

## LABORATORY DETAILS

The laboratory is situated on the 2<sup>nd</sup> Floor of the Liverpool Women's Hospital and is part of the Medical Genetics directorate that also includes the Regional Cytogenetics laboratory and the Regional Clinical Genetics Service (based at the Royal Liverpool Children's Hospital, Alder Hey).

### Postal Address

Regional Molecular Genetics Laboratory,  
Liverpool Women's NHS Foundation Trust,  
Crown Street,  
Liverpool L8 7SS.

**Tel: 0151 702 4228**      **Fax: 0151 702 4226**

**Web:** [www.lwh.me.uk/html/molecular\\_gen.php](http://www.lwh.me.uk/html/molecular_gen.php)

### Laboratory Working Hours

9.00 a.m. – 5.30 p.m. Monday - Friday.  
(No out of hours service is provided).

### Head of Laboratory

Mr Roger Mountford  
Tel: 0151 702 4219  
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### Deputy Head

Dr David Gokhale  
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### Other Scientific staff

Ms Julie Sibbring	Tel 0151 702 4225
Ms Emma McCarthy	Tel 0151 702 4228
Ms Vicky Stinton	Tel 0151 702 4011
Dr Ciaran McAnulty	Tel 0151 702 4011
Mrs Diane Cairns	Tel 0151 702 4228
Dr Andrew Purvis	Tel 0151 702 4011
Ms Lindsey Bradley	Tel 0151 702 4011

E-mail: [DNA.Liverpool@nhs.net](mailto:DNA.Liverpool@nhs.net)

### Quality Manager

Mr Alan Clark  
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### Voice Mail

The Trust voice mail operates on all external lines. When diverted to voice mail, please leave a message and someone from the laboratory will get back to you as soon as possible.

### Complaints

Should you have any comments, suggestions, cause for concern or complaints about the service you receive from the laboratory, please contact the Head / Deputy Head of the Laboratory or the Quality Manager. (Contact details above).

## CURRENT SERVICES

The laboratory uses DNA analysis techniques to carry out testing for a wide range of genetic disorders. A list of current services is given in table 1 on the reverse of this leaflet.

The types of investigation include:

- Confirmation / exclusion of a diagnosis
- Carrier testing and risk assessment in families with a known genetic disorder
- Presymptomatic prediction in individuals at risk of a late-onset genetic disorder
- Prenatal diagnosis of genetic conditions where appropriate.

The laboratory offers testing for a series of "core" disorders plus a set of specialist services for which samples are received on a supra-regional or national basis.

The laboratory is a member of the UK Genetic Testing Network (UKGTN) and will forward DNA samples where appropriate to other UK laboratories for a large range of single gene disorders.

Full details of services available through UKGTN are available at [www.ukgt.nhs.uk](http://www.ukgt.nhs.uk) or by contacting the laboratory. Details of services for rare disorders currently not available in the UK can be found on the web sites [www.orpha.net](http://www.orpha.net) and [www.geneclinics.org](http://www.geneclinics.org) or by contacting the laboratory.

### Gate keeping & Funding of UKGTN Samples

Access to many tests offered in other UK laboratories is restricted to specific referring specialties / clinicians. Please contact the laboratory for details.

The costs of any tests referred to UKGTN centres or abroad will be recharged to the referring Trust.

### Turnaround times

Target turnaround times are given in table 1.

The laboratory works most efficiently by batching up samples and therefore aims to report all non-urgent samples within the stated times.

Urgent work, including all prenatal diagnoses will be completed as soon as possible depending on the techniques required.

Reporting times are typically less than one week for such cases. Please contact the laboratory if a referral is urgent.

### Advice Service

The Head of Laboratory and scientific staff will provide advice on scientific and technical issues. For advice on clinical and counselling issues, please contact the Clinical Genetics Service (0151 702 5000)

## Consent

All genetic testing requires consent. The laboratory assumes that the provision of a sample implies that consent has been obtained by the referring clinician.

