REGIONAL CYTOGENETICS LABORATORY
NORTH-EAST THAMES REGIONAL GENETICS SERVICE

PATIENT SURNAME	OTHER NAMES	NHS No	DOB	SEX

HOSPITAL	CONSULTANT	HOSPITAL No
CLINIC / WARD	TEL No FOR LABORATORY ENQUIRIES	GENETICS No

GP NAME, ADDRESS, POSTCODE **or** PCT CODE	**or** PATIENT POSTCODE	DATE/TIME SAMPLE OBTAINED

CLINICAL DETAILS (eg phenotype, obstetric history, family history):

FOR LAB USE ONLY
LAB No

DATE RECEIVED

DETAILS FOR PRENATAL SAMPLES ONLY:

Down's screen risk: ☐ Gestation at sampling: ☐ Gestation at NT measurement: ☐

STANDARD TESTS (PLEASE TICK ONE)

- ☐ BLOOD KARYOTYPE
- ☐ AMNIOTIC FLUID QF-PCR + KARYOTYPE
- ☐ AMNIOTIC FLUID QF-PCR ONLY
- ☐ AMNIOTIC FLUID KARYOTYPE ONLY
- ☐ CVS QF-PCR + KARYOTYPE
- ☐ CVS QF-PCR ONLY
- ☐ CVS KARYOTYPE ONLY

NHS prenatal testing policy is based on how a patient is funded. For details refer to our website: *http://www.ich.ucl.ac.uk/gosh/clinicalservices /Cytogenetics/InformationforHealthProfessionals*

- ☐ TISSUE KARYOTYPE (Live patient only)
- ☐ TISSUE MLPA (For pregnancy loss)
  - ☐ Suspected Triploidy (e.g. Molar pregnancy)

*For **SPECIMEN REQUIREMENTS** please see overleaf*

ADDITIONAL TESTS (PLEASE TICK IF REQUIRED)

- ☐ Ataxia telangiectasia
- ☐ Bloom syndrome
- ☐ Cri du chat syndrome
- ☐ DiGeorge / VCFS syndrome
- ☐ Fanconi anaemia
- ☐ Kallman syndrome
- ☐ Miller-Dieker syndrome
- ☐ Nijmegen syndrome
- ☐ Smith-Magenis syndrome
- ☐ Williams syndrome
- ☐ Wolf-Hirschhorn syndrome
- ☐ X-linked ichthyosis/steroid sulphatase gene deletion
- ☐ 1p36 deletion syndrome
- ☐ Other tests / special instructions (please specify)

SPECIALIST TESTS:

- ☐ Telomere screening (Clinical Genetics and GOSH Neurology referrals only)
- ☐ Microarray screening (Clinical Genetics and GOSH Neurology referrals only)

CONSENT FOR STORAGE OF BLOOD OR DNA (only necessary for Microarray and Telomere screening)

I consent to storage of my blood or DNA for the purpose of:

☐ *Additional laboratory tests which may provide me or my child with a diagnosis of chromosome imbalance.

☐ *Research investigations to improve diagnostic tests for the future, which may not directly benefit me or my child.

*Please delete if you do not wish to consent.

Signature: …… Date……..

SEE OVERLEAF FOR INSTRUCTIONS AND ADDRESS TO WHICH SAMPLES SHOULD BE SENT

SPECIMEN REQUIREMENTS – all containers must be PLASTIC and STERILE

Specimen	Quantity	Container	Must be received in lab
Blood for karyotype +/- routine FISH	5 ml (1ml for neonates)	Lithium heparin	Same day
Blood for telomere screen* plus karyotype	5 ml (1ml for neonates) 5 ml (1ml for neonates)	EDTA Lithium heparin	Same day
*Samples for telomere screen should be referred by a Clinical Geneticist or GOSH Neurologist			
Blood for microarray** plus karyotype	5 ml (1ml for neonates) 5 ml (1ml for neonates)	EDTA Lithium heparin	Same day
**Samples for microarray screen should be referred by a Clinical Geneticist or GOSH Neurologist			
Fetal/cord blood for karyotype	0.5 ml	Lithium heparin	Same day, by 5.00pm
Amniotic fluid	20 ml	Universal container	Same day, by 5.00pm
Chorionic villus biopsy	10 - 50 mg	Universal container containing 0.9% w/v heparinised saline	Same day, by 3.00pm
Skin biopsy (live patient)	Skin punch 2 mm^2, full thickness	Contact laboratory secretary (020 7829 8870) for specimen container and transport medium	Same day, by 3.00pm
Fetal skin biopsy (post termination/post mortem)	1 cm^2 skin biopsy, full thickness	Universal container. Send dry if possible but in sterile 0.9% saline if delay anticipated	Same day
Products of conception	With chorionic villi or fetal tissues if identifiable	Universal container. Send dry if possible but in sterile 0.9% saline if delay anticipated	Same day
Placental biopsy at cord insertion site	1 cm^3 with chorionic villi or placental membrane	Universal container. Send dry if possible but in sterile 0.9% saline if delay anticipated	Same day

Ideally samples should be received in the laboratory the same day as they are taken; if sample transit is delayed, store at 4ºC

Address all samples and correspondence to:
Regional Cytogenetics Laboratory
NE Thames Regional Genetics Service
Great Ormond Street Hospital NHS Trust
Level 5, York House
37 Queen Square, London WC1N 3BH

For general enquiries
(9.00am – 5.00 pm)
Tel: 020 7829 8870
or 020 7762 6918
Fax: 020 7813 8578
Email: cytogenetics@gosh.nhs.uk

FOR LAB USE ONLY: