Aberdeen Medical Genetics Laboratory
User Information Manual

Updated 23/11/2014:
- Molecular Haematological Oncology reporting times updated

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Medical Genetics – Phone Numbers

General enquiries
- Cytogenetics 01224 553820
- Molecular Genetics 01224 550682
- Clinical Genetics 01224 552120

Prof Zosia Miedzybrodzka 01224 552120
(Service Clinical Director and consultant in Clinical Genetics)

Mr David Stevenson 01224 550931
(Head of Cytogenetics and consultant in Clinical Cytogenetics)

Dr Kevin Kelly 01224 553888
(Head of Molecular Genetics and consultant in Molecular Genetics)

Ms Caroline Clark 01224 559972
(Deputy Head of Molecular Genetics)

Laboratory Address
Specimens should be sent to:
- Medical Genetics Laboratories
- Polwarth Building
- Medical School
- Foresterhill
- Aberdeen
- AB25 2ZD

Laboratory email address
grampian.molgen@nhs.net

Laboratory website
http://www.nhsgrampian.org/medicalgenetics

Laboratory Hours
- Monday – Friday 0900 - 1700 (there is no out of hours service)
Medical Genetics Service

Medical genetics offers a range of laboratory services including; cytogenetic, molecular cytogenetic and molecular genetic investigations on various sample types. Details of these tests and sample requirements are detailed below.

Consent for Genetic Testing

Genetic test results may have implications for relatives and families. DNA samples are normally stored when current diagnostic testing is complete. These issues should be discussed with patients, parents or guardians prior to sending a sample for a genetic test. Telephone advice and consent forms are available from the Department of Medical Genetics (tel. 01224 552120).

Contacting Medical Genetics Laboratory Services

Cytogenetics

For general enquiries, requests for results and to discuss particular cases please phone the Duty Scientist on 01224 553820. The Duty Scientist may not always be available immediately, however, a mail box service is available on this extension and is regularly checked.

Molecular Genetics

For general enquiries, requests for results and to discuss particular cases please phone 01224 550682. The Duty Scientist may not always be available immediately, however, a mail box service is available on this extension and is regularly checked.

Collection of Samples

Please refer to the Laboratory Medicine Clinical Unit Sample Acceptance Policy (available on the intranet).

Completion of Referral Form

The laboratory’s records form part of the genetic register. This is dependent on full details being given on each referral form, i.e. patient CHI number, full name, address, date of birth, sex, full clinical abstract, referring consultant, name and contact number of sender and place to which result is to be sent.

Sample Containers

Use of the correct specimen container is essential. The correct type of container for each sample type or investigation is detailed below. Certain sample types must be transported in sample tubes containing transport medium, these are supplied by the laboratory and are detailed below.
Sample Collection
Asepsis in the collection of specimens is essential. A specimen should be placed in a correctly labelled sterile container, and should reach the laboratory within the intervals indicated (see below). Specimen containers must be clearly labelled with the patient’s full name and CHI number or date of birth.

High Risk Samples
The department should be contacted in advance of forwarding high risk specimens. All packaging and referral forms should be clearly marked as ‘High Risk’.

Infection Hazards
Please help to minimise the risk to laboratory staff and porters by:
(a) discarding cracked tubes or those with tops off,
(b) avoid overfilling and contaminating the outside of containers, and
(c) make sure that tubes and bottles are securely stoppered.

Sample Transportation

Portering Service
Sample containers should be placed in a zip lock specimen bag and the referral form should be placed in the outer pocket of this bag.

Postal Service
Packaging requirements are detailed in appendix 1.2 of the HSE document, ‘Biological Agents: Managing the risks in laboratories and healthcare premises’ (for details, click on the link below):
Referrals for Chromosome Diagnosis

The laboratory can give an effective service if referrals are restricted to those cases where there is a good clinical reason for the examination. The Head of Cytogenetics will be happy to discuss the investigation of individual patients before samples are taken. A full clinical abstract should accompany each sample so that the laboratory can judge which procedures are required.

Time limit for requests for extra tests

It is policy to store fixed cell suspension (where available) for a period of 1 year. Additional FISH testing can be requested during this period – contact the laboratory to check availability of cell suspension.

