OXFORD Molecular Diagnostics Centre

Oxford University Hospitals NHS
NHS Foundation Trust

NATIONAL HAEMOGLOBINOPATHY REFERENCE LABORATORY
John Radcliffe Hospital, Oxford

New Diagnostic Service Announcement

********RARE INHERITED ANAEMIAS*********

The UK National Haemoglobinopathy Reference Laboratory has been providing a referral service for the investigation haemoglobin disorders for 35 years and we are pleased to announce that we have recently expanded our diagnostic service to include rare inherited anemias.

The diagnosis of rare inherited anaemias has traditionally required a complex set of clinical, biochemical and molecular investigations with the result that in some patients the cause of the anaemia can remain unidentified. The advent of next-generation sequencing (NGS) in the form of targeted panels has made investigation simpler as it allows the analysis of multiple genes to be carried out simultaneously. Studies have also shown that these anaemias can be mis-diagnosed when assessment is based on clinical phenotype alone and that making a correct molecular diagnosis can lead to important changes in clinical management of the patient (1).

The Oxford Red Cell Panel

A 51 gene clinical grade NGS (MiSeq) panel which tests for mutations in genes causing:

* **DBA** (Diamond Blackfan Anaemia)
* **CDA** (Congenital Dyserythropoietic Anaemia)
* **Sideroblastic anaemia**
* **Red cell enzyme disorders** (eg. PK, G6PD, GPI, etc.)
* **Red cell membrane disorders** (eg. Hereditary spherocytosis, hereditary elliptocytosis, etc.)

This test is appropriate for
→ patients with a likely diagnosis from the above list, in whom molecular confirmation is sought
→ patients in whom a diagnosis of inherited anaemia is suspected but an exact diagnosis HAS NOT yet been reached
→ family testing of affected individuals with known mutations
→ screening for mutations in the genes on the panel prior to submission for whole genome sequencing through GEL

Accessing the service

A request form for the service is enclosed; additional copies and further information are available at:

www.oxford-translational-molecular-diagnostics.org.uk/

2-4ml EDTA blood sample sent by first class post is sufficient. All samples will receive full haemoglobinopathy testing prior to molecular analysis for rare anaemias. DBA MLPA will also be carried out where appropriate. **For further information please contact the laboratory on 01865 572769 or email:- oxford.molecularhaem@nhs.net**

(1) Roy NBA, Wilson EA, Henderson S, Wray K, Babbs C, Okoli S, et al. A novel 33-Gene targeted resequencing panel provides accurate, clinical-grade diagnosis and improves patient management for rare inherited anaemias. Brit J Haematol, 2016 Oct;175(2):318-330.