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

UKAS MEDICAL 7883

NHS Great Ormond Street Hospital for Children NHS Foundation Trust

NHS University College London Hospitals NHS Foundation Trust

UKAS MEDICAL 8040

Please note that forms received with missing patient identifiers or no referring clinician/facility may not be tested

GENETIC TEST REQUEST FORM

Lab Ref *(lab use only)*		Date & Time Received *(lab use only)*	

Patient Details - *use four patient identifiers*

First name:

Surname:

DOB:

Sex Assigned at birth:

NHS Number: (mandatory)

Hospital No/Your Ref:

Ethnicity:

GOSH Family ID:

Patient Address:

Postcode:

Referring Clinician Details

Referring Clinician: (full name required)

Contact Number:

NHS.net email: (mandatory)

Department:

Hospital: (full hosp. name & address required)

Submitter ID (Outreach):

Referring Consultant: (if different from referring clinician)

Referring Consultant Email:

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.

NHS Patient (England) ☐

NHS Patient (Wales, Scotland, N.I)* ☐

Private/International Patient* ☐

***Billing Address** (If organisation to be invoiced):

Purchase Order No.

***Patient Email Address** (If Self Funding):

Specimen Details	If high risk please specify:	Sample Type	Date / Time Collected	Collected By
High Risk Specimen? Yes ☐ No ☐				

◊Clinical Indication Code: R

Urgent ☐ **Routine** ☐

Reason for referral: ***(please give clinical details & details of previous genetic investigations in the family, if known)***

◊ 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/

Molecular Genetic Testing *(EDTA, except NIPD, see below)*

- ☐ DNA storage ONLY
- ☐ Diagnostic test
- ☐ Carrier test
- ☐ Predictive test
- ☐ NIPD *(PAXgene or Streck cell stabilising tube 20mls)*

Please provide relevant family history above

☐ **Microarray** *(EDTA only)*

If requesting urgent microarray (e.g. pregnancy, infants <3 months) please send a Lithium Heparin as well

☐ **Cytogenetic follow up** *(EDTA & Lithium Heparin)*

Please give the name & GOSH MRN of index patient above or include copy of index patient report

Rapid testing for infants *(Lithium Heparin & EDTA)*

13/18 ☐ 21 ☐ **Aneuploidy (please specify)**

☐ **Presence of SRY (chromosomal sex)**

Karyotype *(Lithium Heparin)*

- ☐ To exclude Turner Syndrome (Short Stature/Amenorrhea ONLY)
- ☐ To exclude Ring 20 (Epilepsy)
- ☐ Azoospermia/Male Infertility/IVF
- ☐ Premature Ovarian Failure/IVF
- ☐ Sample requested by lab

Chromosome Breakage (not Fragile X) *(Lithium Heparin)*

- ☐ Fanconi Anaemia
- ☐ Bloom Syndrome
- ☐ Other—contact the lab

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 (https://bit.ly/2XkBtMu).
- 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:

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. This will lead to delay in testing: 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.

Contact Lab in advance for:

1) **Free fetal (NIPD) analysis please send 20ml blood in Streck or PAXgene ccfDNA cell-stabilising tubes. Glass Streck tubes for NIPD will be accepted.**
2) **RNA Analysis (PAXgene tubes).**

ANY OTHER SAMPLE e.g. prenatal, buccal swab, muscle, skin biopsy, urine - TELEPHONE FOR ADVICE

Shipping Requirements:	Sample Dispatch/Storage:
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'**	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. Samples in Streck Tubes for Non-Invasive Prenatal Diagnosis/Testing must be stored at room temperature and **NOT** refrigerated.

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: (Cytogenetics & Molecular Genetics): gos-tr.norththamesgenomics@nhs.net
(Neurogenetics): ucl-tr.NHNNgenetics@nhs.net

For more information please see our websites:

North Thames GLH: https://www.norththamesglh.nhs.uk/

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