OBSTETRICS AND GYNAECOLOGY
CLINICAL PRACTICE GUIDELINE

Haemoglobinopathy screening in pregnancy

Scope (Staff):	WNHS Obstetrics and Gynaecology Directorate staff
Scope (Area):	Obstetrics and Gynaecology Directorate clinical areas

This document should be read in conjunction with this Disclaimer

Contents

Aims

- To identify and screen women and their partners at risk of haemoglobin disorders and identify fetuses at risk of significant haemoglobin disease
- To offer genetic counselling to women and their partners identified as high risk
- To provide a referral mechanism for women to the Maternal Fetal Medicine (MFM) Service for specialised multidisciplinary obstetric management when there is a confirmed haemoglobin disorder which carries risk for the woman or fetus

Background

Normal haemoglobin contains an iron-containing haem molecule that combines with four globin chains. The major globin chain in adults consists of two alpha (α) chains, encoded by four α globin genes, and two as beta (β) chains, encoded by two β globin genes. Haemoglobinopathies occur as a result of changes in the structure or quantity of these globin genes. The resulting changes in the assembled haemoglobin molecule may result in haemolysis or impaired erythropoiesis.[1, 2]

Haemoglobinopathies are autosomal recessive disorders, indicating that sufferers must inherit abnormalities from both parents. Global estimates suggest that approximately 7% of the world's population are carriers of haemoglobinopathy. They are becoming more prevalent within Australia due to changes in migration patterns.[3]

Thalassaemia

Thalassaemia is caused by mutations in, or deletions of, the α or β genes resulting in decreased globin chain production. It is classified as alpha (α)-thalassaemia when there is impaired synthesis, or beta (β)- thalassaemia when there is impaired β-chain synthesis.[1, 2] Both forms of thalassaemia typically show a reduction in mean corpuscular volume (MCV) and mean corpuscular haemoglobin (MCH) with or without accompanying anaemia. The severity of the disorder will depend on the number of abnormal genes present. A single α gene deletion/dysfunction may have no impact on the patient or red cell indices, whereas deletion of all four α genes results in Bart's hydrops fetalis,[1-8] commonly resulting in intrauterine death unless treated by intrauterine transfusion. Clinically, β-thalassaemia ranges from an asymptomatic carrier state to thalassaemia major characterised by lifelong dependency on blood transfusions and associated complications including iron overload.[4-7] See 'Effects of thalassaemia types' in this document.

Partner testing is required if a woman has thalassaemia trait because co-inheritance with a second thalassaemic typically results in more significant clinical manifestations.[1-4]

Variant haemoglobins

Variant haemoglobins are also caused by mutations in the α or β genes. In contrast to thalassaemia, resulting in reduced globin production, the mutations result in production of a structurally abnormal haemoglobin. There are a large number of

variant haemoglobins with variable clinical impact. Sickle cell disease is a special case, discussed below. Haemoglobin E(HbE) is also common and has significant clinical implications for the fetus if co-inherited with β-thalassaemia trait.[1-7]

Sickle cell disease

Sickle cell disease is characterised by production of an abnormal β-globin chain that forms haemoglobin S (HbS). Those with one abnormal β-globin chain have sickle cell trait; those with sickle cell disease have inherited abnormalities from both parents. Sickle cells have a shortened lifespan, causing chronic haemolytic anaemia, and – under hypoxia and other conditions – may result in acute haemolytic / ischaemic episodes known as sickle cell crises.[1, 2, 4, 7] Maternal effects may include pain, infections, pulmonary complications, anaemia, pre-eclampsia and high rates of caesarean section. The fetus is at risk for spontaneous abortion, preterm birth, intrauterine growth restriction and perinatal death.[1-8]

Geographical distribution of haemoglobin disorders

Historically, the prevalence of α- and β- thalassaemia was high in the Middle East, Mediterranean countries, South East Asia, Indian sub-continent and parts of Africa. A severe form of α-thalassaemia (α^0) is found in South East Asia and parts of the Mediterranean.[1, 2, 4-6] Haemoglobin E (HbE) commonly occurs in the South East Asia and the Indian sub-continent; sickle cell disease is prevalent in tropical Africa.[1, 2, 4-6] See ‘Populations at risk of haemoglobin disorders’ below.

