



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





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

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I. Patient Details (Use <u>FOUR</u> patient identifiers [#])					II. Referring Clinician Details (All MANDATORY*)			
SURNAME [#] :		FIRST NAME [#] :			Referrin	Referring Consultant (Please provide full name)*:		
DATE OF BIRTH#:		Sex at Birth:			•			
NHS Number [#] (Mandatory*):		Hospital No/ Your Ref:			NHS.net email (for queries)*:			
inio italiaci (manadory).		riospitarito, rouriter.			Departm	nent*:		
Ethnicity: GOSH Family ID (If known			(If knov	vn):	Hospital (No initials, please provide full name/address)*:			
Patient Address & Postcode:								
GP Name & Address:					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.			
Consanguineous: Yes No								
Please select <u>ONE</u> option for report and provide details:								
Email (NHS.net email):								
Outreach Portal Submitter ID:								
Post:								
III. Specimen Details If high risk please specify: S			Sa	mple Type	Date / Time Collected		ed	Collected By
High Risk Specimen? Yes No								
°Clinical Indication Code:						u		rgent Routine
THIS FORM IS FOR POSTNATAL NON-WHOLE GENOME SEQUENCING GENETIC TESTS (ALTERNATIVE FORMS LISTED OVERLEAF) Of 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/								
DNA storage (no testing)				EDTA		Contact lab to activate testing on stored DNA		
DNA based testing: Diagnostic Carrier Predictive				EDTA		(Provide R code, eligibility details and clinical utility)		
Karyotype/FISH testing/Fanconi Anaemia/Bloom/ Nijmegen				Lithium Heparin		(Provide R code, eligibility details and clinical utility)		
Rapid testing (infants):	trisomy	13/18 🔲 trisom	ny 21	EDTA / Lithium	Heparin	SRY (chromo	somal 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.								
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								
Details:								
VI. Eligibility — Please	provide d	letails to confirn	n patie	ent meets NHS	E eligibili	ty criteria for th	ne test(s)	requested.
Details:								

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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 RE-QUIREMENT 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 / 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

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