UKAS MEDICAL HOSpital for NHS Foundation	t Ormond Street ital for Children undation Trust Rare & Inherited Disease Genomic Laboratory Level 5 Barclay House 37 Queen Square, London WC1N 3BH University College London Hospitals NHS Foundation Trust									
GENETIC TE				no i	referrin	Referring Clinic			8040	
	Date Re			Re	eferring	Clinician: (full name require				
(lab use only)	(lab use	only)			_		-,			
						Contact Number:				
Patient Details -	use fo	our patient i	Nł	NHS.net email: (mandatory)						
First name: Surname:			Department:							
DOB: Sex Assign			t birth:							
NHS Number: (mandatory) Hospital No			ur Ref:							
			S			Submitter ID (Outreach):				
Ethnicity:	G	OSH Family ID	SH Family ID:		eferring Consultant: (if different from referring clinician)					
Patient Address:					Referring Consultant Email:					
					Referring Clinician: I have discussed genomic testing with					
Postcode:				1	this patient and have retained a record of discussion (see page 2). Consent is not required for DNA storage.					
NHS Patient (England)		*Billing Add	dress (If organisation t	o be	invoic	ed):		Purchase O	rder No.	
NHS Patient (Wales, Scotland,	N.I)*									
Private/International Patient*		*Patient En	nail Address (If Self Fu	ndin	a):					
		- 1								
Specimen Details If hig	h risk p	lease specify:	Sample Type		Date	/ Time Collected		Collecte	d By	
High Risk Specimen? Yes No										
⁶ Clinical Indication Code:	R							Urgent I	Routine	
Reason for referral: (please give clinical details & details of previous genetic investigations in the family, if known) ^o 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	Microarray (EL		TA only)		Karyotype (Lithium Heparin)					
(EDTA, except NIPD, see below)			oarray (e.g. pregnancy, infants nd a Lithium Heparin as well		To exclude Turner Syndron (Short Stature/Amenorrhe					
DNA storage ONLY	<3 n						Azoospermia/Male			
Diagnostic test		Cytogenetic follow up (EDTA & Lithium Heparin)				To exclude Ring 20 (Epilepsy)		Infertility/I	VF	
Carrier test	Carrier test Please give the name & GOSH MRN of index patien above or include copy of index patient report					Premature Ovarian Failure/IVF		Sample req by lab	uested	
Predictive test Rapid testing for infants (Lithium Heparin & EDTA) Chromosome Breakage (not Fragile X) (Lithium Heparin							oarin)			
NIPD (PAXgene or Streck cell stabilising tube)	13/18	21 Aneu	ıploidy (please specify)		Fanconi Anaemia		Bloom Syn	drome	
Please provide relevant family history above		Presence of S	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 (<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: 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 <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					