Table 1 Current services

Disorder + notes	TTT
Adrenoleukodystrophy (X-linked) *Diagnostic index case* *Family studies*	12 weeks 4 weeks
Breast Cancer (familial) *Referrals only accepted from Clinical Genetics Department*	Please enquire
CADASIL *Diagnostic index case* *Presymptomatic test*	8 weeks 3 weeks
Cystic Fibrosis *Diagnostic testing, and carrier detection for 29 common mutations.* *Other rarer mutations by specific request*	3 weeks
DRPLA *Diagnostic and presymptomatic testing for CAG expansion mutation*	4 weeks
Duchenne/Becker Muscular Dystrophy *Diagnostic testing and carrier detection for deletion / duplication mutation plus linkage analysis where necessary*	6 weeks
Factor XI deficiency *Testing for known mutations only*	4 weeks
Familial Adenomatous Polyposis *Diagnostic testing in exon 15 only. Further analysis available though UKGTN* *Presymptomatic testing through Clinical Genetics Dept only*	8 weeks
Friedreich Ataxia *Diagnostic testing and carrier detection for the GAA expansion mutation.* *Point mutation analysis*	4 weeks 12 weeks
Fragile X syndrome *Diagnostic testing and carrier detection for Frax A mutation.* *Frax E testing if specifically requested.*	6 weeks
Gilbert's syndrome *Diagnostic testing for TATAA box mutation.*	4 weeks
Haemochromatosis *Diagnostic testing and family studies for the C282Y mutation.* *H63D mutation by request only.*	4 weeks
HMSN / HNPP *Diagnostic and presymptomatic testing for common 17p duplication / deletion.*	4 weeks
Huntington disease *Diagnostic testing for CAG expansion mutation.* *Presymptomatic testing only accepted from the Clinical Genetics Department*	4 weeks 3 weeks

TTT= Target Turn around time

Disorder + notes	TTT
Leber's Hereditary Optic Neuropathy *Diagnostic testing for 3 common mutations*	6 weeks
MCAD *Diagnostic testing and carrier detection for K329E common mutation*	4 weeks
Mitochondrial mutations *Diagnostic testing for common mutations associated with MELAS, MERRF, NARP.* *Diagnostic testing for mitochondrial rearrangements (muscle biopsy preferred)*	6 weeks
Myotonic Dystrophy *Diagnostic and presymptomatic testing for CTG expansion mutation*	4 weeks
Nail Patella Syndrome *Diagnostic index case* *Family studies*	12 weeks 4 weeks
Norries Disease *Diagnostic index case* *Family studies*	8 weeks 4 weeks
Neuroferritinopathy *Diagnostic and presymptomatic testing for 408insA mutation*	6 weeks
Pancreatitis (Hereditary) *Diagnostic and presymptomatic testing for 3 common PRSS1 mutations*	6 weeks
Peutz-Jegher syndrome *Diagnostic testing and family studies*	Please enquire
Prader Willi/Angelman syndrome *Diagnostic testing using methylation studies.* *Family studies on positive cases.*	4 weeks
Sexing *Analysis of AMXY and SRY sequences. For gender identification (enquire for details)*	4 weeks
Silver-Russell syndrome *Testing for chromosome 7 uni-parental disomy.* *Parental samples required*	6 weeks
Sorsby's Fundus Dystrophy *Diagnostic and presymptomatic testing for common S181C mutation only*	4 weeks
Spinal Muscular Atrophy *Diagnostic and carrier testing for SMN1 deletions.*	4 weeks
Spinal Cerebella Ataxia (types 1, 2, 3, 6, 7) *Diagnostic and presymptomatic testing for SCA 1,2,3,6 mutations.* *SCA7 testing by specific request only.*	4 weeks 4 weeks
Zygosity/Paternity analysis *Zygosity and Paternity testing only available for clinical not social/legal cases*	4 weeks

CPA Accredited

Liverpool Women's Hospital NHS Trust

INFORMATION FOR USERS

CHESHIRE & MERSEYSIDE REGIONAL MOLECULAR GENETICS LABORATORY

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Approved: RM

LABORATORY DETAILS

The laboratory is situated on the 2nd 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 (currently based at Royal Liverpool Children's Hospital Alder Hey).

Postal Address

Regional Molecular Genetics Laboratory,
Liverpool Women's Hospital,
Crown Street,
Liverpool L8 7SS.

Tel: 0151 702 4228
Fax: 0151 702 4226
Web: www.lwh.org.uk/clinical_services/genetics/molecular

Laboratory Working Hours

9.00 a.m. - 5.30 p.m. Monday - Friday

Head of Laboratory

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Deputy Head

Dr David Bourn (until May 2004) Tel: 0151 702 4011
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Quality Manager

Mr Alan Clark Tel: 0151 702 4234
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Other Scientific staff

Ms Julie Sibbring	Tel 0151 702 4225
Ms Emma McCarthy	Tel 0151 702 4225
Dr Ciaron McAnulty	Tel 0151 702 4225
Mrs Diane Cairns	Tel 0151 702 4011

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 below.

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 disorder tests.

Full details of services available through UKGTN are available at the Clinical Molecular Genetics Web site (www.cmgs.org) 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.eddnal.org (European laboratories) and www.geneclinics.org (US laboratories) or by contacting the laboratory.

Funding

The laboratory will endeavour to meet the cost of tests sent to other laboratories through UKGTN but these tests can be very expensive and the laboratory reserves the right not to send inappropriate referrals.
For some disorders, samples will only be sent if the family have been seen by the Clinical Genetics department (the laboratory will notify users in such cases)

Turn around times

Target turn around 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.

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. This includes consent to store the DNA processed from the sample.

DNA storage

DNA from all referrals is currently retained for quality assurance purposes ONLY, unless the request card indicates that permission for this is denied.
The laboratory will also store DNA from patients where no specific genetic test is currently available / required.

Sample types

The preferred sample type is 5mls of EDTA blood (1-2 mls from small children).

Note: This is not the same sample type as the Cytogenetics laboratory, 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 (protocol 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 preferably carried out on chorionic villus samples but cultured 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, 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 current Post Office regulations.

Inappropriate samples

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