Thalassaemia Screening in Pregnancy (Updated March 2009)

FBC/Haemoglobin EPG, and Iron Studies as per ANSC protocol

- **Normal MCV/MCH HbF < 5%**
  - Low risk of having a baby with a haemoglobinopathy
  - PARTNER Normal FBC Normal HbEPG
    - Low risk of having a baby with a haemoglobinopathy
  - PARTNER Normal FBC Abnormal HbEPG
    - REFER for GENETIC COUNSELLING RPAH/TCH 9515 5080

- **HbF ≥ 5%**
  - Test Partner – FBC, HbEPG, Iron studies

- **Low MCV/MCH Increased Hb A2 Consistent with Beta Thalassemia**
  - PARTNER Abnormal FBC Abnormal HbEPG
    - REFER for GENETIC COUNSELLING RPAH/TCH 9515 5080

- **Carrier of a haemoglobinopathy such as HbS, HbE, HbD**
  - Low risk of having a baby with a haemoglobinopathy
  - PARTNER Normal FBC Normal HbEPG
  - PARTNER Normal FBC Abnormal HbEPG
    - REFER for GENETIC COUNSELLING RPAH/TCH 9515 5080

- **Low MCV/MCH Normal Hb EPG (Hb A2) “Carrier of Alpha thalassemia”**
  - Low risk of having a baby with a haemoglobinopathy
  - PARTNER Normal FBC Normal HbEPG
  - PARTNER Normal FBC Abnormal HbEPG
    - REFER for GENETIC COUNSELLING RPAH/TCH 9515 5080

- **Low MCV/MCH Normal Hb EPG (Hb A2) Alpha thalassemia not excluded**
Screening for Haemoglobinopathies in Pregnancy

Current SSWAHS ANC guidelines recommend that all pregnant women have haemoglobinopathy carrier testing, in the first trimester. Screening involves a full blood count (FBC) and haemoglobin electrophoresis (hbepg). Iron studies (which includes ferritin) are essential for the molecular genetics laboratory to interpret molecular testing results. When this screening indicates that the woman is a carrier of a haemoglobinopathy, or screening is inconclusive, her partner should be screened to determine his haemoglobinopathy carrier status. If the woman’s partner is unavailable for testing she should be referred to clinical genetics for a consultation.

It is noted that raised hbf is common in pregnancy. If the hbf is less than 5% with normal MCV and MCH there is no need for the partner to have screening.

When both the woman and her partner are carriers of a haemoglobinopathy, or they both have inconclusive results they should be referred for genetic counselling so possible implications for the pregnancy and molecular testing will be discussed. As molecular testing for haemoglobinopathies is time consuming it is important that at-risk couples are identified as early in the pregnancy as possible.

If you are uncertain about any results you can contact a member of the clinical genetics team on 9515 5080, or fax a copy of the results you are concerned about to 9550 5389.