

## LABORATORY DETAILS

The laboratory offers a comprehensive conventional cytogenetic and molecular cytogenetic diagnostic service for prenatal amniotic fluid and chorionic villus samples, pre- and postnatal blood samples, solid tissues, tumours and haematological malignancies. The laboratory is situated on the 2<sup>nd</sup> Floor of the Liverpool Women's Hospital.

### Postal Address

Cheshire & Merseyside Regional Cytogenetics Laboratory,  
Liverpool Women's Hospital,  
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Liverpool, L8 7SS.

**Tel:** 0151 702 4229

**Fax:** 0151 702 4230

**Web:** [www.lwh.me.uk/html/cytogenetics.php](http://www.lwh.me.uk/html/cytogenetics.php)

### Laboratory Working Hours

9.00 a.m. – 5.30 p.m. Monday - Friday

### Consultant Cytogeneticist / Directorate Lead

Mrs Angela Douglas

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### Consultant Cytogeneticist / Head of Laboratory

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### Deputy Head of Laboratory

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### Quality Manager

Mr Alan Clark

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### Section Heads/ Contacts:

**Prenatal** Michael Groom 0151 702 4234

**Postnatal Blood** Magda Ainscough 0151 702 4234

**Solid Tissue** Joan Getty 0151 702 4229

**Oncology** Una Maye 0151 702 4204

**FISH** Kevin Pearson 0151 702 4229

### Feedback & Enquiries

Should you have any comments, suggestions, cause for concern or complaint about the service you receive from the laboratory, please contact the Head of the Laboratory or the Quality Manager.

## PRENATAL DIAGNOSIS (PND) SERVICES

The laboratory provides a comprehensive conventional and molecular cytogenetic service on the following samples:

- Amniotic Fluid Samples
- Chorionic Villus Biopsies
- Fetal Blood Samples

### AMNIOTIC FLUID (AF) SAMPLES

The laboratory offers a rapid screening service by QF-PCR for the chromosomes 13, 18, 21, X & Y followed by a comprehensive culture & karyotype service detecting most structural anomalies undetectable by the rapid technology.

### Amniotic Fluid Sample Requirements

15 –20 ml Amniotic Fluid in a sterile plastic container.

### Limitations of Test

Heavily bloodstained samples are unsuitable for the rapid screening service

### CHORIONIC VILLUS (CV) SAMPLES

The laboratory offers a direct, provisional rapid aneuploidy (FISH) screening service on appropriate samples with a more comprehensive conventional cytogenetic analysis performed on cultured material.

### Chorionic Villus Sample Requirements\*

Cytogenetic referrals	5-10 mg of Villus tissue
Cytogenetics & Molecular	15 mg of Villus tissue
Biochemical referrals	>20 mg of Villus tissue

*\*The quoted amounts refer to sorted material after the biopsy is dissected out from maternal tissue and decidua.*

**N.B.** All biopsies should be placed in CVS transport medium (available from the laboratory).

### Prenatal Referral Policy

- Serum Screen Positive
- Advanced Maternal Age
- Fetal Anomaly on Ultrasound Scan
- Previous Pregnancy with Chromosome Rearrangement
- Familial Chromosome Rearrangement
- Maternal Anxiety
- Single Gene Disorders

Please discuss with senior member of staff for other referrals.

### FETAL BLOOD SAMPLES (FBS)

The laboratory offers a service for fetal blood samples following appropriate consultation and agreement with a senior member of staff.

### Fetal Blood Sample Requirements

1-2 ml blood, in a ***lithium heparin container***.

## FBS Referral Policy

- Abnormal Ultrasound scan
- Abnormal on AF / CVS Analysis

## POSTNATAL SERVICES

The Laboratory offers a full range of culture / karyotyping, molecular cytogenetic (FISH) & microarray CGH tests which include analysis of microdeletion syndromes and cryptic imbalances.

### Postnatal Blood Sample Requirements

- Neonatal/ Child 1-2 ml
- Adult 5ml

Peripheral blood samples for cytogenetic testing must be collected into ***Lithium Heparin containers***.

### Postnatal Bloods Referral Policy

The Laboratory will accept all appropriate referrals, the major categories being:

- IUGR / Stillbirth
- Congenital Abnormality/ Dysmorphism
- Developmental Delay / Learning Difficulties
- Delayed Puberty
- Sub Fertility / Recurrent Pregnancy Loss
- Family History of Abnormality
- Epilepsy Screening(?)
- Breakage Syndromes (Please consult laboratory for details).

## ONCOLOGY SERVICE

The laboratory offers a wide range of conventional and molecular cytogenetic tests for the investigation of haematological malignancies, including:

- Chronic / Acute Myeloid Leukaemias
- Lymphomas
- Bone Marrow Transplant Monitoring
- Chronic Lymphocytic Leukaemia (FISH only)
- Acute Lymphocytic Leukaemia

### Oncology Sample Requirements

Bone Marrow Aspirate: 1-2 ml in BM transport medium (Available from the laboratory).

