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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** (For information on genes in each panel, please visit: https://panelapp.genomicsengland.co.uk/panels/ ) | **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** ☐ | **Family testing e.g.R375** ☐ | | | |

**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: | |

## 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, x10^12/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) | | **Platelets** (x10^9/L) | |
| **RDW** (%) | | **Haptoglobin** (g/L) | |
| **Haematocrit** (proportion) | | **Bilirubin** (umol/L) | |
| **Reticulocytes** (x10^12/L&%) | | **SGOT/AST** (Serum glutamic oxaloacetic transaminase, IU/L) | |
| **Ferritin** (mlg/L) | | **SGPT/ALT** (Serum glutamic pyruvic transaminase, IU/L) | |
| **Transferrin** (%) | | **LDH** (Lactacte dehydrogenase, IU/L) | |
| **Erythropoietin** (EPO, IU/L) | | **DAT** (Direct antiglobulin test/coombs test) | |

**Blood smear results**

**Bone marrow smear results** (including date analysed)

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## 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 **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. | | | |

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 Regional Genetics Laboratories, Churchill Hospital, Headington, 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.