pathlab



May 2018

Thalassaemia / Haemoglobinopathy Screening

CLINICAL UPDATE

INTRODUCTION

Thalassemia / Haemoglobinopathies are the most common, inherited, single-gene disorder in the world, with high gene frequency within certain geographic / ethnic groups. The disorders fall into two categories: decreased production of haemoglobin (thalassaemia; alpha or beta) or an abnormal form of haemoglobin (variant).

Testing should be used to inform, counsel and prevent the complications of the more severe thalassaemic syndromes and haemoglobinopathies.

With the changing population of New Zealand, an increasing number of patients with thalassemia / haemoglobinopathies are being identified; hence we wish to outline the appropriate investigation pathway.

WHY TEST FOR HAEMOGLOBINOPATHIES

- Sickling disorders are associated with severe life threatening vaso-occlusive crisis, with significant morbidity and mortality
- Some thalassaemia's are incompatible with life i.e. hydrops foetalis
- Thalassaemia major requires lifelong blood transfusions with significant morbidity
- Co-inheritance with other thalassaemia's or variant haemoglobins can cause life threatening events
- Haemolytic anaemias can result from inheritance of a haemoglobinopathy

INDICATIONS IN BLOOD WORK THAT PROMPT TESTING

- Patient(s) from regions with high frequency of clinically significant Haemoglobinopathy i.e. Hb S
- Haemoglobin – Normal or reduced, with MCV – Reduced <80 fL Or MCH – Reduced <27 pg & Iron Studies/ Ferritin – Normal
- (Abnormal HbA1c report)

THALASSAEMIA / HAEMOGLOBINOPATHY SCREENING PATHWAY

The identification of thalassaemia + other clinically significant haemoglobinopathy is a 2-stage process.

Stage One - **Primary** investigations should include:

- CBC & ferritin
- Thalassaemia / Hb'opathy screen – Identification & quantitation of Hb fractions

The majority of clinically significant variants will be detected, with only a minority of requests requiring:

Stage Two – **Confirmatory** / further investigation(s) which may include:

- (Family or partner investigation) & / Or DNA analysis for final clarification.

Stage two investigations are initiated to enable identification of a few mutations and additionally to provide informative counselling for individuals and couples.
Stage two investigations should only be performed at the direction of a haematologist (or as directed by initial first line investigation), not as a primary screen.

THALASSAEMIA / HAEMOGLOBINOPATHY SCREENING - RECOMMENDATION ACTIONS

To screen for the above, ONLY the primary investigations should be requested.
Please ask for a Thalassaemia screen, which requires 2 - 3 x EDTA's and 1 x SST tube.
Confirmatory requests i.e. Thalassaemia gene studies will only be performed at the direction of a Haematologist (& all requests will be triaged).

If you require any further information or clarification please contact a Pathlab Haematologist via our call centre, 07 858 0799.

Alan Neal, Lead of Speciality - Haematology

Please ensure all members of your institution receive a copy of this clinical update.