With changing patterns of migration within Australia and throughout the world, it is important to assess a patient’s ethnic background to assess if they are at risk of haemoglobinopathy. [3-5, 7] In addition, identifying the patient’s ethnic background on the haemoglobinopathy request form also assists the laboratory in directing testing and interpreting the results.[4, 7]

Populations at risk of haemoglobin disorders

Population locations with associated high risk for haemoglobin disorders: [1-5, 7]

Thalassaemia	Sickle cell disease
• African • American/British/Caribbean African • South East Asian and Chinese • Middle Eastern • Pacific Islanders • New Zealand Maori • Southern Europe/Mediterranean • Indian subcontinent • Some northern Western Australian and Northern Territory Australian indigenous communities.	• African • American/British/Caribbean African • Middle Eastern • Southern Europe/Mediterranean • Indian subcontinent • South American

Effects in pregnancy

Effects of thalassaemia types in pregnancy [1-8]

Type of thalassaemia	Genotype	Effect
α+ thalassaemia trait (α thalassaemia minor)	One deleted α gene on one chromosome only (e.g. αα/α –)	Normally asymptomatic. Slight decrease in MCV/MCH
α+ thalassaemia trait (α thalassaemia minor)	Two deleted α genes, one on each chromosome (e.g. α-/α-)	Reduced MCV/MCH +/- mild anaemia
α^0 thalassaemia trait (α thalassaemia minor)	Deletion of two α genes on one chromosome (e.g. αα/--)	Reduced MCV/MCH +/- mild anaemia
Haemoglobin H disease	Total of three deleted α genes (e.g. α-/--)	Moderate microcytic anaemia, hepatosplenomegaly. May require transfusion
Haemoglobin Barts	Complete absence of all α genes (i.e. --/--)	Usually foetal hydrops and death in utero
β thalassaemia minor	One absent (β^0) or impaired (β^+) β gene on one chromosome only (e.g. β/β^0 or β/β^+)	Normally asymptomatic. Reduced MCV/MCH +/- anaemia.
β thalassaemia intermedia	Defined clinically. Various genetic defects (e.g. β^+/β^+ or β^0/β^+)	Variability in symptoms. Moderate microcytic anaemia; hepatosplenomegaly. May require transfusion.
β thalassaemia major	Two absent β genes (i.e. β^0/β^0)	Severe microcytic anaemia. Requires frequent blood transfusions.
Haemoglobin E trait	One normal β gene on one chromosome and one abnormal β gene on the other chromosome (e.g. β/β^E)	Asymptomatic. Possible decreased MCV/MCH
Haemoglobin E disease	Two abnormal β genes (i.e. β^E/β^E)	Mild anaemia, reduced MCV/MCH
Haemoglobin E/β thalassaemia	One absent/impaired β gene (β^0 or β^+) and one abnormal β gene (e.g. β^E/β^0 or β^E/β^+)	Moderate to severe microcytic anaemia. Clinical symptoms vary – potentially thalassaemia intermedia or major.

NB. This list is a guide only. Advice should always be obtained from Haematology in the first instance, particularly in women with coinherited haemoglobinopathy (e.g. α thalassaemia and HbE)

Effect of sickle cell in pregnancy [1-7]

Sickle status	Genotype	Effect
Sickle cell trait	One normal β gene and one abnormal β gene mutation (e.g. β/β^S)	Asymptomatic normally, normal RBC indices. Sickle cell formation can occur in during high fever and significant hypoxia
Sickle cell disease/anaemia	Two abnormal β genes (e.g. β^S/β^S)	Mild to moderate chronic haemolytic anaemia, vaso-occlusion - brain, chest, bones, kidneys, spleen and placenta. Increased maternal and perinatal mortality

NB. This list is a guide only. Advice should always be obtained from Haematology in the first instance, particularly in women with coinherited haemoglobinopathy (e.g. α or β thalassaemia and HbS).

Screening women for haemoglobin disorders

Refer to flow chart at the end of this guideline.

