Oxford Regional Genetics Laboratory
Churchill Hospital
Headington
Oxford OX3 7LE
For sending samples, see address below
Email Enquiries: oxford.molecularhaem@nhs.net Telephone: 01865 572769

NHS
Central & South
Genomic Laboratory Hub
Oxford Genetics Laboratories

Rare Disease in Molecular Haematology Testing Service
Genomic Panel Request Form

Patient Details

NHS No:		**Sex:**	
Surname:		**Address:**	
Forename:			
Date of Birth:		**Postcode:**	
Ethnicity:		**Reference No:**	
Suspected diagnosis:			

Test requested#	**R91 Cytopenia**	☐	**R405 Hereditary Erythrocytosis** (provide JAK2 results)	☐			
	R92 Rare Anaemia	☐	**R406 Thrombocythaemia**	☐			
	R229 Fanconi anaemia	☐	Chromosome breakage studies completed?	☐			
	R313 ELANE	☐	**R259 NBN**	☐	**R338 CSF3R**	☐	**Family testing e.g.R375** ☐

This is the proband: ☐

This is a family member (please complete details below): ☐

Proband name:		Proband date of birth:		
Relationship to proband:		This individual is:	Affected ☐	Unaffected ☐

For information on genes tested in each panel, please visit: https://panelapp.genomicsengland.co.uk/panels/

Requester Details

Clinician:		**Job Title:**	
Email*:		**Phone No:**	
Reporting Address:		**Invoice Address:**	**Same as reporting?** ☐

*Electronic Reporting via Email: The Oxford Genetics Laboratories are now offering the option to receive reports by email. If you would like to receive future reports via this method please provide your email address in the referrer details section (NHS.net email preferred). To set this up, the laboratory will contact you with further information.

Clinical Details

RBC (red blood cell count, x10^12/L)		**WBC** (white blood cell count: x10^9/L)								
HGB (haemoglobin, g/L)		**Neutrophils** (x10^9/L)								
MCV (mean corpuscular volume, fl)		**Lymphocytes** (x10^9/L)								
MCH (mean corpuscular haemoglobin, pg)		**Haptoglobin** (g/L)								
Haematocrit (proportion)		**Bilirubin** (umol/L)								
Reticulocytes (x10^12/L&%)		**SGOT/AST** (Serum glutamic oxaloacetic transaminase, IU/L)								
Platelets (x10^9/L)		**SGPT/ALT** (Setum glutamic pyruvic transaminase, IU/L)								
Ferritin (mlg/L)		**LDH** (Lactacte dehydrogenase, IU/L)								
Transferrin (%)		**DAT** (Direct antiglobulin test/coombs test)								
Erythropoietin (EPO, IU/L)		**HPLC** (%)	HbA:		HbA2:		HbF:		Variant?:	

Blood smear results

Bone marrow smear results (including date analysed)

Clinical Details

Anaemia onset	Fetus ☐	Infant ☐	Child ☐	Adult ☐			
Type	Acute ☐		Chronic ☐		Transfusion Dependent ☐		
Neutropenia onset	Fetus ☐	Infant ☐	Child ☐	Adult ☐			
Type	Acute ☐		Chronic ☐		Transfusion Dependent ☐		
Thrombocytopenia onset	Fetus ☐	Infant ☐	Child ☐	Adult ☐			
Type	Acute ☐		Chronic ☐		Transfusion Dependent ☐		
Jaundice	Prolonged neonatal ☐		Intermittent ☐		Chronic ☐		
Splenomegaly	Yes ☐	No ☐	**Hepatomegaly**		Yes ☐	No ☐	
Pancreatic insufficiency	Yes ☐	No ☐	**Gallstones**		Yes ☐	No ☐	
Dysmorphic facies	Yes ☐	No ☐	**Skeletal, limb or digit abnormalities**		Yes ☐	No ☐	
Developmental delay/learning difficulties	Yes ☐	No ☐	**Any other organ abnormalities**		Yes ☐	No ☐	
Short stature/failure to thrive	Yes ☐	No ☐	**Frequent infections**		Yes ☐	No ☐	
Family History	Yes ☐	No ☐	**Consaguinity**		Yes ☐	No ☐	
JAK2 Results (for R405 Hereditary Erythrocytosis)							
Any other relevant details: (e.g. treatment details, test results, non-haematological findings, transplant histories)							

Sample Information

Sample type (tick):	DNA ☐	EDTA Blood ☐	Date sampled
Labelling standards:	Please label samples with the **patient's**: full name, date of birth, NHS number (or Hospital Number for non-UK referrals). A **minimum** of 2 identifiers must be provided or the sample cannot be accepted for testing.		

Consent

In submitting this sample, the clinician confirms that informed consent has been obtained for: (a) storage and testing (current and future testing as this becomes available) (b) the use of this sample and the information generated from it to be shared with members of the donor's family and their health professionals (if appropriate) and (c) the information generated to be entered onto local and national confidential databases. If specific consent to any of the above is not given, please provide details below. The patient should be advised that the sample may be used anonymously for quality assurance, training and research purposes.

Consent for research: Consent has been obtained for the DNA/RNA of this sample to be used in research/development projects that have been granted ethical approval:	☐

Signed:		**Clinician:**	
		Date:	

Please send samples at room temperature by post or courier to:

Oxford Regional Genetics Laboratories, Churchill Hospital, Headington, Oxford, OX3 7LE