Screening for haemoglobinopathies in pregnancy

Who must comply with this procedure?
All doctors and midwives providing antenatal care or pre-pregnancy advice to couples.

This procedure applies in the following setting:
Women and their partners that are either pregnant or planning a pregnancy.

Definitions

**Partner**: Father of the baby.

**FBE**: Full Blood Examination

**Hb**: Haemoglobin

**MCV**: Mean Corpuscular Volume

**MCH**: Mean Corpuscular Haemoglobin

**HbEPG**: Haemoglobin electrophoresis

Procedure

1. **Offer pre-pregnancy or early antenatal investigations**
   a. Offer all women screening for haemoglobinopathies pre-pregnancy or in early pregnancy.
   b. Offer women a copy of the information sheet produced by Thalassaemia Australia titled “Family Planning.”
   c. Offer all women an FBE, HbEPG, ferritin (if indicated), and DNA analysis (if indicated) performed at Southern Cross Pathology.
   d. Women who have an abnormal FBE (low Hb, MCV or MCH) or HbEPG will automatically be identified by Southern Cross Pathology and a ferritin and DNA analysis will be performed.
   e. Male partners of women with an abnormal FBE and / or HbEPG will also require investigation. These men are to have an FBE, HbEPG, ferritin (if indicated) and DNA analysis (if indicated) performed at Southern Cross Pathology.

2. **Request tests**
   a. The request form must include:
      - Name, address, date of birth and Southern Health UR number (if known) of person having blood test.
      - If a male partner is having blood tests performed the female partner’s details (name, date of birth and Southern Health UR number) are to be included on the request form as this aids the laboratory to match the couple’s results.
   b. For women having hospital based care, including shared care with hospital, initial patient investigations are to be requested at the Midwives Assessment Clinic (MAC) appointment. Women are to be advised to have these antenatal screening tests as soon as possible and at least one week prior to their next appointment at the hospital.
   c. If practitioners choose not to use Southern Cross Pathology they need to understand that ferritin and DNA analysis will not automatically occur and these will need to be ordered separately. Practitioners should be aware that all haemoglobinopathy DNA analysis for
3. Review and communicate results
   a. The referring clinician is responsible to ensure review of each person’s blood results within a reasonable timeframe.
   b. For women having hospital based care, review antenatal screening results including haemoglobinopathy screen, at the next hospital appointment, ie ‘first medical visit. At this visit the partner screen, if required, is ordered. Appropriate arrangements need to be made at this visit for review of partner screen results within two weeks. This review may be in person or by telephone / mail.
   c. Communicate results to the couple and documented in the hospital medical record and the patients hand held antenatal record.

4. Act on results
   a. If a woman’s Hb, MCV, MCH and HbEPG are normal, then the risk of being a carrier of a haemoglobinopathy and having a child affected with a major haemoglobinopathy is low and no further testing is required. However, HbH disease risk may be overlooked unless the partner has an FBE.
   b. If a woman has an abnormal Hb, MCV, MCH or HbEPG and her partner’s results are normal the woman may be a carrier of a haemoglobinopathy but the couple are at low risk of a child with a major haemoglobinopathy.
   c. If the woman and her partner both have abnormal results, especially identification of an abnormal HbEPG and / or DNA analysis has identified a gene mutation, referral to the Medical Therapy Unit at Monash Medical Centre, Clayton is required. The Medical Therapy Unit, Thalassaemia Service, can be contacted by telephone: 95942756 or fax: 95946648.
   d. If the couple have had their results linked at the time of ordering (by following the process outlined above) then the laboratory will issue a summary of the results and any potential implications. This can assist in deciding which couples require referral to the Medical Therapy Unit.
   e. Women with low ferritin levels are to be advised to commence on iron and vitamin C supplementation.
   f. Men with low ferritin levels are to be referred to their General Practitioner for ongoing management.
   g. Offer the appropriate information sheets. The following information sheets are available in English and other languages on the Southern Health website or through Thalassaemia Australia (www.thalassaemia.org.au).
      - Family planning
      - Alpha Thalassaemia
      - Beta Thalassaemia
      - Haemoglobin E
      - Sickle Cell Disease.
   h. Encourage women or their partners identified as having a haemoglobinopathy to contact an appropriate support group eg. Thalassaemia Australia.

- See flow chart for summary
Useful resources
Thalassaemia Australia: www.thalassaemia.org.au.

Document Management

Policy supported: Evidence based clinical care.

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Executive sponsor: Chief Operating Officer

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If this is a hard copy it might not be the latest version of this document. Please see the Southern Health site for current documents.

Disclaimer

The maternity clinical practice procedures and guidelines have been developed having regard to general circumstances. It is the responsibility of every clinician to take account of both the particular circumstances of each case and the application of these procedures and guidelines. In particular, clinical management must always be responsive to the needs of the individual woman and particular circumstances of each pregnancy.

These procedures and guidelines have been developed in light of information available to the authors at the time of preparation. It is the responsibility of each clinician to have regard to relevant information, research or material which may have been published or become available subsequently. Please check this site regularly for the most current version.
Screening for haemoglobinopathies in pregnancy

**Procedure**

Discuss screening and provide information sheet on “Family Planning”

**Pathology tests recommended for the woman**
- FBE
- HbEPG
- Ferritin (if indicated)
- DNA analysis (if indicated)

**NORMAL patient Hb, MCV, MCH and HbEPG**
- Risk of patient being affected by a haemoglobinopathy is low.
- Risk of child being affected by a major haemoglobinopathy is low.
- No further management needed.
- Document results on antenatal record.
- Does not exclude minor alpha thalassaemia carrier state.

**ABNORMAL patient Hb, MCV, MCH or HbEPG**
- Southern Cross Pathology will perform ferritin and DNA analysis.
- Partner screen required – order as above.

**NORMAL partner Hb, MCV, MCH and HbEPG**
- Risk of partner being affected by a haemoglobinopathy is low.
- Risk of child being affected by a major haemoglobinopathy is low.
- Clarify patient condition and manage appropriately.
- Document results on antenatal record.
- Does not exclude HbH disease risk.

**ABNORMAL partner Hb, MCV, MCH or HbEPG**
- Refer couple to Medical Therapy Unit at MMC, Clayton on 9594.2756 (phone) or 9594.6648 (fax).
- Document results on antenatal record.

**If abnormality confirmed requires**
- counselling about patient / partners status
- counselling about potential risk to child
- counselling about testing family members
- appropriate information sheet
- information about support groups.