### DNA storage

DNA from all referrals is currently retained for quality assurance purposes ONLY, unless the request card indicates that permission for storage is denied. Consent is not required for storage for these purposes but it is considered good practice for the referring clinician to inform the patient of this practice.

The laboratory will also store DNA from patients where no specific genetic test is currently available / required.

### Sample types

The preferred sample type is 3mls of **EDTA blood** (1-2 mls from small children).

**Note: This is not the same sample type as for Cytogenetic analysis, which requires Lithium Heparin samples. If in doubt about sample types for a particular test please contact the laboratory.**

DNA results can also be obtained from mouthwash/cheek scrape samples (in normal saline or 4% sucrose or Oragene DNA collection system, details available from the laboratory) and from fresh or frozen tissue samples.

It is also possible to obtain limited results for some assays from blood spots or paraffin embedded fixed tissue samples (please contact the laboratory for further details).

Prenatal diagnosis is usually carried out on chorionic villus samples but amniotic fluid or fetal blood can be used where necessary.

### 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.

### Packaging & Transportation

All samples should be labelled with the Patient's Name, Date of Birth, Postcode, NHS number, 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 - see page 2) including details of family history where relevant.

The sample should be placed in a sealed specimen bag in such a way as to maintain patient confidentiality and to prevent spillage and contamination of couriers and porters. Samples sent through the post should be packaged in accordance with PI 650 and current UN3373 regulations. (See Laboratory web site for further details).

### Inappropriate samples

Clotted blood samples or samples that are inadequately labelled or packaged will not be accepted by the laboratory.

**Table 1 Current services**

Disorder + notes	TTT
<b>Adrenoleukodystrophy (X-linked)</b> <i>Diagnostic testing for mutations in ABCD1 gene</i> <i>Familial mutation studies</i>	8 weeks 2 weeks
<b>Aneuploidy screening (PCR based)</b> <i>Screening for trisomy 13, 18 &amp; 21 plus sex chromosome aneuploidies</i>	3 days
<b>Breast Cancer (familial)</b> <i>BRCA1 &amp; 2: Familial mutation studies only. Referrals only accepted from Clinical Genetics Department</i>	2 weeks
<b>CADASIL</b> <i>Diagnostic testing for mutations in Notch3 gene</i> <i>Familial mutation studies</i>	8 weeks 2 weeks
<b>Cystic Fibrosis</b> <i>Diagnostic testing and carrier detection for 28 common CFTR mutations. Other rarer mutations by specific request</i>	2 weeks
<b>DRPLA</b> <i>Testing for CAG expansion mutation</i>	4 weeks
<b>Duchenne/Becker Muscular Dystrophy</b> <i>Diagnostic testing and carrier detection for deletion / duplication mutation plus linkage analysis where necessary</i>	4 weeks
<b>Dystonia</b> <i>Diagnostic testing for DYT1 mutation in Torsion Dystonia (Service for Myoclonic Dystonia under development)</i>	4 weeks (enquire)
<b>Familial Adenomatous Polyposis</b> <i>APC gene familial mutation studies only. Referrals only accepted from Clinical Genetics Department</i>	2 weeks
<b>Fragile X syndrome</b> <i>Diagnostic testing and carrier detection for Frax A mutation. Frax E testing if specifically requested.</i>	4 weeks
<b>Friedreich Ataxia</b> <i>Diagnostic &amp; carrier testing for the GAA expansion mutation. Point mutation analysis in FXN gene</i>	4 weeks 8 weeks
<b>Gilbert syndrome</b> <i>Testing for TATAA box mutation in UGT1A1 gene.</i>	4 weeks
<b>Haemato-oncology</b> <i>Testing for V617F mutation in JAK2 gene. (JAK2 Exon 12 mutations : contact laboratory)</i> <i>Testing for ITD mutation in FLT3 gene</i> <i>Testing for B cell IgH clonal rearrangements by PCR (T cell clonal rearrangement:testing: contact laboratory)</i>	4 weeks 4 weeks 4 weeks
<b>Haemochromatosis</b> <i>Testing for C282Y mutation and H63D if indicated</i>	4 weeks
<b>Hearing Loss (Non syndromic)</b> <i>Diagnostic testing for Connexin 26 and 30 gene mutations</i> <i>Familial mutation studies for Connexin 26 gene</i> <i>Diagnostic testing for Mitochondrial mutation m.1555A&gt;G</i>	8 weeks 2 weeks 4 weeks
<b>HMSN / HNPP</b> <i>Testing for common 17p duplication / deletion.</i> <i>Testing for mutations in PMP22, MPZ, GJB1 and Mitofusin genes. Familial mutation studies for the above genes</i>	4 weeks 8 weeks 2 weeks
<b>Hereditary Non-Polyposis Colon Cancer (HNPCC)</b> <i>Referrals only accepted from Clinical Genetics Department</i>	Enquire

