

All fields are mandatory. Illegible, unclear or incomplete forms will result in delays or rejection.

CONSENT STATEMENT: It is the referring clinician's responsibility to ensure that the patient/carer knows the purpose of the test and that the sample may be stored for future diagnostic testing. In signing this form the clinician has obtained consent for testing, storage and for the use of this sample and the information gathered from it to be shared with members of the donor's family through their health professionals (if appropriate). The patient should be advised that the sample may be used anonymously for quality assurance and training purposes. **If the patient does not wish information to be shared please write this clearly in the clinical summary box.**

PATIENT DEMOGRAPHICS	
First name:	
Last name:	
DOB:	Gender: Male <input type="checkbox"/> Female <input type="checkbox"/> Other <input type="checkbox"/>
NHS number:	
Hospital no:	Family ref no:
Postcode:	Antenatal: Yes <input type="checkbox"/> No <input type="checkbox"/>
Non-NHSE funded (please attach invoicing details): <input type="checkbox"/>	

PATIENT ETHNICITY	
White:	British <input type="checkbox"/> Irish <input type="checkbox"/> Any Other White Background <input type="checkbox"/>
Mixed:	White And Black Caribbean <input type="checkbox"/> White And Black African <input type="checkbox"/> White And Asian <input type="checkbox"/> Any Other Mixed Background <input type="checkbox"/>
Asian or Asian British:	Indian <input type="checkbox"/> Pakistani <input type="checkbox"/> Bangladeshi <input type="checkbox"/> Any Other Asian Background <input type="checkbox"/>
Black or Black British:	Caribbean <input type="checkbox"/> African <input type="checkbox"/> Any Other Black Background <input type="checkbox"/>
Other Ethnic Groups:	Chinese <input type="checkbox"/> Any Other Ethnic Group <input type="checkbox"/> (please specify: _____)
Not stated <input type="checkbox"/>	Not Known <input type="checkbox"/>

PATHOLOGY RESULTS		Haematology indices	
Iron / liver parameters		Hb	HbF %
Serum Iron		RBC	HbA2 %
Serum TSat		MCV	Hb variant %
Serum Bilirubin		MCH	Absolute Reticulocyte
Serum Ferritin		Platelets	Reticulocyte %
Blood Film comments:			

SAMPLE REQUIREMENTS	
For haemoglobinopathy investigation:	2 x 4 ml EDTA blood
Children and adults (all other tests):	4 ml EDTA blood
Infants:	1 ml EDTA blood
As DNA for Next Generation Sequencing:	3-5µg genomic DNA
As DNA for all other tests:	1-5µg genomic DNA
Date sample collected: _____	

**CLINICAL INFORMATION, FAMILY HISTORY AND CONFIRMATION OF ELIGIBILITY** Is the patient affected?

Please demonstrate how your patient meets the eligibility criteria for this test. Interpretation of results depends on the quality of clinical information provided. Find the eligibility criteria here: <https://www.england.nhs.uk/wp-content/uploads/2018/08/rare-inherited-disease-eligibility-criteria-v7.1-OFFICIAL.pdf>

**Molecular Tests - For Haemoglobinopathy referrals FBC and HPLC results MUST be provided**

<input type="checkbox"/> R93 Hb variant identification	<input type="checkbox"/> R176 Gilbert's genotyping (TA5/6/7/8 repeat)
<input type="checkbox"/> R93 Haemoglobinopathy investigations	<input type="checkbox"/> R95 Hereditary haemochromatosis (HFE - C282Y and H63D variants)
<input type="checkbox"/> R93 Alpha thalassaemia	<input type="checkbox"/> R191 Alpha-1-antitrypsin genotype (S and Z alleles)
<input type="checkbox"/> R93 Beta thalassaemia	<input type="checkbox"/> Thrombophilia genetic screen (please tick all that apply): FVL <input type="checkbox"/> PT <input type="checkbox"/> MTHFR <input type="checkbox"/>
<input type="checkbox"/> Other (please state gene): _____	<input type="checkbox"/> R240/242/244 Familial variant testing (please state familial variant): _____

For further details of each test please refer to the [Synnovis website](#).

**Next Generation Sequencing - Please select which panel(s) are required**

<input type="checkbox"/> R91 Cytopenia (NOT Fanconi anaemia)	<input type="checkbox"/> R96 Iron metabolism disorders	<input type="checkbox"/> R92 Rare Anaemia
<input type="checkbox"/> Neutropenia	<input type="checkbox"/> R168 Porphyria	<input type="checkbox"/> Membranopathy
<input type="checkbox"/> Diamond-Blackfan anaemia	<input type="checkbox"/> R169 Acute intermittent porphyria	<input type="checkbox"/> Enzymopathy
<input type="checkbox"/> Inherited bone marrow failure	<input type="checkbox"/> R170 Variegate porphyria	<input type="checkbox"/> Haemoglobinopathy
<input type="checkbox"/> R313 Neutropenia consistent with ELANE mutations	<input type="checkbox"/> R347 Inherited predisposition to AML	<input type="checkbox"/> Congenital dyserythropoietic anaemia
<input type="checkbox"/> HLH	<input type="checkbox"/> R366 Inherited predisposition to ALL	<input type="checkbox"/> Diamond-Blackfan anaemia
<input type="checkbox"/> Single gene analysis: _____ (name of gene)	<input type="checkbox"/> R405 Hereditary erythrocytosis	<input type="checkbox"/> Sideroblastic anaemia
	<input type="checkbox"/> R406 Thrombocythaemia	<input type="checkbox"/> Haemolytic anaemia
	<input type="checkbox"/> R323 Sitosterolaemia	<input type="checkbox"/> Sitosterolaemia
		<input type="checkbox"/> Megaloblastic anaemia

For details of genes in each subpanel, please refer to the [Synnovis website](#)

CLINICIAN DETAILS	
Requesting clinician / consultant Name: Hospital & department:	Responsible clinician / consultant (if different) Name: Hospital & department:
NHS email: Phone:	NHS email: Phone:

Note: Please ensure the latest version of this request form is used, found on our website: [www.southeastgenomics.nhs.uk](http://www.southeastgenomics.nhs.uk)