

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					
	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 (Lactate 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	Consanguinity	Yes	No
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			
<p>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.</p>			
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