LABUPDATE no. 51

May 2024

GENETIC TESTING FOR HAEMOGLOBINOPATHIES

INTRODUCTION

Haemoglobinopathies are a heterogenous group of disorders caused by inherited defects in the alpha globin (*HBA*) or beta globin (*HBB*) genes. Genetic testing is often necessary to better understand, predict and manage the clinical characteristics of these disorders. Although DNA-based testing is not commonly used as an initial investigation, it is increasingly becoming standard practice to include it as a secondary investigation. It is especially useful in confirming clinically significant disease, for prenatal diagnosis, as well as carrier testing.

Indications for genetic testing include:

- Diagnostic confirmation
- Confirming or excluding carrier status
- Prenatal genetic testing in the context of known parental carrier status
- Heavily transfused individuals where a diagnosis has not previously been confirmed

Genetic testing for thalassaemia and sickle cell disease is now available at Ampath Laboratories. Key information for each test is summarised below.

GENETIC TESTING AVAILABLE AT AMPATH

	Alpha thalassaemia	Beta thalassaemia	Sickle cell disease
Mnemonic	HBAMLPA	HBBSEQ	HBSNGS
Indication	Diagnostic, carrier or pre-natal testing*	Diagnostic, carrier or pre-natal testing*	Diagnostic, carrier or pre-natal testing*
Specimen type	5 ml EDTA blood	5 ml EDTA blood	5 ml EDTA blood
Test methodology	Copy number detection by multiplex ligation-dependent probe amplification (MLPA)	Sanger sequencing and copy number detection by MLPA	Sanger sequencing
Turnaround time	~4 weeks	~6 weeks	~4 weeks
Test requirements	High index of suspicion**	High index of suspicion**	High index of suspicion**
Genetic counselling	Pre- and post-test genetic counselling is available	Pre- and post-test genetic counselling is available	Pre- and post-test genetic counselling is available

* Prenatal testing will only be performed if the familial variant(s) are known and pre-test genetic counselling is essential. The laboratory must be notified before chorionic villous tissue (CVS) or amniotic fluid specimens are collected.

** A family history (including ancestry), full blood count, iron studies and protein-based testing (such as high-performance liquid chromatography (HPLC) or haemoglobin electrophoresis) aids in the interpretation of genetic results and should be provided wherever possible.

Contact the Ampath Genetics Laboratory for information on pricing, genetic counselling services or any other queries at 012 678 0645 of email geneticsclinic@ampath.co.za.