1. Screening and referral process

All antenatal women should be offered screening if they fall into these categories:

- If the woman is of Black African/African Caribbean/African American or Black British origin (irrespective of red cell indices)
- Past history of unexplained anaemia
- Family history of anaemia (unknown cause) or haemoglobinopathy
- If the family originates from a geographical location which puts them at risk of haemoglobin disorders (see previous section) **AND** has a MCV ≤80fL or MCH ≤27pg

2. Women with NO identified risk factors for haemoglobinopathy

Assess the FBP (full blood picture) and ferritin levels (if done) at the booking visit.

- If FBP is normal, reassess FBP at 28 weeks gestation or at time of glucose tolerance test. If the MCV is ≤80fL or the MCH ≤27pg, assess iron studies.
- Treat underlying iron deficiency if ferritin <30ug/L and reassess FBP once iron deficiency is corrected.
- See WNHS Clinical Guideline: Obstetrics and Gynaecology: Anaemia and Iron Deficiency: Management in Pregnancy and Postpartum

3. Women who are at risk of haemoglobinopathy

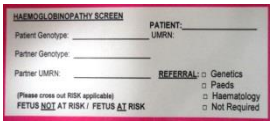
1. Assess ethnic background.
2. Assess FBP and iron studies
3. Determine if haemoglobinopathy screening has been performed previously (e.g. through General Practitioner (GP) or in previous pregnancy). If so, obtain a copy and file the results. If testing was performed outside Australia, or the results are not available, testing should be repeated.
4. If the woman or partner has been found have a haemoglobinopathy, ascertain if the same partner is the father of the current pregnancy. In the case of a new partner, partner studies will be required (see section 4 'Obtaining partner haemoglobin studies' below).
5. All known, suspected or being screened haemoglobinopathies need to be referred to the Clinical Nurse Consultant Patient Blood Management (CNC PBM), via e-referrals ('Haematology CNC PBM').
6. The CNC PBM will then commence a Haemoglobinopathy Management Plan and continue with the processes below.
7. Determine if partner testing has been performed previously (e.g. through GP or in previous pregnancy). Obtain a copy of the results, clearly label as "partner studies" and place in the woman's medical records.
8. If testing has not been performed (or results are unavailable), provide counselling and with maternal consent arrange haemoglobinopathy studies **on the woman only**. Request "FBP, iron studies and haemoglobinopathy studies". The request form should also include the woman's ethnic background to guide interpretation.
9. Women should be directed to have the testing undertaken at a PathWest Laboratory to facilitate full haemoglobinopathy screening including DNA studies.
10. If ferritin levels are <30ug/L initiate treatment for iron deficiency. See WNHS Clinical Guideline: Obstetrics and Gynaecology: Anaemia and Iron Deficiency: Management in Pregnancy and Postpartum. IV iron should not be given until a blood sample has been obtained for haemoglobin studies as this may interfere with result interpretation.
11. If the woman is attending a low risk midwives' clinic and the haemoglobin studies are abnormal, the woman should be referred to the CNC PBM and Team Obstetric clinic for follow-up and counselling. If the haemoglobin studies results are normal the women may then continue antenatal care at the midwives' clinic.
12. If the woman's results identify a haemoglobinopathy which places the fetus at risk, partner studies should be performed urgently (see section 4 'Obtaining partner haemoglobin studies'). Arrange for the woman to be reviewed in the obstetric clinic.
13. If partner test results confirm a haemoglobinopathy which places the fetus at risk, arrange referral to MFM Service for multidisciplinary care. This may also include genetic counselling, physician and haematologist involvement.

