

(WRGL use only):

Investigation(s):

W

DNA loc:

In before?

Initials

Referral reason:

Date of receipt:

**WESSEX REGIONAL GENETICS LABORATORY**

Salisbury District Hospital, Salisbury, Wilts. SP2 8BJ

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**Central & South**  
Genomic Laboratory Hub

# Genomic Medicine Service Rare Disease test referral form

## PATIENT DETAILS

Addressograph label

<b>SURNAME</b>	<b>DATE OF BIRTH</b> _ _ : _ _ : _ _ _ _	<b>SEX</b>	Referring consultant
<b>FORENAME</b>	<b>NHS NUMBER</b>		Hospital / Department
Postcode	Hospital number / Genetics number		Clinician's contact number
NHS England / Other NHS / Private (Address for invoicing if not NHS England):	Additional copies to		Clinician's NHS.net email  @nhs.net

Date of collection \_ \_ : \_ \_ : \_ \_ Collected by:

Priority: Routine  Urgent

*State reason for urgency below*

Sample type: EDTA for all referrals  Lithium Heparin if FISH and/or karyotype required

### **Test Selection: please enter the National Genomics Test Directory clinical indication details.**

Please refer to <https://www.england.nhs.uk/publication/national-genomic-test-directories/>

**National Genomics Test Directory test code(s):** R

Date test requested:

**Name of clinical indication(s):**

**Or select one of the following:**  Diagnostic test for known mutation (R240)  Segregation (R375)  Predictive (R242)  Carrier (R244)

**Speciality of referring clinician:**

**Does this patient fulfil the relevant Rare and Inherited Disease eligibility criteria? Yes / No**

### **Referral reason: please provide full clinical details (including any relevant family history).**

Empty box for referral reason details.

**Details of any previous genetic investigations:**



9005



*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.*

## SAMPLE REQUIREMENTS

### ACCEPTANCE CRITERIA

The Genomic Medicine Service came into operation in England in 2020. The Wessex Regional Genetics Laboratory is part of the Central and South GLH Genomic Laboratory Hub (GLH).

The National Genomic Test Directory specifies which tests are funded by NHS England, together with their eligibility and referral criteria: <https://www.england.nhs.uk/publication/national-genomic-test-directories/>. Please note that any test not included in the National Genomic Test Directory will not be centrally funded and will incur a charge. Please contact the laboratory for further information.

Clinical Genetics services are available if required for advice on rare or unusual cases. Please contact Wessex Clinical Genetics Service, Level G, Princess Anne Hospital, Southampton, SO16 5YA, tel: 02381 206170.

### SAMPLE COLLECTION

#### **For all referrals:**

Please collect **5 ml** of blood taken into an **EDTA** tube. **Mix well** by inverting the tube after collection. For infants, a minimum of **1 ml** is required.

#### **For referrals requiring karyotype and/or FISH analysis:**

Please collect **5 ml** of blood into a **lithium heparin** tube. **Mix well** by inverting tube after collection. For infants, a minimum of **1 ml** is required.

### OTHER TISSUES

Other tissue types may be processed under special circumstances; please contact the laboratory to discuss requirements.

**Tumour tissue for DNA analysis** may be sent as formalin-fixed wax block sections.

**Fresh tissue samples** should be collected in **sterile empty containers**. **DO NOT fix these tissues**.

Details on both the referral form and the sample tube should be **complete and legible**. We reserve the right to refuse to process samples with incomplete, illegible or ambiguous patient information.

Any samples in the wrong tube or medium, or which are subject to significant delay in transit, are liable to be rejected. Blood samples from patients who have had a recent white cell blood transfusion may not be suitable for testing.

### SAMPLE DESPATCH AND TRANSPORT

Sample and referral form should be sent **together** in a secure leak-proof package according to UN P650 packaging instructions, to arrive as soon as possible after collection (e.g. by first class post, courier service or hospital transport) and **within 48 hours for optimum results**. Outside packaging should be clearly labelled '**PATHOLOGICAL SAMPLE FOR DELIVERY TO GENETICS**'.

WRGL opening hours are 9 am - 5.30 pm Mon - Fri; please inform the laboratory of any samples likely to arrive over a weekend or bank holiday, or of anything sent by courier which might arrive outside normal working hours. If there is an unavoidable delay between the sample collection and despatch, blood or tissue may be stored in a refrigerator at 4 °C.

**For current information and to download copies of our referral forms and service guides, please refer to our website: [www.wrql.org.uk](http://www.wrql.org.uk)**