Laboratory testing for Haemoglobinopathies in the Waikato region

SUMMARY

Haemoglobinopathy testing in the Waikato region is available in the Haematology Laboratory, Waikato Hospital. Haemoglobinopathies are inherited disorders and fall into two categories: decreased production of haemoglobin (thalassaemia; alpha or beta) or an abnormal form of haemoglobin (variant). Certain ethnic groups have a higher prevalence of thalassaemias and variant haemoglobins. Testing should be used to inform, counsel and prevent the complications of the more severe thalassaemic syndromes and Haemoglobinopathies.

WHY TEST FOR HAEMOGLOBINOPATHIES:

- Sickling disorders are associated with severe life threatening vaso-occlusive crisis, overwhelming sepsis, splenic sequestration, aplastic crisis, stroke, priapism, pulmonary hypertension, proliferative retinopathy, chronic organ damage
- Some thalassaemias are incompatible with life i.e. hydrops foetalis
- Thalassaemia major requires lifelong blood transfusions
- Co-inheritance with other thalassaemias or variant haemoglobins can cause life threatening events
- Haemolytic anaemias can result from inheritance of a haemoglobinopathy

WHO SHOULD BE TESTED:

- **Family History of thalassaemia or sickle cell**
- **Partners of affected people**
- **Cord bloods** from new-borns of affected parents
- **African, African Caribbean, Middle Eastern , Indian subcontinent** – to look for presence of Hb S
- **Asians, particularly South and South East Asians -** where thalassaemia is endemic
- **Greeks, Mediterranean, South Americans, North and South Europeans -** where thalassaemia and variants are prevalent
- **Pacific Islanders -**where thalassaemia is widespread

DEFINITIVE INDICATIONS IN BLOOD WORK THAT REQUIRE TESTING:

- Haemoglobin – Normal or reduced
- **RBC – Increased** >6 $x10^{12}$/L Males, >5.5$x10^{12}$/L Females
- **MCV – Reduced** <80 fL
- **MCH – Reduced** <27 pg
- Iron Studies/ Ferritin – Normal

SAMPLE TYPES REQUIRED:

- EDTA for haemoglobinopathy screen and CBC
- Plain tube for iron studies including ferritin

References:

1) Ryan K, Bain B, Worthington D, et al. Significant haemoglobinopathies: guidelines for screening and diagnosis. Bjh 2010; 149 (1) 35-49
2) NHS. Sickle Cell and Thalassaemia: Handbook for Laboratories. 2012 Ed3v2
3) Old J et al. Prevention of thalassaemias and other haemoglobin disorders. 2012 Vol 2: Laboratory Protocols Ed:2nd

Contact the haematology laboratory or myself for more information if required.

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