Constitutional samples

Blood Chromosome Diagnosis

- Sterile lithium heparin tube (match the volume of the tube to the volume of the sample)
- 5 - 10 ml (adult) or 1-2ml (child) of venous blood
- Post-mortem cardiac blood and cord blood are also acceptable
- Roll the sample gently to prevent clotting.
- The sample should reach the Laboratory within 24 hours

Fanconi Anaemia Testing

- Sterile lithium heparin tube (match the volume of the tube to the volume of the sample)
- 5 - 10 ml (adult) or 2.5ml (child) of venous blood

In addition to the patient sample send a control sample (5 – 10ml of venous blood) when available

Prenatal Diagnosis

Rapid prenatal diagnosis and full karyotype analysis are carried out on all prenatal samples. A sample of maternal blood is required to complete the rapid diagnosis

Rapid prenatal diagnosis is a molecular test (QF-PCR) used to detect common autosome aneuploidies (13, 18 and 21) and sex chromosome aneuploidy. NB blood staining of amniotic fluid samples can hamper the rapid prenatal test and patients should be advised of this.

Prenatal samples should be forwarded to the laboratory as soon as possible after being taken. If for any reason there is a delay in transportation the samples should be kept at room temperature.

Amniotic Fluid

Prolonged inactivity of the mother, prior to the test, should be avoided to prevent settling of the amniotic fluid cells.
A sample of about 20ml (total volume) of amniotic fluid is required. The containers required can be obtained from the laboratory.

Split the sample as follows:

- 2ml in a sterile, labelled centrifuge tube, accompanied (in the same sample bag) by a 5ml sample of maternal blood in an EDTA tube (for rapid prenatal diagnosis).
- 10-18ml in a sterile, labelled universal container (for conventional cytogenetic analysis)

A separate referral form should accompany each portion of the sample.

**Trophoblastic Villi (CVS)**

A supply of sterile flasks containing CVS transport medium is available on request from the laboratory. These must be used on the day that they are prepared and for that reason the laboratory should be informed in advance of taking the sample. Place each aspirate in a separate transport flask and label each flask.

- 10-20mg of tissue are required
- 5ml sample of maternal blood in an EDTA tube

A portion of the CVS and the blood sample will be forwarded for rapid prenatal diagnosis.

**Solid Tissue (Biopsy and Necropsy)**

A supply of sterile tubes containing tissue transport medium is available on request from the laboratory. These have a limited shelf life which is indicated on the tube.

From stillbirths, neonatal deaths, etc., a surface-sterilised skin sample 10 mm long and 2 mm wide is placed into the medium. Samples from abortions and products of conception should also be sent in transport medium. Recognisable fetal parts and fetuses over 2cm in length should be sent to Pathology in the first instance. If required, they will then forward appropriate material for analysis. Samples should be delivered to the laboratory as soon as possible after being taken.

Samples with suspected common autosomal aneuploidy or sex chromosome aneuploidy may be investigated by QF-PCR. These samples should be collected as above. A sub-sample will be selected and forwarded for rapid aneuploidy screening.

**Referrals for Molecular Cytogenetics (FISH)**

- Molecular cytogenetics can be carried out on all sample types
- Collect these samples as for Chromosome Diagnosis
- The same sample can be used for both cytogenetic and molecular cytogenetic analysis

The request for molecular cytogenetics must be clearly marked on the referral form. Molecular cytogenetics is available for microdeletion syndromes (e.g. DiGeorge).

In addition, it is possible in certain circumstances to analyse samples using formalin fixed paraffin embedded sections of either 2 or 4 microns thick (depending on cellularity). If desired, please contact the laboratory to see if the particular investigation sought is possible by this method.
Referrals for microarray analysis.
Microarray analysis is available for patients with learning difficulties and multiple congenital abnormalities.
  - EDTA tube
  - 5ml of venous blood

Depending on the findings of the microarray analysis, it may be necessary to obtain samples from BOTH parents and a second sample from the patient to complete the study.