4. Obtaining partner haemoglobin studies

1. Ideally partner screening is obtained preconception such that the parents can make an informed decision about risks prior to pregnancy. In the absence of this, maternal results are used to direct partner testing. Concurrent testing of the woman and the partner is only performed when specifically directed by the MFM Service, CNC PBM or Haematology Department.
2. Provide counselling and with paternal consent arrange haemoglobinopathy studies on the partner. Request “FBP, iron studies and haemoglobinopathy studies”. The request form should be completed with the partner’s demographic details; a UMRN will be registered when he attends a PathWest laboratory for testing. In the clinical notes section include his ethnic background, the woman’s details and haemoglobin genotype (i.e. αα/--) and clearly identify **“Partner testing”**.
3. Partners should be directed to have the testing undertaken at a PathWest Laboratory to facilitate full haemoglobinopathy screening.
4. When initiating partner screening create e-referral to Haematology CNC PBM and complete questionnaire and add additional details.

5. Assessing the fetus at risk

1. The risk to the fetus is dependent upon the maternal and paternal genotype. Assessment of the risk is undertaken by the CNC PBM and Haematology department. If the fetus is identified as **high risk** for haemoglobin disease by the CNC PBM and Haematology department, an urgent e-referral is to be created and sent to MFM service via the Obstetric unit providing details of both the woman and her partner’s results.
2. Following review of the results by The MFM Service an appointment will be arranged for the woman to attend KEMH for assessment as soon as possible +/- referral to Genetic Services WA (GSWA) if required.
3. An individual management plan will be developed and documented in the medical records between the MFM Service and the woman/partner. This may include invasive testing, specialised ultrasonography and family testing. A Neonatal Management Plan may also be developed to direct specific management and neonatal testing following delivery of the infant. A copy of the Neonatal Management Plan will be forwarded to the Neonatal Department via the Neonatal Clerical Support team.
4. The haemoglobinopathy screen sticker and MR 036 is completed by the CNC PBM and placed on the MR004.


HAEMOGLOBINOPATHY SCREEN
Patient Genotype: _____ PATIENT: _____
UMRN:
Partner Genotype: _____
Partner UMRN: _____ REFERRAL: □ Genetics
□ Paeds
□ Haematology
□ Not Required
(Please cross out RISK applicable)
FETUS NOT AT RISK / FETUS AT RISK

5. If the fetus is identified as **not at risk f**or haemoglobin disease on review of

the results by Haematology, the CNC PBM will complete the Management Plan and place the MR 036 and sticker in the patient medical notes.

6. Women and current partners who have been screened in previous pregnancies

If women and their current partners have been screened previously within Australia and results are available for both high performance liquid chromatography (HPLC) and DNA analysis, no further testing is required. Obtain a copy of the results and place in the medical records. If the results are not available via the GP, original testing laboratory or within the patient's medical/laboratory records, re-testing is required. See previous sections ('3- Women who are at risk of haemoglobinopathy disorders', '4- Obtaining partner haemoglobin studies' and ' 5- Assessing the fetus at risk'), for maternal and paternal testing as required. If the woman is known to have haemoglobinopathy and presents with a new partner, partner testing should be initiated.

All previously known and tested women must still be referred to the CNC PBM for the current pregnancy.

7. Partners who decline 'paternal' testing, partner no longer in contact with woman or unknown

In a woman with a confirmed haemoglobinopathy, screening is aimed to identify the fetus at risk and enable informed choice regarding management of an affected infant. Partners should be encouraged to participate in screening. However, if after careful discussion the partner declines screening (or is not available for testing) this should be clearly documented in the woman's medical records, and the process below followed:

1. Create and complete e-referral to haematology CNC PBM Haemoglobinopathy and complete questionnaire and add additional details.
2. Haematology Department will review the results. The fetus will be considered to be **at risk** for haemoglobin disease. An e-referral toThe MFM Service may be arrange for the woman to attend KEMH for assessment as soon as possible and, depending on the maternal genotype, consider referral to Genetic Services.
3. An individual management plan will be developed and documented in the medical records. This may include invasive testing, specialised ultrasonography and family testing. A Neonatal Management Plan will be developed to direct specific management and neonatal testing following delivery of the infant. A copy of the Neonatal Management Plan will be forwarded to the Neonatal Department via the Neonatal Clerical Support team.
4. The haemoglobinopathy screen sticker is completed by the CNC PBM and placed on the MR004.

8. Women referred to KEMH for haemoglobinopathy screening

Women are frequently referred to KEMH for assessment of fetal risk when the woman has a confirmed or suspected haemoglobinopathy. The screening process is followed to ascertain the risk to the fetus. Not all women will require delivery and care at KEMH, although women will be required to attend at least one hospital appointment for midwifery and medical review.

Women with identified at risk pregnancies will generally remain under the care of KEMH for antenatal care, delivery and neonatal care.

9. Women who attend KEMH in early pregnancy / non-pregnant with suspected or confirmed haemoglobin disorder

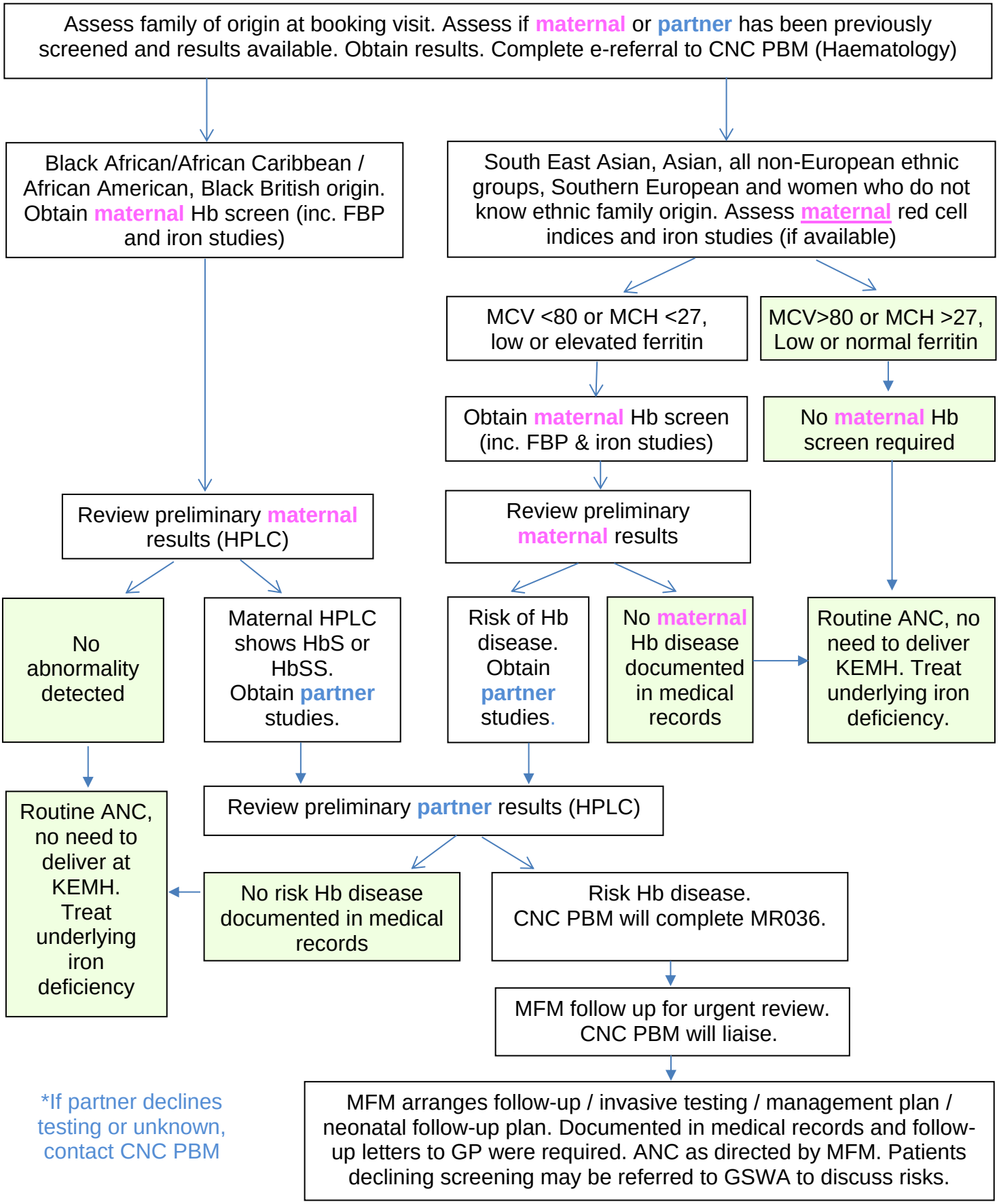
Women who attend KEMH (e.g. Emergency Centre, Outpatient clinics – exclusive of ANC – or Ward 6) with a suspected or confirmed haemoglobin disorder may require additional follow-up dependent on age and childbearing potential. If the woman is likely to become pregnant, then follow-up of results and partner testing (if appropriate), as above, is warranted. If the woman will no longer be attending KEMH for follow-up, the woman's GP should be contacted by letter with the results and instructed to undertake partner testing if the woman has childbearing potential.

