Monash Health Pathology

Bulletin

1st August 2022

RE: DNA testing for alpha thalassaemia

Dear pathology service managers,

With the recent introduction of Medicare rebates which cover DNA testing for alpha thalassaemia, The Thalassaemia & Haemophilia Molecular Reference Laboratory (THMR) at Monash Health Pathology will be changing their testing procedures accordingly. Previously, the laboratory provided testing to Victorian residents referred through private pathology with a mean cell volume of ≤82 and/or a mean cell haemoglobin of ≤28. As of July the 1st 2022, the laboratory will be utilizing the parameter guidelines set by Medicare; a mean cell volume of <80, and/or a mean cell haemoglobin of <28. We will, however, continue to offer testing to both members of a couple where, one member is a proven carrier of a clinically significant haemoglobinopathy irrespective of the other partner's haematological parameters as per our previous protocols. Furthermore, we will continue to provide an interpretative risk assessment for the couple in these scenarios.

Monash Health Pathology and the THMR Laboratory recognises that there are gaps in the new Medicare parameters. For this reason, the THMR Laboratory will continue to provide DNA testing to patients in the following scenarios:

- With normal indices, if their partner is an alpha thalassaemia carrier.
- With normal indices, if there is a family history of alpha thalassaemia.
- Iron deficient patients, where a partner is a confirmed carrier of alpha thalassaemia.
- Iron deficient women of childbearing age, irrespective of pregnancy status.
- Who are not of reproductive age and determining carrier status is of clinical relevance.
- With beta thalassaemia for underlying alpha thalassaemia.
- With unknown haemoglobin variants requiring DNA testing to confirm.
- Diagnosis of a disease state e.g., HbE/beta thalassaemia disease, HbH disease, transfusion and non-transfusion dependent thalassaemia and sickle cell disease/anaemia.
- Couples as part of a disease risk scenario, e.g., HbS homozygosity, HbE/β thal, β thal homozygosity etc.
- All CVS samples, for a possible thalassaemia/haemoglobinopathy syndrome.

Please continue to refer samples to the laboratory that fit one or more of the scenarios described above. However, the DNA testing profile will no longer be offered to patients, which do not fulfil the Medicare benefits schedule criteria, and which do not fit within any of the above scenarios.

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