Oncology Samples
All samples should arrive at the laboratory before 3.00 pm. To allow the investigation of slowly growing cells, it is preferable that samples are not sent on a Friday. Samples for molecular oncology must be received within 1 hour of being taken, and must also be received during normal working hours, to allow appropriate storage prior to further processing. On a Friday, they should not arrive after 4.00 pm, to allow time for appropriate processing.

Bone Marrow Chromosome Diagnosis
  - Sterile tubes containing marrow transport medium (available on request from the laboratory - limited shelf life which is indicated on the tube)
  - The sample of bone marrow should be placed in a tube and gently inverted to prevent clotting
  - Deliver the sample to the laboratory at once

Bone Marrow Molecular Genetic Analysis (RT-PCR)
For screening of common fusion gene transcript detection (BCR-ABL1 in CML & ALL; PML-RARA, RUNX1T1-RUNX1, CBFB–MYH11, Flt3 and NPM1 in AML; MLL-AF4, TEL-RUNX1, E2A-PBX in ALL)
  - EDTA tube
  - 1-2ml of bone marrow
  - Place in a tube and gently invert to prevent clotting
  - Deliver the sample to the laboratory at once, if possible

Tumour and Lymph Node Chromosome Diagnosis
  - Sterile tubes containing lymph node transport medium (available on request from the laboratory - limited shelf life which is indicated on the tube)
  - Tumour samples should be fresh, and free from fat or necrotic tissue.
  - It is essential that the sample is delivered to the laboratory as soon as possible after being taken

Tumour and Lymph Node Molecular Genetic Analysis (DNA-based Clonality PCR)
Tumour or lymph node samples from suspected lymphoproliferations for IGH and/or TCR gene re-arrangement analysis, should be sent as for chromosome diagnosis outlined above.
It is possible to analyse samples using formalin fixed paraffin embedded sections of 5 to 10 microns thick (depending on cellularity).

**Blood Chromosome Diagnosis**
- Sterile tubes containing marrow transport medium (available on request from the laboratory - limited shelf life which is indicated on the tube)
- 10 ml (adult) or 1-2ml (child) of venous blood should be placed in the tube
- Roll gently, to prevent clotting
- Should reach the Laboratory as soon as possible after being taken.

If marrow transport medium is not available sterile lithium heparin tubes will suffice, please match the volume of the tube to the volume of the sample.

**Blood Molecular Genetic Analysis (RT-PCR & DNA-based JAK2 PCR & post-transplant mixed chimaerism analysis)**
For post-treatment monitoring of common fusion gene transcript levels (BCR-ABL1 in CML; and PML-RARA in AML M3)
- EDTA tube
- 10-20ml venous blood
- Gently invert to prevent clotting
- Deliver the sample to the laboratory at once, if possible, especially when RNA extraction is required.

For DNA-based JAK2 PCR of suspected MPN patients
- EDTA tube
- 4-6ml venous blood

For post-transplant PCR studies
- EDTA tube
- 10ml venous blood

**Effusion Chromosome Diagnosis**
- Sterile tubes containing lymph node transport medium (available on request from the laboratory - limited shelf life which is indicated on the tube)
- Place the aspirate in the medium.
- It is essential that the sample is delivered to the laboratory as soon as possible after being taken.

**Molecular Cytogenetic (FISH) Analysis of Oncology Samples**
A number of FISH tests are available for oncology samples. Collect the samples as outlined above and indicate clearly on the referral form that molecular investigation is required.

It is possible in certain circumstances to analyse samples using formalin fixed paraffin embedded sections of either 2 or 4 microns thick (depending on cellularity). If desired, please contact the laboratory to see if the particular investigation sought is possible by this method.
Cytogenetic Sample Turnaround Times

Turnaround times are dependent on the sample type. National guidelines on reporting times are followed where possible and are summarised below. The quoted turnaround times are for conventional and molecular cytogenetics.