Peripheral Blood: 1-5 ml **collected in *Lithium Heparin container***.

(& CLL FISH Testing)  
Lymph Node: Biopsy placed in BM transport medium (Available from the laboratory).

Molecular Genetic: 1.5 ml **collected in EDTA Tests**

## SOLID TISSUES SERVICE

The laboratory provides a tissue culture service for karyotyping, molecular cytogenetics (FISH) testing and storage of cell lines in a liquid nitrogen cell bank.

### Solid Tissue Sample Requirements

Solid tissue samples must be sent in solid tissue transport medium (available from the laboratory). A needle punch biopsy sample is sufficient from live patients. See *Table 1* for further details.

**Table 1. N.B. Tissue samples below should be at least 1 cm<sup>3</sup>.**

Referral Type	Tissues Required - At least one of:
Stillborn / IUD (Within 24hrs of event)	a) Cord blood in Lithium Heparin b) Cardiac blood in Lithium Heparin c) Full-depth skin biopsy and muscle <b>PLUS</b> Placental tissue from cord insertion site with membranes attached.
Stillborn / IUD (Greater than 24hrs of event)	Placental tissue from cord insertion site with membranes attached. <b>PLUS</b> Full-depth skin & muscle biopsy, if possible.
TOP for fetal Abnormality on Ultrasound scan	a) 10-20 ml Amniotic Fluid. b) Placental tissue as above c) Cord Blood in Lithium Heparin d) Cardiac Blood in Lithium Heparin e) Full-depth Skin & Muscle biopsy

### Referral Policy

The Laboratory will accept all appropriate referrals, the major categories are:

- Recurrent Pregnancy Loss / IUD / Stillbirth
- Fetal Abnormality / Confirmation of Prenatal Investigation
- Metabolic Disorder for Biochemical / DNA Assay (Please consult with laboratory for further details).
- Chromosomal Mosaicism investigation
- Tumours (currently specified types by prior agreement)
- Sperm samples (currently specified types by prior agreement)

### FLUORESCENCE IN SITU HYBRIDISATION (FISH) AND MICROARRAY SERVICES

The laboratory employs many DNA FISH probes and has introduced microarray technology to support and extend the scope of its testing repertoire. For further details please contact a senior member of staff.

### Sample requirements

As described under previous headings or slide preparations.

### OTHER INFORMATION

#### Consent

All genetic testing requires consent. The laboratory assumes that the provision of a tissue sample implies that consent has been obtained by the referring clinician. Clinical material is retained after reporting for quality assurance purposes ONLY, unless the request card indicates that permission for this is denied. No additional testing is conducted without clinical request.

### Sample Handling and Storage

All samples should be sent directly to the laboratory. If this is not possible, then they should be stored in a secure refrigerator at +4°C and sent as soon as possible, to arrive at the laboratory within 24hrs of collection.

### Packaging & Transportation

All samples should be labelled with the Patient's Name, Date of Birth, Unit No. and the Date of Collection and be accompanied by a FULLY completed request card (Available from the laboratory or downloadable from the laboratory web site: [www.lwh.org.uk/clinical\\_services/genetics/cytogenetics](http://www.lwh.org.uk/clinical_services/genetics/cytogenetics)). The sample should be placed in a sealed specimen bag in such a way as to maintain patient confidentiality and to prevent spillage and/ or contamination to couriers and porters. Samples sent through the post should be packaged in accordance with current Post Office regulations.

**Please Note:** The viability of samples collected into the wrong containers, stored at extreme temperatures or delayed in transit is impaired and they are prone to failure in culture. Blood Oncology samples with a cell concentration <10<sup>6</sup> / ml and heavily bloodstained bone marrow aspirates are less likely to give successful results.

Because of the nature of the clinical procedure, where possible culture of sub-optimal samples will be attempted however, they are prone to failure. Heavily clotted samples and fixed tissue samples are unsuitable for culture. Samples that are unlabelled and blood that is heavily clotted or collected into EDTA (other than precious samples from babies and for CLL FISH testing) will NOT be processed.

### Turnaround Times

The laboratory aims to meet the following reporting times:

**Table 2.**

Tissue	Target Turnaround Time
<b>Prenatal Samples</b>	
<b>Amniotic Fluid</b>	
• Rapid Screen	4 days
• Final Result	17 days
<b>Chorionic Villus</b>	
• Provisional Direct	4 days
• Final	17 days
• Fetal Blood	10 days
<b>Postnatal Blood</b>	
• Urgent	10 days
• Routine	28 days
<b>Blood Oncology</b>	
• Urgent	10 days
• Routine	21 days
• FISH Only	7 days
<b>Solid Tissue</b>	28 days



# INFORMATION FOR USERS

## CHESHIRE & MERSEYSIDE REGIONAL CYTOGENETICS LABORATORY

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