Management of co-existing iron deficiency

It is important to monitor ferritin levels in pregnant women with a confirmed haemoglobinopathy disorder at booking and at 34 weeks gestation. Iron deficiency should be treated per current guidelines. See WNHS Clinical Guideline: Obstetrics and Gynaecology: Anaemia and Iron Deficiency: Management in Pregnancy and Postpartum

Women may present at booking with an elevated ferritin level and develop iron deficiency as pregnancy progresses. As haemoglobinopathy commonly causes microcytosis[5],the most reliable marker to assess iron deficiency are trends in ferritin (and other parameters available on iron studies) and haemoglobin levels over time. The administration of IV iron will not correct anaemia in iron replete women and should not be given.

Screening women for haemoglobin disorders: Flow chart



*If partner declines testing or unknown, contact CNC PBM

Abbreviations: ANC- Antenatal clinic, FBP- full blood picture, GP- General Practitioner, GSWA- Genetic Services WA, Hb- haemoglobin, HPLC- high performance liquid chromatography, HbS- sickle cell haemoglobin, HbSS- sickle cell disease, MCV- mean cell volume, MCH- mean cell haemoglobin, MFM- Maternal Fetal Medicine

Significant maternal haemoglobinopathy – MFM Service referral

Significant maternal haemoglobinopathies requiring referral and obstetric management by the Department of Maternal Fetal Medicine, KEMH

Pregnant women with significant haemoglobinopathy should have their obstetric care managed through the MFM Service. A multidisciplinary approach, with collaboration between Obstetricians, Haematologists, Neonatologists, Geneticists and Pathologists (regarding laboratory testing) is preferred in these cases.

The following list identifies the women with confirmed significant haemoglobinopathy who require immediate referral to the MFM Department for ongoing care and management on confirmation of pregnancy.

Significant maternal haemoglobinopathies requiring immediate referral to MFM in pregnancy
• Maternal sickle cell anaemia and all other types of sickle cell disease (i.e. HbSS, HbSC, HbSD-Punjab • HbS/β thalassaemia • β thalassaemia intermedia, β thalassaemia major • HbE/β thalassaemia • HbH (α-/--) • Pregnancies with a 1 in 4 risk of significant haemoglobinopathy in the fetus (determined by review of maternal and paternal haemoglobinopathy screening) *

*Pregnancies / fetus considered ‘not at risk’ do **not** require tertiary level obstetric care

No risk of fetal haemoglobinopathy – Sign off by CNC PBM

- The tables below identify situations where the CNC PBM at KEMH may acknowledge results without medical review.
- An asterisk (*) denotes that CNC sign off of 'no fetal risk' will only occur in the absence of other co-inherited haemoglobinopathy. This is also dependent on the number and type of α and/or β gene deletions / mutations in the case of maternal and paternal haemoglobinopathy.
- All other haemoglobinopathy must be reviewed by the MFM Service or Consultant Haematologist as they will have an identified or increased risk of haemoglobinopathy requiring multidisciplinary assessment.