<table>
<thead>
<tr>
<th>Sample type</th>
<th>Recommended turnaround time (days)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Amniotic fluid (full karyotype)</td>
<td>14</td>
</tr>
<tr>
<td>Amniotic fluid (trisomy screen)*</td>
<td>2</td>
</tr>
<tr>
<td>Blood (routine)</td>
<td>28</td>
</tr>
<tr>
<td>Blood (urgent)</td>
<td>10</td>
</tr>
<tr>
<td>CVS (full karyotype)</td>
<td>14</td>
</tr>
<tr>
<td>CVS (trisomy screen)*</td>
<td>2</td>
</tr>
<tr>
<td>FISH constitutional</td>
<td>28</td>
</tr>
<tr>
<td>Microarray analysis</td>
<td>28</td>
</tr>
<tr>
<td>Molecular Haematological Oncology (routine)</td>
<td>21</td>
</tr>
<tr>
<td>Molecular Haematological Oncology (urgent)</td>
<td>14</td>
</tr>
<tr>
<td>Oncology (routine)</td>
<td>21</td>
</tr>
<tr>
<td>Oncology (urgent)</td>
<td>14</td>
</tr>
<tr>
<td>Solid tissue</td>
<td>28</td>
</tr>
<tr>
<td>Solid tissue (trisomy screen)*</td>
<td>2</td>
</tr>
</tbody>
</table>

Also see section- ‘Referrals for DNA diagnosis’

95% of samples should have an issued final report within these times
Referrals for DNA Diagnosis (other than acquired oncology analysis)

The laboratory offers a service of DNA extraction and analysis for genetic diseases. These are summarised in the table below. Clinical enquiries should be directed first to the ON-CALL Clinical Geneticist (01224 552120).

The Head of Molecular Genetics can be contacted on 01224 553888 and can to provide information on the laboratory service and sample collection and storage.

Time limit for requests for extra tests

Providing DNA is available, extra tests can be requested at any time after receipt of the original sample.

Sample Requirements

- Samples of blood (5-10 ml from adults and 1 - 5ml from children) in an EDTA tube
- Unfixed tissue (50-100 mg) in a sterile container are suitable for DNA extraction.

NB DNA cannot be successfully extracted from clotted blood.

Rapid Aneuploidy screening, QF-PCR

- Rapid prenatal diagnosis is achieved by extracting DNA from a 2ml aliquot of amniotic fluid or enzyme digested villi from a CVS.
- A 5ml sample of maternal blood in an EDTA tube is required to complete these studies.
- Samples should be forwarded to the laboratory as soon as possible after sampling (see prenatal samples above for details of sample collection).

NB blood staining of amniotic fluid samples can hamper the rapid prenatal test and patients should be advised of this.

Rapid aneuploidy screening of tissue samples is available (see solid tissue samples above for details of sample collection).

Storage of DNA Samples

When current diagnostic testing is complete, DNA samples are retained in storage in the NE Scotland DNA bank unless otherwise directed. These issues should be discussed with patients, parents or guardians prior to sending a sample for a genetic test. Telephone advice and consent forms are available from the Department of Medical Genetics (tel. 01224 552120).

Scottish Molecular Genetics Consortium

In addition to the disorders listed below many other conditions can be tested through the Scottish Molecular Genetics Consortium. Tests available in Scotland include: Prader Willi / Angelman syndrome, Huntington disease, SCA and Duchenne MD. Please refer to the Clinical Genetics Service (01224 552120) for information.

For information on haemophilia, haemoglobinopathies, connective tissue disorders, rare cystic fibrosis mutation or any other genetic disorder not mentioned contact the laboratory on 01224 559972 or the Clinical Genetics Service on 01224 552120.
Please also refer to the CMGS website [http://www.cmgs.org/](http://www.cmgs.org/)

