

East Anglian Medical Genetics Service

**INFORMATION
FOR
USERS**

Molecular Genetics Laboratory

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**Genetics Laboratories
Molecular Genetics, Box 158
Level 6, Addenbrooke's Treatment Centre
Addenbrooke's Hospital
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www.addenbrookes.org.uk/moleculargenetics

October 2007

Laboratory Working Hours

8.30am-5.30pm Monday-Friday

Acting Head of Laboratory: Becky Treacy
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Genetics Laboratories Director: Dr Jo Whittaker
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Types of investigation

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

Testing is available for the disorders listed overleaf. For some of these disorders, the sample will be sent to another UK laboratory.

All test requests may be reviewed by the genetics service. The laboratory reserves the right not to test samples or send referrals to other laboratories.

Consent

All genetic testing requires consent. It is the responsibility of referring clinicians to ensure appropriate consent has been obtained. DNA is stored from all patients unless consent for this is expressly denied.

DNA storage

DNA from all referrals is currently retained for quality assurance purposes and may be used anonymously for the development of new tests.

Sample types

Adults: 5ml blood in EDTA
Children: 1-5ml blood in EDTA

If testing of other sample types is required please contact the laboratory, prior to sending.

Sample handling

All patient samples must be labelled with:

- **Name**
- **Date of birth**
- **Hospital/NHS number**

Samples should be accompanied by a FULLY completed request card.

Please include details of test, family history, patient address & postcode, GP, referring clinician and unit. Inappropriately labelled samples will not be accepted.

If the sample cannot be sent directly to the laboratory, please store at +4°C and send at the earliest opportunity.

Samples for Prenatal Diagnosis

Please contact the laboratory prior to sending

Reporting Times

Target turn around times are given overleaf. Please contact the laboratory if a test is urgent. Please note: we do NOT report results via the telephone.

Paternity testing

Paternity testing is offered by:

Cellmark Diagnostics	01235 528000
Northgene Ltd	0191 245 3804
Doctors Laboratory	020 7460 4800

Other departments

Clinical Genetics	01223 216446
Cytogenetics (chromosome analysis):	01223 348711

Tests offered:**Reporting time codes 1-6:**

1	<i>Mutation screening of gene(s)</i>	<i>8 weeks</i>
2	<i>Testing for a known mutation</i>	<i>2 weeks</i>
3	<i>Direct test (repeat expansion, dosage)</i>	<i>2 weeks</i>
3a	<i>Direct test (Southern blot analysis)</i>	<i>8 weeks</i>
4	<i>Family studies (linkage analysis)</i>	<i>2 weeks</i>
5	<i>Methylation studies</i>	<i>2 weeks</i>
6	<i>Samples sent to another laboratory</i>	<i>2-8 weeks</i>

Allan Herndon Dudley syndrome (AHDS)*	1, 2
Alzheimer disease (familial cases only)	6
Androgen insensitivity syndrome (AIS)*	1, 2
Angelman syndrome (AS)	4, 5
Breast/Ovarian cancer (familial cases only) Referrals only accepted from Clinical Genetics	1, 2, 6
Congenital adrenal hyperplasia (21-OH)	6
Cystic fibrosis (CF) Delta F508 and 28 common mutations	2
Urgent newborn screening (IRT)	5 days
Rare mutation screening by specific request	6
Donohue syndrome	1, 2
Dentatorubral-Pallidoluysian atrophy (DRPLA)	3
Duchenne/Becker muscular dystrophy Extended mutation screening by specific request	3
Dystonia (DYT1)	6
Emery Dreifuss muscular dystrophy, (X-linked)*	1, 2
Fabry disease	6
Facio-scapula-humeral muscular dystrophy	6
Familial dysalbuminaemic hyperthyroxinaemia R218H, R218P, L66P common albumin mutations:	2
Focal & segmental glomerulosclerosis (FSGS)*	1, 2

Tests offered:

Fragile X syndrome (FraX) FRAX-E testing by specific request	3a
Friedreich ataxia (FRDA) Rare mutation screening by specific request	3
Frontotemporal dementia with Parkinsonism* MAPT and PGRN genes	1, 2
Gaucher disease	6
Gitelman syndrome (GMS)*	1, 2
Glucocorticoid-remediable aldosteronism (GRA)	3
Gonadal dysgenesis (SRY gene)	1, 2
Haemochromatosis (HFE) C282Y and H63D common mutations	2
Haemophilia A and B (Factor VIII & IX deficiency)	6
Hereditary fructose intolerance (HFI)	1, 2
Hereditary non-polyposis colon cancer (HNPCC)	1, 2
Hereditary motor & sensory neuropathy (HMSN/HNPP) Deletion/duplication analysis (PMP22 gene only) Extended mutation screening by specific request only	6
Huntington disease Presymptomatic referral only accepted from Clinical Genetics	3
Incontinentia pigmenti (IP)* Common NEMO deletion	3
Liddle syndrome*	1, 2
Medium-chain acyl CoA dehydrogenase deficiency Common mutation K329E (p.K304E) only	2
Metachromatic leukodystrophy	6
Mitochondrial disorders MELAS, MERRF, LHON, NARP Requires completed clinical information sheet	6

Tests offered:

Multiple endocrine neoplasia Types 1 & 2 Requires completed clinical information sheet	6
Myotonic dystrophy (DM1)	3a
Neurofibromatosis type 1 (NF1)*	1, 2
Neurofibromatosis type 2 (NF2)	6
Oculopharyngeal muscular dystrophy (OPMD)	6
Prader-Willi syndrome (PWS)	4, 5
Polycystic kidney disease (PKD2)*	1, 2
Rabson Mendenhall syndrome	1, 2
Renal tubular acidosis (dRTA)*	1, 2
Retinoschisis, X-linked (RS1)*	1, 2
Sensory neural hearing loss (SNHL) Common mutations only (Connexin 26/30)	6
Spinal bulbar muscular atrophy (Kennedy disease)	3
Spinal muscular atrophy (SMA)	6
Spinocerebellar ataxia (SCA) Types 1, 2, 3, 6, 7 & 17	3
Stickler syndrome	2, 4
Thyroid hormone resistance (THR)	1, 2
Tuberous sclerosis (TSC)*	1, 2
Uromodulin nephropathy (MCKD2/FJHN)	1, 2
Von Hippel-Lindau syndrome*	1, 2
Confirmation of mutation found on a research basis Index case Other family members	8 weeks 2

*further information regarding testing strategy available at www.addenbrookes.org.uk/moleculargenetics

ALL TEST REQUESTS ARE REVIEWED BY THE GENETICS SERVICE