(WGLS use only):

W | | | | | | | |

DNA loc:

In before? Initials

Date of receipt:

Investigation(s):

Wessex Genomics Laboratory Service (Salisbury)
University Hospital Southampton NHS Foundation Trust
Postal address: Salisbury District Hospital, Salisbury, SP2 8BJ
Tel.: +44(0) 2381 207100
E-mail: shc-tr.WRGLdutyscientist@nhs.net
Web: www.wrgl.org.uk

UKAS MEDICAL 9005

Genomic Medicine Service Rare Disease test referral form

PATIENT DETAILS

Addressograph label

SURNAME	**DATE OF BIRTH** _ _ : _ _ : _ _ _ _	**SEX**	Referring consultant (full name)
FORENAME	**NHS NUMBER**		Hospital / Department
Patient's postcode	Hospital number / Genetics number		Clinician's speciality
NHS England / Other NHS / Private (Address for invoicing if not NHS England):	Additional copies to (name, address, speciality)		Clinician's contact number / 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: select a test from the National Genomic Test Directory + provide information on the clinical utility of the requested test

N.B.: Both the **R number and the test code name** (clinical indication) must be provided, the referral must fulfil the associated **eligibility criteria**, and the requested test must have a **clinical utility.** Refer to https://www.england.nhs.uk/publication/national-genomic-test-directories/.

National Genomic Test Directory test code(s): R_____

Test code name(s):

State the clinical utility

- ☐ Patient management (determining therapeutic decisions and/or clinical investigations and/or surveillance programme).
- ☐ Patient, parents, or adult relative reproductive decision making.
- ☐ Unaffected relatives are seeking predictive testing.

For molecular tests (single gene / gene panels) - tick one of the following boxes:

- ☐ Diagnostic full screen (enter R code above)
- ☐ Diagnostic targeted test (R240)
- ☐ Predictive (R242)
- ☐ Segregation (R375)
- ☐ Carrier (R244)
- ☐ DNA storage only (R346)

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

DNA will be stored and not tested unless these details are provided

Details of any previous genetic investigations:

NHS University Hospital Southampton NHS Foundation Trust

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.

NHS Central & South Genomic Laboratory Hub

SAMPLE REQUIREMENTS

ACCEPTANCE CRITERIA

The Genomic Medicine Service came into operation in England in 2020. The Wessex Regional Genetics Laboratory and the Molecular Pathology Department at University Hospital Southampton together form the Wessex Genomics Laboratory Service, within the Central and South Genomics 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, https://www.uhs.nhs.uk/departments/genetics.

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.**
Tissue samples requiring culturing should be collected into tissue transport medium.

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 UN3373 shipment classification and packaging instruction P650, 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'.**

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 ^{o}C.

For current information and to download copies of our referral forms and service guides, please refer to our website: www.wrgl.org.uk