### Genetic Disease Analysis Services Available:

<table>
<thead>
<tr>
<th>Disorder</th>
<th>Service</th>
<th>Genes</th>
<th>Can be referred by</th>
<th>Reporting time (working days)*</th>
</tr>
</thead>
<tbody>
<tr>
<td>ARVC</td>
<td>Mutation screen</td>
<td>PKP2, DSG2, DSP, DSC2</td>
<td>Clinical geneticist/pathologist</td>
<td>40</td>
</tr>
<tr>
<td></td>
<td>Predictive test, mutation known</td>
<td></td>
<td></td>
<td>10</td>
</tr>
<tr>
<td>ARX</td>
<td>Exon 2 duplication</td>
<td>ARX</td>
<td>Clinical geneticist</td>
<td>20</td>
</tr>
<tr>
<td>Breast/ovarian cancer (familial)</td>
<td>Mutation screen</td>
<td>BRCA1, BRCA2, Ovarian also RAD51C, RAD51D</td>
<td>Clinical geneticist, consultant oncologist</td>
<td>40</td>
</tr>
<tr>
<td></td>
<td>Predictive test, mutation known</td>
<td></td>
<td></td>
<td>10</td>
</tr>
<tr>
<td>Brugada / Dilated Cardiomyopathy</td>
<td>Mutation screen</td>
<td>SCN5A</td>
<td>Clinical geneticist/pathologist</td>
<td>40</td>
</tr>
<tr>
<td></td>
<td>Predictive test, mutation known</td>
<td></td>
<td></td>
<td>10</td>
</tr>
<tr>
<td>Charcot Marie Tooth Disease</td>
<td>MLPA and/or mutation screen.</td>
<td></td>
<td>Clinical geneticist/neurologist</td>
<td>40/gene panel</td>
</tr>
<tr>
<td></td>
<td>Predictive test, mutation known</td>
<td></td>
<td></td>
<td>10</td>
</tr>
<tr>
<td>Distal Hereditary Motor Neuropathy</td>
<td>Mutation screen</td>
<td>HSPB1, HSPB3, HSPB8, GARS, BSC12, TRPV4, DNM2, RAB7A, EGR2</td>
<td>Clinical geneticist/neurologist</td>
<td>40/gene panel</td>
</tr>
<tr>
<td></td>
<td>Predictive test, mutation known</td>
<td></td>
<td></td>
<td>Refer to clinical genetics</td>
</tr>
<tr>
<td>Colorectal Cancer</td>
<td>Mutation screen (tumour)</td>
<td>KRAS codons 12,13,61, BRAF codon 600</td>
<td>Oncologist/Pathologist</td>
<td>7</td>
</tr>
<tr>
<td>Cowdens</td>
<td>Mutation screen</td>
<td>PTEN</td>
<td>Clinical geneticist, consultant oncologist</td>
<td>40</td>
</tr>
<tr>
<td></td>
<td>Predictive test, mutation known</td>
<td></td>
<td></td>
<td>Refer to clinical genetics</td>
</tr>
<tr>
<td>CPVT</td>
<td>Mutation screen</td>
<td>RYR2</td>
<td>Clinical geneticist/pathologist</td>
<td>40</td>
</tr>
<tr>
<td></td>
<td>Predictive test, mutation known</td>
<td></td>
<td></td>
<td>Refer to clinical genetics</td>
</tr>
<tr>
<td>Cystic fibrosis</td>
<td>Mutation screen: 29 mutations +/- intron 8 poly-T</td>
<td>CFTR</td>
<td>Hospital specialists</td>
<td>20 non urgent, 7 urgent</td>
</tr>
<tr>
<td>Factor V Leiden</td>
<td>1691 G &gt; A mutation</td>
<td>F5</td>
<td>Any physician</td>
<td>20</td>
</tr>
<tr>
<td>Familial Hypercholesterolaemia</td>
<td>Mutation screen</td>
<td>LDLR, ApoB Ex26, PCSK9 Ex7</td>
<td>Clinical geneticist/consultant lipidologist</td>
<td>40</td>
</tr>
<tr>
<td></td>
<td>Predictive test, mutation known</td>
<td></td>
<td></td>
<td>Refer to clinical genetics</td>
</tr>
<tr>
<td>Fragile X A</td>
<td>FRAXA expansion</td>
<td>FMR1</td>
<td>Hospital specialists</td>
<td>20 non urgent, 7 urgent</td>
</tr>
<tr>
<td></td>
<td>Predictive test, mutation known</td>
<td></td>
<td></td>
<td>Refer to clinical genetics</td>
</tr>
<tr>
<td>Glucocorticoid remediable aldosteronism GRA</td>
<td>Chimaeric gene product</td>
<td></td>
<td>Hospital specialists</td>
<td>20</td>
</tr>
<tr>
<td>Haemochromatosis (familial)</td>
<td>C282Y and H63D mutations</td>
<td>HFE</td>
<td>Any physician</td>
<td>20</td>
</tr>
<tr>
<td>Hereditary and Sensory Autonomic Neuropathy</td>
<td>Mutation screen</td>
<td>SPTLC1</td>
<td>Clinical geneticist/neurologist</td>
<td>40</td>
</tr>
<tr>
<td></td>
<td>Predictive test, mutation known</td>
<td></td>
