



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 patient identifiers or no referring clinician/facility may not be tested

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GENETIC TE	ST F	REQL	JEST	FORM			Referring Clin	ician I	<u>Details</u>	
Lab Ref (lab use only)	Date Re				R	eferring	Clinician: (full name requ	ıired)		
ido use shiy)	(lab asc	Omy)			C	ontact N	Number:			
Patient Details - use four patient identifiers						NHS.net email: (mandatory)				
First name:	Surname:					Department:				
DOB:	s	Sex Assigned at birth:				Hospital: (full hosp. name & address required)				
NHS Number: (mandatory)	l _u	Hospital No/Your Ref:								
(manuatory)		nospital No/ Your Kei:				Submitter ID (Outreach):				
Ethnicity:	GOSH Family ID:				R	Referring Consultant: (if different from referring clinician)				
Patient Address:						Referring Consultant Email:				
						Referring Clinician: I have discussed genomic testing with				
Postcode:							this patient and have retained a record of discussion (see page 2). Consent is not required for DNA storage.			
NHS Patient (England) *Billing Address (If organisation to be invoiced): Purchase Order No.							Purchase Order No.			
NHS Patient (Wales, Scotland, N.I)*										
Private/International Patient* <u>*Patient Email Address</u> (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	
ferrals, please refer to the National Genomic Test Directory for avaic able tests and eligibilit criteria - https://www.england.nhs.uk/publication/national-								www.england.nhs.uk/		
Molecular Genetic Testing		Microarray (EDTA only)				Karyotype (Lithium Heparin)				
(EDTA, except NIPD, see below)	If requesting urgent microarray (e.g. pregnancy, infan				ints		To exclude Turner Syndrome			
DNA storage ONLY	<3 months) please send a Lithium Heparin as well				1		(Short Stature/Amenorrhea ONLY)			
Diagnostic test	Cytogenetic follow up (EDTA & Lithium Heparin)						To exclude Ring 20 (Epilepsy)		Azoospermia/Male Infertility/IVF	
Carrier test	Please give the name & GOSH MRN of index patient above or include copy of index patient report				nt		Premature Ovarian Failure/IVF		Sample requested by lab	
Predictive test	Rapid	Rapid testing for infants (Lithium Heparin & EDTA)				Chromosome Breakage (not Fragile X) (Lithium Heparin)				
NIPD (PAXgene or Streck cell stabilising tube)	13/18	21	Aneu	ploidy (please specif	y)		Fanconi Anaemia		Bloom Syndrome	
Please provide relevant family history above	Presence of SRY (chromosomal sex)			c)		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:	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	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.
and clearly labelled 'diagnostic specimen UN3373'	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

	North Thames GLH: https://www.norththamesglh.nhs.uk/
For more	GOSH Molecular Genetics and Cytogenetics http://www.labs.gosh.nhs.uk/laboratory-services/genetics
information please see our websites:	UCLH Neurogenetics https://www.uclh.nhs.uk/our-services/find-service/neurology-and-neurosurgery/
	neurogenetics/neurogenetics-laboratory

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