UKAS MEDICAL 7883

Rare & Inherited Disease Genomic Laboratory
Great Ormond Street Hospital for Children NHS Foundation Trust
Level 5 Barclay House, 37 Queen Square, London WC1N 3BH

NHS North Thames Genomic Laboratory Hub

**Please note that samples received with incomplete forms will not be tested.**

## I. Patient Details (Use FOUR unique patient identifiers)

| | |
|---|---|
| **SURNAME:** | **FIRST NAME:** |
| **DATE OF BIRTH:** | **Sex at Birth:** - |
| **NHS Number:** | **Genetics / Hospital No:** |
| **Ethnicity:** - | |
| **Patient Address & Postcode:** | |
| **GP Name & Address:** | |

| | |
|---|---|
| ☐ NHS ☐ PRIVATE | **Date received (Lab):** |
| **DATE/TIME COLLECTED:** | **SAMPLE TAKEN BY:** |

## II. Referring Clinician Details (All MANDATORY)

| |
|---|
| **Referring Consultant (Please provide full name):** |
| **NHS.net email (for queries):** |
| **Department:** |
| **Hospital (No initials, please provide full name/address):** |

## III. Reporting Preferences — Please select ONE option for report and provide details.

| | |
|---|---|
| ☐ **Email (NHS.net email):** | |
| ☐ **Outreach Portal Submitter ID:** | |
| ☐ **Post:** | |

## IV. Prenatal Tests — NHS patients will routinely be tested in line with NHS England and JCGM prenatal testing guidelines.

☐ **Amniotic Fluid** ☐ **Chorionic Villus Biopsy** ☐ **Fetal blood**
☐ **(If for microarray, please send maternal blood sample (2ml in plastic EDTA tube)**

☐ **QF-PCR ONLY**
☐ **QF-PCR + MICROARRAY (Please give details of scan anomalies below)**
☐ **QF-PCR + TESTING FOR FAMILIAL GENETIC VARIANT (Please give details)**
☐ **QF-PCR + NON-FAMILIAL GENETIC TESTING (Please discuss with lab before sampling)**

| Down's screen risk | Size of NT | Gestation at NT measurement | Gestation at sampling |
|---|---|---|---|
| | | | |

## V. Reason for Referral — Ultrasound anomalies/ previous genetic investigations in family.

*To prevent delayed or failed test reports you must inform the laboratory if pregnancy is from ovum (egg) donation.*

**Consanguineous:** ☐ Yes ☐ No ☐ Unknown ☐ **Do NOT report fetal sex**

## VI. Solid Tissue Tests (Pregnancy/ Fetal Loss)

☐ **Fetal demise after 24 weeks / stillbirth**
☐ **Third or subsequent miscarriage**
☐ **Pregnancy loss with clinical indication of chromosome abnormality (Details MUST be supplied, please use box above)**

*(Note: For any other referral reason, please indicate funding source.)*

**SAMPLE TYPE:** **SEX (IF KNOWN):** -

If products of conception or a placental sample need to be returned for sensitive disposal please ensure that this is clearly indicated on the referral form and that an appropriate consent form is attached.

Samples containing fetal tissue will be returned for sensitive disposal. Any samples not returned will undergo disposal organised by the laboratory.

**In submitting the sample, the clinician confirms that consent for testing and DNA storage has been obtained.**
**Please use the alternative request form (available on our website – see overleaf) for postnatal referrals.**

**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.
- A record of discussion should be retained within the patient's record. A recommended record of discussion is provided on our website.

## INSTRUCTIONS

| Specimen | Quantity | Container and Actions Required | To arrive in lab |
|---|---|---|---|
| Amniotic fluid | 15-20ml* | Universal container.<br>Email laboratory specimen is being sent. | Same day, by 5pm |
| Chorionic villus biopsy | 15-20mg* | Universal container containing 0.9% w/v heparinised saline.<br>Email to confirm dispatch to laboratory. | Same day, by 5pm |
| Skin biopsy (live patient) | Skin punch 2mm$^3$, full thickness | Universal container. Send in sterile 0.9% saline if possible, dry if not.<br>Email to confirm dispatch to laboratory. | Same day, by 5pm |
| Fetuses | N/A | **Fetuses (24+ week gestation by date or scan) will not be accepted by this laboratory.** | N/A |
| Fetal skin biopsy (post-termination/ post-mortem) | 1cm$^3$ skin biopsy, full thickness | Universal container. Send dry if possible but in sterile 0.9% saline if delay anticipated. | Same day |
| Products of conception (cord/ chorionic villi/ cord/ fetal tissue) | N/A | Universal container. Send dry if possible but in sterile 0.9% saline if delay anticipated. | Same day |
| Placental biopsy at cord insertion site | 1cm$^3$ with chorionic villi or placental membrane | Universal container. Send dry if possible but in sterile 0.9% saline if delay anticipated. | Same day |
| Fetal blood samples or Cordocentesis samples from ongoing pregnancies with multiple congenital abnormalities (MCAs) and from TOPs with MCAs or IUDs | 0.25-2ml | Send in an EDTA blood tube.<br>N.B. a maternal blood sample (2ml in an EDTA blood tube) should be sent if the sample was obtained in utero. | Same day |

*Testing may be compromised if a sub-optimal sample is received and may result in a delayed or failed result.

Please email patient details of any pre-booked prenatal sample or skin biopsy for testing (details below). Samples can be stored and sent at room temperature on the same day, or stored in a fridge overnight. Small samples such as skin biopsies should be placed in sterile saline if being sent the following day.

## Sample must be labelled with:

- Patient's full name (surname and given name)
- Date of birth and NHS number
- Referring Hospital Number
- It is desirable to have the date and time sample was taken

## Shipping Requirements

**Samples MUST BE PACKAGED ACCORDING TO UN PACKING REQUIREMENT PI 650, clearly labelled 'diagnostic specimen UN3373' and be sent to the following address:**

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 / ucl-tr.NHNNgenetics@nhs.net

**For details of all referral criteria and policies please see our website: www.norththamesgenomics.nhs.uk**

CLEAR FORM

## FOR LAB USE ONLY