<td></td>
<td>Refer to clinical genetics</td>
</tr>
<tr>
<td>Disorder</td>
<td>Service</td>
<td>Genes</td>
<td>Can be referred by</td>
<td>Reporting time (working days)*</td>
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<tr>
<td>-------------------------------</td>
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<td>Hyperlipidaemia Type III</td>
<td>Mutation screen</td>
<td>ApoE codons 130, 176</td>
<td>Clinical geneticist, consultant lipidologist</td>
<td>20</td>
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<tr>
<td>Hypertriglyceridaemia</td>
<td>Mutation screen</td>
<td>LPL, Apo C2, Apo A5</td>
<td>Clinical geneticist, consultant lipidologist</td>
<td>40</td>
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<tr>
<td></td>
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<td>GPI-HBP1, LMF1,</td>
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<td>Predictive test, mutation known</td>
<td></td>
<td>Refer to clinical genetics</td>
<td>10</td>
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<tr>
<td>Li Fraumeni</td>
<td>Mutation screen</td>
<td>TP53</td>
<td>Clinical geneticist, consultant oncologist</td>
<td>40</td>
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<td></td>
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<tr>
<td>Long QT syndrome</td>
<td>Mutation screen</td>
<td>KCNQ1, KCNH2, KCNE1, KCNE2, SCN5A, KCNJ2 available upon request</td>
<td>Clinical geneticist/cardiologist/pathologist</td>
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<td>Lung Cancer</td>
<td>Mutation screen (tumour)</td>
<td>EGFR exons 18-21 KRAS</td>
<td>Oncologist/Pathologist</td>
<td>7</td>
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<tr>
<td></td>
<td></td>
<td>codons 12, 13, 61 BRAF codon 600</td>
<td></td>
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<td>Melanoma</td>
<td>Mutation screen (tumour)</td>
<td>BRAF codon 600</td>
<td>Oncologist/Pathologist</td>
<td>7</td>
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<tr>
<td>Myotonic dystrophy</td>
<td>DM1, DM2 gene expansion</td>
<td></td>
<td>Hospital specialists/Ophthalmologists</td>
<td>20 non urgent, 7 urgent</td>
</tr>
<tr>
<td>DM 1 and DM2</td>
<td></td>
<td></td>
<td></td>
<td></td>
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<tr>
<td>Prothrombin 20210A</td>
<td>20210A mutation</td>
<td>F2</td>
<td>Hospital specialist</td>
<td>20</td>
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<td>RNF135</td>
<td>Mutation screen</td>
<td>RNF135</td>
<td>Refer to clinical genetics</td>
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<td>RR-MADD</td>
<td>Mutation screen</td>
<td>ETFDH</td>
<td>Refer to clinical genetics</td>
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<tr>
<td></td>
<td>Predictive test, mutation known</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Rapid prenatal diagnosis</td>
<td>Chromosomes 13, 18 and 21 &amp; sex chromosomes</td>
<td></td>
<td>Hospital specialist</td>
<td>2</td>
</tr>
<tr>
<td>Sickle cell anaemia</td>
<td>Common mutation E7V</td>
<td>HBB</td>
<td>Refer to clinical genetics</td>
<td>10 days - carrier testing</td>
</tr>
<tr>
<td>Torsion dystonia</td>
<td>3bp deletion in DYT1 gene</td>
<td>DYT1</td>
<td>Refer to clinical genetics</td>
<td>20</td>
</tr>
<tr>
<td>Zygosity</td>
<td>Informative marker multiplex</td>
<td></td>
<td>Refer to clinical genetics</td>
<td>20 non urgent, 7 urgent</td>
</tr>
</tbody>
</table>

*Reporting times are set by national agreement with the National Services Division. For any prenatal diagnosis (except trisomy screen) the contracted time is 95% within 7 working days.