

**REGIONAL CYTOGENETICS LABORATORY
NORTH-EAST THAMES REGIONAL GENETICS SERVICE**

PATIENT SURNAME	OTHER NAMES	NHS No	DOB	SEX
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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
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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:

<p>STANDARD TESTS (PLEASE TICK ONE)</p> <p><input type="checkbox"/> BLOOD KARYOTYPE</p> <p><input type="checkbox"/> AMNIOTIC FLUID QF-PCR + KARYOTYPE</p> <p><input type="checkbox"/> AMNIOTIC FLUID QF-PCR ONLY</p> <p><input type="checkbox"/> AMNIOTIC FLUID KARYOTYPE ONLY</p> <p><input type="checkbox"/> CVS QF-PCR + KARYOTYPE</p> <p><input type="checkbox"/> CVS QF-PCR ONLY</p> <p><input type="checkbox"/> CVS KARYOTYPE ONLY</p> <p><small>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</small></p> <p><input type="checkbox"/> TISSUE KARYOTYPE (Live patient only)</p> <p><input type="checkbox"/> TISSUE MLPA (For pregnancy loss)</p> <p><input type="checkbox"/> Suspected Triploidy (e.g. Molar pregnancy)</p> <p><i>For SPECIMEN REQUIREMENTS please see overleaf</i></p>	<p>ADDITIONAL TESTS (PLEASE TICK IF REQUIRED)</p> <p><input type="checkbox"/> Ataxia telangiectasia</p> <p><input type="checkbox"/> Bloom syndrome</p> <p><input type="checkbox"/> Cri du chat syndrome</p> <p><input type="checkbox"/> DiGeorge / VCFS syndrome</p> <p><input type="checkbox"/> Fanconi anaemia</p> <p><input type="checkbox"/> Kallman syndrome</p> <p><input type="checkbox"/> Miller-Dieker syndrome</p> <p><input type="checkbox"/> Nijmegen syndrome</p> <p><input type="checkbox"/> Smith-Magenis syndrome</p> <p><input type="checkbox"/> Williams syndrome</p> <p><input type="checkbox"/> Wolf-Hirschhorn syndrome</p> <p><input type="checkbox"/> X-linked ichthyosis/steroid sulphatase gene deletion</p> <p><input type="checkbox"/> 1p36 deletion syndrome</p> <p><input type="checkbox"/> Other tests / special instructions (please specify)</p>
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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 4°C

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: