

## Rare Disease in Molecular Haematology Testing Service Genomic Panel Request Form

Patient Details			
NHS No:		Sex*:	
Surname:			
Forename:		Address:	
Date of Birth:			
Ethnicity:		Postcode:	
Reference No:			
Suspected diagnosis:			
Test requested (For information on genes in each panel, please visit: <a href="https://panelapp.genomicsengland.co.uk/panels/">https://panelapp.genomicsengland.co.uk/panels/</a> )	R91 Cytopenia	R258 Cytopenia including Fanconi Anaemia	
	R92 Rare Anaemia	R405 Hereditary Erythrocytosis (JAK2 results required)	
	R229 Fanconi anaemia	R406 Thrombocythaemia	
	R313 ELANE	R259 NBN	R338 CSF3R
This testing is: Routine      Urgent      If Urgent please provide reason:			
This is: the proband      a family member      This family member is thought to be: Unaffected      Affected			
Relationship to proband:		Proband genotype:	
Proband name:		Proband date of birth:	

\*Please state if karyotypic and/or phenotypic sex differ from given sex.

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. To set this up, the laboratory will contact you with further information.

Clinical Details	HPLC (%)	HbA:	HbA2:	HbF:	Variant(s)?:
RBC (red blood cell count, x10 <sup>12</sup> /L)			WBC (white blood cell count: x10 <sup>9</sup> /L)		
HGB (haemoglobin, g/L)			Neutrophils (x10 <sup>9</sup> /L)		
MCV (mean corpuscular volume, fl)			Lymphocytes (x10 <sup>9</sup> /L)		
MCH (mean corpuscular haemoglobin, pg)			Platelets (x10 <sup>9</sup> /L)		
RDW (%)			Haptoglobin (g/L)		
Haematocrit (proportion)			Bilirubin (umol/L)		
Reticulocytes (x10 <sup>12</sup> /L&%)			SGOT/AST (Serum glutamic oxaloacetic transaminase, IU/L)		
Ferritin (mg/L)			SGPT/ALT (Serum glutamic pyruvic transaminase, IU/L)		
Transferrin (%)			LDH (Lactate dehydrogenase, IU/L)		
Erythropoietin (EPO, IU/L)			DAT (Direct antiglobulin test/coombs test)		
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	Consanguinity	Yes	No
JAK2 Results (for R405 Erythrocytosis panel)			Chromosome Breakage Results (for Fanconi/NBN)		
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 <b>patient's</b> : full name, date of birth, NHS number (or Hospital Number for non-UK referrals). A <b>minimum</b> of 2 identifiers must be provided or the sample cannot be accepted for testing.			

In submitting this sample the clinician confirms that consent has been obtained for testing and storage.  
Anonymised stored samples may be used for quality control procedures including validation of new genetic tests.

**Please send samples at room temperature by post or courier to:**  
Oxford Genetics Laboratories, Oxford University Hospitals NHS Foundation Trust, The Churchill Hospital, Oxford, OX3 7LE

**Information for Patients**  
Blood samples can be arranged via your GP or the phlebotomy clinic of your local hospital. This form must accompany the sample.  
Following receipt of the sample, laboratory staff are unable to provide information on samples and test results directly to patients or their relatives. Such enquiries should be directed to the referring clinical teams or the GP.