TTT = Target Turn around time

Disorder + notes	TTT
<b>Huntington disease</b> <i>Testing for CAG expansion mutation. Pre-symptomatic</i> <i>Referrals only accepted from Clinical Genetics Department</i>	4 weeks
<b>Kennedy Syndrome (SBMA)</b> <i>Testing for CAG expansion mutation</i>	4 weeks
<b>Leber's Hereditary Optic Neuropathy (LHON)</b> <i>Diagnostic testing for 3 common mutations</i>	4 weeks
<b>Medium chain acyl dehydrogenase deficiency (MCADD)</b> <i>Testing for common c.985A&gt;G mutation only</i>	4 weeks
<b>Mitochondrial disease</b> <i>Testing for common point mutations associated with MELAS, MERRF, NARP. Testing for mitochondrial genomic rearrangements (muscle biopsy preferred)</i>	4 weeks
<b>Myotonic Dystrophy (type 1)</b> <i>Testing for CTG expansion mutation</i>	4 weeks
<b>Myotubular Myopathy (X linked)</b> <i>Diagnostic testing for mutations in MTM1 gene</i> <i>Familial mutation studies</i>	8 weeks 2 weeks
<b>Nail Patella Syndrome</b> <i>Diagnostic testing for mutations in LMX1B gene</i> <i>Familial mutation studies</i>	8 weeks 2 weeks
<b>Neuroferritinopathy</b> <i>Testing for c.408insA mutation in FTL gene</i>	4 weeks
<b>Pancreatitis (Hereditary &amp; Idiopathic)</b> <i>Testing for PRSS1 mutations</i> <i>Testing for CFTR and SPINK1(N34S) mutations</i>	8 weeks 4 weeks
<b>Peutz-Jegher syndrome</b> <i>Diagnostic testing for mutations in STK11 gene</i> <i>Familial mutation studies</i>	8 weeks 2 weeks
<b>PKAN (Hallervorden-Spatz)</b> <i>Diagnostic testing for mutations in PANK2 gene</i> <i>Familial mutation studies</i>	8 weeks 2 weeks
<b>Prader-Willi / Angelman syndromes</b> <i>Diagnostic testing using methylation studies.</i> <i>Family studies on positive cases by microsatellite analysis</i>	4 weeks
<b>Spinal Muscular Atrophy</b> <i>Diagnostic and carrier testing for SMN1 deletions.</i>	4 weeks
<b>Spinal Cerebella Ataxia (types 1, 2, 3, 6, 7, 17)</b> <i>Testing for SCA 1,2,3,6 CAG expansion mutations.</i> <i>SCA7 and SCA 17 testing by specific request only.</i>	4 weeks
<b>Telomeric rearrangements</b> <i>Testing for loss/gain of sub-telomeric regions of all chromosomes by MLPA analysis</i>	4 weeks
<b>Uniparental disomy studies</b> <i>Testing for UPD7 and UPD14. Parental samples required</i>	4 weeks
<b>Uveal Melanoma</b> <i>Testing for rearrangements of chromosomes 1p, 3, 6 and 8 by MLPA analysis</i>	4 weeks
<b>Zygosity / Paternity analysis</b> <i>Zygosity and Paternity testing only available for clinical not social/legal cases</i>	4 weeks



# INFORMATION FOR USERS

## REGIONAL MOLECULAR GENETICS LABORATORY

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