Who we are

Majority owned by the NHS, but with the commercial freedom to invest in innovation, Viapath are on a mission to transform pathology services in the UK. We provide pathology services to the NHS, private hospitals and other organisations both across the country and internationally.

What we do

All our laboratories are either accredited or working towards accreditation by UKAS to ISO15189. To view our laboratory accreditation status please follow this link:

http://www.viapath.co.uk/about-viapath/quality-and-governance/accreditations

TEST OVERVIEW

Description
The alpha thalassaemias are a group of disorders characterised by a reduction in alpha globin synthesis. Each chromosome carries two copies of the alpha globin gene. The laboratory provides a comprehensive service for alpha thalassaemia offering assistance with full blood count (FBC) and HPLC interpretation as well as a definitive diagnostic service. The genetic diagnosis of alpha thalassaemia follows a simple algorithm, testing for common mutations first followed by testing for rarer forms. After each test there is a clinical review. The initial screen is a multiplex assay detecting the common alpha thalassaemia deletions. This assay detects the common alpha plus thalassaemia mutations (alpha 3.7kb and 4.2kb deletions) and the alpha zero thalassaemia mutations (SEA, MED, 20.5kb, FIL deletions). Individuals that are negative for the initial screen undergo alpha globin gene sequencing to detect non-deletional alpha thalassaemia mutations. If negative after sequencing patient samples are screened for rarer deletions using Multiplex Ligation-dependent Probe Amplification (MLPA). In rare cases individuals who are carriers of epsilon gamma delta beta thalassaemia will also have microcytic hypochromic indices and a normal HbA2 percentage; to exclude these carriers a beta locus MLPA assay is performed. In general there is good correlation between the phenotype and the genotype, although this can be obscured by iron deficiency. Antenatal referrals for alpha thalassaemia should be made without waiting for iron results. However, genetic testing can take time and a ferritin level could explain the indices in some cases. The laboratory offers antenatal screening for alpha thalassaemia and can also provide a prenatal diagnostic service for this condition.

Clinical details
Please state if a pregnancy is involved as antenatal work is prioritised. Please identify partner in referral if a fetal risk assessment is required. Please provide full blood count (FBC) and HPLC screening results and iron levels as they become available.

Related condition or disease
- Hb H disease (3 of 4 alpha globin genes affected)
- Hydrops fetalis (4 of 4 alpha globin genes affected)
- Additional alpha globin genes (e.g. Triplicated alpha)

Department
- Special Haematology Department

Laboratory
- Red Cell Centre - Molecular Diagnostics Laboratory

Location
- Viapath at King’s College Hospital

ORDERING INFORMATION

Sample type and Volume required
Volume of blood anticoagulated with EDTA: Adult (16 years and above) 2 x 4 ml, Children (2-15 years) 1 or 2 x 4 ml

Storage and transport
Blood should be stored at 4°C where possible. Send at room temperature by first class post. If possible, please complete

020 7188 7188 (54109)
BusinessDevelopment@viapath.co.uk
Infants (0-2 years) 1 ml. Presence of heparin anticoagulant will inhibit PCR applications. Clotted samples are unsuitable for DNA analysis. For prenatal diagnosis please refer to section for sample requirements.

**Turnaround time**
10 working days from sample receipt. For complex cases where additional tests are required each test will add 10 working days.

the request form attached and send as a hard copy (do not send electronically) with the sample. This will ensure all relevant information is available and will aid us in processing your test.

**Cost**
Please contact Business Development for pricing enquiries.

**Contacts**
Red Cell Centre - Molecular Diagnostics Laboratory
020 3299 1246 / 2265
kch-tr.pnd@nhs.net
c/o Central Specimen Reception
Blood Sciences Laboratory
Ground Floor Bessemer Wing
King’s College Hospital
Denmark Hill
London SE5 9RS
Mon-Fri, 9.00am-5.30pm
How can we help?

We have a number of partnering options to suit your needs, whether you require this specific test or a range of services, we are here to help. Contact one of our friendly Business Development Managers for more information, or visit our website.