North Thames Genomic Laboratory Hub Rare & Inherited Disease Genomic Laboratory Hub Rare & Inherited Disease Genomic Laboratory Level 5 Barclay House 37 Queen Square, London WC1N 3BH NHS Foundation Trust											
GENETIC TE	ST R		FORM		eleliili	Referring Clinici	ian D	Details	0040		
Lab Ref	Date & T	ime		Re	ferring	Clinician: (full name required	d)				
(lab use only)	Receive	d onlv)		6	ntact N	lumbor					
					ontact Number:						
Patient Details - use four patient identifiers					NHS.net email: (mandatory)						
irst name: Surname:				De	Department:						
DOB:	S	ex Assigned at	birth:	Hospital: (full hosp. name & address required)							
NHS Number: (mandatory)	н	ospital No/You	ur Ref:								
False i site u			_	Su	Submitter ID (Outreach):						
Ethnicity:	Ethnicity: GOSH Fa				eferring Consultant: (if different from referring clinician)						
Patient Address:				Referring Consultant Email:							
Postcode:					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) *Billing Address (If organisation to be invoiced): Purchase Order No.						order No.					
NHS Patient (Wales, Scotland, N.I)*											
Private/International Patient*											
Specimen Details If hig	sh risk p	lease specify:	Sample Type		Date	e / Time Collected	_	Collect	ed By		
High Risk Specimen? Yes No											
[◊] Clinical Indication Code: R					Urgent Routin				Routine		
Reason for referral: (please give clinical details & details of previous genetic investigations in the family, if known) For NHS England re- ferrals, please refer to the National Genomic Test Directory for avail- able tests and eligibility criteria - <u>https://</u> www.england.nhs.uk/ publication/national- genomic-test-directories/											
Molecular Genetic Testing		Microarray (EDTA only)			Karyotype (Lithium Heparin)						
DNA storage ONLY	If reque <3 m	esting urgent micro nonths) please sen	array (e.g. pregnancy, infants d a Lithium Heparin as well		To exclude Turner Syndrome (Short Stature/Amenorrhea ONLY)						
Diagnostic test		Cytogenetic follow up (EDTA & Lithium Hengrin)				To exclude Ring 20 (Epilepsy)		Azoospern Infertility/	nia/Male IVF		
Carrier test	Please give the name & GOSH MRN of index patient above or include copy of index patient report			t		Premature Ovarian Failure/IVF		Sample ree by lab	quested		
Predictive test	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	uploidy (please specify)		Fanconi Anaemia		Bloom Syr	ndrome		
Please provide relevant family history above		Presence of SRY (chromosomal sex))		Other—contact the la	b				

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 (<u>https://bit.ly/2XkBtMu</u>).
- > 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 <u>website</u>.

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.

2) RNA Analysis (PAXgene tubes).

ANY OTHER SAMPLE e.g. Prenatal, Buccal swab, Muscle, 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	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: <u>https://www.norththamesglh.nhs.uk/</u>
For more	GOSH Molecular Genetics and Cytogenetics <u>http://www.labs.gosh.nhs.uk/laboratory-services/genetics</u>
information please	UCLH Neurogenetics https://www.uclh.nhs.uk/our-services/find-service/neurology-and-neurosurgery/
see our websites:	neurogenetics/neurogenetics-laboratory