



**Genetic Testing Request Form
Haemoglobinopathies**

Lab Use Only

Lab No:
.....

Date received:
.....

For queries, contact: Sheffield Diagnostic Genetics Service, Tel: 0114 271 7014, Email: sheffield.diagnosticgenetics@nhs.net

1. PATIENT INFORMATION - use sticker if available				2. REFERRING CLINICIAN			
Surname:				Consultant:			
Forename:				Hospital:			
DoB: dd/mm/yyyy		NHS No:		Department:			
Sex:		Hospital No:		Copy report to:			
Address:				Telephone No:			
Postcode:				Contact Email:			
Antenatal patient: YES <input type="checkbox"/> NO <input type="checkbox"/>				Address/ Email for report:			
Please specify gestation of pregnancy:							


3. TESTING REQUIRED Please tick which investigations are required in this patient.		By requesting this test you are confirming that this patient meets the eligibility criteria as defined by the: National Genomic Test Directory
<input type="checkbox"/> Alpha Thalassaemia (<i>HBA1/HBA2</i> Sanger sequencing and dosage analysis by MLPA) <input type="checkbox"/> Beta Thalassaemia (<i>HBB</i> Sanger sequencing and dosage analysis by MLPA) <input type="checkbox"/> HPFH (<i>HBG1/HBG2</i> Sanger sequencing and dosage analysis by MLPA)		R93 (diagnostic) R361 (carrier)
<input type="checkbox"/> Sickle Cell Disease (Targeted testing for HbS variant) NM_000518.4(HBB): c.20A>T, p.Glu7Val		R94 (diagnostic) R362 (carrier)

4. PATIENT'S ETHNICITY/COUNTRY OF ORIGIN:		
This information is important as it informs analytical procedures, and it critical for calculating carrier risks. Please be specific.		
A Mixed – please specify	D Asian – please specify country	G Arabic – please specify
B White – British or Other European	E South East Asian – please specify country	H Don't know
C Mediterranean – please specify country	F Black – please specify country	

5. SAMPLE INFORMATION:	Date Taken: dd/mm/yyyy	High Risk of Infection <input type="checkbox"/> (See guidance notes) If yes please affix label to samples and form and specify.
4ml venous blood sample in an EDTA tube . Lithium Heparin or Serum tubes are unsuitable for testing. Forward the completed referral form and EDTA blood sample to your local Genetics Laboratory (see page 2).		

6. LABORATORY RESULTS Please fill in below or attach copy of own result form. Red cell indices and HPLC/IEF results must be supplied before the sample can be processed.							
Hb (g/L)	RBC(x10 ¹² /L)	MCV (fL)	MCH (pg)	Ferritin (µg/L)	Hb A ₂ (%)	Hb F (%)	Other Hb (%)

7. RELATIVE OF A PATIENT WHO IS AFFECTED WITH, OR A CARRIER OF, A HAEMOGLOBINOPATHY Providing details of the patient's relative will allow us to provide a pregnancy risk specific to this couple.	
Name of partner:	
DOB of partner: dd/mm/yyyy	
Status of relative (affected or carrier):	
Details of the partner's variant (if known):	

 North East and Yorkshire Genomic Laboratory Hub https://ney-genomics.org.uk/	Once taken, samples should be sent to your local Genetics Laboratory Please ensure a minimum of 3 matching identifiers on tubes and form Samples should be packed according to UN3373 / P650 and sent 1 st class post will normally be suitable for DNA extraction. Please store samples at 4°C if they cannot be transported the same day.	
Newcastle Genetics Laboratory	Newcastle Genetics Laboratory Central Parkway Newcastle upon Tyne Tyne and Wear NE1 3BZ	NUTH.DNA@nhs.net
		0191 241 8787/8775/8754
		www.newcastlelaboratories.com/lab_service/laboratory-rare-diseases-services/
Sheffield Genetics Laboratory	Sheffield Diagnostic Genetics Service Sheffield Children's NHS Foundation Trust Western Bank Sheffield S10 2TH	sheffield.diagnosticgenetics@nhs.net
		0114 271 7014
		www.sheffieldchildrens.nhs.uk/SDGS.htm
Leeds Genetics Laboratory	Leeds Genetics Laboratory Genomic Specimen Reception Bexley Wing (Level 5) St James's University Hospital Beckett Street Leeds, LS9 7TF	leedsth-tr.DNA@nhs.net
		0113 206 5419/5205
		www.leedsth.nhs.uk/a-z-of-services/the-leeds-genetics-laboratory/