

Please note that forms received with missing information (patient/ referrer/ test eligibility/ utility) will not be tested.

| I. Patient Details (Use <u>FOUR</u> patient identifiers <sup>#</sup> ) |                            | II. Referring Clinician Details (All <u>MANDATORY</u> *)  |
|--|----------------------------|---|
| SURNAME <sup>#</sup> :   | FIRST NAME <sup>#</sup> :  | Referring Consultant (Please provide full name)*:   |
| DATE OF BIRTH <sup>#</sup> :   | Sex at Birth:              | NHS.net email (for queries)*:   |
| NHS Number <sup>#</sup> (Mandatory*):                                  | Hospital No/ Your Ref:     | Department*:  |
| Ethnicity:   | GOSH Family ID (If known): | Hospital (No initials, please provide full name/address)*:  |
| Patient Address & Postcode:  |                            | Referring Clinician: I have discussed genomic testing with this patient and have retained a record of discussion (see page 2). Consent is not required for DNA storage. |
| GP Name & Address:   |                            |   |

Consanguineous: ☐ Yes ☐ No

Please select ONE option for report and provide details:

|  |  |
|--|--|
| <input type="checkbox"/> Email (NHS.net email):        |  |
| <input type="checkbox"/> Outreach Portal Submitter ID: |  |
| <input type="checkbox"/> Post:                         |  |

| III. Specimen Details   | If high risk please specify: | Sample Type | Date / Time Collected | Collected By |
|---|------------------------------|-------------|-----------------------|--------------|
| High Risk Specimen?<br><input type="checkbox"/> Yes <input type="checkbox"/> No |                              |             |                       |              |

|   |  |
|---|--|
| <sup>o</sup> Clinical Indication Code: <b>R</b> | <input type="checkbox"/> Urgent <input type="checkbox"/> Routine |
|---|--|

**THIS FORM IS FOR POSTNATAL NON-WHOLE GENOME SEQUENCING GENETIC TESTS (ALTERNATIVE FORMS LISTED OVERLEAF)**

<sup>o</sup> For NHS England referrals, please refer to the National Genomic Test Directory for available tests and eligibility criteria:  
<https://www.england.nhs.uk/publication/national-genomic-test-directories/>

|   |                        |  |
|---|------------------------|--|
| <input type="checkbox"/> DNA storage (no testing)   | EDTA                   | Contact lab to activate testing on stored DNA              |
| <input type="checkbox"/> DNA based testing:<br><input type="checkbox"/> Diagnostic <input type="checkbox"/> Carrier <input type="checkbox"/> Predictive | EDTA                   | (Provide R code, eligibility details and clinical utility) |
| <input type="checkbox"/> Karyotype/FISH testing/Fanconi Anaemia/Bloom/Nijmegen  | Lithium Heparin        | (Provide R code, eligibility details and clinical utility) |
| Rapid testing (infants): <input type="checkbox"/> trisomy 13/18 <input type="checkbox"/> trisomy 21   | EDTA / Lithium Heparin | <input type="checkbox"/> SRY (chromosomal sex)             |

| IV. Reason for referral (Please give details of previous genetic investigations in the family, if any. )       |
|--|
|  |
| (For familial/cascade/follow up testing, provide index patient name and DOB, NHS no. or index patient report.) |

| V. Clinical Utility — Please indicate category of Clinical Utility <u>AND</u> provide details.  |
|---|
| <input type="checkbox"/> Patient management (determining therapeutic decisions and/or clinical investigations and/or surveillance programme)<br><input type="checkbox"/> Patient, parents, or adult relative reproductive decision making<br><input type="checkbox"/> Unaffected relatives are seeking predictive testing |
| Details:  |

| VI. Eligibility — Please provide details to confirm patient meets NHSE eligibility criteria for the test(s) requested. |
|--|
| Details:   |

The North Thames GLH Rare and Inherited Genomic Laboratory incorporates the GOSH Molecular Genetic and Cytogenetics services and the UCLH Neurogenetics service. The GOSH laboratory performs all sample handling, DNA extraction and laboratory tests; analysis and reporting is subsequently carried out by each constituent service depending on the test.

### Discussion with patients and family about genomic testing

- > An appropriate discussion of the genomic test and possible implications should take place according to the Consent and Confidentiality in Genomic Medicine guidelines (available on RCPATH website)
- > The patient should be advised that the sample may be used anonymously for quality assurance, research and training purposes, please advise of any restrictions.

### Instructions

The sample tube and referral card must have three matching identifiers to be accepted. Patient's sex at birth must be indicated on the request form.

- **Sample MUST be labelled with:**

- ☐ Patient's full name (surname and given name)
- ☐ Date of birth and NHS number
- ☐ Referring Hospital Number
- ☐ The date and time sample was taken

- **Blood Samples: Mix samples thoroughly for 2 minutes to prevent clotting**

- ◇ 4mls venous blood in plastic EDTA (pink or lavender) bottles (>1ml from neonates)
- ◇ 2mls venous blood in plastic Lithium Heparin (orange or green) bottles (1-2ml from neonates)

- **Lithium Heparin blood samples must be received in lab within 24 hours (refrigerate overnight at 4°C if necessary).**

**NOTE: The following will lead to REJECTION and may require repeat sampling:**

- Samples in glass bottles
- UNLABELLED Samples
- MISLABELLED Samples

**Please note that blood samples taken after HSCT (bone marrow transplant) or after recent blood transfusion are not suitable for genetic testing.**

**Use alternative Test Order Form for:**

- 1) Whole genome sequencing (WGS) from any sample.
- 2) Free fetal (NIPD) analysis (contact lab in advance)
- 3) RNA Analysis (contact lab in advance)
- 4) Prenatal testing (Chorionic Villus, Amniotic Fluid)

**ANY OTHER SAMPLE: e.g. Buccal swab, Muscle, Urine – TELEPHONE FOR ADVICE**

### Shipping Requirements

Samples coming from outside Great Ormond Street Hospital / Institute of Child Health must be packaged in accordance with **UN PACKING REQUIREMENT PI 650** and clearly labelled '**diagnostic specimen UN3373**'.

### Sample Dispatch/ Storage

Samples can be shipped at room temperature. Samples may be stored at room temperature if taken on the day they are to be sent or refrigerated overnight.

### Address to

North Thames GLH, Rare & Inherited Disease Genomic Laboratory  
Specimen Reception, Level 5 Barclay House, 37 Queen Square,  
London WC1N 3BH

**Opening hours:** Monday to Friday 9.00am to 5.30pm (please ensure samples arrive by 5pm)

**Tel (all enquiries):** 020 7829 8870 / 020 7762 6888

**Email:** [gos-tr.norththamesgenomics@nhs.net](mailto:gos-tr.norththamesgenomics@nhs.net) / [ucl-tr.NHNNgenetics@nhs.net](mailto:ucl-tr.NHNNgenetics@nhs.net)

### For more information

North Thames GLH: <https://norththamesgenomics.nhs.uk>

UCLH Neurogenetics: <https://www.uclh.nhs.uk/our-services/find-service/neurology-and-neurosurgery/neurogenetics/neurogenetics-laboratory>