Thalassaemia & Haemoglobinopathy

Introduction:

Thalassaemia & haemoglobinopathies are inherited blood disorder that affects the body's ability to produce haemoglobin, leading to anaemia and other health complications.

Early detection through screening allows parents to make informed decisions about their pregnancy and prepare for any necessary care for their baby. It also provides an opportunity for genetic counselling, helping parents understand the implications and the options available to them.

Actions by LMC's:

If thalassemia or a haemoglobinopathy is suspected* in pregnancy or planning to become pregnant, there are several necessary follow-up actions:

- **Maternal screening**: If either thalassaemia / haemoglobinopathy is suspected, then after informed consent, screening should be undertaken**.
- **Parental Screening**: If a woman is found to be a carrier, the biological father should be offered screening** (We suggest this occurs at the same time as mother being investigated).
- **Referral to Obstetric team**: Couples at risk of having a baby with a significant haemoglobin disorder - see table 1 (Red Orange), should be referred for assessment of risk to baby (See Guidelines for consultation with Obstetric and related medical services – Referral guidelines).

Actions by Obstetric clinicians:

- **(Sickle cell anaemia, HbH disease in pregnancy** – please refer to Haematology)
- **Genetic Services referral / Counselling:**
  If pregnancy with a baby at high risk of a significant haemoglobin disorder, then couples should receive genetic counselling to understand their options – this will be offer / arranged by Obstetric team (Email to: Genetic Services Secretary (ADHB) GenSec@adhb.govt.nz).
- **Fetal monitoring:**
  In most cases of confirmed fetal hemoglobinopathy, fetal diagnosis does not alter ongoing obstetric care, as fetal hemoglobinopathy typically has no adverse effects on the fetus, mother, or course of pregnancy, with some exceptions e.g. HbH disease, Sickle cell disease.

  One exception is cases of alpha thalassemia major where early pregnancy diagnosis and the decision to continue the pregnancy would prompt changes in pregnancy management, such as close fetal monitoring for fetal anaemia and hydrops.

*HbA1c will identify all clinically significant haemoglobin variants e.g. Hb S & increases in HbA2 or Hb F etc. In these situations, the laboratory will contact the requesting clinician to seek permission to perform relevant investigations. However, we will not be able to identify alpha thalassaemia, this will require proactive investigation – unexplained low MCV / MCH i.e. with normal ferritin, especially important in patients from SE Asia, China etc.

For further information please see information leaflet(s), if advice required please contact Pathlab to discuss with one of our Haematologists: 07 858 0795.

** The LMC can initiate / request these investigations i.e. Thalassaemia screen.

Thalassaemia and haemoglobinopathy screening in pregnancy – O&G Magazine

Table 1. Combinations of parental carrier states and their risks to the unborn child.

		Father Carrier of:									
		Hb S	β thalassaemia	δβ thalassaemia	Hb E	Hb O^{Arab}	Hb C	Hb D^{Punjab}	HPFH	Alpha Thalassemia	Not a carrier
Mother carrier of:	Hb S										
	β thalassaemia										
	δβ thalassaemia										
	Hb E										
	Hb O^{Arab}										
	Hb C										
	Hb D^{Punjab}										
	HPFH										
	Alpha Thalassemia	Discuss with Haematologist, risk based upon many variables.									
	Not a carrier										

	Serious risk – Urgent Referral to Obstetric service & genetic counselling.
	Less serious risk – Referral to Obstetric service, may require further investigation.
	No risk – No further action required.

Pathlab information leaflets:

Alpha Thalassaemia - https://bit.ly/3tOGYio
Beta thalassaemia - https://bit.ly/2P3gwms
Delta Beta Thalassaemia - https://bit.ly/3PMmgLg
What is HPPF - https://bit.ly/3uW59hY
Raised HbF - https://bit.ly/3uXHuOe
What is Sickle cell disease - https://bit.ly/3rtH3X5
What is sickle cell trait - https://bit.ly/3qHw12U
What is Haemoglobin E - https://bit.ly/2P1StnO
What is Haemoglobin D - https://bit.ly/3DxXny1
What is Haemoglobin C - https://bit.ly/36tW4Ed