Table 1: Maternal ALPHA globin disorders and paternal status

		Paternal status									
		Normal	α^{+}thal trait (α-/α- or αα/α-)	α^{0} thal trait (αα/--)	HbH disease (α-/--)	β thal trait	β thal int./maj	HbE trait	HbE disease ; HbE + β thal	HbS trait	HbS disease
Maternal status	Normal		CNC	CNC	CNC	CNC	CNC	CNC	CNC	CNC	CNC
	α^{+} thal trait (α-/α- or αα/α-)	CNC	CNC*	MFM	MFM	CNC*	CNC*	CNC*	CNC*	CNC*	CNC*
	α^{0} thal trait (αα/--)	CNC	MFM	MFM	MFM	CNC*	CNC*	CNC*	CNC*	CNC*	CNC*
	HbH disease (α-/--)	CNC	MFM	MFM	MFM	MFM	MFM	MFM	MFM	MFM	MFM

Table 2: Maternal BETA globin disorders and paternal status

		Paternal status									
		Normal	α^{+} thal trait (α-/α- or αα/α-)	α^{0} thal trait αα/--)	HbH disease	β thal trait	β thal int./maj	HbE trait	HbE disease ; HbE + β thal	HbS trait	HbS disease
Maternal status	Normal		CNC	CNC	CNC	CNC	CNC	CNC	CNC	CNC	CNC
	β thal trait	CNC	CNC*	CNC*	CNC*	MFM	MFM	MFM	MFM	MFM	MFM
	HbE trait	CNC	CNC*	CNC*	CNC*	MFM	MFM	MFM	MFM	MFM	MFM
	HbS trait	CNC	CNC*	CNC	CNC*	MFM	MFM	MFM	MFM	MFM	MFM
	β thal int./major	CNC	MFM	MFM	MFM	MFM	MFM	MFM	MFM	MFM	MFM
	HbE disease	CNC	MFM	MFM	MFM	MFM	MFM	MFM	MFM	MFM	MFM
	HbE/β thal	CNC	MFM	MFM	MFM	MFM	MFM	MFM	MFM	MFM	MFM
	HbS disease	CNC	MFM	MFM	MFM	MFM	MFM	MFM	MFM	MFM	MFM

References and resources

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Related WNHS policies, guidelines, procedures and related forms

WNHS Clinical Guideline: Obstetrics and Gynaecology: Anaemia and Iron Deficiency: Management in Pregnancy and Postpartum

Forms

- MR004 Obstetric Special Instruction Sheet
- MR036 Referral for Haemoglobinopathy Screening

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Version history

Version number	Date	Summary
1	Jan 2010	First version. Previously titled 'B1.1.12 Haemoglobinopathies Screening and Referral'. Contact OGD Guideline Coordinator for archived versions.
2	Apr 2014	Screening and referral process updated
3	Oct 2014	Amendment
4	Oct 2015	Flowchart added
5	Apr 2017	Title changed to 'Haemoglobinopathy Screening in Pregnancy' **History**: In April 2017 amalgamated three individual documents on haemoglobinopathy management dating from Jan 2010. **Supersedes**: 1. B1.1.12 Haemoglobinopathy Screening (dated Oct 2014) 2. Assessment of No fetal risk of Hb disease which may be signed off by the CNC Haematology (dated Sept 2016) 3. Significant maternal haemoglobinopathies requiring referral and obstetric management by the Department of Fetal Medicine, King Edward Memorial Hospital (procedure- no date)
6	Sept 2017	Minor amendment- reinserted header
7	Feb 2022	• CNC Haematology changed to CNC Patient Blood Management • e-referral added as method of referring • Background information about variant haemoglobins added, updated tables for effects of thalassaemia types and sickle cell • Women at risk of haemoglobinopathy- changed from assessing ferritin to assessing iron studies • Refer to Haematology CNC PBM via e-referral- ➢ All known, suspected, or being screened, haemoglobinopathies. The CNC PBM will commence a Haemoglobinopathy Management Plan. ➢ When initiating partner screening or if partner testing declined, partner no longer in contact or unknown. Complete questionnaire and add additional details. ➢ All previously known and tested women must still be referred to the CNC PBM for the current pregnancy. • If the fetus is identified as high risk for haemoglobin disease by the CNC PBM and Haematology department, an urgent e-referral is to be created and sent to MFM service via the Obstetric unit providing details of both the woman and her partner's results. • 'not a carrier' changed to 'normal' in sign off by CNC PBM table

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