidy is indicated when:

1. There is a personal or family history of an inheritable chromosomal or genetic disorder.
2. A woman has been screened for aneuploidy by any of the currently available screening methods (see below) and has been identified as being 'at risk'.
3. When a fetal anomaly has been identified by ultrasound scan which is associated with an increased risk of aneuploidy.

# Non-invasive screening tests which are currently available.

## Maternal age

Maternal age is a poor screening method for chromosomal aneuploidy. The Ministry of Health recommends that women are not offered diagnostic testing on the basis of maternal age alone.

## Nuchal translucency

Nuchal translucency is an effective screening method which is performed between 11 and 13+ weeks gestation. This test is arranged by the LMC and is recommended for all women.

This type of ultrasound screening should however be performed in a unit which demonstrates quality control and is preferably a member of the MFM screening programme which uses current software from the Fetal Medicine Foundation, in London.

## Maternal serum screening

Maternal serum screening is another effective screening method, the test is now funded by the New Zealand government. MSS1 consists of testing for PAPP-A and hCG at 9 to 14 weeks. The MSS2 consists of testing for Inhibin A, hCG, Oestriol and AFP at 14 to 20 weeks. The MSS1 is performed as part of the combined test. The Ministry of health has recommended the combined test as the primary screening method. The MSS2 is recommended for those women who are too far advanced in gestation for a combined screen.

## Combined testing

Combined testing consists of a combination of nuchal translucency testing at 11 to 14 weeks and first trimester maternal biochemistry . The risks from the maternal age, nuchal translucency, presence of the nasal bone and maternal biochemistry are combined to give a single risk. At risk of more than 1 in 300 is considered "high risk" and warrants referral for an amniocentesis.

# Invasive testing

Any woman who has a positive non-invasive screening test (1:300 or greater) **must be** offered referral to the MFM service in a timely fashion. If the woman declines referral this should be clearly documented in her hospital medical records and signed by the woman.

The type of invasive testing that will be offered to women depends largely upon the gestation at which the woman is referred. Chorionic villus sampling (CVS) is offered from 11+ week's gestation and amniocentesis is offered from 15+ weeks.

## The referral process

- The referring practitioner is responsible for informing the woman about the need for referral and explaining why invasive testing is indicated.
- All referrals for invasive testing will be triaged by the MFM specialists. The MFM service will decide which invasive tests are appropriate for the woman. The circumstances and the timing of these tests will also be decided by the MFM service.
- If a woman declines invasive testing this will be clearly documented in the woman's hospital medical records.

## Blood tests to be organised by the referring LMC

A copy of the woman's antenatal bloods and her blood group must accompany the referral form, as the MFM service will not be able to schedule an appointment until this information has been made available.

## Discussion and consent process

Written consent will be obtained prior to any invasive procedure being performed.

## Chorionic villus sampling:

Those women who undergo invasive testing prior to 14 weeks will be offered CVS. This procedure is usually performed via the trans-abdominal route, but it may also be performed trans-vaginally.

The trans-abdominal route is sometimes considered unsuitable once the location of the placenta has been verified.

### Trans-abdominal technique

- Sterile technique.
- The placenta is located using ultrasound.
- A 20 gauge needle is inserted into the uterus under direct ultrasound visualisation.
- Aspiration of the chorionic villi is achieved using negative pressure in a 20 to 30 millilitres syringe.

- The adequacy of the sample is assessed by the operator, the MFM coordinator or the cytogenetics laboratory.
- Two attempts are acceptable. Further attempts may be required, but are discouraged as this leads to a higher rate of miscarriage.

**Trans-vaginal technique**

- Sterile technique.
- The woman is placed into lithotomy.
- The placenta is located using ultrasound.
- CVS forceps are introduced into the cervix under direct ultrasound visualisation.
- Two attempts are acceptable. Further attempts may be required, but are discouraged as this leads to a higher rate of miscarriage.

## Amniocentesis:

Amniocentesis is the preferred method of invasive screening if the woman is more than 15 weeks pregnant.

- Sterile technique.
- The amniotic fluid is located using ultrasound.
- A 22 gauge needle is inserted into the uterus under direct ultrasound visualisation.
- Between 10 and 20 millilitres of amniotic fluid is required
- Two attempts acceptable. Further attempts may be required, but are discouraged as this leads to a higher rate of miscarriage.

## Analysis of the CVS and amniocentesis samples

- All samples will be analysed using GTG-banding after standard culture. These results are usually available within 12 to 14 days of the invasive procedure; the PCR results are available within 48 hours.

- Women who undergo invasive testing for aneuploidy on the basis of age alone will **not** be offered rapid analysis of their sample.

- CVS samples may be analysed using a direct preparation method, which allows for a rapid full karyotype where appropriate after discussion with the cytogenetics laboratory.