Action following Antenatal Haemoglobinopathy Screen

Request Haemoglobinopathy Screen if any of:

- High risk ethnicity in patient and partner: South east Asian, Asian, Indian, Sri Lankan, Pakistani, Bangladeshi, Middle Eastern, Mediterranean, Black African, Islander
- MCV <80fL or MCH <26.5pg
- Known haemoglobinopathy carrier, family history of haemoglobinopathy in mother or partner's family

Request
- Full Blood Count
- Haemoglobinopathy screen
- Iron studies

Haemoglobinopathy Screen Result

Beta thalassaemia trait, δβ thalassaemia
or
Abnormal variant detected, such as HbS, HbE, HbC, HbD-Punjab, HbO-Arab, Hb Lepore
or
Hb Constant Spring
or
HbH disease

→ yes → *Further action required*

HbA_2: 2.0 - 3.2% → yes → MCV <80fl or MCH < 26.5pg → yes → Alpha thalassaemia cannot be excluded → *Further action required*

no

HbA_2: 3.3 - 3.7% → yes → Beta thalassaemia cannot be excluded → *Further action required*

HbH bodies detected
or
ICT strip positive

→ yes → *Further action required*

Additional Comments

Molecular studies would be required
Alpha thalassaemia cannot be excluded
Atypical beta thalassaemia cannot be excluded
Partner should be screened

→ *Further action required*

Further Action

If only mother's results available → then **partner** must be **screened** with urgent haemoglobinopathy screen

If 'further action required' from **BOTH** mother AND father's results → **refer urgently** to Genetics or Haematology

Contacts: **RHW** Prenatal Genetic Service Phone: 9382 6098; 9382 6099; 9382 6042 Fax 9382 6038
StG Department of Clinical Genetics Phone: 9113 3635 Fax 9113 3694

Action that will occur through Genetics or Haematology following Antenatal Haemoglobinopathy Screen, when *both* parents involved.

Beta globin problem in both parents (Box A or B)

Box A
HbA_2: 3.3 – 3.9%
Atypical beta thalassaemia cannot be excluded
Molecular studies required

→ *Beta gene molecular testing*

Variant present

Box B
Beta thalassaemia trait
HbS, HbE, HbC, HbD-Punjab, HbO-Arab
δβ thalassaemia, Hb Lepore

→ *Urgent genetic referral*

Alpha globin problem in both parents (Box C or D)

Box C
MCV <80fl or MCH < 26.5pg
HbH bodies detected
ICT strip positive
Alpha thalassaemia cannot be excluded
Molecular studies required
HbE with MCV < 80 or HbE % <27%
HbS with MCV < 80 or HbS % <35%

→ *Alpha gene molecular testing*

2 gene deletion present

Box D
Hb Constant Spring
HbH disease

→ *Urgent genetic referral*