



Great Ormond Street
Hospital for Children
NHS Foundation Trust

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



University College
London Hospitals
NHS Foundation Trust



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

GENETIC TEST REQUEST FORM

Lab Ref <i>(lab use only)</i>	Date Received <i>(lab use only)</i>
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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) <input type="checkbox"/>	*Billing Address (If organisation to be invoiced):	Purchase Order No.
NHS Patient (Wales, Scotland, N.I)* <input type="checkbox"/>		
Private/International Patient* <input type="checkbox"/>	*Patient Email Address (If Self Funding):	

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

◊ Clinical Indication Code: R	Urgent <input type="checkbox"/>	Routine <input type="checkbox"/>
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Reason for referral: <i>(please give clinical details & details of previous genetic investigations in the family, if known)</i>	◊ 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/
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Molecular Genetic Testing <i>(EDTA, except NIPD, see below)</i>	Microarray <i>(EDTA only)</i>	Karyotype <i>(Lithium Heparin)</i>	
	If requesting urgent microarray (e.g. pregnancy, infants <3 months) please send a Lithium Heparin as well	To exclude Turner Syndrome (Short Stature/Amenorrhoea ONLY)	
DNA storage ONLY	Cytogenetic follow up <i>(EDTA & Lithium Heparin)</i>	To exclude Ring 20 (Epilepsy)	Azoospermia/Male Infertility/IVF
Diagnostic test		Premature Ovarian Failure/IVF	Sample requested by lab
Carrier test	Please give the name & GOSH MRN of index patient above or include copy of index patient report	Chromosome Breakage (not Fragile X) <i>(Lithium Heparin)</i>	
Predictive test	Rapid testing for infants <i>(Lithium Heparin & EDTA)</i>	Fanconi Anaemia	
NIPD <i>(PAXgene or Streck cell stabilising tube)</i>	13/18 <input type="checkbox"/> 21 <input type="checkbox"/> Aneuploidy (please specify)	Bloom Syndrome	
Please provide relevant family history above	Presence of SRY (chromosomal sex)	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: Samples in glass bottles will not be accepted
UNLABELLED Samples will not be accepted
MISLABELLED Samples will result in delay**

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

For free fetal (NIPD) analysis please send 20ml blood in Streck or PAXgene ccfDNA cell- stabilizing tubes – Contact Lab in advance

ANY OTHER SAMPLE e.g. Prenatal, Buccal swab, Muscle – 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.

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): gosh.geneticslab@nhs.net / (Neurogenetics): ucl-tr.NHNNgenetics@nhs.net

For more information please see our websites:

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

GOSH Molecular Genetics and Cytogenetics <http://www.labs.gosh.nhs.uk/laboratory-services/genetics>

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